UTS UNIVERSITY OF TECHNOLOGY SYDNEY

# Genomic Information for Families of the Terminally ill (GIFT) Project: Building Evidence for An Intervention Using Mixed Methods

**by Stephanie White**

Thesis submitted in fulfilment of the requirements for the degree of

**Doctor of Philosophy**

under the supervision of Dr Chris Jacobs, Dr Erin Turbitt and Professor Jane Phillips

University of Technology Sydney
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April 2023

# CERTIFICATE OF ORIGINAL AUTHORSHIP

I, Stephanie White, declare that this thesis, is submitted in fulfilment of the requirements for the award of Doctor of Philosophy (95601 Genetic Counselling), in the Graduate School of Health at the University of Technology Sydney.

This thesis is wholly my own work unless otherwise referenced or acknowledged. In addition, I certify that all information sources and literature used are indicated in the thesis.

This document has not been submitted for qualifications at any other academic institution.

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## STATEMENT OF FORMAT

This is a thesis by compilation written in seven chapters. In Chapter 1, an introduction to genetic counselling and testing in palliative care is presented. Chapter 2 presents a critical engagement with relevant literature, including a published, peer-reviewed systematic review manuscript. Chapter 3 describes the principles, philosophy and design of the GIFT Project. Chapter 4 contains two peer-reviewed, published manuscripts from a qualitative study, and Chapter 5 presents a peer-reviewed, published scoping review manuscript. The manuscript in Chapter 6 has been peer-reviewed and a revised version is currently under consideration. This manuscript may not represent the final published work. In Chapter 7, the results of data integration and discussion of the findings are presented. The references from each chapter are presented in a final bibliography at the end of Chapter 7.

Author contributions to each study are detailed in the following section. All other text within this thesis is Stephanie White's original writing, revised with feedback from Dr Chris Jacobs, Professor Jane Phillips and Dr Erin Turbitt.

Four of the five studies have been peer-reviewed and published, with one currently under consideration. The publishing journals have provided permission for inclusion in this thesis as required. Minor formatting changes have been made to these papers for congruency throughout the thesis. For example, all referencing has been changed to Vancouver style. Spelling has been altered to reflect Australian English and table/figure/page numbers have been updated so these flow correctly throughout the thesis. The appendices contain supplementary files for each chapter.

## PUBLICATIONS AND AUTHOR CONTRIBUTIONS

| | |
|---|---|
| **Chapter 2 reference:** White S, Jacobs C, Phillips J. Mainstreaming genetics and genomics: a systematic review of the barriers and facilitators for nurses and physicians in secondary and tertiary care. Genet Med. 2020;22(7):1149-55. | |
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| **Chapter 4a reference:** White S, Turbitt E, Phillips J, Jacobs C. Approaching discussions about genetics with palliative patients, and their families: a qualitative exploration with genetic health professionals. Eur J Hum Genet. 2022:1-8 | |
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| Journal & ranking | Supportive Care in Cancer<br>2021 Impact Factor 3.359, Scimago rating Q2 (oncology) |
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| **Chapter 5 reference:** White S, Virdun C, McErlean G, Phillips J, Jacobs C. Integrating genomics into palliative care: a global scoping review of policy recommendations. Public Health Genomics. 2022:1-15. | |
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| **Chapter 6 reference:** White S, Turbitt E, Rogers K, Tucker K, Best M, Phillips J, et al. A survey of genetic and palliative care health professionals' views towards the integration of genetics into palliative care. Eur J Hum Genet. 2023. (manuscript under review). | |
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I, the undersigned, confirm the statements regarding author contributions are an accurate representation of my involvement with the study or studies within this thesis.

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## RESEARCH DISSEMINATION

**Invited oral presentations**

White S. Genetic information for families of the terminally ill: the GIFT project – A mixed methods PhD. Paper presented at: Genetic Counsellors in Research (GCR) connect monthly virtual seminar, 2022 September 29

White S. Nurse-led research in cancer genetics: a palliative care PhD. Paper presented at: the Cancer Nurses Society of Australia annual conference, 2022 June 18, Brisbane, Australia

White S. Genetics and palliative care: how should we integrate this into care provision? Paper presented at: Palliative Care Nurses Australia virtual conference, 2022 May 3

White S. "It's amazing how many oncology patients have said, 'You're the first person to ask me that question'". Exploring the views and experiences of Australasian palliative care clinicians towards discussing genetics with patients and their families: a qualitative study. Paper presented at: Familial Aspects of Cancer: research and practice virtual meeting, 2021 August 25

White S. Integrating genetic counselling into specialist nurses and physicians' clinical practice: a systematic review of the barriers and facilitators. Paper presented at: NSW Genetic Counselling Education Day, 2019 December 12, Sydney, Australia

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White S, Turbitt E, Phillips J, Jacobs C. "Some families want to know, and some don't want to know, and then where do you sit?" Exploring the views and experiences of Australasian palliative care clinicians towards discussing genetics with patients and their families: a qualitative study. Paper presented at: Ocean Palliative Care virtual conference, 2021 September 8

White S, Jacobs C, Phillips J. Genetics in palliative care:  s systematic review of the barriers and facilitators for nurses and physicians. Paper presented at: Palliative Care Nurses Australia virtual conference, 2020 November 22

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White S. Exploring health professionals' views and experiences of approaching discussions about genetics with people who have palliative care needs (3-minute thesis). Paper presented at: Faculty of Health Research Student Conference 3MT competition, virtual, 2022 June 22

White S. Exploring health professionals' views and experiences of approaching discussions about genetics with people who have palliative care needs (3-minute thesis). Paper presented at: Graduate School of Health 3MT competition, virtual, 2022 June 17 (first prize awarded)

White S, Phillips J, Jacobs C. Genetics in palliative care: progress on a qualitative study exploring the views and experiences of palliative care doctors (poster presentation). Paper presented at: Faculty of Health research student virtual conference, 2020 November 18

White S, Jacobs C, Phillips J. Integrating genetic counselling into specialist nurses and physicians' clinical practice: A systematic review of the barriers and facilitators. Paper presented at: Faculty of Health research student conference, 2019 November 28, Sydney, Australia

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# LIST OF ABBREVIATIONS

| Abbreviation | Definition |
|---|---|
| AACG | Australasian Association of Clinical Geneticists |
| AGREE | Appraisal of guidelines, research and evaluation |
| ANZSPM | Australia and New Zealand Society of Palliative Medicine |
| ASGC | Australasian Society of Genetic Counsellors |
| BCW | Behaviour change wheel |
| BRCA1/2 | Breast cancer gene 1 and 2 |
| CADTH | Canadian Agency for Drugs and Technologies in Health |
| COM-B | Capability, opportunity, motivation behaviour system |
| CINAHL | Cumulative index to nursing and allied health literature |
| CMA | Chromosomal microarray |
| COREQ | Consolidated criteria for reporting qualitative research |
| COVID-19 | Coronavirus disease 2019 |
| DNA | Deoxyribonucleic acid |
| EFA | Exploratory factor analysis |
| ELSI | Ethical, legal, and social implications |
| EIU QOD | Economist Intelligence Unit Quality of death index |
| GC | Genetic counsellor |
| G-HP | Genetic health professional |
| GIFT Project | Genomic information for families of the terminally ill project |
| GRADE | Grading of recommendations, assessment, development and evaluations |
| GSH | Graduate School of Health |
| HGSA | Human Genetics Society of Australasia |
| HREC | Human research ethics committee |
| IVF | In-vitro fertilisation |
| MRC | Medical Research Council |
| NZ | New Zealand |
| PC-HP | Palliative care health professional |
| PCNA | Palliative Care Nurses Australia |
| PCNNZ | Palliative Care Nurses New Zealand |
| PGD | Preimplantation genetic diagnosis |
| PICO | Population, intervention, comparator, outcome |
| PND | Prenatal diagnosis |
| PRISMA | Preferred reporting items for systematic reviews and meta-analyses |
| PROSPERO | International prospective register of systematic reviews |
| QUAL/qual | Qualitative |

| | |
|---|---|
| QUAN/quan | Quantitative |
| REM | Reciprocal engagement model |
| STROBE | Strengthening the reporting of observational studies in epidemiology |
| TDF | Theoretical domains framework |
| UK | United Kingdom |
| USA | United States of America |
| UTS | University of Technology Sydney |
| VUS | Variant of uncertain significance |
| WHO ICCC | World Health Organization Innovative care for chronic conditions |

## KEY TERMS

| Term | Definition |
|---|---|
| Australasia | Australia and New Zealand[1] |
| Clinical geneticist | A medical doctor who has completed, or is currently completing, advanced training in Clinical Genetics with the Royal Australasian College of Physicians[2] |
| Diagnostic genetic testing | Genetic or genomic testing undertaken in a person affected with the purpose of identifying a causative pathogenic variant for the first time in a family |
| Evidence-based intervention | A health-based intervention that has been empirically assessed and shown to improve clinical outcomes |
| Exploratory sequential design | A mixed methods research design with an initial exploratory (qualitative) phase, followed by a generalising (quantitative) phase |
| Family member | A blood relative[1] |
| Genetic counselling | "Genetic counseling is the process of helping people understand and adapt to the medical, psychological and familial implications of genetic contributions to disease. This process integrates the following, (1) Interpretation of family and medical histories to assess the chance of disease occurrence or recurrence, (2) Education about inheritance, testing, management, prevention, resources and research, (3) Counseling to promote informed choices and adaptation to the risk or condition".[3] Genetic counselling can be provided by genetic, and non-genetic, health professionals |
| Genetic counsellor | Allied health professionals who have completed specialised training through a Master of Genetic Counselling program approved by the Human Genetics Society of Australasia |
| Genetic health professional | Genetic counsellors and clinical geneticists[1] |
| Human Genetics Society of Australasia | The peak professional body regulating genetic health professionals, and other professionals working in human genetics, in Australasia |
| Inference | A high-level interpretation (or conclusion) drawn from a single study or data set |
| Interdisciplinary collaboration | The involvement of more than one area of professional expertise to collaborate on a clinical or research issue (e.g., collaboration between genetic counsellors and palliative care health professionals is interdisciplinary) |
| Mainstreaming | Non-genetic health professionals incorporating genetic counselling, or aspects of genetic counselling, into their clinical practice |
| Meta-inference | The conclusions drawn from integrating inferences generated from more than one study |
| Mixed methods research | A type of research that involves the collection, analysis, and integration of qualitative and quantitative data |
| Palliative care | A philosophy of care "that improves the quality of life of patients (adults and children) and their families who are facing the problems associated with life-limiting illness, through the prevention and relief of suffering by means of early identification and correct assessment and treatment of pain and other problems, whether physical, psychosocial or spiritual."[4] |

| | |
|---|---|
| Palliative care context/setting | Any clinical setting in which a person is receiving palliative care from a palliative care health professional. This could include palliative care delivered in the home, hospital, hospice, or elsewhere[1] |
| Palliative medicine doctor | A medical doctor who has completed, or is currently undertaking, advanced training in Palliative Medicine with the Royal Australasian College of Physicians[2] |
| Palliative care health professional | A palliative care nurse or doctor[1] |
| Palliative care nurse | A registered nurse with specialist skills and training in the provision of palliative care[5] |
| Person with palliative care needs | A person, at any life stage, who is receiving palliative care from a palliative care health professional |
| Research participant | A palliative care or genetic health professional who consented to participating in the studies within the GIFT project[1] |
| Pathogenic variant | A disease-causing variant to the DNA code of a gene, defined by the American College of Medical Genetics and Genomics (ACMG) criteria[6] |
| Patient | A person receiving clinical care within a health system |
| Predictive genetic testing | Targeted genetic testing of an unaffected relative for a pathogenic variant that has already been identified in an affected family member |
| Pre-implantation genetic diagnosis | Genetic testing of an embryo for a familial pathogenic variant with the intent of implanting an unaffected embryo |
| Prenatal diagnosis | Invasive testing of a pregnancy using chorionic villus sampling or amniocentesis to identify whether a fetus has inherited a pathogenic variant that has been previously identified in an affected family member |
| Relative | A blood-related family member |

1. These definitions are specific to this thesis and may be defined differently elsewhere.
2. According to the Royal Australasian College of Physicians: https://www.racp.edu.au/
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6. Richards S, Aziz N, Bale S, Bick D, Das S, Gastier-Foster J, et al. Standards and guidelines for the interpretation of sequence variants: a joint consensus recommendation of the American College of Medical Genetics and Genomics and the Association for Molecular Pathology. Genet Med.17(5):405-24.

## ABSTRACT

As genomic testing moves into routine healthcare, including palliative care, understanding the needs of health professionals will maximise the benefits of genomics while minimising potential harms. The GIFT Project aimed to build the Australasian evidence base by understanding the barriers to and facilitators of integrating genomics into the care of people who are palliative and their families.

A systematic review preceded this two-phase exploratory sequential mixed methods project. Searching across three databases, barriers and facilitators from 48 studies were narratively synthesised. Findings suggested health professionals have suboptimal genomics knowledge, and concerns that genomic discussions cause harm. Phase 1 consisted of an interpretive descriptive qualitative study with genetic (G-HPs; *n*=26) and palliative care health professionals (PC-HPs; *n*=14) using semi-structured interviews and focus groups. Data from each cohort was analysed with reflexive thematic analysis. G-HPs described the familial benefits of genomic testing, the discomfort of counselling near end of life, the challenge of getting genomics on the palliative agenda, and a preference for offering DNA storage. PC-HPs described delicately balancing the harms and benefits to patients and families, uncertainty about the palliative care role and responsibilities, and a lack of organisational support. Phase 1 continued with a scoping review of recommendations from 78 genomic and palliative care policies. Just 3% of the policies contained recommendations about integrating genomics into palliative care, highlighting a gap in the policy landscape.

Phase 2 was a cross-sectional, online questionnaire surveying G-HPs (*n*=29) and PC-HPs (*n*=44). Barriers and facilitators were assessed and compared between the two groups. The most frequently selected barrier by both groups was PC-HPs' lack of genetic knowledge (*n*=32/72, 44%). Only a quarter of PC-HPs were 'fairly confident' or 'confident' assessing when to broach a genomics discussion (*n*=8/33, 24%). Developing a genetic referral template (*n*=31, 43%) and fostering close professional relationships were most frequently selected as facilitators (*n*=27, 38%).

To identify the support required to integrate genomics into palliative care, findings from Phase 1 and 2 were integrated using a joint display table to generate four meta-inferences: G-HPs and PC-HPs need support to adapt their practice to the challenges of end of life, development of an interdisciplinary understanding of family-centred care in the palliative–genomic context, a culture of professional collaboration, and further evidence delivered to

policy stakeholders through co-designed, implementation research. Future research could build upon these findings by designing and implementing an intervention to support the integration of genomics into palliative care.

# 1 Introduction

## 1.1 PREAMBLE

Genetic counselling, and genetic and genomic testing (hereafter 'genomic testing') for people with palliative care needs can yield benefits for individuals and their family members. The primary clinical benefit is the ability to guide risk assessment and medical decision making for family members. The psychological benefits for individuals may include addressing existing concerns, making meaning from their illness, and leaving a legacy for their family. For health professionals, engaging in genomic discussions with people who have palliative care needs, particularly near the end of life, can raise several ethical, legal, and social issues. However, the process of navigating discussions about genetic counselling and genomic testing in the palliative context has received little attention.

In this introductory chapter, I explore the relationship between genetic counselling and genomic testing with people who have palliative care needs and their families. I begin by describing the evolution of clinical genomics, including the advances in genomic technology, benefits and challenges of genomic testing, and current genomics service delivery and workforce. I then provide context around genetic counselling and palliative care in Australasia. I discuss the relevance of genetic counselling and testing for people with palliative care needs and their families, and then describe how these factors drove the impetus for this doctoral project. This chapter concludes with the project aim and research questions.

## 1.2 EVOLUTION OF CLINICAL GENOMICS

Genomics has undergone rapid changes in the past 20 years.(1) Technological improvements to genomic testing have fuelled understanding of the association between the genome and disease development. As such, genomic information has growing relevance to clinical care across the lifespan.(2) For individuals, a key benefit of clinical genomics is treatments that target specific genetic variants. For family members, identifying a pathogenic variant in the affected person can provide them with information and options for managing their own future disease risk.(3) To ensure individuals and families benefit from genomic information, health professionals must remain current with the latest improvements and have the knowledge and skills to deliver these benefits to individuals and families.

### 1.2.1 Advances in Genomic Testing Technology

Although this thesis does not focus on genomic technology, a discussion about the evolution of genomic testing in the past two decades provides a backdrop to the integration of genomics into routine healthcare, including palliative care. I review the major updates to genomic sequencing and testing, but this section is not an exhaustive description of all genetic and genomic testing methods. For example, metabolic or biochemical tests, fluorescence in situ hybridisation, Southern blot analysis, and other such tests are beyond scope.

Since completion of the Human Genome Project in 2003, in which the entire human genome was sequenced for the first time, genomics has evolved into a routine technology.(4) Initially costing the Human Genome Project about 3 billion US dollars, a person's whole genome can now be sequenced for close to $1000 US dollars.(5) Plummeting costs have increased the uptake by health professionals and consumers, and genomic testing technologies are now relevant to all stages of the life cycle including prenatal testing, paediatrics, adults, end of life, and even post-mortem. Some examples of genomic testing applications are to investigate abnormal ultrasound findings in a pregnancy, identify a genetic cause for developmental delay in a child, or a familial disease such as cardiomyopathy, cancer or Alzheimer's disease.(2)

Sanger sequencing, in which DNA is interpreted one base pair at a time, is considered by many as the gold standard of DNA sequencing. Sanger sequencing is useful for testing one gene or a known variant in a gene. However, testing one gene (or part of a gene) is slow and expensive for clinical scenarios in which many different genes could be responsible for the clinical problem. In a situation in which many genes need testing, Sanger sequencing limits the ability to obtain a genetic diagnosis. More recently, next-generation sequencing provides the ability to interrogate multiple genes (genomic sequencing) simultaneously, which reduces the time and effort for testing and improves the diagnostic rate.(6). Genomic sequencing is an umbrella term referring to testing where all, or a portion, of a person's genome is analysed using next-generation sequencing technology.(7) This includes sequencing several or many genes related to a clinical presentation (panel testing), all the protein-coding regions (whole-exome sequencing), or all the protein-coding and non-protein-coding regions (whole-genome sequencing).(8)

Obtaining genomic information through cytogenetic techniques (such as karyotype and chromosomal microarray; CMA) has also advanced. The cytogenetic progress made throughout the 20$^{th}$ century revolutionised clinical genomics.(9) Before genomic sequencing

(as described above), cytogenetic techniques were the original genomic tests. Karyotyping, and later G-banded karyotyping, were used to visualise the chromosomes to identify aneuploidy (changes to the number of chromosomes) and copy number variation (large chromosome segmental duplications or deletions).(6) Karyotyping now has many (continued) applications, but the first applications in the 1970s were in prenatal testing for chromosomal conditions, such as Down syndrome, and identifying causes of syndromic intellectual disability. In the 1990s, CMA furthered the ability to detect aneuploidy and copy number variants with the quantitative assessment of the amount of genomic material.(9) Around 2010, CMA, became the preferred method for traditional karyotyping for certain patient groups.(10) CMAs are quicker, cheaper, and less complex, and have a higher diagnostic rate than traditional karyotyping.(10, 11)

### 1.2.2 Benefits of Genomic Information

#### *1.2.2.1 Diagnostic Testing for Individuals*

Alongside the evolution of genomic testing technologies, there have been advances in the clinical utility of genomic information. Since the completion of the Human Genome Project, knowledge about the association between genomic profiles and disease risk has flourished.(7) Diagnostic testing in an affected person involves searching for a causative pathogenic variant using genomic sequencing, cytogenetic techniques, or other methods. For individuals, this means greater diagnostic, prognostic, and treatment information. Common benefits include confirmation of the diagnosis (particularly if there is uncertainty), potential avenues for treatments, information about predicted disease severity and future disease risk, family planning information, and clinical trial participation.(12-16) For example, people with cancer routinely undergo genomic testing to identify therapeutic targets, such as a DNA repair defects.(17, 18) A person with advanced breast or ovarian cancer and an underlying *BRCA1* or *BRCA2* pathogenic variant may be provided with a PARP-inhibitor (olaparib), which has shown promising improvements in overall survival.(12) In addition, this person may learn that they have an increased risk for another primary cancer, helping them to make an informed decision to reduce their future risk with surgical or clinical management. Some clinical examples, alongside the potential benefits of diagnostic and predictive testing are provided in Table 1.

Table 1. Clinical examples to demonstrate the potential clinical benefits of diagnostic and predictive testing.

| **Clinical example** | **Clinical benefits of diagnostic testing for the affected person** | **Clinical benefits of predictive testing for unaffected relatives** | |
|---|---|---|---|
| | | **If positive for pathogenic variant[1, 2]** | **If negative for pathogenic variant** |
| Hereditary breast and ovarian cancer | • Confirm genetic diagnosis<br>• Depending on stage of disease, access to poly-ADP ribose inhibitor<br>• Information about future disease risk<br>• Clinical trial participation<br>• Access to preconception and prenatal genetic testing | • Access to high-risk breast screening<br>• Access to risk-reducing surgery (bilateral mastectomy and bilateral salpingo-oophorectomy).<br>• Access to preconception and prenatal genetic testing<br>• Risk-reducing medication<br>• Counselling for risk reduction through lifestyle factors | • Manage as population risk<br>• No increased risk of transmission to next generation |
| Familial adenomatous polyposis | • Confirm genetic diagnosis<br>• Unlikely to change medical management<br>• Clinical trial participation<br>• Access to preconception and prenatal genetic testing | • Access to high-risk colonoscopy screening and colectomy<br>• Access to preconception and prenatal genetic testing | • Manage as population risk<br>• No increased risk of transmission to next generation |
| Autosomal dominant Alzheimer's disease | • Confirm genetic diagnosis<br>• Unlikely to change medical management<br>• Clinical trial participation | • Participation in clinical trials<br>• Early diagnostic assessment with neurologist<br>• Knowledge to make informed occupational, financial and social decisions<br>• Access to preconception and prenatal genetic testing | • Knowledge that not at increased risk of early-onset Alzheimer's Disease<br>• No increased risk of transmission to next generation |

References: **1.** EviQ. (2020). BRCA1 or BRCA2 – risk management (female) ID: 3814 v.2. Retrieved from https://www.eviq.org.au/cancer-genetics/adult/risk-management/3814-brca1-or-brca2-risk-management-female#cancer-tumour-risk-management-guidelines, **2.** EviQ. (2021). APC (Familial adenomatous polyposis) – risk management ID: 178 v.8. Retrieved from https://www.eviq.org.au/cancer-genetics/adult/risk-management/178-apc-familial-adenomatous-polyposis-risk-ma#cancer-tumour-risk-management-guidelines

#### *1.2.2.2 Predictive Testing for Family Members*

Diagnostic testing is the first step in providing family members the option to undergo predictive testing (Figure 1). For family members, genomic information can help to reduce disease incidence and burden, particularly for people who undergo testing for a known familial pathogenic variant (predictive testing).(3, 19) Predictive testing is a targeted test that involves direct analysis of the pathogenic variant that was identified in the affected person. If no pathogenic variant is identified in the affected person, predictive testing is not possible for relatives. A genetic risk assessment for relatives then reverts to interpretation of the family history, alongside other personal or medical risk factors.

Results from predictive testing for a familial pathogenic variant allow individuals to engage in health behaviours appropriate to their level of risk. With many adult-onset conditions following an autosomal dominant inheritance pattern (meaning each first-degree relative is at 50% risk), it can be pertinent for relatives to make an informed decision about undergoing predictive testing. Unaffected family members who are positive for the pathogenic variant can be offered strategies to reduce or manage their increased risk, while those with negative test results can follow population screening guidelines.(19) Many genetic diseases, such as cancer predisposition syndromes, develop later in life. This gives individuals at increased risk an opportunity to take risk-reducing measures, such as disease screening, surgery, or risk-reducing medication.(20, 21) For example, women with a *BRCA1* or *BRCA2* pathogenic variant, who have an increased lifetime risk of breast and ovarian cancer, may choose to enrol in an established risk management program or participate in clinical trials.(16) In Australia and New Zealand, yearly breast magnetic resonance imaging improves detection of breast cancer at an earlier stage compared with population screening, which stipulates 2-yearly mammograms from 50 years of age.(22) Others may elect to undergo risk-reducing mastectomy and bilateral salpingo-oophorectomy to reduce their risk of breast and ovarian cancer, respectively.(23) High-risk screening programs and risk-reducing surgery are generally indicated for people who have a documented pathogenic variant identified through diagnostic or predictive testing. In the case where relatives cannot access predictive testing because their affected family member did not have diagnostic testing, determining whether their level of risk warrants high-risk measures can be difficult.(24)

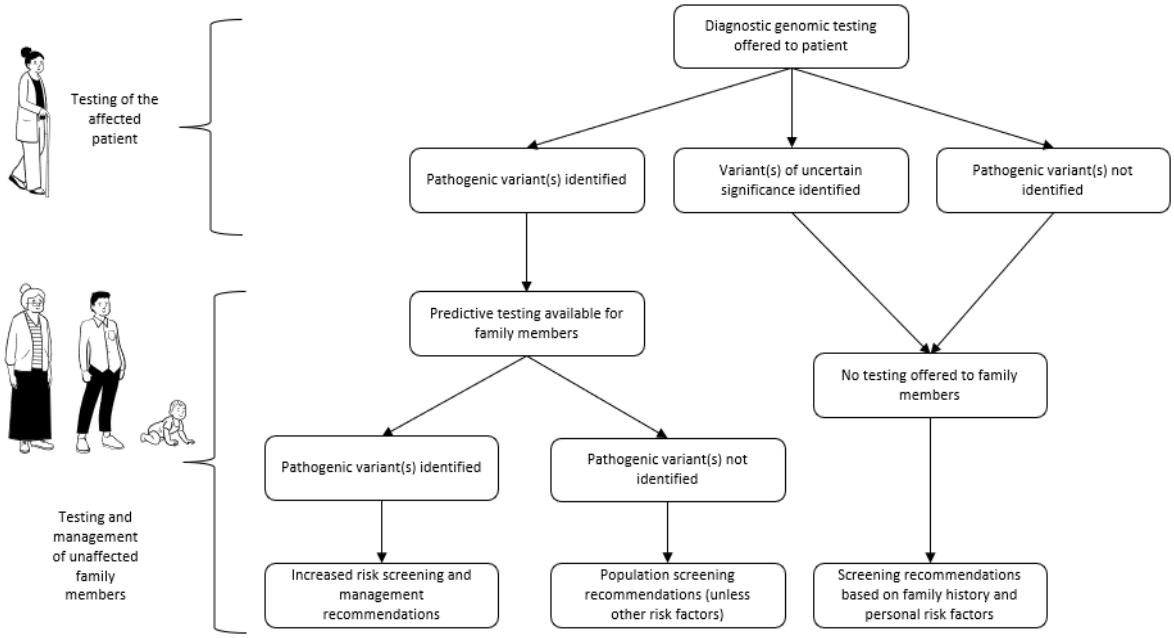


Figure 1. A simplified diagram of the process of offering diagnostic and predictive testing to affected individuals and unaffected family members.

#### *1.2.2.3 Reproductive Benefits*

Predictive testing can also provide reproductive options to people with a pathogenic variant. Some may wish to avoid the risk of a future child inheriting their pathogenic variant.(14) Modern reproductive technologies, including in vitro fertilisation with preimplantation genetic diagnosis (i.e., testing the embryo for a pathogenic variant before implantation) and prenatal diagnosis (e.g., invasive prenatal testing using chorionic villus sampling or amniocentesis) give people the opportunity to prevent the transmission of a pathogenic variant or to terminate an affected pregnancy.(25) In most cases, reproductive options are available only for people who have a documented pathogenic variant in themselves or their relatives. Where reproductive technologies cannot be used, couples who wish to have a child must conceive knowing there is up to a 50% chance that their child will be affected with the same genetic condition, use a donor egg or sperm, or choose to adopt.

#### *1.2.2.4 Personal Benefits*

Much of the literature about clinical benefit focuses on medical or reproductive utility; however, there is growing recognition of the personal or psychological benefits of genomic information. This is sometimes referred to as 'personal utility'.(26) Even in situations where no medical action can be taken to reduce their risk, people report numerous benefits of genomic testing.(14) For individuals and families, genomic testing can improve their knowledge of their

condition, help them cope with health risks, mentally prepare for the future, satisfy altruistic motivations to contribute to research, and provide knowledge for their family members.(27)

### 1.2.3 Challenges of Genomic Information

#### *1.2.3.1 Challenges for the Clinical Setting*

Alongside the clinical and personal benefits of genomic testing is the potential to increase uncertainty or anxiety for some individuals and families. Genetic variants that have an unclear association with disease development (e.g., a variant of uncertain significance; VUS) or are unrelated to the primary purpose of testing (e.g., incidental findings) are challenging for health professionals to interpret, manage, and communicate to individuals and families.(28) In most clinical settings, individuals are informed of the possibility of uncertainty as part of their pre-test consent discussion.(29) Nevertheless, the discovery of a VUS or incidental finding raises ethical questions for health professionals regarding the delivery of uncertain, unsought, and potentially unwanted information.(30) Concerns among health professionals include uncertainties related to the clinical interpretation of the variant and required follow-up, procedures for the return of results, the potential to cause psychological harm, and implications for family members.(31) Internationally, pathogenic variants in genes unrelated to the clinical presentation are deliberately sought,(32, 33) and this raises several additional ethical and practical issues. However, this type of testing is not routinely performed in Australia.(34)

Despite the promise of genomic testing, many individuals and their families will not obtain a genetic diagnosis. Issues related to the accuracy of sequencing technology (e.g., inability to capture all regions of the genome) and variant interpretation mean that many people experience uncertainty and disappointment where a genetic diagnosis is not provided by genomic testing.(7) One option to preserve hope for a genetic diagnosis is to use DNA storage (also referred to as DNA banking). DNA storage is different from biobanking, which stores biological and genetic samples for research.(35) DNA storage involves extracting and storing an individual's genetic material without an accompanying test request. Individuals who receive uninformative or uncertain results may wish to store DNA so their sample can be re-tested in the future. When people die (expectedly or unexpectedly), DNA can be stored by a medical professional (including a coroner) to preserve the opportunity for families to explore whether a genetic cause was responsible for the family member's death.(36)

#### *1.2.3.2 Challenges for the Health System*

The evolution of clinical genomics has resulted in several health system challenges that are yet to be resolved. In Australia and elsewhere, the ability to generate data is outpacing data storage systems.(37) Novel solutions to data storage are an Australian strategic priority as the demand for genomic testing continues to increase.(38) The privacy of a person's genomic information relies on robust data protection. In Australia, the 'patchwork' approach to legal protections between states and territories complicates national efforts towards integration of clinical genomics into routine healthcare.(39 p583) Consumers, genetic health professionals, and professional organisations are concerned about the risk of confidential data being used to discriminate against employment or insurance prospects.(40-42) As an example, the recently introduced MyHealth Record (an Australian Government online portal for health professionals that contains an individual's health information) has the capacity to store genomic test reports. MyHealth Record has fuelled current debate about data and privacy breaches, government access to genomic information, and the potential for misinterpretation of results by non-specialists.(43)

Issues pertaining to equitable access of genomic testing are partly related to fragmented funding systems across Australia.(39) Genomic testing can be accessed through the federal public health funding (Medicare), but approved items represent a fraction of available testing.(44) Outside of Medicare, health professionals with access to other public funds can order genomic testing. To access this public funding, individuals must obtain a tertiary referral to a clinical genetics service. Some individuals access testing through translational research projects, which offer research genomic testing in a clinical setting.(45) In other cases, testing is self-funded, particularly if the criteria for testing through Medicare, public funding, or research are not met. Facilitating self-funded testing is usually at the discretion of the ordering health professional, although individuals can access direct-to-consumer testing.(46) In rare cases, private health funds may reimburse genomic testing.

## 1.3 GENETIC COUNSELLING IN AUSTRALASIA

### 1.3.1 The Genetic Counselling Profession

In Australia and New Zealand (termed here 'Australasia'), genetic counselling is an allied health profession. Genetic counsellors have specialised training in genetics, genomics, communication, and counselling.(47) The profession defines genetic counselling as "a communication process, which aims to help individuals, couples and families understand and

adapt to the medical, psychological, familial, and reproductive implications of the genetic contribution to specific health conditions."(48) The entry-level qualification for employment as a genetic counsellor is a 2-year Master degree, after which the 'Associate Genetic Counsellor' completes a portfolio of work over at least 3 years to become a 'Certified Genetic Counsellor'.(47) Genetic counselling training, certification, and registration in Australia and New Zealand (and in some adjacent Oceanic nations) is overseen by the Human Genetics Society of Australasia.(47)

Genetic counsellors have traditionally worked in public hospital settings alongside clinical geneticists who are medical doctors sub-specialised in clinical genetics.(49) Genetic counsellors interact with a variety of populations, including prenatal, paediatric, adult, and those with palliative care needs.(2) Clinical presentations vary widely and may involve cancer, intellectual disability, and cardiac, neurological, renal, and other conditions. Some genetic counsellors specialise in a clinical area, while others are generalists.(50) Clinical genetic counsellors perform a variety of tasks, including consulting with individuals, couples, and families, eliciting their goals and expectations of genomic information, obtaining relevant personal and familial medical information, making assessments about genetic risk, communicating information in an accessible way, discussing options for genomic testing, obtaining informed consent for testing, interpreting and conveying test results, providing psychosocial support, and more.(51) In addition, genetic counsellors work in non-patient-facing roles, including research, academia, industry, and laboratories.(52)

### 1.3.2 Genetic Counselling Workforce and Delivery of Services

Although the Australian Government has prioritised the integration of genomics into routine healthcare, the 480 genetic counsellors in Australia cannot keep up with demand and this is predicted to worsen with time.(38, 52, 53) As requests for genetic counselling and genomic testing increase, wait times to be seen through public services are growing. For example, in New South Wales, Australia, individuals with a non-urgent referral to a clinical genetic service may wait up to two years to be seen.(50) Appropriately skilled personnel and resourced services are required to deliver the benefits of genomics, but the increase in demand places pressure on existing services.(54).

As genomics evolves into a routine aspect of clinical care, other 'non-genetic' health professionals will need to provide genetic counselling or some aspects thereof. Many genomic tests are moving into routine clinical use, including reproductive carrier screening, non-invasive prenatal testing, and cancer gene testing. The role of a non-genetic health

professional in providing genetic counselling can vary depending on their patient's needs, their resourcing and capability, and links to clinical genetic services. Some may need to assess eligibility for genomic testing, provide information about inheritance, explain genomic test results, or make referrals to clinical genetics services. Australasian and international genomics organisations recommend that genetic counselling accompany genomic testing, regardless of whom it is delivered by.(19, 55, 56) The challenge, therefore, for non-genetic health professionals is to maintain an adequate level of knowledge and skill to integrate aspects of genetic counselling into their practice, in addition to their primary clinical role.(57)

To improve access to genetic counselling and genomic testing, alternative models of care are emerging.(54) Genetic services have traditionally operated a face-to-face, two-appointment model; one appointment for pre-test genetic counselling and one for post-test genetic counselling.(50) Alternatives include genetic counselling via telehealth or offering just one post-test appointment. Some services have embedded a genetic counsellor into a non-genetic service or upskilled non-genetic health professionals to provide genetic counselling (sometimes called 'mainstreaming').(57, 58)

In Australia, mainstreaming is common in the oncology setting. Mainstreaming shifts pre-test counselling and test facilitation from the genomics service to the oncology service.(59) In some settings, mainstreaming is set up as a formal model of care, whereas in others it is ad hoc.(60, 61) Generally, the oncology health professional's role is to discuss genomic testing, obtain informed consent, facilitate testing, deliver the result and, if required, refer the patient to a clinical genetic service. Typically, people are referred if a pathogenic variant or VUS is detected, or if there is a suspicious family history with uninformative testing.(57) Some benefits of mainstreaming include increasing patient and family access to genomic testing, streamlining access to targeted therapies, reducing demand on clinical genetic services, and reducing burden on patients to attend a separate genetic appointment.(62)

Despite the benefits, mainstreaming has its challenges. Oncologists continue to report low confidence in their genomic knowledge, and testing rates can be suboptimal.(63, 64) As a result, many individuals and their families miss the opportunity for genomic testing and to benefit from genomic information. Furthermore, mainstreaming models apply to people with specific cancer types (e.g., non-mucinous ovarian cancer), but individuals with many other cancers can benefit from genomic testing.(65) In other clinical areas, including palliative care and in treating those who have a non-malignant disease, alternative service delivery models are even less well developed.(66) Increasing access to genomics for people with non-

mainstreamed cancers and non-malignant disease requires changes at all levels of the health system.(67)

## 1.4 PALLIATIVE CARE PROVISION IN AUSTRALASIA

### 1.4.1 Defining Palliative Care

Palliative care is a philosophy of care, defined as

> an approach that improves the quality of life of patients (adults and children) and their families who are facing the problems associated with life-limiting illness, through the prevention and relief of suffering by means of early identification and correct assessment and treatment of pain and other problems, whether physical, psychosocial or spiritual.(68 p130)

Palliative care is a relatively young field of specialisation. Modern palliative care is often attributed to the work of Dame Cicely Saunders who, in 1967, founded the first English hospice that combined evidence-based care with compassion and respect for persons. In addition to advocating for adequate pain relief for dying people, she introduced the concept of 'total pain', which encompassed emotional, social, and spiritual dimensions of distress.(69)

The role of the specialist palliative care health professional is varied, but some common responsibilities include assessing a person and family's needs, developing an individualised care plan, providing support to carers, delivering specialised palliative care interventions (e.g., pain or dyspnoea management), supporting transition between health services, assessing and supporting bereavement, and advocating for palliative care provision.(70) Palliative care specialists most commonly care for people with cancer, because of the historic association between palliative care and cancer services.(71) However, there is increasing attention on improving access to specialist palliative care for people with non-malignant disease (e.g., progressive neurological conditions, end-stage renal failure, and cardiomyopathy) and for children with life-limiting illnesses.(72-77)

### 1.4.2 Vulnerability in Palliative Care

The term 'vulnerable' is sometimes used to describe people who are nearing end of life.(78) Providing a conceptual definition of vulnerability in palliative care supports here a commitment to person-centred and strengths-based approaches to care. The use of the term vulnerability in this thesis aligns with a feminist ethics approach of 'inherent vulnerability', which is a shared human condition experienced in various ways and stages of our lives.(79) Inherent vulnerability complements the tenets of the Reciprocal engagement model (REM) of

genetic counselling, one of which is 'patients are resilient' (further discussed in Chapter 7).(80) Inherent vulnerability also links closely to ideas about relational autonomy, which recognises humans as fundamentally social beings who support each other in making decisions about their health.(81)

Inherent vulnerability in palliative care does not reject traditional ideas of autonomy (positing that people make independent, rational decisions without external influence), but rather encourages health professionals to identify and attend to individual and relational sources of pain, discomfort, and distress, while supporting peoples' well-being in a relational way.(82, 83) The risk of contrasting vulnerability and autonomy is that people may be assumed incapable of making autonomous decisions if they are perceived as vulnerable.(79) Deficit-based framing portrays a vulnerable person as helpless, weak, and reliant on others; however, these ideas do not reflect the philosophy of palliative care and are not consistent with the REM.(80, 84)

Acknowledging our shared inherent vulnerability helps to avoid detrimental actions that take advantage of or worsen a person's vulnerability, sometimes termed 'pathogenic vulnerability'.(79) These actions, arising from prejudice, assumptions, or paternalism, result in an individual's oppression and discrimination. Differentiating between inherent vulnerability (a shared human state) and pathogenic vulnerability (harmful actions that worsen a vulnerable person's situation) helps health professionals to articulate their efforts to support people receiving palliative care."(79)

### 1.4.3 Palliative Care in Australia and New Zealand

There are many similarities in palliative care provision between Australia and New Zealand. For example, the primary organisation representing palliative medicine specialists (Australian and New Zealand Society for Palliative Medicine (ANZSPM) spans both countries. ANZSPM outlines shared goals, such as equitable access to individualised and culturally safe care. New Zealand delivers palliative care in the hospice setting more commonly but, in both countries, palliative care is delivered across a variety of settings including hospital and community settings.(85, 86) In addition, specialist palliative care teams are multidisciplinary. Doctors and nurses are a core part of the multidisciplinary team, but people with palliative care needs and families are also cared for by physiotherapists, occupational therapists, social workers, psychologists, and pastoral carers.(84)

### 1.4.4 Managing the Demand for Palliative Care

Similar to clinical genetic services, demand for specialist palliative care currently outweighs capacity.(87) Palliative care delivery in Australia is organised as a three-tiered model, described in Palliative Care Australia's 2018 Palliative Care Service Development Guidelines.(86) The Guideline describes the involvement of palliative care clinical expertise at each tier, but not the training required by the health professionals providing care because they will have a variety of educational backgrounds. Access to the three different tiers of palliative care ought to be based on clinical need, though there are numerous barriers and enablers to appropriate access.(88) The first tier, Level 1 palliative care, is delivered by the treating health professional (e.g., general practitioner, oncologist, or neurologist) and is suitable for people who have an uncomplicated life-limiting illness. Support from specialised palliative care health professionals is usually not required. Level 2 specialist palliative care is also delivered by the treating health professional but with consultative support from a palliative care specialist, such as a palliative medicine physician or specialist nurse. Level 2 is for people who are generally stable but with more complex or fluctuating symptoms related to dying. Level 3 specialist palliative care is delivered by palliative care specialists for people with complicated and distressing symptoms of dying requiring specialist management (e.g., chronic or fluctuating refractory pain, terminal restlessness and agitation, dyspnoea, nausea, vomiting or anorexia, or other physical or psychological issues).(89)

## 1.5 RELEVANCE OF GENOMICS TO PALLIATIVE CARE

### 1.5.1 Relevance to People with Palliative Care Needs

As clinical genetic and palliative care services continue to evolve, so too will the importance of genetic counselling and genomic testing for people with palliative care needs and their families. Many life-limiting conditions, both cancer and non-cancer, are inherited.(65) Some cancers linked to inherited pathogenic variants exhibit tumour characteristics that are associated with poor outcomes. For example, people with breast or prostate cancer and a pathogenic variant in *BRCA1* or *BRCA2* have increased risk of metastasis and a lower survival rate than those without a pathogenic variant.(90, 91) As a result, many of these people will end up as recipients of palliative care.(92)

The extent to which a person receiving palliative care has had the opportunity to access genetic counselling can vary.(93) Some will have had their treating clinician organise genetic counselling and genomic testing before receiving palliative care and, in this case, specialist

palliative care health professionals are unlikely to need to take direct action related to genomics. Some may have been referred to a clinical genetic service or be in the process of genomic testing when they are referred to palliative care. In this case, specialist palliative care health professionals may need to provide information or psychological support related to genomics. Others people will not have been identified as eligible for genomic testing before they receive palliative care.(24) In this situation, specialist palliative care health professionals must play a more active role in identifying people eligible for genomic testing, broaching genomic discussions, collaborating with genetic health professionals, facilitating genomic testing or DNA storage, delivering results, or referring individuals to clinical genetic services.(94)

### 1.5.2 Relevance to the Families of People with Palliative Care Needs

Although not direct recipients of clinical care in the palliative care context, family members benefit from genomic information obtained from their dying relative. Family members at risk of developing a disease because of an inherited pathogenic variant have been termed 'patients in waiting'.(95 p33) Family members may be unaware of their risk because of misunderstandings related to inheritance patterns, the heritability of their relative's disease, or the clinical benefits of genomic testing.(93, 96) Palliative care is often a time when families gather together and share information, which provides an opportunity to collaborate with families to obtain a comprehensive family history, assess the need for genomic testing, discuss the benefits and limitations, and convey the importance of collecting a DNA sample from the affected person to assess the future disease risk of family members.(96, 97)

Both palliative care and genetic counselling professions are committed to family-centred care, although it is not clear how this is operationalised in the genomics context. In Australia, the care of family is intertwined throughout palliative care standards.(70) Palliative Care Australia stipulates "…family and carers work in partnership with the team to communicate, plan, set goals of care and support informed decisions about the care plan" and "the person's family and carers needs are assessed and directly inform provision of appropriate support and guidance about their role."(70 p8) Genetic counselling competency standards set out the expectation that family members' genetic risks are assessed and managed.(51) Genetic counselling models, such as the Reciprocal-engagement model, also highlight the interconnected nature of individuals within their social and familial context.(80)

The practical application of family-centred care within the palliative–genomic setting can raise several ethical, legal, and social complexities, particularly regarding individual autonomy.

For example, a family member may request that a blood sample be obtained from their dying relative for genomic testing, but the dying person may be unable to provide informed consent. Managing these complex scenarios can be challenging for health professionals.(98) The clinical responsibility towards family members is not well defined, despite the importance of family-centred care to the genetic counselling and palliative care professions.(99, 100) Identifying and addressing the support needs of health professionals to allow them to navigate genomic discussions with families will help to improve access to genomic information for those who can benefit from it.(101, 102)

### 1.5.3 The Challenge of Integrating Genomics into the Care of People with Palliative Care Needs and Their Families

For many people with palliative care needs and their families, access to genetic counselling is suboptimal. Palliative care health professionals report that genomics should be addressed with individuals and families before they receive palliative care.(103) However, for several reasons, genomics may not have been raised before this. Non-genetic health professionals (e.g., oncologists) report barriers such as low genomic knowledge and confidence, and limited time with people in the clinic.(63, 64) In a single-centre study, 42% of palliative oncology patients met the criteria for genetic counselling or testing but had not been referred to a clinical genetic service.(93) In the same centre, one-fifth of patients reported a family history of cancer in a research interview, yet a chart review found none of these individual's family histories were documented.(92) In other cases, people refuse conventional treatment or are directly referred to palliative care after a diagnosis because of a poor prognosis.(94) Others may defer or initially decline a discussion about genomics but may wish to address the topic later. Regardless of the reason, missing the opportunity to broach a genomics discussion with people who have palliative care needs reduces the flow-on benefits of genomics to individuals and their families.(24, 104) These challenges represent several potential barriers that prevent the integration of genomics into palliative care in Australasia.

## 1.6 IMPETUS FOR THIS THESIS: GENOMIC INFORMATION FOR FAMILIES OF THE TERMINALLY ILL PROJECT

This chapter has outlined the key concepts that challenge the integration of genomics into the care of people with palliative care needs and their families. However, much remains unknown about the barriers and facilitators in the Australasian setting. Supporting health professionals to integrate genomics into palliative care requires an understanding of the

barriers and facilitators from which an evidence-based intervention can be designed. This thesis describes the Genomic Information for Families of the Terminally ill project (hereafter, the GIFT Project), an exploratory sequential mixed methods project designed to address this gap.

### 1.6.1 GIFT Project Aim

The primary aim of the GIFT Project is to develop an Australasian evidence base of the barriers and facilitators that affect the integration of genomics into care of people with palliative care needs and their families.

### 1.6.2 Research Questions

The GIFT Project sought to answer three primary research questions.

1. What are the barriers and facilitators for genetic and palliative care health professionals towards integrating genomics into the care of people with palliative care needs? (Qualitative question)
2. How do the identified barriers and facilitators compare between genetic and palliative care health professionals? (Quantitative question)
3. What is required to support the integration of genomics into the care of people with palliative care needs and their families? (Mixed methods question)

## 1.7 OVERVIEW OF THESIS CHAPTERS

To assist with navigation of this thesis, an overview of the chapters is provided in Figure 2.

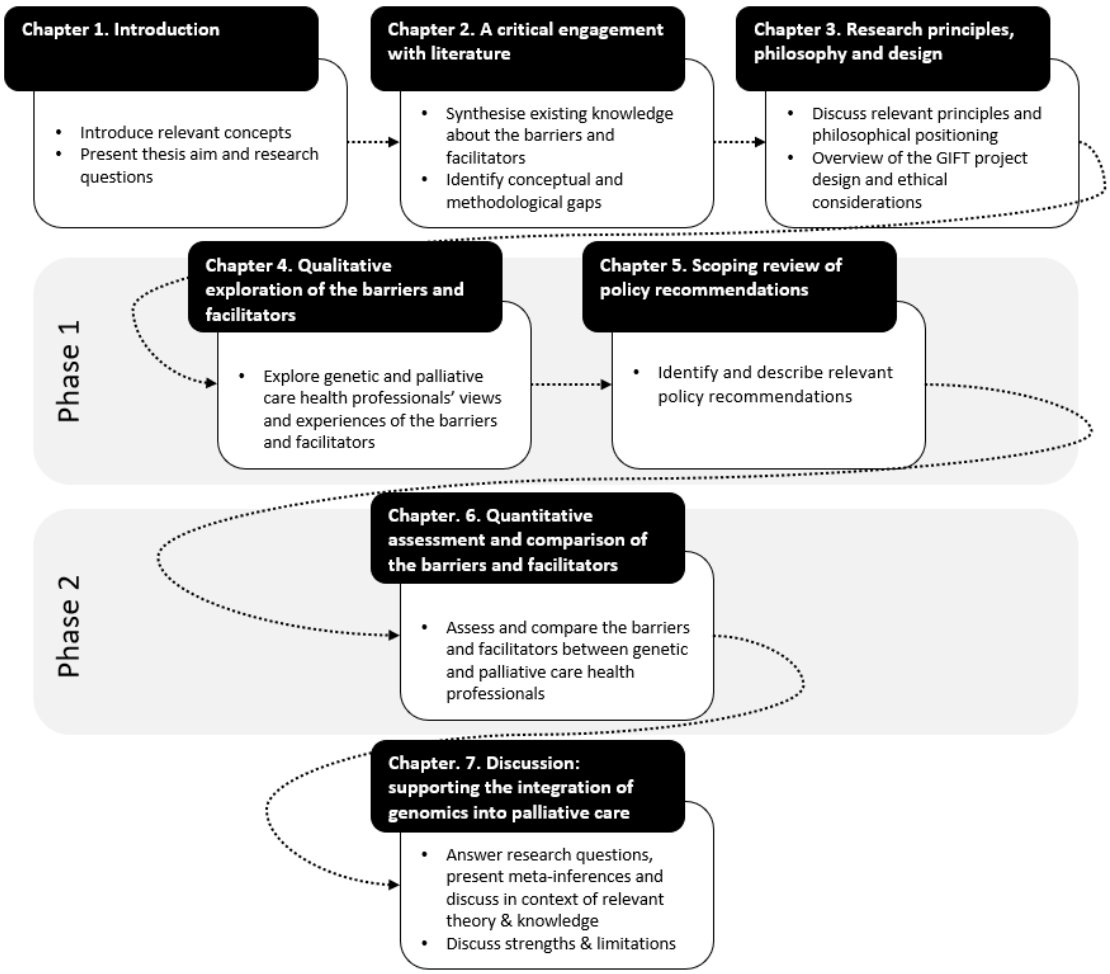


Figure 2. Thesis chapter navigation figure.

## 1.8 CHAPTER SUMMARY

Chapter 1 has described the evolution of clinical genomics, including the advances in genomic testing, and the benefits and challenges of delivering clinical genomics to individuals and families. I provided an overview of genetic counselling and palliative care provision in Australasia and then discussed the relevance of genomics to people with palliative care needs and their families. The challenges of integrating genomics into palliative care underpin the impetus and aim the GIFT Project. In the next chapter, I present a systematic review of the barriers to and facilitators of integrating genomics into clinical practice and a critical engagement of the palliative–genomic literature.

# 2 A Critical Engagement with the Literature

## 2.1 PREAMBLE

In Chapter 2, I build on the concepts introduced in the previous chapter by critically engaging with the relevant literature and identifying the conceptual and methodological gaps for the Genomic Information for Families of the Terminally ill (GIFT) Project to address. I present a peer-reviewed, published, systematic review that synthesises the barriers and facilitators that affect the ability of non-genetic health professionals to integrate genomics into clinical care. Having found limited palliative–genomic literature on an initial search, I broadened the scope of this systematic review to any clinical specialty delivered in a secondary or tertiary setting. Literature from these adjacent fields, such as oncology and cardiology, provided potential barriers and facilitators for the GIFT Project to explore. At the conclusion of the manuscript (section 2.10 onwards), I present an extended discussion that focuses on the barriers and facilitators in the palliative care context and that critically appraises the literature from the palliative–genomic field. The extended discussion was written for this thesis and is not part of the published manuscript. Files related to the planning and conduct of this review are available in appendix B.

## 2.2 INCORPORATING BEHAVIOUR CHANGE THEORY INTO THE SYSTEMATIC REVIEW

In this thesis, health professionals are recognised as key actors in the delivery of clinical care. The Theoretical domains framework (TDF) is a theory of the factors influencing a health professional's behaviour, such as knowledge, skill, and belief about consequences.(105) Each factor in the TDF maps to one of the three components in the Behaviour change wheel (BCW): capability, opportunity, or motivation.(106) The BCW was developed through the critical synthesis of 19 different behaviour change theories and maps different types of interventions to the barriers and facilitators affecting health professionals' behaviour, such as guidelines, regulations, and incentives.(106)

I selected the TDF as an evidence-based framework because of its ability to translate across clinical settings. This was an important feature given that the systematic review would synthesise barriers and facilitators from several specialties. Using the TDF also meant the findings from this review could be translated into interventions to target specific barriers.

## 2.3 MANUSCRIPT 1: A SYSTEMATIC REVIEW OF THE BARRIERS AND FACILITATORS

This systematic review was published in *Genetics in Medicine* (Scimago rating Q1 in the 'genetics' category; 2021 impact factor 8.864).(107) As of 18 March 2023, this study has been cited 40 times. The manuscript in this chapter has undergone minor edits, including changing US spelling to Australian spelling, and 'genetic' to 'genomic' for congruency across the thesis. The table, figure, and page numbers have been changed for coherence. As per the publisher copyright requirements, this is the accepted (not published) version of the manuscript and formal permission was not required for reproduction in a thesis.

Reference: White S, Jacobs C, Phillips J. Mainstreaming genetics and genomics: a systematic review of the barriers and facilitators for nurses and physicians in secondary and tertiary care. Genet Med. 2020;22(7):1149-55.

## 2.4 ABSTRACT

*Purpose:* Genetic and genomic health information increasingly informs routine clinical care and treatment. This systematic review aimed to identify the barriers and facilitators to integrating genetics and genomics into nurses' and physicians' usual practice (mainstreaming).

*Methods:* A search of MEDLINE, EMBASE, CINAHL, and PsycINFO generated 7873 articles, of which 48 were included. Using narrative synthesis, barriers and facilitators were mapped to the Theoretical domains framework (TDF).

*Results:* Barriers were limitations to genetics knowledge and skill, low confidence initiating genomics discussions, lack of resources and guidelines, and concerns about discrimination and psychological harm. Facilitators were positive attitudes toward genomics, willingness to participate in discussions upon patient initiation, and intention to engage in genomics education.

*Conclusion:* Nurses and physicians are largely underprepared to integrate genetic and genomic health information into routine clinical care. Ethical, legal, and psychological concerns

surrounding genomic information can lead to avoidance of genomics discussions. The knowledge–practice gap could limit patients' and families' access to vital genomic information. Building the capacity of the current and next generation of nurses and physicians to integrate genetics and genomics into usual clinical practice is essential if opportunities afforded by precision medicine are to be fully realised.

## 2.5 INTRODUCTION

During the past two decades, the field of human genetics has undergone significant change. The sequencing of the human genome has fuelled understanding of the relationship between genomic variation and human health.(4) Demand is such that clinical nurses and physicians working in a variety of clinical disciplines are now required to integrate genomics into routine care. For example, people with ovarian cancer and a DNA-repair deficiency may be exquisitely responsive to poly-ADP ribose polymerase inhibitors,(12) and cardiologists may consider implantable cardioverter defibrillators for those at risk of sudden cardiac death.(108) Reductions in the cost of genomic testing,(5) and greater public access to and awareness of genomic information,(109) means more people seek genomic information than ever before. Collectively, these changes have prompted the acceleration of genomic information as a critical element of care for many patient populations.

Considering the changing landscape of genetic and genomic (herein referred to as 'genomic' only) opportunities, care pathways for patients to access genomic information need to adapt. Traditionally, access involved referral of patients to tertiary centres for genetic counselling. However, the demands on genetics services are outweighing workforce capacity,(110) with policy makers calling for alternative genomic models of care.(111, 112) One such model is 'mainstreaming', which involves non-genetics nurses and physicians identifying at-risk individuals and initiating genomics discussions,(113) by integrating genomics into practice. Examples include taking a family history, assessing the chance of a genetic condition, organising genomic testing or delivering a genomic test result to a patient. The benefits of identifying individuals with a genetic condition through mainstreaming are three-fold: targeted treatments may be available; a genetic diagnosis may alert the treating specialist to other possible health problems the individual could face; and the individual's relatives can be offered predictive testing (targeted testing for the genetic condition identified in their relative). Predictive testing guides the relative's need for health screening or risk management.

Despite the benefits of genomic health information, translation of research to clinical practice is slow, highlighting the complex and interconnected barriers and facilitators within healthcare pathways.(105) Identifying the underlying barriers and facilitators to nurses and physicians integrating genomics into their practice will lay the groundwork for the development of an evidence-based intervention to encourage behaviour change.(114) The aim of this review was to identify the barriers and facilitators for nurses and physicians working in secondary and tertiary care to integrate genetics and genomics into their usual practice. The secondary aim was to explore the similarities and differences between the specialties and disciplines.

## 2.6 MATERIALS AND METHODS

This systematic review was registered with PROSPERO (CRD42019134752) and conducted in accordance with the Preferred Reporting Items for Systematic reviews and Meta-Analyses (PRISMA) statement (appendix A1).(115) MEDLINE, Cochrane Reviews Database of Systematic Reviews, PROSPERO and the Joanna Briggs Institute Systematic Reviews database were searched to ensure this systematic review would not duplicate existing work.

### 2.6.1 Search Strategy

The search strategy was developed in consultation with an information services librarian. MEDLINE, EMBASE, CINAHL and PsycINFO were searched on 30th August 2019 with no restrictions (appendix A2). Further articles were elicited by backwards searching reference lists of included articles and relevant literature reviews, forwards searching articles using the Web of Science database, and reviewing first author profiles of included articles on ResearchGate (www.researchgate.net).

### 2.6.2 Inclusion and Exclusion Criteria

Inclusion and exclusion criteria were developed using the PICOS framework (appendix A3).(115) Articles were included if they were reported after the first initial human genome sequence was published in February 2001,(4) published in English in a peer-reviewed journal, and reported empirical data on the barriers or facilitators nurses and/or physician encountered when providing genomic information to adults cared for in a secondary or tertiary healthcare setting. Quantitative, qualitative and mixed-method studies were included to incorporate data from varied perspectives. The Royal Australasian College of Physicians Advanced Training Programs were used as a specialty guide to include nurses and doctors who

were most likely to work in secondary and tertiary care.(116) Articles were excluded if they reported on direct-to-consumer genomic testing, pharmacogenomic testing or reproductive carrier testing, or the nurse or physician worked in a primary care, paediatric, prenatal, research or clinical genetics setting. Primary care nurses and doctors were excluded due to the breadth of articles in this area and the existence of previous systematic reviews evaluating genomic interventions in the primary care setting.(117)

### 2.6.3 Screening and Extraction

Following de-duplication, one reviewer (S.W.) screened all articles against the inclusion and exclusion criteria by title and abstract and then by full text (Figure 3). A 20% sample was allocated to a second reviewer (C.J.) at both stages and interrater concordance was calculated using a prevalence-adjusted, bias-adjusted kappa statistic (≥0.7).(118) Disagreements were resolved through discussion. Up to three attempts were made to email authors of articles with missing or ambiguous information.

Data items were predetermined using the Joanna Briggs data extraction instrument (appendix A4).(119) Extraction was performed using QSR International's NVivo Version 12 and exported to an Excel spreadsheet.

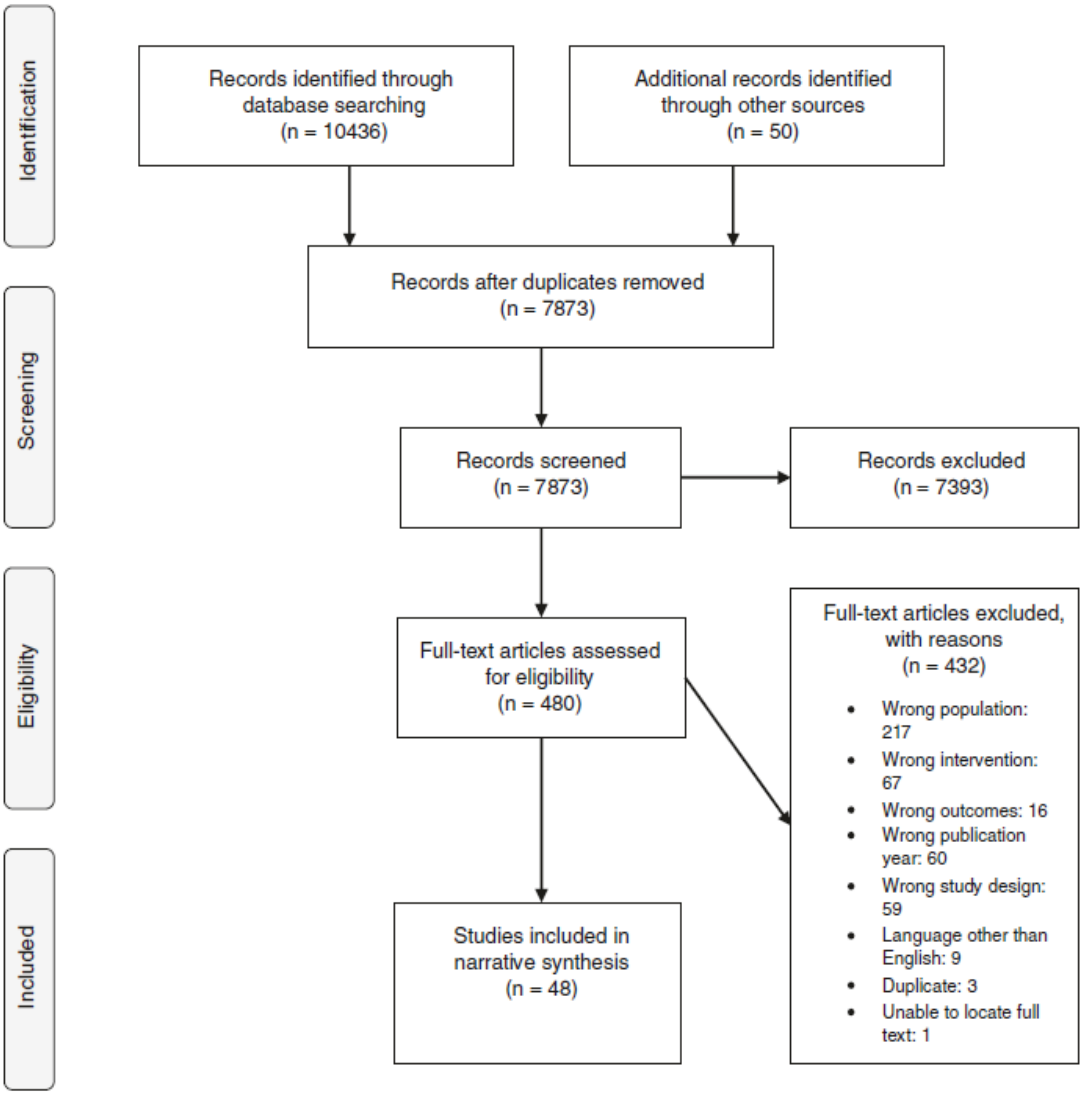


Figure 3. PRISMA flow diagram showing number and reasons for record exclusion. From 10,486 records, 48 were deemed eligible.

### 2.6.4 Risk of Bias

Individual risk of bias assessments were conducted using the QualSyst tool, which was selected because it includes separate qualitative and quantitative assessment matrices (appendix A5).(120) With the aim of including a range of clinical disciplines, articles with a high risk of bias were not excluded. To assess for outcome reporting bias, published study protocols were searched using the World Health Organization's International Clinical Trial's registry platform. No study protocols were identified in the initial systematic search, therefore publication bias could not be assessed.

### 2.6.5 Data Synthesis

Narrative synthesis was performed using the Theoretical domains framework (TDF) to map the barriers and facilitators to higher behavioural domains and components.(121) The TDF is a validated, comprehensive framework describing factors affecting health professional behaviour and can be adapted to diverse clinical contexts.(105) Data generated from studies with varied epistemologies can be challenging to synthesise because they may not be easily comparable. However, narrative synthesis was chosen because of its flexibility to synthesise qualitative and quantitative studies, and the use of the TDF helped to standardise and integrate the extracted data.(122)

Narrative synthesis consists of four inter-related, non-linear steps (121). The first step was the development of a preliminary theory prior to the database search. A visual map of the potential barriers and facilitators was developed based on our initial readings, which helped to refine the review question, search strategy, and inclusion criteria. The second step was a preliminary synthesis of data extracted from the included studies. Extracted data items were grouped into themes. If the data item did not adequately correspond to an existing theme, a new theme was created. Each theme was mapped to a TDF domain and the frequency of each domain was calculated as a percentage of the total number of articles. The TDF domains sit within the Behaviour change wheel's Capability, opportunity, motivation behaviour system (COM-B),(106) and these components were used to organise and describe the results. This process is represented in Figure 4. The third step was to explore relationships within and between studies, including the differences between nurses and physicians and between clinical disciplines, which were described narratively. The last step was assessing the robustness of the synthesis. We aimed to provide transparency of the synthesis by including risk of bias scores alongside individual studies (appendix A6) and identifying strengths and limitations of the overall review (described in section 2.8.2)."

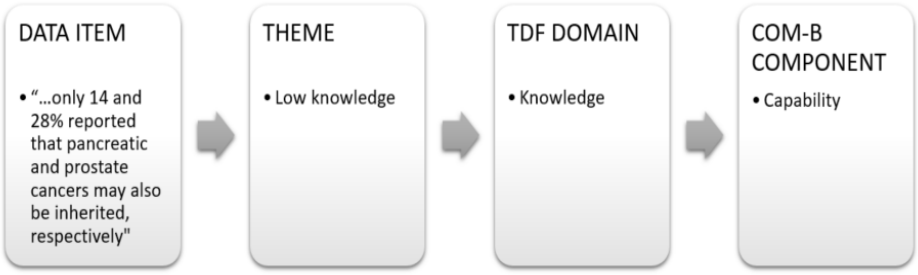


Figure 4. An example of the data synthesis process, from extracted data to COM-B component.
NB. TDF: Theoretical domains framework, COM-B: Capability, opportunity, motivation behaviour system

## 2.7 RESULTS

### 2.7.1 Study Characteristics

Nearly all of the 48 included articles were from high income countries (see appendix A6 for a summary of included articles; *n*=45, 94%). Half of the articles originated from the United States of America (USA) (*n*=25, 52%) and involved oncology nurses or physicians (*n*=24, 48%). The majority of articles were surveys (*n*=38, 79%), which largely used novel, unvalidated instruments (*n*=37/38, 97%). Three-quarters of the articles only included physicians (*n*=35, 73%), were published after 2011 (*n*=36, 75%), and were assessed as having a low risk of bias (0.67 – 1.0; *n*=35, 73%). There were no significant differences in reported barriers and facilitators between quantitative, qualitative and mixed methods articles.

### 2.7.2 Factors Influencing Integration of Genetic Counselling

Most articles (*n*=40, 83%) reported both barriers and facilitators, while a small number only reported the facilitators (*n*=5, 10%),(123-127) or the barriers (*n*=3, 6%).(128-130) Themes were broadly associated with nurses' and physicians' capability (*n*=44, 92%), opportunity (*n*=39, 81%) and motivation (*n*=38 articles, 79%) to integrate genomics into practice (see appendix A7 for a summary of the identified barriers and facilitators).

### 2.7.3 Capability to Integrate Genomics into Practice

*Knowledge and Skill*: 27 articles (56%) explored nurses' and physicians' knowledge of genomics,(103, 113, 129, 131-154) while 41 articles (85%) reported on their skills.(103, 113, 124-128, 130-138, 140-149, 151-165) While nurses and physicians routinely engaged in discussions about genomics with their patients,(125, 127, 132, 134, 141, 144, 149, 151, 154-158) all demonstrated limited understanding of general genomic concepts, and/or concepts relevant to their specialty.(129, 131-140) Despite knowledge deficits, nurses and physicians did engage in discussions about genomics with their patients.(125, 127, 132, 134, 141, 144, 149, 151, 154-158) In some specialties, family history information was routinely obtained,(124, 132, 133, 135, 136, 141, 142, 149, 151, 154-157) although the extent of the family history was not always adequate.(131, 134, 135, 138, 142, 159) A smaller number of articles reported that physicians did assess genetic risk,(126, 133, 137, 151, 152, 154, 162, 163) however, confidence in family history and individual risk assessment was low.(103, 113, 132, 135, 138, 140, 141, 145-147, 151, 155, 159-161) Four articles reported an inverse relationship between years of clinical practice and level of knowledge.(132, 135, 136, 141)

Oncologists and neurologists were most likely to order genomic testing. There were no reports of nurses or physicians from other specialties ordering testing.[25, 26, 39, 49, 53, 59, 60, 64-66] Most nurses and physicians had low awareness of genomic tests relevant to their area of practice.(103, 131, 133, 141-145) They also had difficulty interpreting a genomic test result.(131, 134, 147-149, 151, 153)

### 2.7.4 Opportunity to Integrate Genomics into Practice

*Environmental context & resources*: 39 articles (81%) explored the impact of environmental context and resources on nurses' and physicians' ability to integrate into practice. Nurses and physicians infrequently referred patients to clinical genetics services,(103, 131, 132, 134-137, 139, 141, 153, 155-157, 162) primarily because of the prohibitive cost of accessing genomic testing,(137, 142, 146-148, 162-167) lack of resources,(135, 137, 140, 141, 146, 155, 163, 164, 167) absence of guidelines,(129-131, 147, 157, 158, 163) and of lack of time to initiate a genomics discussions.(139, 140, 146, 155, 160, 161, 167) Some nurses and physicians had concerns about the privacy of genomic information or the process of informed consent.(113, 140, 145, 146, 154, 166, 168) However, if patients raised questions or concerns about genomics, nurses and physicians did engage in these discussions.(113, 127, 137, 139, 144, 145, 147, 151, 157, 162, 164)

A small number of articles reported nurses and physicians actively avoided or refused to discuss genomics with their patients, where they felt genomics was not relevant to clinical care and there may be potential negative consequences of genomic information.(124, 131, 139, 142, 146) For example, some palliative care clinicians considered their clinical setting as inappropriate to initiate discussions about genomics and were disappointed when this had not been addressed previously.(103, 113) Nurses and physicians reported the value of close working relationships or collaboration with clinical genetics professionals.(113, 131, 132, 135, 137, 140, 151, 158, 161, 164, 167)

### 2.7.5 Motivation to Integrate Genomics into Practice

*Belief about consequences*: In total, 26 articles (54%) explored nurses' and physicians' belief about consequences. Nurses and physicians are cognisant of the potential medical benefit that genomic information can provide for patients,(123, 137, 146, 147, 151, 157, 160, 161, 165, 167, 168) but this was tempered by concerns about the risk of psychological harm, such as inducing feelings of guilt or hopelessness.(113, 128, 139, 140, 145, 146, 148, 151) The potential benefit to relatives was described, including clarifying family members' risks and providing

screening or family planning options.(113, 131, 141, 146, 149, 154, 157) Some nurses and physicians worried about the emotional impact of genomic information on the family.(103, 113, 128, 139, 148, 151) There were additional concerns about insurance and employment discrimination based on a genomic test result.(132, 143, 145, 146, 148, 149, 166)

*Goals & Professional Role*: Goals of the nurse or physician was explored by 11 articles (23%), while 14 articles (29%) reported views on professional roles. Nurses and physicians had mixed feelings about whether genomic information contributed to their clinical goals for the patient or aligned with their views about their professional role. Genomic information was not always perceived as particularly useful in the clinical setting.(139, 142, 147, 160, 161, 163, 164, 167) Genomic information was described as irrelevant by nurses and physicians in certain clinical disciplines, such as ophthalmology,(139) and by particular professionals, such as breast surgeons.(160, 161) Viewing genomics as irrelevant to clinical practice appeared to foster an active resistance to integrating genomics into practice.(139, 160, 161) In contrast, nurses' and physicians' were confident in their competence to provide genomic information,(113, 124, 141-143, 149, 151, 154, 155, 160, 161, 164) and in their view that genomic information provision was appropriate within their clinical role.(113, 123, 127, 132, 134, 148, 149, 151, 160, 161, 166) However, nurses and physicians were uncomfortable about providing genomic health information to 'at-risk' relatives of their patients.(134, 139, 151, 158, 160, 161)

*Intention & Optimism*: Intention of the nurse or physician was explored by 16 articles (33%), while 14 articles (29%) reported on optimism. Nurses and physicians expressed positive attitudes towards genomics,(113, 123, 127, 135, 137, 140, 157, 160, 161, 166, 168) reported their beliefs about the future benefit of genomic information for patients and society as a whole,(125, 132, 139, 149, 157, 166) and regarded genomic health information as an inevitable major factor in clinical care in the future.(123, 140, 144, 160, 161) Nurses and physicians expressed their intention to engage in continuing professional education, demonstrating the need for increased genomic literacy. Most nurses and physicians preferred clinically relevant education in the form of workshops, lectures or online content.(113, 124, 131-133, 136, 138, 139, 145, 147, 149, 155, 158-161) Descriptions of nurses' intentions to pursue further genomics education were more prevalent than articles reporting physicians' intentions.

## 2.8 DISCUSSION

This systematic review identified that, while there are a number of indicators that nurses and physicians are engaging with and have positive attitudes towards genomics, there are also significant barriers that prevent them from doing this on a routine basis.

Consistent with previous reports,(169) this review identified that nurses and physicians under-refer patients who require, or may require, assessment of their genetic risk based on their diagnosis or family history. Although there are likely to be a number of additional precursors to low referral rates, many nurses and physicians lack adequate genomics knowledge. Nurses' and physicians' low confidence in engaging in discussions about genomics or performing genomics-related tasks (such as obtaining family history information, performing a risk assessment or interpreting a genomic test result) suggests an awareness of their limited knowledge. While it has been suggested that few nursing and medical undergraduate degrees adequately prepare graduates to integrate genomic health information into their clinical practice,(170, 171) this review found that more recent nursing and physician graduates had better genomics knowledge scores than their more experienced colleagues.(132, 135, 136, 141) Although an inverse relationship between years of practice and knowledge has been reported previously,(172) this finding suggests educators are recognising the importance of graduates having adequate genomics knowledge and incorporating this into undergraduate programs. It was noted, however, that articles describing nurses' skill set were less prevalent than articles describing the abilities of physicians. For nurses and physicians who did not receive adequate genomics education in basic training or who trained a long time ago, accessing continuing professional development can be marred by financial and scheduling barriers.(173)

Collaborative relationships between the nurse or physician and clinical genetic professionals was highlighted in this review as a valuable resource, with the potential to improve access to genomics education and increase the number of appropriate referrals to clinical genetics services.(174) Nonetheless, while some nurses and physicians do feel capable of raising and discussing relevant genomic health information with their patients, others appear to engage reactively to their patient's request for genomic information or may feel obligated to initiate discussions where there are medical management implications dependent on a genomic test result.(175) Articles describing nurses' views about the appropriateness of genomics within their role were more prevalent than articles describing physicians' views.

Although issues of knowledge, skill, training and resources are playing a significant role, other important factors contribute to nurses and physicians' capacity to integrate genomic information into their practice.

Concerns about the ethical, legal and psychological aspects of genomic information appear to critically inform their motivation to integrate genomics into practice. Depending on the nurse's or physician's views, motivation to integrate genomics into practice may vary. Pleasingly, a substantial number reported the potential positive effect of genomic health information, such as personalising and improving medical management or providing risk advice to relatives who can benefit from screening or risk-reducing interventions.(12) However, only a small number of nurses and physicians feel genomic information can be improve psychological wellbeing.(113, 140, 148) Concerns about the potential for genomic information to inflict psychological harm on patients were frequently reported, despite genetic counselling demonstrating an ability to reduce anxiety and improve accuracy of perceived genetic risk.(176)

Ethical and legal considerations, such as insurance or employment discrimination resulting from inappropriate sharing of genomic information, were also raised. While these concerns have been reported elsewhere by research participants and the general public, sharing of genomic data is widely considered to be a necessary step to improve understanding of the genetic basis of disease and future medical care.(177) In this genomics era, government bodies are moving to develop ethical and legal safeguards for individuals and families, however, these processes can lag behind scientific developments and require refinement even after implementation.(178) Meanwhile, nurses and physicians who have significant ethical, legal or psychological concerns about genomic information may actively avoid initiating conversations about genomics with their patients.(139) Sidestepping the opportunity to explore a patient's genomic concerns may mean a vital opportunity is missed, particularly in specialties like palliative care, which represent the final chance to collect valuable patient knowledge about family history or a DNA sample which could benefit their relatives.(179)

### 2.8.1 Implication for Future Research

The majority of articles included in this review utilised an unvalidated survey to capture the barriers and facilitators faced by nurses and physicians in integrating genomics in their practice. Development of a validated tool to assess genomics practice, attitudes and

knowledge could be considered in future research, to enable more accurate comparisons between different specialties and disciplines.

To ensure patients and families have appropriate access to genomic health information, nurses and physicians need to successfully integrate genomics into their practice.(114) To achieve this aim, there is a need for further research to understand the context-specific barriers and facilitators (for example in palliative care, oncology and neurology) and develop evidence-based, theory-informed interventions.

### 2.8.2 Limitations

Limitations of this review relate to both the individual articles and review methodology. As discussed above, almost all quantitative reports used novel, unvalidated measures. To represent a range of disciplines and specialties, articles with high risk of bias were included, although their findings were present in other articles. Given resource issues, only English-language articles were included. The review was strengthened by adhering to the PRISMA guidelines and the use of a theoretical framework to map and synthesise outcomes.(105, 115) Although the findings of this review are not necessarily novel, synthesising the literature to date will assist the genomics implementation field in developing theory-informed, evidence-based interventions.

## 2.9 CONCLUSION

Building the capacity of nurses and physicians to integrate genetics and genomics into routine clinical care is essential if opportunities afforded by precision medicine are to be fully realised. Many nurses and physicians have limited knowledge and skills about genetics and genomics, do not feel confident addressing these issues with patients and lack resources and guidelines to direct them. Apprehension about ethical, legal and psychological impacts of genomic information influence willingness to engage in genomics discussions, unless requested by patients. This review identified potential behavioural targets to inform the development of theory-informed, evidence-based interventions to facilitate the integration of genomics into nurses' and physicians' usual care. Such interventions will need to be tailored to the specific clinical setting.(114)

## 2.10 REVIEW AND CRITICAL APPRAISAL OF PALLIATIVE–GENOMIC LITERAURE

In this section, I provide a focused appraisal of the palliative–genomic context by reviewing (a) the relevant literature included in my published systematic review, (b) literature not included in the systematic review because it did not meet the eligibility criteria, and (c) literature that has been published since the last literature search conducted for the systematic review (conducted on 30 August 2019). This section begins with a review of the barriers and facilitators to integrating genomics into palliative care. I then critically appraise the literature to identify the methodological and conceptual gaps.

### 2.10.1 Barriers and Facilitators in the Palliative Care Context

#### *2.10.1.1 Perspectives of Palliative Care Health Professionals*

A small body of literature reports several barriers and facilitators from the perspectives of palliative care health professionals.(180) A frequently reported barrier among palliative care nurses and doctors is low knowledge and confidence about genomics.(103, 113, 128, 133, 138, 181, 182) In particular, palliative care physicians report low knowledge of and experience with DNA storage.(133) Specific training about genomics (133, 138, 181) and the development of guidelines or web tools (133, 138, 181, 182) are suggested to improve their capability. Palliative care health professionals appear to refer their patients or relevant family members to clinical genetic services infrequently.(24)

Palliative care health professionals' attitudes towards genomics are variable, even within the same study. Some palliative care health professionals report the relevance and importance of genomics to palliative care,(103, 113, 127, 128, 181), whereas others have reservations about the appropriateness of the palliative care setting.(113, 128, 181) Concerns about distressing patients and families with genomic information are common.(103, 113, 128, 181) A desire for genomics to be addressed earlier in patient care may stem from a sense that genomics detracts from delivering quality palliative care.(128, 181, 183)

#### *2.1.1.1 Perspectives of Genetic Health Professionals*

The involvement of genetic health professionals in the care of people with palliative care needs is not well documented.(180) The proportion of people with palliative care needs as part of a genetic health professional's workload is not clear, although many report having been involved in addressing the genomic needs of a dying person or their family.(184) The addition of a genetic health professional to the palliative care team may help to address patients' and

families' questions about disease heritability, reduce uncertainty about the future, and facilitate adaption after learning about difficult genomic information.(185) In addition, genetic health professionals could provide support to palliative care health professionals to assess eligibility for genomic testing and support facilitation of genomic testing or DNA storage.(84, 85) However, further training may be needed to address genetic health professionals' discomfort surrounding end-of-life discussions.(184, 186, 187) Some have suggested that genetic health professionals may be able to initiate referrals to palliative care if they can identify unmet physical or psychological needs in a person with a life-limiting illness.(94)

#### *2.1.1.2 Perspectives of People with Palliative Care Needs and Families*

The literature about the views of people with palliative care needs and their families is limited. Some people have suspicions of disease heritability, concern for the health of family members, and a sense of relief following a genetic risk assessment.(92, 183) One study found that people who suspected they had a genetic cause for their cancer were three times more likely to be assessed as high risk,(92) whereas other studies have seen a low awareness of the role of genetics in disease development.(93, 188) Individuals appear to value genomic discussions because genetic knowledge improves their understanding of their disease and satisfies altruistic motivations to help others.(189) Family members of palliative individuals show general support for genetic counselling or testing, provided that this aligns with the palliative person's wishes.(96) In general, people with palliative care needs appear to be accepting of genomic discussions in the palliative care setting, but further research would strengthen the evidence base.(92, 93)

### 2.10.2 Critical Appraisal of the Palliative–Genomic Literature

There is limited evidence of the barriers and facilitators to integrating genomics into palliative care. Within the existing literature, there is problematic heterogeneity in the outcomes measured, methodological approaches, and results. Currently, limited inferences can be drawn about the overarching barriers and facilitators, particularly for the Australasian context.

Most of the evidence has arisen from countries outside of Australia; namely the USA,(92-94, 101, 133, 184, 185, 187, 188, 190-192) UK,(103, 113, 128, 181, 183, 189, 193) and Canada.(96, 127, 138, 182) Only six articles included in my systematic review were relevant to the clinical palliative care context,(103, 113, 127, 128, 133, 138) although several relevant articles were not eligible for inclusion. These included commentaries,(101, 179, 185) case

studies,(75, 94, 191), literature reviews,(180, 192-194), and unpublished doctoral theses.(181, 183) Only seven articles have been published in the past 5 years.(92, 93, 96, 113, 127, 138, 182) In a rapidly evolving environment such as clinical genomics, up-to-date evidence about the barriers and facilitators is essential to the design of an appropriate clinical intervention. Furthermore, only one article validated their survey instrument and used mixed methods to augment their findings.(103)

Some of the older articles (more than 10 years old) describe the inappropriateness of the palliative care setting to broaching genomics,(103, 128, 181) whereas the more recent literature seems to report this less. Uncertainty about the palliative care role in addressing genomics is a persistent issue.(113, 138) Clarifying health professionals' views about the appropriateness of the palliative care setting and their role in addressing genomics is required. Several articles have reported in-depth, qualitative findings related to the potential benefits and harms of genomic discussions, though confidence in these findings would improve by testing these themes with quantitative (or other) methodologies.(96, 113, 128)

## 2.11 CHAPTER SUMMARY

In this chapter, I presented a peer-reviewed, published, systematic review that synthesised the literature about the barriers and facilitators related to health professionals' capability, opportunity, and motivation to integrate genomics into their practice. After the systematic review, I focused upon the barriers and facilitators in the palliative care context and critically appraised the literature from the palliative–genomic field. Barriers to genomics integration include low knowledge and confidence. Concerns about the appropriateness of the palliative care setting and uncertainty about the palliative care role need to be resolved. The ability to draw conclusions for the Australasian context is impeded by the limited and heterogenous evidence. In the next chapter, I present the principles, philosophical approach, conceptual framework, and methodology selected to begin to address the gaps identified.

# 3 Research Principles, Philosophy and Design

## 3.1 PREAMBLE

Chapter 3 explores the theoretical concepts that underpin this thesis and links these concepts to methodological decisions made in the Genomic Information for Families of the Terminally ill (GIFT) Project. I start by exploring the theory related to intervention design, including the principles of implementation science and then explore my philosophical positioning and application of a conceptual framework. The chapter then moves into a more detailed description of the GIFT Project's objectives and design. I outline the exploratory sequential mixed methods approach, including the methods used for data connection and integration. Lastly, I explore the ethical considerations.

The methods used in individual studies are described briefly in this chapter in the context of the overarching design, but further details are provided in corresponding study chapters as part of the published, peer-reviewed manuscripts.

## 3.2 LAYING THE FOUNDATION FOR AN INTERVENTION

### 3.2.1 The Role of Implementation Science

Implementation science is the study of designing, implementing, and monitoring interventions within complex systems to improve the uptake of evidence-based medicine.(195) In contrast to implementation studies, implementation science evaluates the acceptance and impact of an intervention rather than the clinical effects. Broadly, there are three types of implementation science studies: process evaluations (study of the factors affecting implementation), formative evaluations (feeding back observations to the study team to refine the intervention), and summative evaluations (collating information about the uptake and impact of an intervention at the end of a study).(196) A process evaluation, in which observations are made through the collection of qualitative and quantitative data, can guide the development of an intervention by identifying the barriers and facilitators in a specific context. An intervention is then designed to target those barriers and facilitators.

The use of implementation science theory, models, and frameworks can support researchers to generate transferrable evidence by building on existing knowledge.(196) Integrating theory into the research design helps to organise complex health systems and delineate the roles of various actors and stakeholders.(86, 197) A health system framework, such as the World Health Organization Innovative care for chronic conditions framework (WHO ICCC; discussed further in section 3.4), is one such theoretical approach that can be used to conceptualise the potential barriers and facilitators for evaluation.(67)

According to the British Medical Research Council (MRC), the core elements of intervention design are to first understand the context of the intervention, program theory (the presumed effect and outcome of the intervention), views of stakeholders, key uncertainties, required refinements to the intervention (if one exists), and economic considerations (Figure 5).(102) Research teams develop this core knowledge before the intervention is developed, implemented, evaluated, and monitored for effectiveness, harms, acceptability, cost-effectiveness, scalability, and transferability.(102)

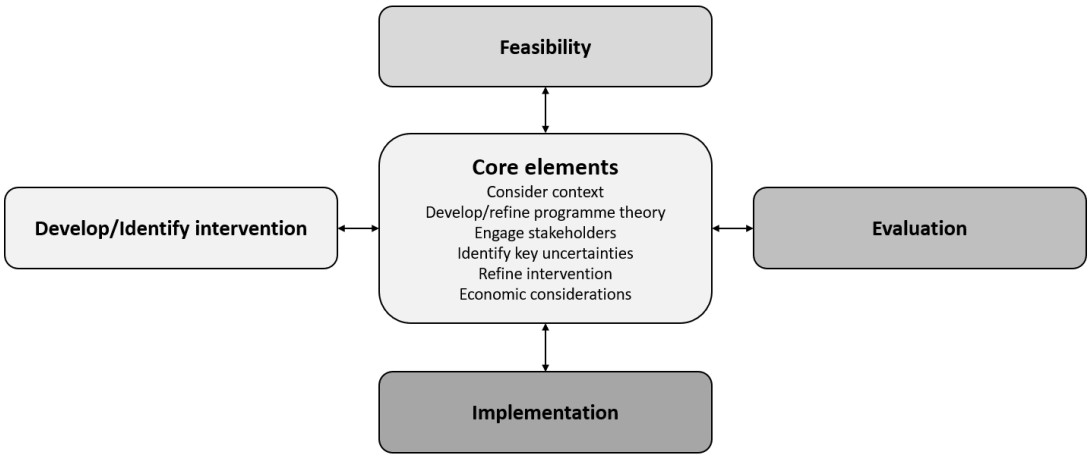


Figure 5. Framework for developing and evaluating complex interventions. Adapted from the British Medical Research Council Guidance on the design and evaluation of complex interventions.*(102)* NB. No permissions are required to reproduce or adapt this figure as it has been published under a Creative Commons license.

### 3.2.2 Applying the Principles of Implementation Science and Intervention Design to Define the Research Gap

Many of the 'core elements' listed in the MRC guidance cannot be identified within the existing literature in the palliative–genomic field. The barriers and facilitators in the Australasian context are not well described, and most studies have arisen from the USA, Canada, and the UK.(180) Although international evidence can be helpful for beginning to

identify and understand the barriers and facilitators in Australasia, an assessment of the local context is required.(102) The lack of Australasian evidence means that knowledge of the other core elements are also missing; that is, program theory cannot be developed without understanding the local barriers and facilitators, local stakeholders have not been engaged, key uncertainties have not been explored, and economic considerations have not been assessed. Each of these gaps represents an opportunity to build the evidence base for an intervention to support the integration of genomics into palliative care within Australasia.

In addition to the lack of Australasian knowledge, the principles of implementation science provide the methodological justifications for building the current evidence base. Of the available evidence, only one study used a mixed methods approach.(103) Process evaluations using mixed methods research draws on the exploratory strengths of qualitative research and progresses findings into more generalisable insights. For this reason, mixed methods research is recommended by expert groups for the development of an evidence base for healthcare interventions.(102)

## 3.3 PHILOSOPHICAL POSITIONING

Reflecting on my philosophical position as a researcher led me to explore the tension I felt between 'traditional' schools of research. On one hand, I believe in the value of establishing the 'truth'. This is reflected in my clinical work as a genetic counsellor, whereby I accept certain medical information to be objectively accurate and useful to individuals and families. On the other hand, my interactions with individuals and families taught me that truth is interpreted through a complex lens comprising many sociocultural factors. I was torn by these initial reflections and felt as though I needed to choose between a strictly post-positivist or constructivist position. I could see the benefits and limitations of the two schools of thought and felt confident both approaches could produce valuable evidence if applied in the right context. This led me to the philosophical position of pragmatism. Adopting a pragmatic position meant I did not need to hold to one worldview but rather could embrace multiple philosophies. I had previously viewed post-positivism and constructivism as opposing realities, but embracing pragmatism helped me to see these two worldviews as complementary.

Pragmatism gave me the language to articulate my position towards important philosophical concepts, such as ontology, epistemology, axiology, and rhetoric.(198) I resonated with pragmatism because, ontologically, I believe that individuals and communities experience reality in infinitely unique ways. However, research findings should generate

useable information to benefit these individuals and communities; therefore, truth needs to be accepted within the context in which it is produced. Epistemologically, I believe that to unearth the truth, diverse insights from individuals and communities must be obtained and explored. Regarding axiology, I believe research is value laden. It is impossible (and arguably, not desirable) to remove my existing views and prior experiences completely from the influence on my research. Reflecting on the biases I hold as a clinician, I committed to reflective practices (such as reflexive journalling and debriefing with supervisors) and to position myself explicitly within my research to ensure transparency. The rhetoric I use throughout this thesis has been chosen to remain person centred and to reflect the varied experiences, backgrounds, and views of participants. In keeping with modern inclusive language, I preference phrasing such as 'person with palliative care needs' to convey my view that people receiving palliative care are not simply 'palliative care patients' but are valuable individuals who are receiving palliative care.

### 3.3.1 Philosophical Approach to Qualitative Research

Qualitative research is an essential, humanising component of healthcare research that illuminates a social justice agenda, rouses a moral conscience, gives a voice to and advocates for vulnerable and underrepresented groups, and identifies life-saving actions and harmful practices.(199 p58-9) Beginning the GIFT Project with a qualitative phase enabled me to capture and explore a broad range of perspectives from genetic and palliative care health professionals. This exploratory approach was important because of the limited evidence about the barriers and facilitators.(200)

The genetic counselling research discipline is relatively new and uses diverse qualitative approaches.(201) I selected interpretive description as a conceptual label for the qualitative methods used in the GIFT Project.(202) Originating from the nursing discipline, interpretive description is a practical approach to qualitative research that encourages the generation of clinically relevant findings. The purposes of interpretive description are to answer real-world questions in the context of the available evidence and to communicate the findings to relevant stakeholders in a practical way.(202 p40)

### 3.3.2 Philosophical Approach to Quantitative Research

Quantitative research has long been considered the gold-standard for generating knowledge because of traditional philosophical assumptions that there is an objective, observable truth. Researchers with post-positivist philosophical positioning tend to be drawn

to quantitative research, in which reliability and objectivity are held in high regard.(203) Rigorous quantitative research can triangulate findings developed with other methods (such as qualitative research) and improve generalisability.

More recently, the role of axiology in quantitative research has been challenged. Post-positivists have been criticised for assuming that the researcher's role and values do not influence their findings.(204) Critics have argued that interpretations of quantitative data are subject to the researcher's values and biases and that this influences how data are understood and presented.(204) Philosophical approaches to quantitative research can be less well articulated, which can make it difficult to judge whether bias has influenced the findings.(203, 205) Agreeing with these axiological arguments against a post-positivist approach, I continued to position myself pragmatically and used reflexive practices throughout the quantitative phase of the GIFT Project.(200 p183-5)

### 3.3.3 Interdisciplinary Positioning

Although this is primarily a thesis about genetic counselling, I have integrated interdisciplinary terminology, knowledge, and research traditions from palliative care, clinical genetics, and implementation science. This thesis has benefited from the input of several disciplines; however, there are challenges to communicating effectively with interdisciplinary audiences, which I address below.

Using terminology that all groups share a common understanding of is a significant challenge. One example is using 'genomic(s)' to refer to both 'genetic' and 'genomic' testing. Despite the technical differences between these terms, the use of multiple terms can impede clarity. Unless the context requires a different term, I use 'genomic' for brevity and to reflect the forward perspective of integrating genomic medicine into routine healthcare. I use 'genetic' to refer to 'genetic counselling', as this is the commonly accepted terminology related to the genetic counselling profession.

Another example is terminology to describe a person receiving palliative care. People with palliative care needs traverse a range of clinical scenarios and settings, which can make it difficult to encompass these differences within one phrase. The person may be receiving or ceasing active treatment (e.g., chemotherapy). They may be in their last months, weeks, days, or hours of life, or be living with a chronic, life-limiting illness. The person may be receiving care from a palliative care specialist or a non-specialist health professional. They may be an

elderly person, a young adult, or a child. Unless stated otherwise in this thesis, a 'person with palliative care needs' refers primarily to adults receiving care from a palliative care specialist.

Conveying appropriate conceptual detail to interdisciplinary audiences is an additional challenge. In this thesis, I have sought to provide enough detail to demonstrate my grasp of relevant concepts, but not so much detail that the main messages for a non-specialist audience are lost.

Lastly, while writing in the third person (or passive voice) is common in nursing and medical scientific literature, I preference the active voice wherever possible.(206) The purpose of using the active voice is to convey transparency about my role and decisions while conducting this doctoral research.(207)

## 3.4 CONCEPTUAL FRAMEWORK

The use of theory, in the form of a conceptual framework, is often applied in mixed methods research. Conceptual frameworks can provide an explanatory scaffolding, which helps healthcare researchers to design their study.(208 p43-44) Frameworks can be used to guide the development of appropriate research questions and lend credence to findings. For these reasons, applying a conceptual framework to the design, analysis, and synthesis of the GIFT Project was intended to enhance the rigour of my research and to anticipate the type of barriers and facilitators that would need exploration.

I selected the World Health Organization Innovative care for chronic conditions (WHO ICCC) Building Blocks For Action framework as a conceptual framework.(67) The WHO ICCC framework describes the interrelated factors affecting healthcare delivery for chronic disease care, as opposed to acute care (such as accident and emergency care). The chronic nature of genetic conditions and palliative care made this a suitable framework to apply to the GIFT Project.

The framework comprises three levels, described as the micro-, meso-, and macro-level factors. The factors at each of these levels influence the provision of care to people with chronic conditions. The micro-level refers to patient–provider interactions that value individuals as partners in their care. The meso-level refers to healthcare organisations and communities, including the organisation of health services and delivery of education to health professionals. The macro-level refers to the policy environment, including government investment and legislation.

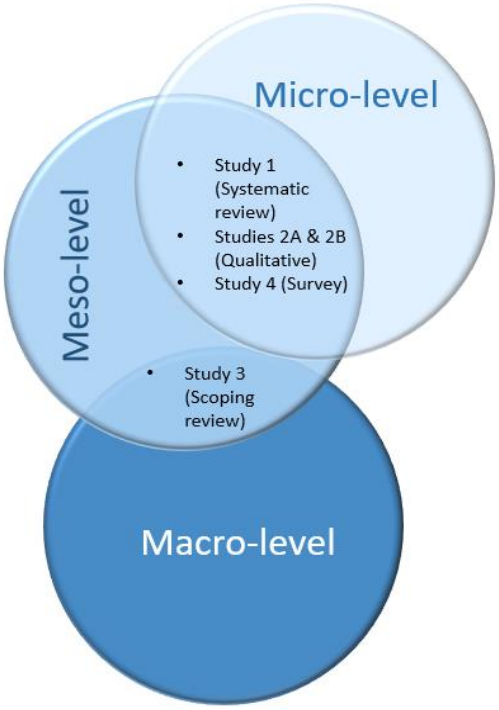


Figure 6. The WHO ICCC framework levels (micro-, meso-, and macro-levels) and corresponding studies

The WHO ICCC framework was applied to the thesis in several ways. Before conducting the systematic review, the framework helped me to conceptualise the potential barriers and facilitators in preparation for the narrative synthesis. As I progressed to primary data collection, I scaffolded my interview and survey questions to capture data across the three levels so not to miss important findings. I used the framework as a lens in my qualitative analysis to guide the organisation of findings and themes. The framework was the impetus for a scoping review of policy recommendations after identifying that the macro-level factors were inadequately captured by the qualitative study. As intended, the studies within this thesis have generated evidence relevant to each level of the WHO ICCC framework (Figure 6.).

## 3.5 RESEARCH OBJECTIVES

Three research objectives were developed to meet the thesis aim and answer the research questions (as defined in chapter 1) of the GIFT Project.

1. Explore the barriers and facilitators to integrating genomics into the care of people with palliative care needs (qualitative objective)
2. Compare the perceptions of the identified barriers and facilitators between genetic and palliative care health professionals (quantitative objective)
3. Identify what is required to support the integration of genomics into the care of people with palliative care needs and their families (mixed methods objective)

## 3.6 RESEARCH DESIGN

### 3.6.1 Overview of the GIFT Project

The GIFT Project was a pre-intervention process evaluation conducted over four years using an exploratory sequential mixed methods design to generate an evidence base of the barriers to and facilitators of the integration of genomics into the care of people with palliative care needs and their families (Figure 7).(208 p223) I adopted a broad approach to examine the support required at each level of the health system.(67)

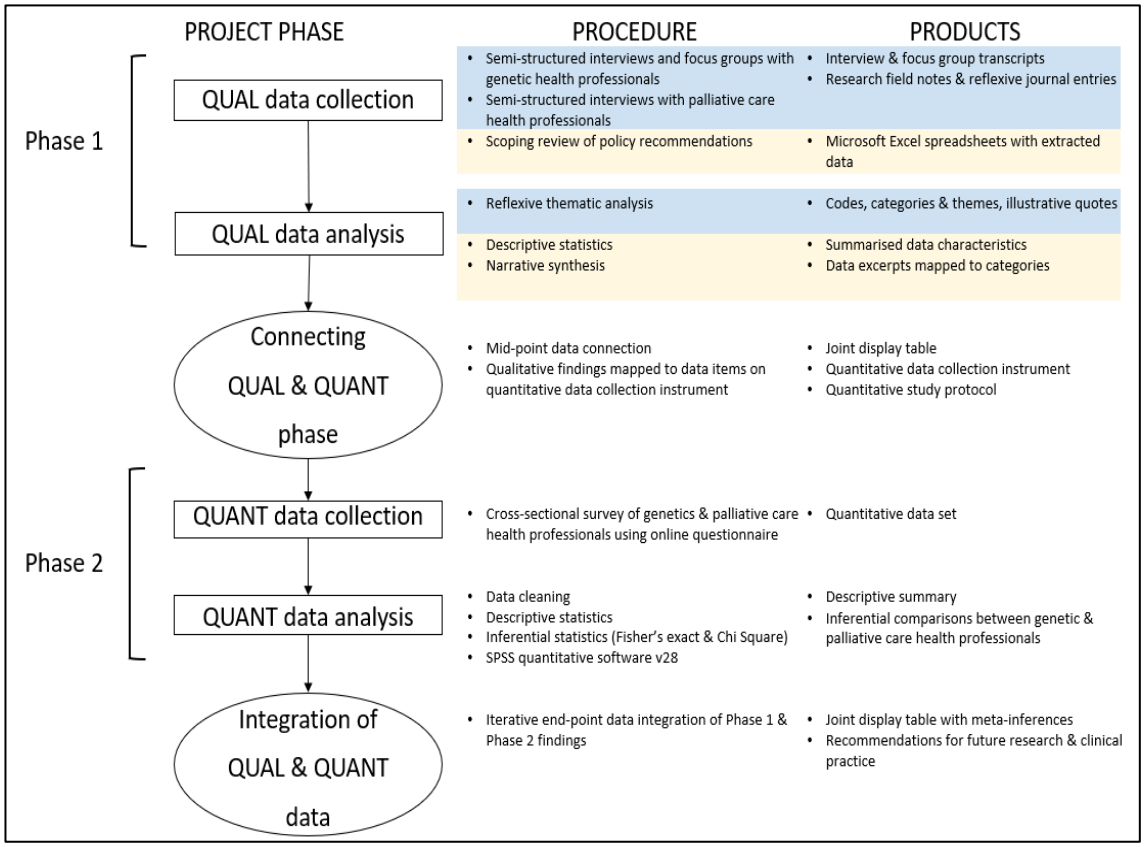


Figure 7.Visual representation of the GIFT Project procedures and products using an exploratory sequential mixed methods design. Blue shading represents the qualitative study and yellow shading represents the scoping review. QUAL: qualitative, QUANT: quantitative

Each of the component studies of the GIFT Project was designed to fulfil a research objective (Figure 8). Objective 1 was met through Phase 1, which comprised the qualitative study and scoping review. Objective 2 was met through Phase 2, which comprised the online questionnaire. Objective 3 was fulfilled through the process of integrating the qualitative and quantitative data from Phase 1 and Phase 2.

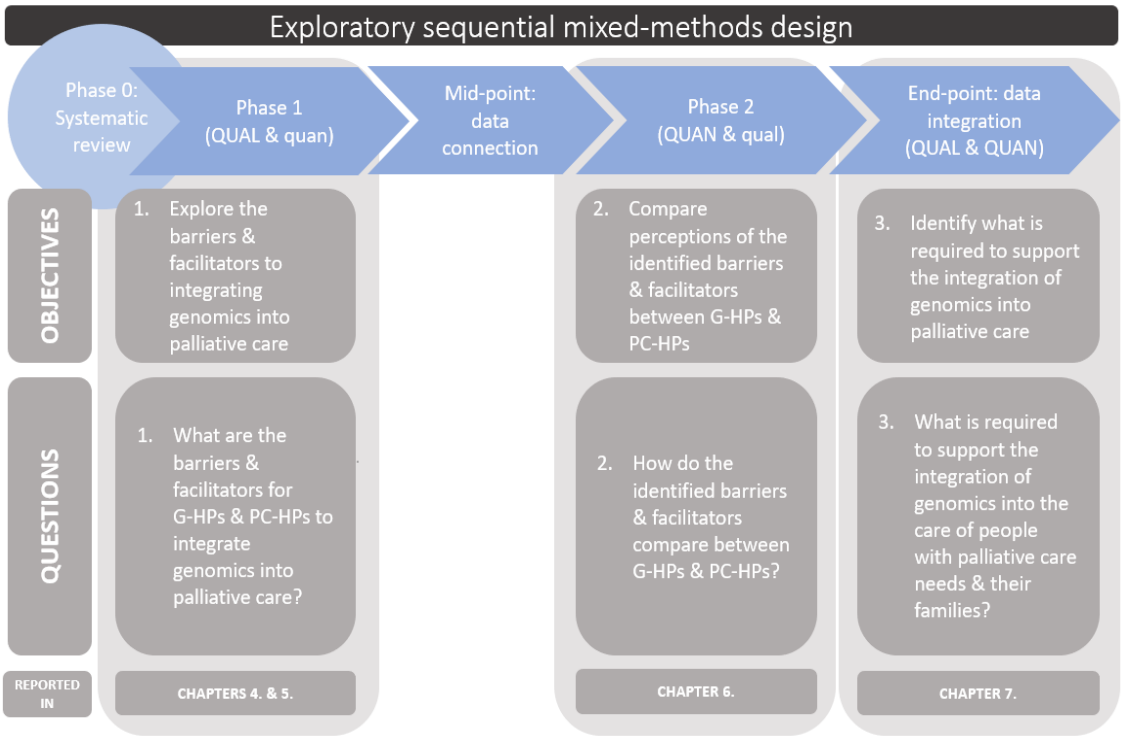


Figure 8. The three research objectives each aligned with a different research question and phase of the GIFT Project. Abbreviations: G-HP=Genetic health professional, PC-HP=Palliative care health professional

### 3.6.2 Setting and Participants

The GIFT Project was conducted at the University of Technology Sydney (UTS) located in Sydney, New South Wales, Australia. Although some study investigators are affiliated with external universities and hospitals, no external organisations (academic, clinical, or otherwise) were involved in the conduct of the GIFT Project.

Health professionals are recognised as key actors in the delivery of healthcare, and their behaviour is considered crucial to the design, implementation, and monitoring of a future intervention. The health professionals recruited into the studies in this thesis were genetic health professionals (defined as genetic counsellors and clinical geneticists) and palliative care health professionals (defined as specialist palliative care nurses and doctors). The eligibility criteria for inclusion in the studies in Phase 1 and Phase 2 are shown in Table 2.

Table 2. Eligibility characteristics of participants in the studies of the GIFT Project.

| **Participant group** | **Inclusion criteria** | **Exclusion criteria** | |
|---|---|---|---|
| Palliative care nurses | Registered nurses who provide direct nursing or who manage nurses who provide direct nursing care to people with palliative care needs and families in Australia or New Zealand. This may include but is not limited to direct clinical care, research, teaching, or policy development. | • Student nurses<br>• Enrolled nurses | • Unable to speak adequate English to engage meaningfully in an interview or focus group<br>• Nurses, medical doctors, and GCs who have not met their professional registration (or equivalent) requirements as stipulated by their relevant professional boards within the past 5 years |
| Palliative medicine doctors | Medically trained interns, residents, registrars, or consultants whose majority role (≥50% of clinical workload) is to provide palliative care/medicine in Australia or New Zealand | • Medical students | |
| Clinical geneticists / Genetic doctors | Medically trained interns, residents, registrars, or consultants or other appropriately trained medical doctors whose majority role (≥50% of clinical workload) is to provide clinical genetics or familial cancer services in Australia or New Zealand | | |
| Genetic counsellors (GCs) | GCs with Part 1 certification with the HGSA** (or who are registered with the HGSA) and were involved in providing genetic counselling services in Australia or New Zealand | • Student GCs<br>• GCs who have only worked in a non-clinical role | |

** Please note, Part 1 Certification for genetic counsellors has become an outdated professional description since designing the GIFT Project. The equivalent description is included in brackets. GC: Genetic counsellor; HGSA: Human Genetics Society of Australasia

The decision to capture barriers and facilitators related to the Australian and New Zealand setting was made primarily because of the potential to recruit genetic and palliative care health professional within the one strategy. Two of the health professional organisations that facilitated participant recruitment (Human Genetics Society of Australasia, and Australian and New Zealand Society of Palliative Medicine) span both Australia and New Zealand. Though other Oceanic nations (such as Vanuatu and Fiji) can be incorporated into the definition of Australasian, I decided not to include them because of the small numbers of genetic and palliative care specialists in these countries, and the difference in health service delivery.

Participants were eligible regardless of their work setting (e.g., public or private) as I wanted the findings of the GIFT Project to capture potentially diverse views and experiences across different health services. Participants could work in any genetic or palliative care setting, whether it be community, in-patient, out-patient, hospice, or elsewhere. Participants were eligible if they had delivered care to a person with palliative care needs with any disease type, including those with malignant and/or non-malignant disease.(73) By capturing the barriers and facilitators relevant to all people receiving palliative care, the findings were intended to benefit a broader palliative care population rather than limiting the benefits to those with a cancer diagnosis.

### 3.6.3 Mixed Methods Research

Mixed methods research is defined as the collection and integration of qualitative and quantitative data. A benefit of mixed methods research is the ability to enhance the strengths and offset the weaknesses of qualitative and quantitative research, while coming to a higher understanding of the research phenomena. Several mixed methods approaches are possible, depending on the objective, sequence of data collection, number and timing of studies, emphasis placed on the qualitative or quantitative phase, and the point at which data integration occurs. Data integration is the process of generating meta-inferences by integrating qualitative and quantitative data. A variety of tools and methods can be used for data integration including joint display tables, data merging, or narrative integration.(209)

Within mixed methods research, greater emphasis on one type of data is denoted using upper-case letters (e.g., QUAL or QUAN), and lesser emphasis is denoted by lower-case letters (e.g., qual or quan).(208 p62-3) In Phase 1 of the GIFT Project, qualitative data were given more weighting than quantitative (QUAL > quan). In Phase 2, quantitative data were given more weighting than the brief qualitative data (collected in free text response boxes; QUAN > qual). During data integration, both data types were given equal weighting. The data types given preference in each study, aligned with the phase, objective, and methods, are displayed in Table 3.The GIFT project displayed by phase, objective, method, data type, and manuscript title.Table 3.

### 3.6.4 Rationale for an Exploratory Sequential Mixed Methods Design

An exploratory sequential mixed methods design was best suited to the GIFT Project for several reasons. Firstly, the mixed methods design aligned with my pragmatic worldview. Pragmatism is inextricably linked with mixed methods research because of the flexibility to

choose different methodologies to answer research questions rather than making choices because of beliefs about a method's superiority or disciplinary traditions.(208 p39-43). In the GIFT Project, a qualitative approach was taken to explore the barriers and facilitators (Objective 1) because of the limited Australasian evidence and the opportunity to explore the topic deeply. To assess further and compare the initial findings between genetic and palliative care professionals (Objective 2), a quantitative approach was selected. Identifying support needs (Objective 3) required an integrated approach through the generation of meta-inferences to integrate the qualitative and quantitative findings.

Secondly, an exploratory sequential approach was best suited to my thesis aim, which was to generate an evidence base. The initial qualitative phase (Phase 1) generated rich data and contributed to filling the gap in the literature about genetics in palliative care, particularly in Australasia and from the perspectives of genetic health professionals. Phase 2 progressed the knowledge generated in Phase 1 by testing our data with quantitative methods. Here I aimed to advance the exploratory findings into usable, generalisable evidence that could enhance clinical practice.

Lastly, the decision to use an exploratory sequential mixed methods design was influenced by practical considerations. Practicality is equally important as methodological justifications because research needs to be feasible.(208 p89) Mixed methods approaches that use a 'concurrent' design, whereby qualitative and quantitative data are collected at the same time, are better suited to teams with greater resourcing capability. In the GIFT Project, a sequential approach was selected because, as a single researcher, it was not feasible for me to conduct multiple studies at once.

Table 3.The GIFT project displayed by phase, objective, method, data type, and manuscript title.

| Phase | Research objective | Study method | Data type | Manuscript title |
|---|---|---|---|---|
| Phase 0 | Synthesise literature about barriers and facilitators to identify conceptual and methodological gaps | Systematic review | quan & qual | Mainstreaming genetics and genomics: a systematic review of the barriers and facilitators for nurses and physicians in secondary and tertiary care |
| Phase 1 (QUAL & quan) | 1. Explore the barriers and facilitators to integrating genomics into the care of people with palliative care needs | Interpretive descriptive qualitative study with (a) genetic health professionals (semi-structured interviews and focus groups) and (b) palliative care health professionals (semi-structured interviews) | QUAL & quan | (a) Approaching discussions about genetics with palliative patients, and their families: a qualitative exploration with genetic health professionals |
| | | | | (b) Views and experiences of palliative care clinicians in addressing genetics with individuals and families: a qualitative study |
| | | Scoping review | qual & quan | Integrating genomics into palliative care: a global scoping review of policy recommendations |
| Phase 2 (QUAN & qual) | 2. Compare the perceptions of the identified barriers and facilitators between genetic and palliative care health professionals | Questionnaire survey study | QUAN & qual | A survey of genetic and palliative care health professionals towards the integration of genetics into palliative care |
| End-point data integration | 3. Identify what is required to support the integration of genomics into the care of people with palliative care needs and their families | Integration of findings from Phases 1 and 2 using joint display tables and generation of meta-inferences | QUAL & QUAN | N/A |

### 3.6.5 Data Integration

#### *3.6.5.1 Mid-Point Data Connection: Modifying an Existing Questionnaire to Meet the Needs of the GIFT Project*

A key feature of the exploratory sequential design is the building or connecting of the qualitative phase to the quantitative phase.(208 p84-93) 'Data connection' refers to the process of mapping findings from the first phase to the design of the next phase, as opposed to 'data integration', which involves mixing qualitative and quantitative data to assess congruence and produce meta-inferences (described further in section 3.6.5.2).

Phase 1 identified several barriers and facilitators that affect genetic and palliative care health professionals that were then assessed using quantitative methods in Phase 2.(210) The questionnaire used in Phase 2 was a pre-existing survey designed in 2018 by Dr Chris Jacobs, Professor Jane Phillips, Associate Professor Megan Best, Dr Kathy Tucker, Associate Professor Alison McEwen, and Dr April Morrow. The content was informed by a literature review that explored the barriers to and facilitators integrating genomics into palliative oncology.(180) In 2019, a Master of Genetic Counselling student, Grace Phillips, was assigned the questionnaire as a thesis project. The study initially intended to recruit participants from Australia, New Zealand, and the UK; however, delays to international site approvals prevented the timely distribution of the questionnaire. With further complications related to COVID-19, the Master's thesis project was redesigned as a pilot study.

Following completion of the unpublished pilot study, I incorporated this questionnaire into my doctoral thesis. The questionnaire had a natural congruence to my thesis aim and design. However, the data items on the questionnaire had not been designed based on my Phase 1 findings. To bridge this gap, I developed a joint display table to cross-check the existing items on the questionnaire with the data from the systematic review and Phase 1 (appendix B1).(210) If an existing item mapped to my Phase 1 findings, the item was retained. If a finding from Phase 1 was not represented, a data item was added to the questionnaire. For example, the original questionnaire did not have an item assessing potential facilitators, so a checkbox list of facilitators was added. The format of additional questions matched existing questions for congruency. For example, the 'facilitators' item was formatted to reflect a similar existing item to assess potential challenges. In consultation with supervisors, I did not remove any items from the questionnaire (even if there were no corresponding findings from Phase 1), as the pilot study had deemed the questionnaire feasible.

#### *3.6.5.2 End-Point Data Integration*

Data integration is a defining feature of mixed methods research and is characterised by the deliberate mixing or transformation of qualitative and quantitative data to generate meta-inferences. Several methods and tools to facilitate data integration are available.(211) I have used a joint display table to visualise and integrate across the four studies of the GIFT Project (appendix B2).(210) The systematic review findings were not included in the end-point joint display table so that meta-inferences were specific to the palliative–genomic context.

Before data integration, data within each study were subjected to intra-method analyses. For example, qualitative data were analysed with reflexive thematic analysis, and the quantitative data were analysed with descriptive and inferential statistics. Once the qualitative, scoping review and quantitative data had been analysed separately, the findings were mapped systematically using a Microsoft Excel spreadsheet. Each column corresponded to a different study, and data within the rows were mapped to similar concepts across studies. I engaged in an iterative and reflexive process of integrating findings across studies and drawing meta-inferences.

The process of integration required a deep engagement in the data. At first, many of the individual data items were reviewed and compared across studies. I generated inferences from the individual data items within each study, and this produced several 'high-level inferences'. Then, these high-level inferences were interpreted across the different studies to develop meta-inferences. The inferences and meta-inferences progressed from detail-oriented descriptions to broader conceptualisations. Regular meetings with my supervisors provided opportunities to discuss and reflect on my interpretations of the meta-inferences, which in turn progressed each iteration of the joint display table. I also determined whether each study converged, and noted whether findings across each study were confirmed, enhanced understanding, or were contradictory.(208 p227-34)

## 3.7 ETHICAL CONSIDERATIONS

The GIFT Project was guided by The National Statement on Ethical Conduct in Human Research, which is a set of guidelines for the ethical design, conduct, and dissemination of research involving humans in Australia.(212) Ethical approval for all aspects of this thesis was granted by the University of Technology Sydney Human Research Ethics Committee. Study approval numbers and ethical considerations for individual studies are mentioned within

corresponding manuscripts. Throughout the GIFT Project, I adhered to UTS policies that dictate proper data management and storage procedures (appendix B3).

Several stakeholders are anticipated to benefit from the findings of the GIFT Project, including individuals and families, health professionals, government agencies, professional organisations, health services, and hospitals. At the individual level, groundwork for an intervention to improve access to genomic information will provide clinical and psychological benefits to individuals and family members. At the health professional level, addressing the challenges of genomics in a palliative care situation could improve job satisfaction and support provided by health services, and help to reduce the risk of burnout. At the health service and policy level, research in the Australasian context will increase awareness among policy makers about incorporating relevant guidance for health services.

### 3.7.1 Ethical Considerations of the Aim, Objectives, and Design

The aim and questions of this thesis were developed with the principles of beneficence, non-maleficence, and justice in mind. At the beginning of my doctoral program, I had hoped to design an intervention. However, finding little evidence to underpin the design of an intervention, I focused on building the evidence base. Proceeding to developing an intervention would not best serve the palliative care population because such an intervention would not have been informed by theory. In a worst-case scenario, an ill-informed intervention could cause harm to this vulnerable population. People with palliative care needs and their families deserve an evidence-based intervention that improves access to genomic health information.(102)

A robust research design is an important ethical consideration because it reduces waste of researcher resources and ensures respect of the participant's time and expertise.(212) The exploratory sequential design meant I could explore thoroughly the myriad barriers and facilitators affecting genetics in palliative care before assessing these findings in a larger population. The ground-up approach meant I could capture the nuances relevant to the Australasian context in Phase 1 and increase the likelihood that the data collection tool used in Phase 2 would reflect participants' views and experiences. Recognising that people with palliative care needs and their families can be in a vulnerable situation, I felt that obtaining health professionals' views in the first instance was of low risk in this initial exploration.

Online data collection through interviews, focus groups, and questionnaires provided an opportunity for individuals from diverse geographical, financial, or personal circumstances to

participate. Logistical barriers to participation in an in-person interview, focus group, or survey, such as the inability to travel, lack of time to attend a face-to-face interview, or being in a rural area, were overcome by offering virtual options. Participants could schedule the interview at a time and place convenient to them. For some, this meant being able to participate from their workplace rather than having to ask for leave to attend a face-to-face interview or focus group. For others, interviews were scheduled at a time convenient to their family commitments. Online data collection can be a barrier to people who are uncomfortable or unfamiliar with technology, or without access. However, given health professionals were recruited during the era of COVID-19, when telehealth services were commonly used, I felt the risk of exclusion based on technology was low.

### 3.7.2 Pre-Existing Relationships

My role as a clinical genetic counsellor meant there was high likelihood I would have pre-existing relationships with people who volunteered as participants in my studies. Pre-existing relationships have the potential to infringe on the autonomy of participants by unduly coercing people to participate.(212) In addition, part of the recruitment strategy in the qualitative study was to send invitation emails to the research team's known contacts. I applied for an ethics amendment with this approach to help supplement low response rates, particularly from palliative care health professionals. I acknowledged the risk of emailing known contacts, including the potential for coercion and harm to existing relationships. I took several steps to mitigate the risk of coercion with participants as follows.

- During recruitment, I reiterated the voluntary nature of participation, their right to withdraw at any time, and that non-participation would not harm their relationship with me, the research team, or UTS.
- Participants were informed the focus group or interview could be conducted by another member of the research team, rather than myself, if they preferred.
- I used a template email (approved for use by the ethics committee) that included a statement about their right not to respond and that non-response or non-participation would not harm their relationship with the research team or UTS.
- A maximum of two emails per contact were sent.

### 3.7.3 Risk Mitigation for Participants

Risk to participants through participation in an interview, focus group, or questionnaire included the potential for discomfort or inconvenience.(212) Participants could have

experienced psychological discomfort, embarrassment, or harm to their reputation by articulating an opinion or describing an experience in a focus group. Participants could have become distressed if a question evoked a painful, shameful, or otherwise negative emotion. At the beginning of the interview, focus group, or questionnaire, these risks were addressed as part of a verbal consent script or participant information statement. Focus group participants were reminded to be respectful of the opinions of others and confidentiality. During the qualitative data collection, I used my clinical counselling skills to monitor participants for negative responses or emotions through verbal and non-verbal communication. For the qualitative study, a distress and safety protocol was developed to manage participant distress (appendix C6).

## 3.8 CHAPTER SUMMARY

This chapter has described the theoretical concepts underpinning the GIFT Project. The overarching concepts included intervention design, including the principles of implementation science, pragmatic philosophy, and the WHO ICCC conceptual framework. I outlined the exploratory sequential mixed methods design of the GIFT Project, rationales for the methodological decisions, and ethical considerations. In the next chapter, I present the beginning of Phase 1 of the GIFT Project. Two peer-reviewed and published manuscripts describe the initial qualitative exploration of the barriers and facilitators affecting the integration of genomics into palliative care.

# 4 Qualitative Exploration of the Barriers and Facilitators

## 4.1 PREAMBLE

Chapter 4 describes the beginning of Phase 1 of the Genomic Information for Families of the Terminally ill (GIFT) Project with the qualitative exploration of the barriers and facilitators. The two manuscripts within this chapter arose from one qualitative study with two cohorts: genetic health professionals and palliative care health professionals. This study addressed the micro-level (patient–provider interactions) and meso-level (organisational level) factors of the World Health Organization Innovative care for chronic conditions (WHO ICCC) framework. The study design, procedures, and methods were similar for the two groups, except that the data from each cohort were analysed and reported separately. This approach preserved the nuanced differences between the two groups for assessment in Phase 2.

Following the conclusion of the first manuscript, an extension of the results with an additional theme related to genetic health professionals' views on DNA storage is presented (section 4.9 to 4.11). In consultation with my supervisors, these results were not reported in the published manuscript because they fell outside the scope of the manuscript, which had a limited word count and aimed to tell a cohesive story. However, these findings are relevant to the thesis aim and were included in the 'mid-point data connection' process for assessment in Phase 2. Files related to the planning and conduct of the qualitative study are available in appendix C.

## 4.2 MANUSCRIPT 2. QUALITATIVE EXPLORATION WITH GENETIC HEALTH PROFESSIONALS

This manuscript was published in *European Journal of Human Genetics* in 2022 (Scimago rating Q1 in the 'genetics' category, 2021 impact factor 5.351).(213) No permissions were required to reprint this manuscript because it was published under a Creative Commons license. Minor edits have been made, including changing 'genetic' to 'genomic' (except in the manuscript title) and updating table headings and page numbers for congruency across the thesis.

Reference: White S, Turbitt E, Phillips J, Jacobs C. Approaching discussions about genetics with palliative patients, and their families: a qualitative exploration with genetic health professionals. Eur J Hum Genet. 2022:1-8

## 4.3 ABSTRACT

Genomic information can provide clinical benefits to families of people with palliative care needs. However, integration of genomics into mainstream medicine has not focused on palliative populations. We explored the views and experiences of genetic health professionals in addressing genomics with people receiving palliative care, and their families. We conducted an interpretive descriptive qualitative study with genetic counsellors and clinical geneticists using interviews and focus groups. Findings were generated using reflexive thematic analysis. Three themes were identified: 1) Focusing on the benefit to the family, 2) The discomfort of addressing genomics near end of life and 3) "It's always on the back-burner": challenges to getting genomics on the palliative care agenda. Participants discussed the familial benefit of genomics in palliative care alongside the challenges when patients are near end of life. They perceived genomics as low priority for palliative care due to misunderstandings related to the value of genomic information. Acknowledging the challenges in the palliative care context, genetic health professionals want improved service leadership and awareness of the familial benefits of palliative-genomic testing. Strong leadership to support genetic health professionals in addressing these barriers is needed for the benefits of genomic information to be realised.

## 4.4 INTRODUCTION

Identifying the genetic contribution of a palliative patient's condition has utility for their family, as it offers the possibility to reduce morbidity and mortality if preventative measures are indicated and actioned.(24) However, up to a quarter of patients with life-limiting conditions, and their relatives, may be missing the opportunity for genetic counselling or testing prior to the terminal phase of their disease.(169) While genetic and genomic testing ('genomic testing') close to end of life is unlikely to benefit the patient, it could help family members assess their own genetic risk and make health, reproductive, social and financial decisions.(14, 16) Health professionals providing active treatment may not identify eligible patients for genomic testing early in their disease trajectory because of low genomics knowledge, competing clinical priorities or patients bypassing traditional treatment

pathways.(214) Palliative care then becomes the final opportunity to conduct a genomic risk assessment and, if indicated, collect a DNA sample for the future benefit of family members.

Genetic counselling guidelines recommend adopting a person and family-centred approach, which suggests genetic health professionals should be conscious of the unique ethical and practical issues affecting people with palliative care needs, and their families.(215) However, little is known about the views and experiences of the workforce providing genetic counselling and testing to people with palliative care needs. First-person accounts and peripheral reports describe the emotional impact of genetic health professionals' close proximity to death and grief.(184, 185) Further work is needed to understand the views and experiences of genetic health professionals when discussing genomic issues with people with palliative care needs, and their families.

Alternatives to traditional clinical genetics pathways, including mainstreaming (non-genetic health professionals managing genomic testing) and translational genomic research, are changing the nature of genetic health professionals' interactions with people with palliative care needs and their families.(160, 216) Genetic health professionals generally support new delivery models because of the rapid integration of genomics into mainstream medicine and increasing workforce pressure.(54, 217) However, mainstreaming into palliative care appears to have received less attention, despite patients' interest to discuss genomic testing to address existing concerns about their family members' future disease risk.(92, 93) Some palliative care health professionals report having the capability to discuss genomics with their patients, others have varied opinions on the relevance of genomics to palliative care and concerns about causing harm, but most desire further education and support to improve their confidence.(127, 218) Understanding the views of genetic health professionals about delegating responsibility and providing support to palliative care health professionals will build evidence for an intervention designed to support the integration of genomics into the palliative care setting.(102) Therefore, we aimed to explore genetic health professionals' views and experiences of integrating genetics and genomics into the care of people with palliative care needs, and their families, including their perceptions of the barriers and suggestions to support integration.

## 4.5 MATERIALS AND METHODS

### 4.5.1 Design

We used an interpretive descriptive qualitative study design with online focus groups and semi-structured interviews.(202) These findings are a sub-set of data from a broader qualitative study that additionally recruited palliative care nurses and doctors (reported elsewhere).(218) The study protocol was pre-registered: https://osf.io/h4gt9/.

### 4.5.2 Theoretical Approach

Due to the limited evidence about genomics in palliative care, we selected an inductive approach to explore the boundaries of our participants' views and to ensure our findings were grounded in the data.(219 p9-10) Underpinned by a pragmatic epistemology, we aimed to generate findings relevant to the Australasian setting, but that could also inform stakeholders in comparable countries.(102)

### 4.5.3 Participants and Sampling

We invited Australian and New Zealand genetic counsellors and clinical/cancer geneticists (including trainees) via their professional organisations (see appendix C9). Organisations sent an email blast or included the invitation in their newsletter. We published the invitation on our professional Twitter accounts and asked participants to snowball the invitation.

### 4.5.4 Data Instrument and Collection

We developed a semi-structured focus group and interview guide informed by the World Health Organization Integrated Care for Chronic Conditions framework and existing literature.(67, 214) We piloted the guide with two genetic health professionals who suggested reordering two questions and clarifying whether we wanted responses about malignant and non-malignant cohorts. We asked about experiences of genomic discussions with people with palliative care needs and their families, barriers and facilitators, and perceptions of palliative care health professionals and organisations' roles (e.g., hospital, health service) in integrating genomics into the palliative care setting (interview guide in appendix C10).

Participants completed a demographic survey to provide context to their responses. We opted not to collect specific geographical location (e.g., state) or qualification/training status (e.g., clinical geneticist vs. clinical genetics fellow) to reduce the chance of participant

identification. We conducted semi-structured interviews and focus groups via Zoom (except one in-person interview) due to COVID-19 limitations.(220)

We prioritised focus groups to encourage fluidity of ideas, but offered one-on-one interviews if the individual preferred.(221) S.W conducted all individual interviews and moderated two focus groups and J.P moderated one focus group. Either S.W, C.J or J.P acted as an 'observer' at each focus group to take notes and to provide feedback and a summary to the moderator.(222) Interviews and focus groups were audio- and video-recorded on Zoom, transcribed verbatim and de-identified.(220) We returned transcripts to participants to check them for accuracy. We made a pragmatic decision to discontinue data collection when no new information related to the research questions was being identified in subsequent interviews. We acknowledge there is always potential for additional insights from continued data collection and agree with arguments that declarations of data saturation are incongruent with reflexive thematic analysis.(223)

### 4.5.5 Data Analysis

Though we acknowledge the methodological differences between focus groups and individual interviews, we offered both options to our participants. We prioritised focus groups to encourage fluidity of ideas, expecting that some participants may have limited experience of addressing genetics with individuals and families. We encouraged the exchange of ideas but did not explicitly analyse the interactions between focus group participants. Participant responses were analysed in the same way regardless of whether they were captured by a focus group or individual interview.

Using inductive reflexive thematic analysis, we employed NVivo V12 to code transcripts and Microsoft Excel and Word to develop themes relevant to the research question.(224, 225) S.W led the analysis. C.J. co-coded two transcripts to engage with S.W. about data interpretation and resulting codes. We revised codes over several iterations before organising into initial themes. S.W. and E.T. met weekly over ten weeks to discuss and develop themes, with monthly input from the C.J. and J.P. We actively sought disconfirming cases.

### 4.5.6 Reflexivity

As a clinical genetic counsellor, I (S.W.) considered myself an 'insider' to the participants (226). The advantage of being an insider is easier access to participants, shared language and concepts, and a rich understanding of the topic with the potential to notice important

subtleties. However, the disadvantage is the potential to introduce bias from pre-formed opinions and lack of objectivity. To develop my reflexivity, I kept a journal to explore my reactions, thoughts, and feelings throughout the research process, from study conception, through to participant interviews, data analysis and writing up the findings. Though this diary did not form part of the analysed data, it enabled me to critically view my interpretations and assumptions. Several times I reviewed the journal contents with the research team to garner their perspectives and deepen my insights during data analysis. I drew upon the collective qualitative research training of the study team and engaged in critical discussions about theme development to ensure our findings were true to our participants' views.

### 4.5.7 Ethics

We recorded verbal consent (verbal consent script in appendix C11). Participants with a pre-existing relationship with the interviewer were given additional reminders that non-participation would not harm their relationship with the research team. The University of Technology Sydney Human Research Ethics Committee granted ethics approval (ETH20-5046/20-5347).

## 4.6 RESULTS

We conducted three focus groups (two with four and one with five participants) and 13 one-on-one interviews between October and December 2020, totalling 26 genetic health professional participants. Focus groups lasted between 54 to 58 minutes (average 56 minutes) and interviews lasted between 21 to 52 minutes (average 29 minutes). There were ten (38.46%) medical doctors (clinical/cancer geneticists and clinical/cancer fellows/trainees) and 16 genetic counsellors (61.54%). Most were female (*n*=21, 80.77%), worked in a metropolitan area (*n*=21, 80.77%), public setting (*n*=23, 88.46%) and had 0-5 years' experience (*n*=11, 42.31%) (Table 4). Two additional genetic counsellors expressed interest in participating; one was lost to follow-up and one did not participate due to scheduling issues.

Participants had palliative experience predominantly in cancer genomics, and to a lesser extent in general clinical genetics, research and neonatal intensive care settings. Most interactions occurred in outpatient clinics. Some patients had genetic counselling during active treatment with supportive palliative care, while others were in their last days or weeks of life. For patients at end of life, DNA storage, rather than genomic testing, was more common.

Three themes were identified: 1) Focusing on the benefit to the family, 2) The discomfort of addressing genomics near end of life and 3) “It’s always on the back-burner”: challenges to getting genomics on the palliative care agenda. The third theme consists of two subthemes: a) Burden of proof: instilling the value of genomics in palliative care, and b) “Individuals can only do so much”: finding solutions in the absence of service leadership.

Table 4. Participant demographics (*N*=26)

| Participant | Sex | Age range | Discipline | Years of experience | Work location[2,3,4] | Work sector |
|---|---|---|---|---|---|---|
| P1* | Female | 31-45 | Genetic counsellor | 6-10 | Regional | Public |
| P2* | Female | 31-45 | Genetic counsellor | 6-10 | Metropolitan | Public |
| P3^ | Female | 46-60 | Genetic counsellor | >15 | Metropolitan | Public |
| P4* | Male | 31-45 | Genetic counsellor | 0-5 | Metropolitan | Public |
| P5 | Female | 31-45 | Genetic counsellor | 6-10 | Regional | Public |
| P6 | Female | 31-45 | Genetic counsellor | 0-5 | Metropolitan | Public |
| P7* | Female | 18-30 | Genetic counsellor | 0-5 | Metropolitan | Public |
| P8* | Female | 18-30 | Genetic counsellor | 0-5 | Metropolitan | Public |
| P9^ | Female | 31-45 | Genetic counsellor | 0-5 | Metropolitan | Public |
| P10^ | Female | 31-45 | Genetic counsellor | 11-15 | Metropolitan | Public |
| P11 | Female | 46-60 | Genetic counsellor | >15 | Rural | Public |
| P12^ | Female | 18-30 | Genetic counsellor | 0-5 | Regional | Public |
| P13 | Female | 31-45 | Genetic counsellor | 6-10 | Metropolitan | Public |
| P14 | Female | 18-30 | Genetic counsellor | 0-5 | Metropolitan | Public |
| P15 | Female | 31-45 | Genetic counsellor | >15 | Metropolitan | Public / private[5] |
| P16 | Female | 31-45 | Genetic counsellor | 11-15 | Metropolitan | Public |
| P17~ | Male | 31-45 | Medical doctor[1] | 0-5 | Metropolitan | Public |
| P18~ | Female | 31-45 | Medical doctor | 0-5 | Metropolitan | Public |
| P19~ | Male | >60 | Medical doctor | >15 | Metropolitan | Public |
| P20 | Female | 31-45 | Medical doctor | 0-5 | Metropolitan | Public |
| P21~ | Female | 31-45 | Medical doctor | 0-5 | Metropolitan | Public |
| P22 | Female | >60 | Medical doctor | >15 | Regional | Public / private[5] |
| P23 | Male | 46-60 | Medical doctor | >15 | Metropolitan | Public |
| P24 | Female | 46-60 | Medical doctor | 11-15 | Metropolitan | Public |
| P25 | Male | 46-60 | Medical doctor | >15 | Metropolitan | Private |
| P26 | Female | >60 | Medical doctor | >15 | Metropolitan | Public |

*Focus group one ^Focus group two, ~Focus group three. 1. All medical doctors were either clinical geneticists, cancer geneticists or clinical genetics/cancer genetics fellows or trainees. 2. Metropolitan: Within a major capital city (also known as 'urban'). 3. Regional: A city or town that lies outside of a major capital city. 4. Rural: All areas that lie outside of metropolitan or regional areas. 5. Equal mix of public & private work

### 4.6.1 Focusing on the Benefit to the Family

Participants described the importance of the family unit when discussing genomics with people with palliative care needs. They explained the main reason for testing was to elucidate relevant genomic information for relatives, rather than for the patient's clinical benefit.

> *It's a very different consult to our regular consults. It's not so much about that patient, but the family, and a lot of the discussion is probably more so with the family (P1)*

Their experience was that relatives often initiated referrals to the genetics service and were engaged in learning about their risk. Participants built relationships with these families, providing continuity of care before and after the patient's death.

> *But the best thing about it is we will actually […] form relationships with not just that individual, […] you actually form a relationship with a family (P18)*

Some participants described the legal and ethical challenge of family-centred care when health systems preference individual autonomy over familial benefits. They described cases where they could not discuss relevant genomic information with the family, because they did not have consent from the palliative patient.

> *An issue […] is when there's issues with consent to share information with other relatives. So, just say a patient has died, we've got the contact for someone in the family [but] they're not the engaged person, […] there's a niece or someone more distantly related who is more engaged in the process. I think that's a big issue (P2)*

Participants were sensitive to families' vulnerability in an end-of-life context, but most thought they were grateful for the opportunity to discuss the genetic implications of their relative's disease. There was also a sense from participants that a family-centred approach was in-line with the palliative patient's wishes.

> *There are often classic examples of needing to give people the chance to tell their story, […] they're usually really grateful to have the opportunity to reflect on what's going on and what it means for other people in the family (P16)*

Participants had experience working with families who were unaware a DNA sample from their late relative would have helped evaluate their own genetic risk, which meant relatives could only be provided with empirical risk information rather than an individualised

assessment. They noted the family's frustration that genomics had not been discussed while their relative was alive.

> *Parents, who I saw […] for genetic counselling, where there wasn't enough information were really quite cross with their doctors. That it hadn't been presented to them in a way that they understood the information would be helpful to them for the future (P19)*

Participants favoured addressing genomics with families while their relative was still alive so they did not miss the opportunity of obtaining a DNA sample for the family's benefit.

> *So bringing it up […] obviously there's a lot of distress going on, but I think it's […] probably not more distressful than losing the opportunity and then the family not having had that opportunity (P10)*

### 4.6.2 The Discomfort of Addressing Genomics Near End of life

Most participants thought people with palliative care needs wanted to engage with genomics to leave a legacy, make meaning from their illness and for reassurance their family would have access to important information. However, they acknowledged the value of genomic information depended on patients' and families' personal values, which was difficult to assess when they were providing genetic counselling near end of life.

> *The other biggest challenge is […] you don't have rapport and they don't know who you are […] we're just arriving at the very, very last minute […] you feel like an intruder in something that is such a private thing (P15)*

Approaching family members to discuss genomics could be challenging because the conversation was taking place at an emotionally difficult time.

> *In these circumstances, you usually want someone who's a close genetic relative, […] and then you're talking to them at what's usually a really awful time, initiating that conversation at that point is quite difficult (P7)*

However, there was a sense among participants that it was important to recognise and overcome their own feelings of discomfort about having genomic discussions with people with palliative care needs, and their families.

> *We all have to overcome our own discomfort in raising these issues with vulnerable people. And that takes quite a bit of doing (P19)*

Participants found navigating discussions with people with palliative care needs more difficult when important information was missing from their referral, such as prognosis, competency, family dynamics or circumstances.

> *One thing that I have found challenging is […] figuring out who's the appropriate person [to contact] in the family. And sometimes that's not really made clear on the referral (P7)*

Participants remarked that people with palliative care needs do not want to have a detailed discussion about genomic testing, even if they are willing to have a DNA sample collected.

> *Some people are just so unwell that they don't want to […] have an appointment. They're happy to have the test, but they just don't want to go into everything (P13)*

Discussions to obtain consent for genomic testing were managed by reducing or simplifying the information imparted. Participants wanted to convey the most important concepts, while not overburdening patients with irrelevant information. However, they described feeling conflicted about whether they were fulfilling their duty to obtain informed consent.

> *I'm not ever going to make someone listen to me […] if they're not interested. But the thing that makes me uncomfortable is that even if I'm confident that they're on board […] that they're actually signing a piece of paper, which states that they understand things, which I really know that they don't (P12)*

### 4.6.3 "It's Always on the Back-Burner": Challenges to Getting Genomics on the Palliative Care Agenda

#### *4.6.3.1 Burden of proof: instilling the value of genomics in palliative care (subtheme)*

Participants conveyed their sense that genomics is not a priority for palliative care health professionals because of misunderstandings related to the value of genomic information. Some speculated that late referrals to palliative care (for example, from oncology) might affect the palliative care health professionals' ability to identify the need for a genomics discussion. Nonetheless, participants wished genomics were higher on their priority list so discussions could occur as early as possible in the patient's disease trajectory.

*So genetics, […] it never really has a priority. It's always on the back burner, […] it's not as [much a] quantifiable benefit as […] other areas of acute medical practice (P25)*

Participants thought palliative care health professionals might avoid discussions about genomics because they believe another specialist has already addressed it, have concerns about harming patients or do not see genomics as relevant or part of their role.

*You know, I've heard things said […] to families and patients, "Do you really want to spend your last days focusing on whether this might be hereditary or not, instead of just enjoying what time you have left?", which is really disconcerting to hear, because I think both can be done (P24)*

Noting palliative care health professionals' expert communication skills, participants thought basic genomics education, particularly related to the importance of the proband sample and process of DNA storage, could be sufficient to prepare them for genomics discussions.

*I think some of it comes from a misunderstanding that we actually need to test the person with the cancer diagnosis in the first place to get any useful information for the family (P1)*

Participants felt responsible for providing education, but found it difficult to find time to deliver ongoing, concise and targeted education, due to the various cancer types and non-malignant conditions palliative care health professionals' encounter.

*I guess, it's on us [to be] finding the channels to get in there, to let people know that we're here […] There's so many MDT meetings that we could be attending, but, you know, I can't be everywhere at once (P5)*

Participants described the value they could add to conversations about genomics with people with palliative care needs and families. However, some wanted to improve their own palliative care knowledge to ensure they managed these discussions appropriately.

*From a genetic counselling point of view, I would be keen to […] have had more training in the space. I think those, particularly end-of-life conversations, they're quite confronting (P14)*

#### *4.6.3.2 "Individuals can only do so much": finding solutions in the absence of service leadership (subtheme)*

While a few participants described well-integrated services, most reported their services do not recognise the value of genomic information to people with palliative care needs and families, with inadequate funding to develop solutions to existing barriers.

> *They come along and they say, "Yes, we want to help you, but there's no money". So, I think it's that recognition that genetics […] is actually an integral part of all of these streams of medicine (P11)*

Without clear leadership, participants noted that people with palliative care needs (particularly those in private hospitals or from rural areas) were missing the opportunity to address genomics and wondered whether telehealth could help people with palliative care needs overcome these inequities. Some described patients and family members overcoming access barriers by taking the initiative to seek out genomic testing for themselves.

> *I find that often when successful in the private setting, it's because the family is motivated […] and very proactive in making sure the blood is collected […] So that's often how it's circumnavigated (P1)*

Participants valued a multidisciplinary approach to care, but portrayed a lack of collaboration, communication and professional relationships between palliative care and genetic health professionals. They described feeling powerless as individuals in overcoming these barriers.

> *We've been in this building for three and a half years and I still have not worked out ways […] to get those buy-ins and having any kind of meaningful get together and 'here's what you are, here's what we can offer' and so on (P3)*

Participants suggested several strategies to overcome barriers and support integration of genomics into palliative care (Table 5). These included workflow strategies, such as embedding a genetic counsellor within a palliative care team, tools to assess eligibility for genomic testing, such as a red flag checklist for new hospice admissions, and integrating genomic guidance into relevant policy

Table 5. Strategies suggested by participants to support integration of genetics into palliative care

| SUGGESTED STRATEGY | SUPPORTING QUOTE |
|---|---|
| **Workflow strategies** | |
| Provide enough time and opportunity for patients and their families to consider whether genetic testing is right for them | I think it should [be] over multiple bites at the cherry. You know, just introduce the concept or explore the concept and then allow time to pass and answer questions as appropriate (P24) |
| Consider having a specialised or embedded genetic counsellor available for the palliative care service | I think that it's quite important for genetic counsellors to have areas they specialise in, where professionals can call on them for advice. Because I think in a palliative care setting, you almost don't need a physician because the diagnosis has been done (P20) |
| Encourage a palliative care health professional to champion genetics from the inside | You need [… ] somebody in palliative care who thinks it's important […] and it's not just got to be a doctor, it's got to be the nurses. You really need somebody in nursing, who thinks it's important (P26) |
| Encourage genetic and palliative care health professionals to attend the same multidisciplinary team (MDT) meetings | I think MDT meetings are the easiest way to integrate us in. Because I don't think every department has the resources to have a genetic counsellor on staff, but the MDTs are an excellent opportunity to […] build the contacts to be able to have those discussions with each other (P5) |
| Liaise directly with palliative care health professionals who are involved in the patient's care when a referral is received | Once I have spoken to the nurses or the physicians who are actually involved with that patient's palliative care planning, they have been extremely helpful […] in terms of organizing and carrying out a more satisfactory consultation for this family (P4) |
| **Strategies and tools to assess eligibility for genetic & genomic testing** | |
| Screen patients on admission to palliative care or hospice with a checklist, family history questionnaire, red flag document or digital application. | I would have thought some sort of triaged model with red flags, […] check around any questions about family risks, and maybe you'd even […] tailor it to the fact that people have children. That's more likely to be at the front of their mind then if they don't (P23) |
| Provide written material about genetics to patients and their families | What I would like to see is[…] a sort of pack that both for […] doctors and for families around when family members are dying, that kind of almost raises some of those questions by default and then families can pick and choose (P17) |

| | |
|---|---|
| Ask patient and their family if they have any unmet need related to genetics | Maybe just checking with the patient […] “So have you been referred to genetics?”, “Has someone raised this with you that it could be hereditary?” [or] “OK, I can potentially be that liaison person, check in with genetics”. Because some people do forget that they've had anything through us (P13) |
| Consider reoffering the opportunity to people with palliative care needs and families to discuss genetics. | In that case […] we'd seen her previously and […] she either declined testing or hadn't gotten around to having the blood taken and then realized the clock was ticking. And so desperately wanted to have the blood taken (P5) |
| **Service improvement strategies** | |
| Generate leadership by reflecting the value of genetics in relevant policy and/or guidelines | I think it would help if there was a national strategy on the integration of genomics into palliative care. […] I think it is quite important that you do have some sort of national leadership (P25) |
| Use telehealth services for patients receiving palliative care | One of the biggest barriers is that they're too unwell, or that it's just adding a burden to their appointments, so being able to stay at home […] in general I would say it's probably been really positive for patients in general, but probably palliative care in particular (P13) |
| Improve capability of electronic medical records to share information between services | So how do they get access to medical records that sometimes might span over years? […] there's a suggestion: electronic records that actually talk to each other. […] You can take a considerable amount of time to wade through health records to see if genetics has already been covered (P24) |

## 4.7 DISCUSSION

We described three themes that illustrate the perceived clinical and psychological benefits of genomic information to families: the discomfort genetic health professionals can experience when providing genetic counselling near end of life, health professional and organisational level barriers preventing integration of genomics into palliative care and potential strategies to overcome these.

Genetic health professionals emphasize the familial benefit, as opposed to the individual benefits, of genomic information in the palliative care context.(100, 227) The benefits described reflected broad definitions of utility, encompassing potential clinical and personal benefits (for example, the genomic test is of psychological value to the patient and family).(27, 228) Discussion of the familial benefits overlapped with descriptions of family-centred care and relational approaches to autonomy in the palliative care literature, as a philosophy that centres the individual within their social system.(81, 99, 229, 230) However, frameworks to operationalise family-centred approaches to care in Western health systems are often missing, putting health professionals in the difficult position of executing individualistic processes, despite knowing that families are integral to patient care.(231) The shared family-focused philosophies of palliative care and clinical genetics could be harnessed to design a family-centred intervention to support integration of genomics into palliative care.

Understanding a patient and family's goals of genetic counselling or testing is key to building a trusting foundation to support shared decision-making.(229) Previous research describes deteriorating patient health, heightened emotions and limited time as barriers to discussing genomics, but our findings go further by suggesting these factors affect genetic health professionals' ability to build relationships with and elicit the patient's and family's goals of genetic counselling and testing.(180) Families are often dealing with complex issues and making numerous decisions in the end-of-life stage, so despite the benefits of genomic information, health professionals can be uncomfortable broaching difficult discussions.(128, 232) Given the nature of interactions genetic health professionals have with grief and loss, additional training about palliative care may help to manage their discomfort.(184).

Furthermore, we heard genetic health professionals adapt their approach to obtaining informed consent for genomic testing by reducing or simplifying information they convey to terminally ill patients. Patients at end of life may have decreased capacity to engage in complex discussions due to illness or delirium.(98) In addition, genetic health professionals may be considering and responding to the contextual 'function' of consent.(233) For people with palliative care needs, the primary aim may be to establish any objection to genomic testing rather than focus on the individual clinical implications of the result.(101) To tailor appropriate approaches to genetic counselling, discussions about genomic testing and the associated medico-legal processes, further enquiries into patients' and family members' preferences for delivery, timing and content of discussions about genomics is urgently needed.(96)

Akin to previous literature, our participants advocated for genomics to be introduced earlier in the patient's disease course, rather than at end of life.(103) However, palliative care health professionals appear to be subject to well-known barriers to integration of genomics, such as low knowledge and confidence.(127, 133, 138, 218) Our participants echoed a general willingness to assist with improving palliative care health professionals' genomics knowledge, but this was contingent upon time and resource constraints.(234) Our findings suggest genetic health professionals (in)ability to implement strategies to overcome structural barriers (for example, embedding a genetic counsellor within a palliative care team) was affected by a lack of funding and low awareness of genetic services at the organisational level.(67) Demonstrating the economic value of genomic testing for the benefit of relatives to organisations is complex.(26) While cascade testing rates are typically used to assess familial value, a more nuanced analysis combining health economics with ethical, legal and social implications may better illustrate the significance of genomic information, improve funding and support health professionals to implement strategies to support integration of genomics into palliative care.(235)

### 4.7.1 Strengths and Limitations

This study combined a theory-informed instrument with an inductive approach to data analysis, allowing us to benefit from existing knowledge about genomic integration, while developing data-driven themes relevant to the palliative care context. A qualitative approach enabled exploration of participants' views and experiences in this understudied area; however, generalisability is limited. Participants with strong views may have self-selected to participate,

skewing the data with positive attitudes towards palliative-genomic integration, while negative or neutral views may not be represented. Most participants had less than five years of experience, were female, and working in public and metropolitan/urban settings. While some of these characteristics represent a large portion of the genomic workforce in Australasia, views from diverse groups may not be captured here.(52)

### 4.7.2 Practice and Research Implications

Genetic health professionals and policy stakeholders can use these findings to increase awareness of the challenges genetic health professionals face when discussing genomics with people with palliative care needs and families. Generalisability of our findings would improve if these themes were tested in a larger, quantitative study. Interventions to support integration of genomics into palliative care could harness the shared family-centred philosophies of clinical genetics and palliative care. Research with people with palliative care needs and their families is required to understand their needs regarding genomic information.

## 4.8 CONCLUSION

We identified three main themes that illustrate the centrality of the family when providing genetic counselling to people with palliative care needs and their families, the discomfort of managing genomic issues near end of life, and highlight the practice barriers that are unlikely to be overcome without improved leadership to increase funding and implement targeted strategies. Cross-boundary collaboration between palliative care and genetics could focus on the shared value of family-centred care, while further research should elucidate the economic and personal value of genomic information to families to demonstrate the benefit of investing in the integration of genomics into palliative care.

## 4.9 EXTENDED RESULTS

As detailed in the preamble, I present here an additional theme generated from the genetic health professional data set called “Store now, talk later: a DNA storage approach at end of life”.

In situations in which genomics is addressed close to the patient’s end of life, participants articulated that storing DNA, rather than facilitating genomic testing, was their preferred approach.

> *I always found a useful default position with a family was to say to them “Look, we don't need to talk about it now,* [but] *we do need to get some samples. We don't have to do anything with them except store them. And it can be discussed later.” (P19)*

Offering DNA storage was thought to be consistent with a family-centred approach to care at the end of life.

> *I think there are situations where we really could just be having a quick DNA storage conversation and then meeting with the family in a few months’ time. Particularly if it’s not going to change anything for the person in palliative care. So, at that point, it’s really thinking about family. What this might mean for them in terms of their screening. (P14)*

Though nearing their end of life, there was a sense from participants that most people were interested in providing a DNA sample for their family to use in the future.

> *I think sometimes if it's just for their family, they think that their task is to provide the DNA so the test can be done and, […] well they're doing it to share it with their family. So, they're very willing for the information to be shared (P12)*

Participants thought it was not in the family’s best interest to be introduced to the genetics team for the first time when their relative was close to the end of life.

> *In those* [end-of-life] *circumstances, we often get a big push from treating teams to come and see the families and talk about the genetics. And my personal view is that's not the right time for most families, if any, to be seeing people that they've never met before…to be talking about something that's not acutely relevant and that they're not even focusing on (P24)*

Participants explained that offering DNA storage reduces the amount of information that needs to be conveyed to the person with palliative care needs.

> *So having that opportunity to actually talk through* [DNA storage] *I think was somewhat useful, but his [the palliative patient's] care factor was probably minimal at that point. So, I think that there's still a role for a very truncated* [discussion] *as opposed to following our normal agenda (P3)*

Participants saw no need for family members to engage in complex genomic discussions in the end-of-life situation. DNA storage was thought to offer family members the opportunity to participate in discussions about genomic testing at a more appropriate time.

> *There's probably more of a push about at least storing DNA so that we can have that discussion down the track when some of the stressors have kind of, unfortunately, […] gone away. So, people have the time to really take a deep breath and make a bit more of an informed decision about whether to have testing (P2)*

Participants were happy to be guided by the family's needs and timing of the follow up.

> *if it's really down to those last hours or days, we might say, "Look, let's store some DNA and we'll get the contact details for your children or your spouse or whoever it might be. And we'll have a chat with them in another month or so and just touch base initially and go with their timeframe." (P3)*

One participant worried that holding brief conversations with family members about DNA storage could be misunderstood and lead to inaccurate assumptions about their disease risk.

> *I think they may assume more* [testing has been done] *as they were "going to take some blood from genetic testing from mum and we never heard anything. So, it must be fine." I think that nuanced conversation […] may get lost, maybe misunderstood or misconstrued. That's what worries me (P23)*

Participants wondered if practical barriers could be overcome if treating doctors had the capability to facilitate DNA storage.

> *This could have been a much more straightforward circumstance if that doctor knew that they could arrange DNA storage.* […] *I didn't have a* [phone] *number for her. I'd never spoken to her before. So, it was just all*

*these quite practical type things that I think could be fixed. It's already a difficult conversation to have anyway (P2)*

To build capability, participants thought palliative care health professionals would require education about DNA storage.

*So, education I think is hugely important and* […] *giving them easy pathways to* […] *the patient who just says, "Yes, I want to store my DNA, but I really don't want to think about it". Just so they've just got the forms handy, and they can have a minimal dialogue with DNA stored, and it can be dealt with down the track (P22)*

## 4.10 EXTENDED DISCUSSION

The findings here suggest that in most end-of-life situations, genetic health professionals prefer to facilitate DNA storage over ordering genomic testing. One reported benefit of DNA storage was reducing the burden of information on dying people and their family members. As people approach the end of life, they may have reduced or fluctuating cognition and be unable to engage in an informed consent discussion for genomic testing.(236) For family members, the high emotions associated with supporting a dying family member can impede comprehension and decision-making capacity.(96)

Compared with genomic testing, gaining consent for DNA storage can be a relatively simple discussion conducted with people (if conscious and lucid) or family members.(101) Many of the discussion points on genomic testing consent forms, such as insurance implications of genomic testing, can be omitted as no testing is being requested.(101) Conveying information about the possible results and clinical implications of genomic testing can be delayed until the family decides (after their relative has died) what testing they wish to pursue, if any. This is congruent with the perceptions of relatives, who did not perceive that palliative care is the appropriate time to be worrying about their own health.(96)

Some participants' preference was for palliative care or treating health professionals to facilitate DNA storage, although they wanted to retain responsibility for the follow-up of family members to discuss genomic testing. One reason may be because of their view that obtaining consent for DNA storage is a simple discussion. Genetic health professionals, however, may lack understanding of palliative care health professionals' capacity to engage in discussions about DNA storage. Palliative care health professionals' awareness of DNA storage appears to

be low.(133) Many do not feel confident applying their knowledge or skills to facilitate DNA storage or even be aware of this as a possibility.(133) At least initially, genetic health professionals may need to collaborate with palliative care health professionals to support the latter's development.

The findings suggest that genetic health professionals do not want to be introduced to families for the first time when people are close to the end of life. In the cancer setting, individuals prefer to engage in a genomics discussion with their treating health professional.(237) The palliative care realm does not contain similar data, although relatives of dying people have reported that palliative care health professionals are not equipped to answer their questions about genomics.(96) Patients have expressed their concerns about the heritability of their disease, particularly in relation to the impact this may have for future generations.(92, 183) A genetic health professional, in collaboration with the palliative care team, may be needed to meet the informational needs of individuals and family members and to reassure them that their concerns are being actioned, and that relatives will be assessed and managed appropriately. However, further exploration of individuals' and families' perspectives would be valuable. Establishing the roles and responsibilities of genetic and palliative care health professionals, alongside the wishes of people with palliative care needs and their relatives, is needed before implementing a DNA storage approach to care.

## 4.11 EXTENDED CONCLUSION

Offering DNA storage, as opposed to genomic testing, at the end of life has the potential to overcome some of the barriers to genomic testing for the clinical benefit of family members. Storing DNA could simplify the consent and procedures associated with integrating genomics into palliative care for people near the end of life. However, further work is needed to explore the responsibilities of genetic and palliative care health professionals, and whether this approach meets the needs of dying people and their families.

## 4.12 MANUSCRIPT 3. QUALITATIVE EXPLORATION WITH PALLIATIVE CARE HEALTH PROFESSIONALS

This paper was published in *Supportive Care in Cancer* in 2021 (Scimago rating Q2 in the 'oncology' category, 2021 impact factor 3.359).(238) Reprinted by permission from Springer-Verlag GmbH: Springer Nature [*Supportive Care in Cancer*].(218) As per publisher requirements, this is the final accepted version rather than the published version (license to reproduce is available in appendix C12). Minor edits have been made, including changing 'genetic' to 'genomic', 'clinician(s)' to 'health professional(s)' (except in the manuscript title), and updating table headings and page numbers for congruency across the thesis. Please note that, because of the word limit for this journal, representative quotes were placed in tables rather than embedded into the main text.

Reference: White S, Phillips J, Turbitt E, Jacobs C. Views and experiences of palliative care clinicians in addressing genetics with individuals and families: a qualitative study. Support Care Cancer. 2022;30(2):1615-24.

## 4.13 ABSTRACT

*Purpose:* A proportion of people with palliative care needs unknowingly have a genetic predisposition to their disease, placing relatives at increased risk. As end of life nears, the opportunity to address genomics for the benefit of their family narrows. Health professionals face numerous barriers addressing genomic issues, but there is limited evidence from the palliative care health professional perspective. Our aims are to 1. Explore the views and experiences of palliative care health professionals in addressing genomics with patients and their families, and 2. Generate suggested strategies that support integration of genomics into palliative care.

*Methods*: An interpretive descriptive qualitative study using semi-structured interviews with palliative care doctors and nurses (*N*=14).

*Results:* Three themes were identified: 1. Harms and benefits of raising genomics: A delicate balancing act, 2. Navigating genomic responsibility within the scope of palliative care, 3. Overcoming practice barriers: A multipronged approach. Participants described balancing the benefits of addressing genomics in palliative care against potential harms. Responsibility to address genomic issues depends on perceptions of relevance and the scope of palliative care. Suggestions to overcome practice barriers included building genomic-palliative care

relationships, multi-layered genomics education, developing clinical resources and increasing organisational support.

*Conclusions:* Integrating aspects of genomics is feasible but must be balanced against potential harms and benefits. Palliative care health professionals were uncertain about their responsibility to navigate these complex issues to address genomics. There are opportunities to overcome barriers and tailor support to ensure people nearing end of life have a chance to address genomic issues for the benefit of their families.

## 4.14 INTRODUCTION

Rapid advances in our understanding of the role of genomics in common and rare cancers and non-malignant disease means an increasing number of people with palliative care needs may require a discussion about the genetic contribution to their illness.(24) When genomics is not addressed, family members may miss important genomic information to make medical, social and reproductive decisions.(14, 23, 25) Health professionals (doctors and nurses), including those in palliative care, are being asked to integrate complex genomic knowledge into practice and address these issues with people with palliative care needs.(112, 239) This could include: identifying people with a genetic condition, having discussions about genomics, obtaining family history information, conducting a genetic risk assessment, organising DNA storage or testing, conveying genomic test results or making referrals to clinical genetics services. Interventions to support health professionals to integrate genomics into practice are available, but research is lacking in the palliative care context.(180, 240)

Despite the push for genomics to be introduced early in a person's diagnosis (particularly in oncology, where genomic testing guides targeted treatments), up to 40% of eligible people are not referred for genetic counselling or testing.(93, 188) Oncology health professionals have limited time, knowledge and resources to identify everyone who should be offered genomic testing.(59, 241) Some people with cancer bypass oncology services because of a late diagnosis or refusal of conventional treatment, while others initially decline or delay a discussion about genomics.(101, 242) For people with non-malignant conditions (e.g. neuro- or cardio-genetic conditions), pathways to genetic counselling or testing may be even less clear.(147, 157)

Although there are potential psychological benefits of genomic information for people with palliative care needs,(183) the clinical value is in assessing the genetic risk of relatives. (191) Life-limiting genetic conditions (such as familial cancer and neurodegenerative diseases) can

be inherited in an autosomal dominant way, meaning first-degree relatives (i.e., children, siblings, parents) have a 50% chance of having the same genetic predisposition. Identifying a genetic mutation in the affected person offers relatives an opportunity for targeted genetic testing to see whether they have inherited the same predisposition.(19) Relatives at increased risk may choose to engage in disease screening or risk-reducing interventions, or use genomic information to plan financial, occupational, social and reproductive decisions.(14, 16, 23, 25) If end of life is the final opportunity to identify people dying with a genetic condition, it is important palliative care health professionals feel capable and supported to appropriately integrate genomics into their practice for the benefit of family members.

Barriers and facilitators related to opportunity, capability and motivation impact health professionals' abilities to appropriately integrate genomics into practice.(214) Evidence from the USA, UK and Canada suggest palliative care health professionals consider their genomics knowledge, skills and confidence to be low, believe genomics should have been addressed during active treatment and have concerns about genomic information causing psychological distress.(103, 113, 128, 133, 138) While international evidence is a helpful guide to predict what the issues may be locally, generating evidence about the barriers and facilitators specific to the cultural, geographical and socio-political environment in Australasia is a critical step towards developing an evidence-based intervention to support palliative care health professionals to integrate genomics into practice.(114)

Our aims, therefore, are to explore the views and experiences of Australasian palliative care health professionals towards addressing genomics with patients, and their families; and to generate their suggested strategies to improve integration of genomics into palliative care.

## 4.15 MATERIALS AND METHODS

### 4.15.1 Theoretical Perspectives and Design

This qualitative study, underpinned by an interpretive descriptive perspective, used semi-structured interviews to provide a straightforward, yet in-depth description of genomics in palliative care.(202) Pragmatism led us to an inductive approach to capture varied realities of our participants and ensure findings were grounded in data.(219 p9-10) Using an implementation science approach, we explored the boundaries of this topic in the Australasian setting to produce contextually useful results to generate evidence for an intervention.(196) The study protocol was registered at https://osf.io/h4gt9/.

### 4.15.2 Participants and Recruitment

A purposive sample of Australian and New Zealand palliative care health professionals were recruited via professional organisations and social media. Several organisations circulated invitations twice to their members (appendix C9). Email invitations were sent to known contacts of the research team and participants were asked to snowball the invitation to supplement low response rates. We were unable to record the number of non-responders or reasons for not responding.

### 4.15.3 Data Instrument and Collection

The semi-structured interview guide was informed by a systematic review,(214) and the World Health Organisation Innovative care for chronic conditions framework.(67) Minor modifications to the interview guide introduction (e.g. explaining 'integrating/addressing genetics') and question order were made following two pilot interviews. Participants were asked open questions about prior experiences and challenges of addressing genomics, perceptions of the palliative care and health organisation role and ways to overcome perceived barriers. Prompts were used if participants required clarification (see appendix C13).

We offered both focus groups and interviews, but all participants preferred an individual interview. Interviews were held on Zoom, audio-recorded and transcribed verbatim.(220) C.J conducted one interview (participant had a pre-existing relationship with S.W), while S.W conducted the remainder. Transcripts were de-identified, returned to participants to check them for accuracy, and coded in NVivo V12. Data saturation was achieved when no new information was elicited from participants.(223) A reflexive diary was maintained throughout the research process to encourage reflection and consider the impact of internal biases and preconceptions on the data.(243) Participants provided demographic information via an online survey to give context to their responses

### 4.15.4 Data Analysis

To inductively develop themes, we utilised the six non-linear steps of thematic analysis.(225) S.W led the analysis. E.T and C.J each co-coded two transcripts. Open coding was applied to sentences and paragraphs. Codes were revised (collapsed, expanded, renamed) over several iterations before being grouped into initial themes. Themes were further developed through discussion of our interpretations and reflections in weekly team meetings.

### 4.15.5 Research Team

S.W is a PhD candidate, certified genetic counsellor and registered nurse with training in qualitative research methodology. C.J is a female senior lecturer within the Master of Genetic Counselling program at the University of Technology Sydney (UTS), registered genetic counsellor and registered nurse with qualitative research experience. J.P is a professor of palliative nursing with extensive mixed methods research and PhD supervision experience. E.T is a lecturer in the Master of Genetic Counselling program at UTS with qualitative research experience.

### 4.15.6 Ethics

The UTS Research Ethics Office (ETH20-5046) approved the study.

## 4.16 RESULTS

Fourteen one-on-one interviews were undertaken with palliative care doctors (trainees and physicians, *n*=10) and nurses (*n*=4; Table 6). Most participants were female (12/14, 86%) and worked in a metropolitan area (9/14, 64%). Interviews were conducted between August 2020 and February 2021, lasting 31 to 56 minutes (averaging 40 minutes). Participants predominantly drew upon their experiences, however a small proportion of responses were based on hypothetical situations.

Three themes were identified: 1. Harms and benefits of raising genomics: A delicate balancing act, 2. Navigating genomic responsibility within the scope of palliative care (including two subthemes) and 3. Overcoming practice barriers: A multipronged approach.

Table 6. Demographic characteristics of participants (*N*=14).

| Participant | Sex | Age range | Discipline | Years of PC experience | Work location[1,2,3] | Work sector |
|---|---|---|---|---|---|---|
| P1 | Female | 46-60 | Doctor | >15 | Regional | Public |
| P2 | Female | 31-45 | Doctor | 6-10 | Metropolitan | Public |
| P3 | Male | 31-45 | Doctor | 6-10 | Metropolitan | Public |
| P4 | Female | 46-60 | Doctor | >15 | Regional | Public |
| P5 | Female | 46-60 | Doctor | >15 | Regional | Public |
| P6 | Female | 46-60 | Doctor | 6-10 | Regional | Public |
| P7 | Female | 31-45 | Doctor | 6-10 | Metropolitan | Public |
| P8 | Female | 46-60 | Doctor | >15 | Metropolitan | Public |
| P9 | Male | 31-45 | Doctor | 0-5 | Metropolitan | Public |
| P10 | Female | 46-60 | Doctor | >15 | Metropolitan | Public |
| P11 | Female | 46-60 | Nurse | >15 | Metropolitan | Public |
| P12 | Female | 18-30 | Nurse | 6-10 | Metropolitan | Private |
| P13 | Female | >60 | Nurse | >15 | Rural | Public |
| P14 | Female | 46-60 | Nurse | >15 | Metropolitan | Public/ private[4] |

NB: 1. Metropolitan: Within a major capital city. 2. Regional: A city or town that lies outside of a major capital city. 3. Rural: All areas that lie outside of metropolitan or regional areas. 4. Equal mix of public & private work. PC: Palliative care.

### 4.16.1 Harms and Benefits of Raising Genomics: A Delicate Balancing Act

Participants conveyed their appreciation for the complexity of genomics, particularly in the palliative care context. Deciding whether to raise genomics requires consideration of a number of psychological, medical and ethical factors, with most participants wary genomic information may cause psychological harm (Table 7; quote 1.1). Participants were concerned that raising genomics may result in feelings of guilt, blame and uncertainty for individuals and families (quote 1.2). For families with complex relationships or conflicting opinions about the value of genomic information, knowing how, when or if to address genomics was particularly difficult for participants (quote 1.3).

Some participants reported that discussions about genomics could harm the therapeutic relationship between the health professional and their patient (quote 1.4). Others did not think of genomics as harmful, explaining that positive framing helps individuals understand the potential benefits of genomic information for the family (quote 1.5).

Some participants reported that people often have altruistic motivations to engage in a discussion about genomics and are relieved they can offer genomic information to their relatives (quote 1.6). A few reported initiating conversations about genomics with relatives and found that questions about genomic risk were often already on the relative's mind. Most believed relatives had a right to genomic information that would affect future health decisions. Although these participants reported that these conversations could be difficult, they found families generally appreciated the information (quote 1.7).

Participants described their ethical responsibilities when deciding whether to address genomics, including an obligation to respect a person's autonomy in their right to accept or decline a discussion about genomics (quote 1.8). Ethical obligations extended to obtaining consent to engage in discussions about genomics with relatives or collecting biological samples for genomic testing. The cognition of the person with palliative needs played a major role in assessing whether consent could or should be obtained, and from whom. Most participants agreed that once cognition reduced, the legal medical proxy could provide consent for genomic testing or DNA storage (quote 1.9).

Table 7. Representative quotes for theme 1. Harms and benefits of raising genomics: a delicate balancing act.

| **Theme component** | **Representative quote** | **Quote #** |
|---|---|---|
| Deciding to raise genomics is complex and may cause harm | "There's a very intricate balance because […] a lot of these patients, they worry about being a burden to their family members. We're trying to give them a sense of dignity […] so we don't necessarily even say that or approach that topic [of genomics]." (P3) | 1.1 |
| Discussing genomics can cause negative feelings | "One other thing I think is really important is the profound guilt that people have, if they've unknowingly given something to their children. I don't think that can be understated […] it can be a real point of concern in terms of someone's spiritual health. Like […] they've in some way, put a hex on their family." (P6) | 1.2 |
| Families can be complex | "The thing is with families […] some want to know, and some don't want to know and then where do you sit?" (P13) | 1.3 |
| Discussing genomics can harm the therapeutic relationship | "Sometimes you can lose your therapeutic alliance by pushing those things [discussions about genomics]." (P6) | 1.4 |
| Positive framing can influence the way genomics is perceived | "I think if you frame the whole thing in terms of being a gift to the family, that you could definitely help patients see it as a good thing to do, rather than something to be feared." (P10) | 1.5 |
| Some people have altruistic motivations to discuss genomics | "Certainly a lot of patients […] they feel they can't give anything to their family. And this, they can reduce the risk of cancers going forward and help inform your family about what to do next. Usually there's a big sigh of relief thereafter." (P9) | 1.6 |
| Most families do want to know genomic information, even if it's difficult to talk about | "You have to start to […] gather some strategies for some of those discussions with families, because often families do have a whole bunch of stuff going on at the same time and families, in my experience, almost universally do want to know about genetic conditions. Even if it's painful, they still want to know about it." (P2) | 1.7 |
| Health professionals respect the autonomy of individuals to accept or decline a genomics discussion | "So, you might introduce the idea that this could be helpful for the family. But basically if they don't want to do it, that's entirely their right to decline." (P5) | 1.8 |
| Consent for genomic testing or storage can be obtained from a legal proxy | "The man had dementia, I think we had to have the consent of a EPOA [Executive Power of Attorney] to take the blood. And, and we did that. And I mean, he was literally dying." (P13) | 1.9 |

### 4.16.2 Navigating Genomic Responsibility Within the Scope of Palliative Care

Participants' assessment of their responsibility to address genomics was described relative to their views and experiences of the scope of palliative care. Participants explored the relevancy of genomics and how genomics aligns within the role and goals of palliative care Most believed that integrating genomics into the evolving palliative care role was feasible, describing aspects of genomics they felt were appropriate to integrate and the boundaries of their responsibility (Table 8; quote 2.1).

#### *4.16.2.1 (Ir)Relevancy of Genomics to Palliative Care*

Reports varied among participants about how frequently their work interfaced with genomics. Some reported regular discussions about genomics, but most said this occurs infrequently. Nonetheless, many recounted multiple conversations about the role of genes in disease development, caring for people with genetic conditions or helping to organise genomic investigations (quote 2.2). Perceived relevancy of genomics to palliative care also varied among participants. Those with limited genomics exposure conveyed the irrelevancy of genomics to most patients and their practice. Participants who perceived genomics as relevant often described a formative clinical or personal experience that changed their perception that genomics related to their goals as palliative care health professionals (quote 2.3). Some participants explained that they expected genomics to have already been discussed prior to the person requiring palliative care (quote 2.4).

Engagement with ongoing professional education about genomics was not a priority for participants who described genomics as irrelevant. Instead, they selected other educational topics when choosing professional development opportunities (quote 2.5). Participants who reported active engagement in ongoing genomics education did so because they felt genomics was relevant to their practice or they had a special interest in the genomics field (quote 2.6).

#### *4.16.2.2 Aligning Genomics Within the Role and Goals of Palliative Care*

Participants explored their responsibility to address genomics within the role and goals of palliative care. Using the World Health Organisation definition of palliative care, some explained that addressing genomics was contrary to the 'relief of suffering', while others thought addressing genomic risk was part of an 'impeccable assessment' (Table 8; quotes 2.7 and 2.8).

Universally, individualised discussions about genomics were considered integral to the person and family centred approach of palliative care (quote 2.9). Participants explained that their ability to engage in difficult conversations and to provide holistic, family centred care were transferrable skills they could use to have conversations about genomics. However, most felt they could not apply these skills without an increase in their genomic knowledge (quote 2.10).

While many participants reported being involved in discussions about genomics, most described their preference to play a supportive role rather than be the primary drivers of these conversations (quote 2.11). However, participants described a stronger responsibility to address genomics if they suspected the individual had not yet had the opportunity. Several groups were identified as more likely to have genomics missed prior to palliative care: people who did not receive oncology care because of a late diagnosis or poor prognosis, those who decline conventional treatment, and people from low socioeconomic or rural areas (quote 2.12). A small number of participants reported a duty to check genomics had been addressed regardless because, in their experience, genomics was often not discussed prior to palliative care. (quote 2.13)

Participants explored their responsibility to address the genomic concerns of family members. Most explained they were unable to engage in a detailed genetics assessment because the relative is not their patient. Instead, participants directed relatives to their own doctors for advice.

Table 8. Representative quotes for theme 2. Navigating genomic responsibility within the scope of palliative care.

| Theme component | Representative quote | Quote # |
|---|---|---|
| Integrating genomics into palliative care is feasible | "[Palliative care] is one of the younger specialties around and the way I see it, we're still finding our feet as to who we are and what we do […] so I think it's still an evolving field and it certainly is still on the cards to add this [addressing genomics] on among the multiple things that are being done in palliative care." (P9) | 2.1 |
| **Subtheme: (Ir)Relevancy of genomics to palliative care** | | |
| The frequency of genomics discussions in palliative care varied | "Oh look, certainly not once a month [does genomics come up]. But a few times […] a year […] I mean, in terms of saying […] 'Look, it's nothing you ate, it's nothing you did […] it's not your fault. You just got cancer, it's bad luck' kind of thing […] to a certain extent […] we do talk about genetics [in] that setting." (P8) | 2.2 |
| The perceived relevance of genomics to palliative care varied among participants | "Cause I think, if not the first time ever, I think 'Genetic[s], how is it even relevant to palliative care?' We talked about it and go, 'Oh yeah, indeed it is relevant!' It's this idea of, 'Is it even relevant?' And needing to build that bridge in the mind of clinicians, that actually it is relevant to achieve the goal of care." (P3) | 2.3 |
| Genomics should have been addressed prior to palliative care | "Honestly, I really expect the medical oncologist to do that […] I expect that the medical oncologist has seen a person, diagnosed them with cancer and part of that diagnostic workup is their genetic predisposition and […] whether or not the family then needs to have genetic testing." (P7) | 2.4 |
| Health professionals will not engage with ongoing genomics education if they don't deem it relevant to care | "I guess […] not a lot of continuing education on genetics because it doesn't seem to be relevant to most people when they choose what their medical education for the year is. They aren't keeping up in genetics, and I certainly don't blame them […] they don't seem very connected, unless you're in paediatrics." (P4) | 2.5 |
| Active genomics education engagement arose from a special interest | "I suppose in terms of ongoing training, because I'm fascinated by it, I look at it and […] I keep abreast of it, but I wonder if other people kind of know how this area is evolving." (P6) | 2.6 |
| **Subtheme: Aligning genomics within the role and goals of palliative care** | | |
| Some felt genomics aligned with the goals of palliative care, while others did not | "I'm not convinced we need to take this up, or spend too much attention on it, because we've got to spend attention on getting right what it is we're meant to be doing, which is prevention, relief of suffering, to help people live well 'til they die. That's what my focus is." (P14) | 2.7 |
| | "If you apply the WHO type of definition of palliative care, and it's around 'impeccable assessment', I think that actually asking the patient or their family if there's any unmet need […] I have in practice come across families who have been really worried about inheritance but haven't had that opportunity to ask yet, so I feel it's important that […] I actually incorporate that into routine questioning." (P1) | 2.8 |

| | | |
|---|---|---|
| Discussions about genomics should be individualised | “The piece that palliative care will play in those discussions […] could be really varied and I think probably need to be individualised to the family.” (P2) | 2.9 |
| Palliative care health professionals have transferrable skills to integrate genomics, but want more knowledge | “We very much do pride the discipline on being holistic and including family members. And if we're letting people down by not addressing this, that there's undiagnosed, unaddressed, psychospiritual stress in family members because palliative care doctors don't know how to have conversations about genetic testing or who to refer […] then I think that would be a great enabler for us to up-skill.” (P7) | 2.10 |
| Participants want palliative care to support genomics discussions, not drive them | “I kind of see us more of playing […] that linking up role […] I think that our role is probably more in that sort of secondary supportive piece or in flagging families interested with the [genetic] service.” (P2) | 2.11 |
| Participants were concerned that some people miss out on a genomics discussion prior to palliative care | “I think there is a group, and there's a lot of patients here in this area where […] they’re very late presenters. They might get a one-off appointment with an oncologist who just says ‘No treatment’. And my concern here is trying to figure out, who does the genetic stuff?” (P13) | 2.12 |
| Health professionals should always check if genomics has been addressed | “I think that the responsibility of me is to ask […] if there’s anything suspicious, I’ll say ‘Have you ever had an opportunity to talk about the genetics of this or whether this is inherited?’, ‘Are you worried about that kind of thing?” And it’s amazing, like anecdotally, I can’t give you a figure, it’s amazing how many patients have said to me, ‘You’re the first person to ask me that question’. Even oncology patients.” (P1) | 2.13 |
| Addressing the genomic concerns of the relatives is not in the health professional’s scope | “So you might just tease it out a little bit, but essentially […] because that relative is not our patient, we often explain to them about the familial cancer clinic and then ask them to go to their GP to get a referral.” (P5) | 2.14 |

### 4.16.3 Overcoming Practice Barriers: A Multipronged Approach

Participants reported several barriers preventing them building capability in genomics, but explained the first step towards overcoming these barriers is to demonstrate an unmet need for genomics in palliative care (Table 9; quote 3.1).

Participants described the impact of professional relationships with genetics health professionals, or lack thereof, on their ability to access genetics services (quote 3.2). A few described their relationship with the oncologist as a barrier to addressing genomic issues (quote 3.3). Participants suggested co-locating palliative care and genetics departments to build better relationships, improve communication, access timely advice and encourage a multidisciplinary approach to care (quote 3.4).

Participants reported having low genomics knowledge. For some, their level of confidence depended on whether they were assessing genetic risk for a malignant or non-malignant disease. However, most thought practical knowledge, including how to access genetic services within their organisation, was just as important as theoretical genomic concepts (quote 3.5). Participants had a variety of suggestions to improve their genomics knowledge, including a genomics module in palliative care trainee curricula, a compulsory oncology term during palliative medicine physician training, lectures delivered by genetics health professionals and inclusion of genomic research in palliative care conferences and journals (quote 3.6).

Participants felt healthcare organisations could do more to support integration of genomics into palliative care through funding, education and raising awareness. A lack of research into the feasibility and acceptability of genomics in palliative care, particularly from the perspective of people with palliative care needs and their families, was identified as an important gap (quote 3.7). Participants suggested a simple, practical, accessible, web-based genomics guideline for palliative care, as well as consumer-friendly information about genomics to provide to people with palliative care needs (quote 3.8).

Table 9. Representative quotes for theme 3. Overcoming practice barriers: a multipronged approach

| Theme component | Representative quote | Quote # |
|---|---|---|
| Building genomics capability in palliative care first requires demonstrating an unmet need | "So that's the biggest barrier, is a lack of integration into comprehensive care. But we can't get that until we demonstrate unmet need." (P1) | 3.1 |
| Professional relationships impact health professionals' ability to address genomic issues | "On our wards, the clinicians who know how to contact the genetics teams […] they tend to have positive outcomes and you know, get that blood test done […] whereas those who don't know how to contact the genetics team […] those ones just probably put in [the] too hard basket and move on to the next patient instead of thinking twice about it." (P9) | 3.2 |
| Professional relationships between palliative care and oncology health professionals can impact willingness to integrate genomics | "I would personally feel uncomfortable recommending that a patient go and get genetic testing. I'd feel like I'd be stepping on the medical oncologist toes, or I'd probably call the medical oncologist and ask them, 'Should this person be having genetic testing based on their relative's diagnosis?'" (P7) | 3.3 |
| Co-locating genetics and palliative care departments could improve integration | "There's something about the physical space […] about just being able to pop across and be like, 'Hey, there's a patient with this, that, and the other, would I refer them to you? Is that someone that you'd want to see?'" (P7) | 3.4 |
| Participants feel their genomics knowledge is low | "I think education is a big one […] not just about the topic of genetics, but it's also about like how to communicate with families and […] identifying whose role it is and even just practical things, like […] what are the genetic services available to identify if there is a genetic element or not". (P12) | 3.5 |
| Genomics education should be delivered in palliative care forums | "I think education of people in palliative care about the implications of genetic knowledge [would be helpful]. And that would mean just getting these articles published […] in palliative care journals and try to present the information at palliative care conferences, because that's how people already in the field tend to get their education." (P10) | 3.6 |
| Organisations could be doing more to help integrate genomics into palliative care | "One of the key issues of us not being able to do much, it's lack of access, the lack of awareness […] lack of research results to be distributed and lack of therefore funding. And I think from the health services and policy level, they have a lot they can contribute to help that." (P3) | 3.7 |
| A practical genomics guideline for palliative care would help health professionals | "I think that also having things like quick tips or written information or […] some locus where you could go quickly and get some broad information, even just around the practicalities of referrer." (P2) | 3.8 |

## 4.17 DISCUSSION

This study identified how palliative care health professionals are required to concurrently balance the harms and benefits of genomic information and navigate their responsibility when addressing genomics with their patients, as well as practical strategies to overcome practice barriers.

Our findings show that underlying the challenges faced by palliative care health professionals appears to be an uncertainty about their role in addressing genomics.(103, 113) Although we did not identify a primary cause for this uncertainty, changes to genomic testing guidelines over the last several years, particularly in cancer genomics, may be contributing.(59, 161) Genomic mainstreaming pathways, whereby non-genetics specialists (including oncology health professionals) order genomic testing to guide decisions about treatment, could be driving an assumption that all people receiving palliative care have already had a discussion about genomics.(103, 181) Cancer mainstreaming models, however, typically target distinct tumour types (such as high-grade serous ovarian cancer),(59) meaning people with non-mainstreamed tumours may not automatically be offered genomic testing.

Varied levels of genomic knowledge among oncology health professionals,(244) (and other specialists, such as neurologists),(157, 163) suggest that, along with our findings, palliative care health professionals may be falsely led to believe that their patients have already had a genomics discussion. Although palliative care health professionals do not always see genomics as their primary responsibility, assuming genomics has been previously addressed may inadvertently miss opportunities to provide psychologically and medically valuable genomic information to individuals and families. We have highlighted the holistic skill set of palliative care health professionals, including the ability to approach difficult discussions and facilitate communication among families, as a solid foundation from which to assess and address any unmet genetic needs.(103, 113) To increase confidence in these discussions, however, palliative care health professionals want a better grasp of theoretical and practical genomic concepts.(103, 113, 128, 133, 138, 183)

Implementation scientists recommend a multi-faceted approach to improving health professionals' capability, and this is reflected in our findings.(117, 234, 245) While fostering relationships between palliative care and genetics colleagues provides immediate, everyday support,(133) education embedded at each level of training and development may cultivate

relevancy.(234) Importantly, this could instil knowledge about why an asymptomatic relative's genetic risk assessment is markedly improved with access to the palliative person's genetic information.(19) However, successful changes to health professional practice need to look beyond education.(246) For example, we reported the discomfort health professionals have extracting genomic information from the person with palliative care needs for the family's benefit, because the relatives are not the health professionals' patients. It remains unclear whether holistic support of the family extends to addressing concerns about their own genomic risk.(247)

A health professionals' ability to share genomic information between relatives in the palliative care context is ethically and legally complex.(113, 128) People with palliative care needs may be cognitively unable to engage in genomics discussions or provide informed consent for DNA testing or storage.(191) Family members may disagree about the value of genomic information or their relative's dying wishes.(190) In our study, palliative care health professionals were committed to a person-centred approach to genomics discussions. Nonetheless, they echoed calls for healthcare organisations to provide them with support to navigate the complicated, time-pressured scenarios that can arise in the palliative care context.(133, 138)

### 4.17.1 Strengths and Limitations

This study adds to our understanding of the challenges of addressing genomics for people with palliative care needs. A key strength is the study design being informed by a systematic review and a theoretical framework.(67, 214) While we captured a range of views, recruitment of palliative care nurses was challenging and a greater voice would have provided further reassurance that a full array of perspectives was captured. Furthermore, most participants were female and worked in a metropolitan area. Although the palliative care workforce in Australia is largely represented by these two groups, there may be diverse views that could yield additional barriers and facilitators not reported here.(248)

### 4.17.2 Practice and Research Implications

Drawing practice implications from qualitative research can be difficult because of limitations in generalisability or determining causation. However, given the limited evidence about genetics in palliative care, a qualitative exploration was deemed appropriate to investigate the scope of this topic. Healthcare organisations could develop resources to help palliative care health professionals locate their local genetics service.

Future research opportunities include examining the barriers and facilitators identified here on a larger, quantitative scale with a view to designing and testing an intervention to support palliative care health professionals to integrate genomics into practice. Exploring the value of genomic information with people who have palliative care needs and their families would fill an important literature gap.

## 4.18 CONCLUSIONS

Three overarching themes collectively describe the decision-making process and challenges palliative care health professionals face when deciding whether to address genomic issues in practice. Health professionals balance the potential harms and benefits of genomic information to their patients, navigate whether it is their responsibility to raise genomics and must overcome numerous practice barriers. Potential strategies to build the capability of palliative care health professionals include fostering genomic-palliative care health professional relationships, developing clinical resources, increasing organisational support for genomics integration and imbedded and ongoing genomics education.

## 4.19 CHAPTER SUMMARY

This chapter presented two peer-reviewed, published manuscripts. Both manuscripts arose from one qualitative study involving two health professional cohorts. For the genetic health professional cohort, four themes described focusing on the benefit to the family, the discomfort of addressing genomics at the end of life, the challenge of placing genomics on the palliative care agenda, and a DNA storage approach at the end of life. For the palliative care health professionals, three themes described balancing the harms and benefits of genomics, navigating genomic responsibility, and overcoming practice barriers. The findings in this study relate primarily to the micro-level (patient–provider interactions) and meso-level (healthcare organisation) of the WHO ICCC framework. In the next chapter, Phase 1 of the GIFT Project continues with a scoping review of policy recommendations that explores some of the macro-level (policy environment) factors.

# 5 Scoping Review of Policy Recommendations

## 5.1 PREAMBLE

In Chapter 5, I present a peer-reviewed, published scoping review manuscript. This scoping review identified and described policy recommendations related to the integration of genomics into the care of people with palliative needs and their families. The findings explore some of the 'macro-level' factors as described by the World Health Organization Innovative care for chronic conditions (WHO ICCC) framework and build on genetic and palliative care health professionals' reported dissatisfaction with organisational support articulated in the qualitative study. Files related to the development and conduct of the scoping review are in appendix D.

## 5.2 MANUSCRIPT 4. A SCOPING REVIEW OF POLICY RECOMMENDATIONS

This paper was published in *Public Health Genomics* in 2022 (Scimago rating Q3 in 'public health, environmental and occupational health' category; 2021 Impact Factor 2.132).(249) I selected this journal to target a genomics audience interested in the policy environment. No permissions were required to reprint this manuscript because it was published under a Creative Commons license. Minor edits have been made, including updating table and figure headings for congruency across the thesis.

Reference: White S, Virdun C, McErlean G, Phillips J, Jacobs C. Integrating genomics into palliative care: a global scoping review of policy recommendations. Public Health Genomics. 2022:1-15.

## 5.3 ABSTRACT

*Background:* Genomics has growing relevance to palliative care where testing largely benefits relatives. Integrating genomics into palliative care has not received the critical attention it requires. Health professionals report a lack of policy guidance to support them to overcome practice barriers to identify people with palliative care needs who are eligible for genetic testing, provide genetic counselling and facilitate genetic testing or DNA storage.

*Summary:* To identify policy recommendations related to: (1) integrating genomics into the care of patients with palliative care needs and their families, and (2) care of the family unit, we performed a scoping review of palliative care and genomic policies. Two of 78 policies recommended integrating genomics into palliative care. Six palliative care policies mentioned genomics in background information but were without relevant recommendations.
No genomics policies mentioned palliative care in the background information. Across all policies, "Delivering Family-Centred Care" was the most frequent recommendation related to care of the family unit (*n*=62/78, 79.5%).

*Key Messages:* We identified a policy gap related to integrating genomics into palliative care. Without policy guidance, health services are less likely to commit funding towards supporting health professionals. Without funding, delivering the benefits of genomics to patients and relatives is more difficult for health professionals. Framing recommendations about genomics as family-centred care may resonate with genomic and palliative care stakeholders. These findings highlight an opportunity to improve the policy landscape and access to genomic information for patients with palliative care needs. We call for incorporation of appropriate recommendations into palliative care and genomic policy.

## 5.4 INTRODUCTION

Genetics and genomics (herein referred to as 'genomics') has growing relevance to most areas of healthcare, including palliative care, as the genetic basis for disease increasingly influences treatments, risk management, reproductive options and social decisions.(2) The ability of health professionals to identify people with palliative care needs who may have an inherited pathogenic variant, provide genetic counselling, facilitate genomic testing (or DNA storage for future testing) and support family communication has utility for both the individual and family. For the individual, genomic testing may help them access personalised therapies , while unaffected family members can have predictive testing to inform future disease risk and screening or risk-reducing options.(3, 12) Additionally, genomic testing has utility beyond medical decision-making (often termed 'personal utility').(27) Genomic information has the potential to yield psychological benefits for patients with palliative care needs; providing answers for the cause of an illness, a sense of control, and relief at knowing family members may be able to avoid the same disease.(185, 189) For the clinical and personal benefits of palliative–genomic testing to be realised, integrating genomics into the care of people with palliative care needs must be added to the palliative care agenda.(180)

Genetic and palliative care health professionals have identified a lack of guidance and organisational support to overcome barriers to integrating genomics into the care of people with palliative care needs and their families.(218, 250) In the palliative care context, heightened patient and family emotions, deteriorating patient health and cognition, and variable genomic attitudes and knowledge may influence a health professional's decision to initiate a discussion about genomic testing.(128) Although some palliative care health professionals have concerns about initiating genomic discussions with patients who have palliative care needs and their families,(113) there is no evidence of psychological harm resulting from genomic discussions.(92) In fact, addressing existing concerns that patients with palliative care needs have about their relatives' future disease risk may yield positive psychological benefits.(183) In either case, offering genomic testing to a person at end of life for the benefit of family members highlights the uncertain ethical and legal terrain of palliative care health professionals' duty to the family, particularly where there are complex family dynamics.(98) Furthermore, the absence of support from health services leaves health professionals alone to manage the complex ethical, legal and social implications of approaching discussions about genomics with people who are palliative (and their families), particularly as they near end of life.(113) When these barriers prevent patients with palliative care needs from accessing genomic testing before they die, their DNA and family history knowledge are irretrievably lost, which in turn , impacts the quality of information relatives are provided with about future disease risk and management.(24)

Positive public policy for genomics in palliative care will support health professionals to deliver the benefits of genomics to patients and families. Implementation science theories (such as Michie and colleagues' "Behaviour change wheel") highlight policy as an important influence on health professionals' capability (e.g. having the knowledge to patients eligible for genomic testing), opportunity (e.g. processes in place to enable DNA storage at end of life) and motivation (e.g. belief they are acting in patients and families' best interests).(106) Other frameworks demonstrate the relationship between the macro- (policy environment), meso- (health services and professional organisations) and micro-level (patient-provider interactions) factors that affect the success of health intervention implementation.(67) For instance, policy recommendations ideally stimulate funding to overcome barriers and develop local guidelines to support health professionals integrate genomics into their practice. For families to benefit from the genomic testing of their dying affected relative, supportive policy at a government or organisational level is first needed to generate the flow-on effects to health services and

professionals.(251). Governments and professional organisations are publishing policies that articulate the significance of genomics to routine medical care, but it is not known whether existing policies acknowledge the benefits of palliative–genomic testing or address the practical and ethical challenges health professionals face in the palliative care context.

To investigate the policy support available for palliative and genetic health professionals, we performed a scoping review to identify and map current policy recommendations about the integration of genomics into the care of people with palliative care needs, including recommendations related to care of the family unit. We sought to answer the following questions:

1. What global policy guidance is available that describes the integration of clinical genetic and genomic health information into the care of people with palliative needs and their families?
2. What recommendations in palliative care and genetic/genomic policies regarding care of the family unit are relevant to the integration of clinical genetic and genomic health information into the care of people with palliative needs and their families?

## 5.5 METHODS

### 5.5.1 Design

A scoping review, using the methodology described by the Joanna Briggs Institute, was selected to map and describe global policy recommendations related to genomics in palliative care, rather than evaluate impact or effectiveness of recommendations.(252) We used the World Health Organization Innovative care for chronic conditions framework as an initial conceptual framework, which guided us to explore the 'macro' policy environment.(67) Reporting items aligned with the PRISMA-ScR extension (Appendix D2).(253) An a-priori review protocol was published on Open Science Framework (https://osf.io/5eumn/) and updated in January 2022 when the second review question was added. The review team (consisting of palliative care and genetic counselling experts with experience in systematic and scoping reviews) developed the second review question following initial exploration of the extracted recommendations about care of the family unit. We identified an opportunity to determine whether recommendations about care of the family unit could reveal common policy ground between palliative care and genomics.

### 5.5.2 Eligibility Criteria

Eligibility criteria were developed using the Population, Concept and Context framework.(252) For the purpose of this review, we used the term 'policy', but sought to include a range of governance documents, including 'frameworks', 'strategies', 'standards' or similar. Policies were required to focus on the provision of palliative care or clinical genetic and genomic services (including genetic counselling). We did not include clinical practice guidelines for singular conditions as we aimed to examine the broader policy environment (for example, service development frameworks were included, while care guidelines for terminal breast cancer were excluded). Eligible policies were published in English between 2010 – 2022 and authored by national or state (or equivalent) governments or their agencies, or international, national or state-based professional palliative care, clinical genetics/genomics or genetic counselling organisations. To retrieve policies from countries with the infrastructure to integrate genomics into palliative care, we included policies from the top 20 countries ranked by the Economic Intelligence Unit Quality of Death Index, for the quality of their palliative care provision.(254) The full eligibility criteria is available in appendix D3.

### 5.5.3 Information Sources and Search

The search strategy was co-designed with an information scientist (S.S) and peer-reviewed at a genetic counselling research seminar. The strategy consisted of three approaches: (1) database search, (2) web-search (3) emailing key informants. The database and web-search strategies are available in appendix D4. We repeated the database and web-search twice: once for palliative care policies and once for genetic and genomic policies. The search was run on the 1st June 2020 and repeated on the 21st February 2022.

1. Database Search: Three databases (Medline, EMBASE and CINAHL) were interrogated using nesting and Boolean operators to combine relevant terms, such as 'Guideline' and 'Health Policy' with 'Genetic Counseling', 'Genetic Testing', or with 'Palliative Care'. The database search was supplemented by hand-searching the Canadian Agency for Drugs and Technologies in Health grey literature tool.(255) Database records were exported to, and deduplicated in EndNote.(256)
2. Web-Search: We designed a systematic web-search to retrieve non-commercially published documents.(257) Using Google, we constructed a single-line search using nesting and Boolean operators based on the Medline strategy. To reduce the potential bias of geo-locating algorithms, we used incognito mode and cleared caches and

cookies prior to running the search.(257) Based on the number of retrieved pages identified on a test run, we made a pragmatic decision to limit the search to the first ten pages of returned results.(258) The results were captured by copying the web-site name and URL into a Microsoft Excel spreadsheet so the same results could be screened by more than one reviewer.(259) On advice from the information scientist, we ran two additional searches with the "type:PDF" function (one for palliative care and one for genetic/genomic policies), to increase the sensitivity of the search towards retrieving policy documents.

3. Emails to Key Informants: To capture any missed policies, we emailed key informants to cite their local and/or national palliative care or genetics/genomics policy. Palliative care key informants were identified in the contact list in the Economist Intelligence Unit Quality of Death Index.(254) Genetic and genomic key informants were identified via the Transnational Alliance of Genetic Counseling, consultation with experts, authors identified in this review and targeted, country-specific web searching.(260)

Forward-searching (using Web of Science database) and backward-searching (reviewing reference lists) was conducted on all policies meeting inclusion criteria.

### 5.5.4 Selection of Sources of Evidence

S.W. & C.J. piloted the eligibility criteria by independently screening 25 randomly selected policies. Changes to criteria included specifying that at least 50% of the policy must be relevant to palliative care or genetic/genomic service provision. S.W. & G.M. then independently screened 20% of the records at title and abstract, and full text screening using Covidence and Microsoft Excel.(259, 261) For records arising from the web-search, title and abstract screening involved reviewing the web-page, and full text screening involved reviewing the web-site in full. We also followed any potentially relevant internal or external web-links (snowballing). With biostatistician consultation (K.R), we used a Prevalence-Adjusted Bias-Adjusted Kappa statistic and achieved substantial inter-rater agreement (>0.7) before S.W. screened the remainder of the records independently.(118, 262)

### 5.5.5 Data Items and Charting

We used a modified Joanna Briggs Institute data extraction instrument with pre-determined data items (see appendix D5). In addition to policy characteristics (e.g., author, year, country), we extracted verbatim recommendations with their relevant heading and page number. Ten policies were randomly selected for S.W., G.M. & C.J. to independently pilot the

extraction tool. Changes included adding extraction fields, including organisation's jurisdiction (e.g. state, national, international) and population age group (e.g. paediatric, adult, all ages). S.W. independently extracted recommendations about genetics and genomics from palliative care policies, recommendations about palliative care from genetic and genomic policies and recommendations about care of the family unit from both palliative care and genetic and genomic policies. G.M. reviewed extracted data from 20% of the included policies and verified accuracy.

### 5.5.6 Critical Appraisal

In line with scoping review guidance, we did not perform critical appraisal assessments on individual policy documents because (a) policies are not primary research articles and (b) to our knowledge, a validated critical appraisal tool for policy documents does not exist.(258) However, to embed a quality check into our eligibility criteria, we required policies to be evidence-based and include a description of the method by which the policy was developed (informed by the AGREE-tool).(263)

### 5.5.7 Mapping and Synthesis

To determine what global policy guidance was available for the integration of genomics into the care of people with palliative needs, policies were grouped by region, policy focus (palliative care or clinical genetics and genomics), jurisdiction (state or equivalent, national or international) and population age group (paediatric, adult or all ages). The presence or absence of recommendations related to integration of genomics into palliative care was tabulated and narratively summarised.

To determine which recommendations regarding care of the family unit were relevant to the integration of genomics into the care of people with palliative needs, S.W. and C.V. independently grouped recommendations about care of the family unit into one of three categories: (a) Relevant to palliative care only, (b) Relevant to genetics and genomics only, or (c) Relevant to both palliative care and genetics and genomics. Initial agreement was 67.44%. S.W. and C.V. then collaboratively assessed and sorted each recommendation into the appropriate category using a broad approach. For example, we broadly interpreted the recommendation "If services cannot meet the family's needs, appropriate referrals are made" as referring to any need (e.g. physical, psychological or social) and could therefore apply to both palliative care and genetic and genomic services. S.W. further sorted the recommendations relevant to both palliative care and genetics and genomics into descriptive

categories. C.V. reviewed the descriptive categories and provided feedback, including the suggestion to collapse and rename some of the categories. Once categories were finalised, we calculated the percentage of each category as a proportion of the total number of policies. Initially, a granular-level matrix with all recommendations relevant to both palliative care and genetics and genomics were cross-tabulated with each policy. Individual recommendations were grouped into categories to demonstrate which policies included recommendations from each category (note: this did not represent how frequently the category showed up in each policy). We used descriptive statistics to calculate the presence of each category across all policies, using proportions (n) and percentage (%) of the total number of policies (N). We additionally stratified by policy focus (palliative care or genetics and genomics) and region. To visually represent the proportion of each category across policies (stratified by region), the biostatistician (K.R.) generated a "heat map" using 'R' software.(264, 265) A narrative synthesis accompanies the visual results.(121)

### 5.5.8 Ethics

The University of Technology Sydney Research Ethics Office waived the requirement of ethics approval for this study. However, the reviewers were mindful of contacting key informants as part of this project. A maximum of three email attempts were made to each person and no direct quotes are included.

## 5.6 RESULTS

In total, 78 global policies were included (see PRISMA-flow diagram in Figure 9). The majority were palliative care policies (*n*=61, 78.21%) with a country-level focus (*n*=41, 52.56%) and relevant to all ages (*n*=58, 74.36%). Australian policy accounted for one-quarter (*n*=20, 25.64%) of the included policies (Table 10).

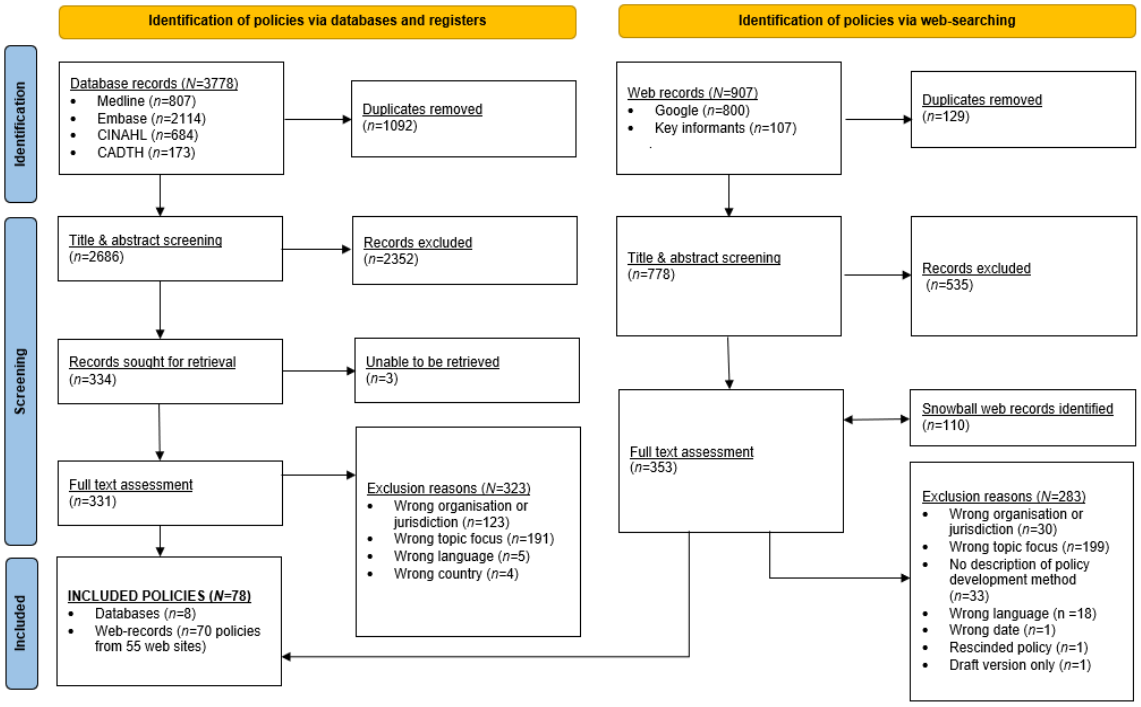


Figure 9. PRISMA-flow diagram demonstrating the number of retrieved records, inclusion and exclusion numbers and reasons for exclusion. From 4685 records, 78 policies were included in the final review. Abbreviations: CINAHL=Cumulative Index to Nursing and Allied Health Literature; CADTH=Canadian Agency for Drugs and Technologies in Health

Table 10. Summary of included policies (*N*=78), listed by region and publication year.

| EIU QOD index rank | Region | Simplified citation | Policy focus | | Geographical jurisdiction | | | Population scope | | | Recommendations | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | | | Palliative Care | Genomic | State | National | International | Paediatric | Adult | All Ages | Integration of genomics into palliative care | Care of family recommendations apply to palliative care & genomics |
| NA | Europe | Fellmann et al. (2019) | | ✓ | | | ✓ | | | ✓ | X | ✓ |
| | | Oliver et al. (2016) | ✓ | | | | ✓ | | | ✓ | X | ✓ |
| | | Tuffrey-Wijne et al. (2015) | ✓# | | | | ✓ | | ✓ | | X | ✓ |
| | | van der Steen et al. (2014) | ✓ | | | | ✓ | | ✓ | | X | ✓ |
| | | Van El et al. (2013) | | ✓ | | | ✓ | | | ✓ | X | ✓ |
| | | Van El and Cornel (2011) | | ✓ | | | ✓ | | | ✓ | X | X |
| NA | Global | World Health Organization (2018a) | ✓# | | | | ✓ | | | ✓ | X | ✓ |
| | | World Health Organization (2018b) | ✓# | | | | ✓ | ✓ | | | X | ✓ |
| | | Parikh et al. (2017) | | ✓ | | | ✓ | | | ✓ | ✓ | ✓ |
| 1 | United Kingdom | Her Majesty's Government (2020) | | ✓ | | | ✓ | | | ✓ | X | ✓ |
| | | National Institute for Health and Care Excellence (2019) | ✓ | | | | ✓ | | ✓ | | X | ✓ |
| | | Hospice UK (2017) | ✓ | | | | ✓ | | | ✓ | X | ✓ |
| | | National Institute for Health and Care Excellence (2017) | ✓ | | | | ✓ | ✓ | | | X | ✓ |
| | | National Institute for Health and Care Excellence (2016) | ✓ | | | | ✓ | ✓ | | | X | ✓ |
| | | Leadership Alliance for the Care of Dying People (2014) | ✓ | | | | ✓ | | | ✓ | X | ✓ |

| | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|
| | | National Institute for Health and Care Excellence (2011) | ✓ | | | ✓ | | ✓ | | X | ✓ |
| *1* | *England* | National Palliative and End of Life Care Partnership (2021) | ✓ | | | ✓ | | | ✓ | ✓ | ✓ |
| | | Palliative Care for People with Learning Disabilities (2017) | ✓ | | | ✓ | | | ✓ | X | ✓ |
| | | Department of Health (2016) | ✓ | | | ✓ | | | ✓ | X | ✓ |
| | | National End of Life Care Programme (2011) | ✓ | | | ✓ | | ✓ | | ✓ | ✓ |
| *1* | *Wales* | Welsh Government (2017) | | ✓ | | ✓ | | | ✓ | X | X |
| *1* | *Scotland* | Dumfries and Galloway Integration Joint Board (2020) | ✓# | | ✓* | | | ✓ | | X | ✓ |
| | | Glasgow City Health and Social Care Partnership (2018) | ✓ | | ✓* | | | | ✓ | X | ✓ |
| *1* | *Northern Ireland* | Department of Health Social Services and Public Safety (2010) | ✓ | | | ✓ | | ✓ | | X | ✓ |
| 2 | Australia | South Australian Health (2021) | ✓ | | ✓ | | | | ✓ | X | ✓ |
| | | WA Department of Health (2021) | ✓# | | ✓ | | ✓ | | | X | ✓ |
| | | NSW Health (2019) | ✓ | | ✓ | | | | ✓ | X | ✓ |
| | | Department of Health (2018) | ✓ | | | ✓ | | | ✓ | X | ✓ |
| | | Palliative Care Australia (2018a) | ✓ | | | ✓ | | | ✓ | X | ✓ |
| | | Palliative Care Australia (2018b) | ✓ | | | ✓ | | | ✓ | X | ✓ |
| | | WA Department of Health (2018) | ✓ | | ✓ | | | | ✓ | X | ✓ |
| | | Department of Health and Human Services: Tasmanian Government (2017) | ✓ | | ✓ | | | ✓ | | X | ✓ |
| | | Department of Health (2017) | | ✓ | | ✓ | | | ✓ | X | ✓ |

| | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|
| | | Department of Health and Human Services (2017) | | ✓ | ✓ | | | | ✓ | X | X |
| | | NSW Health (2017) | | ✓ | ✓ | | | | ✓ | X | ✓ |
| | | Australian Commission on Safety and Quality in Health Care (2016) | ✓ | | | ✓ | ✓ | | | X | ✓ |
| | | Department of Health and Human Services (2016) | ✓ | | ✓ | | | | ✓ | X | ✓ |
| | | Australian Commission on Safety and Quality in Health Care (2015) | ✓ | | | ✓ | | ✓ | | X | ✓ |
| | | QLD Health (2015) | ✓ | | ✓ | | | | ✓ | X | ✓ |
| | | Health (2013) | ✓ | | ✓ | | | | ✓ | X | ✓ |
| | | Palliative Care New South Wales (2012) | ✓ | | ✓ | | | | ✓ | X | ✓ |
| | | Department of Health (2012) | ✓ | | ✓ | | | | ✓ | X | ✓ |
| | | National Health and Medical Research Council (2011) | ✓ | | | ✓ | | ✓ | | X | ✓ |
| | | Department of Health (2011) | ✓ | | ✓ | | | | ✓ | X | ✓ |
| 3 | New Zealand | Hospice New Zealand (2019) | ✓ | | | ✓ | | ✓ | | X | ✓ |
| | | Ministry of Health (2017a) | ✓ | | | ✓ | | ✓ | | X | ✓ |
| | | Ministry of Health (2017b) | ✓ | | | ✓ | | ✓ | | X | ✓ |
| | | National Health Council (2015) | | ✓ | | ✓ | | | ✓ | X | ✓ |
| 4 | Ireland | National Clinical Programme for Palliative Care (2019) | ✓ | | | ✓ | | ✓ | | X | ✓ |
| | | HSE Primary Care Division (2017) | ✓ | | | ✓ | | ✓ | | X | ✓ |
| | | Irish Hospice Foundation, Irish College of General Practitioners, and Health Service Executive (2011) | ✓ | | | ✓ | | | ✓ | X | ✓ |

| | | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| | | Irish Hospice Foundation (2010) | ✓ | | ✓ | | ✓ | X | ✓ |
| 8 | Netherlands | IKNL and Palliactief (2017) | ✓ | | ✓ | | ✓ | X | ✓ |
| | | ZonMw (2015) | ✓ | | ✓ | | ✓ | X | ✓ |
| 9 | United States of America | National Coalition for Hospice and Palliative Care (2018) | ✓ | | ✓ | | ✓ | X | ✓ |
| | | Hampel, Bennett, Buchanan, Pearlman, and Wiesner (2015) | | ✓ | ✓ | | ✓ | X | ✓ |
| 10 | France | Aviesan (2016) | | ✓ | ✓ | | ✓ | X | ✓ |
| 11 | Canada | Ministry of Health (2021) | ✓ | | ✓ | | ✓ | X | ✓ |
| | | Ontario Palliative Care Network (2019) | ✓ | | ✓ | | ✓ | X | ✓ |
| | | Genome Canada (2019) | | ✓ | | ✓ | ✓ | X | X |
| | | Genome British Columbia (2019) | | ✓ | ✓ | | ✓ | X | X |
| | | Health Canada (2018) | ✓ | | | ✓ | ✓ | X | ✓ |
| | | Government of New Brunswick (2018) | ✓ | | ✓ | | ✓ | X | ✓ |
| | | Canadian Hospice Palliative Care Association (2015) | ✓ | | | ✓ | ✓ | X | ✓ |
| | | Alberta Health Services (2014) | ✓# | | ✓ | | ✓ | X | ✓ |
| | | Department of Health and Wellness (2014) | ✓ | | ✓ | | ✓ | X | ✓ |
| | | Canadian Hospice Palliative Care Association (2013) | ✓ | | | ✓ | ✓ | X | ✓ |
| | | Ministry of Health (2013) | ✓ | | ✓ | | ✓ | X | ✓ |
| 12 | Singapore | Ministry of Health (2018) | | ✓ | | ✓ | ✓ | X | ✓ |
| | | Singapore Hospice Council (2015) | ✓ | | | ✓ | ✓ | X | ✓ |
| | | Lien Centre for Palliative Care (2012) | ✓ | | | ✓ | ✓ | X | ✓ |

| | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| 14 | Japan | The Japanese Association of Medical Sciences (2011) | | ✓ | | ✓ | | | | ✓ | X | ✓ |
| 15 | Switzerland | Swiss Academy of Medical Sciences (2018) | ✓ | | | ✓ | | | | ✓ | X | ✓ |
| | | Federal Office of Public Health (2014) | ✓ | | | ✓ | | | | ✓ | X | ✓ |
| | | Swiss Academy of Medical Sciences (2013) | ✓ | | | ✓ | | | | ✓ | X | X |
| | | Federal Office of Public Health (2012) | ✓ | | | ✓ | | | | ✓ | X | ✓ |
| | | Federal Office of Public Health (2010) | ✓ | | | ✓ | | | | ✓ | X | X |
| 20 | Finland | Ministry of Social Affairs and Health (2015) | | ✓ | | ✓ | | | | ✓ | X | ✓ |
| | | **TOTAL** | **61** | **17** | **22** | **40** | **16** | **5** | **15** | **58** | **2** | **71** |

NB. Policies marked with an asterix (#) mentioned genetics or genomics in their background information. Policies marked with a star (*) have a council area jurisdiction but were included because they originate from Scotland which does not have states/provinces. Full citations of all policies included in this review is in appendix D6. Abbreviations - EIU QOD: Economist Intelligence Unit Quality of Death, NA: Not applicable

### 5.6.1 Integration of Genomics into Palliative Care

Of the 78 policies, only two (2.56%) included recommendations about integrating genomics into palliative care (Figure 10).(266, 267) The first, an international genomics policy, recommended palliative care involvement when planning care for people with mitochondrial disease.(266) The second, an English palliative care policy for patients with neurological disease, recommended being aware of the psychological impact of a positive family history on the patient, including fear of the disorder and of their children developing the same disease.(267) Of the 61 palliative care policies, only six (9.84%) mentioned genomics in the background information, and none of these incorporated genomics into their recommendations.(76-78, 268-270) The background information in these six policies illustrated the increased likelihood of a genetic cause in palliative children and examples of genetic conditions.(76-78, 268-270) One policy described the impact of life-limiting, congenital anomalies, including pain, social isolation, stigmatisation and a lack of resources to provide long-term palliative care.(77) In three of the six policies that mentioned genomics in the background, genomic information was referred to in policies, or sections of the policy, about paediatric palliative care.(76, 77, 270) Excerpts of the background information are in appendix D7. None of the genomic policies mentioned palliative care in their background information.

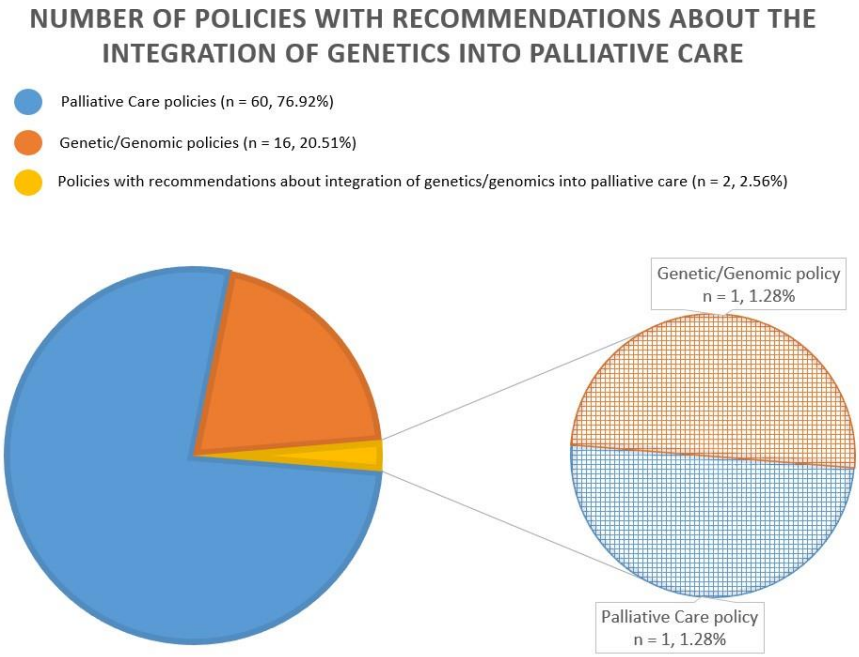


Figure 10. Two of 78 policies included in this review included recommendations about integrating genomics into the care of people with palliative care needs.

### 5.6.2 Care of the Family Unit

Almost all policies (*n*=72/78, 92.31%) had recommendations about care of the palliative patient's family. We identified 168 unique recommendations, 55 of which were relevant to palliative care only, five relevant to genetics and genomics only and 108 recommendations relevant to both palliative care and genetics and genomics. Recommendations relevant to both palliative care and genetics and genomics were grouped into 11 descriptive categories (Table 11).

The most frequent category overall (n=62/78, 79.5%), including by region (n=10/18, 55.56%) was "Delivering Family-Centred Care", although only 29.41% (n=5/17) of genomic policies included this category compared to 93.44% (n=57/61) of palliative care policies. This category described the importance of attending to family members' psychological, social and spiritual needs. The second most prevalent category overall was "Governance & Policy" (n=53/78, 67.9%), which recommended care for families be enshrined in policy and enacted by health services. The least mentioned category overall (n=5/78, 6.4%) and by region (n=4/18, 22.22%) was "Physical & Symptom Care", which related to assessing and managing family members' physical health.

In addition to "Delivering Family-Centred Care", genomic policies gave equal attention to "Ethical Care" (n=5/17, 29.41%) and "Governance & Policy" (n=5/17, 29.41%). "Ethical Care" recommendations described the ethical obligations health professionals have towards family members. For example, that discussions surrounding consent for genomic testing must include implications for family members. As for the palliative care policies, their other focus was on recommendations related to "Governance & Policy" (n=48/61, 78.69%) and "Informational Needs" (n=43/61, 70.49%). "Informational Needs" recommendations described health professionals' duty to respond to each family's unique informational needs by assessing family members' information requirements and provide information in an accessible way (Figure 11).

Table 11. Recommendations related to care of the family unit grouped into 11 categories.

| Category heading | Category description | Frequency of category in policies overall (*N*=78) | Frequency of category in PC policies (*n*=61) | Frequency of category in genomic policies (*n*=17) |
|---|---|---|---|---|
| | | *n* (%) | | |
| Delivering family-centred care | Health professionals deliver family-centred care, recognising the important role families' play, and identify when family members may need to be recipients of care to support their emotional, social, and physical needs | 62 (79.49) | 57 (93.44) | 5 (29.41) |
| Governance and policy | Care is organised under relevant government and organisational policy and enacted through health services to foster a supportive and responsive environment. Families are partners in identifying areas for improvement. | 53 (67.95) | 48 (78.69) | 5 (29.41) |
| Informational needs | Health professionals assess, provide, and respond to families individualised informational needs | 46 (58.97) | 43 (70.49) | 3 (17.65) |
| Bereavement care | Health professionals identify and support family members through simple and complex grief reactions to their loss | 37 (47.44) | 37 (60.66) | 0 (0) |
| Ethical care | Health professionals have a duty of care to be aware of their ethical obligations and aim to uphold principles of autonomy, beneficence, non-maleficence, and justice in their practice | 34 (43.59) | 29 (47.54) | 5 (29.41) |
| Communication skills and processes | Health professionals are supported by processes that enhance their skills to communicate efficiently and empathically families | 29 (37.18) | 29 (47.54) | 0 (0) |
| Assessment and care planning | Health professionals perform assessments to ensure care planning is individualised, responsive, and appropriate to the family's needs | 27 (34.62) | 27 (44.26) | 0 (0) |
| Research and feedback | Institutions and health professionals are aware improvements will result from developing appropriate outcome measures, inviting feedback from families, and partnering with families in research | 26 (33.33) | 25 (40.98) | 1 (5.88) |
| End-of-life care | Health professionals engage in important conversations with the family when the palliative person is close to death | 18 (23.08) | 18 (29.51) | 0 (0) |
| After death care | Health professionals must manage administrative and supportive processes after the palliative person has died | 8 (10.26) | 7 (11.48) | 1 (5.88) |
| Physical and symptom care | Health professionals ensure that family members' physical needs are assessed and cared for | 5 (6.41) | 5 (8.20) | 0 |
| NB. Categories are listed in order of the frequency they were identified in policies overall, where 'n' is the number of policies that included recommendations within the category. The frequency of the category by palliative care (PC) and genomic policy is also displayed. | | | | |

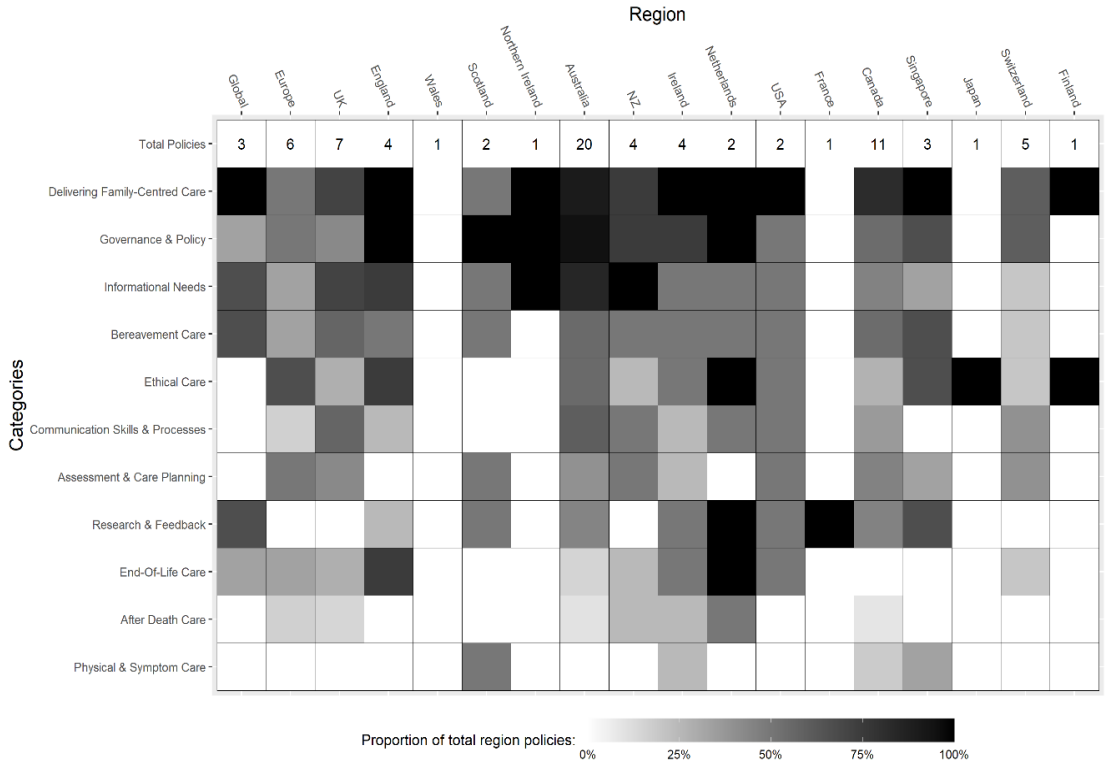


Figure 11. The degree of shading in this heat map represents the proportion of policies (as a percentage of the total policies in that region) that included recommendations about care of the family unit. Policies are grouped by region on the x-axis, and the descriptive categories of recommendations related to care of the family unit are listed on the y-axis.

## 5.7 DISCUSSION

This global scoping review of policy recommendations complements the evolving dialogue about patients',(92, 93) families',(96) and health professionals' (113, 138, 218) experiences and views of the barriers and facilitators affecting integration of genomics into palliative care by examining the policy environment. We have identified and mapped recommendations related to the integration of genomics into the care of people with palliative care needs. A policy gap was evident, with only two of 78 policies explicitly including recommendations to integrate genomics into the care of patients with palliative care needs and their families. We also mapped recommendations about care of the family unit, finding that "Delivering Family-Centred Care" was a key recommendation across both palliative care and genomic policies.

Implementing genomics into the palliative care setting requires policy action from the meso- (i.e. health services and professional organisations) and macro-level (i.e. government).(67) Our review suggests the palliative care profession is falling behind other medical specialties, such as oncology,(271) neurology,(272) and cardiology,(273) which have

published documents highlighting the importance of genomics to their patient groups. Despite this, translation of genomics into routine care is slow and there are numerous reasons why health professionals across specialties do not broach genomics with their patients.(214) If genomics is not addressed by treating specialists, palliative care becomes the final point to collect DNA from the affected person for the family's benefit before the patient dies and the opportunity is lost.(24) Palliative care and genomic organisations need to communicate this duty to their health professionals, as guiding relatives to engage in appropriate levels of screening or risk-reduction is a key clinical and economic benefit offered by genomics.(94, 274) Palliative care and genomic health professionals have called for organisational support, including initial tertiary genomic education, continuing professional development, co-locating palliative care and genetic teams within health services and developing point-of-care guidelines to identify high-risk patients.(138, 218) Resources and funding are essential to the success of these strategies but health services are unlikely to commit these without a positive policy environment.(251) We have demonstrated a need for policy that articulates the importance of a genomics discussion before the palliative person dies, acknowledges the complexities and challenges, and delivers potential solutions to support health professionals.

One reason policy guidance may be lacking is that demonstrating the economic value of genomics in palliative care is challenging. Traditionally, research assessing the economic value of genomics to the family unit is measured through rates of predictive testing and changes in an individual's health behaviour.(3) To generate economic evidence in the palliative care context, researchers must overcome difficulties related to ethical concerns (e.g. satisfying institutional review boards that their research will not unduly harm vulnerable people) and logistical hurdles (e.g. patients dying prior to research participation).(236) To holistically assess the value of genomics to families and fill this important gap, health economists have suggested enriching economic evaluations with ethical, legal and social implication (ELSI) research.(235) As genomics continues to revolutionise healthcare, we see a need for palliative care implementation research to demonstrate the economic value of genomic testing to families, alongside the clinical, psychological and social benefits.

Our review corroborated the value of family-centred care to palliative care and clinical genetics, finding that recommendations related to "Delivering Family-Centred Care" were the most prevalent category of recommendations across both palliative care and genomic policies.(99, 100) These recommendations illustrate the importance of attending to family members' psychological, social and spiritual needs. Elements of family-centred care align with

the personal utility of genomic information, such as satisfying altruistic motivations to protect their relatives from future disease, reducing the family's uncertainty of the future, providing a sense of control and making meaning through findings answers.(183, 275) Leveraging this common ground offers policy makers an avenue to frame the benefits of genomics as family-centred care, so relevant recommendations resonate with both palliative care and clinical genetic and genomic stakeholders.

With accumulating evidence demonstrating the value of genomics in palliative care, it is timely for palliative care and genomic policy makers to develop policy recommendations about integrating genomics into palliative care, so the clinical and psychological benefits of genomics can be realised. We call for a clear policy stance that communicates the important of committing funding and resources towards supporting health professionals to address genomics with patients who have palliative care needs and their families.

## 5.8 STRENGTHS AND LIMITATIONS

This review addresses a gap in our understanding of how genomics is conceived in the context of palliative care and is strengthened by adherence to established scoping review guidance. In addition, the multi-pronged search strategy (in particular, the web-search) identified relevant policies through commercial and non-commercial publishers, as opposed to relying solely on academic databases. However, web-searching methods are described vaguely in scoping review guidelines, meaning we relied on other researchers' published experiences to develop our own procedures. In addition, we took steps to reduce the web-searching "bubble-effect" (which is the tendency to retrieve web records within the searcher's location), but there appeared to be comparatively more web results from our home country (Australia).(276) Regarding eligibility criteria, our resources limited us to English-language policies, so we may have missed relevant recommendations from policies in other languages. Lastly, to maintain feasibility of the review, we focused on palliative care and genomic policies; however, there may be related recommendations in policies in adjacent medical fields (such as oncology or obstetrics/gynaecology).

## 5.9 CONCLUSION

The dearth of policy recommendations related to the integration of genomics in the care of people with palliative care needs and their families is an identified gap. Without a clear policy stance, health services are unlikely to support health professionals to navigate the

complexities of integrating genomics into routine palliative care. Delivering family-centred care was a prevalent existing recommendation across both palliative and genomic policies. Policy makers urgently need to harness this common ground to frame the benefits of genomics as family-centred care, to ensure recommendations resonate with both palliative care and genomic stakeholders. To realise the potential clinical, psychological, social and economic benefits of genomic medicine in palliative care, we call on policy makers to incorporate recommendations about the integration of genomics in palliative care to communicate the importance of allocating resources and funding to health services.

## 5.10 CHAPTER SUMMARY

In this chapter, a scoping review in the form of a peer-reviewed, published manuscript was presented. The scoping review identified a lack of high-level policy guidance related to the integration of genomics to palliative care and an opportunity to frame future recommendations as 'family-centred care'. In the next chapter, I move into Phase 2 of the GIFT Project. Chapter 6 presents an assessment and comparison of the barriers and facilitators between genetic and palliative care health professionals that were identified in Phase 1.

## 5.11 MEETING OBJECTIVE 1: EXPLORE THE BARRIERS TO AND FACILITATORS OF INTEGRATING GENOMICS INTO THE CARE OF PEOPLE WITH PALLIATIVE CARE NEEDS

The first objective of the GIFT Project has now been met. The qualitative study in Chapter 4 explored the barriers and facilitators from the perspectives of genetic and palliative care health professionals.(218, 250) The qualitative findings aligned predominantly with the micro- and meso-level factors of the WHO ICCC framework. The scoping review presented in Chapter 5 explored the policy environment (macro-level) by identifying and describing genomic and palliative care policy recommendations. The inferences and meta-inferences generated from and across these studies are discussed further in Chapter 7 where the research questions are answered.

# 6 Quantitative Assessment and Comparison of the Barriers and Facilitators

## 6.1 PREAMBLE

In Chapter 6, I present Phase 2 of the Genomic Information for Families of the Terminally ill (GIFT) Project. Phase 2 comprised a quantitative assessment and comparison of the identified barriers and facilitators between genetic and palliative care health professionals. The development of the survey, through the process of mid-point data connection, was described in Chapter 3. From section 6.10 onwards, I present an extended methods, results, and discussion section related to DNA storage for people near their end of life. The DNA storage approach was suggested by genetic health professionals in Phase 1 (Chapter 4) but was not reported in the corresponding qualitative manuscript. Similarly, the results related to DNA storage were beyond scope of the quantitative manuscript because of the limited word count and need to tell a cohesive story. However, these findings have relevance to the thesis aim and objectives, and were incorporated into the mid- and end-point data integration processes.

## 6.2 MANUSCRIPT 5. A SURVEY OF GENETIC AND PALLIATIVE CARE HEALTH PROFESSIONALS

In December 2022, I submitted a manuscript describing the study in this chapter to the *European Journal of Human Genetics* (2021 impact factor 5.531, Scimago rating Q1 in 'genetics' category)(213). The manuscript has been peer reviewed. The journal advised they will consider a revised version that addresses the reviewers' comments. Comments from reviewers pertained mostly to the low response rate in this study and suggested being more explicit about the limitations of the findings. A revised version of the manuscript was submitted to the journal on April 15, 2023, and is currently under consideration. In this chapter, I present the revised and resubmitted version of the manuscript. Minor edits have

been made. I have changed ‘genetic’ to ‘genomic’ (except in the manuscript title), and updated table, figure, and page numbers. Please note, in the survey, the term ‘DNA testing’ was used to describe genetic and genomic testing. In addition, the survey described the clinical storage of DNA as ‘DNA banking’. I have changed the terminology here to ‘DNA storage’ for congruency across the thesis.

Reference: White S, Turbitt E, Rogers K, Tucker K, Best M, Phillips J, et al. A survey of genetic and palliative care health professionals' views towards the integration of genetics into palliative care. Eur J Hum Genet. 2023. (manuscript under consideration).

## 6.3 ABSTRACT

Genetic counselling and testing have utility for people with palliative care needs and their families. However, genetic and palliative care health professionals have described difficulties initiating palliative-genetic discussions. Between March and July 2022, we received *n*=73 surveys (6% response rate) from genetic and palliative care health professionals in Australia and New Zealand that assessed and compared barriers and facilitators. The main perceived barrier to both groups was palliative care health professionals’ lack of genetic knowledge (44%). Most palliative care health professionals were “not at all confident” performing several activities, including discussing DNA storage (52%) and knowing their legal responsibilities when sharing genetic information (58%). The most frequently selected facilitator for genetic health professionals was fostering close relationships with palliative care health professionals (52%), while palliative care health professionals indicated a genetic referral template (51%) would be of assistance. Almost all participants agreed genetic discussions do not undermine the central ethos of palliative care (87%). Fewer palliative care health professionals considered themselves well situated to have genetic discussions with a palliative patient’s family compared to genetic health professionals (*p*=0.014). Our results suggest that genetic and palliative care health professionals support integrating genetics into palliative care, although refinement of the palliative care health professionals’ role in this process is required. We have identified intervention targets to overcome barriers related to knowledge and confidence, which ought to be integrated into future interventions designed to support health professionals deliver the benefits of genetic information to people with palliative care needs and their families.

## 6.4 INTRODUCTION

Genetic counselling and testing can yield important information for individuals and families at all stages of life.(2) In the palliative care setting, the clinical benefit is predominantly for relatives, rather than the patient having testing. Providing a patient who has palliative care needs with the opportunity to engage in genetic counselling (and if indicated, DNA storage or genomic testing) can ultimately help family members to assess and manage future disease risk by, for example, engaging in recommended screening or risk-reducing surgery.(16)

Additionally, patients with palliative care needs may experience personal or psychological benefits beyond those related to clinical intervention. Addressing patients' pre-existing concerns about genetic risk may resolve unmet psychological needs, assist in making meaning from their illness, provide reassurance that family members are receiving relevant information, and support altruistic motivations to help others.(94, 113, 185) Despite these benefits, several barriers (discussed further below) and a lack of evidence-based support prevent genetic and palliative care health professionals from initiating discussions about genomics with patients who have palliative care needs.(214). Developing a robust evidence base will tailor support for health professionals to identify patients eligible for genomic testing to provide genetic counselling before the patient dies and the opportunity to gather genomic information is lost. In so doing, family members will have better access to predictive genetic testing and a more personalised genomic risk assessment.(3)

As the demand for genetic counselling and testing increase, so too does pressure on existing genetic services.(54) Targeted efforts to improve the capability of 'non-genetic' health professionals aim to improve access and delivery of genetic services to patients and families.(277, 278) An understanding of the barriers and facilitators relevant to each context will support the development of appropriate interventions.(102) In the palliative care context, the small body of evidence leaves several gaps.(180) For example, a commonly reported barrier is low genomics knowledge and confidence amongst palliative care health professionals.(113, 133, 138) These descriptions (i.e., 'low confidence') are not specific enough to inform the development of an intervention to support palliative care health professionals in the areas in which they feel deficient. By defining the activities requiring support, directed interventions can be implemented for the greatest impact.(102)

Another gap is found in the evidence describing genetic and palliative care health professionals' views of the potential harms and benefits of genomic discussions.(103, 113, 128,

181, 218, 250, 279) Qualitative studies have begun to illustrate the motivations underlying health professionals' decision making in the palliative context, such as the possibility for genomic discussions to cause psychological harm to patients and families. To advance our understanding, themes about harms and benefits should be examined with quantitative methods to determine whether reported attitudes are generalisable. However, to our knowledge, there does not appear to be any triangulation of the qualitative descriptions of harms and benefits.(127, 133, 138)

Another area requiring further examination is reports from health professionals that the palliative care context is an 'inappropriate' place to discuss genomics.(128) (20) Other work places less emphasis on this and tends to report palliative care health professionals' uncertainty about the role they play in addressing genomics.(218) Additionally, there is a scarcity of evidence from the genetic health professional perspective about how whose role it is to broach and facilitate genetic counselling and testing for patients with palliative care needs.(250) Eliciting genetic and palliative care health professionals' views about their role in integrating genomics into palliative care may enhance the provision of genomics to this population.

To fill these gaps and further existing knowledge about the barriers and facilitators, we aimed to assess and compare the experience, confidence, and attitudes of genetic and palliative care health professionals towards addressing genomics with patients who have palliative care needs and their families.

## 6.5 MATERIALS AND METHODS

We designed a quantitative, cross-sectional survey study that assessed genetic and palliative care health professionals' views towards integrating genomics into the care of people with palliative care needs and their families. We took a broad approach to defining a 'palliative care' setting, by including any setting in which palliative care could be delivered (e.g., community, hospital, hospice). A study protocol is available on the Open Science Framework (https://osf.io/n6dfh; appendix E1). Reporting items align with the STROBE statement (appendix E9).(280) We did not hold a priori hypotheses as this was an exploratory study with limited theoretical or empirical data available on which to base predictions.

### 6.5.1 Participants and Recruitment

Participants were eligible if they were a (a) palliative care health professional or (b) genetic health professional. We defined health professionals as medical doctors, registered nurses, and genetic counsellors. To ensure responses were relevant to clinical practice, participants were required to be currently, or have previously, worked in a clinical area.

We estimated a potential sample size of 1390 participants based on a 30% response rate from the estimate population of genetics and palliative health professionals, which would allow estimates of prevalence with a 95% CI half-width of <1%, and to test for differences in an indicator between subgroups (with at least 500 members) of 10% absolute difference with 0.9 power.

We began recruiting participants through professional organisations in March 2022 and closed the survey on the 21st of July 2022. Three palliative care organisations (Australia and New Zealand Society of Palliative Medicine, Palliative Care Nurses Australia, and Palliative Care Nurses New Zealand) and one genetic organisation (Human Genetics Society of Australasia, including two of its special interest groups: Australasian Society of Genetic Counsellors and Australasian Association of Clinical Geneticists) advertised the survey link to their members via email blasts and online newsletters. Organisations circulated the invitation up to three times. Health professionals self-selected to participate. On the survey landing page, participants selected whether they were a palliative care or genetic health professional, and this directed them to the relevant survey based on their specialty.

### 6.5.2 Instrument

We searched APA PsycTests for relevant validated scales.(281) We identified one scale that assessed hospice nurses' perceptions of the importance of genomics to care and confidence performing 'genomic-related activities'.(103) However, this measure was not designed for genetic and non-genetic health professionals and was therefore not suitable. Instead, we developed two online surveys using REDCap software;(282) one for palliative care health professionals and the other for genetic health professionals (appendices E5 & E6). The survey item development was informed by recent literature review findings,(180, 214) our previous qualitative interviews and focus groups of genetic and palliative care health professionals (*n*=40)(218, 250) and underpinned by the World Health Organization Innovative care for chronic conditions framework.(67) Across the two surveys, most items had the same or similar wording to enable comparison between the genetic and palliative care groups.

Participants were given a modified Likert scale (i.e., never, occasionally, sometimes, usually, or always) to indicate the frequency of performing genomic activities. Previous training and experience were assessed by selecting the most appropriate answer from a predefined list. For some items, response totals are greater than 100% because participants could select more than one option. Likert scales assessed confidence (1=not all confident to 5=confident) and attitudes (1=strongly disagree to 5=strongly agree). A list of previously identified barriers (*n*=18), facilitators (*n*=13), and resources or tools (*n*=12) were provided. Participants were instructed to select up to three responses from each list as their 'main challenges' and 'most helpful' facilitators, resources, or tools. At the beginning of the survey, we defined DNA storage and testing as a clinical activity, rather than research. We were not able to distinguish for each question whether participants were responding hypothetically or from experience.

The survey was piloted with 19 participants (four palliative care and 15 genetic health professionals), of which four participated in a qualitative interview to provide feedback on the survey readability, acceptability, and usability. Participants wanted improved clarity about what was intended by the question "What is your ethnicity?". In response, we replaced this with three additional questions related to country of birth, cultural background and language spoken at home.(283) We also incorporated their suggestion to include a 'not applicable' option to most questions.

### 6.5.3 Data Analysis

After closing the survey, we summarised categorical variables with numbers and percentages stratified by profession (i.e., palliative care health professional or genetic health professional). We compared professions' demographic variables and responses to identical questions about requests for initiation of genomic testing, confidence with genomic activities and attitudes towards integrating genomics into palliative care. Comparisons were made using Fisher-Freeman-Halton Exact Test or Chi-Square Test (using the exact test where there was an expected cell count <5), with statistical significance set at $\alpha<0.05$. For questions that were not designed to be compared, results were described with summary statistics. Where there was item non-response, we used listwise-deletion to deal with missing data. For summary statistics and comparisons, we used SPSS version 28.(284) For visualisations, we used R (version 4.1.3)(264) and ggplot.(265)

### 6.5.4 Ethics

The University of Technology Sydney Human Research Ethics Office granted ethical approval for this study (ETH19-2408/21-5854). The survey landing page provided participants with information about the study (appendix E4). Consent was implied by completion of the survey.

## 6.6 RESULTS

We received 80 responses, of which seven were blank. Emails containing the survey invitation were opened by a maximum of 1,438 potential participants, equalling an approximate response rate of 6%. We were unable to collect reasons for non-participation from non-responders. Eighteen participants provided partially completed responses that we included in the analysis, therefore the frequency counts vary between items.

### 6.6.1 Demographics

Demographic data are presented in Table 12. Of the 73 surveys containing data, 60% (*n*=44/73) were completed by palliative care health professionals and 40% were completed by genetic health professionals (*n*=29/73). Fifty-five participants provided demographic data (75%). Most participants were female (*n*=51/55, 93%), born in Australia (*n*=37/55, 67%), working in the public sector (*n*=43/55, 78%) and in a metropolitan location (*n*=44/55, 80%). There were no significant differences between palliative care and genetic health professionals' gender (*p*=0.624), age (*p*=0.686), years since qualification (*p*=0.74), years of experience in specialist area (*p*=0.367), work sector (*p*=0.316) or location (*p*=0.113). More genetic health professionals held a master's degree than palliative care health professionals (*p*=0.001).

Table 12. Demographic results overall and stratified by health profession.

| Demographic variable | Overall (*n*=55)* | Genetic HP (*n*=24) | Palliative care HP (*n*=31) |
|---|---|---|---|
| | n (%) | n (%) | n (%) |
| **Gender** | | | |
| Female | 51 (93) | 23 (96) | 28 (90) |
| Male | 4 (7) | 1 (4) | 3 (10) |
| Non-binary | 0 (0) | 0 (0) | 0 (0) |
| Prefer not to disclose | 0 (0) | 0 (0) | 0 (0) |
| **Age** | | | |
| 20-24 | 2 (4) | 2 (8) | 0 (0) |
| 25-34 | 12 (22) | 6 (25) | 6 (19) |
| 35-44 | 16 (29) | 7 (29) | 9 (29) |
| 45-54 | 11 (20) | 4 (17) | 7 (23) |
| 55-64 | 13 (24) | 5 (21) | 8 (26) |
| >65 | 1 (2) | 0 (0) | 1 (3) |
| **Country of birth** | | | |
| Australia | 37 (67) | 19 (79) | 18 (58) |
| New Zealand | 2 (4) | 0 (0) | 2 (6) |
| England | 9 (16) | 1 (4) | 8 (26) |
| Scotland | 2 (4) | 1 (4) | 1 (3) |
| Other | 5 (9) | 3 (13) | 2 (6) |
| **Country of work** | | | |
| Australia | 48 (87) | 24 (100) | 24 (77) |
| New Zealand | 7 (13) | 0 (0) | 7 (23) |
| **Cultural background/ethnicity** | | | |
| None | 2 (4) | 1 (4) | 1 (3) |
| Australian | 38 (69) | 19 (79) | 19 (61) |
| New Zealand | 2 (4) | 0 (0) | 2 (6) |
| English | 6 (11) | 0 (0) | 6 (19) |
| Irish | 3 (5) | 1 (4) | 2 (6) |
| Scottish | 2 (4) | 1 (4) | 1 (3) |
| Chinese | 3 (5) | 0 (0) | 3 (10) |
| Other or prefer not to say | 10 (18) | 7 (29) | 3 (10) |
| **Language at home** | | | |
| English | 54 (98) | 23 (96) | 31 (100) |
| Other | 1 (2) | 1 (4) | 0 (0) |
| **Profession** | | | |
| Medical | 20 (36) | 4 (17) | 16 (52) |
| Nursing | 15 (27) | 0 (0) | 15 (48) |
| Genetic Counselling | 20 (36) | 20 (83) | 0 (0) |
| **Highest qualification** | | | |
| PhD | 4 (7) | 2 (8) | 2 (6) |
| Master's degree | 20 (36) | 15 (63) | 5 (16) |
| Bachelor's degree | 19 (36) | 3 (13) | 16 (52) |
| Diploma | 6 (11) | 1 (4) | 5 (16) |
| Professional qualification | 6 (11) | 3 (13) | 3 (10) |
| **Years since qualification** | | | |
| Less than 2 years | 5 (9) | 4 (17) | 1 (3) |
| 2 – 5 years | 3 (5) | 2 (8) | 1 (3) |
| 6 – 10 years | 13 (24) | 5 (21) | 8 (26) |
| 11 – 15 years | 11 (20) | 7 (29) | 4 (13) |
| More than 15 years | 23 (42) | 6 (25) | 17 (55) |
| **Years in specialist area** | | | |

| | | | |
|---|---|---|---|
| Less than 2 years | 9 (16) | 5 (21) | 4 (13) |
| 2 – 5 years | 9 (16) | 3 (13) | 6 (19) |
| 6 – 10 years | 12 (22) | 3 (13) | 9 (29) |
| 11 – 15 years | 11 (20) | 7 (29) | 4 (13) |
| More than 15 years | 14 (25) | 6 (25) | 8 (26) |
| **Work sector** | | | |
| Public | 43 (78) | 18 (75) | 25 (81) |
| Private | 3 (5) | 2 (8) | 1 (3) |
| Public and private | 7 (13) | 2 (8) | 5 (16) |
| Other | 2 (4) | 2 (8) | 0 (0) |
| **Work location** | | | |
| City/metro/urban | 44 (80) | 22 (92) | 22 (71) |
| Regional | 8 (15) | 1 (4) | 7 (23) |
| Rural | 2 (4) | 1 (4) | 1 (3) |
| Other | 1 (2) | 0 (0) | 1 (3) |
| **Work setting** | | | |
| Hospital | 34 (62) | 21 (88) | 13 (42) |
| Hospice | 9 (16) | 0 (0) | 9 (29) |
| Community clinic | 4 (7) | 0 (0) | 4 (13) |
| Home care | 3 (6) | 0 (0) | 3 (10) |
| Independent clinic | 3 (6) | 1 (4) | 2 (6) |
| Other | 2 (4) | 2 (8) | 0 (0) |
| *Demographic information was missing from 18 surveys, therefore *n*=55. | | | |

### 6.6.2 Previous Experience in Genomics and Palliative Care

Palliative care health professionals indicated that they perform the following three activities at least 'occasionally': taking a family health history (*n*=34/43, 79%), drawing a three-generation pedigree (*n*=22/43, 51%) and making a genomic risk assessment (*n*=15/43, 35%). The most common time to take a family health history was when the patient commenced palliative care (*n*=21/43, 49%). Half of the palliative care health professionals (*n*=22/42, 52%) indicated that in the previous year, they had not checked if their patient, or their relatives, had already had an opportunity to discuss genomics before coming under their care.

Most genetic and palliative care health professionals indicated requests for genomic testing come from oncology health professionals (*n*=21/55, 38%), followed by family members (*n*=14/55, 25%). Only 2% (*n*=1/55) indicated that requests for genetic testing come from palliative care health professionals. Many genetic health professionals had been involved with facilitating DNA storage or testing for a person receiving palliative care (*n*=23/28, 82%). Among these participants, the most frequently selected time that they became involved was when the patient was close to death (*n*=11/23, 48%).

### 6.6.3 Previous Training in Genomics and Palliative Care

Almost all palliative care health professionals had not received previous training in genomic risk assessment or testing (*n*=40/44, 91%) but the majority (*n*=30/40, 75%) were interested in receiving training. For those who were not interested (*n*=10/40, 25%), reasons included having other educational priorities (*n*=3/10, 33%), genomics not being relevant to their work (*n*=3/10, 33%), lack of time (*n*=2/10, 20%) or being retired or close to retirement (*n*=2/10, 20%). More than half of the genetic health professionals had previously received training in communicating with patients at end of life or bereaved families (*n*=17/29, 59%). All genetic health professionals without previous training were interested in receiving training (*n*=12/12, 100%).

### 6.6.4 Confidence Integrating Genomics into Palliative Care

A third of palliative care health professionals were 'fairly confident' or 'confident' with contacting their local genetics service (*n*=11/33, 33%) and a quarter were 'fairly confident' or 'confident' assessing when to broach a genomics discussion (*n*=8/33, 24%). Fewer palliative care health professionals were 'fairly confident' or 'confident' identifying patients who were eligible for genomic testing (*n* = 4/33, 12%) and responding to family members' questions about genomics (*n*=4/33, 12%). Most genetic health professionals were 'fairly confident' or 'confident' when communicating with patients (*n*=15/25, 60%) and families (*n*=20/25, 80%) at end of life.

When comparing the two health professional groups (Figure 12), palliative care health professionals reported lower confidence than genetic health professionals when discussing DNA storage (*p*=0.001) or testing (*p*=0.001), facilitating or taking a DNA sample (*p*=0.001), disclosing genomic results to people with palliative care needs (*p*=0.001) or bereaved family members (*p*=0.001) and navigating legal responsibilities when sharing genomic information in the palliative context (*p*=0.003).

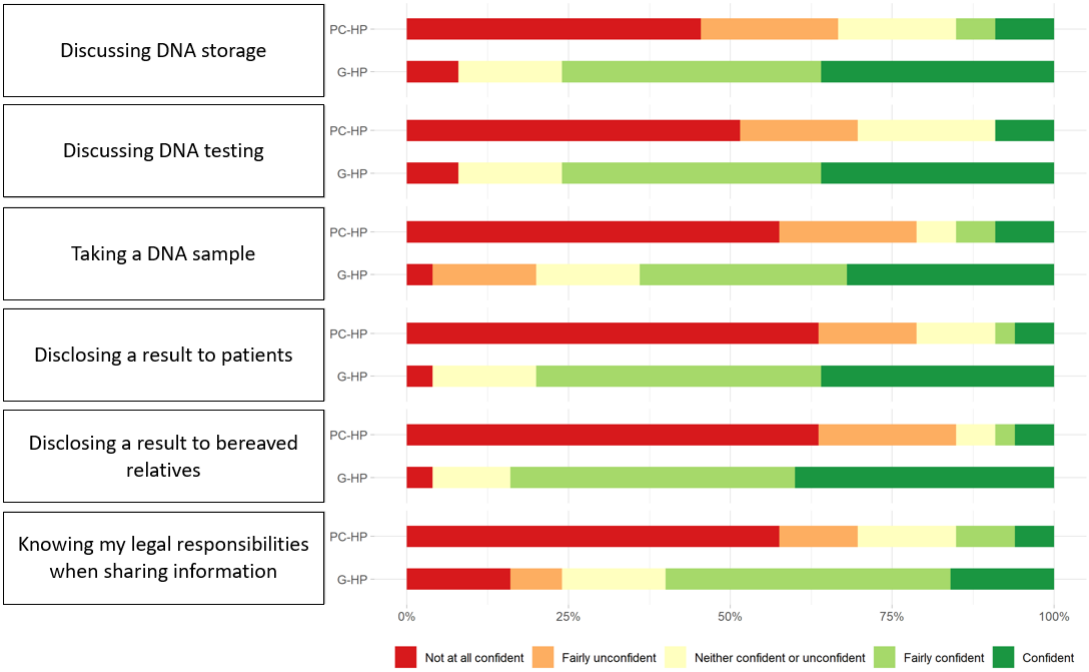


Figure 12. Palliative care (PC-HP, *n*=33) and genetic health professionals' (G-HP, *n*=25) confidence engaging with genomic activities

### 6.6.5 Perceived Barriers and Facilitators

The most frequently selected barrier by genetic and palliative care health professionals was palliative care health professionals' lack of knowledge about DNA storage/testing (Table 13; *n*=32/72, 44%). Genetic health professionals selected the under-referral of people with palliative care needs to genetic services as a barrier more frequently than palliative care health professionals (*p*=0.001). Palliative care health professionals selected 'identifying eligible patients' as a barrier more frequently than genetic health professionals (*p*=0.046).

Genetic health professionals considered fostering closer working relationships between palliative care health professionals and genetic health professionals a more important facilitator than palliative care health professionals (*p*=0.041). Palliative care and genetic health professionals frequently selected the development of a specific referral template as a facilitator (*p*=0.145).

Of nine resources or tools to support palliative-genomic DNA storage or testing, the most frequently selected by both groups was 'support from a specialist genetics service or colleague' (*n*=33/72, 46%), although this was more frequently selected by genetic health professionals (*p*=0.006).

Table 13. Participants' top three perceived barriers, facilitators, and resources or tools they have found useful.

| Item description | Overall (*n*=72) | G-HP (*n*=29) | PC-HP (*n*=43) |
|---|---|---|---|
| | n (%) | n (%) | n (%) |
| **Top 5 barriers** | | | |
| PC-HPs' lack of knowledge | 32 (44) | 13 (45) | 19 (44) |
| Identifying eligible patients | 19 (26) | 4 (14) | 15 (35) |
| Conflicting priorities between providing palliative care and genomic testing | 15 (21) | 6 (21) | 9 (21) |
| Under-referral of people with palliative care needs to genetics | 15 (21) | 12 (41) | 3 (7) |
| Urgency of the situation/referral | 13 (18) | 8 (28) | 5 (12) |
| **Top 5 facilitators** | | | |
| Developing a specific genetic referral template for people with palliative care needs | 31 (43) | 9 (31) | 22 (51) |
| Fostering closer working relationships between PC-HPs and G-HPs | 27 (38) | 15 (52) | 12 (28) |
| G-HPs deliver education to PC-HPs | 25 (35) | 11 (38) | 14 (33) |
| Embedding a genetic counsellor in the palliative care team | 17 (24) | 8 (28) | 9 (21) |
| PC-HPs and G-HPs attend the same multidisciplinary team meetings | 15 (21) | 11 (38) | 4 (9) |
| **Top 5 resources or tools** | | | |
| Support from a specialist genetics service or colleague | 33 (46) | 19 (66) | 14 (33) |
| Support from a palliative care colleague | 15 (21) | 9 (31) | 6 (14) |
| I have not found any resources or tools helpful | 10 (14) | 3 (10) | 7 (16) |
| Other/no experience | 10 (14) | 1 (3) | 9 (21) |
| Clinical decision-making algorithm or guideline | 9 (13) | 3 (10) | 6 (14) |

Items are ranked in order of the most frequently selected across both genetic (G-HP) and palliative care health professional (PC-HP) groups. The full lists of barriers, facilitators & resources or tools are in appendix E8.

### 6.6.6 Attitudes Towards Genomics in Palliative Care

Nearly all the genetic (*n*=23/24, 96%) and palliative care health professionals (*n*=30/31, 97%) 'agreed' or 'strongly agreed' that people with palliative care needs may experience positive emotional benefits from genetic counselling or testing (*p*=1.0; Figure 13)**.** The majority of genetic (*n*=23/24, %) and palliative care health professionals (*n*=30/31, %) 'agreed' or 'strongly agreed' that genetic testing may be important for surviving family members (*p*=0.687). Most genetic (*n*=23/24, 96%) and palliative care health professionals (*n*=25/31, 81%) 'strongly disagreed' or 'disagreed' that discussing DNA storage/testing with people

receiving palliative care undermines the central ethos of palliative care (*p*=0.286). More genetic health professionals disagreed (*n*=12/24, 50%) that DNA storage/testing will have been discussed prior to palliative care than palliative care health professionals (*n*=8/31, 26%, *p*=0.018). Palliative care health professionals (*n*=10/31, 32%) disagreed more frequently than genetic health professionals (*n*=1/24, 4%) that 'palliative care health professionals are well placed to have discussions about DNA storage/testing with family members' (*p*=0.014). Genetic health professionals more frequently disagreed (*n*=13/24, 54%) that privacy and discrimination concerns make DNA storage/testing discussions difficult compared to palliative care health professionals (*n*=6/31, 19%; p=0.01). More genetic health professionals agreed (*n*=10/24, 42%) that palliative care health professionals should revisit genomics discussions with people with palliative care needs if they initially decline compared to palliative care health professionals (*n*=4/31, 13%; *p*=0.039). For the statement 'The family of a palliative patient have a right to know if they are at risk of developing a genetic disease, regardless of the palliative patient's wishes,' the majority of genetic (*n*=10/24, 42%) and palliative care health professionals (*n*=17/31, 55%) neither agreed nor disagreed (*p*=0.562).

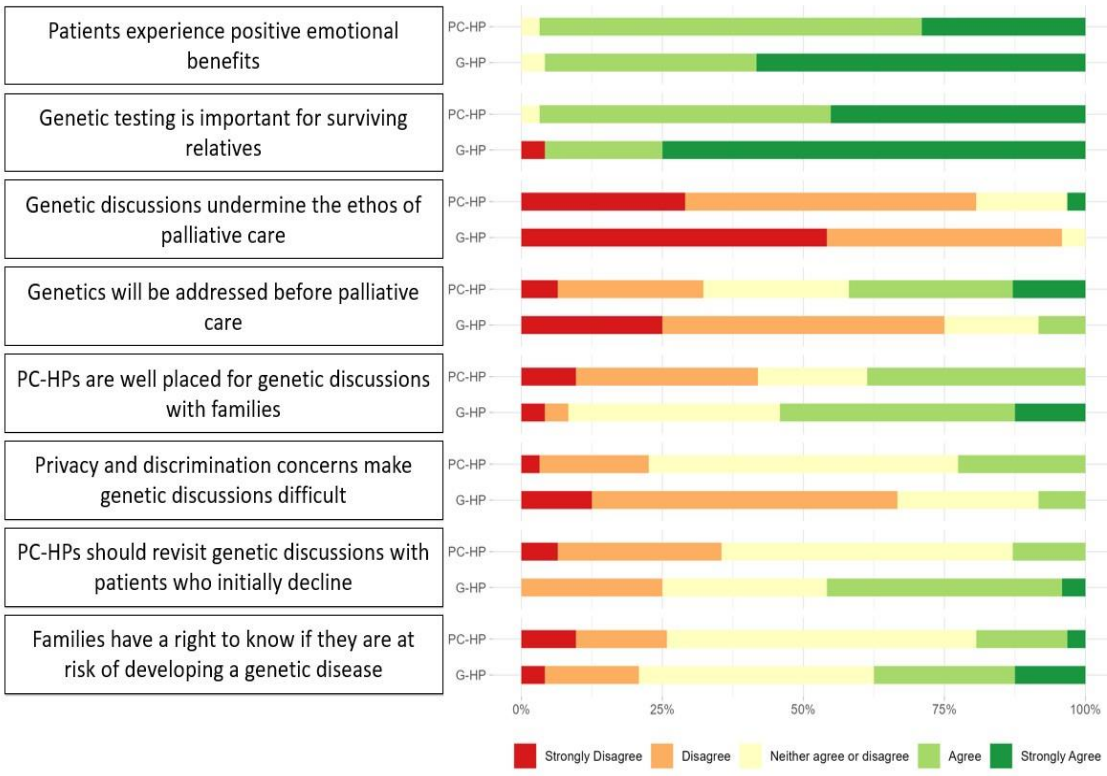


Figure 13. Palliative care (PC-HP, *n*=31) and genetic health professionals' (G-HP, *n*=24) agreement with statements related to the integration of genomics into palliative care

More genetic health professionals disagreed (*n*=12/24, 50%) that DNA storage/testing will have been discussed prior to palliative care than palliative care health professionals (*n*=8/31, 26%, *p*=0.018). Palliative care health professionals (*n*=10/31, 32%) disagreed more frequently than genetic health professionals (*n*=1/24, 4%) that 'palliative care health professionals are well placed to have discussions about DNA storage/testing with family members' (*p*=0.014).

Genetic health professionals more frequently disagreed (*n*=13/24, 54%) that privacy and discrimination concerns make DNA storage/testing discussions difficult compared to palliative care health professionals (*n*=6/31, 19%; *p*=0.01). More genetic health professionals agreed (*n*=10/24, 42%) that palliative care health professionals should revisit genetics discussions with people with palliative care needs if they initially decline compared to palliative care health professionals (*n*=4/31, 13%; *p*=0.039).

## 6.7 DISCUSSION

This survey found that genetic and palliative care health professionals are supportive of integrating genomics into the care of people with palliative care needs and their families, although some differences in opinion regarding the role of palliative care health professionals were noted. We identified knowledge and confidence barriers along with intervention targets, including relationship building between genetic and palliative care health professionals, and potential improvements to referral processes.

Palliative care health professionals reported low levels of confidence when engaging with genomic activities, consistent with previous reports.(113, 133, 138) Our results further current understanding about palliative care health professionals' lack of confidence by identifying potential areas where support may improve capability. For example, a targeted educational approach that focuses on broaching DNA storage and testing discussions, facilitating DNA collection, and understanding the legalities of sharing genomic health information may be of greater support to palliative care health professionals than delivering general genomics education.

Despite their low confidence, most palliative care health professionals would at least occasionally obtain a family health history from their patient. Less frequently, palliative care health professionals were drawing a three-generation pedigree or conducting a genomic risk assessment. One way to support palliative care health professionals' engagement and confidence in pedigree drawing as the basis of a genomic risk assessment may be to leverage

their existing skill and knowledge about genograms. Within palliative medicine, genograms are often used to document family structures, relationships, and other social information.(285) Genetic health professionals' expertise makes them well placed to deliver education about pedigree-drawing and risk assessment to palliative care health professionals. In keeping with previous efforts to upskill non-genetics health professionals, our findings indicated that education delivered by genetic health professionals would be highly valued.(57)

Genetic health professionals indicated that patients with palliative care needs were under-referred to genetic services. At least two audits of referrals to genetic services reported related findings. One audit found that 22% of unaffected relatives referred for risk assessment were received 11 years (on average) after the last affected individual had died.(24) A second audit investigated the referrals of 45 individuals who died while awaiting a genetics appointment. They estimated the health of 133 first-degree relatives was moderately or significantly impacted by their family member failing to receive a genetics appointment.(104) These suboptimal practices may explain why genetic health professionals in our cohort were less likely to think that genomics will have been addressed prior to patients receiving palliative care (i.e., by treating clinicians, such as oncologists) compared to palliative care health professionals.

Another possible explanation, given the close links between palliative care and oncology, is the impact of cancer mainstreaming models upon palliative care health professionals' assumptions about genetic referral practices. For example, it is now common in Australia for gynaecological oncologists to organise germline breast cancer gene testing rather than referring the patient to a genetic service.(57) Palliative care health professionals may therefore assume that all genomic needs will be addressed prior to referral to palliative care, and not consider it to be their responsibility. However, mainstreaming models only target patients with distinct tumour types (e.g., high-grade serous ovarian carcinoma) and there is a lack of mainstreaming in other specialties, such as neurology or cardiology.(286) Furthermore, research has shown that, even when people with cancer receive germline results through mainstreamed testing, oncologists report low confidence explaining implications to family members.(63) If this is the case, we suggest palliative care health professionals check all people receiving palliative cares' genomic needs, particularly for those with 'non-mainstreamed' malignancies or a non-malignant disease. However, our findings suggest palliative care health professionals are not routinely verifying this information.

Our results raise questions about genetic and palliative care health professionals' views on the palliative care role in addressing genomics. In keeping with recent reports, most palliative care health professional participants agreed they had some responsibility to address genomics with their patients, although less certainty was evident when considering their responsibility to family members.(218) If, as our findings show, family members are often initiating genomic discussions, a better understanding of how palliative care health professionals respond to family members queries is needed. As family-centred care is a central tenet of palliative care, it seems appropriate for palliative care health professionals to address family members' genomic concerns or refer them to an appropriate provider for more complex discussions.(99) In contrast, genetic health professionals agreed that palliative care health professionals were well placed for discussions about genomics with family members. We suggest there may be a mismatch between what genetic health professionals expect of palliative care health professionals and what is happening in practice. Incorporating communication about genomics to family members may therefore also be an important topic to include in an educational intervention. Future research to understand palliative care health professionals' views and experiences of communicating about genomics with family members, as opposed to patients, would provide a valuable insight into the content of these discussions, reasons for discomfort and avenues for support.

Our results did not confirm previous concerns from palliative care health professionals about the potential harms of addressing genomics, such as a negative psychological impact to patients and relatives.(287) The benefits of genomic information for people with palliative care needs and their families were almost unanimous. Participants rejected the idea that the palliative care context is an 'inappropriate' place to discuss genomics, which contrasts with previous qualitative work.(128, 218) Although we do not discount these potential harms, our results may reflect a shift in attitudes as genomics in routine medical care becomes more widely accepted.(38)

While a referral template was selected most frequently as a facilitator, genetic health professionals were also interested in interventions that preceded the point of referral. The facilitators supported by genetic health professionals are similar to interventions implemented in various mainstreaming models, including fostering collaborative relationships, embedding a genetic counsellor in the palliative care team, and multidisciplinary team meetings.(58, 59) It is possible that genetic health professionals were more likely to emphasise these 'collaborative' interventions because of a belief that genomics is not valued by palliative care health

professionals.(250) Genetic health professionals may view collaborative working as an opportunity to educate palliative care health professionals about the familial benefits of genetic counselling and testing, in addition to facilitating referrals. Interestingly, our findings do not support the suggestion that palliative care health professionals do not value genetic counselling and testing. Rather, palliative care health professionals simply desire practical and educational support.

## 6.8 STRENGTHS AND LIMITATIONS

Although several advertisements were sent through national organisations, our sample was small and self-selected, so may be subject to non-response/selection bias. The validity of comparisons would have been improved with larger cohorts. Our findings may not be generalisable (that is, the data presented here may not represent their source groups) or represent diverse attitudes towards integrating genomics into palliative care. Further work to understand reasons for the low response rate could provide valuable insights, including whether the topic was perceived as unimportant or if it were a result of survey fatigue (several participants dropped out half-way through the survey).

Our ability to conduct the planned statistical analysis, including ordinal logistic regression for Likert responses, was also impacted by the small sample size. Furthermore, though participants reported their engagement with genomic activities, such as taking a family history, we were unable to determine the quality or content of these activities.

The professional organisations who circulated invitations to the members for participation were only able to share limited demographic summaries. For example, one organisation could only share the number of members who were qualified or in-training. As a result, we could not reliably assess the representativeness of our sample. Genetic health professionals were more likely to hold a master's degree than palliative care health professionals. This could be explained by the proportion of genetic counsellors in the sample, for whom a Master degree has been the entry level qualification since 2010.(47)

Despite these limitations, our evidence begins to fill the thematic and methodological literature gaps in this understudied area. Further work with patients who have palliative care needs, and their families, would be likely to identify additional barriers and facilitators to understand and support integration of genomics into their care.

## 6.9 CONCLUSION

Genetic and palliative care health professionals both support the integration of genomics into the routine care of people with palliative care needs and their families. Building the confidence of palliative care health professionals through the delivery of education by genetic health professionals, inter-specialty collaboration, and development of a specific genetic referral template is an important first step. Defining the role of palliative care health professionals in addressing genomics with family members requires further work. Our findings shine a light on the existing barriers and facilitators to integrating genomics into the care of people with palliative care needs with a view to developing targeted interventions. In doing so, the benefits of genetic counselling and testing can be realised by patients with palliative care needs and their families.

## 6.10 EXTENDED METHODS

The numerical data presented in this extended section were analysed in the same way as the main body results. Categorical variables are presented as numbers and percentages stratified by profession. Participants were asked to rate their level of 'comfort' with palliative care health professionals performing six different actions related to DNA storage at the end of -life using a 5-point Likert scale from 'very uncomfortable' [1] to 'very comfortable' [5]. Comfort is a complex, multifaceted concept that has previously been used to measure health professional behaviour in morally challenging scenarios.(288) The Fisher–Freeman–Halton exact test or chi-square test (using the exact test where there was an expected cell count <5) were used to compare results between health professional groups. An $\alpha$ value < 0.05 was considered significant. For items with no response, I used listwise deletion to deal with missing data. IBM SPSS Statistics (version 28),(29) R (version 4.1.3),(30) and ggplot(31) were used for statistical analyses and visualisation.

After rating their level of comfort with the DNA storage actions on the Likert scale, participants were invited to write a free text answer in response to "…any further thoughts you have about palliative care health professionals performing these actions." Participants could respond in their own words and there was no word limit. I analysed the responses using content analysis.(289 p243-72) I categorised the responses into concepts based on key words or phrases, such as 'education'. Responses could be categorised into more than one category. The number and content of each concept are presented narratively.

## 6.11 EXPLORATORY FACTOR ANALYSIS

At the conclusion of data collection, the possibility of conducting an Exploratory factor analysis (EFA) was discussed among the study team, which included a biostatistician. EFA is an analytical method used to test the psychometric properties of a newly developed scale or measure. EFA tests the appropriateness of combining individual data items (or questions in a questionnaire) into theoretical constructs, called 'latent' or 'unobservable' factors.(290) EFA can be used to determine which items have a high degree of co-variance and are therefore explained by the same underlying factor. Eigenvalues and scree plots are then used to determine the number of factors that explain a group of variables is assessed, and these factors can then be developed into subscales for future use. There are several reasons one may consider using EFA, including the development of theoretical constructs, simplifying data sets, evaluating construct validity, and determining the relationship between different variables.(291)

The reason for considering EFA in this study was to reduce the number of variables, simplify the data set, and improve data analysis and interpretation. There were two reasons for wanting to simplify the *whole* data set post hoc; that is, EFA was considered for data from the entire survey and not just the data presented here in this extended section. Firstly, the data set showed that several participants dropped out of the survey at the half-way point, which could indicate that the survey was too long. The addition of items to the original questionnaire (as described in Chapter 3) meant there were more items than when initially piloted. EFA could have been used to reduce the number of items into underlying factors and doing so would have improved the survey for future use. Secondly, interpreting individual data items was difficult because the items had not been conceptualised as constructs when the survey was developed. Although some items grouped easily into theoretical concepts (e.g., items related to previous training or education), others were more difficult (e.g., items related to attitudes). EFA could have been used to improve the confidence that the grouped items had a high degree of co-variance and were related to the same construct (an indicator of construct validity).

Ultimately, EFA was deemed not to be feasible because of two issues with the data set. Firstly, the sample size in this study was too small. The recommended sample size required for EFA varies, and some sources state that >200 participants are required.(291, 292) Others quote minimum sample sizes based on the number of items, such as a ratio of 10:1, meaning

10 participants are needed for each item.(293 p355-6) In either case, the sample size in this study was not powered adequately for EFA. Secondly, EFA had not been built into the design of the survey. Evaluating groups of items as constructs requires a dedicated effort to develop these constructs as part of the survey design.(294) Theoretical knowledge about different constructs and input from a variety of stakeholders should be integrated beforehand. In future studies, I will assess whether EFA should be built into the study design at an early stage in consultation with a biostatistician instead trying to use it as a post hoc solution to reduce the number of data items and simplify interpretation.

## 6.12 EXTENDED RESULTS

The results within this section were written for this thesis and are not a part of the manuscript in this chapter. These findings build upon the extended results presented in Chapter 4 (section 4.9).

### 6.12.1 Assessing Comfort with a DNA Storage Approach to People Near the End of Life

Six items were used to assess genetic and palliative care health professionals' comfort with palliative care health professionals performing actions related to DNA storage (Figure 14). More genetic health professionals indicated they were 'somewhat' or 'very comfortable' (*n*=23/24, 96%) with palliative care health professionals introducing DNA storage to the patient or family than were palliative care health professionals (*n*=15/31, 48%, *p*<.001). More genetic health professionals indicated they were 'somewhat' or 'very comfortable' (*n*=21/24, 88%) with palliative care health professionals obtaining consent for DNA storage than were palliative care health professionals (*n*=12/31, 39%, *p*<.001). More genetic health professionals indicated they were 'somewhat' or 'very comfortable' (*n*=24/24, 100%) with palliative care health professionals facilitating DNA sample collection than were palliative care health professionals (*n*=14/31, 45%, *p*<.001). More genetic health professionals indicated they were 'somewhat' or 'very comfortable' (*n*=23/24, 96%) with palliative care health professionals organising for the DNA sample to be stored than were palliative care health professionals (*n*=12/31, 39%, *p*<.001). More genetic health professionals indicated they were 'somewhat' or 'very comfortable' (*n*=23/24, 96%) with palliative care health professionals instructing the family how to follow-up with the genetic service than were palliative care health professionals (*n*=17/31, 55%, *p*<.001). More genetic health professionals indicated they were 'somewhat' or 'very comfortable' (*n*=22/24, 92%) with palliative care health professionals communicating the

follow-up plan to the genetic service than were palliative care health professionals (*n*=17/31, 55% *p*=.003).

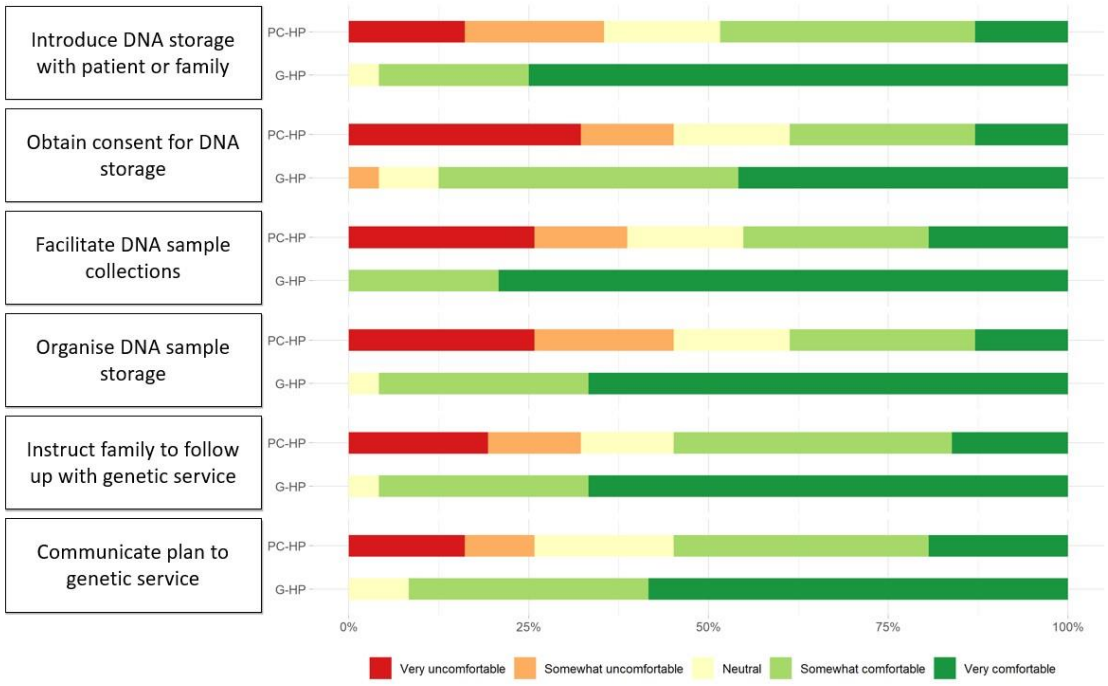


Figure 14. Genetic (G-HP; *n*=24) and palliative care (PC-HP; *n*=31) health professionals' comfort with PC-HPs performing each step of the DNA storage approach for people at the end of life

### 6.12.2 Free Text Responses to the DNA Storage Approach

Of the participants who completed the DNA storage Likert scale, 11 palliative care health professionals (*n*=11/31, 34%) and eight genetic health professionals (*n*=8/24, 33%) responded to the free text response that asked participants for further comments about a DNA storage approach to care for people near the end of life.

Six palliative care health professionals detailed their discomfort with performing actions related to DNA storage. They indicated they were not currently comfortable performing these actions but expected their comfort levels would improve if they received relevant education. One participant explained they did not think DNA storage should be part of their role, one was unsure about the legalities and practicalities of post-mortem genomic testing, and one was concerned about managing testing for the family after their patient had died. Two other participants commented that they hoped genomics would be discussed before the end-of-life phase.

All eight comments from the genetic health professionals were in support of a DNA storage approach. Five participants considered palliative care health professionals well placed to

initiate discussions about DNA storage. Two supported the approach because it removed pressure from families and health professionals to organise genomic testing "at a difficult time". Two thought palliative care health professionals may need additional education to facilitate DNA storage and that working collaboratively would better serve patients with palliative care needs and their families. One participant highlighted the importance of genetic services being responsible for the follow up and management of families.

## 6.13 EXTENDED DISCUSSION

DNA storage could be offered to people near the end of life as an alternative to organising genomic testing. Genetic health professionals support this approach because it reduces the burden of conveying complex information to vulnerable patients and families, and postpones complex discussions until after the palliative person has died when families may be ready to discuss genomic testing.(250)

These findings lend further support to DNA storage being the preferred approach to end-of-life care by genetic health professionals. Their comfort in delegating DNA storage to the palliative care team may reflect their belief in palliative care health professionals' expertise in facilitating difficult discussions with dying people and their families.(250) Genetic health professionals may also be willing to delegate this task because of the difficulty navigating genomic discussions near the end of life, particularly when they are introduced to individuals and families for the first time in this setting.(250)

Palliative care health professionals are less comfortable with managing DNA storage for dying people, although this discomfort could be explained by low knowledge of DNA storage processes. Some palliative care health professionals may not be familiar with DNA storage, and this survey may have been their first encounter with the concept.(133) DNA storage was not mentioned by palliative care health professionals in Phase 1 of the GIFT Project.(218). Collaborative professional relationships, as suggested here, have been noted previously by both genetic and palliative care health professionals as providing support for them to meet the genomics needs of people with palliative care needs and their families.(218, 250) Further research to explore the perceptions of palliative care health professionals about DNA storage may help to elucidate how this approach aligns within their professional role and goals.

Despite the purported benefits of DNA storage, there are several potential issues. Taking biological samples for DNA storage may inadvertently mislead families to believe genomic

testing has occurred and an assumption their disease risk is low if they are not contacted with a result. Other families that are ineligible for publicly funded testing could be inappropriately referred by palliative care to the genetic service, leading to frustration. Additionally, clear pathways for DNA storage need to be devised so that genetic and palliative care health professionals, people with palliative care needs, and their families have a common understanding of the process.(36) Referral pathways for family members will need to be established. All of these, and other issues, should be the focus of future research.

A limitation of these findings is that without further context or explanation, genetic and palliative care health professionals may have interpreted the items about the DNA storage approach differently. Incorporating EFA into the design of these items may have improved the construct validity. Cognitive interviewing or further pilot testing may have increased the reliability and validity of the survey.(295) In future, a more detailed proposition about DNA storage could be co-designed with relevant stakeholders including, but not limited to, people with palliative care needs, families, and genetic and palliative care health professionals. Testing this proposition would help to elucidate details about the responsibility of facilitating the DNA storage approach, where collaboration could be helpful between palliative care and genetic health professionals, and the process of ongoing follow-up and management of families.

## 6.14 EXTENDED CONCLUSION

DNA storage has potential as an approach to care for people near the end of life and their families. Genetic health professionals are more comfortable with a palliative care-led DNA storage approach than are palliative care health professionals. Supporting genetic and palliative care health professionals with interdisciplinary collaboration and education could help to bridge the knowledge and practice gaps. Further research to explore palliative care health professionals' views and incorporate the views of stakeholders will help to build the necessary detail into a proposed DNA storage approach.

## 6.15 CHAPTER SUMMARY

In this chapter, a peer-reviewed manuscript of a quantitative, online, survey study was presented. The findings showed that genomics in palliative care is generally supported by genetic and palliative care health professionals but that there are barriers to its effective integration. Implementing a palliative care-led DNA storage approach has potential, but

further work is needed to understand the feasibility and acceptability. In the next and final chapter, the data from Phase 1 and Phase 2 are integrated to generate meta-inferences. The research questions are answered and discussed in the context of the relevant literature. The strengths and limitations of the GIFT Project are explored, and the thesis concludes with several recommendations for clinical practice and future research.

## 6.16 MEETING OBJECTIVE 2: COMPARE THE PERCEPTIONS OF THE IDENTIFIED BARRIERS AND FACILITATORS BETWEEN GENETIC AND PALLIATIVE CARE HEALTH PROFESSIONALS

The study in this chapter has met the second objective of the GIFT Project. Phase 2 has now concluded. The quantitative survey assessed and compared perceptions of the barriers and facilitators between genetic and palliative care health professionals.(296) The inferences generated from this study in relation to the thesis research questions are addressed in Chapter 7.

# 7 Discussion: Supporting the Integration of Genomics into Palliative Care

## 7.1 PREAMBLE

In Chapter 7, I begin by answering the research questions set out in Chapter 1. I provide a high-level summary of questions 1 and 2 (with further detail in corresponding chapters). Research question 3 is answered using the meta-inferences generated through the integration of the qualitative and quantitative data produced in Phases 1 and 2. I discuss the meta-inferences in the context of the literature and then examine the strengths and weaknesses of the Genomic Information for Families of the Terminally ill (GIFT) Project. I present recommendations for clinical practice and future research, discuss the contribution of this thesis to the field, and conclude with a chapter summary and thesis conclusion.

## 7.2 ANSWERING THE RESEARCH QUESTIONS

In Chapter 1, three research questions were defined. The questions were answered through the completion of a corresponding objective (as detailed in Chapter 3), which aligned to a particular phase of the GIFT Project. Research question 1 was answered through the qualitative exploration of the perceptions of genetic and palliative care health professionals about the barriers and facilitators (Chapter 4), and the scoping review's exploration of policy recommendations (Chapter 5). Research question 2 was answered by quantitatively comparing the barriers and facilitators between genetic and palliative care health professionals (Chapter 6). Research question 3 is answered in this chapter through a presentation of the four meta-inferences produced through the end-point integration of the qualitative and quantitative data generated in Phases 1 and 2. A high-level summary is provided for research questions 1 and 2. Though there is some conceptual overlap, the answers to question 1 and 2 are intended to be descriptive, while the answer to question 3 is action-oriented.

### 7.2.1 Research Question 1: What are the Barriers and Facilitators for Genetic and Palliative Care Health Professionals Towards Integrating Genomics into the Care of People with Palliative Care Needs?

The end-of-life situation challenges genetic health professionals to uphold patient autonomy where the primary benefit for genomic testing is for family members. Genetic health professionals approach the concept of family-centred care through a genomics lens, meaning the primary purpose of offering genomic testing is for the benefit of family members rather than the person with palliative care needs. DNA storage is preferred over genomic testing to overcome some of the perceived practical and ethical tensions when addressing genomics with people who are facing the end of life. Genetic health professionals are inadequately supported by their organisations to overcome the challenges of integrating genomics into the care of people with palliative needs.

Palliative care health professionals weigh the benefits and harms of genomic information when people are near the end of life. The primary concern is the individual's needs, rather than prioritising the family's need for genomic information. Palliative care health professionals want better support from their healthcare organisations to facilitate interdisciplinary care and educational opportunities. Currently, palliative care health professionals are shouldering the responsibility of genomics integration without adequate organisational support.

The policy environment does not appear to be conducive to health services allocating funding and resources to support genetic or palliative care health professionals to integrate genomics into their practice. Family-centred care is a shared clinical and professional goal between palliative care and genetic health professionals.

### 7.2.2 Research Question 2: How do the Identified Barriers and Facilitators Compare Between Genetic and Palliative Care Health Professionals?

Similarities and differences in the perceptions of the barriers and facilitators were identified between genetic and palliative care health professionals. Both groups value genomic information for people with palliative care needs and their families but see palliative care health professionals' lack of genomic knowledge as a barrier to integration. Developing a referral template was suggested by both groups. Palliative care health professionals have less confidence than genetic health professionals in addressing their patients' genomic needs, although genetic health professionals see palliative care health professionals as well placed to have these genomic discussions. Genetic health professionals support palliative care health

professionals facilitating DNA storage, but palliative care health professionals appear to be uncomfortable with this responsibility. Further education about DNA storage is desired.

### 7.2.3 Research Question 3: What is Required to Support Genetic and Palliative Care Health Professionals to Integrate Genomics into the Care of People with Palliative Needs and Their Families?

To answer the third research question, an end-point joint display table (appendix B2) was used to generate meta-inferences through the integration of qualitative and quantitative data from Phases 1 and 2 of the GIFT Project (Figure 15).(218, 250, 296, 297) The process of generating meta-inferences also met the corresponding research objective (to identify what support is required). Integrating the high-level inferences across individual studies underpinned the iterative development of meta-inferences related to the support required to integrate genomics into the care of people with palliative care needs and their families. Further detail about the methods of the end-point data integration was provided in Chapter 3. In the next section, I describe the meta-inferences and answer research question 3.

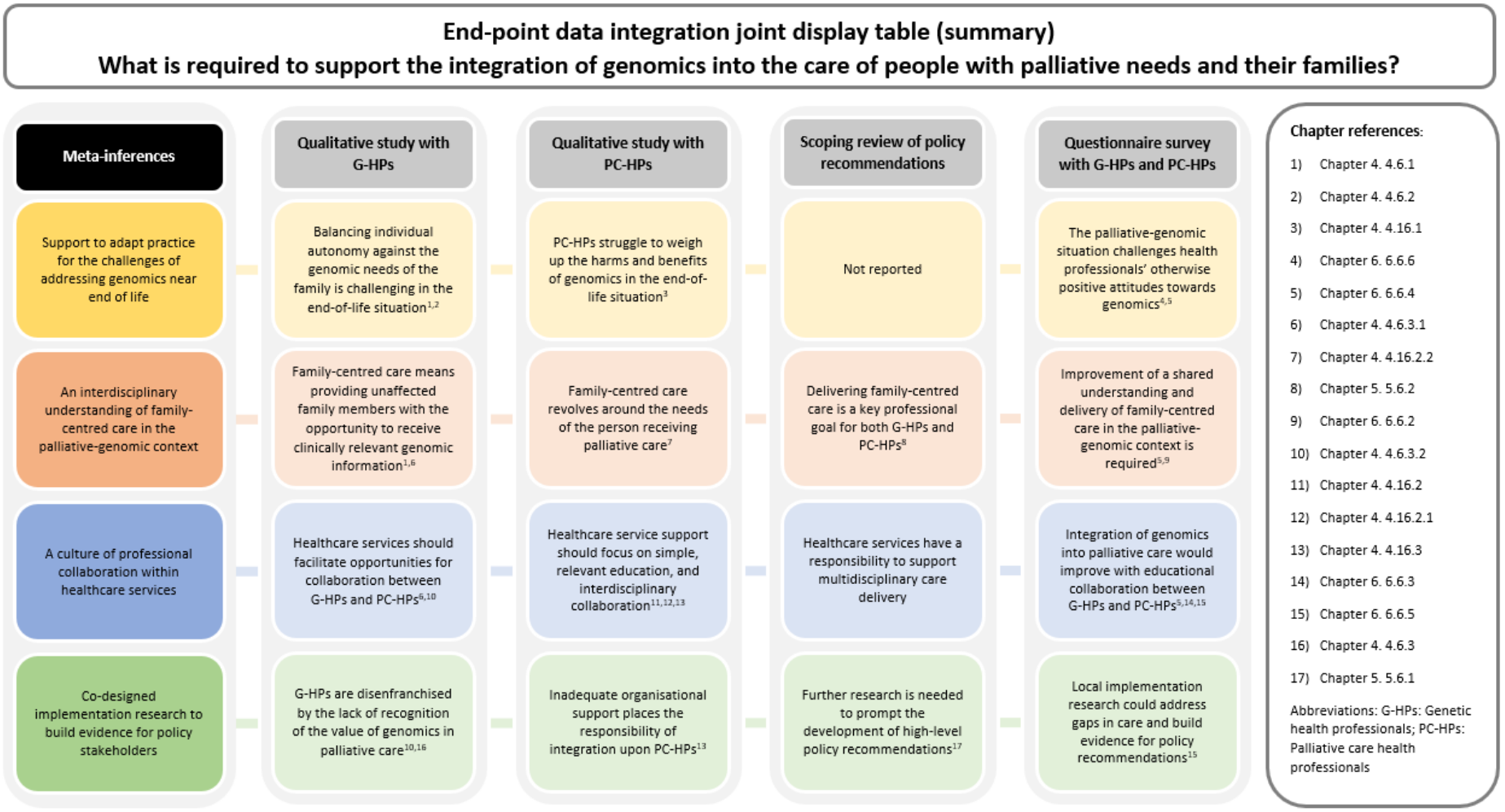


Figure 15. Summary of the joint display table (full table in appendix B2). Inferences from each study are integrated to develop the meta-inferences to answer research question 3.

## 7.3 WHAT IS REQUIRED TO SUPPORT THE INTEGRATION OF GENOMICS INTO PALIATIVE CARE?

Four meta-inferences illustrate the support required for genetic and palliative care health professionals to integrate genomics into the care of people with palliative care needs and their families. Although there is some conceptual overlap between the findings, each finding broadly aligns to the micro-, meso-, or macro-level of the World Health Organization Innovative care for chronic conditions (WHO ICCC) framework (Figure 16).(67) As these findings are underpinned by the integration of Phases 1 and 2, the individual studies within Phases 1 and 2 are not cited in the following meta-inference sections.

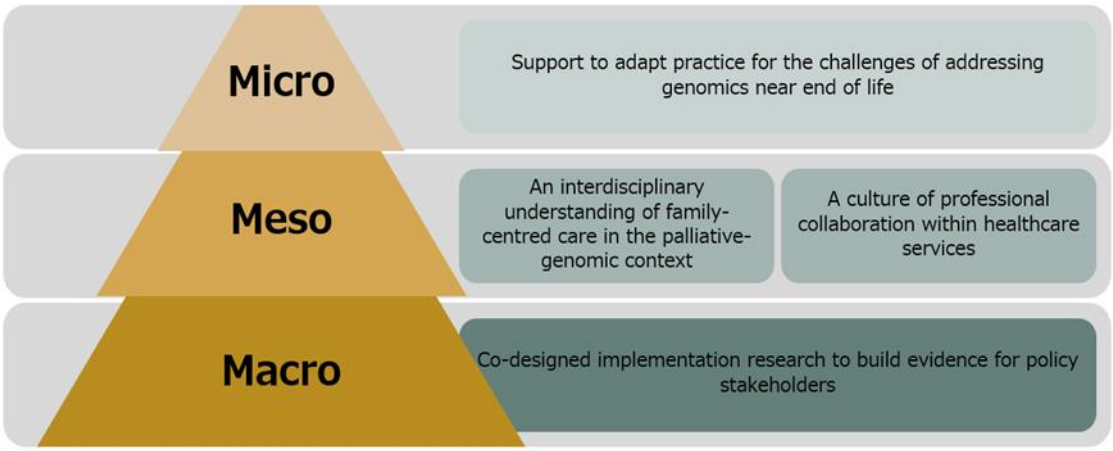


Figure 16. The four meta-inferences related to research question 3 align with a different level of the World Health Organization Innovative care for chronic conditions framework.

### 7.3.1 Meta-Inference 1: Supporting Practice Adaptions to Address Genomics Near the End of Life

Genetic and palliative care health professionals need support to adapt to the challenges of addressing genomics for people nearing the end of life and their families. The associated pressures, such as limited time and inability to engage in complex discussions with the person receiving palliative care require health professionals to navigate changes to their usual practice and subsequent bioethical tensions.

Genetic health professionals truncate their interactions with dying people by confining discussions to what they perceive as pertinent information. This contrasts with usual practice, whereby genetic health professionals obtain informed consent through in-depth discussions of individualised benefits and limitations of genomic testing. Exploring the values and wishes of

individuals and families is the backbone to informed decision making about proceeding with or declining testing. However, reduced capacity to engage in complex genomic discussions means informed consent from a dying person may not be possible. The inability of genetic health professionals to practise in their usual way generates uncertainty about whether genomic testing is in the person's and family's best interests. Despite this tension, genetic health professionals believe genomic testing should be offered, even when autonomy cannot be guaranteed, to provide clinical benefit to relatives. Genetic health professionals viewed family members as 'patients in waiting' and demonstrated a clinical responsibility to ensure relatives had accurate information about their own risk. Balancing the clinical benefit of genomic testing to family members over the autonomy of the patient is a key ethical dilemma for genetic health professionals. Guidance and education about the end-of-life situation may help genetic health professionals uphold the autonomy of a dying patient in the context of familial genomic benefit.

When palliative care health professionals decide whether to broach a genomics discussion, they do so with an uncertainty of whether this contradicts their clinical role and goals. Palliative care health professionals often navigate sensitive discussions at the end of life. Any decision, genomics related or not, is in pursuit of palliative care health professionals' primary clinical goal to improve a person's quality of life and relieve suffering. Concerns stemmed from uncertainty about whether a genomics discussion warranted the potential harm to individuals and families. The relevant ethical principles are beneficence (that genomics will provide benefit) and non-maleficence (that genomics will not cause harm). For palliative care health professionals, the uncertainty about whether the benefits of genomic information outweigh the potential burden inflicted on individuals requires a detailed understanding of the benefits and limitations of genomic testing. Palliative care health professionals' low genomics knowledge impacts on their ability to make this decision confidently. Therefore, it may be that low knowledge and confidence about genomics influences palliative care health professionals' ethical decision making when broaching genomics with people near the end of life. Education about the clinical relevance, benefits, and harms of genomic testing may help palliative care health professionals navigate the bioethical tensions experienced in the palliative–genomic situation.

Without guidance to navigate these adaptions, people receiving palliative care and their families are at risk of receiving inconsistent care. Although some genetic and palliative care health professionals may be able to overcome these challenges on their own, guidance and

education to promote safe adaptions for the end-of-life situation will support health professionals to provide consistent, evidence-based care. Further work to define the exact nature of this support, and who will be responsible for delivering it, should be explored in each unique context (e.g., hospital, department/s, hospice, community practice). The appropriate type, level, and source of support will depend on a variety of factors at the micro-, meso-, and macro-levels. The barriers and facilitators identified in the GIFT Project are a starting point for local implementation research to design future interventions targeted at the right barriers. The bioethical tensions and practice adaptions for genetic health professionals differ from the tensions and adaptions for palliative care health professionals. Support must be tailored to the unique needs of each speciality. At a minimum, clinical practice guidelines should be developed to validate the end-of-life challenges and to communicate the expectations of health professionals and services. Ideally, health services should be responsible for identifying support needs; however, in the short-term, health professionals and/or researchers (individuals or groups) may need to lead this assessment.

### 7.3.2 Meta-Inference 2: An Interdisciplinary Understanding of Family-Centred Care in the Palliative–Genomic Context

Providing family-centred care is a key tenet of genetic and palliative care delivery. However, there is currently no shared understanding of family-centred care in the palliative–genomic context (Figure 17). For genetic health professionals, family-centred care means attending to the genomic needs of the family. Genetic health professionals describe a clinical responsibility towards family members because they provide genetic counselling to families after the person receiving palliative care has died. This may be why genetic health professionals were in support of again offering a genomics discussion, even if the individual had declined initially. Genetic health professionals want families to have access to genomic information that has clinical relevance for them.

For palliative care health professionals, delivering family-centred care revolves around the needs of the palliative patient. Supporting family members recognises that the well-being of the family is an important aspect of the patient's health. Palliative care health professionals' approach is to centre their patient's medical and psychological well-being in the context of their family rather than focus on the family's needs. Palliative care health professionals seem less likely to focus on the genomic needs of individual family members, particularly if these contradict the wishes of their patient. Palliative care health professionals are concerned about

causing psychological harm by insisting on a genomic discussion for the benefit of the family. In some cases, conversations are avoided altogether.

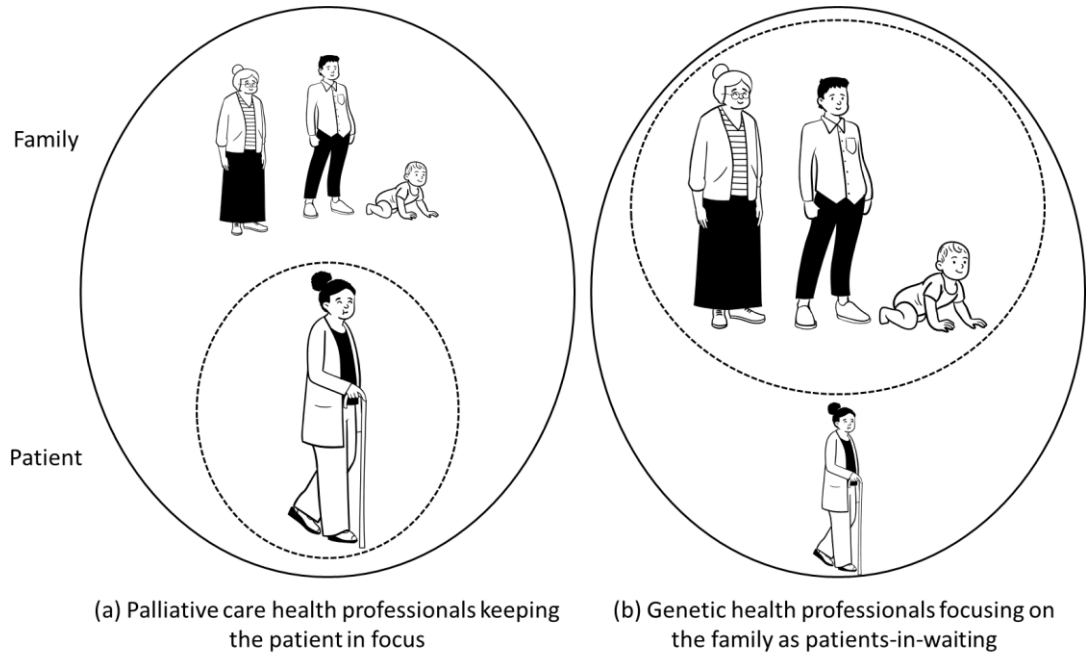


Figure 17. Differences in the conceptualisation of family-centred care according to (a) palliative care health professionals and (b) genetic health professionals. The health professionals' primary focus is illustrated by the dotted lines.

There is the opportunity to develop a shared, interdisciplinary understanding of what family-centred care means in the palliative–genomic context to meet the individual's and family's genomic needs, and to define the roles and responsibilities of genetic and palliative care health professionals. Palliative care health professionals' role in the delivery of genomics could become illuminated through jointly defining the boundaries of their genomic responsibility towards patients and their family members. Genetic health professionals could explore how prioritisation of the family's genomic needs could impact the palliative person and their family. In addition, a shared understanding could underpin the development of a future intervention by aligning genetic and palliative care heath professionals' goals. Collaborative activities between genetic and palliative care health professionals across clinical, academic, and policy settings should focus on family-centred care as a shared mental model while working towards refinement of family-centred care in the palliative–genomic setting.

### 7.3.3 Meta-Inference 3: A Culture of Professional Collaboration Within Healthcare Services

Improving professional collaboration and relationships will help to foster interdisciplinary collaboration between genetic and palliative care health professionals. There appears to be underutilisation of cross-boundary genetic and palliative care expertise to improve the delivery of genomics to people receiving palliative care and their families. Individual health professionals are under-resourced to do this and require dedicated support from their health service.

Developing a collaborative working culture requires attention at the organisational level. Delivering multidisciplinary care supports equity and access of care to individuals and families. Several strategies could be applied by health services to foster collaboration, such as allocating funding for a genetic counsellor to be embedded within a palliative care team, identifying a palliative care health professional as a 'genomics champion' (i.e., someone who encourages and educates their peers about genomics), or encouraging both genetic and palliative care health professionals to attend the same multidisciplinary meetings. The appropriate intervention for each health service will need to be established by implementation research at the local level.

Health services could support collaborative interactions alongside traditional educational interventions. Genetic and palliative care health professionals value the sharing of knowledge through formal or informal interdisciplinary discussions. Palliative care health professionals prefer education that is practical, clinically relevant, and accessible in their existing forums, rather than in-depth education about theoretical genomic concepts. As opposed to attending formal lectures or courses about genomics, informative, collaborative interactions may therefore be more acceptable to palliative care health professionals. Health services could trial interdisciplinary collaboration as a low-stakes initiative before committing funding or resources to more complex or involved educational interventions. As the interdisciplinary relationships progress, formal educational opportunities may be more acceptable. Bringing together genetic and palliative care health professionals with existing bonds into a formal educational opportunity, such as a workshop, could continue to build professional relationships and develop a shared understanding of family-centred care.

### 7.3.4 Meta-Inference 4: Co-Designed Implementation Research to Build Evidence for Policy Stakeholders

Generating evidence for policy stakeholders could begin to be addressed by local implementation research. The barriers and facilitators identified in the GIFT Project can be assessed in the local context to design and trial a suitable intervention. Involvement of an implementation scientist will provide the opportunity and expertise to measure outcomes related to the uptake and success of the intervention alongside outcomes related to clinical benefit. Small-scale implementation research can have a real-world impact for patients, families, and health professionals, create awareness at the local and organisational level, generate momentum for further funding, and contribute to the growing evidence of the value of integrating genomics into the care of people with palliative care needs and their families.

Health professionals are currently shouldering the responsibility of integrating genomics into the care of people with palliative needs and their families. A lack of organisational support places health professionals under pressure to overcome individually the challenges faced in the palliative–genomic setting. Health professionals desire dedicated support from their health services to improve the delivery of genomics to people with palliative care needs and their families as part of the broad genomics implementation landscape.

An absence of high-level policy related to the integration of genomics into palliative care is likely to contribute to the lack of organisational support. With high-level policy recommendations, health services may be more likely to provide dedicated support to health professionals. However, changes to policy recommendations require a suite of evidence demonstrating the value of genomics to people with palliative care needs and families. Several evidence gaps remain, including knowledge of the economic, clinical, ethical, legal, and social benefits of genomics in palliative care. Building evidence to lobby policy stakeholders for change will require long-term research engagement.

Research should be conducted by interdisciplinary teams and co-designed with a variety of stakeholders to capture diverse needs. In particular, the voices of patients with palliative care needs and their families are a critical evidence gap. Discussions about genomics with people with palliative care needs and families, particularly at the end of life, have the potential to upset or harm people who are in a vulnerable situation. Exploring the needs of patients and families, including how, when, or if discussions about genomics occur in the palliative care context, will guide the design of future research and intervention design.

## 7.4 DISCUSSING THE MAIN FINDINGS IN THE CONTEXT OF EXISTING THEORY AND KNOWLEDGE

Several concepts and theories are relevant to the findings in the GIFT Project, including genetic counselling and palliative care theory, ethical frameworks, behaviour change theory, and evidence from broader genomic implementation projects (Figure 18).

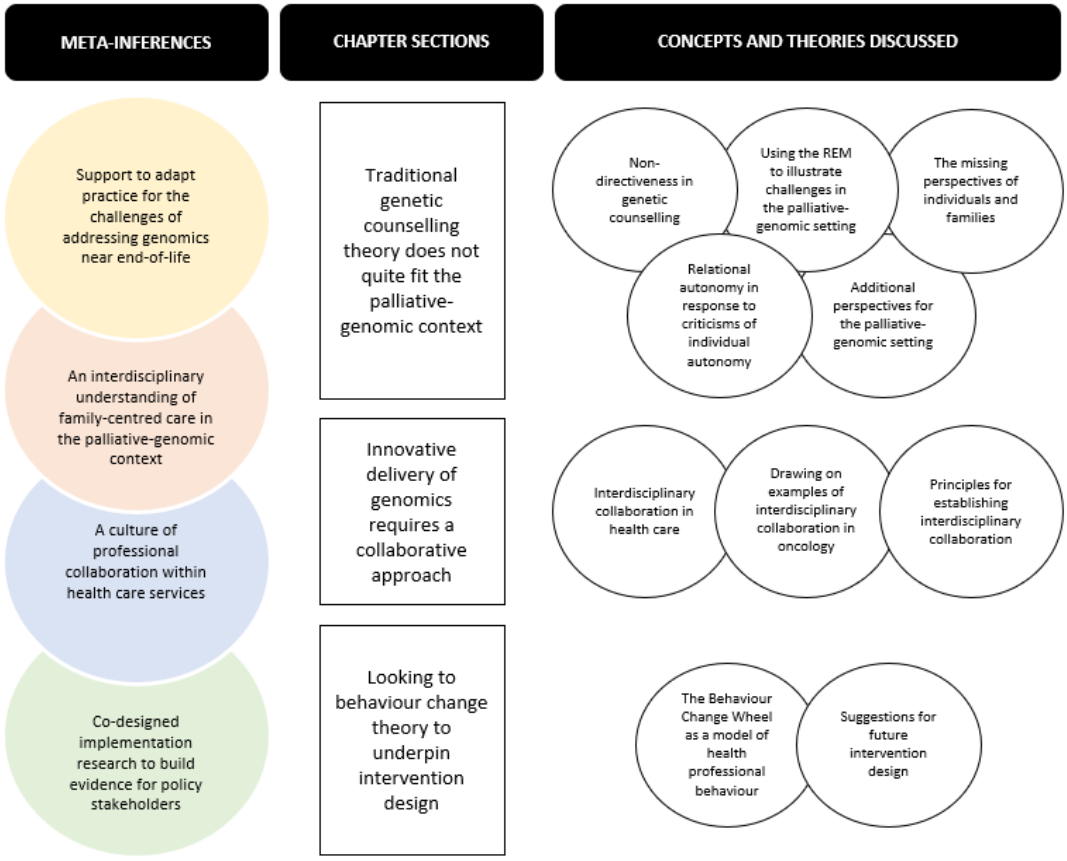


Figure 18. The concepts and theories discussed to provide context to the meta-inferences in this thesis. NB: This is not a conceptual map but is provided to assist with the navigation of section 7.4. Abbreviation: REM=Reciprocal-engagement model

### 7.4.1 Traditional Genetic Counselling Theory and Frameworks Do Not Quite Fit the Palliative-Genomic Context

The history and underlying theory guiding genetic counselling and palliative care practice provide several insights into the challenges faced in the palliative–genomic situation.

#### *7.4.1.1 Non-Directiveness in Genetic Counselling*

Several theories and models guide genetic counselling. Underlying many of them is the concept of 'non-directiveness'. Non-directiveness is an approach to counselling first introduced

by Carl Rogers in the 1950s, and this was adopted by the genetic counselling profession to support autonomous decision making.(298) Providing non-directive genetic counselling was a protective mechanism against eugenic practices, such as Nazi-led eradication of people with disabilities and paternalistic attitudes (e.g., physicians instructing parents to terminate pregnancies affected with aneuploidy).(299) Non-directiveness is practised by exploring a person's views and experiences, providing value-neutral information about clinical options, and supporting people to make decisions that are consistent with their values.

#### *7.4.1.2 Relational Autonomy in Response to Criticisms of Individual Autonomy*

Though individual autonomy remains an important ethical and legal concept, relational approaches to autonomy have gained traction in both genetic counselling and palliative care literature.(81, 300) Where individual autonomy places the focus upon a single person's capacity, wishes, and needs, relational autonomy is a reconceives autonomy as a way to acknowledge and value the inherent personal, familial, social, and cultural connections each person has. Proponents of relational autonomy argues prioritising an individual's autonomy ignores the fact that people are not always rational and independent, that decisions are made by considering others or impacted by others, that bioethical frameworks are too closely linked to Western ideals, that privileging autonomy discriminates against people with disabilities, and that individual autonomy is conceived of as an all-or-nothing concept.(81) Adopting a relational approach, often in the form of shared-decision making, positions people within their social systems and encourages, rather than limits, external influences on decision making.(301) Practising in a relational way also encourages health professionals to engage with the people their patient is emotionally connected to.(302) However, there are risks and challenges associated with relational autonomy. These include infringing on the strongly held wishes of an individual, collusion against individuals, 'silence conspiracy' (in which family members withhold information from the unwell person), a lack of confidentiality, limited time to engage with multiple family members, and increasing family members' stress by involving them in decision making.(81) To balance the benefits and risks, health professionals need skills in relational communication and social dynamics.

In the palliative–genomic situation, where a person's cognition may be reduced or fluctuates, prioritising individual autonomy can inappropriately force unwell patients to make decisions alone or exclude them from decision making if deemed incompetent.(81) One example is the use of informed consent documents that prioritise individual autonomy. Some have criticised these documents as perpetuating unhelpful practices and suggest changes to

reflect a more relational approach, although specific suggestions are lacking in the palliative–genomic context.(303) Changing documents and processes to allow health professionals to practise in a relational way provides more flexibility to assess and meet the individual's and family's needs.(304) Doing so may help to reduce the burden on individuals (or surrogate decision makers) to make difficult decisions about genomic testing in isolation.

#### *7.4.1.3 Using the Reciprocal-Engagement Model to Illustrate Genetic Counselling Challenges in the Palliative-Genomic Situation*

The Reciprocal-engagement model (REM) is one of the few genetic counselling-specific models of practice.(80) Developed by genetic counselling experts in the USA in 2007, the REM comprises five central tenets that incorporate individual and relational elements: [1] the genetic counsellor–patient relationship is integral to genetic counselling; [2] genetic information is key; [3] patient autonomy must be supported; [4] patients are resilient; and [5] patient emotions make a difference. These tenets encompass a variety of concepts, such as interpersonal communication, information delivery, and ethical practice, including respect for persons. The REM places the relationship between the genetic counsellor and the individual in the centre of the model, which means that the relational focus is predominantly between these two parties. The relational aspects between the individual and their family are implied (e.g., "patient emotions make a difference"), whereas the model is more explicit about individual autonomy (e.g., "patient autonomy must be supported".)(80)

The challenges described by genetic health professionals in the palliative–genomic situation can be examined in relation to the role of autonomy in the REM (Table 14). Near end of life, there is a lack of time and opportunity to build relationships with patients, deliver genomic information, explore options and resilience, and acknowledge difficult emotions. When practice is underpinned by models such as the REM with an explicit ethical instruction, the end-of-life situation increases pressure to deliver care that upholds these tenets. Infringements on ethical principles are a risk factor for moral distress and health professional burnout.(305, 306) An increased awareness of the ethical tensions in the palliative–genomic setting offers opportunities to provide preventive support to genetic and palliative care health professionals. Formal bioethical support can be preventive (ethics training or projects), proactive (ethics screening, informal consultations), reactive (ethics consultation for a difficult cases), or retrospective (debriefing or supervision).(307) Further work within clinical services to determine the appropriate form of bioethical support and a model of genetic counselling that aligns with ethically defensible practice in the end-of-life situation are required.

Table 14. Comparison of genetic counselling between the traditional context and the palliative–genomic context, using the Reciprocal-engagement model (REM).

| **REM tenet** | **Traditional genetic counselling context*** | **Palliative–genomic context** |
|---|---|---|
| The genetic counsellor-patient relationship is integral to genetic counselling | • Individuals are able to actively participate in relationship building<br>• There is time available to build relationships | • Individuals may be unable or unwilling to participate in relationship building<br>• There may be limited time available to build relationships with individuals or families |
| Genetic information is key | • Individuals are usually able to receive and interpret genomic information<br>• There is time available to explain complex genomic information | • Individuals may be unable or unwilling to receive complex information<br>• There may be limited time to convey complex genomic information |
| Patient autonomy must be supported | • Genetic counselling promotes an individual's autonomy by describing and exploring responses to all clinical options<br>• Individuals (competent adults) provide informed consent for their testing | • Limited time or possibility to explore options that promote autonomy<br>• Patient may be unable to provide informed consent because of reduced cognition<br>• Consent for genomic testing may need to be provided by a surrogate decision maker** |
| Patients are resilient | • Individuals are viewed as autonomous humans<br>• Sources of support are explored | • People are viewed as vulnerable individuals<br>• Limited time/opportunity to explore supports |
| Patient emotions make a difference | • Emotions are explored and acknowledged<br>• Counselling strategies are used to support emotions | • Limited time/opportunity to explore emotions<br>• Emotions may be heightened, making rational exploration more difficult for genetic health professionals |

NB. * Traditional genetic counselling context refers to providing genetic counselling to a person who has cognitive capacity and is not expected to imminently die
** This is only a difference in the adult setting. In paediatric contexts, informed consent is always provided by a surrogate decision maker (i.e., legal guardian).

#### *7.4.1.4 Additional Perspectives for the Palliative–Genomic Setting*

As genomic service delivery evolves, so too must the theoretical models that underpin genetic counselling practice. Non-directive approaches may not be appropriate for every genetic counselling situation, including the palliative-genomic context.(308) In an age of increasing genomic complexity, patients and families may need assistance interpreting information and making decisions, especially if heightened emotions impede decisions that

may lead to benefits for the family.(308) An ethical framework published in 2020 offers an approach to assist genetic health professionals evaluate whether non-directiveness helps or hinders by balancing individual autonomy with other important concepts, including the relational and familial implications of genetic counselling and genomic testing.(229) Frameworks such as these offer a theoretical basis for genetic health professionals to consider how practice meets their ethical obligations in the palliative–genomic setting. For patients near the end of life, providing simple information accompanied by a suggestion to store DNA may be the most ethical approach. The decision for patients (or family members) is then unilateral. The first question is whether the dying person (or surrogate decision maker) has any objection to collection of a blood sample for DNA storage. If there is no objection, the next question is whether the dying person (or surrogate decision maker) consents to the future use of this sample for genomic testing for the benefit of relatives.(101)

In the end-of-life situation, collaboration with palliative care health professionals can offer genetic health professionals insight into an individual's and family's values about genomic testing.(309) Discomfort related to the inability to explore the palliative person's (or family's) values before offering DNA storage (or genomic testing, if appropriate) could be addressed by drawing on the knowledge of palliative care health professionals. Nurses, in particular, have a unique perspective from the bedside where they may have more frequent conversational interactions with individuals and families.(310) A deeper understanding of an individual's or family's receptiveness to genomic information could be available to genetic health professionals through the development of professional relationships and collaboration with palliative care health professionals. Gleaning insights from trusted palliative care colleagues may reduce the discomfort experienced by genetic health professionals because of the lack of opportunity to explore the individual's and family's values.

#### *7.4.1.5 The Missing Perspective: Patients with Palliative Care Needs and Their Families*

The perspectives of people with palliative care needs and families is a critical evidence gap. Although health professionals' perspectives offer important insights, these make up just one piece of palliative–genomic implementation picture.(102) Health professionals are aware that conversations about genomics with people receiving palliative care (and their families), particularly near the end of life, may stir up difficult emotions.(96) Introducing the idea that a family member may be at risk of the same disease can be a confronting proposition. When handled poorly, conversations can harm individuals and families, which is a direct contradiction to the palliative care philosophy.(311)

Literature from the neonatal genomic testing context, in which parents are offered rapid genomic testing for their critically unwell newborn to guide treatment or withdrawal of treatment, may offer some insights into the needs of families in a highly emotive, time-pressured, and potentially palliative situation. Parents explain that it is difficult to take in information with heightened emotions and described the importance of having a plain language summary to take away and read afterwards.(312) The importance of adopting a family-centred (or in this case, 'parent-centred') approach was evident. Some parents consented to testing easily, whereas others required more time to work through their concerns, including feelings of guilt for having potentially passed on a genetic condition to their child.(313) More generally, people with palliative care needs and their families value clear and effective communication.(314) Involving family members in important discussions appears to improve a person's satisfaction with the quality of their palliative care.(315, 316) Avoiding or delaying difficult discussions does little to support the needs of individuals and families.(317)

The few studies that have elucidated individuals' or families' views about genomics in the palliative care context have produced discordant results. On one hand, some people had existing suspicions that their disease had a hereditary basis,(183) but others had never heard of genomic testing or DNA storage.(93) Individuals reported being comfortable engaging in genomics discussions(92) and valued the opportunity to address existing concerns about genetic risk and to leave a legacy for future generations.(183, 189, 318, 319) However, relatives felt it was generally inappropriate to raise genomics in the palliative care setting.(96) Overall, it seems that people with palliative care needs value the opportunity to discuss genomics, but a more robust evidence base is needed before sound conclusions can be drawn. Establishing the views and needs of palliative people and their families is critical to ensuring that any future intervention is ethically and clinically appropriate.

### 7.4.2 Innovative Delivery of Genomics Requires a Collaborative Approach

Integrating genomics into palliative care is one piece of the broader genomic implementation picture. As mentioned in Chapter 1, innovative care delivery models are being trialled to improve access to genomics for people across a range of clinical areas. Underpinning many of these models are the collaborative relationships genetic health professionals form with other healthcare teams. Collaboration between genetic and palliative care health professionals is desired, although the nature of this collaboration needs to be defined. This

section provides examples of teams that genetic and palliative care health professionals operate within, the benefits and drawbacks of collaborative working, and potential avenues for collaborative work based on others' experiences of integrating genomics into routine medical care.

#### *7.4.2.1 Interdisciplinary Collaboration in Health Care*

Collaborative work with interdisciplinary teams is an essential component of healthcare.(320) Interdisciplinary teams deliver care in a variety of contexts, including different patient populations (e.g., patients with cancer) and stages of care (e.g., transition between services). Interdisciplinary collaboration is protective by improving the quality and safety of care, patients' experiences, and clinical outcomes.(321) Fragmented teams reduce the quality and safety of patient care through miscommunication or misunderstandings, delays in access to care, inadequate health professional expertise, and access barriers (e.g., people having to attend multiple appointments).(321). Genetic health professionals play a part in many interdisciplinary teams, including acute neonatal care teams to facilitate rapid genomic testing to guide treatment or withdrawal of care in sick neonates,(322) multidisciplinary tumour board teams to advise oncology teams if a patient meets criteria for genomic testing,(160) and neurogenetic teams to facilitate testing and provide counselling to individuals and families affected by neurogenetic conditions.(66) Palliative care health professionals often work within oncology interdisciplinary teams to identify and deliver care to people with cancer who would benefit from palliative care input (although they may also work with other non-cancer teams).(323)

#### *7.4.2.2 Drawing on Examples of Collaborative Interventions in the Oncology Setting*

Despite calls for interdisciplinary collaboration in the palliative–genomic setting, there is a lack of published interventions.(75, 94) However, insights could be gleaned from the multi-component oncology interventions that have demonstrated a significant improvement in rates of genomic testing or genetic counselling.(240) For example, the inclusion of a patient navigator in a multi-component intervention, whose role is to work between the genetic and oncology teams to identify patients eligible for testing, increased referrals of women with breast cancer for genetic counselling from 69% to 91% and genomic testing from 59% to 86% across the study period.(324) In another study, genetic counsellors and colorectal surgeons co-reviewed immunohistochemistry results from people with colorectal cancer to determine if a referral was warranted.(325) Significant increases in referrals and attendance to genetic

counselling and higher rates of genomic testing and identification of pathogenic variants in affected individuals were reported compared with their control model.(325) Embedding genetic counsellors into gynaecology oncology clinics has also shown a significant increase in referrals of women with gynaecological malignancies to the genetic service.(58) In another study, embedding a genetic counsellor, in addition to a specialised referral template and regular multidisciplinary case reviews, increased patient referrals to the genetic service from 54% to 85%.(59)

#### *7.4.2.3 Principles for Establishing Interdisciplinary Collaboration*

Effective teams are underpinned by a shared mental model, which is defined as "a common understanding of the situation, the plan for treatment, and the roles and tasks of the individuals in the team".(326) The need for a shared mental model is supported by the findings in this thesis, in that developing a shared understanding of family-centred care is likely to support the integration of genomics into palliative care. Once a shared mental model is established, effective teams require leadership, team monitoring, 'backup behaviour' (support to help other members achieve their tasks), adaptability, and 'team orientation' (the belief that the team's goal is more important than the individual's goals).(320) Collaborative approaches to care are valued in clinical genomics because they lead to improvements in patient care.(327)

When establishing a team, it is important to identify educational, psychological, and organisational barriers.(326) Educational barriers can relate to the clinical knowledge required to form a shared goal as well as the knowledge required to function effectively as a team. Psychological barriers refer to the negative perceptions of other health professionals or an overly positive view of their own profession. Examples of organisational barriers include inconsistent paperwork between departments or geographical distance between team members. Providing training about teamwork (such as inclusive and democratic practices) and supporting teams with relevant protocols and procedures can help to overcome barriers.(328) For the palliative–genomic setting, further work to elucidate the barriers to and facilitators of interdisciplinary collaboration could begin to understand how genetic and palliative care health professionals can work together to address the needs of people receiving palliative care and their families.

### 7.4.3 Looking to Behaviour Change Theory to Inform Intervention Development

In healthcare, a desired outcome relies on a health professional behaving in a particular way. One might assume that if a genetic or palliative care health professional identifies a person requiring a genomics discussion, they would then provide the option for genetic counselling and DNA storage or genomic testing (if appropriate). However, such assumptions do not acknowledge the complexity of health professional behaviour. Exploring this complexity is essential, as behaviour directly impacts the clinical benefit available to relatives through an accurate assessment of their disease risk. Although this thesis did not aim to design an intervention, the findings can be examined in relation to behaviour change theory to inform future interventions.

#### *7.4.3.1 The Behaviour Change Wheel as a Model of Health Professional Behaviour*

Understanding the factors influencing a health professional professional's behaviour can underpin the design of an intervention that targets that behaviour. The Behaviour change wheel (BCW) and Capability, opportunity, motivation behaviour model (COM-B) maps behavioural domains (e.g., knowledge, skill, or beliefs about consequences) to overarching components (e.g., knowledge and skill map to 'capability').(106) Each component is linked to an intervention designed to target the behavioural domain (e.g., 'capability' barriers can be address with educational interventions). With an absence of theory specific to the palliative–genomic setting, the BCW offers a theory-informed approach to suggest interventions that target the barriers and facilitators identified in the GIFT Project.

#### *7.4.3.2 Suggestions for Future Intervention Design*

A predominant barrier to integrating genomics into palliative care is the low genomic knowledge of palliative care health professionals, which in turn may reduce palliative care health professionals' motivation to broach a genomics discussion.(218, 250, 296) The BCW explains that barriers related to capability, including poor knowledge, influence a health professionals' motivation to engage in a behaviour. Interventions to address capability are education (provision of knowledge), training (practising of skills), and enablement (increasing means and reducing barriers to increase capability or opportunity).(106) A multicomponent intervention incorporating these interventional functions may help to improve practice by supporting palliative care health professionals' capability and motivation.(102) For example, palliative care health professionals may not be aware of the possibility of DNA storage or how to facilitate this, but providing education could improve their capability, and in turn, increase

their motivation to integrate genomics into the care of people with palliative care needs and their families.(133)

The BCW also links behavioural components to different policy interventions. The BCW backs the development of guidelines to enhance capability, as suggested in Chapter 4.(106, 218, 250) Capability issues can also be addressed by developing regulatory and legislative guidance.(106) However, health professionals may be less receptive to regulation and legislation in the palliative–genomic context, in which the flexibility to remain person and family centred is highly valued.

## 7.5 RECOMMENDATIONS

This section summarises the recommendations of the GIFT Project for clinical practice and future research. These recommendations are not listed in order of importance. These are high-level recommendations that can be adopted and further refined in more specific settings.

### 7.5.1 Recommendations for Clinical Practice

- Health professionals should remain person and family centred when engaging in discussions about genomics with people who have palliative care needs.
- For people nearing the end of life, health professionals should consider offering DNA storage rather than genomic testing.
- Health professionals and health services should identify and deliver the support required to help genetic and palliative care health professionals navigate practice adaptions when addressing genomics near the end of life.
- Health services should develop local clinical practice guidelines to support health professionals to navigate genomics in the end-of-life situation.
- Health services should provide bioethical support to genetic and palliative care health professionals to navigate ethical tensions when addressing genomics with people nearing the end of life.
- Health professionals and health services should develop a shared understanding of family-centred care for the palliative–genomic situation.
- Health services should provide tangible support to build interdisciplinary connections and collaborative relationships between genetic and palliative care health professionals.
- Policy stakeholders should consider developing high-level recommendations for organisations or health services to provide support to health professionals.

### 7.5.2 Recommendations for Future Research

- Conduct research with people who have palliative care needs and their families about their views and experiences of genetic counselling, genomic testing, and DNA storage.
- Clinical audit/s or similar that estimate the number of people receiving palliative care whose disease has an underlying genetic aetiology, and who have had, or require, genetic testing.
- Design and trial a multicomponent intervention, including a DNA storage approach for addressing genomics at the end of life.
- Design, test, and monitor a clinical practice guideline for the end-of-life situation.
- Develop and analyse the acceptability of a shared family-centred care model, including how acceptable this model is to people with palliative care needs and their families.
- Explore the barriers to and facilitators of effective collaboration and interdisciplinary teamwork between genetic and palliative care health professionals.
- Collaborative research with interdisciplinary health professionals, researchers, academic institutions, and healthcare organisations to establish local support needs (including barriers and facilitators) through implementation research.
- Conduct an economic analysis of outcomes for individuals seeking advice about the personal clinical implications of a deceased relative's condition, including the impact of suboptimal risk assessments for people who are unable to access relevant genetic information.
- Conduct a policy stakeholder consultation to determine the implications of integrating recommendations about genomics in palliative care into relevant policy documents.

## 7.6 STRENGTHS AND LIMITATIONS

Here I detail the strengths and limitations of the GIFT Project. Strengths and limitations pertaining to the individual studies are detailed in corresponding manuscripts.

### 7.6.1 Strengths

The GIFT Project was strengthened by using theory, which was important given the limited evidence about genomics in palliative care. The overarching framework (the WHO ICCC) provided a conceptual lens at every stage, including planning, data collection, analysis, and integration. Using the Theoretical domains framework and COM-B models to synthesise the data in the systematic review provided an evidence-based framework and spurred the exploration of implementation science theories relevant to the palliative-genomic context.

Incorporating implementation science principles into the planning, design, and conduct of the GIFT Project set up the next stage of intervention design.

Conducting the GIFT Project with an exploratory sequential approach allowed a deep exploration of the Australasian context primarily from the perspectives of health professionals before testing these findings with quantitative methods. The combination of different methods, including the deliberate integration of qualitative and quantitative data, has begun to fill the conceptual and methodological gaps in the literature.

The GIFT Project benefited from a variety of viewpoints. The guidance provided at regular meetings with my interdisciplinary supervisory team (including expertise in genetic counselling, social science, and palliative care) provided varied perspectives that encouraged me to engage in critical reflections of my assumptions and biases. In addition, the studies in this thesis were presented as 'works in progress' for peer-review at the Genetic Counselling Research Seminar in the Graduate School of Health at UTS on several occasions. I presented work to the Improving Palliative, Aged and Chronic Care through Clinical Research and Translation consumer advisory group for feedback. I have presented various aspects of this thesis at conferences (full details listed on pages vii–viii). Except for the manuscript in Chapter 6 (which is currently under consideration at the *European Journal of Human Genetics)*, all manuscripts have been published following external, independent peer review.

### 7.6.2 Limitations

One of the limitations of the GIFT Project was the inability to incorporate views of additional stakeholders, namely the views of people with palliative care needs and their families. One way to overcome this would have been to set up an ongoing relationship with a consumer advisory group in addition to the presentations made to consumers (as above). Unfortunately, given the interruptions caused by COVID-19, this did not eventuate.

The decision to include genetic and palliative care health professionals from New Zealand alongside Australian health professionals (collectively referred to as Australasia) was made because of the organisational jurisdiction of the Human Genetics Society of Australasia and the Australian and New Zealand Society of Palliative Medicine. However, health professionals from New Zealand were underrepresented, and the findings may not be generalisable to their setting.

In exploratory sequential research, participants from the first phase generally do not participate in the next phase.(208 p192-3) However, with a small population, obtaining adequate numbers can be more difficult if participants in the qualitative study are excluded from other studies. In the GIFT Project, I was unable to cross-reference participants in the questionnaire survey without infringing on their anonymity. Therefore, it is possible that some participants in the qualitative research took part in the quantitative survey.

The scope of palliative care in this thesis was intentionally broad to capture the barriers to and facilitators of a range of conditions (including malignant and non-malignant disease), life stages (including patients receiving palliative care alongside curative treatment, to those at the end of life), and delivered by different disciplines (including medical and nursing disciplines but not allied health or other disciplines). However, this broad approach has several limitations. Firstly, given the tendency for palliative care to be associated with cancer, the findings here may be more representative of the barriers and facilitators to integrating genomics into the care of palliative people with cancer, rather than other life-limiting disease (such as neurodegenerative conditions). Secondly, some of the main findings relate more specifically to the end-of-life context, where the challenges to genomics integration are intensified. The findings may be less applicable to health professionals caring for patients who are not expected to die imminently. Thirdly, although some participants had experience in paediatric palliative care, the adult context is likely to be overrepresented here. Lastly, other health professionals (such as social workers) who are involved in the delivery of palliative care may be integrating genomics into their practice and could have shared their insights. Further work to assess the barriers and facilitators for people with different life-limiting diseases, at different life stages and ages, and provided by different health disciplines would be valuable.

## 7.7 CONTRIBUTION TO THE FIELD

### 7.7.1 Building Evidence to Overcome the Challenge of Genomic Implementation

The implementation of genomics into routine healthcare is a complex challenge that will require creative approaches for each unique context. This thesis, and the published papers within it, have contributed to the knowledge base of genomics implementation by building on relevant theory (e.g., related to health professional behaviour) and methodological approaches to implementation research (e.g., demonstrating the use of an exploratory sequential design). Some of the challenges described in this thesis are relevant to genomics implementation more broadly. The importance of synthesising the barriers and facilitators

affecting health professional behaviour in genomics implementation is evidenced by the 40 citations (as of January 2023) of the systematic review manuscript in this thesis. Those who have cited my work used the findings to support their work in a variety of clinical areas (e.g., nephrology, oncology, psychiatry, and pharmacogenomics)(329-332) and research endeavours (e.g., workforce support for non-genetic health professionals).(333, 334)

### 7.7.2 Raising Awareness of the Palliative-Genomic Context

Despite efforts to implement genomics into routine healthcare, the focus on people with palliative care needs and their families remains suboptimal. In the age of personalised medicine, where genomic testing is often performed to guide targeted therapies, it can be easy to lose sight of settings where the clinical benefits of genomics are indirectly related to family members, rather than the person having the tests. However, the published work from this thesis appears to be increasing awareness of the challenges of the palliative-genomic situation. An Australian group has cited my research in a paper that discusses the hope and certainty genomics provides for children with palliative needs and their families.(335) In addition, I was invited to present to a plenary session about integrated care at the Palliative Care Nurses Australia annual congress in 2022. Several of my abstracts have been selected and presented as talks and posters at several palliative care, genetic, and oncology conferences during my candidature (see pages vii-viii for further details).

### 7.7.3 Bolstering the Genetic Counselling Research Discipline

Genetic counselling research continues to strengthen, and many genetic counsellors now perform research as part of their role.(52) The discipline displays diverse research capabilities and interests that use qualitative, quantitative, and mixed methods approaches. This thesis has provided an example of an exploratory sequential mixed methods design in the genetic counselling discipline, explicit philosophical positioning, and the use of theory. In 2022, I was invited to present the methodological aspects of the thesis to the Genetic Counsellors in Research Connect group, a national group of genetic counsellors who are working or interested in research. When participating in genetic conferences, I highlight the methodological aspects as integral to the clinical implications to demonstrate the importance of rigor in developing evidence-based recommendations.

## 7.8 CHAPTER SUMMARY

This chapter began with a high-level summary of how this thesis answered research questions 1 and 2 in previous chapters. Research question 3 was answered in this chapter through the presentation of four meta-inferences. I then discussed the meta-inferences in relation to the existing literature and followed this with several recommendations for clinical practice and future research. I explored the strengths and limitations, and finished by discussing the contribution of this thesis to the field.

## 7.9 THESIS CONCLUSION

This thesis has begun to fill the conceptual and methodological gaps in the palliative–genomic literature. As genomics continues to be adopted into routine healthcare, integrating genomics into the care of people with palliative needs and their families requires dedicated attention. Genetic and palliative care health professionals need support to safely adapt practice and navigate the bioethical tensions in the end-of-life situation. Development of a shared understanding of family-centred care is required, and healthcare services must support interdisciplinary teams to work collaboratively for optimal care delivery. Co-designed implementation research at the local level will help to build a suite of evidence to lobby policy stakeholders to develop relevant recommendations. Incorporating these elements into a multi-component intervention should support best practice for genetic and palliative care health professionals to integrate genomics into the care of people with palliative care needs and their families.

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# 8 Appendices

# Appendix A: supplementary files related to systematic review

## CONTENTS

SUPPLEMENTARY FILE A1: PRISMA CHECKLIST

| Section/topic | # | Checklist item | Reported on page # |
|---|---|---|---|
| TITLE | | | |
| Title | 1 | Identify the report as a systematic review, meta-analysis, or both. | 18 |
| ABSTRACT | | | |
| Structured summary | 2 | Provide a structured summary including, as applicable: background; objectives; data sources; study eligibility criteria, participants, and interventions; study appraisal and synthesis methods; results; limitations; conclusions and implications of key findings; systematic review registration number. | 18 |
| INTRODUCTION | | | |
| Rationale | 3 | Describe the rationale for the review in the context of what is already known. | 19 |
| Objectives | 4 | Provide an explicit statement of questions being addressed with reference to participants, interventions, comparisons, outcomes, and study design (PICOS). | 19-20, Appendix A3 |
| METHODS | | | |
| Protocol and registration | 5 | Indicate if a review protocol exists, if and where it can be accessed (e.g., Web address), and, if available, provide registration information including registration number. | 20 |
| Eligibility criteria | 6 | Specify study characteristics (e.g., PICOS, length of follow-up) and report characteristics (e.g., years considered, language, publication status) used as criteria for eligibility, giving rationale. | 20, Appendix A3 |
| Information sources | 7 | Describe all information sources (e.g., databases with dates of coverage, contact with study authors to identify additional studies) in the search and date last searched. | 20 |
| Search | 8 | Present full electronic search strategy for at least one database, including any limits used, such that it could be repeated. | Appendix A2 |
| Study selection | 9 | State the process for selecting studies (i.e., screening, eligibility, included in systematic review, and, if applicable, included in the meta-analysis). | 21 |
| Data collection process | 10 | Describe method of data extraction from reports (e.g., piloted forms, independently, in duplicate) and any processes for obtaining and confirming data from investigators. | 21 |
| Data items | 11 | List and define all variables for which data were sought (e.g., PICOS, funding sources) and any assumptions and simplifications made. | Appendix A4 |

| | | | |
|---|---|---|---|
| Risk of bias in individual studies | 12 | Describe methods used for assessing risk of bias of individual studies (including specification of whether this was done at the study or outcome level), and how this information is to be used in any data synthesis. | 22 & Appendix A5 |
| Summary measures | 13 | State the principal summary measures (e.g., risk ratio, difference in means). | NA |
| Synthesis of results | 14 | Describe the methods of handling data and combining results of studies, if done, including measures of consistency (e.g., $I^2$) for each meta-analysis. | 22-23 |

SUPPLEMENTARY FILE A2: MEDLINE SEARCH STRATEGY

| | Term/s # | Terms | Yield |
|---|---|---|---|
| Concept 1 | 1 | Nurse's Role/ or nurs*.mp. | 695758 |
| | 2 | Nurses/ | 37265 |
| | 3 | General Practitioners/ or general practitioner*.mp. | 48773 |
| | 4 | Neurology/ or neurolog*.mp. | 388934 |
| | 5 | Oncology Nursing/ or Oncologists/ or oncolog*.mp. | 146450 |
| | 6 | Physicians/ or Physician's Role/ or physicia*.mp. | 528705 |
| | 7 | palliative care.mp. or Palliative Care/ | 60046 |
| | 8 | neuropathology.mp. or Neuropathology/ | 12032 |
| | 9 | Medical Oncology/ | 16658 |
| | 10 | Surgeons/ or surgeon*.mp. | 180524 |
| | 11 | Health Personnel/ or health personnel.mp. | 163597 |
| | 12 | Primary Health Care/ or primary health care.mp. | 84382 |
| | 13 | Terminal Care/ or terminal care.mp. or hospice.mp. or end of life care.mp. | 40274 |
| Concept 2 | 14 | Genetics, Medical/ or genetic*.mp. or Genetics/ | 3709688 |
| | 15 | Genomics/ or genomic*.mp. | 290410 |
| | 16 | Genetic Services/ | 482 |
| Concept 3 | 17 | Pedigree/ or pedigree.mp. | 87096 |
| | 18 | Risk Assessment/ or risk assessment.mp. | 270318 |
| | 19 | Referral and Consultation/ | 62872 |
| | 20 | Patient Education as Topic/ | 81885 |
| | 21 | Medical History Taking/ or anamnesis.mp. | 23368 |
| | 22 | family history.mp. | 54442 |
| | 23 | Genetic Counseling/ or genetic counsel?ing.mp. | 23496 |
| | 24 | Genetic Testing/ or gen* test*.mp. or genetic screening.mp. or DNA banking.mp. | 58833 |
| | 25 | Precision Medicine/ | 14380 |
| | 26 | Patient Care/ or patient care.mp. | 175719 |
| | 27 | Education, Professional/ | 2674 |
| | 28 | Continuing, Medical, Education/ or Continuing, Nursing, Education/ or refresher courses.mp. | 46853 |
| Concept 4 | 29 | Knowledge/ or Health Knowledge, Attitudes and Practice/ | 112420 |
| | 30 | Attitude/ or Behavior/ | 72202 |
| | 31 | “Attitude of Health Personnel”/ | 115272 |
| | 32 | Practice Patterns, Physicians’/ or medical practice.mp. | 71606 |
| | 33 | Practice Patterns, Nurses/ or nursing practice.mp. | 18109 |
| | 34 | Perception/ or perception*.mp. | 397850 |
| | 35 | Clinical Competence/ or clinical competenc*.mp. | 88255 |
| | 36 | Professional Competence/ or professional competenc*.mp. | 24802 |

| | | | |
|---|---|---|---|
| | 37 | Professional Practice/ | 16427 |
| | 38 | genomic literacy.mp. | 35 |
| | 39 | experienc*.mp. | 998178 |
| | 40 | 1, 2, 3, 4, 5, 6, 7, 8, 9, 10, 11, 12 or 13 | 2039621 |
| | 41 | 14, 15 or 16 | 3760771 |
| | 42 | 17, 18, 19, 20, 21, 22, 23, 24, 25, 26, 27 or 28 | 837379 |
| | 43 | 29, 30, 31, 32, 33, 34, 35, 36, 37, 38 or 39 | 1703072 |
| | 44 | 40, 41, 42 and 43 | 3007 |

## SUPPLEMENTARY FILE A3: PICOS ELIGIBILITY CRITERIA FOR SYSTEMATIC REVIEW

| | INCLUSION | EXCLUSION |
|---|---|---|
| **Population** | • Qualified specialist nurses and/or physicians caring for adult patients<br>AND<br>• Providing secondary or tertiary health care<br>AND<br>• 50% or more of the population must represent nurses and physicians<br><br>• The following clinical areas will be included:<br>  ○ Cardiology (adult)<br>  ○ Dermatology<br>  ○ Endocrinology<br>  ○ Gastroenterology<br>  ○ Clinical hematology<br>  ○ Medical oncology<br>  ○ Nephrology<br>  ○ Neurology<br>  ○ Ophthalmologists<br>  ○ Palliative medicine<br>  ○ Psychiatric medicine<br>  ○ Radiation Oncology<br>  ○ Respiratory Medicine<br>  ○ Rheumatology<br>  ○ Surgical oncology | • Midwives<br>• Allied Health Professionals (including genetic counselors, physiotherapists, speech pathologists, occupational therapists, orthoptists, dietitians, dental hygienist, social workers, pharmacists, medical radiation practitioners, chiropractors, podiatrists, optometrists, osteopaths and Chinese medicine practitioners)<br>• Patient, consumer or client groups<br>• Health professionals working in a non-clinical setting (eg. academic institutions, government, laboratory, research)<br>• The clinical specialty of the nurses and/or physicians are not indicated or are unclear<br>• Specialist nurses or physicians working within the following clinical specialty areas:<br>  ○ Addiction medicine<br>  ○ Adolescent and Young Adult Medicine<br>  ○ Clinical genetics (including Familial Cancer)<br>  ○ Clinical pharmacology<br>  ○ Community child health<br>  ○ Emergency medicine<br>  ○ General and acute care medicine<br>  ○ Obstetrics/Gynecology<br>  ○ Pediatric, neonatal or perinatal medicine<br>  ○ Geriatric medicine<br>  ○ Clinical immunology and allergy<br>  ○ Infectious diseases<br>  ○ Nuclear medicine<br>  ○ Occupational and environmental medicine<br>  ○ Public health medicine<br>  ○ General rehabilitation medicine<br>  ○ Sexual health medicine<br>• Nurses or physicians working in a primary care setting. This may include<br>  ○ General practitioners<br>  ○ Family physicians<br>  ○ General internists (unless specifically stated they work within a secondary, tertiary or hospital setting)<br>  ○ Obstetrician/gynecologist<br>  ○ Practice nurses<br>• Sample of nurses and physicians obtained from a laboratory database |

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| **Intervention** | • Provision of genetic counseling which may include one or more of the following<br>  ○ Taking a family health history<br>  ○ Assessing genetic risk<br>  ○ Identifying an individual with or at risk of a genetic condition<br>  ○ Initiating a discussion with a patient or family member about genetics or genomics<br>  ○ Providing genetic health information to a patient or family member<br>  ○ Organizing genetic or genomic testing<br>  ○ Interpreting genetic or genomic test results<br>  ○ Discussing and/or organizing DNA banking<br>  ○ Delivering a genetic or genomic test result to a patient or their family member<br>  ○ Facilitating adjustment to a genetic condition or a genetic or genomic test result<br>  ○ Referring an individual or family to a genetics service | • Measuring effect of an intervention to assist nurses and/or physicians to incorporate genetics and genomics into their practice<br>• Phenomena of interest is unrelated to genetic counseling<br>• Reproductive genetic counseling. This may include<br>  ○ Abnormal ultrasound findings in pregnancy<br>  ○ Prenatal screening tests (such as first trimester screening or Non-invasive prenatal screening)<br>  ○ Prenatal diagnostic tests or preimplantation genetic diagnosis<br>  ○ Reproductive carrier screening (eg. discussion or testing of recessive conditions for the purpose of determining the health of a couple's offspring)<br>  ○ Genetic counseling for pharmacogenetic testing<br>• Genetic testing related to public health such as population screening or newborn screening<br>• Molecular tumor testing, except if the study explicitly discusses heritable pathogenic variants via molecular tumor testing<br>• Genetic counseling related to direct-to-consumer genetic testing<br>• Genetic assessment relating to the clinicians own genetic risk<br>• Genetic counseling in the research setting<br>• Management of an individual or family based on a genetic risk assessment or genetic test (eg. breast MRI, risk-reducing surgeries, chemoprevention, environmental modifiers, pacemakers, beta-blockers) |

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| **Outcomes** | • The primary focus (defined as 50% or more of the reported outcomes) is related to the integration of genetics and genomics into clinical practice<br>• Reporting of barriers and facilitators towards incorporating genetics and genomics into usual clinical practice. This may include<br>• Attitudes towards incorporating genetics and genomics into usual practice<br>• Behaviors in relation to incorporating genetics and genomics into usual practice<br>• Knowledge which impacts on the ability to incorporate genetics and genomics into usual practice<br>• Heterogeneous populations (>10% of participants of sample do not meet inclusion criteria) must have outcome data reported separately for each clinical specialty | • The primary focus (defined as 50% or more of the reported outcomes) is not related to the barriers and facilitators towards integration of genetics and genomics into clinical practice<br>• Description of clinical services<br>• Reporting of patient presentation, characteristics or phenotype<br>• Attitudes, knowledge or behavior related to variant reclassification, updated guidelines or availability of new genetic testing for patients who have already received genetic testing<br>• Attitudes, knowledge or behavior related to hypothetical scenarios<br>• Outcomes for heterogeneous populations (>10% of participants of sample do not meet inclusion criteria) are not reported separately for each clinical specialty |
| **Study design** | • Quantitative design<br>• Written in English language<br>• Published after February 2001<br>• Qualitative design<br>• Mixed-methods design | • Written in a language other than English<br>• Published before February 2001<br>• Non-primary research including<br>  ○ Reviews (eg. systematic, scoping, narrative)<br>  ○ Editorials<br>  ○ Expert opinion<br>  ○ Conference abstract<br>  ○ Protocols |

## SUPPLEMENTARY FILE A4: DATA EXTRACTION ITEMS WITH EXAMPLES.

| NB: Extraction tool based on the Joanna Briggs Institute evidence synthesis manual (as referenced in manuscript) to demonstrate the pre-determined data items | |
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| **Data item** | **Extraction examples** |
| Author, year, journal | Smith, Robert; 2019; Journal of Genetic Counselling |
| Methodology: | Qualitative, quantitative or mixed-methods |
| Method: | Grounded theory, ethnography, descriptive, experimental |
| Phenomena of interest: | Family history taking, risk assessment, genetic testing |
| Setting | Primary, secondary or tertiary care, educational institution |
| Geographical: | Australia, Australasia, United Kingdom, Europe |
| Cultural: | Targeted population (eg. Jewish, Christian, vegan) |
| Participants' discipline: | Nurses or doctors |
| Participants' specialty: | Cardiology, oncology, neurology |
| Data analysis: | Thematic analysis, descriptive statistics |
| Outcomes: | "23% of cardiologists felt their genetics knowledge was lacking" |
| Authors conclusions: | Themes, subthemes, limitations |
| Comments: | Funding sources |

## SUPPLEMENTARY FILE A5: QUALSYST CHECKLIST FOR RISK OF BIAS/CRITICAL APPRAISAL

| *NB. Tool based on quality assessment from (Kmet 2004)* | | | |
|---|---|---|---|
| Date of assessment: | | Name: | |
| First author: | | Publication year: | |
| Title: | | | |

| **QUALITATIVE** | | | |
|---|---|---|---|
| **Criteria** | **YES (2)** | **PARTIAL (1)** | **NO (0)** |
| Question / objective sufficiently described? | | | |
| Study design evident and appropriate? | | | |
| Context for the study clear? | | | |
| Connection to a theoretical framework / wider body of knowledge? | | | |
| Sampling strategy described, relevant and justified? | | | |
| Data collection methods clearly described and systematic? | | | |
| Data analysis clearly described and systematic? | | | |
| Use of verification procedure(s) to establish credibility? | | | |
| Conclusions supported by the results? | | | |
| Reflexivity of the account? | | | |
| TOTAL | | | |

| **QUANTITATIVE** | | | |
|---|---|---|---|
| **Criteria** | **YES (2)** | **PARTIAL (1)** | **NO (0)** |
| Question / objective sufficiently described? | | | |
| Study design evident and appropriate? | | | |
| Method of subject/comparison group selection or source of information/input variables described and appropriate? | | | |
| Subject (and comparison group, if applicable) characteristics sufficiently described? | | | |
| If interventional and random allocation was possible, was it described? | | | |
| If interventional and blinding of investigators was possible, was it reported? | | | |
| If interventional and blinding of subjects was possible, was it reported? | | | |
| Outcome and (if applicable) exposure measure(s) well defined and robust to measurement / misclassification bias? Means of assessment reported? | | | |
| Sample size appropriate? | | | |
| Analytic methods described/justified and appropriate? | | | |
| Some estimate of variance is reported for the main results? | | | |
| Controlled for confounding? | | | |
| Results reported in sufficient detail? | | | |
| Conclusions supported by the results? | | | |
| TOTAL | | | |

SUPPLEMENTARY FILE A6: SUMMARY OF INCLUDED ARTICLES

| Author Year Country | Aim | Design<br><br>Measure | Participants<br><br>Response rate (RR) | Main findings | Barrier/Facilitator: TDF domains *(themes)* | Risk of bias |
|---|---|---|---|---|---|---|
| Paller<br><br>2019<br><br>USA | To describe practice patterns and attitudes of urological oncologists towards germline genetic testing for men with advanced prostate cancer | Single survey<br><br>Novel instrument (unvalidated) | *N*=26 oncologists<br><br>RR 44% | Germline testing has therapeutic relevance for patients with metastatic prostate cancer. 38% of urologic oncologists referred to genetic counsellors, while the remainder took partial or full responsibility for genetic information provision. 62% considered germline testing for all patients with metastatic prostate cancer. | Barriers: Environmental context & resources *(lack of time, resources, links to genetics services, cost of testing)*; Goals *(genetic testing not useful)*<br><br>Facilitators: Environmental context & resources *(having written resources);* Belief about consequences *(medical benefit to patient)* | 0.72 |
| Klepek<br><br>2019<br><br>USA | To characterize neurologists' practices and attitudes towards genetic testing in Amyotrophic Lateral Sclerosis (ALS) | Single survey<br><br>Novel instrument (unvalidated) | *N*=77 neurologists<br><br>RR 30% | Neurologists with positive attitudes towards genetic testing were more likely to offer genetic testing to 'sporadic' ALS patients (p = 0.0001). Links to a genetic counsellor improved attitude scores (p = 0.03) but did not increase testing to 'sporadic' ALS patients (p = 0.49), early onset ALS (p = 0.71) or those with ALS and dementia (p = 0.99). | Barriers: Environmental Context & Resources (l*ow referral rates; lack of guidelines*)<br><br>Facilitators: Skill (*genetics discussions, family history taking, ordering genetic testing*); Environmental context & resources (p*atient request*); Optimism (p*ositive attitude, future benefit*); Belief about Consequences (b*enefit for patient and relatives*) | 0.83 |
| Hallowell<br><br>2019<br><br>UK | To describe medical oncologists' and breast nurses' and physicians' views regarding implementation of a treatment-focused genetic | Semi-structured interviews | *N*=13 [6 (46%) breast surgeons; 1 (8%) breast care nurse; 6 (46%) medical oncologist]<br><br>RR 42% | Breast surgeons were reluctant to integrate genetics and genomics into their role. In contrast, medical oncologists were enthusiastic, citing the medical benefit to their patients and feeling capable of integrating genetics and genomics into their practice. | Barriers: Skill (*low confidence*); Memory, attention & decision processes (g*enetics detracts from other concerns*); Environmental context & resources (*lack of time*); Goals (*genetic testing not useful, not relevant*); Professional role (r*esistant to change, not appropriate to counsel asymptomatic relatives*) | 0.85 |
| Wright<br><br>2019 | | Semi-structured interviews and | | | | 0.85 |

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| UK | testing pathway (TFGT) | observational field notes | | | Facilitators: Optimism (*positive attitudes, integration of genetics is inevitable*); Belief about consequences (*benefit for patient*); Professional role (*appropriate for clinician to provide genetic health information*); Belief about capability (*competent to provide genetic health information*); Intention (*to engage in further education [online content preferred]*) | |
| Cleophat<br><br>2019<br><br>Canada | To ascertain frequency of and views about discussions of inherited cancer in palliative care | Single survey<br><br>Novel instrument (unvalidated) | *N*=64 [29 (45%) palliative care physicians; 35 (55%) palliative care nurses<br><br>RR 9% | Approximately 50% of the nurses and almost all physicians had been questioned by patients or relatives and in some instances had instigated these conversations themselves. Respondents felt these conversations were relevant to the palliative care setting and feasible. Barriers were not explored. | Barriers: NA<br><br>Facilitators: Skill (*genetics discussions*); Environmental context & resources (*patient request*); Optimism (*positive attitudes*); Professional role (*appropriate for clinician to provide genetic health information*) | 0.5 |
| Murciano-Goroff<br><br>2018<br><br>USA | To examine characteristics of oncologists who order *BRCA1* and *BRCA2* testing for breast cancer patients | Single survey<br><br>Novel instrument (unvalidated) | *N*=732 [508 (69%) medical oncologists; 224 (31%) breast surgeons)<br><br>RR 18% | Ordering of *BRCA1* and *BRCA2* testing was associated with higher innovativeness scores (p = 0.001), older age (p = 0.02), insurance coverage (p < 0.001), seeing >50% breast cancer patients in practice (p = 0.007). Lower rates of testing were associated with having >5% of patients not covered by health insurance (p = 0.05) | Barriers: Environmental context & resources (*cost of testing*)<br><br>Facilitators: Skill (*experience with somatic testing, seniority, ordering genetic testing*); Belief about consequences (*benefit for patient*); Belief about capabilities (*innovativeness increases capability*) | 0.78 |
| Loss<br><br>2018<br><br>Germany | To explore ophthalmologists' knowledge and relevance of the genetics of Age-related Macular Degeneration (AMD), their views about genetic tests and genetic | Semi-structured interviews | *N*=30 ophthalmologists<br><br>RR not specified | Hospital-based ophthalmologists had better knowledge and were more interested in the genetics of AMD than community-based ophthalmologists, who appeared to actively avoid discussing or learning more about this topic. | Barriers: Knowledge (*low knowledge scores*); Behavioural Regulation (*avoid discussions; avoids further education*); Environmental context & resources (l*ow referral rates, lack of time*); Goals (*genetic testing not useful, not relevant to care*); Belief about consequences (*genotype/phenotype relationship not established, early diagnosis not valued, psychological impact on patient & relatives*); | 0.7 |

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| | communication practices | | | | Professional role (*not appropriate to counsel asymptomatic relatives*); Reinforcement (*no expectation from patients*)<br><br>Facilitators: Environmental context & resources (p*atient request*); Optimism (f*uture benefits*); Intention (t*o engage in genetics education [workshops preferred]*)) | |
| Gonthier<br><br>2018<br><br>Canada | To assess palliative care clinicians' knowledge and explore experience discussing genetics | Single survey<br><br>Novel instrument (unvalidated) | *N*=28 [9 (32%) palliative care physicians, 19 (68%) palliative care nurses]<br><br>RR not specified | All physicians and two-thirds of nurses had discussed inherited cancer with patients or families previously. Both groups self-reported and demonstrated low knowledge but interest in further education. | Barriers: Knowledge (*Low knowledge scores*): Skill (*Low confidence obtaining family history & performing risk assessment*);<br><br>Facilitators: Intention (*To engage in education [online content or workshops preferred])* | 0.72 |
| Dearing<br><br>2018<br><br>UK | To explore perceptions and comfort of providing genetics services to palliative patients. Determine useful resources or educational training programs familiarity with their local Clinical Genetics Service | Semi-structured interviews | *N*=6 [4 (67%) palliative care nurses; 2 (33%) palliative care physicians]<br><br>RR 58% | Participants had concerns about discussing genetics in the palliative care setting but this did not necessarily act as a barrier. Palliative care nurses and physicians recognized the benefits of genetic counseling to the patient and family, but lacked confidence in their knowledge and skills to routinely integrate this into practice | Barriers: Knowledge (*Low awareness of genetic tests*); Skill (*Low confidence, uncertain how to refer*); Environmental context & resources (*Setting inappropriate, genetics should have already been addressed; poor links to genetics services, relatives have right not to know*); Belief about consequences (*Psychological impact on patient & relatives*)<br><br>Facilitators: Environmental context & resources (*Patient request, right to genetic information*); Optimism (*Positive attitudes*); Belief about consequences (*Important for relatives, psychological benefit for patients*); Professional role (*appropriate for clinician to provide genetic health information*); Belief about capabilities (*Competent to* | 0.7 |

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| | | | | | *provide genetic health information*); Intention (*Interest in further education*) | |
| Katz 2018 USA | To determine the attributes of breast surgeons which influence ordering of genetic testing for patients with newly diagnosed breast cancer | Single survey<br><br>Novel instrument (unvalidated) | *N*=377 breast surgeons<br><br>RR 77% | In an author developed "Tendency to Test" model (which included referral and testing practices and confidence discussing pros and cons of genetic testing), patients who saw surgeons at the 5th percentile had a 26% chance of receiving a genetic test, while patients who saw a surgeon at the 95th centile had a 72% chance of receiving a genetic test. | Barriers: Knowledge (*Difficulty interpreting test result*); Environmental context & resources (*Low referral rates*)<br><br>Facilitators: Skill (*Ability to recognize high risk features*) | 0.78 |
| Kurian 2017 USA | | | | | | 0.56 |
| Vajda<br><br>2017<br><br>UK | To determine the factors which influence neurologists to recommend clinical genetic testing for patients with Amyotrophic Lateral Sclerosis (ALS) | Single survey<br><br>Novel instrument (unvalidated) | *N*=167 neurologists (*n*=139 neurologists specialized in ALS care)<br><br>RR not specified | ALS specialized neurologists ($p = 9.1 \times 10^{-5}$) and those who saw ≥30 ALS patients/year ($p = 1.2 \times 10^{-4}$) more likely to offer diagnostic genetic testing for familial ALS and sporadic ALS than those not specialized in ALS or who saw fewer patients, respectively. ALS specialized neurologists ($p = 2.83 \times 10^{-5}$) and those who saw ≥30 ALS patients/year ($p = 1.7 \times 10^{-7}$) were more likely to offer predictive genetic testing to asymptomatic relatives than those who were not specialized in ALS or saw less than 30 patients a year, respectively. | Barriers: Skill (*Performing a risk assessment);* Environmental context & resources (*Lack of resources & guidelines, cost of testing*); Goals (*genetic testing not useful*); Belief about consequences (*early diagnosis not valued*)<br><br>Facilitators: Skill (*Recognizing high risk features, experience with genetic conditions, ordering genetic testing*); Environmental context & resources (*right to genetic information*) | 0.67 |
| Jacher<br><br>2017<br><br>USA | To determine use, knowledge and attitudes of nurses and physicians treating patients with Pulmonary Arterial Hypertension (PAH) towards genetic | Single survey<br><br>Novel instrument (unvalidated) | *N*=167 [79 (76.8%) pulmonologists; 24 (23%) cardiologists; 64 (29%) nurses)<br><br>RR not specified | Although attitudes were generally positive, physicians were less likely to agree that genetic counseling or testing should be offered to those with or at risk of PAH ($p < 0.04$) or that genetic testing is important for medical management than non-physicians ($p = 0.027$). Cardiologists' patients asked about genetic testing more frequently ($p = 0.019$) and had more favorable attitudes | Barriers: Knowledge (*low knowledge scores*); Environmental context & resources (*low referral rates, lack of resources, cost of testing*); Goals (*genetic counseling unnecessary*)<br><br>Facilitators: Skill (r*ecognizing high risk features*); Environmental context & resources (*patient request, collaboration with genetic services*); Optimism (*positive* | 0.83 |

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| | health information and testing | | | towards predictive genetic testing than pulmonologists (p = 0.031). | *attitudes*); Belief about consequences (*benefit to patients*) | |
| Choi<br><br>2017<br><br>Korea | To evaluate practice patterns related to hereditary ovarian cancer among Korean gynecologic physicians | Single survey<br><br>Novel instrument (unvalidated) | *N*=50 gynecology oncologists<br><br>RR not specified | Awareness of hereditary ovarian cancer among gynecology oncologists was generally high, with 76% routinely taking a family history and 87 - 94% offering genetic testing to appropriate patients. Of the respondents, 79% understood the definition of a variant of uncertain significance. | Barriers: Environmental context & resources (*privacy concerns);*<br><br>Facilitators: Knowledge (*high knowledge scores);* Skill (*able to recognize high risk features, having genetics discussions & obtaining family history*); Belief about consequences (*important for relatives*); Belief about capabilities (*competent to provide genetic health information*) | 0.45 |
| Arthur<br><br>2017<br><br>USA | To understand genetic testing practices and attitudes towards genetic testing and counseling among neurologists treating patients with ALS | Single survey<br><br>Novel instrument (unvalidated) | *N*=43 neurologists<br><br>RR 32% | 90% of neurologists ordered genetic testing in familial cases, while 30% ordered testing for sporadic cases. 98% provided genetic counseling and 91% would change their attitude if gene therapy became available. | Barriers: NA<br><br>Facilitators: Skill (*ordering genetic testing, engaging in discussions about genetics);* Optimism (*future benefit of genetic information)* | 0.61 |
| Parikh<br><br>2016<br><br>USA | To assess how family history of patients with stage II CRC influences medical oncologists' selection of Lynch Syndrome (LS) testing | Single survey<br><br>Novel instrument (unvalidated) | *N*=327 medical oncologists<br><br>RR 46% | Although rates of genetic and molecular Lynch Syndrome testing increased with strength of family history, oncologists did not routinely adhere to recommendations for universal mismatch repair immunohistochemistry screening of all colorectal cancers diagnosed under 50 years | Barriers: Knowledge (*Low awareness of genetic tests*); Skill (*difficulty performing genetic risk assessment*)<br><br>Facilitators: Skill (*ability to recognize high risk features*) | 0.83 |
| Jenkins<br><br>2016<br><br>UK | To explore the relevance and demand for psychiatric genetic counseling in the | Mixed-methods: Survey and semi-structured interviews | Survey: *N*=32 [23 (72%) psychiatric nurses; 9 (28%) psychiatric | 94% of psychiatric nurses and consultants believed a genetic counseling service for psychiatric patients would have some relevance or be very relevant. 44% felt they had little | Barriers: Knowledge (*uncertain what genetic counseling involves, low knowledge*); Skill (*Low confidence, uncertain how to refer to genetics services*); Environmental context & resources (*Lack of* | 0.8 |

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| | UK, identify possible benefits and barriers and investigate whether integration of genetic counseling and clinical psychiatry is desirable or feasible. | Novel instrument (unvalidated) | consultants]: RR 32%<br><br>Interviews: *N*=9 [3 (33%) psychiatric nurses; 6 (67%) psychiatric consultants]: RR 28% | knowledge about the genetics of psychiatric conditions, and 44% stated they had only a little information about genetics provided in their education or training. The benefits of a combined psychiatric-genetics clinic were verbalized, but the actualization of this is hampered by low patient demand, concern for further patient stigmatization and limited predictive ability of psychiatric genetic testing. | *time & resources, patients may be unable to consent*); Belief about consequences (*Over-medicalization and psychological to patient, genotype/phenotype relationship not well established*)<br><br>Facilitators: Environmental context & resources (*collaboration with genetics services*); Belief about consequences (*Positive attitudes, genetic information to inevitably inform patient care*) | |
| Gray<br><br>2016<br><br>USA | To explore the impact of introducing whole exome sequencing (WES) on oncologists. | Mixed-methods: Survey and semi-structured interviews<br><br>Novel instrument (unvalidated) | *N*=27 oncologists<br><br>RR 100% | ≥90% of oncologists were confident performing a number of genetics tasks, like taking a family history, identifying high risk families and communicating this to patients. However, they were less confident attending the psychological impact of genetic information, and concerned about being overwhelmed with information from WES. | Barriers: Knowledge (*Low awareness of genetic tests*); Skill (*Low confidence in abilities*); Environmental context & resources (*palliative care setting not appropriate*); Belief about consequences (*genotype/phenotype relationship not well established, psychological impact on patient and relatives*); Professional Role (*not appropriate to counsel asymptomatic relatives*)<br><br>Facilitators: Skill (*discussions, family history taking, ability to recognize high risk features, experience with somatic testing*); Environmental context & resources (*patient request, right to information, collaboration with genetics services*); Belief about consequences (*benefit to patient*); Professional role (*appropriate for clinician to provide genetic health information*); Belief about capabilities (*competent to provide genetic health information*) | 0.61 |
| Ferraro<br><br>2016 | To investigate genetic epilepsy testing practices | Single survey | *N*=85 neurologists | 78% of neurologists have ordered genetic testing in the last two years and 51% of neurologists' patients had | Barriers: Environmental context & resources (l*ack of resources, cost of testing*); Goals (*genetic testing not useful* | 0.28 |

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| USA | and beliefs among adult neurologists | Novel instrument (unvalidated) | RR not specified | requested testing. 88% had access to a genetics clinician | Facilitators: Skill (*ordering genetic testing*); Environmental context & resources (*patient request, collaboration with genetics services*); Belief about capabilities (*competent to provide genetic health information*) | |
| Prolla<br><br>2015<br><br>Brazil | To assess oncology nurses' knowledge and practice patterns related to hereditary breast cancer | Single survey<br><br>Novel instrument (unvalidated) | *N*=137 oncology nurses<br><br>RR 89% | 69% of oncology nurses were uncertain about what genetic counseling involves and 83% had not referred their patients, despite 81% routinely obtaining a family history. 99% of participants wanted further genetics education, with 62% preferring lectures | Barriers: Knowledge (*Low knowledge scores, uncertain what genetic counseling involves*); Skills (*Uncertain how to refer to genetics service, performing risk assessment*); Memory, attention & decision processes (*low knowledge associated with graduation year*); Environmental context & resources (*low referral rates)*<br><br>Facilitators: Skill (*routinely obtaining family history*); Intention (*to engage in genetics education [lectures preferred]*) | 0.44 |
| Eccles<br><br>2015<br><br>UK | To assess interpretation and management of a variant of uncertain significance (VOUS) in a breast cancer setting | Single survey<br><br>Novel instrument (unvalidated) | *N*=155 [63 (41%) medical oncologists; 54 (35% radiation oncologists; 38 (24%) breast surgeons]<br><br>RR 19% | Breast cancer nurses and physicians were aided when the testing laboratory provided detailed variant interpretation. Breast surgeons were more confident interpreting genetics reports than medical oncologists (p = 0.03) and more confident understanding a VUS than radiation oncologists (p = 0.003) | Barriers: Knowledge (*low awareness of genetic tests, uncertainty interpreting results*)<br><br>Facilitators: Environmental context & resources (*complex family history, detailed genetic test report assists interpretation*) | 0.83 |
| Zhou<br><br>2014<br><br>Australia, New Zealand | To investigate the perceived roles and competencies of psychiatrists in genetic risk communication | Single survey<br><br>Novel instrument (unvalidated) | *N*=140 psychiatrists<br><br>RR not specified | 96% of psychiatrists had a moderate to strong belief that genetics influenced psychiatric health, but only 43% agreed or strongly agreed their medical training had prepared them to discuss the genetic aspects of disease with their patients and 13% would feel competent ordering genetic testing. However, | Barriers: Knowledge (*Low awareness of genetic tests, uncertainty interpreting results*); Belief about consequences (*concerns about insurance discrimination*)<br><br>Facilitators: Skill (*obtaining family history, discussing genetics*); Optimism (*future benefits*); Belief about consequences | 0.94 |

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| | | | | psychiatrists strongly believed it to be their role to discuss the influence of genetics with their patients, when compared with medical geneticists and genetic counselors (p = 0.05). | (*benefit for family*); Professional role (*appropriate for clinician to provide genetic health information*); Belief about capabilities (*competent to provide genetic health information*); Intention (*to engage in genetics education [workshops preferred*) | |
| Tanabe<br><br>2014<br><br>Japan | To describe practice and awareness of hereditary breast and ovarian cancer (HBOC) in Japan and to identify areas for improvement in HBOC care | Single survey<br><br>Novel instrument (unvalidated) | *N*=307 gynecology oncologists<br><br>RR 50% | 93% of gynecology oncologists were interested in HBOC and 98% considered HBOC when consulting with patients. However, less than 1 in 5 provided genetic counseling, and less than 1 in 7 referred eligible patients onto genetic services Academic centers, dedicated cancer hospitals and centers with strong links with genetic services were more likely to integrate genetic counseling into their practice (p < 0.05) | Barriers: Knowledge (*Low knowledge scores*); Skill (*low confidence, family history taking, genetic risk assessment*); Memory, attention & decision processes (*low knowledge associated with graduation year*); Environmental context & resources (*low referral rates, lack of resources*)<br><br>Facilitators: Skill (*exposure to genetic conditions, family history taking*); Memory, attention & decision processes (*recent graduate);* Environmental context & resources (*collaboration with genetics services, working at an academic hospital*) | 0.83 |
| Salm<br><br>2014<br><br>USA | To report neurologists' and psychiatrists' genetic testing practices, attitudes, and knowledge | Single survey<br><br>Novel instrument (unvalidated) | *N*=535 [163 (30%) neurologists; 372 (70%) psychiatrists]<br><br>RR 15% | 74% of neurologists and 14% of psychiatrists had ordered genetic testing in the previous 6 months. 68% of this group thought genetic testing should be used more frequently. Half of respondents were concerned about harming the patient psychologically. Neurologists were more likely to order tests, have patients ask about genetic testing and feel more confident discussing and organizing testing (p < 0.05). | Barriers: Skill (*Low confidence*); Environmental context & resources (*Privacy concerns*); Belief about consequences (*genetic discrimination, psychological impact on patient*)<br><br>Facilitators: Knowledge (*high knowledge scores*); Skill (*Knowledge of ordering logistics, ordering genetic testing*); Environmental context & resources (*patient request*); Intention (*further education desired*) | 0.78 |
| Nippert<br><br>2014 | To describe breast surgeons' actual and preferred practice patterns | Single survey | *N*=927 breast surgeons<br><br>RR 37% | Breast surgeons from the Netherlands, Germany and UK routinely discussed family history with their patients. Physicians from France and Germany did | Barriers: Knowledge (*Uncertainty interpreting test result*); Skill (*obtaining family history*); Behavioural regulation (*avoid discussions*); Environmental context | 0.83 |

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| Germany, France, UK, Netherlands | for patients with concerns about inherited breast cancer | Novel instrument (unvalidated) | | not obtain paternal history as frequently compared to the Netherlands and UK. UK breast surgeons referred to clinical genetics for almost all aspects of genetic counseling, while those in the Netherlands, Germany and France preferred to provide genetic counseling themselves ($p < 0.05$) | & resources (*low referral rate*); Professional role (*not appropriate to counsel asymptomatic relatives, genetic counseling responsibility of genetics service*)<br><br>Facilitators: Skill (*discussions about genetics*); Professional role (*appropriate for clinician to provide genetic health information*) | |
| Monahan<br><br>2014<br><br>UK | To describe gastroenterologists' practice patterns related to inherited colorectal cancer | Single survey<br><br>Novel instrument (unvalidated) | *N*=365 [163 (45%) gastroenterologist, 144 (39%) colorectal surgeon, 58 (16%) oncologist] | 62% of physicians believed they could recognize appropriate patients for referral to clinical genetics but only 32% 'always' ordered mismatch repair immunohistochemistry for patients with bowel cancer diagnosed under 50 years. 41% wanted clear guidelines, pathways and support networks. | Barriers: Skill (*difficulty performing risk assessment*); Environmental context & resources (*lack of guidelines*)<br><br>Facilitators: NA | 0.67 |
| Klitzman<br><br>2014<br><br>USA | To examine the views and use of genetic tests among psychiatrists | Single survey<br><br>Novel instrument (unvalidated) | *N*=372 psychiatrists<br><br>RR 7% | 52% of psychiatrists have discussed genetics their patients, either by their own volition or because patients raise it. 14% had ordered a genetic test in the previous 6 months, although a sizeable minority believed testing was available for major depression (20%), obsessive-compulsive disorder (10%) and suicidality (7%). | Barriers: Knowledge (*low knowledge regarding relevant tests)*<br><br>Facilitators: Skill (*discussing genetics*); Environmental context & resources (*patient request*); Optimism (*genetic information is inevitable*) | 0.83 |
| Beitsch<br><br>2014<br><br>USA | To assess the current practice of breast surgeons in ordering breast cancer genetic testing | Single survey<br><br>Novel instrument (unvalidated) | *N*=907 breast surgeons<br><br>RR 35% | 54% of surgeons routinely ordered *BRCA1* and *BRCA2* testing and 52% felt confident performing pre- and post-genetic counseling. 63% routinely obtained a 3-generation family history. 84% expressed a desire for further genetics education. | Barriers: NA<br><br>Facilitators: Skill (*taking family history, ordering testing*); Belief about capabilities (*competent to provide genetic health information*); Intention (*to engage in further genetics education*) | 0.56 |
| Burcher<br><br>2013 | To ascertain the attitudes of oncology health | Single survey | *N*=149 [40 (27%) breast surgeons; | The large majority of nurses and physicians believed TFGT provided diagnostic clarification (84%) and risk | Barriers: NA | 0.94 |

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| Australia | professionals towards TFGT for women with breast cancer | Novel instrument (unvalidated) | 46 (31%) oncologists; 63 (42%) breast care nurses]<br><br>RR not specified | clarification (96%) although nurses were more likely to believe TFGT was useful compared to oncologists and breast surgeons (p = 0.045). 47% felt that breast surgeons are the most appropriate professional to offer TFGT. | Facilitators: Optimism (*positive attitudes, genetic information is inevitable*); Belief about consequences (*benefit to patient*); Professional role (*appropriate for clinician to provide genetic health information*) | |
| Prochniak<br><br>2012<br><br>USA | To assess practice patterns for hereditary colorectal cancer, assess risk assessment and factors which influence decision to refer to clinical genetics or organize genetic testing independently | Single survey<br><br>Novel instrument (unvalidated) | *N*=290 [185 (64%) gastroenterolo-gists; 105 (36%) colorectal surgeons] | Physicians who graduated longer ago were more likely to refer high risk patients to outside clinical genetics services (p = 0.01), while physicians with formal training in genetics were less likely to refer high-risk patients to clinical genetics services (p =0.03). Physicians with higher knowledge scores were not more likely to have ordered genetic testing or referred to clinical genetics. Physicians who refer to clinical genetics rather than order genetic testing independently were more concerned about genetic discrimination, value the expertise of clinical genetics and endorsed their ability to conduct a risk assessment (p < 0.05) | Barriers: Knowledge (*low knowledge of relevant tests*); Belief about consequences (*discrimination concerns*)<br><br>Facilitators: Skill (*experience with somatic testing*); Environmental context & resources (*existence of guidelines*); Belief about capabilities (*competent to provide genetic health information*) | 0.83 |
| Burke<br><br>2012<br><br>UK | To identify areas of genetic knowledge deficiency in hemophilia nurses | Two surveys<br><br>Novel instruments (unvalidated) | First survey: *N*=58 hemophilia nurses, RR 75%<br><br>Second survey: *N*=17 hemophilia nurses, RR 41% | 98-100% of hemophilia nurses rated various genetics tasks as relevant to their practice (identifying individuals with genetic conditions, taking a family history, recognizing mode of inheritance and making a referral to clinical genetics). Only 58% felt it was relevant for them to order a genetic test. 88% noted they had learnt about genetics "on-the-job", but would prefer formal training sessions that included applied genetics information. | Barriers: Skill (*low confidence, obtaining family history, perform risk assessment*)<br><br>Facilitators: Intention (*to engage in further education [workshops preferred]*) | 0.55 |

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| Quillin<br><br>2011<br><br>USA | To describe palliative clinicians' DNA banking practices and their desired resources to assist with this practice | Single survey<br><br>Adapted "National Cancer Institute's Physician Survey on Cancer Susceptibility Testing" survey | *N*=49 palliative oncology physicians<br><br>RR 37% | 11% of palliative oncologists were aware of commercial DNA banking and 39% did not feel qualified to offer this to their patients. 18% (*n*=9) correctly responded to all knowledge questions. | Barriers: Knowledge (*low knowledge scores, low awareness of relevant tests*)<br><br>Facilitators: Skill (*assessing genetic risk*); Intention (*to engage in further education [online content & genetic counselor on staff preferred]*) | 0.78 |
| Lillie<br><br>2011<br><br>UK | To explore nurses' perceptions of addressing familial cancer assessment within the palliative care context | Semi-structured interviews | *N*=10 palliative care nurses<br><br>RR unknown (sampling strategy not specified) | Palliative care nurses were concerned about distressing patients and relatives by discussing genetic information. Innovative training methods could improve nurses' knowledge, skills and confidence in addressing genetic issues at the end of life. | Barriers: Skills (*assessing genetic risk*); Memory, attention and decision processes (*genetics detracts from more important issues*); Environmental context & resources (*palliative care inappropriate setting*); Belief about consequences (*psychological impact on patient & relatives*); Professional role (*genetic counseling responsibility of genetics clinicians*)<br><br>Facilitators: NA | 0.65 |
| Kelly<br><br>2011<br><br>USA | To describe nurses' knowledge and attitudes about family history assessment of CRC | Structured interviews<br><br>Novel instrument (unvalidated) | *N*=16 gastroenterology nurses<br><br>RR not applicable (convenience sample) | 87% (*n*=14) of gastroenterology nurses overestimated the prevalence of inherited CRC. 25% (*n*=4) described lack of guidelines as a barrier. | Barriers: Knowledge (*low knowledge scores*); Environmental context & resources (*lack of guidelines*)<br><br>Facilitators: NA | 0.44 |
| Graves<br><br>2011<br><br>USA | To explore perceptions of genetic counseling and testing for African American | Semi-structured interviews | *N*=13 [8 (62%) medical oncologists; 5 (38%) breast surgeons] | Medical oncologists and surgeons perceived African American women as having unique barriers to accessing genetic counseling or testing due to potential psychological harm to themselves or their families, cost of | Barriers: Knowledge (*uncertainty interpreting test result*); Skill (*performing risk assessment*); Environmental context & resources (*cost of testing*); Belief about consequences (*insurance discrimination,* | 0.65 |

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| | women at high-risk for breast cancer | | RR not applicable (snowball sampling) | counseling or testing and concerns about insurance discrimination. Increasing the number of ethnically and culturally diverse clinicians may improve clinical genetics access for these groups. | *psychological impact on patients & relatives*)<br><br>Facilitators: Environmental context & resources (*family support*); Belief about consequences (*psychological benefit for patient*); Professional role (*appropriate for clinician to provide genetic health information)* | |
| Bonter<br><br>2011<br><br>Canada | To describe cardiologists' and oncologists' practice patterns, attitudes and barriers towards ordering genetic testing | Single survey<br><br>Novel instrument (unvalidated) | *N*=194 [102 (53%) cardiologists; 92 (47%) oncologists]<br><br>RR 10% | Canadian oncologists & cardiologists recognized the benefits of genetic information for their patients, but faced a number of barriers to integration. Oncologists were more likely to have graduate training in genetics and agreed that genetic test results influenced patient management than cardiologists ($p<0.05$), but were also more likely to agree results took too long to be useful | Barriers: Knowledge (*uncertainty interpreting test result*); Skill (*low confidence*); Environmental context & resources (*lack of guidelines, cost of testing*); Goals (*genetic testing not useful*); Belief about consequences (*genotype/phenotype relationship not established*)<br><br>Facilitators: Skill (*ordering testing*); Environmental context & resources (*patient request*); Belief about consequences (*benefit for patient*); Intention (*to engage in further education*) | 0.72 |
| Metcalfe<br><br>2010<br><br>UK | To determine adult hospice nurses' perception of and confidence in carrying out genetics-related activities within a palliative care context | Mixed-methods<br><br>Single survey and semi-structured interviews<br><br>Novel instrument (unvalidated) | Survey: *N*=328 palliative care nurses, RR 29%<br><br>Interviews: *N*=8 palliative care nurses (clinical educators) | Palliative care nurses rated most genetic-related issues as very important, but lacked confidence in integrating genetics into their practice. Participants with previous genetics training were more confident and rate the importance of genetics-related activities higher than those with no previous training ($p<0.01$). Senior palliative care nurses had better levels of confidence in carrying out genetics-related activities than staff nurses ($p < 0.05$) | Barriers: Knowledge (*low knowledge regarding relevant tests*); Skill (*low confidence, genetic risk assessment*); Behavioural regulation (*genetic counseling should be provided by a physician*); Environmental context & resources (*genetic health information provision should have already occurred, low referral rates*); Belief about consequences (*psychological impact on relatives*) | 0.94 |

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| | | | | | Facilitators: Skill (*senior staff more confident*); Belief about capabilities (*confident addressing psychosocial issues*) | |
| Claybrook<br><br>2010<br><br>USA | To investigate oncologists' use of genetic services for colorectal cancer patients and their families and determine barriers for referral to genetics services | Single survey<br><br>Novel instrument (unvalidated) | *N*=53 oncologists<br><br>RR 35% | While 96% of oncologists were interested in genetics services for their patients and 83% knew of an appropriate genetics service, only 58% had made a referral in the past. Of the 42% who had not referred previously, the most common reason was not having an appropriate patient to refer. | Barriers: Skill (*uncertain how to make a referral, genetic risk assessment*); Environmental context & resources (*low referral rates, cost of testing*)<br><br>Facilitators: Skill (*ability to recognize high risk patients*); Environmental context & resources (*patient request*) | 0.78 |
| Grant<br><br>2009<br><br>USA | To assess views about type 2 diabetes genetic testing related to diabetes prediction and lifestyle adherence | Single survey<br><br>Novel instrument (unvalidated) | *N*=175 diabetes nurses and physicians<br><br>RR 12% | Diabetes nurses and physicians had more positive attitudes about genetic testing in general and would instigate early treatment based on a 'high-risk' result when compared to generalists (p <0.03). | Barriers: Environmental context & resources (*privacy concerns*)<br><br>Facilitators: Optimism (*positive attitudes*); Belief about consequences (*medical benefit for patient*) | 0.83 |
| Hoop<br><br>2008<br><br>USA | To understand psychiatrists' preparedness to provide genetic services<br><br>a | Single survey<br><br>Novel instrument (unvalidated) | *N*=45 psychiatrists<br><br>RR 48% | Psychiatrists were more likely to refer patients to clinical genetics if they had intellectual disability or multiple congenital abnormalities in addition to their mental illness than patients who did not have additional features (p < 0.03). No psychiatrists who trained > 5 years ago (*n*=27/45, 60%) felt competent to deliver genetic counseling in comparison to those with more recent training (p <0.02) Psychiatrists who trained within the last 5 years had better knowledge of genetic testing laboratories (p < 0.01). | Barriers: Knowledge (*low knowledge regarding relevant tests*); Skill (*low confidence*); Memory, attention & decision processes (*low knowledge associated with graduation year*); Environmental context & resources (*low referral rates, lack of resources*)<br><br>Facilitators: Skill (*family history taking, discussion about genetics*); Memory, attention & decision processes (*recent graduate);* Environmental context & resources (*complex phenotype increases chance of referral*); Belief about consequences (*benefit to family*); Professional role (*appropriate for clinician to provide genetic counseling to patient &* | 0.94 |

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| | | | | | | *relatives*); Belief about capabilities (*competent to provide genetic health information*) | |
| | b | To ascertain psychiatrists' attitudes towards genetic testing and ethical/legal patient safeguards | | | Psychiatrists had generally positive attitudes towards genetic testing for psychiatric conditions but were cognizant of ethical issues such as privacy, insurance and discrimination. Those who had genetics training within the last 5 years viewed genetic testing as less useful than those without genetics training within the last 5 years ($p < 0.01$). | Barriers: Environmental context & resources (*privacy concerns, cost of testing*); Belief about consequences (*insurance & employment discrimination*)<br><br>Facilitators: Optimism (*positive attitudes, future benefits*) | 0.83 |
| Agnese<br><br>2006<br><br>USA | To determine surgical oncologists' knowledge and use of cancer genetic counseling services | | Single survey<br><br>Novel instrument (unvalidated) | *N*=364 oncologists<br><br>RR 24% | Of the respondents, 98% obtain family history information and 94% discuss hereditary cancer syndromes routinely. 83% had access to genetic counseling services at their institution and 38% referred to these services frequently. 60% had personally ordered genetic testing for their patients previously. | Barriers: Environmental context & resources (*low referral rates*)<br><br>Facilitators: Skill (*taking family history, genetics discussions, ordering genetic testing*) | 0.55 |
| Van Langen<br><br>2005<br><br>Netherlands | To investigate views of Dutch cardiologists about cardiogenetic roles, responsibilities and needs | | Single survey<br><br>Novel instrument (unvalidated) | *N*=189 cardiologists<br><br>RR 33% | 43% of cardiologists felt it was their sole responsibility to inform patients about HCM while 41% preferred to share responsibility with geneticists. 35% saw ordering genetic testing for HCM patients as their responsibility while 53% did not feel responsible for organizing genetic testing for asymptomatic relatives. 35% preferred geneticists to discuss risks to offspring. Experienced | Barriers: Environmental context & resources (*lack of guidelines*); Professional role (*not appropriate to counsel asymptomatic relatives*)<br><br>Facilitators: Skill (*genetics discussions*); Environmental context & resources (*collaboration with genetics services*); Intention (*to engage in further education [workshops & online content preferred]*) | 0.72 |

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| | | | | cardiologists preferred to collaborate with geneticists when organizing genetic testing (p = 0.021). | | |
| Finn<br><br>2005<br><br>USA | To examine psychiatrists' knowledge and attitudes regarding the use of genetic information. | Single survey<br><br>Novel instrument (unvalidated) | *N*=352 psychiatrists<br><br>RR 54% | Median psychiatric genetic knowledge scores were 5/12 (42%) and median general genetic knowledge scores were 4/9 (44%). Those who had more recently graduated had better knowledge than those who graduated longer ago (p<0.05). Psychiatrists with higher knowledge scores also felt more competent to deliver genetic counseling and were less likely to be directive in their counseling than those with lower knowledge (p<0.05). | Barriers: Knowledge (*low knowledge scores*); Skill (*low confidence*); Memory, attention & decision processes (*low knowledge associated with graduation year)*; Environmental context & resources (*low referral rates*); Belief about consequences (*insurance & employment discrimination*)<br><br>Facilitators: Knowledge (*high knowledge scores*); Skill (*family history taking, genetics discussions*); Memory, attention & decision processes (*recent graduate)*; Environmental context & resources (*complex phenotype increases chance of referral*); Optimism (*future benefits*); Professional role (*appropriate for clinician to provide genetic health information*); Intention (*to engage in further education [workshops, online content & scientific journals preferred]*) | 0.89 |
| Cox<br><br>2004<br><br>Canada | To understand nurse and physician knowledge and value of hereditary aspects of polycystic kidney disease (PKD), and how this influences discussions and attitudes towards predictive testing and renal screening | Semi-structured interviews and focus group | *N*=8 [5 (63%) nephrologists; 3 (37%) nephrology nurses]<br><br>RR not applicable (purposive sampling) | Nephrologists and nephrology nurses had concerns about the utility of genetic counseling and testing in PKD, and the psychological impact genetics can have on patients and families, but did appreciate the potential benefit of genetic information if it relates to medical management or family planning. | Barriers: Knowledge (*uncertain what genetic counseling involves*); Skill (*low confidence*); Memory, attention & decision processes (*genetics detracts from more important issues*); Behavioural regulation (*avoid discussions*); Environmental context & resources (*lack of time & resources, privacy concerns, cost of testing*); Goals (*genetic counseling unnecessary, not relevant to care*); Belief about consequences (*early diagnosis not valued, insurance discrimination, psychological* | 0.75 |

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| | | | | | *impact on patient*); Reinforcement (*previous negative experience with genetic service*)<br><br>Facilitators: Environmental context & resources (*written resources for patients*); Belief about consequences (*important for family, medical benefit for patient*) | |
| Van Langen<br><br>2003<br><br>Netherlands | To investigate Dutch cardiologists' knowledge, practices and educational needs regarding genetics and Hypertrophic Cardiomyopathy (HCM) | Single survey<br><br>Novel instrument (unvalidated) | *N*=197 cardiologists<br><br>RR 33% | 41% of cardiologists did not inform their patients about the genetic aspect of HCM. Self-rated knowledge scores varied between 3.3-5.1/10. Cardiologists with higher self-rated knowledge were more likely to have subspecialized (p = 0.06), to discuss genetic implications for children (0.06) and to have a working relationship with a clinical geneticist (p = 0.07). 94% desired regular refresher courses regarding genetics and HCM. | Barriers: Knowledge (*low knowledge scores & relevant tests, uncertainty interpreting results*); Skill (*obtaining family history, genetic risk assessment*); Behavioural regulation (*avoid discussions*); Environmental context & resources (*low referral rates, lack of guidelines, poor links to genetics services*)<br><br>Facilitators: Environmental context & resources (*collaboration with genetics services*): Belief about consequences (*important for family*); Intention (*to engage in further education [workshops or online content preferred]*) | 0.83 |
| Batra<br><br>2002<br><br>USA | To investigate knowledge and attitudes of gastroenterologists toward genetic services for colorectal cancer patients and their families | Single survey<br><br>Novel instrument (unvalidated) | *N*=258 adult gastroenterologists<br><br>RR 33% | 99% of gastroenterologists did obtain family history information but only 39% collected a three-generation pedigree. Those reluctant to recommend genetic testing did not perceive testing as standard of care and believe testing has low utility (p < 0.05). Reluctance to refer for genetic counseling was associated with perceived benefits and cost, although these physicians were more likely to provide their own genetic counseling (p <0.05) | Barriers: Knowledge (*low knowledge regarding relevant tests*); Skill (*obtaining family history, genetic risk assessment*); Behavioural regulation (*avoid discussions*); Environmental context & resources (*cost of testing*); Goals (*genetic counseling unnecessary*, *genetics genetic testing not useful*); Reinforcement (*not standard of care*)<br><br>Facilitators: Skill (*taking family history*); Belief about capabilities (*competent to provide genetic health information*) | 0.67 |

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| Culver<br><br>2001<br><br>USA | To assess oncologists' opinions about genetic testing and measured extent to which oncologists refer patients to clinical genetics services | Single survey<br><br>Novel instrument (unvalidated) | *N*=135 oncologists [59 (43%) medical; 36 (27%) radiation; 30 (22%) hematologic; 7 (5%) gynecological; 2 (2%) surgical; 1 (1%) urological]<br>RR 51% | 93% obtained a three-generation pedigree. 79% discussed genetic testing with their patients but only 22% had offered to organize testing. 16% had previously referred a patient to a genetics service. 61% did not have necessary resources or staff to offer genetic testing. 76% felt they were more knowledgeable about genetics than other nurses and physicians. Those who felt more knowledgeable were more likely to have the necessary resources to offer genetic testing ($p = 0.03$) and greater interest in continuing education ($p < 0.001$). | Barriers: Skill (*low confidence*); Behavioural regulation (*genetic counseling should be provided by a physician*); Environmental context & resources (*low referral rates, lack of time & resources*); Goals (*genetic counseling unnecessary*);<br><br>Facilitators: Skill (*family history taking, genetics discussions*); Belief about capabilities (*competent to provide genetic health information*); Intention (*to engage in further education*) | 0.67 |

SUPPLEMENTARY FILE A7: DESCRIPTION OF THE BARRIERS AND FACILITATORS MAPPED USING THEORETICAL DOMAINS FRAMEWORK

| TDF domain | TDF domain definition[1] | Barriers identified | | Facilitators identified | |
|---|---|---|---|---|---|
| | | n (%) of articles | Themes | n (%) of articles | Themes |
| Knowledge | An awareness of the existence of something | 25 (52%) | Low general and specific (to clinical discipline) knowledge<br>Low awareness of relevant genetic tests<br>Low understanding of genetic test results | 3 (6%) | Higher genetic knowledge levels improve ability to discuss genetic issues with patients |
| Skill | An ability or proficiency acquired through practice | 25 (52%) | Low confidence related to genetic counseling tasks (obtaining family history, assessing genetic risk, organizing genetic testing, interpreting genetic test result) | 30 (63%) | Some family history information is obtained<br>Discussions about genetics with patients do occur Oncologists and neurologists report ordering genetic testing<br>Experience ordering tumor genetic testing feel better equipped to provide integrate genetic and genomic mainstreaming |
| Memory, attention & decision processes | The ability to retain information, focus selectively on aspects of the environment and choose between two or more alternatives | 8 (17%) | Discussions about genetics detracts attention away from more important topics | 3 (6%) | Recently graduated nurses and physicians have higher knowledge scores and feel better equipped to integrate genetics and genomics into practice than their more experienced counterparts |
| Behavioural regulation | Anything aimed at managing or changing objectively observed or measured actions | 7 (15%) | Physicians make an active decision not to discuss genetics with their patients where they consider the negative consequences and see no clinical benefit | 0 (0%) | Not reported |
| Environmental context & resources | Any circumstance of a person's situation or environment that discourages or encourages the development of skills and abilities, independence, social competence, and adaptive behaviour | 35 (73%) | Referral rates to clinical genetics services are low, due to lack of time, resources and guidelines<br>The cost of genetic testing is perceived as prohibitive<br>The potential for privacy breaches of genetic information | 23 (48%) | When prompted by patients, most nurses and physicians will engage in genetic counseling<br>Close working relationships or collaboration with clinical genetics services may improve confidence to integrate genetics and genomics into practice |

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| Social influence | Those interpersonal processes that can cause individuals to change their thoughts, feelings or behaviours | 0 (0%) | Not reported | 0 (0%) | Not reported |
| Belief about consequences | Acceptance of the truth, reality or validity about outcomes of a behaviour in a given situation | 16 (33%) | The psychological impact of genetic information on patient and relatives<br>The potential for insurance and employment discrimination | 18 (38%) | The medical or psychological benefit of genetic information for their patient and/or relatives |
| Optimism | The confidence that things will happen for the best or that desired goals will be obtained | 0 (0%) | Not reported | 14 (29%) | Positive attitudes towards genetic information<br>Genetic information benefits society as a whole<br>Genetic information will be integrated into routine clinical care in the future |
| Professional role and identity | A coherent set of behaviours and displayed personal qualities of an individual in a work setting | 7 (15%) | Counseling asymptomatic relatives about genetic risk is not perceived as part of the nurse or physician's role | 11 (29%) | Providing genetic health information is appropriate in the context of the nurse's or physician's clinical role |
| Reinforcement | Increasing the probability of a response by arranging a dependent relationship, or contingency, between the response and a given stimulus | 4 (8%) | The perception that patients don't expect to be provided with genetic information | 0 (0%) | Not reported |
| Goals | Mental representation of outcomes or end states that an individual wants to achieve | 11 (23%) | Where genetic information is not useful for patient care, nurses and physicians are less likely to engage in discussions about genetics | 0 (0%) | Not reported |
| Intentions | A conscious decision to perform a behaviour or a resolve to act in a certain way | 0 (0%) | Not reported | 16 (33%) | Recognition of the important of genetic information and intention to engage in further genetics education that is relevant to their clinical discipline.<br>Lectures, workshops or online content delivery preferred |

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| Belief about capabilities | Acceptance of the truth, reality, or validity about an ability, talent, or facility that a person can put to constructive use | 0 (0%) | Not reported | 0 (0%) | Belief that nurses and physicians are capable of providing genetic counseling with appropriate training and support |
| Emotion | A complex reaction pattern, involving experiential, behavioural, and physiological elements, by which the individual attempts to deal with a personally significant matter or event) | 0 (0%) | Not reported | 0 (0%) | Not reported |
| 1 Cane J, O'Connor D, Michie S. Validation of the theoretical domains framework for use in behaviour change and implementation research. Implement Sci. 2012;7(37):1-17. | | | | | |

# Appendix B – supplementary files related to research design

## CONTENTS

SUPPLEMENTARY FILE B1: MID-POINT DATA CONNECTION JOINT DISPLAY TABLE

| **Study 1: Systematic review (QUANT)** | **Study 2a: Genetic HPs (QUAL)** | **Study 2b: PC HPs (QUAL)** | **Study 2: Scoping review (QUANT)** | **Study 4: PC & Genetic HP Questionnaire (QUANT; green: both surveys, blue: genetic survey, yellow: PC survey)** | **Existing or new item** | **Justification for modification or addition** |
|---|---|---|---|---|---|---|
| Not mentioned | *Participants described the value they could add to conversations about genetics with palliative patients and families. However, some wanted to improve their own palliative care knowledge to ensure they manage these discussions appropriately. "From a genetic counselling point of view, I would be keen to […] have had more training in the space. I think those, particularly end-of-life* | Not mentioned | Not mentioned | **3. Have you ever received any training in communicating with patients at the end of life or with bereaved families?** | Existing | NA |
| | | | | Yes (go to Q3a) | | |
| | | | | No (go to Q3d) | | |
| | | | | **3a. What area(s) did you study? (Please indicate all that are relevant.)** | Existing | |
| | | | | Communicating with cancer patients | | |
| | | | | Communication skills with patients at the end of life | | |
| | | | | Bereavement counselling | | |
| | | | | Other (please explain) | | |
| | | | | **3b. What type of training have you received in communicating with patients at the end of life** | Existing | |

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| | *conversations, they're quite confronting"* | | | **or with bereaved families? (Please indicate all that are relevant.)** | | |
| | | | | Degree/diploma | | |
| | | | | Short course/module over at least two sessions | | |
| | | | | One-off lecture/seminar/workshop | | |
| | | | | On-line short course or Massive Online Open Course (MOOC) | | |
| | | | | Course/session on communication with people receiving palliative care as part of another course or study day | | |
| | | | | Course/session on bereavement counselling | | |
| | | | | Private study (e.g. reading papers) | | |
| | | | | Other (please explain) | | |
| | | | | **3c. How long has it been since you last received training or education in communicating with patients at the end of life or with bereaved families?** | Existing | |
| | | | | Less than 12 months | | |
| | | | | 1 to 2 years | | |
| | | | | 3 to 5 years | | |

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| | | | | 6 to 10 years | | |
| | | | | More than 10 years | | |
| | | | | **3d. Are you interested in receiving training in communicating with patients at the end of life or with bereaved families?** | New | Determine preferred education modality |
| | | | | Yes (go to Q3e) | | |
| | | | | No (go to Q3f) | | |
| | | | | **3e. What type of training would you like to receive in communicating with patients at the end of life or with bereaved families? (Please indicate all that are relevant.)** | New | |
| | | | | Degree/diploma | | |
| | | | | Short course/module over at least two sessions | | |
| | | | | One-off lecture/seminar/workshop | | |
| | | | | On-line short course or Massive Online Open Course (MOO | | |
| | | | | Course/session on communication with people receiving palliative care as part of another course or study day | | |
| | | | | Course/session on bereavement counselling | | |

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|---|---|---|---|---|---|---|
| | | | | Private study (e.g. reading papers) | | |
| | | | | Other (please explain) | | |
| | | | | **3f. What is the main reason you are not interested in receiving this training?** | New | Assess reasons for not wanting education (i.e. barriers to education) |
| | | | | Lack of time to receive training | | |
| | | | | I have other education/training priorities | | |
| | | | | I already know a lot about communicating with patients at end-of-life & bereaved families | | |
| | | | | Communicating with patients at end-of-life & bereaved families is not relevant to my work | | |
| | | | | Other (please explain) | | |
| | | | | **3g. Please add any comments about the training you have received or would find helpful in communicating with patients at the end of life of with bereaved families (free text response)** | Existing | NA |

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| *Nurses and physicians expressed their intention to engage in continuing professional education, demonstrating the need for increased genetic literacy. Most nurses and physicians preferred clinically relevant education in the form of workshops, lectures, or online content* | *Participants conveyed their sense that genetics is not a priority for palliative care health professionals because of misunderstandings related to the value of genetic information.* | *Participants had a variety of suggestions to improve their genetics knowledge, including a genetics module in palliative care trainee curricula, a compulsory oncology term during palliative medicine physician training, lectures delivered by genetics clinicians and inclusion of genetic research in palliative care conferences and journals* | *Health professionals are supported by processes that enhance their skills to communicate efficiently and empathically families* | **3. Have you ever received any training in family history risk assessment, genetic testing and/or genomic testing?** | Existing | NA |
| | | | | Yes (go to Q3a) | | |
| | | | | No (go to Q3d) | | |
| | | | | **3a. What area(s) did you study? (Please indicate all that are relevant.)** | Existing | |
| | | | | Family history risk assessment | | |
| | | | | Genetics and/or genetic testing | | |
| | | | | Genomics and/or genomic testing | | |
| | | | | Other (please explain) | | |
| | | | | **3b. What type of training have you received in family history risk assessment, genetic testing and/or genomic testing? (Please indicate all that are** | Existing | |
| | | | | Degree/diploma | | |
| | | | | Short course/module over at least two sessions | | |
| | | | | One-off lecture/seminar/workshop | | |
| | | | | On-line short course or Massive Online Open Course (MOO | | |

| | | | | | | |
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| | | | | Courses/sessions on genetics/genomics as part of another course or study day | | |
| | | | | Private study (e.g. reading papers) | | |
| | | | | Other (please explain) | | |
| | | | | **3c. How long has it been since you last received training or education in family history risk assessment, genetic testing and/or genomic testing?** | Existing | |
| | | | | Less than 12 months | | |
| | | | | 1 to 2 years | | |
| | | | | 3 to 5 years | | |
| | | | | 6 to 10 years | | |
| | | | | More than 10 years | | |
| | | | | **3d. Are you interested in receiving training in family history risk assessment, genetic testing and/or genomic testing?** | New | Determine preferred education modality |
| | | | | Yes (go to Q3e) | | |
| | | | | No (go to Q3f) | | |
| | | | | **3e. What type of training would you like to receive in family history risk assessment, genetic testing and/or genomic** | New | |

| | | | | | | |
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| | | | | **testing? (Please indicate all that are relevant)** | | |
| | | | | Degree/diploma | | |
| | | | | Short course/module over at least two sessions | | |
| | | | | One-of lecture/seminar/workshop | | |
| | | | | On-line short course or Massive Online Open Course (MOO | | |
| | | | | Courses/sessions on genetics/genomics as part of another course or study day | | |
| | | | | Private study (e.g. reading papers) | | |
| | | | | Other (please explain) | | |
| | *Participants thought palliative care health professionals might avoid discussions about genetics because they ... do not see genetics as relevant or part of their role. "You know, I've heard things said […] to families and patients, "Do you* | *Engagement with ongoing professional education about genetics was not a priority for participants who described genetics as irrelevant. Instead, they selected other educational topics when choosing professional* | | **3f. What is the main reason you are not interested in receiving this training?** | New | Assess reasons for not wanting education (i.e. barriers to education) |
| | | | | Lack of time to receive training | | |
| | | | | I have other education/training priorities | | |
| | | | | I already know a lot about genetics/genomics | | |
| | | | | Genetics/genomics is not relevant to my work | | |

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| | *really want to spend your last days focusing on whether this might be hereditary or not, instead of just enjoying what time you have left?", which is really disconcerting to hear, because I think both can be done"* | *development opportunities* | | Other (please explain) | | |
| NA | NA | NA | NA | **3g. Please add any comments about the training you have received or would find helpful about genetics/genomics (free text respons** | Existing | NA |
| Not mentioned | *Most genetic health professionals' palliative care had experience with storing DNA at end-of-life when the patient was close to death* | Not mentioned | Not mentioned | **4. Have you ever been involved in facilitating DNA banking/testing for people receiving palliative care?** | Existing | NA |
| | | | | Yes | | |
| | | | | No | | |
| | | | | Not sure | | |
| | | | | **4a. In your experience, at what point did you usually become involved?** | Existing | |
| | | | | When the patient commences palliative care | | |

| | | | | | | |
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| | | | | When the patient is close to death | | |
| | | | | After the patient has died | | |
| | | | | It has never been raised in my experience | | |
| | | | | Not sure | | |
| | | | | Other (please specify) | | |
| *In some specialties, family history information was routinely obtained,although the extent of the family history was not always adequate. A smaller number of articles reported that physicians did assess genetic risk, however, confidence in family history and individual risk assessment was low* | Not mentioned | *Reports varied among participants about how frequently their work interfaced with genetics. Some reported regular discussions about genetics, but most said this occurs infrequently.* | *The second, an English palliative care policy for patients with neurological disease, recommended being aware of the psychological impact of a positive family history on the patient, including fear of the disorder and of their children developing the same disease* | **4. Please indicate how often you are involved in the following activities in your practice: (never, occassionally, sometimes, usually, always)** | Existing | NA |
| | | | | Taking a family health history | | |
| | | | | Drawing a three-generation family tree (pedigre | | |
| | | | | Making a genetic risk assessment | | |
| | | | | **5. In your experience, at what point in the patient's palliative care trajectory is a family health history usually taken?** | Existing | NA |
| | | | | When the patient commences palliative care | | |
| | | | | When the patient is close to death | | |
| | | | | After the patient has died | | |

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| | | | | Family health history is not taken<br>Not sure<br>Other (please explain) | | |
| *However, if patients raised questions or concerns about genetics, nurses and physicians did engage in these discussions.* | *Their experience was that relatives often initiated referrals to the genetics service and were engaged in learning about their risk.* | *A few reported initiating conversations about genetics with relatives and found that questions about genetic risk were often already on the relative's mind.* | Not mentioned | **5. In your experience who usually initiates the request for genetic testing?**<br>Patients<br>Family members<br>Palliative care health professionals<br>Genetics health professionals<br>Oncology health professionals<br>It has never been raised in my experience<br>Not sure<br>Other (please specify) | Existing | NA |
| NA | NA | NA | NA | *Please note, for the purposes of this survey: DNA banking, as opposed to DNA testing, is the process of obtaining a DNA sample (usually blood, saliva or buccal) and storing this sample in a laboratory, without any request for DNA testing. Please consider DNA banking for future* | New | For a shared understanding of the terms used in this survey (i.e. DNA banking) |

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| | | | | *CLINICAL use only. This means the DNA would be used for future DNA testing to help understand genetic risk for relatives and NOT used for research purposes.* | | |
| | | | | **6a. Please indicate approximately how often you have been involved in the following activities in the last 12 months: (zero, once or twice, three to five times, six to ten times, more than 10 times, I have not worked clinically in the last 12 months)** | See individual items | |
| Not mentioned | *Noting palliative care health professionals' expert communication skills, participants thought basic genetics education, particularly related to the importance of the proband sample and process of DNA banking, could be* | *PC HPs recounted multiple conversations about the role of genes in disease development, caring for people with genetic conditions or helping to organise genetic investigations* | Not mentioned | Identifying a patient receiving palliative care who is eligible for DNA banking/testing. | Existing | NA |
| Not mentioned | | | | Initiating a discussion about DNA banking/testing with a patient receiving palliative care or their relative. | Existing | NA |

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| | *sufficient to prepare them for genetics discussions.* | | | | | |
| *Nurses and physicians reported the value of close working relationships or collaboration with clinical genetics professionals* | *Participants valued a multidisciplinary approach to care, but portrayed a lack of collaboration, communication and professional relationships between palliative care and genetic health professionals.* | *Participants described the impact of professional relationships with genetics clinicians, or lack thereof, on their ability to access genetics services. Participants suggested co-locating palliative care and genetics departments to build better relationships, improve communication, access timely advice and encourage a multidisciplinary approach to care* | | Providing advice to a palliative care health professional about one of their patients | Modified | Simplified wording and mirrored structure/word choice to enable comparison to the PC HP survey |
| | | | | Seeking advice from a genetics health professional about one of your palliative patients | Modified | Simplified wording and mirrored structure/word choice to enable comparison to the PC HP survey |
| *Nurses and physicians infrequently referred patients to clinical genetics services…* | *Some speculated that late referrals to palliative care (for example, from oncology) might affect the palliative* | *Nonetheless, many recounted multiple conversations about the role of genes in disease development, caring* | | Receiving a referral from a palliative care health professional for a patient receiving palliative care | New | To determine how often genetic HPs recalled receiving referrals for PC patients and to mirror PC survey to enable comparison |

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| | *care health professionals' ability to identify the need for a genetics discussion* | *for people with genetic conditions or helping to organise genetic investigations* | | Referring a patient to specialist genetics services. | Existing | NA |
| *Oncologists and neurologists were most likely to order genetic testing. There were no reports of nurses or physicians from other specialties ordering testing. Most nurses and physicians had low awareness of genetic tests relevant to their area of practice. They also had difficulty interpreting a genetic test result.* | Not mentioned | | | Taking consent for DNA banking/testing from a patient | Existing | NA |
| | | | | Taking consent for DNA banking/testing from a relative of a patient receiving palliative care. | Existing | NA |
| | | | | Facilitating collection of a DNA sample from a patient receiving palliative care | Modified | Changed wording from 'taking DNA' to 'faciliating collection' because pilot study participants explained they did not personally collect the sample, but were involved in organising the collection |
| | | | | Disclosing genetic/genomic test results to a patient receiving palliative care. | Existing | NA |
| | | | | Disclosing a deceased patient's genetic/genomic test results to bereaved relatives | Existing | NA |

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| | | | | Providing genetic counselling to a patient receiving palliative care | New | Reports of frequency of genetic HPs involvement with PC patients varied, so this question assessed frequency |
| Not mentioned | | | | Cared for a palliative patient with an underlying genetic condition | New | Reports of frequency of PC HPs involvement with patients with genetic conditions varied, so this question assessed frequency |
| Not mentioned | | *Clinicians should always check if genetics has been addressed: "I think that the responsibility of me is to ask […] if there's anything suspicious, I'll say 'Have you ever had an opportunity to talk about the genetics of this or whether this is inherited?', 'Are you worried about that kind of thing?" And it's amazing, like anecdotally, I can't give you a figure, it's amazing how many patients have said to* | | Checked if my palliative patient had already had an opportunity to discuss genetics before coming under my care (eg. reviewing medical records, asking referrer or asking patient or relative directly) | New | Assessed how often PC HPs would check if their patients had genetics addressed, based on reports in phase 1 |

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| | | *me, 'You're the first person to ask me that question'. Even oncology patients."* | | | | |
| *Nurses and physicians reported the value of close working relationships or collaboration with clinical genetics professionals* | *Participants valued a multidisciplinary approach to care, but portrayed a lack of collaboration, communication and professional relationships between palliative care and genetic health professionals.* | *Participants described the impact of professional relationships with genetics clinicians, or lack thereof, on their ability to access genetics services. Participants suggested co-locating palliative care and genetics departments to build better relationships, improve communication, access timely advice and encourage a multidisciplinary approach to care* | Not mentioned | **7. Which scenario best describes the availability of specialist genetic services for your current clinical area?**<br>Specialist genetics services and palliative care services are embedded within the same hospital or group of hospitals.<br>Specialist genetics services and palliative care services are not embedded within the hospital or group but are accessible to each other.<br>Specialist genetics services and palliative care services are NOT accessible to each other. | Existing | NA |

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| | | | | Palliative care services have access to private genetics services only | | |
| | | | | Other (please explain) | | |
| | | | | Not sure | | |
| Not mentioned | *Discussions to obtain consent for genetic testing were managed by reducing or simplifying the information imparted. Participants wanted to convey the most important concepts, while not overburdening patients with irrelevant information. However, they described feeling conflicted about whether they were fulfilling their duty to obtain informed consent. I'm not ever going to make someone listen to me […] if they're not interested. But the thing that makes me* | Not mentioned | Not mentioned | **8. In your experience, how is consent for DNA banking or testing obtained and documented?** | Existing | NA |
| | | | | Verbal consent only (undocumented) | | |
| | | | | Verbal consent (documented in the patient's clinical records) | | |
| | | | | Written consent using locally available paperwork (but not a formal genetics consent form) | | |
| | | | | Written consent using a formal genetics consent form | | |
| | | | | In my experience consent has not been taken | | |
| | | | | Other | | |

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| | *uncomfortable is that even if I'm confident that they're on board […] that they're actually signing a piece of paper, which states that they understand things, which I really know that they don't ".* | | | | | |
| | | | | **9. In your experience, what do you consider to be the main challenges for palliative care/genetics health professionals in facilitating DNA banking/testing? *These response options have all been identified as challenges in prior studies. For the purpose of identifying priority areas, we are requesting you nominate your top three challenges** | See individual items | |

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| *While nurses and physicians routinely engaged in discussions about genetics with their patients, most demonstrated limited understanding of general genetic concepts, and/or concepts relevant to their specialty.* | *Noting palliative care health professionals' expert communication skills, participants thought basic genetics education, particularly related to the importance of the proband sample and process of DNA banking, could be sufficient to prepare them for genetics discussions* | *Participants reported having low genetics knowledge. For some, their level of confidence depended on whether they were assessing genetic risk for a malignant or non-malignant disease.* | Not mentioned | Identifying eligible patients | Existing | NA |
| *Nurses and physicians infrequently referred patients to clinical genetics services, primarily because ... of lack of time to initiate a genetics discussion.* | *Some speculated that late referrals to palliative care (for example, from oncology) might affect the palliative care health professionals' ability to identify the need for a genetics discussion* | *Nonetheless, they echoed calls for healthcare organisations to provide them with support to navigate the complicated, time-pressured scenarios that can arise in the palliative care context.* | Not mentioned | Urgency of the situation/referral | Existing | |

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| *A small number of articles reported nurses and physicians actively avoided or refused to discuss genetics with their patients, where they felt genetics was not relevant to clinical care and there may be potential negative consequences of genetic information.For example, some palliative care clinicians considered their clinical setting as inappropriate to initiate discussions about genetics...* | *Participants thought palliative care health professionals might avoid discussions about genetics because they believe another specialist has already addressed it, have concerns about harming patients or do not see genetics as relevant or part of their role.* | *Participants explored their responsibility to address genetics within the role and goals of palliative care. Using the World Health Organization definition of palliative care, some explained that addressing genetics was contrary to the 'relief of suffering', while others thought addressing genetic risk was part of an 'impeccable assessment'.* | Not mentioned | Conflicting priorities between providing palliative care and facilitating genetic testing | Existing | |
| *Some nurses and physicians had concerns about the privacy of genetic information or the process of informed consent* | *Discussions to obtain consent for genetic testing were managed by reducing or simplifying the information imparted. Participants wanted to convey the most* | *The cognition of the person with palliative needs played a major role in assessing whether consent could or should be obtained, and from whom.* | Not mentioned | Obtaining informed consent | Existing | |

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| | *important concepts, while not overburdening patients with irrelevant information. However, they described feeling conflicted about whether they were fulfilling their duty to obtain informed consent.* | | | | | |
| *Nurses and physicians are cognizant of the potential medical benefit that genetic information can provide for patients, but this was tempered by concerns about the risk of psychological harm, such as inducing feelings of guilt or hopelessness. The potential benefit to relatives was described, including clarifying family members' risks and* | *Approaching family members to discuss genetics could be challenging because the conversation was taking place at an emotionally difficult time.* | *Participants were concerned that raising genetics may result in feelings of guilt, blame and uncertainty for individuals and families.* | Not mentioned | Discomfort with initiating discussions about DNA storage/testing with patients or families | Existing | |

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| *providing screening or family planning options. Some nurses and physicians worried about the emotional impact of genetic information on the family.* | | | | | | |
| *Most nurses and physicians had low awareness of genetic tests relevant to their area of practice. They also had difficulty interpreting a genetic test result.* | *Participants conveyed their sense that genetics is not a priority for palliative care health professionals because of misunderstandings related to the value of genetic information* | *Participants reported having low genetics knowledge. For some, their level of confidence depended on whether they were assessing genetic risk for a malignant or non-malignant disease. However, most thought practical knowledge, including how to access genetic services within their organisation, was just as important as theoretical genetic concepts* | Not mentioned | Palliative care health professionals' lack of knowledge about DNA banking/testing or procedures | Existing | |

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| | Not mentioned | Not mentioned | Not mentioned | Genetics health professionals' lack of knowledge of the procedure for consent and DNA storage | Existing | |
| *However, if patients raised questions or concerns about genetics, nurses and physicians did engage in these discussions* | *Participants were sensitive to families' vulnerability in an end-of-life context, but most thought they were grateful for the opportunity to discuss the genetic implications of their relative's disease. There was also a sense from participants that a family-centred approach was in-line with the palliative patient's wishes.* | *A few reported initiating conversations about genetics with relatives and found that questions about genetic risk were often already on the relative's mind.* | Not mentioned | The views or expectations of the family | Existing | |
| *Nurses and physicians infrequently referred patients to clinical genetics services, primarily because of...lack of resources…* | *While a few participants described well-integrated services, most reported their services do not recognise the value of genetic information to palliative patients and families, with* | *Participants felt healthcare organisations could do more to support integration of genetics into palliative care through funding, education and raising awareness* | Not mentioned | Lack of availability of specialist genetics services | Existing | |

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| | *inadequate funding to develop solutions to existing barriers.* | | | | | |
| *Nurses and physicians reported the value of close working relationships or collaboration with clinical genetics professionals.* | *Participants valued a multidisciplinary approach to care, but portrayed a lack of collaboration, communication and professional relationships between palliative care and genetic health professionals. They described feeling powerless as individuals in overcoming these barriers.* | *Participants described the impact of professional relationships with genetics clinicians, or lack thereof, on their ability to access genetics services. Participants suggested co-locating palliative care and genetics departments to build better relationships, improve communication, access timely advice and encourage a multidisciplinary approach to care* | Not mentioned | Communication difficulties between genetics and palliative care services | Existing | |

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| *Nurses and physicians had mixed feelings about whether genetic information contributed to their clinical goals for the patient or aligned with their views about their professional role.* | *Participants thought palliative care health professionals might avoid discussions about genetics because they believe another specialist has already addressed it, have concerns about harming patients or do not see genetics as relevant or part of their role.* | *Participants explored their responsibility to address genetics within the role and goals of palliative care. Using the World Health Organization definition of palliative care, some explained that addressing genetics was contrary to the 'relief of suffering', while others thought addressing genetic risk was part of an 'impeccable assessment'* | Not mentioned | Conflicting views within the palliative care team about the utility of DNA banking/testing for palliative patients, and their families | Modified | Expanded upon 'conflicting views' to make the barrier more explicit |
| *Nurses and physicians infrequently referred patients to clinical genetics services, primarily because of...lack of resources…* | *Participants suggested several strategies to overcome barriers and support integration of genetics into palliative care (Table 2). These included workflow strategies, such as embedding a genetic counsellor within a palliative care team, tools to* | *Participants felt healthcare organisations could do more to support integration of genetics into palliative care through funding, education and raising awareness* | *Resources and funding are essential to the success of these strategies but health services are unlikely to commit these without a positive policy environment* | Lack of resources | Existing | NA |

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| | *assess eligibility for genetic testing, such as a red flag checklist for new hospice admissions, and integrating genetic guidance into relevant policy.* | | | | | |
| *Nurses and physicians are cognizant of the potential medical benefit that genetic information can provide for patients, but this was tempered by concerns about the risk of psychological harm, such as inducing feelings of guilt or hopelessness....Some nurses and physicians worried about the emotional impact of genetic information on the family.* | *Approaching family members to discuss genetics could be challenging because the conversation was taking place at an emotionally difficult time.* | *Deciding whether to raise genetics requires consideration of a number of psychological, medical and ethical factors, with most participants wary genetic information may cause psychological harm. Participants were concerned that raising genetics may result in feelings of guilt, blame and uncertainty for individuals and families.* | Not mentioned | Distress of the patient or family members | Existing | NA |

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| Not mentioned | *Participants found navigating discussions with palliative patients more difficult when important information was missing from their referral, such as prognosis, competency, family dynamics or circumstances* | *For families with complex relationships or conflicting opinions about the value of genetic information, knowing how, when or if to address genetics was particularly difficult for participants* | Not mentioned | Complex family dynamics or relationships | New | Included from findings in PC HP qual study |
| Not mentioned | Not mentioned | *Some participants reported that discussions about genetics could harm the therapeutic relationship between the clinician and their patient* | Not mentioned | Concern that a discussion about genetics could harm the therapeutic relationship | New | Included from findings in PC HP qual study |

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| *Nurses and physicians infrequently referred patients to clinical genetics services…* | *Participants conveyed their sense that genetics is not a priority for palliative care health professionals because of misunderstandings related to the value of genetic information. Some speculated that late referrals to palliative care (for example, from oncology) might affect the palliative care health professionals' ability to identify the need for a genetics discussion. Nonetheless, participants wished genetics were higher on their priority list so discussions could occur as early as possible in the patient's disease trajectory.* | *Reports varied among participants about how frequently their work interfaced with genetics. Some reported regular discussions about genetics, but most said this occurs infrequently.* | Not mentioned | Under-referral of palliative patients to genetics services | New | Included from findings in genetic HP qual study |
| *A small number of articles reported nurses and* | Not mentioned | *...Some reported regular discussions about genetics, but* | Not mentioned | In my experience DNA storage or genetic/genomic testing | Existing | NA |

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| *physicians actively avoided or refused to discuss genetics with their patients…* | | *most said this occurs infrequently.* | | has not ever been considered | | |
| *Nurses and physicians infrequently referred patients to clinical genetics services, primarily because of the prohibitive cost of accessing genetic testing…* | NA | NA | NA | Other (please explain) | Existing | (Cost unlikely to be identified as a main barrier in the Australasian setting due to the universal healthcare system) |
| | | | | **10. Please indicate your level of confidence about the following: (not at all confident to confident)** | See individual items | |
| Not mentioned | *Participants described the value they could add to conversations about genetics with palliative patients and families. However, some wanted to improve their own palliative care knowledge to ensure they manage these discussions appropriately. "From a genetic counselling point of view, I would* | Not mentioned | Not mentioned | Communicating with patients at the end of life | Existing | NA |
| Not mentioned | | Not mentioned | *Health professionals deliver family-centered care, recognising the important role families' play and identifying when* | Communicating with the families of patients who are at the end of life | Existing | NA |

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| | *be keen to […] have had more training in the space. I think those, particularly end-of-life conversations, they're quite confronting"* | | *family members may need to be recipients of care to support their emotional, social and physical needs* | | | |
| *A smaller number of articles reported that physicians did assess genetic risk, however, confidence in family history and individual risk assessment was low.* | *Noting palliative care health professionals' expert communication skills, participants thought basic genetics education, particularly related to the importance of the proband sample and process of DNA banking, could be sufficient to prepare them for genetics discussions* | *Participants reported having low genetics knowledge. For some, their level of confidence depended on whether they were assessing genetic risk for a malignant or non-malignant disease.* | Not mentioned | Identifying patients who may be eligible for DNA banking/testing | Existing | NA |
| *Oncologists and neurologists were most likely to order genetic testing. There were no* | *Most participants thought palliative patients wanted to engage with genetics to leave a legacy,* | *While many participants reported being involved in discussions about genetics, most* | Not mentioned | Discussing DNA banking with patients or their families | New | Created new item to specifically test for confidence towards discussing DNA banking (not testing) |

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| *reports of nurses or physicians from other specialties ordering testing. Most nurses and physicians had low awareness of genetic tests relevant to their area of practice.* | *make meaning from their illness and for reassurance their family would have access to important information. However, they acknowledged the value of genetic information depended on patients' and families' personal values, which was difficult to assess when they were providing genetic counselling near end-of-life.* | *described their preference to play a supportive role rather than be the primary drivers of these conversations* | Not mentioned | Discussing DNA testing with patients or their families | New | Created new item to specifically test for confidence towards discussing DNA testing (not banking) |
| Not mentioned | Not mentioned | *However, most thought practical knowledge, including how to access genetic services within their organisation, was just as important as theoretical genetic concepts* | Not mentioned | Contacting genetics service | Existing | NA |

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| Not mentioned | *Approaching family members to discuss genetics could be challenging because the conversation was taking place at an emotionally difficult time....However, there was a sense among participants that it was important to recognise and overcome their own feelings of discomfort about having genetic discussions with palliative patients, and their families* | Not mentioned | Not mentioned | Facilitating collection of a DNA sample from a patient receiving palliative care | Modified | Changed wording from 'taking DNA' to 'faciliating collection' because genetic HP pilot study participants explained they did not personally collect the sample, but were involved in organising the collection |
| *Oncologists and neurologists were most likely to order genetic testing. There were no reports of nurses or physicians from other specialties ordering testing. Most nurses and physicians had low awareness of genetic tests* | *Noting palliative care health professionals' expert communication skills, participants thought basic genetics education, particularly related to the importance of the proband sample and process of DNA banking, could be sufficient to prepare* | *Nonetheless, many recounted multiple conversations about the role of genes in disease development, caring for people with genetic conditions or helping to organise genetic investigations* | Not mentioned | Taking a DNA sample for banking or testing | Existing | NA |

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| *relevant to their area of practice. They also had difficulty interpreting a genetic test result.* | *them for genetics discussions* | | | | | |
| | *Genetics clinicians preferred to retain responsibility to oversee genetic testing, explaining their expertise about the implications of genetic testing benefited families.* | *While many participants reported being involved in discussions about genetics, most described their preference to play a supportive role rather than be the primary drivers of these conversations* | Not mentioned | Disclosing genetic/genomic test results to palliative care patient | Existing | NA |
| | | | Not mentioned | Disclosing genetic/genomic test results to bereaved families | Existing | NA |
| *However, nurses and physicians were uncomfortable about providing genetic health information to at-risk relatives of their patients.* | Not mentioned | *Participants explored their responsibility to address the genetic concerns of family members. Most explained they were unable to engage in a detailed genetics assessment because the relative is not their patient. Instead, participants directed relatives to their own doctors for advice.* | *Health professionals deliver family-centered care, recognising the important role families' play and identifying when family members may need to be recipients of care to support their emotional, social and physical needs* | Knowing how to respond if a family member asks me questions about their genetic disease risk | New | New item to assess aspects of potential role that PC HP feel confident with (i.e. responding to family member questions) |

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| *Some nurses and physicians had concerns about the privacy of genetic information or the process of informed consent* | *Some participants described the legal and ethical challenge of family-centred care when health systems preference individual autonomy over familial benefits. They described cases where theycould not discuss relevant genetic information with the family, because they did not have consent from the palliative patient.* | *Families can be complex: "The thing is with families […] some want to know, and some don't want to know and then where do you sit?"* | Not mentioned | Knowing my legal responsibility when sharing health information with family members when a patient is terminal or after they have died | New | Assess genetic HP finding among both groups that legal implications increase complexity of discussions |
| Not mentioned | *Genetics clinicians preferred the timing of follow up discussions about genetic testing to be guided by the family, highlighting the importance of palliative care clinicians making a clear plan with families to contact the genetics service when they are ready.* | *For families with complex relationships or conflicting opinions about the value of genetic information, knowing how, when or if to address genetics was particularly difficult for participants* | *Health professionals perform assessments to ensure care planning is individualised, responsive and appropriate to the family's needs* | Assessing an appropriate time to broach a discussion about genetics | New | Assess PC HPs views of the genetic HP finding that they trust PC HPs judgement about when to raise genetic discussions |

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| | | | | **11. What resources or tools have you found helpful when facilitating DNA banking/testing in the palliative care setting? (Please indicate all that apply)** | | |
| *Nurses and physicians infrequently referred patients to clinical genetics services, primarily because of...lack of resources…* | *Participants suggested several strategies to overcome barriers and support integration of genetics into palliative care (Table 2). These included workflow strategies, such as embedding a genetic counsellor within a palliative care team, tools to assess eligibility for genetic testing, such as a red flag checklist for new hospice admissions, and integrating genetic guidance into relevant policy.* | *Participants felt healthcare organisations could do more to support integration of genetics into palliative care through funding, education and raising awareness. A lack of research into the feasibility and acceptability of genetics in palliative care, particularly from the perspective of people with palliative care needs and their families, was identified as an important gap (quote 3.7). Participants suggested a simple, practical, accessible,* | *Care is organised under relevant government and organisational policy and enacted through health services fostering a supportive and responsive environment…* | Web-based risk assessment tool | Existing | NA |
| | | | | Smart phone App | | |
| | | | | Support from a palliative care colleague | | |
| | | | | Contact with specialist genetics services | | |
| | | | | Educational brochures | | |
| | | | | Telephone information hotline, | | |
| | | | | Face to face education (go to Q11a) | | |
| | | | | Online education | | |
| | | | | Clinical decision-making algorithm | | |
| | | | | Clinical practice guidelines (go to Q11b) | | |
| | | | | I have not found any resources or tools helpful | | |
| | | | | Other (please explain) | | |
| | | | | **11a. How frequently did you receive "face to face education" for DNA banking/testing in the** | Existing | NA |

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| | | *web-based genetics guideline for palliative care, as well as consumer-friendly information about genetics to provide to people with palliative care needs.* | | **palliative care setting? (free text respons** | | |
| | | | | **11b. Which clinical practice guidelines did you find useful? (free text respons** | Existing | NA |
| | | | | **12. What additional resources or tools would be helpful, if any? (free text response)** | Existing | NA |
| | | | | **13. Please indicate to what extent you agree or disagree with the following statements: (Strongly disagree to strongly agree)** | See individual items | |
| *For example, some palliative care clinicians considered their clinical setting as inappropriate to initiate discussions about genetics and were disappointed when this had not been addressed previously.* | *Participants were sensitive to families' vulnerability in an end-of-life context, but most thought they were grateful for the opportunity to discuss the genetic implications of their relative's disease. There was also a sense from participants that a family centred approach was in-line* | *Participants explored their responsibility to address genetics within the role and goals of palliative care. Using the World Health Organization definition of palliative care, some explained that addressing genetics was contrary to the 'relief of suffering', while others thought addressing genetic* | *The most frequent category overall (n=62/78, 79.5%), including by region (n=10/18, 55.56%) was "Delivering Family-Centred Care", although only 29.41% (n=5/17) of genomic policies included this category compared to 93.44% (n=57/61)* | Discussing DNA banking/testing with people receiving palliative care undermines the central ethos of palliative care in providing comfort and support at an emotionally vulnerable time | Existing | NA |

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| | *with the palliative patient's wishes* | *risk was part of an 'impeccable assessment'.* | *of palliative care policies. This category described the importance of attending to family members' psychological, social and spiritual needs.* | | | |
| *Nurses and physicians are cognizant of the potential medical benefit that genetic information can provide for patients, but this was tempered by concerns about the risk of psychological harm, such as inducing feelings of guilt or hopelessness..* | *Participants were sensitive to families' vulnerability in an end-of-life context, but most thought they were grateful for the opportunity to discuss the genetic implications of their relative's disease.* | *Others did not think of genetics as harmful, explaining that positive framing helps individuals understand the potential benefits of genetic information for the family...Some participants reported that people often have altruistic motivations to engage in a discussion about geneticsand are relieved they can offer genetic information to their relatives* | Not mentioned | Patients may experience positive emotional benefits from being able to give a sample for DNA banking/testing for the possible future benefit of their relatives | Existing | NA |

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| | *Most participants thought palliative patients wanted to engage with genetics to leave a legacy, make meaning from their illnessand for reassurance their family would have access to important information. However, they acknowledged the value of genetic information depended on patients' and families' personal values, which was difficult to assess when they were providing genetic counselling near end-of-life.* | *Participants were concerned that raising genetics may result in feelings of guilt, blame and uncertainty for individuals and families* | Not mentioned | Discussing DNA banking/testing may cause distress to the families by making them assume/fear their fate is pre-determined. | Existing | NA |
| *For example, some palliative care clinicians considered their clinical setting as inappropriate to initiate discussions about genetics and were disappointed when this had not* | *Participants thought palliative care health professionals might avoid discussions about genetics because they believe another specialist has already addressed it…* | *Some participants explained that they expected genetics to have already been discussed prior to the person requiring palliative care* | Not mentioned | DNA banking/testing will have been discussed by other health professionals before the patient is referred to palliative care | Existing | NA |

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| *been addressed previously.* | | | | | | |
| *For example, some palliative care clinicians considered their clinical setting as inappropriate to initiate discussions about genetics and were disappointed when this had not been addressed previously.* | *Participants were sensitive to families' vulnerability in an end-of-life context, but most thought they were grateful for the opportunity to discuss the genetic implications of their relative's disease. There was also a sense from participants that a family centred approach was in-line with the palliative patient's wishes...Nonetheless, participants wished genetics were higher on their priority list so discussions could occur as early as possible in the patient's disease trajectory.* | *Participants explored their responsibility to address genetics within the role and goals of palliative care. Using the World Health Organization definition of palliative care, some explained that addressing genetics was contrary to the 'relief of suffering', while others thought addressing genetic risk was part of an 'impeccable assessment'.* | Not mentioned | The priority in palliative care is to improve quality of life and relieve suffering, therefore it is not an appropriate time to discuss DNA banking/testing | Modified | Changed wording from "to facilitate a good death" to "improve quality of life and relieve suffering" to mirror the WHO palliative care definition |

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| *The potential benefit to relatives was described, including clarifying family members' risks and providing screening or family planning options.* | *Participants described the importance of the family unit when discussing genetics with palliative patients. They explained the main reason for testing was to elucidate relevant genetic information for relatives, rather than for the patient's clinical benefit.* | *Most believed relatives had a right to genetic information that would affect future health decisions. Although these participants reported that these conversations could be difficult, they found families generally appreciated the information.* | Not mentioned | DNA banking/testing of the patient may be important for the surviving relatives | Existing | NA |
| *For example, some palliative care clinicians considered their clinical setting as inappropriate to initiate discussions about genetics and were disappointed when this had not been addressed previously...Nurses and physicians had mixed feelings about whether genetic information contributed to their clinical goals for the patient or aligned* | *Participants thought palliative care health professionals might avoid discussions about genetics because they believe another specialist has already addressed it, have concerns about harming patients or do not see genetics as relevant or part of their role.* | *While many participants reported being involved in discussions about genetics, most described their preference to play a supportive role rather than be the primary drivers of these conversations* | Not mentioned | It is not the responsibility of health professionals in palliative care to discuss DNA banking/testing | Existing | NA |

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| *with their views about their professional role.* | | | | | | |
| *Genetic information was not always perceived as particularly useful in the clinical setting. Genetic information was described as irrelevant by nurses and physicians in certain clinical disciplines, such as ophthalmology, and by particular professionals, such as breast surgeons. Viewing genetics as irrelevant to clinical practice appeared to foster an active resistance to integrating genetics into practice.* | *Participants thought palliative care health professionals might avoid discussions about genetics because they believe another specialist has already addressed it, have concerns about harming patients or do not see genetics as relevant or part of their role.* | *Perceived relevancy of genetics to palliative care also varied among participants. Those with limited genetics exposure conveyed the irrelevancy of genetics to most patients and their practice. Participants who perceived genetics as relevant often described a formative clinical or personal experience that changed their perception that genetics related to their goals as palliative care clinicians* | Not mentioned | DNA banking/testing is not appropriate for people receiving palliative care because it will not help the patient | Existing | NA |

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| | | | | **14. Please indicate to what extent you agree or disagree with the following statements: (Strongly disagree to strongly agree)** | | |
| *However, nurses and physicians were uncomfortable about providing genetic health information to at-risk relatives of their patients.* | *Genetics clinicians recommended that, instead of referring terminal patients to the genetics service, palliative care clinicians should introduce the idea of obtaining and storing a DNA sample for future use.* | *Participants explored their responsibility to address the genetic concerns of family members. Most explained they were unable to engage in a detailed genetics assessment because the relative is not their patient. Instead, participants directed relatives to their own doctors for advice* | *Health professionals deliver family-centered care, recognising the important role families' play and identifying when family members may need to be recipients of care to support their emotional, social and physical needs* | Palliative care health professionals are well placed to have discussions about DNA banking/testing with the family members of a palliative patient | New | New item to assess phase 1 finding that PC HP are responsible for discuss genetic issues with family members |
| *Some nurses and physicians had concerns about the privacy of genetic information or the process of informed consent* | Not mentioned | Not mentioned | Not mentioned | Privacy and discrimination concerns (e.g. insurance and employment discrimination) make discussions about DNA banking/testing with palliative patients and their families difficult | New | Assess whether systematic review findings apply to PC context |
| Not mentioned | *Participants were sensitive to families' vulnerability in an end-of-life context,* | *Although these participants reported that these conversations could* | Not mentioned | Families generally appreciate being told genetic information that is relevant to their health | New | Assess phase 1 finding about families wanting genetic information |

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| | *but most thought they were grateful for the opportunity to discuss the genetic implications of their relative's disease.* | *be difficult, they found families generally appreciated the information* | | | | |
| Not mentioned | *However, they acknowledged the value of genetic information depended on patients' and families' personal values, which was difficult to assess when they were providing genetic counselling near end-of-life.* | *Universally, individualised discussions about genetics were considered integral to the person and family-centred approach of palliative care* | *Health professionals perform assessments to ensure care planning is individualised, responsive and appropriate to the family's needs* | Discussions about DNA banking/testing need to be individualised to the palliative patient, and their families | New | Confirm phase 1 finding that discussions must be individualised, rather than standardised |
| *Nurses and physicians expressed positive attitudes toward genetics, reported their beliefs about the future benefit of genetic information for patients and society as a whole, and regarded genetic health information as an inevitable major factor in* | Not mentioned | *Most believed that integrating genetics into the evolving palliative care role was feasible, describing aspects of genetics they felt were appropriate to integrate and the boundaries of their responsibility* | Not mentioned | As time goes on, genetic health information will inevitably become part of the palliative care health professionals' scope of practice | New | Assess phase 1 uncertainty about the future role of PC HPs in genetic discussions |

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| *clinical care in the future* | | | | | | |
| Not mentioned | *Consider reoffering the opportunity to palliative patients and families to discuss genetics: "In that case […] we'd seen her previously and […] she either declined testing or hadn't gotten around to having the blood taken and then realised the clock was ticking. And so desperately wanted to have the blood taken"* | *Participants described their ethical responsibilities when deciding whether to address genetics, including an obligation to respect a person's autonomy in their right to accept or decline a discussion about genetics* | Not mentioned | If a patient declines a discussion about DNA banking/testing, palliative care health professionals should revisit this discussion with palliative patients at a later date to see if they've changed their mind | New | Assess whether phase 1 finding extends to PC HPs intiating a repeat genetic discussion and genetic HPs' belief that discussions should be reoffered |
| Not mentioned | *Some participants described the legal and ethical challenge of family-centred care when health systems preference individual autonomy over familial* | *Most believed relatives had a right to genetic information that would affect future health decisions. Although these participants reported* | Not mentioned | The family of a palliative patient have a right to know if they are at risk of developing a genetic disease, even if it means having a difficult conversation about DNA | New | Assess varied views from phase 1 about the family's right to genetic information |

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| | *benefits. They described cases where they could not discuss relevant genetic information with the family, because they did not have consent from the palliative patient.* | *that these conversations could be difficult, they found families generally appreciated the information* | | banking/testing with a palliative patient | | |
| Not mentioned | | *Participants described their ethical responsibilities when deciding whether to address genetics, including an obligation to respect a person's autonomy in their right to accept or decline a discussion about genetics* | Not mentioned | The wishes of the palliative patient to engage in a discussion about DNA banking/testing are to be respected, no matter the wishes of the family | New | Assesse views from phase 1 about respecting patients' decisions about engaging in genetic discussions |
| | | | | **15. In your opinion, which of these might help palliative care health professionals to discuss genetics with palliative patients, and their families? *These response options have all been identified as facilitators in prior studies. For the purpose of identifying priority areas, we are** | New | No existing question to assess perceived facilitators |

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| | | | | **requesting you nominate your top three facilitators** | | |
| Not mentioned | *Participants valued a multidisciplinary approach to care, but portrayed a lack of collaboration, communication and professional relationships between palliative care and genetic health professionals. They described feeling powerless as individuals in overcoming these barriers. We've been in this building for three and a half years and I still have not worked out ways […] to get those buy-ins and having any kind of meaningful get together and 'here's what you are,* | *Participants suggested co-locating palliative care and genetics departments to build better relationships, improve communication, access timely advice and encourage a multidisciplinary approach to care* | Not mentioned | Physically co-locating palliative care and genetics health professionals within a hospital or organisation | | Suggested by PC HPs in phase 1 |

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| | *here's what we can offer' and so on"* | | | | | |
| Not mentioned | *Participants suggested several strategies to overcome barriers and support integration of genetics into palliative care (Table 2). These included workflow strategies, such as embedding a genetic counsellor within a palliative care team, tools to assess eligibility for genetic testing, such as a red flag checklist for new hospice admissions, and integrating genetic* | *Participants felt healthcare organisations could do more to support integration of genetics into palliative care through funding, education and raising awareness* | Not mentioned | Developing a specific referral template for palliative care patients to the genetics service that includes relevant family member details | | Possible strategy to overcome reported referral difficulties in phase 1 |
| Not mentioned | | Not mentioned | Not mentioned | Embedding a genetic counsellor in the palliative care team | | Suggested by genetic HPs in phase 1 |

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| | *guidance into relevant policy.* | | | | | |
| Not mentioned | *Encourage genetic and palliative care health professionals to attend the same multidisciplinary team (MDT) meetings: "I think MDT meetings are the easiest way to integrate us in. Because I don't think every department has the resources to have a genetic counsellor on staff, but the MDTs are an excellent opportunity to […] build the contacts to be able to have those discussions with each other"* | *Participants described the impact of professional relationships with genetics clinicians, or lack thereof, on their ability to access genetics services. Participants suggested co-locating palliative care and genetics departments to build better relationships, improve communication, access timely advice and encourage a multidisciplinary approach to care* | Not mentioned | Having both palliative care and genetics health professionals attending the same multidisciplinary team meetings | | Suggested by PC HPs in phase 1 |
| *Nurses and physicians reported the value of close working relationships or collaboration with* | *Participants valued a multidisciplinary approach to care, but portrayed a lack of collaboration, communication and professional* | | Not mentioned | Fostering a closer working relationship between palliative care and genetics health professionals | | Suggested by PC/genetic HPs in phase 1 |

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| *clinical genetics professionals* | *relationships between palliative care and genetic health professionals. They described feeling powerless as individuals in overcoming these barriers.* | | | | | |
| Not mentioned | *Participants felt responsible for providing education, but found it difficult to find time to deliver ongoing, concise and targeted education, due to the various cancer types and non-malignant conditions palliative care health professionals' encounter.* | Not mentioned | Not mentioned | Delivering genetics education to palliative care health professionals, including ways of sensitively communicating with patients and families about genetics | | Assess perceived value of genetic HPs delivering the education desired in phase 1 |
| *Nurses and physicians expressed their intention to engage in continuing professional education, demonstrating the need for increased genetic literacy. Most nurses and* | Not mentioned | *Participants had a variety of suggestions to improve their genetics knowledge, including a genetics module in palliative care trainee curricula, a compulsory oncology* | Not mentioned | Receiving genetics education from genetics health professionals, including ways of sensitively communicating with patients and families about genetics | | |

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| *physicians preferred clinically relevant education in the form of workshops, lectures, or online content.* | | *term during palliative medicine physician training, lectures delivered by genetics clinicians and inclusion of genetic research in palliative care conferences and journals.* | | | | |
| Not mentioned | *While a few participants described well-integrated services, most reported their services do not recognise the value of genetic information to palliative patients and families, with inadequate funding to develop solutions to existing barriers...Without clear leadership, participants noted that palliative patients (particularly those in private hospitals or from rural areas) were missing the* | *Participants felt healthcare organisations could do more to support integration of genetics into palliative care through funding, education and raising awareness. A lack of research into the feasibility and acceptability of genetics in palliative care, particularly from the perspective of people with palliative care needs and their families, was identified as an important gap.* | *Of the 78 policies, only two (2.56%) included recommendations about integrating genomics into palliative care...Of the 61 palliative care policies, only six (9.84%) mentioned genomics in the background information, and none of these incorporated genomics into their recommendations.* | Policy guidance detailing how and when to discuss DNA banking/testing with palliative patients and their families | | Reported as gap in phase 1 qual and identified as gap in phase 1 scoping review |

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| | *opportunity to address genetics...Generate leadership by reflecting the value of genetics in relevant policy and/or guidelines: "I think it would help if there was a national strategy on the integration of genomics into palliative care. […] I think it is quite important that you do have some sort of national leadership"* | | | | | |
| Not mentioned | *Some described patients and family members overcoming access barriers by taking the initiative to seek out genetic testing for themselves: "I find that often when successful in the private setting, it's because the family is motivated […] and very proactive in making sure the* | Not mentioned | Not mentioned | Empowering palliative patients, and their families, to seek out DNA banking/testing for themselves | | Suggested by genetic HPs in phase 1 |

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| | *blood is collected […] So that's often how it's circumnavigated."* | | | | | |
| *Nurses and physicians reported the value of close working relationships or collaboration with clinical genetics professionals* | *Participants valued a multidisciplinary approach to care, but portrayed a lack of collaboration, communication and professional relationships between palliative care and genetic health professionals.* | Not mentioned | Not mentioned | Speaking directly to the palliative care health professional about the palliative patient | | Assess whether direct collaboration is valued as suggested in phase 1 |
| | Not mentioned | *Participants described the impact of professional relationships with genetics clinicians, or lack thereof, on their ability to access genetics services* | Not mentioned | Speaking directly to the genetics health professional about the palliative patient | | |

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| Not mentioned | *Improve capability of electronic medical records to share information between services: "So how do they get access to medical records that sometimes might span over years? […] there's a suggestion: electronic records that actually talk to each other. […] You can take a considerable amount of time to wade through health records to see if genetics has already been covered."* | *Participants felt healthcare organisations could do more to support integration of genetics into palliative care through funding, education and raising awareness.* | Not mentioned | Improving the capability of electronic medical records to share relevant information between health professionals | | Possible strategy to overcome reported challenges in inter-discplinary communication identified in phase 1 |
| *Nurses and physicians reported the value of close working relationships or collaboration with clinical genetics professionals* | *Liaise directly with palliative care health professionals who are involved in the patient's care when a referral is received: "Once I have spoken to the nurses or the physicians who are actually involved with that patient's palliative care planning, they have* | Not mentioned | Not mentioned | Receiving assistance from palliative care health professionals to facilitate collection of a DNA sample from a palliative patient | | Assess whether direct collaboration is valued as suggested in phase 1 |

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| | *been extremely helpful […] in terms of organising and carrying out a more satisfactory consultation for this family."* | | | | | |
| | Not mentioned | *Participants described the impact of professional relationships with genetics clinicians, or lack thereof, on their ability to access genetics services* | Not mentioned | Collaborating with genetics health professionals to facilitate collection of a DNA sample from a palliative patient | | |
| Not mentioned | Not mentioned | *"I'm not convinced we need to take this up, or spend too much attention on it, because we've got to spend attention on getting right what it is we're meant to be doing, which is prevention, relief of suffering, to help people live well 'til they die. That's what my focus is."* | Not mentioned | We shouldn't be discussing DNA banking/testing with palliative patients, or their families | | Control item |

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| NA | NA | NA | NA | Other (please explain) | | Opportunity to suggest other facilitators |
| *For example, some palliative care clinicians considered their clinical setting as inappropriate to initiate discussions about genetics and were disappointed when this had not been addressed previously...Nurses and physicians had mixed feelings about whether genetic information contributed to their clinical goals for the patient or aligned with their views about their professional role...In contrast, other nurses and physicians were confident in their competence to* | *When palliative care clinicians find themselves in a situation where genetics is being addressed when the patient is close to end-of-life, genetics clinicians clearly articulated a preferred model of care. Genetics clinicians recommended that, instead of referring terminal patients to the genetics service, palliative care clinicians should introduce the idea of obtaining and storing a DNA sample for future use. Then, if the patient and family are agreeable,* | *Clinicians respect the autonomy of individuals to accept or decline a genetics discussion "So, you might introduce the idea that this could be helpful for the family. But basically if they don't want to do it, that's entirely their right to decline." Ethical obligations extended to obtaining consent to engage in discussions about genetics with relatives or collecting biological samples for genetic testing. The cognition of the person with palliative needs played a major role in assessing whether* | Not mentioned | **16. Previous studies with genetics health professionals have suggested that they believe patients close to end of life should be offered DNA banking by palliative care health professionals, rather than referring them to the genetics service for DNA testing. We would like to understand your views about how this would work in practice. Please indicate your comfort with palliative care health professionals performing the following actions: (Very uncomfortable to very comfortable)**<br>Introduce the idea of DNA banking with the patient and/or the family | New | Potential model described by genetic HPs in phase 1. Questions added to assess participants attitudes towards the various steps within this model |

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| *provide genetic information and, in their view, genetic information provision was appropriate within their clinical role* | *palliative care clinicians should obtain appropriate consent for DNA storage, facilitate collection of the DNA sample, make a plan with the family about following up with the genetics service and communicate this plan to the genetics service. Genetics clinicians explained their reasoning behind this model of care, finding that in their experience, introducing the genetics team at end of life was not in families' best interests. They added that storing a DNA sample, as opposed to organising genetic testing, reduced* | *consent could or should be obtained, and from whom. Most participants agreed that once cognition reduced, the legal medical proxy could provide consent for genetic testing or DNA storage...Participants explored the relevancy of genetics and how genetics aligns within the role and goals of palliative care. Most believed that integrating genetics into the evolving palliative care role was feasible, describing aspects of genetics they felt were appropriate to integrate and the boundaries of their responsibility:* | | Obtain consent for DNA banking from the patient, or appointed representative<br>Facilitate collection of DNA sample<br>Organise for the DNA sample to be banked<br>Instruct the family how to follow up with the genetics service<br>Communicate family follow up plan to the genetics service | | |

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| | *patient and family distress associated with considering genetic testing at the end of life. Genetics clinicians did not see the need for family members to consider the complex implications of genetic testing at an emotionally difficult time. Clinicians acknowledged that, while stored DNA does not last forever, it gave relatives time to think about whether they wanted to proceed with genetic testing and to consult the genetics service when they felt ready. In terms of obtaining consent for DNA storage, genetics clinicians explained that their expertise is not required because it is a simpler discussion compared to obtaining consent* | *"“[Palliative care] is one of the younger specialties around and the way I see it, we’re still finding our feet as to who we are and what we do […] so I think it’s still an evolving field and it certainly is still on the cards to add this addressing genetics] on among the multiple things that are being done in palliative care.”...Some participants explained that they expected genetics to have already been discussed prior to the person requiring palliative care...Participants explained that their ability to engage in difficult conversations and to provide holistic, family-centred care were transferrable skills they could use* | | **16a. Please describe any further thoughts you have about palliative care health professionals performing these actions (free text response)** | New | |

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| | *for genetic testing. They also saw benefit for the patient, in eliminating the need for a lengthy consent discussion about the implications of genetic testing that ultimately had no consequences for their own health. Genetics clinicians preferred the timing of follow up discussions about genetic testing to be guided by the family, highlighting the importance of palliative care clinicians making a clear plan with families to contact the genetics service when they are ready. For palliative care clinicians offering follow up appointments with bereaved relatives, this was noted as an opportunity to* | *to have conversations about genetics. However, most felt they could not apply these skills without an increase in their genetic knowledge. While many participants reported being involved in discussions about genetics, most described their preference to play a supportive role rather than be the primary drivers of these conversations.* | | | | |

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| | *remind family members that a DNA sample had been stored for future use. Genetics clinicians preferred to retain responsibility to conduct genetic testing, explaining their expertise about the implications of genetic testing benefited families.* | | | | | |

SUPPLEMENTARY FILE B2: END-POINT DATA INTEGRATION JOINT DISPLAY TABLE

**Joint display table to answer research question 3**

What is required to support genomic and palliative care health professionals integrate genomics into the care of people with palliative care needs and their families?

| Qualitative data: interviews and focus groups with genetic health professionals (GHP; QUAL) | Qualitative data: interviews with palliative care health professionals (PC HP; QUAL) | Qualitative and quantitative data: scoping review of policy recommendations (qual & quan) | Quantitative data: questionnaire survey of genetic and palliative care health professionals (QUAN) | Data Convergence | Meta-Inference |
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| **HIGH-LEVEL INFERENCE**<br>• ***Balancing individual autonomy against the genomic needs of the family is challenging in the end-of-life situation***<br>**DRAWN FROM THEME:**<br>Focusing on the benefit to the family<br>The discomfort of addressing genomics near end-of-life.<br>**DATA SUMMARY:**<br>Usually, navigating the potential benefits and harms of a genomic discussion is managed by taking the time to explore patients' values. However, | **HIGH-LEVEL INFERENCE**<br>• ***PC HPs struggle to weigh up the harms and benefits of genomics in the end-of-life situation***<br>**DRAWN FROM THEME:**<br>Harms and benefits of raising genomics: a delicate balancing act.<br>**DATA SUMMARY:**<br>Broaching genomics with palliative patients challenges PC HPs usual practice because raising genomics could potentially contradict their clinical goal by causing harm to the individual and/or family. In deciding whether to broach | Not reported | **HIGH-LEVEL INFERENCE**<br>• ***The palliative–genomic situation challenges health professionals' otherwise positive attitudes towards genomics***<br>**DRAWN FROM CATEGORY:**<br>Attitudes towards genomics in palliative care<br>Confidence integrating genomics into palliative care<br>**DATA SUMMARY:**<br>Despite positive attitudes towards genomics, both PC and GHPs are | Confirm and enhance understanding | *Support to adapt practice to the challenges of addressing genomics with people near end of life and their families*<br>Health professionals require specialised clinical and ethics support to navigate the challenges of addressing genomics at end of life. There is a shared discomfort among genetic and palliative care health professionals arising from the required practice modifications to usual care. Health professionals' usual practice is underpinned by bioethical principles, which are challenged by several factors in the end-of-life setting. The difficulty navigating clinical practice and ethical principles may explain the cognitive dissonance between reported |

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| for unwell patients or those nearing end-of-life, there is inadequate time to explore these values. GHPs must reduce or simplify the amount of information they impart. However, GHPs can be uncomfortable with these changes to practice because they are uncertain if they have done enough to obtain informed consent. Health systems that prioritise the autonomy of the individual make end-of-life discussions more difficult because of the ethical and legal challenge of engaging relatives in clinical decision making where patients' cognition is reduced. Despite these challenges, GHPs feel responsible for overcoming their discomfort to focus on the needs of the patient and family. | a discussion about genomics, PC HPs must weigh up the potential benefits of genomics with the potential medical and psychological harms. PC HPs make these decisions in the context of their ethical and legal responsibilities to patients and their families; their primary goal is to improve their patient's quality of life and minimise suffering. | | uncertain about how to apply these benefits when the wishes of the patient and family are in conflict. GHPs think PC HPs are well placed to engage in these complex discussions, but PC HPs are not confident engaging in discussions about DNA banking or testing. | | positive attitudes towards genomics yet feeling uncomfortable about engaging in discussions about genomics with patients nearing end of life. |
| **HIGH LEVEL INFERENCE**<br>• ***Family-centred care means providing unaffected family members with the opportunity to receive clinically relevant genomic information*** | **HIGH LEVEL INFERENCE**<br>• ***Family-centred care revolves around the needs of the person receiving palliative care.***<br>**DRAWN FROM THEMES:** | **HIGH LEVEL INFERENCE**<br>• ***Delivering family-centred care is a key professional goal for both GHPs & PC HPs.***<br>**DRAWN FROM DATA:** | **HIGH LEVEL INFERENCE**<br>• ***Improvement of a shared understanding and delivery of family centred care in the palliative-genomic context is required*** | Confirm and enhanced understanding | *An interdisciplinary understanding of family-centred care in the palliative-genomic context*<br>While there are differing interpretations of 'family-centred care', this is a shared principle between genomics and palliative care. Improving upon health |

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| **DRAWN FROM THEMES:**<br><br>Focusing on the benefit to the family.<br><br>Burden of proof: instilling the value of genomics in palliative care<br><br>**DATA SUMMARY:**<br><br>GHPs adopt a family centred approach to meet the clinical and psychological needs of the family. This is operationalised by preferencing DNA storage to DNA testing. Storing DNA gives GHPs an opportunity to explore family member's needs before proceeding with testing. Building relationships with families supports their goal in providing relevant clinical information to unaffected family members. | Aligning genomics within the role and goals of palliative care<br><br>**DATA SUMMARY:**<br><br>For PC HPs, family centred care revolves around the needs of the palliative patient. While PC HPs are supportive of patients and families having the opportunity to discuss genomics, the needs of the patient are prioritised over the needs of family members. Addressing the genomic needs of family members, without the involvement of the patient, is not within the palliative care scope of practice. | Almost all policies (*n* = 72/78, 92%) had recommendations about care of the patient's family. Most of these genomic and palliative care policies recommended "Delivering Family-Centred Care" (*n* = 62/78, 80%). | **DRAWN FROM CATEGORY:**<br><br>Confidence integrating genomics into palliative care<br><br>Previous experience in genomics and palliative care<br><br>**DATA SUMMARY:**<br><br>PC HPs are frequently obtaining a family health history from their patient, but are less frequently checking with their patient, or their relatives, if they have had an opportunity to discuss genomics. Family members are thought to be interested in a discussion about genomics by raising the topic with their dying relative's healthcare professional. However, PC HPs are not confident responding to family member's questions about genomics, discussing DNA banking or testing with family members or disclosing results to bereaved family members. | | professionals and organisations understanding of 'family-centred care' in the palliative-genomics context may help identify further intervention targets. Underpinning future interventions, such as a DNA storage approach, with a family centred care philosophy may help to communicate the relevance of the intervention and improve clinical uptake. |

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| | | | Meanwhile, most GHPs reported feeling confident with providing genetic counselling palliative care patients and families. | | |
| **HIGH LEVEL INFERENCE:**<br>• ***Healthcare services should facilitate opportunities for collaboration between genomic and palliative care health professionals***<br>**DRAWN FROM THEMES:**<br>Burden of proof: instilling the value of genomics in palliative care<br>"Individuals can only do so much": finding solutions in the absence of service leadership<br>**DATA SUMMARY:**<br>GHPs believe PC HPs do not prioritise genomic information among their other responsibilities because they don't understand the value, don't feel it is their responsibility or are worried about the potential harms. GHPs feel | **HIGH LEVEL INFERENCE:**<br>• ***Healthcare service support should focus on simple, relevant concepts, and interdisciplinary collaboration***<br>**DRAWN FROM THEMES:**<br>Navigating genomic responsibility within the scope of palliative care<br>(Ir)Relevancy of genomics to palliative care<br>Overcoming practice barriers: a multipronged approach<br>**DATA SUMMARY:**<br>Previous experience with genomics influences perceptions about relevancy. Without frequent exposure or a formative experience, PC HPs view genomics as irrelevant and do not perceive genomics as their responsibility. Viewing | **HIGH LEVEL INFERENCE:**<br>• ***Healthcare services have a responsibility to support multidisciplinary care delivery***<br>**DRAWN FROM DATA:**<br>67.95% (*n*=53/78) policies included recommendations related to Governance and policy, of which 36% *(n*=19/53) specified that care should be delivered by multidisciplinary teams. | **HIGH LEVEL INFERENCE:**<br>• ***Integration of genomics into palliative care would improve with educational collaboration between GHPs and PC HPs***<br>**DRAWN FROM CATEGORIES:**<br>Confidence integrating genomics into palliative care<br>Previous training in genomics and palliative care<br>Perceived barriers & facilitators<br>**DATA SUMMARY:**<br>Despite selecting education as a top facilitator, few PC HPs have engaged in genomics education with PC HPs' lack of genomic | Confirmed and enhanced understanding | *A culture of professional collaboration within healthcare services*<br>Health services can support the integration of genomics into palliative care by facilitating professional collaboration between genomic and palliative care health professionals. Building professional relationships will provide an opportunity to establish meaningful connections that benefit palliative patients and their families. As relationships improve, opportunities to engage in more formal education may arise. A common understanding of shared and differing clinical goals would identify areas where the two professions could complement each other's work. Collaboration at the local level could identify suitable intervention targets to ensure the needs of both palliative care patients and their family members are being met. |

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| responsible for educating PC HPs but are limited by practical barriers, such as lack of time and resources<br><br>GHPs expressed the value of collaborative professional relationship between GHPs & PC HPs in delivering client-centred care and want further education about how to manage palliative care discussions. | genomics as irrelevant reduces the likelihood of engaging in genomics education. As a result, PC HPs feel their genomic knowledge is low. Practical information, such as how to contact the local genomic service, delivered in existing palliative care forums, as well as building professional relationships with GHPs is desired. Connections to clinical genomics services may improve confidence in contacting their local genomic service for support and basic genomic knowledge. | | knowledge as the main perceived barrier. Across all genomic activities, PC HPs reported low confidence. More GHPs have engaged with palliative care education and feel confident providing genetic counselling to patients and families. Both groups are interested in learning about the others' specialty. Fostering closer working relationships between genomic and palliative care health professionals, and the support of a genomics colleague were highlighted as important facilitators | | |
| **HIGH LEVEL INFERENCE:**<br><br>• ***GHPs are disenfranchised by the lack of recognition of the value of genomics in palliative care.***<br><br>**DRAWN FROM THEMES:**<br><br>"*It's always on the back-burner*": challenges to getting genomics on the palliative care agenda | **HIGH LEVEL INFERENCE:**<br><br>• ***Inadequate organisational support places the responsibility of integration upon palliative care health professionals***<br><br>**DRAWN FROM THEMES:** | **HIGH LEVEL INFERENCE:**<br><br>• ***Further research is needed to prompt the development of high-level policy recommendations.***<br><br>**DRAWN FROM DATA:**<br><br>*Integration of Genomics into* | **HIGH LEVEL INFERENCE:**<br><br>• ***Local implementation research could address gaps in care and build evidence for policy recommendations***<br><br>**DRAWN FROM CATEGORY:** | Confirmed and enhanced understanding | *Co-designed implementation research to build evidence for policy stakeholders*<br><br>A lack of organisational support for genomics in palliative care is underpinned by an inadequate evidence base that demonstrates the value to stakeholders. Transdisciplinary research that demonstrates the value of genomics in palliative care is needed to lobby policy |

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| "Individuals can only do so much": finding solutions in the absence of service leadership<br><br>**DATA SUMMARY:**<br><br>Clinical genomics is perceived as an unimportant area of health care across all levels of the health system (micro, meso and macro). GHPs are frustrated and fatigued by needing to justify the value of genomic information in the palliative care context and wish there was better top-down leadership, so patients do not have to advocate for themselves.<br><br>Service leadership is crucial to ensure all patients and families are receiving equitable care. While GHPs have several ideas for strategies to overcome barriers, inadequate recognition and funding from the health service level are the main barriers to integrated care. | Overcoming practice barriers: a multipronged approach.<br><br>**DATA SUMMARY:**<br><br>PC HPs believe that health care organisations do not meaningfully support the integration of genomics into palliative care. PC HPs suggest that supporting integration requires funding, education and raising awareness. In the absence of organisational support, PC HPs suggest grass-roots initiative could support the development of professional relationships with their GHP colleagues. Improved connections to clinical genomics services may improve integration at a local level. | *Palliative Care; n =* 2/78 policies recommended integrating genomics into palliative care & *n* = 6/61 PC policies mentioned genomics in background information but were without relevant recommendations.<br><br>**DATA SUMMARY:**<br><br>Although some PC policies acknowledge genomic conditions as a feature in their patients, policy guidance is lacking. Without a positive policy environment, health services won't commit funding to support HPs. Policy makers may need further evidence about the clinical, psychological and economic benefits of instilling genomics into palliative care. | Perceived barriers & facilitators<br><br>**DATA SUMMARY:**<br><br>GHPs highlighted gaps in care: they believe PC patients are under-referred to genomic services and were less likely to think that DNA banking/testing will have been discussed prior to palliative care. Point-of-care documents (such as a referral template) may facilitate genomics into PC. Support from genomics colleagues most useful tool/resource | | stakeholders, so all patients and families receive equitable care. |

## SUPPLEMENTARY FILE B3: DATA MANAGEMENT AND STORAGE

**Data Management and Storage**

UTS has several policies relevant to the management and storage of data that I familiarised myself with for the purpose of The GIFT Project. This includes the Data Governance Policy, Privacy Policy, Records Management Policy, Research Policy and Information Security Policy.

**Protection of Personal Information**

The protection of personal and/or identifiable information of my study participants was my top priority. During the qualitative study in Phase 1, participants consented to provide their name, age, phone number and email address. It was essential for me to collect these details so I could correspond with the participant to organise their interview or focus group. To protect my participants' personal information, I ensured their details were held in strict confidence and not shared with any third party. Their personal information was stored in a password protected folder on my UTS student OneDrive account. Files containing personal information were accessed only by me as required on a password protected, UTS-owned computers. If details were shared between the research team (for example, when creating online calendar invitations for focus group participants), only the research teams' official UTS emails, or other affiliated university emails, were used. Personal data was removed as soon as practical from working documents (e.g., interview transcripts).

As detailed in Chapter 5, part of the scoping review search strategy was to contact 'key informants' and ask them to identify relevant local or national policy documents related to genetics, genomics or palliative care. Key informants' emails were accessed through publicly accessible documents and sites and therefore not subject to the same responsibility of protection. However, I was careful not to arbitrarily amplify the key informants' emails. A Microsoft Excel spreadsheet detailing the key informant's name, email, and the date and content of email contact to and from the informant was maintained as a study record. I stored this on my UTS student OneDrive account. Names and emails of key informants were not published within the resulting manuscript.

Phase 2 did not require the collection of any personal or identifiable data, as all survey data was anonymised.

**Management of Study Records**

All files were stored on my UTS student OneDrive account. A Research Data Management Plan was created and stored in 'Stash' (UTS data management planning software).

The following data types were produced during this thesis:

- Excel spreadsheets (.xlsx)
- Video and audio recordings (.mp4)
- Focus group and interview transcripts, verbal consent acknowledgement forms (.docx)
- NVivo V12 data analysis software files (.nvp)
- SPSS files (.sav)
- PowerPoint presentations (.pptx)
- EndNote Libraries (.enl)
- PDF versions of study materials, manuscripts, journal submissions etc (.pdf)
- Reflexive journal (hard copy diary, no identifiable information recorded)

Upon completion of the studies in this thesis, data was archived to the UTS eResearch Store for a period of five years. In accordance with UTS policy, all files containing personal, identifiable, or sensitive information will be destroyed with the assistance of a UTS data librarian after five years.

# Appendix C: supplementary files related to qualitative study

## CONTENTS

SUPPLEMENTARY FILE C1: QUALITATIVE STUDY PROTOCOL

# Genetics and genomics in palliative care: exploring the views and experiences of palliative care and genetic clinicians

Sponsor:

Translational Cancer Research Network (TCRN)

PhD Scholarship Top-Up Award

## Project team roles and responsibilities

Coordinating Principal Investigator
Name: Professor Jane Phillips
Position: Director of IMPACCT (Improving Palliative, Aged and Chronic Care Through Clinical Research and Translation)
Institution: University of Technology Sydney
Responsibilities: Oversee planning, conducting and reporting of the study

Principal Investigator
Name: Stephanie White
Position: PhD candidate, Associate Genetic Counsellor, Registered Nurse
Institution: University of Technology Sydney, Royal North Shore Hospital
Responsibilities: Primary researcher planning, conducting and reporting of the study

Associate Investigator
Name: Dr Chris Jacobs
Position: Senior Lecturer in Genetic Counselling, Registered Genetic Counsellor, Registered Nurse
Institution: University of Technology Sydney
Responsibilities: Contribute to planning, conducting and reporting of study

## SUMMARY

| | |
|---|---|
| **Study Title** | Genetics and Genomics in Palliative Care: Exploring the Views and Experiences of Palliative Care and Genetics Clinicians' |
| **Objectives** | To explore palliative care and genetics clinicians' 1) views and experiences of the barriers and facilitators of integrating genetics and genomics into routine care of people with palliative needs (and their families), and 2) suggested strategies to improve genetic and genomic integration into palliative care |
| **Study design** | Interpretive descriptive qualitative study involving focus groups and semi-structured interviews with purposively sampled palliative care and genetics clinicians. |
| **Planned sample size** | N = 40 - 60 |
| **Inclusion criteria** | - Registered nurses and physicians who are currently and directly involved in providing palliative care to patients in Australasia (Australia and New Zealand)<br><br>- Qualified genetic counsellors and physicians who are currently and directly involved in providing genetic counselling, clinical genetics and familial cancer service in Australasia |
| **Study procedures** | Several professional organisations will send potentially eligible participants an email invitation to participate. After reading the Participant Information Sheet and providing informed consent, participants will be invited to a virtual Zoom focus group or semi-structured interview. Utilising a semi-structured guide, we will explore participant views and experiences of genetics and genomics in palliative care. Focus groups and interviews will last up to one hour. |
| **Analysis considerations** | When saturation is reached, data collection will cease. Transcribed focus groups and interviews will be managed using NVivo V12 software. Researchers will employ thematic analysis, as described by Braun and Clarke (2006). |
| **Study duration** | Recruitment and data collection is predicted to take six - nine months (September 2020 – May 2021). Analysis and reporting is expected to take a further six months and be complete by August 2021. |

## BACKGROUND AND RATIONALE

Rapid advances in our understanding of genetic contribution to disease, genomic testing technologies and development of precision medicines has had a major impact on routine medical care in the last two decades (Boyle & De Boeck, 2013; Lander et al., 2001; Maron et al., 2007). Most medical specialties, including palliative care, have not historically integrated genetics and genomics (herein referred to solely as 'genomic/s') but are now under pressure to learn and apply genomic information into their practice (Dearing & Taverner, 2018). Front-line nurses and physicians with no or limited formal genomics education are tasked with identifying individuals with a genetic condition, performing pre-test counselling, organizing genetic testing, interpreting the result and communicating the impact of the result for the patient and relatives (Aday & Macrae, 2017; Dodson & Lewallen, 2011). Nurses and physicians feel under-equipped to integrate genomics into practice but do see the benefit of genomic information and are willing to learn more for the benefit of their patients (White, Jacobs, & Phillips, 2020).

Although identifying an underlying genetic condition in a person with palliative care needs changes little for their own health or management, it can be significant for their relatives. Despite this major benefit, genomics is largely missing from the palliative care agenda (Morrow, Jacobs, Best, Greening, & Tucker, 2018). This may be, in part, due to the additional benefits of integrating genomics into other clinical areas, such as oncology, in which there has been a surge of resources (Flynn, Cusack, & Wallen, 2019; Hallowell et al., 2019; Long et al., 2018; Rahman et al., 2019). Comparatively, it would seem that palliative care has flown under the radar. While it may be too late for people with palliative care needs to directly benefit from genetic testing, 'predictive testing' (targeted genetic testing for an identified familial pathogenic variant) in relatives is the most accurate way to assess whether that relative is living with the same genetic risk (Konstantinopoulos & Matulonis, 2018; Kuchenbaecker et al., 2017; Le et al., 2015). Relatives at increased risk can be offered risk-reducing or surveillance options, which can drastically improve their long-term health (Kuchenbaecker et al., 2017).

Palliative care nurses and physicians ('clinicians') have expressed concerns about integrating genomics into practice, citing low knowledge and confidence in talking about genomics (Dearing & Taverner, 2018; Gonthier et al., 2018; Ingelby, 2015; A. Lillie, K, 2009; A. K. Lillie, Clifford, & Metcalfe, 2011; Metcalfe, Pumphrey, & Clifford, 2010; J. M. Quillin, Bodurtha, Siminoff, & Smith, 2011). Some clinicians are concerned genomic discussions may distress individuals and families, while detracting attention away from 'good' palliative care (Dearing & Taverner, 2018; Ingelby, 2015; A. K. Lillie et al., 2011; Metcalfe et al., 2010). Despite this, people with palliative care needs report existing suspicions of a genetic predisposition, concern for family members, psychological benefit following a genetics discussion and have taken it upon themselves to discuss genetics with their palliative clinician (Cleophat et al., 2019; Daniels, Urbauer, Stanley, Johnson, & Lu, 2009; Dearing & Taverner, 2018; John Martin Quillin, Bodurtha, Siminoff, & Smith, 2010). Meanwhile, genetics clinicians (medical geneticists and genetic counsellors) appear to be supportive of genomic integration into routine care (Daniels, Burzawa, Brandt, Schmeler, & Lu, 2011; Dwarte, Barlow-Stewart, O'Shea, Dinger, & Terrill, 2018).

Australasian studies examining palliative clinicians' views about genomics are missing from the literature. Without local data, state and federal governments' efforts to integrate genomics into routine medical care are likely to fail (Craig et al., 2008; Department of Health, 2017; NSW

Ministry of Health, 2017). Despite the benefits of genomic information, translation of recommendations to clinical practice is slow, highlighting the complex and interconnected barriers and facilitators within healthcare pathways (Cane, O'Connor, & Michie, 2012). Understanding the action required (from the perspectives of palliative and genetics clinicians) is a critical step towards making appropriate recommendations to stakeholders, so individuals and families do not miss out on important genetic information.

## OBJECTIVES AND RESEARCH QUESTIONS

### Study objectives

To explore palliative care and genetics clinicians':

1. Views and experiences of the barriers and facilitators of integrating genetics and genomics into the routine care of people with palliative needs, and/or their families
2. Suggested strategies to improve genetic and genomic integration into palliative care

### Research questions

Primary question

1. What are palliative care and genetics clinicians' views and experiences of integrating genetics and genomics when providing care for people with palliative needs?

Secondary questions

1. What are the views and experiences of palliative care and genetics clinicians' providing genetic counselling and/or genetic testing to people receiving palliative care, and their families?
2. What are the barriers and facilitators to integrating genetics and genomics into the care of people with palliative needs and their families?
3. What action needs to occur to support the integration of genetics and genomics into the care of people with palliative needs, and their families?

## STUDY DESIGN

This is an exploratory, cross-sectional, interpretive descriptive qualitative study using online focus groups and semi-structured interviews with palliative care and genetics clinicians'. Qualitative methods have been selected to capture rich data about participants' views and experiences in this under-researched area (Creswell, 2014, p. 4).

## EXPECTED STUDY DURATION

Advertising and recruitment for this study is expected to commence in September 2020. Recruitment and data collection should take approximately six to nine months, between September 2020 and May 2021. At the point of data saturation (ie. when no new information arises from continued focus groups or interviews), data collection will cease. Data analysis and reporting the findings (in the form of a manuscript for publication, thesis chapter and conference presentations) is expected to take six months, between February 2021 and August 2021.

## DATA SOURCE AND POPULATION

Purposive sampling will be used to maximise variation and ensure the participants are able to provide relevant information and answer the research questions (Morse, 2003, pp. 884-885). By considering the study objective, sample specificity, the use of theory, likely quality of

interview dialogue and analysis strategy, a sample size of approximately 40 – 60 participants is likely to be sufficient (Malterud, Siersma, & Guassora, 2016). To illustrate these considerations, our broad study objective may mean a greater volume of data are needed reach saturation. However, the participants (palliative care and genetics clinicians') are specific to the research objective and questions, reducing the number of participants needed. Furthermore, using the WHO Integrated Care for Chronic Conditions framework may reduce the number of participants needed by incorporating the most relevant elements into the interview dialogue (World Health Organization, 2002). In regards to the interview dialogue quality, I am an experienced communicator but a novice researcher. Although I will engage in focus group moderation and interview training prior to conducting data collection, the number of participants may need to be higher to compensate for my inexperience. Lastly, this study will be a cross-case analysis, increasing the number of participants required (Malterud et al., 2016). Table 1 describes the number of participants to be recruited from each professional organisation.

*Table 1. Sample source and size summary. The different types of participants are listed against the professional organisations from which they will be sampled and the estimated sample size of each group*

| Participant type | Professional organisation | Sample size |
|---|---|---|
| Palliative care nurse | Palliative Care Nurses Australia<br>Palliative Care Nurses New Zealand | 8 - 16 |
| Palliative care doctor | Australian and New Zealand Society of Palliative Medicine | 8 - 16 |
| Genetic counsellor | Human Genetics Society of Australasia<br>Australasian Society of Genetic Counsellors | 8 - 16 |
| Clinical geneticist (including cancer geneticist and trainees) | Human Genetics Society of Australasia<br>Australasian Association of Clinical Geneticists | 8 - 16 |

Demographic data will be collected to check variation in the sample and provide context to quotes and themes. Demographics include gender (male, female or non-binary), age (18 – 30, 31 – 45, 46 – 60, >60) specialty (palliative care, clinical genetics or genetic counselling), discipline (nurse, physician or genetic counsellor), years of practice (0 - 5 years, 6 – 10 years, 11 – 15 years, >15 years), area of practice (rural, regional or urban), work sector (private, public or mix of public and private).

**RECRUITMENT**

Nurses, genetic counsellors and physicians will be recruited via online invitations (Appendix 1 & 2). Six organisations will be asked to circulate online invitations to their members: Palliative Care Nurses Australia (PCNA); Palliative Care Nurses New Zealand (PCNNZ); Australian and New Zealand Society of Palliative Medicine (ANZSPM); Human Genetics Society of Australasia (HGSA); Australasian Society of Genetic Counsellors (ASGC) and; Australasian Association of Clinical Geneticists (AACG). If agreeable, organisations will circulate an email invitation to their members up to three times. Members of the research team will advertise recruitment through their personal and affiliated social media platforms, such as Twitter.

Interested participants will be asked to contact me for further information and to express interest in participating. At this point, I will send the participant the Participant Information Statement (PIS; Appendix 3), link to the RedCAP demographic survey and answer any questions the participant may have (National Institutes of Health, 2020). I will arrange a date

and time for focus group or interview with the participant, and email these details (including a zoom link) to the participant.

After discussing participation by phone or email, I will create a Zoom 'meeting' and email the invitation link to the participant. To ensure security and confidentiality, I will configure the Zoom meeting so that, upon entry, the participant is directed initially to a virtual 'waiting room' where they will wait until I admit them to the meeting. If the participant wishes to withdraw from the study before, during or after participation, their personal information will be removed from all study records. If they have already participated in the focus group when they indicate a preference to withdraw, none of their direct quotes will be reported (National Health and Medical Research Council, 2007).

**ELIGIBILITY CRITERIA**

<table>
<tr><th>Population group</th><th>Inclusion criteria</th><th colspan="2">Exclusion criteria</th></tr>
<tr><td>Palliative care nurses</td><td>Registered nurses who provide direct nursing or manage nurses who provide direct nursing care to palliative care patients and families in Australia or New Zealand</td><td>Student nurses<br><br>Enrolled nurses</td><td rowspan="4">Unable to speak adequate English to meaningfully engage in an interview or focus group</td></tr>
<tr><td>Palliative medicine doctors</td><td>Medically trained interns, residents, registrars or consultants whose majority role (≥50% of clinical workload) is to provide palliative care/medicine to patients in Australia or New Zealand</td><td rowspan="2">Student doctors</td></tr>
<tr><td>Genetics doctors</td><td>Medically trained interns, residents, registrars or consultants or other appropriately trained medical doctors whose majority role (≥50% of clinical workload) is to provide a clinical genetics or familial cancer services in Australia or New Zealand</td></tr>
<tr><td>Genetic counsellors (GC)</td><td>GCs who have attained Part 1 certification with the Human Genetics Society of Australasia and are involved in providing genetic counselling services in Australia or New Zealand</td><td>Student GCs<br><br>GCs who have only worked in a non-clinical role</td></tr>
</table>

**CONSENT**

I will utilise a verbal consent script at the beginning of each focus group or interview. If the participant wishes to have a copy of this script template, this will be sent to them by email. A script template will be saved with each participants details and agreement.

The potential participants will be reminded at up to four separate time-points that their participation is voluntary and they are free to withdraw from participating in the focus group or interview at any time. These time points are when:

1 Reading the PIS and consent form
2 The researcher calls the participant to discuss their involvement in the study

3 Receiving the focus group or interview confirmation email
4 They present for the focus group or interview (prior to commencing)

## DATA COLLECTION INSTRUMENT

The data collection instrument is an interview schedule developed by the research team based on findings from the author's existing systematic review examining barriers and facilitators for nurses and physicians in secondary and tertiary care towards integrating genomics into practice and the World Health Organization (WHO) Integrated Care for Chronic Conditions framework (White et al., 2020; World Health Organization, 2002)(Appendix 5). The schedule will remain the same across focus groups and interviews so the same questions are explored regardless of whether data was generated through a focus group or interview.

## DATA COLLECTION

Participants will be offered a focus group initially, however, if the participant is unable to attend or does not wish to participate in the focus group, a one-on-one semi-structured interview will be offered at a time and date mutually agreed upon by myself and participant. Focus groups and interviews will be conducted via the online video-conferencing platform, Zoom (Zoom [computer software], 2020) at a time and date decided by myself and participant. I will digitally record the focus groups and interviews using the in-built recording function in Zoom software. I will download the audio and video files from the focus groups to assist with transcription. I will only download the audio file from interviews. Participants can choose whether to have their video on-or-off for the duration of the focus group or interview.

### Focus groups

Online focus groups promote discussion and fluidity of ideas between participants, reduce social desirability bias and overcome logistical barriers such as scheduling issues, travel costs, inconvenience and (Morgan, 1996; Walker, 2013; Woodyatt, Finneran, & Stephenson, 2016). Focus groups will have 4 – 6 participants from the same professional discipline. For example, one focus group will have only palliative care nurse participants, while another will have only genetic counsellors. Homogenous characteristics with focus groups (in this case, related to occupation) aim to increase the comfort of participants and therefore encourage open, free-flowing discussion (Liamputtong, 2011). Within the same occupational focus groups, I will aim to select participants who have a variety of experience, to create opportunities for exploration of differing views (Liamputtong, 2011; Morgan, 1996). Focus groups will run for approximately one hour.

For the first 2 – 3 focus groups, an experienced member of the research team (J.P or C.J) will be present to observe me. Following the focus group, we will reflect upon what went well and areas in which I could improve for next time.

The total number of focus groups will depend on the preference of the individual participants (ie. whether they choose to participate in a focus group or interview), however we expect between 8 – 16 focus groups (2 – 4 focus groups per professional group, based on 4 participants per focus group).

### Semi-structured interviews

If a participant cannot attend or prefers not to participate in a focus group, they will be offered a one-on-one semi-structured interview. One-on-one interviews permit a deep exploration of

participants' responses, allow greater flexibility around scheduling and can overcome social desirability bias (Morris, 2015). Interviews are expected to last 30 – 60 minutes.

The total number of interviews will depend upon the preferences of individual participants, however we expect approximately 16 interviews (four participants from each professional group).

## DATA ANALYSIS

Transcribed focus groups and interviews will be imported to NVivo V12 software for management throughout analysis (QSR International, 2020). We have selected thematic analysis has been selected as a rigorous, yet flexible approach to elicit themes from the data (Braun & Clarke, 2006). Thematic analysis is commonly used in health research, as it provides a relatively straightforward method to qualitative data analysis and can be applied to a variety of methodological approaches (Braun & Clarke, 2006). For health researchers who are not bound to a particular disciplinary analytical tradition, thematic analysis offers accessibility to novice researchers while also capable of generating a rich description of the data set (King, 2004).

There are many approaches to thematic analysis depending on the epistemological position of the researcher. Although this study sits within a larger pragmatic mixed-methods project, we have adopted an interpretive descriptive approach to this qualitative study. As such, we will utilise a data-driven inductive approach to analysis, rather than a framework or theory-driven deductive approach. An inductive approach involves generating codes and themes from the data. With little existing evidence about the research topic, utilising an inductive approach aims to avoid inadvertently missing important themes, particularly those that are relevant to the Australasian context.

An interpretive descriptive approach to thematic analysis will derive themes from the data by following the six non-linear steps outlined by Braun and Clarke (2006). Prior to commencing data analysis, the nature of a 'theme' was considered and discussed. For the most part, we will search for latent themes (ie. the underlying meaning or motivation to a response or action), although there is likely to be some themes which will be more semantic (ie. surface meaning of the response)(Braun & Clarke, 2006).

I will document a contemporaneous audit trail to enable an explicit account of how thematic analysis was applied in this study and detail this process in the final report. At this stage I expect each step to involve:

- Familiarising with the data - transcribing recordings, reading and re-reading transcripts, re-listening to audio-recordings. Transcribed focus group and interview transcripts will be sent to the participants for checking (Cypress, 2017; Yardley, 2008b).
- Generating initial codes - looking for repeated patterns within and across the data and grouping these into codes. Sentences or groups of sentences that refer to one concept will be coded to as many codes as relevant. Sections of data can be re-coded or un-coded as required.
- Searching for themes – codes will be examined, grouped and constructed into initial themes. A number of tools may be used, such as a mind map linking similar codes into broader themes. I will actively search for disconfirming cases (Yardley, 2008b)

- Reviewing themes – initial themes will be critically examined to ensure they accurately represent the data. This involves returning to the raw data and revising codes or themes as necessary
- Defining and naming themes – themes will be finalised alongside a rich and thick description of the themes, ensuring to include all of the complexity instilled by the codes and data
- Producing the report – the final report will narratively describe the overall themes i have generated from the data. This will make up the results section of the research report.

After familiarising with the data, I will engage in peer-review at every stage. At stage 2, this will involve one or two members of the research team experienced in qualitative research reading at least two transcripts in full, making notes about their initial thoughts and inductively assigning their own codes to the data. We will then meet to discuss the meaning we have derived from the data and the codes we independently assigned to each transcript. We will discuss whether there are missing or superfluous codes, or if codes need redefining. At stage 3, I will re-review the codes and discuss the groupings and initial themes with the experienced members of the research team. At stage 4, I will review and discuss the final themes and supporting data with the experienced member of the research team. At stage 6, the research report will be critically analysed by the research team to ensure congruence between the data, codes, themes and narrative. The purpose of peer-review is to encourage reflexivity by stimulating discussion, challenge my assumptions and ensure I can justify the decisions made during the analysis process.

## VALIDITY

In all aspects of the methods described, we have strived to conduct and produce valid, rigorous research. There are a number of frameworks by which to examine rigour in qualitative research. The earliest framework introduced by Lincoln and Guba (1985) described 'trustworthiness'; a concept to which qualitative researchers could achieve by ensuring their research was credible, transferable, dependable, and confirmable. These concepts have evolved to move away from parallels of conventional quantitative criteria and towards concepts that reflect and embrace the inherent interpretive nature of qualitative research. A more modern 'validity' framework by Yardley (2008a) is comprised of four key principles: sensitivity to context, commitment and rigour, coherence and transparency and, impact and importance. We prospectively selected this framework to consider various aspects of the research and reflect upon decisions in an effort to produce good quality qualitative research. Our application of this framework to this study is discussed below.

*Sensitivity to context:* In designing this study, there were several considerations pertaining to context to consider.

I acknowledged the possible effects of my clinical experience and preconceived ideas. Possible impact points included data collection, such as how I ask questions and respond to participant's answers, and data analysis, such as confirmation bias of certain data points while ignoring other important pieces of data. Reflexivity and peer-review are utilised to reduce these effects.

I was aware that participant's might inadvertently provide socially desirable responses if they were aware of my clinical role as a genetic counsellor. As much as practically possible, I will not disclose my clinical profession to participants and strive for respectful, naïve inquiry at all times.

Participants comfort to disclose difficult or uncomfortable information may be impacted by three factors. Firstly, many will choose to conduct their interviews from their place of work where they may be unable to obtain a private space. Being at work could also impact on their ability to remain focused during the interview. Secondly, the COVID-19 pandemic means interviews and focus groups are to be conducted via video-conferencing software. While there are benefits in terms of convenience, the loss of the intangible 'human-ness' of conversation might reduce participants comfort in disclosing difficult information. Thirdly, to the palliative care participants who are aware of my background, I am likely to be viewed as an 'outsider'. Participants might feel I cannot understand their perspectives or feel unwilling to fully open up and share thoughts and feelings. In contrast, genetics professionals are likely to view me as an 'insider', so may feel more willing to disclose uncomfortable information.

*Commitment and rigour:* An in-depth engagement with the topic is demonstrated through my commitment to produce evidence about genetics in palliative care through this and other studies (including a systematic review and quantitative study). Prolonged engagement with the data will be achieved by being the primary researcher conducting interviews and focus groups, maintaining a reflexive journal and research audit trail (Lichtman, 2011). The peer review process described above also contributes to validity by encouraging reflexivity and an in-depth analysis of data.

*Coherence and transparency:* I considered at length my epistemological stance, the methods and purpose of this study to ensure this was a coherent and logical research approach. My assumptions and positions will be made transparent in the research report so readers have adequate information to judge the validity of the research findings. Findings will be reported in accordance with the 'Consolidated Criteria for Reporting Qualitative Research' (COREQ) checklist (Tong, Sainsbury, & Craig, 2007).

*Impact and importance:* We aim to produce important and practical findings for relevant stakeholders. These groups include the clinical genetics and palliative care communities, wider health care organisations and patients and families who may be impacted by the failure of our health system to adequately manage those who simultaneously have palliative and genetic health care needs.

## ETHICAL CONSIDERATIONS

Ethical approval will be sought from the UTS Research Ethics office.

### Study benefits

The findings from this study are likely to fill an evidence gap, which will benefit a number of stakeholders. This includes government agencies, national palliative care, clinical genetics and genetic counselling organisations, health services and hospitals. Importantly, a long-term benefit is for individuals and families to have greater access to genetic health information, to optimise their health and well-being.

### Study risks

Risk to individuals participating in this study are low, although there may be discomfort or inconvenience (National Health and Medical Research Council, 2007). Participants may experience psychological discomfort or embarrassment because of verbalising an opinion or experience in a focus group, which impacts their reputation among the other participants. It is possible a participant could become distressed during the focus group or interview, if a question evokes a painful, shameful or otherwise negative emotion, related to genetics or genomics in their work or personal life. To manage this risk, at the beginning of the focus group, I will address the potential for negative emotions and encourage respect for other's opinions and confidentiality. Participants will be reminded of the voluntary nature of their involvement and their right to end participation at any time without question or penalty. I will monitor the participants for negative emotions and respond accordingly.

As I hold a dual clinical role, there is a risk that a participant may feel coerced into participation through a pre-existing professional relationship. If I know a participant, I will provide additional information during the recruitment phase about their right to not participate and provide reassurance that their non-participation will not harm our pre-existing relationship. Consent can be obtained from another member of the research team if required. If desired by the participant, the focus group or interview can be conducted by another team member.

We have developed a crisis support protocol in the event that a participant becomes distressed because of this research. If the participant is a current employee of a health service, we will suggest they contact their local Employee Assistance Program for counselling. We will provide contact details for Lifeline and Beyond Blue. I will then discuss the event with doctoral supervisors and follow up with the participant as deemed appropriate by the research team.

**CONFIDENTIALITY AND PRIVACY**

Personal and identifiable information, such as name, age, phone number and email address, will be recorded to contact participants and organise their participation. Any personal or identifiable information will be:

- Held in strict confidence, stored in a password protected folder on UTS OneDrive and not shared with any third party
- Accessed only by the research team on password protected, UTS-owned computers
- Not be transferred between computers by email or external storage device (eg. USB or hard-drive)
- De-identified as soon as practically possible to ensure working documents (which may need to be transferred between members of the research team) do not contain identifiable information

**DATA STORAGE AND RETENTION**

Data will be managed according to the UTS Guidelines for the Management of Research Data. A comprehensive research data management plan is documented in 'Stash' (UTS data management planning software). The following data types will be produced during this study:

- Excel spreadsheets (.xlsx)
- Consent forms and emails (.pdf)
- Video and audio recordings (.mp4)
- Focus group and interview transcripts (.docx)

- NVivo v12 data analysis software files (.nvp)
- Field notes (hard copy diary)

Working files will be stored on my password protected UTS OneDrive account. OneDrive folders that contain personal, identifiable or sensitive information will be password protected. This includes, but is not limited to, contact information, consent forms and transcripts.

Emails to potential and actual participants will only originate from my UTS Microsoft Outlook account. After sending or receiving an email, the email will be downloaded, saved as a PDF document and stored in a password-protected folder on the OneDrive system. The original email will be deleted from my Outlook account.

Upon completion of this study, all data will be archived to the UTS eResearch Store for a period of five years. After this time, all files containing personal, identifiable or sensitive information will be destroyed with the assistance of a UTS data librarian.

**CONFLICTS OF INTEREST**

If the participants are aware of my dual role as a researcher and clinical genetic counsellor, they may ask for personalised genetic health information before, during or after the focus group or interview. I will politely refrain from providing genetic health information to any participant in my role as a researcher. I will provide the participant with information about how to access a clinical genetics service for these questions to be addressed. I will address questions of a general nature as they arise.

C.J and J.P have no conflicts of interest. There are no financial or other benefits arising from the conduct of this study for any of the researchers.

**FUNDING**

This project is funded by a Translational Cancer Research Network PhD Scholarship Top-up Award, supported by the Cancer Institute NSW. SW is the recipient of a PhD stipend scholarship from the Translational Cancer Research Network (http://www.tcrn.unsw.

edu.au/). The TCRN holds no conditions on the conduct or outcome of this research.

**RESEARCH OUTCOMES**

The outcome of this research will be:

1) Publication in a quality, peer-reviewed journal
2) Presentations at academic conferences in the form of poster or oral presentation
3) Contribute to S.W.'s doctoral thesis.

There is no anticipated secondary use of the data. Results will not be returned to participants, except in the format of a published manuscript.

**PUBLICATION PLAN**

Target Journal: Palliative Medicine (Impact factor: 4.956)

Word limit: 3000

# SUPPLEMENTARY FILE C2: INITIAL HREC APPROVAL FOR QUALITATIVE STUDY (ETH20-5046)



## FW: HREC Approval Granted - ETH20-5046

Stephanie White <Stephanie.White@uts.edu.au>
Tue 8/4/2020 8:31 AM
To: Steph White <Stephanie.A.White@student.uts.edu.au>
1 attachments (309 KB)
Ethics Application.pdf;

**From:** Research.Ethics@uts.edu.au
**Sent:** Tuesday, August 4, 2020 8:30:55 AM (UTC+10:00) Canberra, Melbourne, Sydney
**To:** Research Ethics; Jane Phillips; Stephanie White; Steph White
**Subject:** HREC Approval Granted - ETH20-5046

Dear Applicant

**Re: ETH20-5046 - "Genetics and Genomics in Palliative Care: Exploring the Views and Experiences of Palliative Care and Genetic Clinicians"**

Thank you for your response to the Committee's comments for your project. The Committee agreed that this application now meets the requirements of the National Statement on Ethical Conduct in Human Research (2007) and has been approved on that basis. You are therefore authorised to commence activities as outlined in your application.
You are reminded that this letter constitutes ethics approval only. This research project must also be undertaken in accordance with all UTS policies and guidelines including the Research Management Policy.

Your approval number is UTS HREC REF NO. ETH20-5046.

Approval will be for a period of five (5) years from the date of this correspondence subject to the submission of annual progress reports.

The following standard conditions apply to your approval:
· Your approval number must be included in all participant material and advertisements. Any advertisements on Staff Connect without an approval number will be removed.
· The Principal Investigator will immediately report anything that might warrant review of ethical approval of the project to the Ethics Secretariat (Research.Ethics@uts.edu.au).
· The Principal Investigator will notify the UTS HREC of any event that requires a modification to the protocol or other project documents, and submit any required amendments prior to implementation. Instructions on how to submit an amendment application can be found here.
· The Principal Investigator will promptly report adverse events to the Ethics Secretariat. An adverse event is any event (anticipated or otherwise) that has a negative impact on participants, researchers or the reputation of the University. Adverse events can also include privacy breaches, loss of data and damage to property.
· The Principal Investigator will report to the UTS HREC annually and notify the HREC when the project is completed at all sites. The Principal Investigator will notify the UTS HREC of any plan to extend the duration of the project past the approval period listed above through the progress report.

· The Principal Investigator will obtain any additional approvals or authorisations as required (e.g. from other ethics committees, collaborating institutions, supporting organisations).
· The Principal Investigator will notify the UTS HREC of his or her inability to continue as Principal Investigator including the name of and contact information for a replacement.
This research must be undertaken in compliance with the Australian Code for the Responsible Conduct of Research and National Statement on Ethical Conduct in Human Research.

You should consider this your official letter of approval. If you require a hardcopy please contact the Ethics Secretariat.

If you have any queries about your ethics approval, or require any amendments to your research in the future, please don't hesitate to contact the Ethics Secretariat and quote the ethics application number (e.g. ETH20-xxxx) in all correspondence.

Yours sincerely,
Prof Beata Bajorek
Chairperson
UTS Human Research Ethics Committee
C/- Research Office University of Technology Sydney
E: Research.Ethics@uts.edu.au
*Ref: E38*

## SUPPLEMENTARY FILE C3: OUT OF SESSION AMENDMENT HREC APPROVAL

**RE: Amendment enquiry: ETH20-5046 - Updated Personnel and Changes to working under Section 6**

Quinn Nguyen <NhuQuynh.Nguyen@uts.edu.au>on behalf of Research Ethics research.ethics@uts.edu.au

Fri 2/12/2021 8:50 AM

**To**: Steph White <Stephanie.A.White@student.uts.edu.au>; Research Ethics research.ethics@uts.edu.au

**Cc:** Chris Jacobs <Chris.Jacobs@uts.edu.au>; Erin Turbitt Erin.Turbitt@uts.edu.au

Hi Steph,

Our apologies for the delayed response. I have discussed your amendment with my team and since this is a minor amendment, we can approve these changes via email. Chris and Erin: you have been copied in this email for your information. If you have any queries or concerns regarding this amendment, please feel free to let me know. If I can be of any further assistance, please feel free to let me know.

Kind regards

Quinn

Quinn Nguyen (Ms)

Research Ethics Administrator

University of Technology Sydney

T. +61 (02) 9514 9772

PO Box 123 Broadway NSW 2007 Australia

Visit the Research pages of Staff Connect for lots of useful information essential for researchers.

Book an ethics clinic.

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**From:** Steph White <Stephanie.A.White@student.uts.edu.au>

**Sent:** Wednesday, 10 February 2021 9:29 AM

**To:** Research Ethics research.ethics@uts.edu.au

**Subject**: Amendment enquiry: ETH20-5046

Hi there,

My name is Stephanie, I am a PhD candidate with GSH. I just had a chat on the phone with one of your staff about an ethics amendment for change of supervisor. She mentioned I could email you about this change rather than submit a formal amendment. However, I do also want to slightly change the eligibility criteria for one of my participant groups, so if I still need to submit an amendment for this purpose, I am happy to. The woman I spoke with wasn't sure so

suggested sending an email for advice in the first instance. I'll list a summary of the changes here:

- Change Prof Jane Phillips from chief investigator to associate investigator (she has recently left UTS, but remains an adjunct professor. She has taken a job with QUT).
- Change Dr Chris Jacobs from Associate Investigator to Chief Investigator.
- Add Dr Erin Turbitt as Associate Investigator (she is new personnel to the project)
- Change the wording of the following inclusion and exclusion criteria (listed under section 6: Recruitment of participants)
    - From "registered nurses directly involved in providing palliative care to patients in Australasia (Australia and New Zealand)", to "Registered nurses directly involved in palliative care in Australasia (Australia and New Zealand). This may include but is not limited to direct clinical care, research, teaching or policy development.
    - "from "Nurses, medical doctors and genetic counsellors who have not been in clinical practice for more than 5 years", to "Nurses, medical doctors and genetic counsellors who have not met their professional registration (or equivalent) requirements as stipulated by their relevant professional boards within the last five years"

The inclusion/exclusion criteria wording change is in response to recognising that many registered nurses who are experts in palliative care may not be providing direct clinical care, but rather working in academic institutions or policy development. Our previous inclusion criteria excluded these valuable participants without good justification.

As I said, very happy to put this in an amendment if required. Once I've received your advice, I'll include the relevant supervisors on this email chain so they're aware which process pathway we'll go down.

Kind regards,

Stephanie White

PhD Candidate, University of Technology Sydney

Genetic Counsellor [FHGSA], RN

# UTS HREC Approval - ETH20-5347

To: Steph White
Wed 9/23/2020 7:48 AM

Ethics Application.pdf
**190 KB**
Reply
Forward

**From:** Research.Ethics@uts.edu.au
**Sent:** Wednesday, September 23, 2020 7:48:00 AM (UTC+10:00) Canberra, Melbourne, Sydney
**To:** Research Ethics; Steph White; Jane Phillips; Stephanie White; Chris Jacobs
**Subject:** UTS HREC Approval - ETH20-5347

Dear Applicant

**Re: ETH20-5347 - "Genetics and Genomics in Palliative Care: Exploring the Views and Experiences of Palliative Care and Genetic Clinicians"**

The HREC Expedited Review Committee reviewed your amendment application for your project and agreed that the amendments meet the requirements of the NHMRC National Statement on Ethical Conduct In Human Research (2007). I am pleased to inform you that the Committee has approved your request to amend the protocol as follows:

EXPLAINER: existing participant (EP) is an individual who has already consented & participated in this study. Potential participants (PP) are individuals or groups of individuals known to the EP, who has not yet agreed to participate in this research but may be interested. Four changes to recruitment: 1. Snowball sampling: The researchers will ask EPs (past & future) if they know any PP. If so, the researcher will ask whether the PP has reasonably consented to being contacted by the researcher. 1a. If there is no consent, the researcher can forward a standard email & Participant Information Statement (written for snowball recruitment - see attachments) to the EP, who can forward this to PP. 1b. If there is consent (ie. the EP explicitly states the PP is aware of the study, interested in participating & has provided consent to the EP to share their contact details), then the researcher can contact the PP directly. The EP will be asked if they consent to the standard email naming them as a referrer. If they do not consent to this, the researcher will utilise strategy 1a instead. 2. Circulation of invitation via any relevant organisation who agrees to distribute to their membership, in addition to those specified in the original application. The invitation circulated would be the same as that already approved in the original application. 3. Share the invitation with known contacts of the research team: Members of the research team could forward invitations to PP, or ask the PP to circulate the invitation more widely to other PP. All processes for managing known relationships in the original application will be upheld. The research team would not email their known contacts more than twice with the research invitation. A standard email template will be used (see attachments) 4. Advertise participation in presentations at national/international conferences (see attachments).

This amendment is subject to the standard conditions outlined in your original letter of approval. You are reminded that this letter constitutes ethics approval only. This research project must also be undertaken in accordance with all UTS policies and guidelines including

the Research Management Policy.

You should consider this your official letter of approval. If you require a hardcopy please contact the Research Ethics Secretariat.

To access this application, please click here, a copy of your application has also been attached to this application

If you wish to make any further changes to your research, please contact the Research Ethics Secretariat in the Research Office.

In the meantime I take this opportunity to wish you well with the remainder of your research.

Yours sincerely,

Prof Beata Bajorek
Chairperson
UTS Human Research Ethics Committee
C/- Research Office
University of Technology Sydney
T: (02) 9514 2478
Research.Ethics@uts.edu.au | Website
PO Box 123 Broadway NSW 2007

Ref: E13-3

## SUPPLEMENTARY FILE C5: RISK MATRIX FOR QUALITATIVE STUDY (SUBMITTED WITH HREC APPLICATION)

| Code | Risk | Magnitude | Likelihood | Mitigation |
|---|---|---|---|---|
| 1a | Participants may experience psychological discomfort through their involvement in a focus group. They may feel embarrassed/ashamed when verbalising a professional opinion or experience in front of others, particularly if another participant disagrees with their opinion. | We expect the magnitude of risk to be inconvenience or discomfort. Professional conflict within a focus group discussion may cause a participant discomfort if they feel they are not being listened to, their views are not valid or 'incorrect'. However, professional debate or disagreement within a clinical setting (such as a hospital or health service) is a common phenomenon and therefore this is not expected to produce significant harm for participants. | We expect the likelihood of this risk to be possible. The purpose of a focus group is to generate convergent and divergent opinions on a topic, so some professional disagreements are expected. However, even if these disagreements occur, we expect these to be of a professional nature and unlikely to generate a more negative emotion than discomfort. | Participants will be made aware that participation is voluntary (written within the PIS and verbally reminded), so that if they experience a negative emotion they may choose to withdraw at any time without consequence.<br>At the beginning of the focus group, the facilitator will acknowledge the possibility of be differing opinions within the group. The facilitator will remind all participants to engage in professional discourse and to respect other's opinions |
| 1b | Focus group participant/s may breach the privacy and confidentiality of other participant/s by disclosing the group member's identities to outside parties | We expect the magnitude of risk to be inconvenience or discomfort. This is because the research topic is professional and clinician's participating in health research is common. Therefore, a privacy breach is unlikely to be personally or professionally harmful to the participant. | We expect the likelihood of this risk to be possible. Although all participants will be reminded of maintaining each other's privacy and confidentiality prior to the commencement of the, the researcher cannot guarantee the participants' will adhere to these principles. | Prior to the focus group, the researcher will inform the participants they are able to complete the focus group or interview with their video on-or-off and are able to change their Zoom display name, if they wish. At the beginning of the focus group, the researcher will remind the participants of each other's privacy and confidentiality and request they do not speak about other's identities or comments outside of the focus group. |
| 1c | A focus group participant's reputation may be at risk if they make statements of a negative nature within the focus group | We expect the magnitude of risk to be harmful, although this would depend on the nature of the comment. A statement by a | We expect the likelihood of this risk to be slight. Participants will be aware they are involved in research with health professionals, and are therefore | At the beginning of the focus group, the researcher will remind participants that the focus group is a professional forum and everyone |

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|---|---|---|---|---|
| | discussion (eg. disclosure of unsafe or illegal conduct, express politically incorrect views) | participant, which is viewed negatively by the group, could have an ongoing professional impact on the participant. | more likely to moderate their behaviour and comments to be socially acceptable within their professional culture. | is expected to treat others, within and outside of the focus group, with respect and dignity. They will also be reminded that the researcher will maintain their privacy and confidentiality, within the confines of the law |
| 2a | Participants may feel self-conscious in a focus group or interview, or experience feelings of inadequacy if they perceive they are not able to respond to the facilitator/interviewer's questions | We expect the magnitude of risk to be inconvenience or discomfort. Clinicians are experienced professionals who are expected to communicate with people from a variety of occupations and backgrounds, so communicating with researchers is likely to be a skill they already possess. They are also experienced in conversing on health-related topics, answering questions from other professionals and patients/family members, and having an awareness of their scope of practice. Therefore, being asked questions about a topic they may be unfamiliar with is likely to be a concept they are familiar with and unlikely to cause harm. | We expect the likelihood of this risk to be possible. Self-consciousness is a common emotion. Furthermore, existing literature suggests some palliative care nurses and doctors have low knowledge of genetics and genomics. Therefore it is possible the palliative care nurses and doctors may feel unequipped to answer the interviewer's questions. | Participants will have read the PIS, which notes the researchers are interested in participants with a range of views and experiences. This will be reiterated at the beginning of the focus group or interview. The researcher will monitor the participant for signs of self-consciousness or inadequacy (eg. negative comments about their answers or performance, downcast eyes, fidgeting). If these are noted, the researcher will (as deemed appropriate) normalise these feelings, ask the participant if they'd like to skip the question, remind the participant they are free to answer as many questions as they wish, and/or to withdraw at any point. |
| 2b | Participants may experience a negative emotion if a question reminds them of a painful experience with genetics or genomics in their professional or personal life. These negative emotions may include shame, distress, anger, offence, sadness or discomfort. | We would categorise the magnitude of risk as discomfort or harmful. Depending on the degree or the difficult or painful memory or experience (whether professional or personal), the participant may experience harm if they are reminded of this through the course of the focus group or interview. | We expect the likelihood of this risk to be possible. Existing literature suggests palliative care nurses and doctors feel underequipped to integrate genetics and genomics into practice, which means it is possible the participant/s may have had a professional experience which they did not feel prepared to manage. Similarly, it is possible genetics clinicians may have | At the beginning of the focus group or interview, the researcher will acknowledge the potential for negative emotions to arise during or after the focus group or interview. The researcher monitor the participant/s during the focus group and interview for signs of distress. If signs of distress are noted, the researcher will ask the participant if |

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| | | | had an experience of providing genetic care to a dying person which produced a negative emotion. | they wish to skip the question or stop their participation. Further details and steps related to this risk are summarised in the Distress and Safety Protocol (attached). |
| 2c | As the researcher primarily conducting the data collection (Stephanie White) also holds a clinical role, it is possible the researcher and participant may have a pre-existing relationship, and therefore the participant may feel coerced into being involved with the study (discussed more in the next section). | We would categorise the magnitude of risk as discomfort. They may experience an internal conflict by feeling coerced into giving up their time for research which they do not wish to participate in, for professional or personal reasons. | We expect the likelihood of this risk to be possible. The researcher conducting the focus groups and interviews (Stephanie White) is relatively junior in her field, which is likely to reduce the chance of this risk occurring. | Potential participants will be required to contact the researcher to express their interest in being involved in the research, which aims to reduce coercion (instead of the researcher approaching potential participants). All potential participants, including those known to the researcher previously, will be told, in writing and verbally, that their participation is voluntary |
| 2d | Participants will be required to give up their time to participate in a focus group or interview for which they will not be compensated | We would categorise the magnitude of risk as inconvenience. Without compensation for their time, participant may feel inconvenienced by losing time which could have been otherwise spent on professional or personal matters. However, given the time committment is expected to be maximum two hours (including time to read PIS, complete demographic survey, schedule interview/focus group and review transcript), the participant is not expected to experience discomfort or harm as a result. | We expect the likelihood of this risk to be unavoidable, as their participation cannot proceed unless they agree to commit the time required to participate | Although the time commitment of participants is unavoidable, they will be informed of the amount of time required for the focus group or interview and other related activies, in writing and verbally, prior to scheduling their participation. |
| 2e | The researchers' may inadvertently breach the privacy or confidentiality of a participant | We would categorise the magnitude of risk as discomfort, harmful or painful. The extent of risk of an inadvertent privacy breach at the hands of the researcher would | We expect the likelihood of this risk to be slight. The research team has considered this risk carefully and developed a data management plan, | Mitigation of this risk is detailed in section 8. Briefly, any documents containing personal or identifying information will be deidentified as soon as possible. All working |

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| | | depend at which stage the identifying information was made accessible to outside parties, how much information was disclosed and the nature of the information. In the worst case scenario, a participant's demographic information, personal details (such as name and email address) and responses to focus group or interview questions could be linked. This would result is a serious breach of trust between the participant, researchers and UTS, and depending on the nature of the data, may result in harm to the participant's personal or professional life. | which mitigates this risk (discussed further in section 8). | documents will be kept on a secure server (UTS OneDrive). No documents containing identifiable details will be transferred between the researchers by email or external storage (such as USB), and will not be shared with any third parties.<br>At completion of the research, all research data will be stored on a secure long-term server (UTS eResearch Store) for a period of 5 years.<br>Any research outputs (journal articles, conference presentations) will not include participant's identifying details |

## SUPPLEMENTARY FILE C6: DISTRESS AND SAFETY PROTOCOL

**Genetics and Genomics in Palliative Care: Exploring the Views and Experiences of Palliative Care and Genetic Clinicians**

**Distress and Safety Protocol: Health care professional (nurse, doctor or allied health)**

**UTS HREC approval number: ETH20-5406/5347**

The following protocol will be put in place should a participant became distressed and require either additional or ongoing assistance. Any member of the research team enacting this protocol will discuss this with the research team and Chief Investigator.

**PRIOR to the commencement of any focus group or interview:**

- Information regarding the availability of support and/or counselling services available (should it be required) will be provided to all potential and actual study participants in the participant information sheet (PIS).
- The researcher will provide potential and actual participants with information about the risks and benefits of the research prior to commencement of the focus group or interview. This information will be available in the PIS and the researcher will be available to answer any additional questions so individuals are informed when providing consent.

**Strategies to assist those distressed DURING an interview:**

If a participant becomes uncomfortable or distressed while discussing any topic during the interview, the researcher will take the following action:

- Suggest the interview be paused and/or terminated.
- If the participant wishes this to happen, the interview will be ceased.
- A member of the research team who is a health professional (Stephanie White, Dr Chris Jacobs or Professor Jane Phillips) will be contacted to spend time with the participant, provide support and help within their scope of practice, and to discuss their concerns. This contact will likely be via telephone or zoom
- After consulting the chief investigator (Professor Jane Phillips), a recommendation will be made that the participant speak to a counselling professional or relevant support service to discuss their concerns, and referred if they agree. The options include contacting:
    - Their own doctor
    - Counselling through their own employee assistance program (eap, if available). The researcher can assist the participant to access their relevant eap.
    - An accessible, anonymous support service such as lifeline (telephone 13 11 14) or beyond blue (telephone 1399 22 46 36)

**Strategies to assist those distressed POST an interview:**

In follow-up:

- A follow-up telephone call will be made by the chief investigator (Professor Jane Phillips) the following day to ensure that the participant is well and to determine feasibility of a follow-up interview if one is planned.
- The researcher involved in the interview will contact the UTS Research Ethics office to determine whether an adverse event report is required

# SUPPLEMENTARY FILE C7: PARTICIPANT INFORMATION SHEET FOR GENETIC HEALTH PROFESSIONALS

**PARTICIPANT INFORMATION SHEET (Genetic health professionals)**

**Genetics and Genomics in Palliative Care: Exploring the Views and Experiences of Palliative Care and Genetics Clinicians**

**UTS HREC REFERENCE NUMBER ETH20-5046/5347**

## WHO IS DOING THE RESEARCH?

My name is Stephanie White and I am a PhD candidate at UTS. My supervisors are Professor Jane Phillips (Professor of Palliative Nursing, UTS) and Dr Chris Jacobs (Senior Lecturer in Genetic Counselling).

## WHAT IS THIS RESEARCH ABOUT?

This research is to find out about exploring the views and experiences of palliative care and genetic health care professionals in discussing genetic health information with people (and/or their families) who have palliative care needs.

## FUNDING

Funding for this project has been received from the Translational Cancer Research Network.

## WHY HAVE I BEEN ASKED?

You have been invited to participate in this study because you are a genetic counsellor or doctor working in a clinical genetics or familial cancer setting, and you work in Australia or New Zealand.

## IF I SAY YES, WHAT WILL IT INVOLVE?

If you decide to participate, I will invite you to participate in a 1-hour online focus group that will be audio- and video-recorded and transcribed. If you prefer, you can participate in 1-hour individual online semi-structured interview that will be audio recorded and transcribed. After the focus group or interview, you will be sent the transcript to check it is accurate. However, if you don't want to check the transcript for accuracy, you don't have to. The researcher will use the recordings of your interview or focus group to accurately transcribe the sessions. Once transcription is complete, the recordings will be destroyed. We will ask whether you can recommend other potential participants for the study.

## ARE THERE ANY RISKS/INCONVENIENCE?

Yes, there are some risks/inconvenience. This includes giving up your time to participate, for which you won't be remunerated. Questions about past experiences could bring up upsetting memories, or make you feel embarrassed or uncomfortable. If you participate in a focus group, other members of the group may know your identity, meaning your anonymity cannot be guaranteed. Forwarding on research invitations or contacts may be inconvenient or cause distress. The researchers will do their best to mitigate these risks. If you become upset or distressed because of participation in this study, the researchers can help you access counselling or support.

## DO I HAVE TO SAY YES?

Participation in this study is voluntary. It is completely up to you whether or not you decide to take part.

## WHAT WILL HAPPEN IF I SAY NO?

If you decide not to participate, it will not affect your relationship with the researchers or the University of Technology Sydney. If you wish to withdraw from the study once it has started, you can do so at any time without having to give a reason, by contacting Stephanie White (stephanie.white@uts.edu.au).

If you withdraw from the study, we will destroy your interview recordings and transcripts. Focus group recordings will not be destroyed, but your responses in the transcript will be removed. However, it may not be possible to withdraw your data from the study results if these have already had your identifying details removed.

If you decide to leave the research project, we will not collect additional personal information from you, although personal information already collected will be retained to ensure that the results of the research project can be measured properly and to comply with law. You should be aware that data collected up to the time you withdraw will form part of the research project results. If you do not want them to do this, you must tell them before you join the research project.

## CONFIDENTIALITY

By signing the consent form, you consent to the research team collecting and using personal information about you for the research project. All this information will be treated confidentially. It will be kept on a secure, encrypted UTS server. Data is managed in accordance with relevant guidelines and policies, including the UTS Guidelines for the Management of Research Data, the UTS Research Management Policy and the Australian Code for the Responsible Conduct of Research. Your information will only be used for the purpose of this research project and it will only be disclosed with your permission, except as required by law.

We plan to discuss and publish the results at academic conferences and in peer-reviewed publications to further our collective understanding of the views and experiences of genetics health professionals towards genetics in palliative care. In any publication, information will be provided in such a way that you cannot be identified.

## WHAT IF I HAVE CONCERNS OR A COMPLAINT?

If you have concerns about the research that you think my supervisor or I can help you with, please feel free to contact us by email (stephanie.white@uts.edu.au).

You will be given a copy of this form to keep.

**NOTE:**

This study has been approved in line with the University of Technology Sydney Human Research Ethics Committee [UTS HREC] guidelines. If you have any concerns or complaints about any aspect of the conduct of this research, please contact the Ethics Secretariat on ph.: +61 2 9514 2478 or email: Research.Ethics@uts.edu.au], and quote the UTS HREC reference number. Any matter raised will be treated confidentially, investigated and you will be informed of the outcome.

## SUPPLEMENTARY FILE C8: PARTICIPANT INFORMATION SHEET FOR PALLIATIVE CARE HEALTH PROFESSIONALS

**PARTICIPANT INFORMATION SHEET (Palliative care health professionals)**

**Genetics and Genomics in Palliative Care: Exploring the Views and Experiences of Palliative Care and Genetics Clinicians**

**UTS HREC REFERENCE NUMBER ETH20-5046/5347**

### WHO IS DOING THE RESEARCH?

My name is Stephanie White and I am a PhD candidate at UTS. My supervisors are Professor Jane Phillips (Professor of Palliative Nursing, UTS) and Dr Chris Jacobs (Senior Lecturer in Genetic Counselling).

### WHAT IS THIS RESEARCH ABOUT?

This research is to find out about exploring the views and experiences of palliative care and genetic health care professionals in discussing genetic health information with people (and/or their families) who have palliative care needs.

### FUNDING

Funding for this project has been received from the Translational Cancer Research Network.

### WHY HAVE I BEEN ASKED?

You have been invited to participate in this study because you are a registered nurse or doctor working in palliative care, and you work in Australia or New Zealand.

### IF I SAY YES, WHAT WILL IT INVOLVE?

If you decide to participate, I will invite you to participate in a 1-hour online focus group that will be audio- and video-recorded and transcribed. If you prefer, you can participate in 1-hour individual online semi-structured interview that will be audio recorded and transcribed. After the focus group or interview, you will be sent the transcript to check it is accurate. However, if you don't want to check the transcript for accuracy, you don't have to. The researcher will use the recordings of your interview or focus group to accurately transcribe the sessions. Once transcription is complete, the recordings will be destroyed. We will ask whether you can recommend other potential participants for the study.

### ARE THERE ANY RISKS/INCONVENIENCE?

Yes, there are some risks/inconvenience. This includes giving up your time to participate, for which you won't be remunerated. Questions about past experiences could bring up upsetting memories, or make you feel embarrassed or uncomfortable. If you participate in a focus group, other members of the group may know your identity, meaning your anonymity cannot be guaranteed. Forwarding on research invitations or contacts may be inconvenient or cause distress. The researchers will do their best to mitigate these risks, but if you become upset or distressed because of participation in this study, the researchers can help you access counselling or support.

### DO I HAVE TO SAY YES?

Participation in this study is voluntary. It is completely up to you whether or not you decide to take part.

### WHAT WILL HAPPEN IF I SAY NO?

If you decide not to participate, it will not affect your relationship with the researchers or the University of Technology Sydney. If you wish to withdraw from the study once it has started, you can do so at any time without having to give a reason, by contacting Stephanie White (stephanie.white@uts.edu.au).

If you withdraw from the study, we will destroy your interview recordings and transcripts. Focus group recordings will not be destroyed, but your responses in the transcript will be removed. However, it may not be possible to withdraw your data from the study results if these have already had your identifying details removed

If you decide to leave the research project, we will not collect additional personal information from you, although personal information already collected will be retained to ensure that the results of the research project can be measured properly and to comply with law. You should be aware that data collected up to the time you withdraw will form part of the research project results. If you do not want them to do this, you must tell them before you join the research project.

## CONFIDENTIALITY

By signing the consent form, you consent to the research team collecting and using personal information about you for the research project. All this information will be treated confidentially. It will be kept on a secure, encrypted UTS server. Data is managed in accordance with relevant guidelines and policies, including the UTS Guidelines for the Management of Research Data, the UTS Research Management Policy and the Australian Code for the Responsible Conduct of Research. Your information will only be used for the purpose of this research project and it will only be disclosed with your permission, except as required by law.

We plan to discuss and publish the results at academic conferences and in peer-reviewed publications to further our collective understanding of the views and experiences of palliative care health professionals towards genetics in palliative care. In any publication, information will be provided in such a way that you cannot be identified.

## WHAT IF I HAVE CONCERNS OR A COMPLAINT?

If you have concerns about the research that you think my supervisor or I can help you with, please feel free to contact us by email (stephanie.white@uts.edu.au).

You will be given a copy of this form to keep.

**NOTE:**

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## SUPPLEMENTARY FILE C9: RECRUITMENT DETAILS

**Recruitment Strategy**

- The Human Genetics Society of Australasia and two of its special interest groups (Australasian Society of Genetic Counsellors and Australasian Association of Clinical Geneticists) circulated an email invitation to their members on two occasions (invitation 1)
- Three palliative care and palliative medicine organisations circulated email invitations to their members on two occasions: Palliative Care Nurses Australia (PCNA), Palliative Care Nurses New Zealand, and the Australian and New Zealand Society of Palliative Medicine (invitation 2)
- An invitation was emailed to delegates of the PCNA Annual Conference 2020 (invite 2) and a conference presentation given by Stephanie White at the 2020 PCNA conference included a slide inviting contact from the audience (invitation 3).
- Two multidisciplinary research organisations circulated an email invitation to their members on two occasions: Translational Cancer Research Network (TCRN) and Maridulu Budyari Gumal (formerly known as SPHERE) (invitation 1 and 2)
- A social media invitation was advertised through the research teams' individual and associated professional Twitter accounts (invitation 4)
- To supplement low response rates, an ethics amendment (ETH20-5347) allowed the research team to
    - Circulate the email invitation to their known contacts (invitation 5)
    - Ask participants to forward the research invitation to any eligible and potentially interested clinicians (invitation 6)
    - Circulate the invitation to any eligible and potentially interested clinicians (invitation 7)

1. **Email Invitation to Genetics Health Professionals**. This invitation was circulated by the Human Genetics Society of Australasia, Australasian Society of Genetic Counsellors and Australasian Association of Clinical Geneticists.

**Genetics in palliative care: We want to know what you think about discussing genetics with people who have palliative care needs**

Integration of genetics into routine medical care has quickly become a hot topic for policy makers, health care organisations and consumers. We need to add your voice to the conversation, to understand the frontline barriers and facilitators to discussing genetics with people who have palliative care needs.

If you are a genetic counsellor, clinical geneticist or other doctor working in a clinical genetics or familial cancer setting, we invite you to participate in an online focus group or interview. We want to talk to people with a range of views and experience. If you are interested in participating or more information, please contact Stephanie White (stephanie.white@uts.edu.au)

This research is being conducted by Stephanie White, a PhD candidate at the University of Technology Sydney, supervised by Professor Jane Phillips and Dr Chris Jacobs. Ethical approval has been granted by the University of Technology Sydney Research Ethics office (UTS HREC reference number ETH20-5046).

2. **Email Invitation to Palliative Care Health Professionals.** This invitation was circulated by the Australia and New Zealand Society for Palliative Medicine, Palliative Care Nurses Australia and Palliative Care Nurses New Zealand.

**Genetics in palliative care: We want to know what you think about discussing genetics with people who have palliative care needs**

Integration of genetics into routine medical care has quickly become a hot topic for policy makers, health care organisations and consumers. We need to add your voice to the conversation, to understand the frontline barriers and facilitators to discussing genetics with people who have palliative care needs.

If you are a registered nurse or doctor who works with people with palliative care needs, we invite you to participate in an online focus group or interview. We want to talk to people with a range of views and experience. If you are interested in participating or more information, please contact Stephanie White (stephanie.white@uts.edu.au)

This research is being conducted by Stephanie White, a PhD candidate at the University of Technology Sydney, supervised by Professor Jane Phillips and Dr Chris Jacobs. Ethical approval has been granted by the University of Technology Sydney Research Ethics office (UTS HREC reference number ETH20-5046).

3. **Conference Invitation.** Text for PowerPoint slide at PCNA 2020 conference**.**

We are still recruiting

If you're interested in participating, please contact Stephanie White

stephanie.white@uts.edu.au

Twitter: @_Steph__White_

4. **SOCIAL MEDIA INVITATION. THE RESEARCH TEAM WILL ADVERTISE THIS 276-CHARACTER INVITATION VIA THEIR PERSONAL AND AFFILIATED TWITTER ACCOUNTS.**

"What do you think about discussing genetics with people with palliative care needs? Aust/NZ palliative care nurses and doctors, genetic counsellors and clinical geneticists: You are invited to participate in an online focus group or interview. DM or email me (stephanie.white@uts.edu.au) to find out more. Please RT!"

5. **STANDARD EMAIL TEMPLATE INVITATION TO RESEARCHERS' KNOWN CONTACTS**

Dear *name of potential participant*

I hope this email finds you well. I'm contacting you about a study I'm involved in called "Genetics and genomics in palliative care: Views and experiences of palliative care and genetics clinicians". It's a qualitative study that involves one-hour focus groups or interviews. I've attached the Participant Information Statement (PIS) if you're interested in reading more.

The reason for reaching out is two-fold:

1. If you're interested in participating yourself, you can contact Stephanie White (PhD candidate at UTS) at stephanie.white@uts.edu.au

2. If you know of anyone who may be interested in participating, you could forward them this email along with the PIS, and they may contact Stephanie directly.

I appreciate you are busy, so please know that you are under no obligation to do either of these two things. If you choose not to participate or forward the invitation, it will not affect your relationship with me, the research team or UTS.

If I don't hear back from you, I'll reach out just once more in a couple of weeks. However, if I don't hear from you after that, I won't email you again about this.

I hope you're staying safe and well.

All the best,

*name of researcher*

**6. STANDARD EMAIL TEMPLATE INVITATION TO EXISTING PARTICIPANT – SNOWBALL RECRUITMENT**

Dear *name of participant*

Thank you kindly for participating in our study. As we discussed, we are asking participants whether they are willing to share the research invitation with other eligible individuals (palliative care nurses and doctors, genetic counsellors and clinical geneticists in Australia and New Zealand). You are under no obligation to share this invitation if you don't want to.

If you know of anyone who may be interested, please forward this email to them, including the Participant Information Statement attached. If they are interested in participating, they can contact me directly at stephanie.white@uts.edu.au.

Thanks again and all the best.

Warm regards,

Stephanie.

**7. STANDARD EMAIL TEMPLATE INVITATION TO POTENTIAL PARTICIPANT – SNOWBALL RECRUITMENT**

Dear *name of potential participant*

My name is Stephanie White and I am contacting you because your colleague, *name of referrer*, provided your contact details as someone who may be interested in participating in research I am conducting. The study is called "Genetics and genomics in palliative care: Views and experiences of palliative care and genetics clinicians". As you are a *insert name of discipline*, I would be interested in hearing your views on this topic. Participation involves a one-hour interview or focus group. I have attached the Participant Information Statement for further information.

If you are interested in participating, you can contact me at stephanie.white@uts.edu.au (or simply reply to this email). Participation is completely voluntary, and choosing not participate will not affect your relationship with *insert name of referrer*, myself, the research team or UTS.

If I don't hear from you, I'll re-send this email once more in a couple of weeks. If I don't hear from you after that, I won't contact you again about this. You are under no obligation to respond and your contact details will not be stored or shared us.

Warm regards,

Stephanie White.

# Genetics Clinician Interview/Focus Group Guide

## Welcome

- Welcome each participant, introduce facilitator, observer and each other

## Overview

- Thank everyone for their participation
- Explain purpose of focus group – to hear about views and experiences of integrating genetics into the care of people (and their families) with palliative needs, particularly to understand the barriers, facilitators and possible strategies to overcome these barriers. Describe how results will be used – to develop an evidence base about genetics in palliative care
- Explain why they have been invited and selected to participate – because they are a genetic counsellor or doctor working in genetics

## Consent questions

- *Refer to verbal consent script template*

## Guidelines

- If you haven't already, please complete the demographics survey as soon as possible
- There are no right or wrong answers, only differing points of view. Even if you don't agree with others, you must listen respectfully as others share their views
- Please respect each other's privacy and confidentiality by refraining from speaking about the focus group outside of this forum
- As we are audio-recording, please ensure one person speaking at a time. Provide brief zoom etiquette instructions
- We're on a first name basis
- If possible, please turn off mobile phones or turn to silent. If you must respond to a call, please do so as quietly as possible and re-join us as soon as you can.
- My role as facilitator will be to guide the discussion among the group
- Thank everyone again. Ask each person to introduce themselves, their professional role and how long they have been working in their field

## Opening question (round-robin)

1. Can you please tell me about your experience of genetics in the context of palliative care clients, or their families?

## Transitional questions

1. In your experience, what have been some of the challenges of discussing genetics with palliative clients or their families? (*prompts: what made this challenging? What were the successes? What made them successes?)*

2. What role do you think palliative care health professionals play in addressing genetics with palliative clients or their families? *(prompts: different roles for nurses/doctors/allied health? Any intervention more appropriate than others? Current role vs. Future role? Could/should it be part of the role? Why/why not?)*
3. How do you think palliative clients and their families should be provided with genetic health information in an ideal world? (*prompts: timing, service, discipline)*
4. What is the role, if any, of larger organisations such as the hospital, professional palliative care organisations or government in helping palliative clients and families access genetic information?

Key questions

1. What are the main barriers and facilitators of discussing genetics with palliative clients and their families? (*prompts: what are the benefits/harms to clients/families? Impact on workforce/workflow?)*
2. What are the facilitators/enablers?
3. If palliative care health professionals were asked by their service to start integrating genetics into their clinical practice, what would be the impact for you as a genetics clinician? (*prompts: how would you feel? What would they need to make this happen? Educational needs? Impact on patients/families? Resource/staffing needs? Guidance?)*
4. Do you think it is appropriate and feasible for palliative care health professionals to integrate genetics with their clients and/or families?

Ending question

1. With all we have talked about today in mind, what is the most important point you'd like me to know about genetics in palliative care?
2. Is there anything you'd like add?

Summary and closing

1. *Facilitator/observer to summarise discussion.* Is this an accurate summary of what we have discussed?
2. *Facilitator to re-state the purpose of focus group discussion.* Is there anything we have missed? Any further thoughts anyone would like to add?

Snowball/recruitment question

1. Are there other individuals or groups who may be interested in participating in this research?
    A. (if participant says yes) you're under no obligation to do so, but if you're willing, would you forward the research invitation to these individuals or groups? Alternatively, if you have permission to share their names and email addresses, i could forward them the research invitation myself.
    B. (if participant says no) no problem, thank you for your time.

Thank everyone for coming. Ask if participant/s would like a summary of results once data analysis complete (explain email address will be retained).

*Please note: If the participant wishes to have a one-on-one interview instead, this schedule will be modified for that purpose.*

## SUPPLEMENTARY FILE C11: VERBAL CONSENT SCRIPT TEMPLATE

**UTS HREC REFERENCE NUMBER ETH20-5046/5347**

| | | | |
|---|---|---|---|
| **Interviewee number:** | | **Date:** | |
| **Interviewer:** | | **Time:** | |

*Key: Underlined areas indicate areas in which the script may differ, depending on whether consent the participant is consenting to a focus group or interview.*

"Thank you for agreeing to speak with me today about genetics in palliative care. The focus group/interview will take approximately 60 minutes. If at any point, you feel that you would rather not go on with the focus group/interview that is fine too".

*[Wait for participant to confirm they are happy to continue, otherwise thank them for their time.]*

"Thank you. Now I just need to confirm some information about you, and I'm going to start recording. This will help us to accurately record the group discussion/your answers to the questions, but all this information will remain completely confidential. Is that OK?"

"First, I need to ask you some questions to confirm that you consent to participating. Remember, even after you've answered these questions, you can withdraw your consent at any time during the interview. However, it may not be possible to withdraw your data from the study results if these have already had your identifying details removed".

The consent questions are: *(request response from each participant if in focus group):*

| **Question** | **Yes** | **No** |
|---|---|---|
| Have you read the information contained in the participant information sheet? | | |
| Have you had an opportunity to ask questions and are you satisfied with the answers you have received? | | |
| Do you understand that there may be risks, such as the inconvenience of providing your time for this research and the potential to feel discomfort by the questions or your/others' responses? | | |
| Do you understand that the research will produce reports, academic work or articles? | | |
| Do you freely agree to participate in this activity, with the understanding that you may withdraw at any time? | | |
| Do you agree to having this interview audio recorded and transcribed? | | |

(If answered NO to any of these – clarify and/or discontinue interview)

"If you have any concerns about the research you can contact myself, Stephanie White, or another member of the research team, Dr Chris Jacobs or Professor Jane Phillips."

"If you would like to talk to someone who is not connected with the research, you may contact the Research Ethics Officer on 02 9514 9772 or Research.ethics@uts.edu.au and quote this number: UTS HREC Approval Number ETH20-5046/5347."

*Record if the participant declines to provide verbal consent:*

*Interview no. _____ read the verbal consent script (or had it read to them) and agreed to participate on date: _____ time: _____ .*

Supplementary file C12: license from publisher to reproduce manuscript (chapter 4b reference)

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Licensee: Springer-Verlag GmbH, DE (the 'Licensee')
Journal Name: Supportive Care in Cancer (the 'Journal')
Manuscript Number: JSCC-D-21-00978R1
Proposed Title of Article: Views and experiences of palliative careclinicians in addressing genetics with individuals and families: a qualitative study (the 'Article')
Author(s) [Please list all named
Authors]: Stephanie White, Stephanie A. White, Jane Phillips, Erin Turbitt, Chris Jacobs (the 'Author')
Corresponding Author Name: Stephanie White

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## SUPPLEMENTARY FILE C13: data collection instrument (palliative care group)

### Palliative Care Clinician Focus Group/Interview Guide

### Welcome

- Welcome participant, introduce self

### Overview

- Thank participant for their participation
- Explain purpose of interview – to hear about views and experiences of integrating genetics into the care of people (and their families) with palliative needs, particularly to understand the barriers, facilitators and possible strategies to overcome these barriers. Describe how results will be used – to develop an evidence base about genetics in palliative care
- Describe what is meant by 'integrating genetics into practice' or 'addressing genetics with patients and families'. Explain I will using this phrasing throughout the interview. What I mean is any action you or a colleague might take at work to address a genetic issue with a client or relative. This may include discussing a genetic concern, taking a family history, organising a referral to a genetics service, storing DNA or ordering genetic testing, or making health recommendations to someone based on their relative's medical problem, for example.
- Explain why they have been invited and selected to participate – because they are a nurse/doctor working in palliative care

### Consent questions

- Refer to verbal consent script template

### Guidelines

- If you haven't already, please complete the demographics survey as soon as possible
- There are no right or wrong answers
- We are audio-recording and may need to allow for audio delays. Provide brief zoom etiquette instructions
- We're on a first name basis
- If possible, please turn off mobile phones or turn to silent. If you must respond to a call, please do so and re-join us as soon as you can.
- Ask participant to introduce themselves, their professional role and how long they have been working in their field

### Opening question

2. Can you please tell me about your experience of genetics in palliative care?

### Transitional questions

5. Generally speaking, what role do you think palliative care health professionals play in addressing genetics with palliative clients or their families? *(prompts: different roles for nurses/doctors/allied health? Any intervention more appropriate than others? Current role vs. Future role? Could/should it be part of the role? Why/why not?)*

6. If you suspected a client had a genetic condition, which might have implications for their relative's health, how would you feel about addressing this? (*prompts: how would you address it? What action would you take? Who would you get involved? What would help you to address this?)*
7. If you were approached by a client, or their relative, with a concern that they had a genetic condition, how would you manage this? *(prompts: would you feel comfortable addressing this? What action would you take?)*
8. In your experience, what have been some of the challenges of addressing genetics with palliative clients or their families? (*prompts: what made this challenging?)*
9. How do you think palliative clients and their families should be provided with genetic health information? (*prompts: timing, service, discipline)*
10. What is the role, if any, of larger organisations such as the hospital, professional palliative care organisations or government in helping palliative clients and families access genetic information?

Key questions

5. What are the main barriers and facilitators of discussing genetics with palliative clients and their families? (*prompts: what are the benefits/harms to clients/families? Impact on workforce/workflow?)*
6. If you were asked by your service to start integrating genetics into your clinical practice, what impact would this have on you? (*prompts: how would you feel? What would you need to make this happen? Educational needs? Resource/staffing needs? Guidance?)*
7. Do you think it is appropriate and feasible for palliative care health professionals to discuss genetics with their clients and/or families?

Ending question

3. With all we have talked about today in mind, what is the most important thing you'd like me to know about genetics in palliative care?

Closing question

3. Is there anything we have missed? Any further thoughts you would like to add?

Snowball/recruitment question

2. Are there other individuals or groups who may be interested in participating in this research?
    A. (if participant says yes) you're under no obligation to do so, but if you're willing, would you forward the research invitation to these individuals or groups? Alternatively, if you have permission to share their names and email addresses, i could forward them the research invitation myself.
    **B.** (if participant says no) no problem, thank you for your time.

Finishing

Thank participant for coming. Ask if participant would like a summary of results once data analysis complete (explain email address will be retained).

# SUPPLEMENTARY FILE C14: COREQ CHECKLIST FOR MANUSCRIPTS IN CHAPTER 4

| Consolidated Criteria for Reporting Qualitative Studies (COREQ): 32-item checklist. Adapted from: Tong, A., P. Sainsbury, and J. Craig, Consolidated criteria for reporting qualitative research (COREQ): A 32-item checklist for interviews and focus groups. *International Journal for Quality in Health Care*, 2007. 19(6): p. 349-57. DOI: 10.1093/intqhc/mzm042 | | | |
|---|---|---|---|
| **No. Item** | **Guide questions/description** | **Chapter 4a reference (genetic health professionals)** | **Chapter 4b reference (palliative care health professionals)** |
| | | **pg. #** | **pg #** |
| **Domain 1: Research team and reflexivity** | | | |
| **Personal Characteristics** | | | |
| 1. Interviewer/ facilitator | Which author/s conducted the interview or focus group? | 55-56 | 75 |
| 2. Credentials | What were the researcher's credentials? E.g. PhD, MD | Not explicit in chapter 4, though this was provided in the title page of submitted manuscript. Available on page v of thesis. | |
| 3. Occupation | What was their occupation at the time of the study? | 56 | 75-76 |
| 4. Gender | Was the researcher male or female? | Not explicit in chapter 4, though this was provided in the title page of submitted manuscript. | 75-76 |
| 5. Experience and training | What experience or training did the researcher have? | 56 | 75-76 |
| **Relationship with participants** | | | |
| 6. Relationship established | Was a relationship established prior to study commencement? | 56 | 75 |
| 7. Participant knowledge of the interviewer | What did the participants know about the researcher? e.g. personal goals, reasons for doing the research | 56 | 75 |
| 8. Interviewer characteristics | What characteristics were reported about the interviewer/facilitator? e.g. Bias, assumptions, reasons and interests in the research topic | 56 | 75-76 |
| **Domain 2: Study design** | | | |
| **Theoretical framework** | | | |
| 9. Methodological orientation and Theory | What methodological orientation was stated to underpin the study? e.g. grounded theory, discourse analysis, ethnography, phenomenology, content analysis | 54-55 | 74 |
| **Participant selection** | | | |
| 10. Sampling | How were participants selected? e.g. purposive, convenience, consecutive, snowball | 55 | 75 |

| | | | |
|---|---|---|---|
| 11. Method of approach | How were participants approached? e.g. face-to-face, telephone, mail, email | 55 | 75 |
| 12. Sample size | How many participants were in the study? | 56-57 | 76 |
| 13. Non-participation | How many people refused to participate or dropped out? Reasons? | 57 | NA |
| **Setting** | | | |
| 14. Setting of data collection | Where was the data collected? e.g. home, clinic, workplace | 55-56 | 75 |
| 15. Presence of non-participants | Was anyone else present besides the participants and researchers? | 55-56 | NA |
| 16. Description of sample | What are the important characteristics of the sample? e.g. demographic data, date | 57-58 | 76 |
| **Data collection** | | | |
| 17. Interview guide | Were questions, prompts, guides provided by the authors? Was it pilot tested? | 55-56 & Appendix C10 | 75 & Appendix C13 |
| 18. Repeat interviews | Were repeat interviews carried out? If yes, how many? | NA | NA |
| 19. Audio/visual recording | Did the research use audio or visual recording to collect the data? | 55-56 | 75 |
| 20. Field notes | Were field notes made during and/or after the interview or focus group? | 55-56 | 75 |
| 21. Duration | What was the duration of the interviews or focus group? | 56-57 | 76 |
| 22. Data saturation | Was data saturation discussed? | 55-56 | 75 |
| 23. Transcripts returned | Were transcripts returned to participants for comment and/or correction? | 55-56 | 75 |
| **Domain 3: Analysis and findings** | | | |
| **Data analysis** | | | |
| 24. Number of data coders | How many data coders coded the data? | 56 | 75 |
| 25. Description of the coding tree | Did authors provide a description of the coding tree? | NA (not congruent with reflexive thematic analysis) | NA (not congruent with reflexive thematic analysis) |
| 26. Derivation of themes | Were themes identified in advance or derived from the data? | 56 | 75 |
| 27. Software | What software, if applicable, was used to manage the data? | 56 | 75 |
| 28. Participant checking | Did participants provide feedback on the findings? | NA (no consent) | |
| **Reporting** | | | |
| 29. Quotations presented | Were participant quotations presented to illustrate the themes / findings? Was each quotation identified? e.g. participant number | 56-65 | 76-84 |
| 30. Data and findings consistent | Was there consistency between the data presented and the findings? | 65-66 | 85-86 |

| | | | |
|---|---|---|---|
| 31. Clarity of major themes | Were major themes clearly presented in the findings? | 57 | 76 |
| 32. Clarity of minor themes | Is there a description of diverse cases or discussion of minor themes? | 56 | 79-80 (variation in views described) |

# Appendix D- supplementary files for scoping review

## CONTENTS

SUPPLEMENTARY FILE D1: PROTOCOL FOR SCOPING REVIEW

# Integrating genetics and genomics in the care of people with palliative needs: A protocol for a scoping review of policy

Part of the GIFT project (Genetic Information for Families of the Terminally Ill)

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**Conflicts of interest** The authors declare no conflicts of interest

**Update information:** Two changes to this protocol were made on the 21st January 2022. These occurred after S.W completed data extraction. The changes relate to the recommendations extracted about 'care of the family'. The research team discussed how to synthesise these recommendations to provide a concise and practical summary to the reader.

The first addition was to create a second review question: *What recommendations in palliative care and genetic/genomic policies regarding care of the family are relevant to the integration of clinical genetic and genomic health information into the care of people with palliative needs, and their families?* The rationale behind adding this question is to examine the 'care of the family' data with a clinical genetics & palliative care lens to generate commonalities between the palliative care and genetic policy guidance, given there had been little common ground identified in the review process to date.

The second change was an additional data synthesis step that would answer the new review question. The research team agreed to group the extracted recommendations related to 'care of the family' into one of three categories: 1) relevant only to palliative care, 2) relevant only to clinical genetics and 3) relevant to both palliative care and genetics. Further detail is provided in 3.8: Data synthesis.

**DEFINITION OF KEY TERMS**

Clinical Genetics: "…a diagnostic service and 'genetic counselling' for individuals or families with, or at risk of, conditions which may have a genetic basis." (1)

Genetic counselling: "…a communication process, which aims to help individuals, couples and families understand and adapt to the medical, psychological, familial and reproductive implications of the genetic contribution to specific health conditions." (2)

Genomics: "A branch of biotechnology concerned with applying the techniques of genetics and molecular biology to the genetic mapping and DNA sequencing of sets of genes or the complete genomes…" (3)

Government: "The body of persons that constitutes the governing authority of a political unit or organization…" (4)

Organisation: "A group of people who work together in an organized way for a shared purpose." (5)

Policy: "…a high-level overall plan embracing the general goals and acceptable procedures especially of a governmental body." (6)

Palliative care: "…person and family-centred care provided for a person with an active, progressive, advanced disease, who has little or no prospect of cure and who is expected to die, and for whom the primary goal is to optimise the quality of life." (7)

Scoping review: "A variation of a systematic review that aims to "map the literature on a particular topic or research area and provide an opportunity to identify key concepts; gaps in the research; and types and sources of evidence to inform practice, policymaking, and research." (8)

Web page: "…a set of data or information which is designed to be viewed as part of a website." (9)

Website: "a collection of publicly accessible, interlinked Web pages that share a single domain name." (10)

**BACKGROUND**

Integrating genetics and genomics into the routine care of people with palliative care needs, may be critical in improving health outcomes for their family members (11). Although genomics and palliative care can, at first glance, feel akin to mixing oil and water, a closer look at these clinical specialties reveals important similarities; both fields aim to optimise life and care for the individual in the context of their family unit (2, 7). Despite these parallels, genomics in usual palliative care practice is underutilized (12). Palliative care nurses and doctors ('palliative care clinicians') report a lack of guidance in applying clinical genetics for the benefit of people with palliative needs, and their families (13). Without clear policy guidance, families will continue to miss the opportunity to obtain valuable genetic information to help them live long and healthy lives. Additionally, clinicians who provide clinical genetics services (such as clinical geneticists and genetic counsellors ['genetics clinicians']) are likely to encounter people with palliative needs, and their families, in their work (14, 15). There are specific considerations when working with these individuals and families, including obtaining informed consent for genetic testing from people who may be in their final days or hours of life, and returning genetic test results to the family after their relative has died (14, 15).

National and international government agencies are recognizing the importance of integrating genomics into routine medical care, but it is not clear how relevant this guidance is to the palliative care context (16-18). There is no clear overlap between policy documents which outline the broad challenges of integrating genetic health information, with the specific considerations for using genetics or genomics in palliative care, including the barriers faced by palliative care and genetics clinicians (19, 20). Emerging evidence reveals the challenges palliative care clinicians face, such as concern of causing additional distress to individuals and families, or detracting attention away from their main goal of providing good palliative care (19, 21, 22). Additional ethical concerns about obtaining a DNA sample from the person with palliative needs for the benefit of their relatives, result in a complex balance of individual and family harms and benefits for the palliative care clinician to navigate (19). High-level policy guidance, in addition to other interventions, should filter directly to healthcare organisations who provide palliative care and clinical genetics services. In turn, front-line health care professionals such as palliative care and genetics clinicians can obtain the support and guidance needed to navigate their responsibilities and provide quality health care (23). At this time, the existence or content of global policy related to the integration of genomics into palliative care is unclear (24).

**OBJECTIVE**

To identify and describe current national and international policy about integrating clinical genetics and genomics into the care of people with palliative needs, and their families.

**METHODS**

A scoping review guided by the Joanna Briggs Institute and reporting items aligned with the PRISMA-ScR extension (25, 26). A scoping review is considered the most appropriate methodology to identify, map and describe the policy environment related to genetics and genomics in palliative care (26).

**Review Questions**

1. What national and international policy guidance is available that describes the integration of clinical genetic and genomic health information into the care of people with palliative needs, and their families?
2. What recommendations in palliative care and genetic/genomic policies regarding care of the family are relevant to the integration of clinical genetic and genomic health information into the care of people with palliative needs, and their families?

**Eligibility Criteria**

Inclusion and exclusion criteria developed using the Population, Concept, Context (PCC) framework (**Error! Reference source not found.**). To identify and map policy relevant to the reviewers context, this review will only include policies from the top 20 countries listed in the Economist Intelligence Unit Quality of Death Index (27).

**Information Sources and Search Strategy**

We will identify eligible policies through three main information sources:

- Database search
- Web search
- Emails to key informants

The database and web search will be co-designed with an information scientist experienced in health database searching and peer-reviewed, to ensure inclusion of appropriate terms and no important terms are omitted (28). Searching will be supplemented by hand-searching key government and organisational websites suggested by key informants or revealed by the database or web search. Forward-chaining, using Web of Science, and back-chaining, by scanning reference lists of eligible articles, will be conducted (29).

**Database Search**

The reviewers will search Medline, EMBASE and CINAHL by combining Medical Subject Heading (MeSH) terms and keywords related to policy, palliative care, clinical genetics and genetic counselling with Boolean operators (Appendix B). Test searches supervised by the information scientist yielded a large number of results. To balance maintaining a comprehensive search, yielding relevant policies and managing large numbers, a pragmatic decision was made to limit the search by publication date (2010 – current) and to English language results (30). The reviewers will modify the Medline search strategy for the other databases and incorporate alternative terms identified in other databases into the Medline search strategy.

**Web Search**

Web searching enables inclusion of 'grey literature' which resides outside of traditional academic databases (31). The Cochrane handbook recommends including grey literature to augment and improve review findings (32). As there is no 'gold standard' for web searching, the strategy was informed through consultation with an information scientist, reviewing articles that utilise a web search and peer-review (28, 33-35).

The web search will consist of two stages (Appendix C).

- Firstly, the reviewers will interrogate Google (www.google.com) by combining keywords related to policy, palliative care, clinical genetics and genetic counselling

with Boolean operators. Guidance for deciding how many web results to review is lacking, therefore, a pragmatic decision to review the first ten pages of results only (equalling 100 results per Google search) was made through consultation with the study team and reviewing articles which utilised web searching (33, 34).

- Secondly, we will utilise the Canadian Agency for Drugs and Technologies in Health (CADTH) "Grey Matters: a practical tool for searching health-related grey literature" to conduct systematic hand-searching of eligible national government and organisational websites (36).

**Emails to Key Informants**

The review team will email key palliative care, clinical genetics and genetic counselling informants from eligible countries. The purpose of these questions is to identify policy documents missed by the search or in development. Key palliative care informants from eligible countries will be identified through the contact index list in the EIU QOD Index (27). We will identify key clinical genetics and genetic counselling informants through the Transnational Alliance of Genetic Counselling contact list, consultation with experts, organisations identified in this review and targeted web searching (33, 34, 37).

The email will include a brief introduction and the review objective, followed by three questions:

1. Could you please list or link any high-level policy (including strategy, vision, framework, standards) written by national government or organisations that pertain to the provision or delivery of palliative care/clinical genetics/genetic counselling services?
2. Are you aware of any policy regarding the provision or delivery of clinical genetics or genomics to people with palliative care needs (or their families)?
3. Is there another key informant who works in this area that I should contact?

**RECORD MANAGEMENT**

All working files will be stored on the UTS-approved Microsoft OneDrive and backed up on an external hard drive.

We will export the records yielded from Medline, EMBASE and CINAHL .ris format, upload to Endnote software and de-duplicate using the Bramer method (38, 39). Unique records will be exported from Endnote in .xml format and uploaded to Covidence for record selection (40).

The reviewers will document records from the web search in a Microsoft Excel 2016 spreadsheet, including the date of the search, search terms, number of records, title of each individual record and URL address. We will download and save documents (.pdf or .docx) arising from the web search in a dedicated folder that corresponds with the search from which it was located.

The reviewers will save emails to and from key informants as .pdf files in a dedicated folder and utilise a Microsoft Excel 2016 spreadsheet to document the date of contact/s, responses to questions and details of suggested key informants to contact.

**RECORD SELECTION**

**Piloting and Inter-Rater Reliability**

Twenty-five records will be randomly selected for two reviewers to pilot the eligibility criteria, with ≥75% agreement considered adequate to commence screening (41). Alterations, additions or removal of eligibility criteria will be made as required (32).

Two reviewers will screen 20% of records at each stage of record selection. The first stage will be title and abstract or 'first-pass' and the second stage will be full text screening or 'second-pass' (explained further below)(32). When the two reviewers obtain adequate agreement using a Cohen's kappa statistic (adequate agreement = ≥0.7), one reviewer will complete the remaining record selection (42). If discrepancies are unable to be resolved through discussion between two reviewers, a third reviewer will be asked to decide (32). If there is missing information affecting the ability to assess eligibility, the reviewers will contact the author to a maximum of three email attempts.

**Title and Abstract Screening/First-Pass Screening**

We will perform title and abstract screening on all database records, which involves reading the document title and abstract to determine whether it meets eligibility criteria. For the web and hand-search, we will perform 'first-pass' screening, which involves reading the title and web page. A record will progress to full-text/second-pass screening if it meets inclusion criteria, has missing or ambiguous information, or cannot be excluded based on title and abstract, or first-pass screening.

**Full Text Screening/Second-Pass Screening**

Database records that progress to full text screening will be read in full against eligibility criteria. For the web search records, the web page and web site (which the web page belongs to) will be explored in full for eligible documents. Reviewers will record reasons for exclusion recorded in Covidence, Endnote or Microsoft Excel. For ease of comparison, the reviewers will exclude ineligible records in a hierarchical way:

a) Wrong population
b) Wrong context
c) Wrong concept

**CRITICAL APPRAISAL**

The purpose of a scoping review is to identify and map evidence, rather than critique evidence quality (25). Furthermore, we have not identified a validated appraisal tool for policy documents. Therefore, the reviewers will not conduct a formal critical appraisal of individual documents. However, the eligibility criteria (**Error! Reference source not found.**) specifies that policies must based on empirical evidence and include an explanation of the method by which they were written, which aims to filter out policies of low quality in the record selection process (43).

**DATA ITEMS AND CHARTING**

A standardised data extraction instrument from the Joanna Briggs Institute (JBI; Template Source of Evidence Details, Characteristics and Results Extraction Instrument) will be utilised (44). Data items will include: citation details; publishing organisation; country; area of speciality (eg. palliative care or clinical genetics); presence or absence of information related to genetics/genomics in palliative care; content of information related to genetics/genomics in palliative care (if present); key strategies to promote integration of genetics and genomics into

routine medical care; presence or absence of information related to care of family; content of information related to care of family (if present; Appendix 4).

At the conclusion of full text screening, two reviewers will pilot the JBI data extraction instrument by randomly selecting and extracting data from 10 documents (41). Adjustments to the instrument will be made as necessary, with revisions outlined and justified in the final review (25, 32).

**DATA SYNTHESIS**

The reviewers will group policies by country, to provide a global overview of policy guidance related to genetics and genomics in palliative care, and by speciality, to determine the number of palliative care or genetics organisations that address genetics and genomics in palliative care. The data will be presented in tabular form to provide a clear mapping of the presence or absence and content of genetics integration into palliative care (25). The outcomes will also be narratively synthesised, as described by Popay, Roberts (45). This involves four non-linear steps: 1) theory generation, 2) developing a preliminary synthesis, 3) exploring relationships between the data and 4) assessing the robustness of the synthesis.

To explore commonalities between palliative care and genetic/genomic policies, we synthesised the recommendations about 'care of the family' by categorising them into one of three groups: 1) relevant only to palliative care, 2) relevant only to clinical genetics and 3) relevant to both palliative care and genetics. A palliative care expert (C.V) with experience in systematic reviews and environmental scanning independently co-categorised the recommendations with S.W.

**ETHICS**

This study does not require formal ethical approval, as per consultation with the UTS Research Ethics office. However, the reviewers are mindful of contacting key informants as part of the search strategy. If key informants do not respond to the reviewer's email, this will not adversely affect their relationship with the study team or the University of Technology Sydney. If the key informant has not responded after three email attempts, the reviewers will not contact the informant again. The reviewers will not use direct quotes of the key informants in any output from this review. If any ethical issues arise during the course of this study, the reviewers will discuss this with the review team and if necessary, consult the UTS Research Ethics office.

**PUBLICATION PLAN**

We will submit the final review manuscript to a leading, peer-reviewed palliative medicine journal.

Journal name: Palliative Medicine

Impact factor: 4.956

Word limit: 5,000

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# SUPPLEMENTARY FILE D2: PRISMA SCOPING REVIEW EXTENSION CHECKLIST

| SECTION | ITEM | PRISMA-ScR CHECKLIST ITEM | REPORTED ON PAGE # |
|---|---|---|---|
| **TITLE** | | | |
| Title | 1 | Identify the report as a scoping review. | 88 |
| **ABSTRACT** | | | |
| Structured summary | 2 | Provide a structured summary that includes (as applicable): background, objectives, eligibility criteria, sources of evidence, charting methods, results, and conclusions that relate to the review questions and objectives. | 88-89 |
| **INTRODUCTION** | | | |
| Rationale | 3 | Describe the rationale for the review in the context of what is already known. Explain why the review questions/objectives lend themselves to a scoping review approach. | 89-91 |
| Objectives | 4 | Provide an explicit statement of the questions and objectives being addressed with reference to their key elements (e.g., population or participants, concepts, and context) or other relevant key elements used to conceptualize the review questions and/or objectives. | 91 |
| **METHODS** | | | |
| Protocol and registration | 5 | Indicate whether a review protocol exists; state if and where it can be accessed (e.g., a Web address); and if available, provide registration information, including the registration number. | 91 |
| Eligibility criteria | 6 | Specify characteristics of the sources of evidence used as eligibility criteria (e.g., years considered, language, and publication status), and provide a rationale. | 91-92 & Appendix C3 |
| Information sources* | 7 | Describe all information sources in the search (e.g., databases with dates of coverage and contact with authors to identify additional sources), as well as the date the most recent search was executed. | 92-93 |
| Search | 8 | Present the full electronic search strategy for at least 1 database, including any limits used, such that it could be repeated. | Appendix C4 |
| Selection of sources of evidence† | 9 | State the process for selecting sources of evidence (i.e., screening and eligibility) included in the scoping review. | 93 |
| Data charting process‡ | 10 | Describe the methods of charting data from the included sources of evidence (e.g., calibrated forms or forms that have been tested by the team before their use, and whether data charting was done independently or in duplicate) and any processes for obtaining and confirming data from investigators. | 93 |
| Data items | 11 | List and define all variables for which data were sought and any assumptions and simplifications made. | 93 & Appendix C5 |

| SECTION | ITEM | PRISMA-ScR CHECKLIST ITEM | REPORTED ON PAGE # |
|---|---|---|---|
| Critical appraisal of individual sources of evidence§ | 12 | If done, provide a rationale for conducting a critical appraisal of included sources of evidence; describe the methods used and how this information was used in any data synthesis (if appropriate). | 94 |
| Synthesis of results | 13 | Describe the methods of handling and summarizing the data that were charted. | 94 |
| **RESULTS** | | | |
| Selection of sources of evidence | 14 | Give numbers of sources of evidence screened, assessed for eligibility, and included in the review, with reasons for exclusions at each stage, ideally using a flow diagram. | Figure 9. PRISMA flow diagram, pg 95 |
| Characteristics of sources of evidence | 15 | For each source of evidence, present characteristics for which data were charted and provide the citations. | Table 10, pg 97-101 & Appendix C6 |
| Critical appraisal within sources of evidence | 16 | If done, present data on critical appraisal of included sources of evidence (see item 12). | Not applicable |
| Results of individual sources of evidence | 17 | For each included source of evidence, present the relevant data that were charted that relate to the review questions and objectives. | Table 10, pg 97-101 & Appendix C6 |
| Synthesis of results | 18 | Summarize and/or present the charting results as they relate to the review questions and objectives. | 95-104 |
| **DISCUSSION** | | | |
| Summary of evidence | 19 | Summarize the main results (including an overview of concepts, themes, and types of evidence available), link to the review questions and objectives, and consider the relevance to key groups. | 105-106 |
| Limitations | 20 | Discuss the limitations of the scoping review process. | 106-107 |
| Conclusions | 21 | Provide a general interpretation of the results with respect to the review questions and objectives, as well as potential implications and/or next steps. | 107 |
| **FUNDING** | | | |
| Funding | 22 | Describe sources of funding for the included sources of evidence, as well as sources of funding for the scoping review. Describe the role of the funders of the scoping review. | Not described in thesis. Statement available in open access manuscript |

# SUPPLEMENTARY FILE D3: SCOPING REVIEW ELIGIBILITY CRITERIA

| | 1. Inclusion | 2. Exclusion |
|---|---|---|
| **1. Population** | 1.1.1 Policies written by international, national and state-based professional palliative care, clinical genetics or genetic counselling organisations<br><br>1.1.2 Policies written by international, national and state-based government agencies (including agencies that are funded by national or state governments to produce policy/guidelines eg. EviQ, NICE) | 2.1.1 Policies written by organisations that are not concerned with palliative care, clinical genetics or genetic counselling<br><br>2.1.2 Policies written by organisations, author/s or government agencies which do not have an international, national or state focus or jurisdiction (eg. local health districts or single institutions)<br><br>2.1.3 Policies written by patient, family or consumer organisations/support groups<br><br>2.1.4 Policies written by organisations that have a disease specific focus (eg. European Cystic Fibrosis Society) |
| **2. Concept** | 1.2.1 Policy documents, which may be described as a policy, framework, strategy, vision, standard, guideline or other<br><br>1.2.2 Documents which provide international, national or state guidance about provision of palliative care, clinical genetics or genetic counselling services, where at least 50% of the document is related to palliative care or genetics<br><br>1.2.3 Document is based on empirical evidence, as evidenced by a statement from the authors or references to evidence within the policy<br><br>1.2.4 The authors describe the method by which the policy was developed<br><br>1.2.5 No evidence of a more recent version of the publication<br><br>1.2.6 Written in the English language<br><br>1.2.7 Publication date after 2010<br><br>**OR**<br><br>1.2.8 Review date within the last ten years (2010 - current) | 2.2.1 Documents which do not provide policy guidance about palliative care, clinical genetics or genetic counselling services, where less than 50% of the policy is related to palliative care or genetics<br><br>2.2.2 The policy is not based on empirical evidence, evidenced by no statement from the authors or no references to evidence within the policy<br><br>2.2.3 There is no description in the policy of the method by which it was developed<br><br>2.2.4 Policies which are identified in the database search but are not present on the publishing organisation's website<br><br>2.2.5 Policy still in development or not yet published<br><br>2.2.6 Policy has been rescinded or superseded by an updated version<br><br>2.2.7 Publication date **AND** review date not prior to 2010<br><br>2.2.8 Written in a language other than English<br><br>2.2.9 The policy is a duplicate of a document which has already been included<br><br>2.2.10 The policy is about laboratory or research practices, pharmacogenetics, specific treatment practices (eg. pain management, radiotherapy, acupuncture) or a single disease process |

| | | |
|---|---|---|
| | | 2.2.11 The policy is about spiritual care<br><br>2.2.12 The policy relates to prenatal care<br><br>2.2.13 The policy has a disease specific focus (eg. Guidelines related to genetic testing for a single condition)<br><br>2.2.14 The policy relates to bereavement care only |
| **3. Context** | 1.3.1 Policies from the top 20 countries listed in the Economist Intelligence Unit Quality of Death Index (see Appendix A) | 2.3.1 Policy documents from other countries, that are not the top 20 countries listed in the Economist Intelligence Unit Quality of Death Index (see Appendix A) |

# SUPPLEMENTARY FILE D4: SEARCH STRATEGIES FOR MEDLINE DATABASE AND WEB-SEARCH

| **Medline search** | | |
|---|---|---|
| **PALLIATIVE CARE POLICY SEARCH** | | |
| Search # | MeSH terms and keywords | # of records |
| 1 | Practice Guideline/ or Guideline/ or Practice Guidelines as Topic/ or Societies, Medical/ or Health Policy or Quality of Health Care/ | 361157 |
| 2 | Palliative Care/ or "Hospice and Palliative Care Nursing"/ or Palliative Medicine/ or Hospice Care/ or Terminal Care/ or Terminally Ill/ | 89491 |
| 3 | (guideline* or standard* or polic* or consensus or strategy or vision or framework).ti | 409191 |
| 1,2 & 3 | *Combined searches 1, 2 & 3 with Boolean operator:* "AND" | 893 |
| 1,2 & 3 with limits | *Applied the following limits to the combined search: "*2010-current", "English language" | 447 |
| 1,2 & 3 with limits | *Applied the following limits to the combined search when re-ran on 21st February 2022:* "2020 – current", "English language" | 57 |
| **GENETIC/GENOMIC POLICY SEARCH** | | |
| Search # | MeSH terms and keywords | # of records |
| 1 | Genetic Counseling/ or genetic counsel?ing.mp. or Genetic Testing/ or Genetics, Medical/ or DNA banking.mp. or DNA storage.mp | 76747 |
| 2 | Practice Guideline/ or Guideline/ or Practice Guidelines as Topic/ or Societies, Medical/ or Health Policy or Quality of Health Care/ | 361157 |
| 3 | (guideline* or standard* or polic* or consensus or strategy or vision or framework).ti | 409191 |
| 1,2 & 3 | *Combined searches 1, 2 & 3 with Boolean operator:* "AND" | 656 |
| 1,2 & 3 with limits | *Applied the following limits to the combined search: "*2010-current", "English language" | 382 |
| 1,2 & 3 with limits | *Applied the following limits to the combined search when re-ran on 21st February 2022:* "2020 – current", "English language" | 55 |

| Google search | | |
|---|---|---|
| **Search #** | **Search terms** | **# of records (first ten pages reviewed only, equalling 100 results per search)** |
| **PALLIATIVE CARE POLICY SEARCH** | | |
| 1 | (“Palliative Care” OR “Palliative Medicine” OR “Hospice Care” OR “Terminal Care” OR “Terminally Ill”) AND (“Practice Guideline” OR “Guideline” OR “Health Policy” OR "Standard" OR "Consensus" OR "Strategy" OR "Vision" OR "Framework") AND (Government OR “Government Agency” OR “Medical Society”) | 16000000 |
| 2 | (“Palliative Care” OR “Palliative Medicine” OR “Hospice Care” OR “Terminal Care” OR “Terminally Ill”) AND (“Practice Guideline” OR “Guideline” OR “Health Policy” OR "Standard" OR "Consensus" OR "Strategy" OR "Vision" OR "Framework") AND (Government OR “Government Agency” OR “Medical Society”)(type:.pdf) | 21590000 |
| **GENETIC/GENOMIC POLICY SEARCH** | | |
| 3 | (“Genetic Counseling” OR “Genetic Counselling” OR “Genetic Screening” OR “Genetic Testing” OR “Medical Genetics” OR “Clinical Genetics” OR “DNA banking” OR “DNA storage”) AND (“Practice Guideline” OR “Guideline” OR “Health Policy” OR "Standard" OR "Consensus" OR "Strategy" OR "Vision" OR "Framework") AND (Government OR “Government Agency” OR “Medical Society”) | 37200000 |
| 4 | (“Genetic Counseling” OR “Genetic Counselling” OR “Genetic Screening” OR “Genetic Testing” OR “Medical Genetics” OR “Clinical Genetics” OR “DNA banking” OR “DNA storage”) AND (“Practice Guideline” OR “Guideline” OR “Health Policy” OR "Standard" OR "Consensus" OR "Strategy" OR "Vision" OR "Framework") AND (Government OR “Government Agency” OR “Medical Society”)(type:.PDF) | 399000 |

# SUPPLEMENTARY FILE D5: DATA EXTRACTION TOOL

| Example of data extraction sheet version 3 (18.11.2020) to illustrate the predetermined data items. Modified Joanna Briggs Institute extraction tool from evidence synthesis manual |
|---|
| **SECTION 1: COMPLETE FOR ALL POLICIES** |
| **Data extraction item** |
| Date |
| Reviewer |
| Document source |
| Title |
| Year |
| Year planned for review |
| Country |
| International, national or state? |
| Publishing organisation |
| Palliative care or genetics/genomics document? |
| Adult, paediatric or both |
| **SECTION 2: COMPLETE FOR ALL PALLIATIVE CARE POLICIES** |
| 1. Is genetics or genomics mentioned in the background information? (Yes/no) |
| 1a. Content of background genetic information (Include heading and page number copy & paste) |
| **SECTION 3: COMPLETE FOR ALL GENETICS/GENOMICS POLICIES** |
| 2. Is palliative care mentioned in the background information? (Yes/no) |
| 2a. Content of background palliative care information (Include heading and page number; copy & paste) |
| 3. Does the document address key strategies to promote integration of genetics and genomics into routine medical care? (Yes/No) |
| **SECTION 4: COMPLETE FOR ALL POLICIES** |
| 4. Does the document address the integration of genetics/genomics into palliative care as part of the strategy/recommendations/policy direction? (Yes/No) |
| 4a. If yes, what is the content? (Include heading and page number; copy & paste) |
| 5. Is care of the family mentioned in the background information? (Yes/No) |
| 5a. If yes, content of background family information (Include heading and page number; copy & paste) |
| 6. Is care of the family part of the document's strategy/ recommendations/policy direction? (Yes/No) |
| 6a. Content of family strategy/recommendations/policy directions (Include heading and page number; copy & paste) |

# SUPPLEMENTARY FILE D6: FULL CITATIONS OF ALL POLICY DOCUMENTS

| EIU QOD: Economist Intelligence Unit Quality of Death | | |
|---|---|---|
| **EIU QOD index rank** | **Region** | **Full citation** |
| NA | Europe | Fellmann, F., van el, C. G., Charron, P., Michaud, K., Howard, H. C., Boers, S. N., . . . Association for European Cardiovascular Pathology. (2019). European recommendations integrating genetic testing into multidisciplinary management of sudden cardiac death. *European Journal of Human Genetics* 27(12), 1763-1773. doi:10.1038/s41431-019-0445-y |
| | | Oliver, D., Borasio, G. D., Caraceni, A., De Visser, M., Grisold, W., Lorenzl, S., . . . Voltz, R. (2016). A consensus review on the development of palliative care for patients with chronic and progressive neurological disease. *European Journal of Neurology*, 23(1), 30-38. doi:http://dx.doi.org/10.1111/ene.12889 |
| | | Tuffrey-Wijne, I., McLaughlin, D., Curfs, L., Dusart, A., Hoenger, C., McEnhill, L., . . . Oliver, D. (2015). Defining consensus norms for palliative care of people with intellectual disabilities in Europe, using Delphi methods: A White Paper from the European Association of Palliative Care. *Palliative Medicine*, 30(5), 446-455. doi:http://dx.doi.org/10.1177/0269216315600993 |
| | | van der Steen, J., Radbruch, L., Hertogh, C. M. P. M., de Boer, M. E., Hughes, J. C., Larkin, P., . . . on behalf of the European Association of Palliative Care. (2014). White paper defining optimal palliative care in older people with dementia: A Delphi study and recommendations from the European Association for Palliative Care. *Palliative Medicine*, 28(3), 197-209. doi: 10.1177/0269216313493685 |
| | | Van El, C. G., Cornel, M. C., Borry, P., Hastings, R. J., Fellmann, F., Hodgson, S. V., . . . on behalf of the ESHG Public and Professional Policy Committee. (2013). Whole-genome sequencing in health care. *European Journal of Human Genetics*, 21(6), 580-584. doi:http://dx.doi.org/10.1038/ejhg.2013.46 |
| | | Van El, C. G., & Cornel, M. C. (2011). Genetic testing and common disorders in a public health framework: Recommendations of the European Society of Human Genetics. *European Journal of Human Genetics*, 19(4), 377-381. doi:10.1038/ejhg.2010.176 |
| NA | Global | World Health Organization. *Integrating palliative care and symptom relief into primary health care: a WHO guide for planners, implementers and managers*. Geneva, Switzerland: World Health Organization; 2018 [Available from: https://apps.who.int/iris/bitstream/handle/10665/274559/9789241514477-eng.pdf?ua=1.] |
| | | World Health Organization. *Integrating palliative care and symptom relief into paediatrics: A WHO guide for health planners, implementers and managers*. Geneva, Switzerland: World Health Organization; 2018 [Available from: https://apps.who.int/iris/bitstream/handle/10665/274561/9789241514453-eng.pdf?ua=1.] |
| | | Parikh, S., Goldstein, A., Karaa, A., Koenig, M. K., Anselm, I., Brunel-Guitton, C., . . . Chinnery, P. F. (2017). Patient care standards for primary mitochondrial disease: A consensus statement from the mitochondrial medicine society. *Genetics in Medicine*, 19(12), 1-18. doi:http://dx.doi.org/10.1038/gim.2017.107] |
| 1 | United Kingdom | Her Majesty's Government. *Genome UK: The future of healthcare.* United Kingdom: Department of Health and Social Care, Department for Business, Energy & Industrial Strategy, Office for Life Sciences, and Lord Bethell of Romford; 2020 [Available from: https://assets.publishing.service.gov.uk/government/uploads/system/uploads/attachment_data/file/920378/Genome_UK_-_the_future_of_healthcare.pdf.] |
| | | National Institute for Health and Care Excellence. *End of life care for adults: service delivery.* London, United Kingdom: NICE; 2019 [Available from: https://www.nice.org.uk/guidance/qs13/resources/end-of-life-care-for-adults-pdf-2098483631557.] |
| | | Hospice UK. *Transforming hospice care: A five-year strategy for the hospice movement 2017 to 2022*. London, UK: Hospice UK; 2017 [Available from: https://www.hospiceuk.org/docs/default-source/about-us-documents-and-files/hospice-uk-strategy-2017-2022.pdf?sfvrsn=6.] |
| | | National Institute for Health and Care Excellence. *End of life care for infants, children and young people.* London, United Kingdom: NICE; 2017 [Available from: https://www.nice.org.uk/guidance/qs160/resources/end-of-life-care-for-infants-children-and-young-people-pdf-75545593722565.] |

| | | |
|---|---|---|
| | | National Institute for Health and Care Excellence. *End of life care for infants, children and young people with life-limiting conditions: planning and management.* London, United Kingdom: NICE; 2016 [Available from: https://www.nice.org.uk/guidance/ng61/resources/end-of-life-care-for-infants-children-and-young-people-with-lifelimiting-conditions-planning-and-management-pdf-1837568722885.] |
| | | Leadership Alliance for the Care of Dying People*. One chance to get it right.* London: UK Government; 2014 [Available from: https://assets.publishing.service.gov.uk/government/uploads/system/uploads/attachment_data/file/323188/One_chance_to_get_it_right.pdf.] |
| | | National Institute for Clinical Excellence. *End of life care for adults*. London, United Kingdom: NICE; 2011 [Available from: https://www.nice.org.uk/guidance/qs13/resources/end-of-life-care-for-adults-pdf-2098483631557.] |
| *1* | *England* | National Palliative and End of Life Care Partnership. *Ambitions for Palliative and End of Life Care: A national framework for local action 2021-2026.* London, UK: NHS England; 2021 [Available from: https://acpopc.csp.org.uk/system/files/documents/2021-05/FINAL_Ambitions-for-Palliative-and-End-of-Life-Care_2nd_edition.pdf.] |
| | | Palliative Care for People with Learning Disabilities. *Delivering high quality end of life care for people who have a learning disability*. London, UK: NHS England,; 2017 [Available from: https://www.england.nhs.uk/wp-content/uploads/2017/08/delivering-end-of-life-care-for-people-with-learning-disability.pdf.] |
| | | Department of Health. *Our Commitment to you for end of life care: The Government Response to the Review of Choice in End of Life Care*. London, UK: NHS Finance and Operations,; 2016 [Available from: https://assets.publishing.service.gov.uk/government/uploads/system/uploads/attachment_data/file/536326/choice-response.pdf.] |
| | | National End of Life Care Programme*. End of life care in long term neurological conditions a framework for implementation*. London, UK: NHS England; 2011 [Available from: https://www.nai.ie/assets/98/E29C88A6-9CA5-06B3-E74D285E3C0695A2_document/End_20life_20care_20long_20term_20neuro_20conditions.pdf.] |
| *1* | *Wales* | Welsh Government. *Genomics for precision medicine strategy*. Wales, UK: Welsh Government; 2017 [Available from: https://gov.wales/sites/default/files/publications/2019-04/genomics-for-precision-medicine-strategy.pdf.] |
| *1* | *Scotland* | Dumfries and Galloway Integration Joint Board*. A Plan for Palliative Care*. Dumfries, Scotland: Dumfries and Galloway Health and Social Care; 2020 [Available from: https://dghscp.co.uk/wp-content/uploads/2020/09/Agenda-Item-8-Appendix-1-Final-Draft-Plan-for-Palliative-Care-1.pdf.] |
| | | Glasgow City Health and Social Care Partnership. *Palliative & End of Life Care Plan 2018-2023.* Glasgow, Scotland: Glasgow City HSCP; 2018 [Available from: https://www.yoursupportglasgow.org/media/23104/glasgow-hscp-palliative-care-plan-2018-23.pdf.] |
| *1* | *Northern Ireland* | Department of Health Social Services and Public Safety. *Living Matters Dying Matters A Palliative and End of Life Care Strategy for Adults in Northern Ireland*. Northern Ireland, UK: Department of Health Social Services and Public Safety; 2010 [Available from: https://www.health-ni.gov.uk/sites/default/files/publications/dhssps/living-matters-dying-matters-strategy-2010.pdf.] |
| 2 | Australia | South Australian Health. *South Australia's Palliative Care Strategic Framework.* Adelaide, SA, Australia: Government of South Australia; 2021 [Available from: https://www.sahealth.sa.gov.au/wps/wcm/connect/54c48d50-0124-471a-a1fb-ba7daa0e0c79/21064.3+Palliative+Care+Strategic+Framework-2022-2027-WEB.pdf?MOD=AJPERES&CACHEID=ROOTWORKSPACE-54c48d50-0124-471a-a1fb-ba7daa0e0c79-nSjyFKq. |
| | | WA Department of Health. *Western Australian Paediatric Strategy for End-of-Life and Palliative Care 2021-2028.* Perth, WA, Australia: Government of Western Australia; 2021 [Available from: https://ww2.health.wa.gov.au/~/media/Corp/Documents/Health-for/End-of-Life/Paediatric-Strategy-for-EoLPC.pdf?mc_cid=45a56d34f1&mc_eid=6bf2e9428b.] |
| | | NSW Health. *End of Life and Palliative Care Framework 2019-2024.* Sydney, NSW, Australia: NSW Government; 2019 [Available from: https://www.health.nsw.gov.au/palliativecare/Publications/eol-pc-framework.pdf. |
| | | Department of Health. *National Palliative Care Strategy.* 2018 Canberra, ACT, Australia: Australian Government; 2018 [Available from: https://www.health.gov.au/resources/publications/the-national-palliative-care-strategy-2018. |
| | | Palliative Care Australia. *Palliative Care Service Development Guidelines.* Griffith, ACT, Australia: PCA; 2018 [Available from: https://palliativecare.org.au/wp-content/uploads/dlm_uploads/2018/02/PalliativeCare-Service-Delivery-2018_web-1.pdf.] |

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|---|---|---|
| | | Palliative Care Australia. *National Palliative Care Standards.* Griffith, ACT, Australia: PCA; 2018 [Available from: https://palliativecare.org.au/publication/standards/.] |
| | | WA Department of Health. *WA End-of-Life and Palliative Care Strategy 2018–2028.* Perth, WA, Australia: Government of Western Australia; 2018 [Available from: https://ww2.health.wa.gov.au/~/media/Files/Corporate/general-documents/Health-Networks/Palliative-care/WA-End-of-life-and-Palliative-Care-Strategy-2018-2028.pdf.] |
| | | Department of Health and Human Services. *Compassionate communities: A Tasmanian Palliative Care Policy Framework 2017 – 2021.* Hobart, Tasmania, Australia: Tasmanian Government; 2017 [Available from: https://www.health.tas.gov.au/__data/assets/pdf_file/0006/249333/Tasmanian_Palliative_Care_Policy_Framework_v16.pdf.] |
| | | Department of Health. *National Health Genomics Policy Framework 2018 – 2021.* Canberra, ACT, Australia: Australian Government; 2017 [Available from: https://www.health.gov.au/resources/publications/national-health-genomics-policy-framework-2018-2021.] |
| | | Department of Health and Human Services. *Genetic and genomic healthcare for Victoria 2021: Improving the health and wellbeing of Victorians.* Melbourne, Victoria, Australia: Victorian Government; 2017 [Available from: https://www.health.vic.gov.au/sites/default/files/migrated/files/collections/policies-and-guidelines/g/genetic-and-genomic-healthcare-in-victoria-2021.pdf.] |
| | | NSW Health. *Genomics Strategy.* Sydney, NSW, Australia: NSW Government; 2017 [Available from: https://www.health.nsw.gov.au/services/Publications/nsw-health-genomics-strategy.pdf. |
| | | Australian Commission on Safety and Quality in Health Care. *National consensus statement: essential elements for safe and high-quality paediatric end-of-life care.* Sydney, NSW, Australia: ACSWHC; 2016 [Available from: https://www.safetyandquality.gov.au/publications-and-resources/resource-library/national-consensus-statement-essential-elements-safe-and-high-quality-paediatric-end-life-care. |
| | | Department of Health and Human Services. *Victoria's end of life and palliative care framework: A guide for high-quality end of life care for all Victorians.* Melbourne, Victoria, Australia: Victorian Government 2016 [Available from: https://www.health.vic.gov.au/sites/default/files/migrated/files/collections/policies-and-guidelines/v/victorias-end-of-life-and-palliative-care-framework.pdf.] |
| | | Australian Commission on Safety and Quality in Health Care. *National consensus statement: essential elements for safe and high-quality end-of-life care.* Sydney, NSW, Australia: ACSWHC; 2015 [Available from: https://www.safetyandquality.gov.au/publications-and-resources/resource-library/national-consensus-statement-essential-elements-safe-and-high-quality-end-life-care.] |
| | | QLD Health. *Statewide strategy for end-of-life care.* Brisbane, QLD, Australia: Queensland Government; 2015 [Available from: https://www.health.qld.gov.au/__data/assets/pdf_file/0022/441616/end-of-life-strategy-full.pdf.] |
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# SUPPLEMENTARY FILE D7: EXTRACTS OF RELEVANT RECOMMENDATIONS AND BACKGROUND INFORMATION

| NB. Table contains excerpts of data extracted from policies that included recommendations about the integration of genetics and genomics into palliative care, and excerpts that highlight the genetic and genomic information extracted from background sections in palliative care policies. | | | | |
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| **REGION** | **AUTHOR** | **YEAR** | **TITLE** | **EXCERPTS OF CONTENT RELATED TO GENETICS IN PALLIATIVE CARE** |
| **POLICIES THAT ADDRESS THE INTEGRATION OF GENETIC/GENOMIC INFORMATION INTO PALLIATIVE CARE** | | | | |
| Global | Mitochondrial Medicine Society | 2017 | Patient care standards for primary mitochondrial disease: a consensus statement from the Mitochondrial Medicine Society | Table 2 (page 5)<br>Other specialist consultations to consider at time of diagnosis and at 1–2 year intervals as needed based on symptoms: Palliative care<br>Gastroenterology – Recommendations (page 10)<br>Decisions regarding gastrostomy or jejunostomy tube insertion should be made in close consultation with a gastroenterologist and, in some cases, palliative care<br>Nephrology – Recommendations (page 11)<br>Dialysis for end-stage renal disease should be considered palliative, and patients receiving dialysis should ideally also be considered for renal transplantation |
| England | National End of Life Care Programme | 2011 | End of life care in long term neurological conditions a framework for implementation | Holistic care - psychosocial and spiritual aspects (page 24)<br>Psychosocial care: As someone faces the diagnosis and then the progression of a life limiting condition there will be many emotional and psychological issues that come to the fore, including: Fear of the disorder. It may be unknown to the person and their family, or there may be a family history of the disease, with memories of these experiences. Either will be a frightening prospect Concerns as to the possibility of children being affected by the disease. This may be a specific concern for some – with Huntington's disease there is a risk because it is genetic - and for others when the genetic basis of the disease may be less clear or not even an issue at all<br>Multiple losses for both the person and their family involving many of the items listed above. This, and the changing family roles involved, may be profound |
| **PALLIATIVE CARE POLICIES THAT DESCRIBE GENETICS/GENOMICS IN BACKGROUND INFORMATION** | | | | |
| Australia | WA Department of Health | 2021 | Western Australian Paediatric Strategy for End-of-Life and Palliative Care 2021-2028 | Unique characteristics of paediatric palliative care (page 8)<br>Genetic counselling: More than one child in the family may be affected, and there may be a need for genetic counselling<br>Main groups of life-limiting conditions for children: Table 1 (page 14)<br>Life-threatening conditions for which curative treatment may be feasible but can fail: Children with cancer when treatment fails. Irreversible organ failure where transplantation is not an option or where transplantation has failed. |

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| | | | | Life-limiting conditions where premature death is inevitable. However, there may be long periods of intensive treatment aimed at prolonging life and allowing participation in normal activities: Examples include complex cardiac disease and Duchenne muscular dystrophy (DMD). Ongoing research and medication improvements have meant that some people with Cystic Fibrosis are surviving into their 40s and beyond. A similar trend is seen with DMD.<br>Life-limiting, progressive conditions without curative treatment options, where treatment is exclusively palliative and may commonly extend over many years: Examples include neurodegenerative conditions (e.g. Batten disease), metabolic conditions (e.g. mucopolysaccharidoses) and neuromuscular conditions.<br>Priority One: Care is accessible to everyone, everywhere (page 16)<br>The majority of referrals to paediatric palliative care are for non-cancer diagnoses and include conditions such as neurological, metabolic or chromosomal abnormalities. |
| Scotland | Dumfries and Galloway Integration Joint Board | 2020 | A Plan for Palliative Care | 4.3.5 Learning disability (page 29)<br>People with a learning disability have a shorter life expectancy compared to the general population largely due to life limiting illnesses being more prevalent in this group. These include<br>• higher levels of particular cancers including stomach and bowel cancer and<br>• higher risk of developing heart conditions, dementia and leukaemia (specific to people with Down's Syndrome) |
| Global | World Health Organization | 2018 | Integrating palliative care and symptom relief into primary health care: a WHO guide for planners, implementers and managers | What is palliative care? (page 6)<br>Many countries also lack rehabilitation medicine specialists and services and long-term care facilities to care for people with non-life-threatening but serious disabilities such as paraplegia or quadriplegia or those due to brain injuries or congenital anomalies |
| Global | World Health Organization | 2018 | Integrating palliative care and symptom relief into paediatrics: A WHO guide for health planners, implementers and managers | What is palliative care? (page 6)<br>Many countries also lack rehabilitation medicine specialists and services and long-term care facilities to care for children with non-life-threatening but serious disabilities such as paraplegia or quadriplegia or those due to brain injuries or congenital anomalies<br>Types of health conditions (page 8)<br>The wide range of childhood illnesses increases the difficulty of providing PPC services that meet each child's needs. Further, many paediatric genetic or congenital conditions are rare and not seen in adults, the symptoms may differ in each child and there may be no clear diagnosis or prognosis<br>Table 3: Populations that need PPC (page 10)<br>Children with progressive life-threatening conditions for which no curative treatment is available: Spinal muscular atrophy, Duchenne's muscular dystrophy |

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| | | | | Neonates who are severely premature or have severe congenital anomalies: Severe prematurity, anencephaly, congenital diaphragmatic hernia, trisomy 13 or 18<br>Table 4: Condition that commonly generate a need for PPC (page 10)<br>Genetic conditions: Neurologic conditions (progressive neurological deficits and disability), Sickle cell disease and anaemia (pain crises, bone necrosis), Connective tissue disorders (chronic pain)<br>Children who suffer without a clearly life-threatening condition (page 12)<br>Whether the disability is due to a traumatic injury, congenital anomaly or genetic condition, pain and social isolation and stigmatization are common |
| Europe | European Association of Palliative Care | 2015 | Consensus Norms for Palliative Care of People with Intellectual Disabilities in Europe | Cause of death (page 23)<br>There are also higher rates of dementia in the population of people with intellectual disabilities and a higher incidence of Alzheimer's Disease associated with Down Syndrome. A Swedish study found that dementia was a main or contributing cause of death in 30% of older people with Down Syndrome. The genetic link between Down Syndrome and dementia is thought to be due to the presence of the third chromosome 21 which is associated with the production of the beta-amyloid protein which has been found in the brain of people with Alzheimer's dementia |
| Canada | Alberta Health Services | 2014 | Palliative and End of Life Care: Alberta Provincial Framework | Pediatric-Focused Services (page 14)<br>In most pediatric palliative care programs, 70% or more of the children on the program have chronic neurological or genetic (often multi-system) conditions and a much smaller percentage have cancer |

# Appendix E: supplementary files related to quantitative study

## CONTENTS

SUPPLEMENTARY FILE E1: QUANTITATIVE STUDY PROTOCOL

# Investigating the barriers and facilitators to genetic and genomic testing for people receiving palliative care in Australia and New Zealand: A survey study protocol

**Study team**

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**Background and rationale**

For people dying from an inherited disease, palliative care may be the last opportunity to store a DNA sample for future use by their families and offspring so that they can accurately appraise their individual risk of developing the same condition. Banking a DNA sample from a dying relative may enable family members to access an accurate risk assessment and make informed decisions about managing their own risk and the risk to their children, for example, engaging in appropriate screening or risk reducing surgery. Although testing may not alter the dying person's treatment, it is vital that the opportunity to store a blood sample for possible future genetic testing of relatives is not lost.

A large proportion of people receiving palliative care will have a diagnosis of cancer. Up to 15% of common cancers are due to a pathogenic variant.(1). Most hereditary common cancers, such as breast, ovarian and colorectal cancer are dominantly inherited, whereby each first-degree relative of an affected individual is at 50% risk of having inherited the mutation. In addition to cancer, there are other adult-onset life-limiting disorders that are due to a gene mutation, such as motor neurone disease and Huntington’s disease.

People receiving palliative care have expressed positive feelings about discussing genetic testing, such as reassurance, interest and a sense of altruism.(2-4), yet, discussion of genetics is largely missing from the palliative care agenda,(5) and eligibility for genetic testing is frequently not identified in palliative care.(3, 6) There are many possible barriers to discussing genetic testing in this setting, including lack of confidence and knowledge,(7) limited awareness of how to access genetics services,(8, 9) uncertainty about whose responsibility it is to raise the issue with patients and families,(8) and concern about causing additional distress.(10) As a recent commentary on the integration of genomic testing into healthcare noted, 'It is probable that all health care practitioners will be engaging with genomic medicine in time and as it is 'mainstreamed' at scale, health professionals will increasingly be asked to communicate and manage the results from genomic testing.'(11)

Although a small qualitative study has identified the need for close links with clinical genetics and education for palliative care health professionals,(9) few studies have investigated communication about genetic or genomic testing in palliative care in Australia and New Zealand from the perspective of palliative care and genetics health professionals. Therefore, the aim of this study is to increase understanding of palliative care and genetic health professionals' current practice, views, experiences, barriers and facilitators, in order to build an evidence base for an intervention to support these health professionals to address genetics with palliative patients and their families.

**Research questions & objectives**

<u>Descriptive questions</u>

- How frequently do palliative care and genetic health professionals estimate their engagement with various genetic activities with palliative patients and their families?
- How do palliative care and genetic health professionals rate their confidence when engaging in genetic activities with palliative patients and their families?
- What do palliative care and genetic health professionals perceive as the most important barriers and facilitators to engaging in genetic activities with palliative patients and their families?
- How do palliative care and genetic health professionals rate their agreement with statements pertaining to the:
    - harms and benefits of addressing genetics with palliative patients and their families

    - appropriateness of the palliative care setting
    - role of palliative care health professionals
    - approaching discussions about genetics
    - balancing individual autonomy with familial rights
- How do palliative care and genetic health professionals rate their level of comfort with palliative care health professionals being responsible for initiating and managing DNA storage with palliative patients at end-of-life?

Inferential questions

- What are the similarities and differences in the above responses between palliative care and genetic health professionals?

**Objectives**

- To descriptively analyse the frequency of engagement in genetic activities, confidence performing genetic activities, perceived barriers and facilitators, agreement with factors affecting engagement with genetic activities and comfort with palliative care health professionals initiating and managing DNA storage at end-of-life
- To compare the similarities and differences between genetic and palliative care health professionals' frequency of engagement in genetic activities, confidence performing these activities, perceived barriers and facilitators, agreement with factors affecting engagement with genetic activities and comfort with palliative care health professionals initiating and managing DNA storage at end-of-life.
- To develop recommendations for practice and future research that contribute to the evidence base for the development of an intervention that supports genetic and palliative care health professionals to address genetics with palliative patients and their families

**Expected outcomes and significance**

The outcome of this study will contribute to Stephanie White's PhD thesis to develop an evidence base around the barriers and facilitators towards integrating genetics and genomics into the care of people with palliative care needs, and their families. Long term, we hope this study will inform the development of an intervention to build the capacity of palliative care health professionals to deliver genetic counselling to dying people and their families and contribute to the development of guidelines for the provision of genetic testing in healthcare

settings outside of specialist genetics services.

**Methods**

The study will use a cross sectional on-line survey using REDcap software to reach palliative care and genetics health professionals.

***Eligibility criteria***

This study will collect data from two health professional groups: i) palliative care health professionals and ii) genetics health professionals.

i) Palliative care health professionals are defined as medical doctors or nurses who specialise or sub-specialise in the provision of palliative care to patients diagnosed with a life-limiting illness. Students (ie. those completing base level medical or nursing degrees) are not eligible.

ii) Genetics health professionals are defined as medical doctors who specialise or sub-specialise in clinical genetics (including familial cancer), and genetic counsellors who have completed the minimum degree required in their country to practice as a genetic counsellor. Students (ie. medical students completing their base level medical training and genetic counselling students) are not eligible.

***Sample***

We will be using a stratified sampling technique. We have identified potential participants through health professional groups, with a potential pool of 4635 persons that could participate in the survey. Based on similar previous research studies we expect a 30% response rate, equaling a sample of 1390 participants. We will have a high level of precision (half-width of 95% CI < 1%, simple asymptotic method) when estimating prevalence of the key indicators for the entire sample, and also have good levels of precision for country (precision=1.4%) and professional status (precision=3%) for the smallest groups in these strata. We will have 0.8 power to test an absolute difference of 12.4% in prevalence of an indicator between sub-groups with at least 250 members, and 10% absolute difference between sub-groups with 500 members.

***Recruitment***

The survey will be distributed to palliative care and genetics health professionals in Australia and New Zealand via national health professional organisations. The following organisations have agreed to circulate the link to the survey to their members:

- Human Genetics Society of Australasia (and associated special interest groups: Australasian Society of Genetic Counsellors and Australasian Association of Clinical Geneticists)
- Australian and New Zealand Society of Palliative Medicine
- Palliative Care Nurses Australia
- Palliative Care Nurses New Zealand

If other professional organisations are identified, they will be approached to ask if they will assist in recruitment.

***Procedure***

Depending on the policy/procedures of the organization, members will receive a link to the survey through an email invitation or advertised within a regular newsletter from their organisation. How many (if any) follow up emails sent will depend on the organisation.

Each organisation will be asked:

- To circulate either an invitation OR a link and asked whether they will be willing to send out 2 or 3 reminders.
- To provide the total number of email addresses they send the survey to.
- If their website is able to detect how many people open an email.

To minimise the risk of bias, a limit will be put on the survey software so only one survey can be filled on the same computer, reducing duplication.

***Measures***

A self-administered RedCap survey has been developed by the study team for palliative care and genetics health professionals based on the literature and the authors' systematic review and qualitative interviews. The survey collects nominal and ordinal data through radio buttons, checkboxes, Likert scales and free text boxes related to the following topics:

- Demographic data: organization they received invitation from, gender, age group, ethnicity, profession, years since qualification, highest academic achievement, specialty (including primary specialty), job title, years working in specialty, country, work sector, work setting, extent of training in either genetics/genomics (for PC health professionals) or end-of-life (EOL)/bereavement communication training (for genetics health professionals), time since completing genetics/genomics/EOL/bereavement training, country, whether there is an embedded genetic service at their institution

- Palliative care health professionals specific data collection will include: how often they are involved in genetic risk assessments (always, usually, sometimes, occasionally, never), how often they've been involved in various genetic activities related to identifying, discussing or managing patients/families in the last 12 months, when genetics discussions tend to take place, who initiates genetic testing, availability of genetics services in their clinical area, how genetic consent is obtained, the three main challenges of facilitating genetic banking/testing, level of confidence about various genetic activities related to identifying, discussing or managing patients/families, which tools/resources are useful in facilitating DNA banking/testing, the extent to which they agree or disagree with various statements about genetics discussions, the appropriateness and impact on patients and families and the palliative care role in addressing genetics, the most helpful facilitators for supporting PC health professionals to discuss genetics and level of comfort about a DNA banking model of care for patients at end-of-life
- Genetics health professionals specific data collection will include: if and when (in the patient's journey) they have been involved in organized DNA banking/testing, who initiated testing, how often they've been involved in various genetic activities related to identifying, discussing or managing patients/families in the last 12 months, availability of genetics services receiving PC, how genetic consent is obtained, the three main challenges of facilitating genetic banking/testing, level of confidence about various genetic activities related to discussing or managing palliative patients/families, which tools/resources are useful in facilitating DNA banking/testing, the extent to which they agree or disagree with various statements about genetics discussions, the appropriateness and impact on patients and families and the palliative care role in addressing genetics, the most helpful facilitators for supporting PC health professionals to discuss genetics and level of comfort about a DNA banking model of care for patients at end-of-life

***Data analysis***

We will conduct a descriptive analysis of the study by calculating the mode and percentages for nominal data, and the median and inter-quartile range for the ordinal data. We will estimate prevalence of the key indicators with a 95% confidence interval across all the survey respondents. Data management and analysis will be completed in appropriate statistical package (e.g. SPSS or SAS).

Inferential statistics will be used to compare responses between occupational groups:

genetic health professionals and palliative care professionals. A/Professor Kris Rogers (statistician) is providing statistical support. A detailed analysis plan has been developed. Likely tests include Chi-square tests and ordinal logistic regression (to compare Likert scale responses between occupational groups). Fisher's exact test will be used if response rates are below what is anticipated.

***Practical issues***

Recruitment is dependent on the policy of each organisation. Recruitment may be a challenge with time poor health professionals. Nurses are known to be poor survey responders. To address this issue, we have piloted the survey with genetic counsellors and palliative care nurses and doctors to ensure the surveys take less than 20 minutes to complete and made adjustments to questions based on pilot participants suggestions to improve clarity. If the response rate is poor, our ethics approval allows us to approach further organisations to distribute the survey and increase participation.

**Start date and duration**

Ethical approval was received in February 2022. Survey distribution is expected to commence in April 2022. The survey will remain open for 4 – 6 weeks and close in May 2022. Data analysis will commence in May/June 2022 and aim to be complete by September 2022. A manuscript ready for publishing should be complete by December 2022.

**Dissemination**

Findings will be disseminated to each organisation for circulation to their members. In addition, the findings will be published in peer-reviewed journals relevant to the target audience and presented within the Graduate School of Health, at the Human Genetics Society of Australasia conference and at a palliative care conference. The findings will also be disseminated via Twitter to influential tweeters within the various disciplines.

**Funding**

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SUPPLEMENTARY FILE E2: RESEARCH INVITATIONS FOR GENETIC AND PALLIATIVE CARE HEALTH PROFESSIONALS

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## New survey invitation: Talking about genetics with people who have palliative care needs, and their families

As a genetics health professional, you may have discussed genetic or genomic testing with people who have palliative care needs or their family members. In this new survey study, we are interested in learning about your views and experiences of these discussions. Even if you have no or limited experience in speaking to people with palliative care needs, we are still interested to understand your views.

If you are a genetic counsellor, clinical geneticist or other health professional working in genetics**, you are invited to participate in an anonymous, online survey** to share your views. The survey will take approximately 10-20 minutes to complete.

**Please follow this link to complete the survey:**

**https://redcap.link/jo2gxpz4**

This study is being conducted by Stephanie White, PhD candidate, at the University of Technology Sydney. The UTS Research Ethics Office has approved this research (ETH18-2408).

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## New survey invitation: Providing palliative care to people with a genetic condition

As a palliative care doctor, you may have cared for people and their family members who have a genetic condition. In this new survey study, we are interested in learning about your views and experiences of discussions about genetics in the palliative setting. Even if you have no or limited experience in these conversations, we are still interested to hear your views.

If you are a palliative care doctor, **you are invited to participate in an anonymous, online survey** to share your views. The survey will take approximately 10-20 minutes to complete.

**Please follow this link to complete the survey:**

**https://redcap.link/jo2gxpz4**

This study is being conducted by Stephanie White, PhD candidate, at the University of Technology Sydney. The UTS Research Ethics Office has approved this research (ETH18-2408).

---

New survey invitation: Providing palliative care to people with a genetic condition

As a palliative care nurse, you may have cared for people and their family members who have a genetic condition. In this new survey study, we are interested in learning about your views and experiences of discussions about genetics in the palliative setting. Even if you have no or limited experience in these conversations, we are still interested to hear your views.

If you are a palliative care nurse, **you are invited to participate in an anonymous, online survey** to share your views. The survey will take approximately 10-20 minutes to complete.

**Please follow this link to complete the survey:**

**https://redcap.link/jo2gxpz4**

This study is being conducted by Stephanie White, PhD candidate, at the University of Technology Sydney. The UTS Research Ethics Office has approved this research (ETH18-2408).

## Your ethics application has been approved as low risk - ETH21-5854

research.ethics@uts.edu.au <research.ethics@uts.edu.au>
Mon 14-Feb-22 1:59 PM
To:
Research Ethics <research.ethics@uts.edu.au>;
Stephanie White <Stephanie.White@uts.edu.au>;
Chris Jacobs <Chris.Jacobs@uts.edu.au>

Cc:
Jules McConnochie <[redacted]@uts.edu.au>;
Adam Maurizi <Adam.Maurizi@uts.edu.au>;
Eddy Dharmadji <Eddy.Dharmadji@uts.edu.au>;
Karen Gomez <Karen.Gomez@uts.edu.au>;
Rebekah Tatian <Rebekah.Tatian@uts.edu.au>;
Toby Newton-John <Toby.Newton-John@uts.edu.au>

1 attachments (270 KB)
Ethics Application.pdf;

Dear Applicant

**Re: UTS HREC Ref. No. ETH21-5854 - "Investigating the Barriers and Facilitators to Genetic and Genomic Testing for People Receiving Palliative Care in Australia, New Zealand and the United Kingdom"**

Your local research office has reviewed the amendment application for your above-named project and agreed that the amendments meet the requirements of the National Statement on Ethical Conduct In Human Research (2007).

I am pleased to inform you that your amendment has been approved as follows:

Personnel: Change research student to Stephanie White. Include Dr Erin Turbitt as Co-Investigator. Remove Grace Phillips as research student.

Research instrument: A number of additional domains for assessment have been added to the existing research instrument. These have been added in the form of additional response options and two new questions. For the committees convenience, we have attached a summary table with the relevant research questions and responses for modification, the proposed modifications to the question and responses and a justification for the modification is attached to this amendment. Please see the attachment called "Proposed survey modifications" for full details regarding the proposed changes.

Participant material: We wish to make changes to the Participant Information Sheet to reflect the updates to study personnel . An updated copy is attached.

Recruitment of participants: We propose to widen the pool of potential participants in Australia and New Zealand by distributing the survey through any organisation that

agrees in the future, to send the survey to it's members on our behalf. We will request formal, written approval by any organisation that we approach and will only approach organisations that would reasonably have a membership that are eligible to participate in this study. If the committee requests, we will send these agreements to the committee as organisations agree.

This amendment is subject to the standard conditions outlined in your original letter of approval. You are reminded that this letter constitutes ethics approval only. This research project must also be undertaken in accordance with all UTS policies and guidelines including the Research Management Policy.You should consider this your official letter of approval. If you require a hardcopy please contact your local research office.To access this application, please click here. A copy of your application has also been attached to this email. If you have any queries about this approval, or require any amendments to your approval in thefuture, please do not hesitate to contact your local research office or the Ethics Secretariat(Research.Ethics@uts.edu.au).
----------Ref: 12e

# SUPPLEMENTARY FILE E4: PARTICIPANT INFORMATION SHEET (SURVEY LANDING PAGE)

## Investigating the barriers and facilitators to genetic/genomic testing for people receiving palliative care in Australia, New Zealand and the United Kingdom

[UTS HREC REF NO. ETH19-2408/21-5854]

**What is the research study about?**

The purpose of this research/online survey is to investigate the existing facilitators and barriers to genetic/genomic testing in the adult palliative care setting.

You have been invited to participate because you are a member of a professional organisation of healthcare professionals specialising in either palliative care, genetics or genomics. We are interested in understanding your experiences and views of discussing genetics with people who have palliative care needs, and their families.

**Who is conducting this research?**

My name is Stephanie White and I am a PhD candidate at UTS. The research team includes experts in genetic counselling, clinical genetics, palliative care medicine and nursing and medical ethics.

**Other members of our study team:**

Dr Chris Jacobs, Senior Lecturer, Genetic Counselling, UTSAssociate Professor Alison McEwen, Head of Genetic Counselling, UTSProfessor Jane Phillips, Professor of Palliative Care Nursing, UTSAssociate Professor Kathy Tucker, Consultant Clinical Geneticist, Prince of Wales Hospital (POWH), SydneyDr Erin Turbitt, Lecturer, Genetic Counselling, UTSDr Megan Best, Associate Professor of Bioethics, Institute for Ethics and Society, The University of Notre DameDr April Morrow, Genetic Counsellor, POWH, SydneyEligibility Criteria

You have been invited to participate because you work or have worked as a palliative care or genetics health professional. Please do not complete this survey if you have never worked as a health care professional in palliative care or genetics/genomics.

**Do I have to take part in this research study?**

Participation in this study is voluntary. It is completely up to you whether or not you decide to take part.

If you decide to participate, you will be directed to complete an online survey that will take 10 to 20 minutes of your time. You will be asked some demographic questions, such as about your professional and clinical area, genetics/genomics education and views and experiences of DNA banking/testing in adult palliative care. Please answer all the questions that you are directed to. If you receive the survey from more than one organisation, please only complete it once.

You can change your mind at any time and stop completing the survey

without consequences.

**Are there any risks/inconvenience?**

It is possible that the questionnaire could cause upset by reminding participants of an uncomfortable experience of raising a difficult issue with patients or families or leads to feelings of embarrassment or regret at not having discussed genetic testing with potentially eligible patients or families. If you experience feelings of distress as a result of participation in this study you can let

the researcher know and they will provide you with assistance.

Alternatively, please contact your usual workplace support provider, occupational health service, or General Practitioner. Accessible, anonymous support is also available from support organisations as follows: In the UK, the number for Samaritans is 116 123 In Australia, the number for Lifeline is 13 11 44 In New Zealand, the number for Lifeline is 0800 543 354

**What will happen to information about me?**

Access to the online questionnaire is online through this link. Submission of the online questionnaire/s is an indication of your consent. By responding "Yes" to "I have read the information above and I agree to taking part in this survey", you consent to the research team collecting and using personal information about you for the research project. All this information will be treated confidentially. The data will be securely stored on a password protected university computer with access limited to the research team. Your information will only be used for the purpose of this research project, except as required by law.

We plan to publish results in a peer-reviewed academic journal, and to disseminate findings to health professionals, patient groups and policy makers through conference presentations and publications. Results from this research will be included in Stephanie White's thesis. In any publication, information will be provided in such a way that you cannot be identified.

**What if I have concerns or a complaint?**

If you have concerns about the research that you think the researcher can help you with, please feel free to contact Stephanie White by email at stephanie.white@uts.edu.au. If you would like to talk to someone who is not connected with the research, you may contact: UTS Research Ethics Officer, Phone: 02 9514 9772 E-mail: research.ethic@uts.edu.au And quote UTS HREC REF NO. ETH18-2408.

This study has been reviewed by and received ethics clearance through University of Technology Sydney's Human Research Ethics Committee.

If you've received this survey from more than one organisation, please only complete it once.

---

Please choose which group of health professions your occupation would fall under. You will then be directed to the relevant survey.

- ○ Palliative care health professionals
- ○ Genetics health professionals
- ○ None of the above

---

I have read the information above and I agree to taking part in this survey

- ○ Yes
- ○ No

SUPPLEMENTARY FILE E5: SURVEY FOR GENETIC HEALTH PROFESSIONALS



# Barriers and facilitators to genetic testing in palliative care: Survey for Genetics Health Professionals

1. Are you now working, or have you previously worked, in a clinical area?
   - ◯ Yes
   - ◯ No

2. Which organisation sent you the link to the survey that you are completing?
   - ◯ Human Genetics Society of Australasia (HGSA)
   - ◯ Australasian Society of Genetic Counsellors (ASGC)
   - ◯ Australasian Association of Clinical Geneticists (AACG)
   - ◯ British Society for Genetic Medicine (BSGM)
   - ◯ Association of Genetic Nurses and Counsellors (AGNC)
   - ◯ Clinical Genetics Society (CGS)
   - ◯ European Society of Human Genetics (ESHG)
   - ◯ Other

Please explain.

_____

The following questions are to understand your experiences and views about discussing DNA banking/testing with patients receiving palliative care, and their families.

We will ask you some demographic questions at the end of the survey.

Please note:

DNA banking, as opposed to DNA testing, is the process of obtaining a DNA sample (usually blood, saliva or buccal) and storing this sample in a laboratory, without performing any DNA testing. As you answer these questions, please consider DNA banking for CLINICAL use only. This means the DNA would be used for future DNA testing to help understand genetic risk for relatives and NOT used for research purposes.

| | |
|---|---|
| Have you ever received any training in communicating with patients at the end of life or with bereaved families? | ◯ Yes<br>◯ No |
| What area(s) did you study? (Please indicate all that are relevant.) | ☐ Communicating with cancer patients<br>☐ Communication skills with patients at the end of life<br>☐ Bereavement counselling<br>☐ Other |
| Please explain. | _____ |
| What type of training have you received in communicating with patients at the end of life or with bereaved families? (Please indicate all that are relevant.) | ☐ Degree/diploma<br>☐ Short course/module over at least two sessions<br>☐ One-off lecture/seminar/workshop<br>☐ On-line short course or Massive Online Open Course (MOOC)<br>☐ Course/session on communication with people receiving palliative care as part of another course or study day<br>☐ Course/session on bereavement counselling<br>☐ Private study (e.g. reading papers)<br>☐ Other |
| Please explain. | _____ |
| How long has it been since you last received training or education in communicating with patients at the end of life or with bereaved families? | ◯ Less than 12 months<br>◯ 1 to 2 years<br>◯ 3 to 5 years<br>◯ 6 to 10 years<br>◯ More than 10 years |
| Are you interested in receiving training in communicating with patients at the end of life or with bereaved families? | ◯ Yes<br>◯ No |

| | | |
|---|---|---|
| | What type of training would you like to receive in communicating with patients at the end of life or with bereaved families? (Please indicate all that are relevant.) | ☐ Degree/diploma<br>☐ Short course/module over at least two sessions<br>☐ One-off lecture/seminar/workshop<br>☐ On-line short course or Massive Online Open Course (MOOC)<br>☐ Course/session on communication with people receiving palliative care as part of another course or study day<br>☐ Course/session on bereavement counselling<br>☐ Private study (e.g. reading papers)<br>☐ Other |
| | Please specify | _____ |
| | What is the main reason you are not interested in receiving this training? | ◯ Lack of time to receive training<br>◯ I have other education/training priorities<br>◯ I already know a lot about communicating with patients at end-of-life & bereaved families<br>◯ Communicating with patients at end-of-life & bereaved families is not relevant to my work<br>◯ Other |
| | Please specify | _____ |
| | Please add any comments about the training you have received or would find helpful in communicating with patients at the end of life of with bereaved families. | _____ |
| 4. | Have you ever been involved in facilitating DNA banking/testing for people receiving palliative care? | ◯ Yes<br>◯ No<br>◯ Not sure |
| 4a. | In your experience, at what point did you usually become involved? | ◯ When the patient commences palliative care<br>◯ When the patient is close to death<br>◯ After the patient has died<br>◯ It has never been raised in my experience<br>◯ Not sure<br>◯ Other |
| | Please explain. | _____ |
| 5. | In your experience, who usually initiates requests for DNA testing? | ◯ Patients<br>◯ Family members<br>◯ Palliative care health professionals<br>◯ Genetics health professionals<br>◯ Oncology health professionals<br>◯ It has never been raised in my experience<br>◯ Not sure<br>◯ Other |
| | Please explain. | _____ |

6a. Please indicate approximately how often you have been involved in the following activities in the last 12 months.

| | | Zero | Once or twice | Three to five times | Six to 10 times | More than 10 times | I have not worked clinically in the last 12 months |
|---|---|---|---|---|---|---|---|
| a. | Identifying a patient receiving palliative care who is eligible for DNA banking/testing. | ○ | ○ | ○ | ○ | ○ | ○ |
| b. | Initiating a discussion about DNA banking/testing with a patient receiving palliative care or their relative. | ○ | ○ | ○ | ○ | ○ | ○ |
| c. | Providing advice to a palliative care health professional about one of their patients | ○ | ○ | ○ | ○ | ○ | ○ |
| d | Receiving a referral from a palliative care health professional for a patient receiving palliative care | ○ | ○ | ○ | ○ | ○ | ○ |
| e. | Taking consent for DNA banking/testing from a patient receiving palliative care. | ○ | ○ | ○ | ○ | ○ | ○ |
| f. | Taking consent for DNA banking/testing from a relative of a patient receiving palliative care. | ○ | ○ | ○ | ○ | ○ | ○ |

6b. Please indicate approximately how often you have been involved in the following activities in the last 12 months.

| | | Zero | Once or twice | Three to five times | Six to 10 times | More than 10 times | I have not worked clinically in the last 12 months |
|---|---|---|---|---|---|---|---|
| g. | Facilitating collection of a DNA sample from a patient receiving palliative care | ○ | ○ | ○ | ○ | ○ | ○ |
| h. | Disclosing genetic/genomic test results to a patient receiving palliative care. | ○ | ○ | ○ | ○ | ○ | ○ |
| i. | Disclosing a deceased patient's genetic test results to bereaved relatives. | ○ | ○ | ○ | ○ | ○ | ○ |
| j. | Providing genetic counselling to a patient receiving palliative care | ○ | ○ | ○ | ○ | ○ | ○ |

7. Which scenario best describes the availability of specialist genetics services for people receiving palliative care in your area?

- ○ Specialist genetics services and palliative care services are embedded within the same hospital or group of hospitals.
- ○ Specialist genetics services and palliative care services are not embedded within the hospital or group but are accessible to each other.
- ○ Specialist genetics services and palliative care services are NOT accessible to each other.
- ○ Palliative care services have access to private genetics services only
- ○ Not sure

8. In your experience how is consent for DNA banking or testing obtained and documented?

- ○ Verbal consent only (undocumented)
- ○ Verbal consent (documented in the patient's clinical records)
- ○ Written consent using locally available paperwork (but not a formal genetics consent form)
- ○ Written consent using a formal genetics consent form
- ○ In my experience consent has not been taken
- ○ Other

Please explain.

---

9. In your experience, what do you consider to be the main challenges for genetics health professionals in facilitating DNA storage/testing for people receiving palliative care? (Please select up to 3 responses.)

* Please note: These response options have all been identified as challenges in prior studies. For the purpose of identifying priority areas, we are requesting you nominate your top three challenges

- ☐ Identifying eligible patients
- ☐ Urgency of the situation/ referral
- ☐ Conflicting priorities between providing palliative care and facilitating genetic testing
- ☐ Obtaining informed consent
- ☐ Discomfort with initiating discussions about DNA storage/testing with patients or families in palliative care
- ☐ Palliative care health professionals' lack of knowledge about DNA banking/testing or procedures
- ☐ Genetics health professionals' lack of knowledge of the procedure for consent and DNA storage
- ☐ The views or expectations of the family
- ☐ Lack of availability of specialist genetics services
- ☐ Communication difficulties between genetics and palliative care services
- ☐ Conflicting views within the palliative care team about the utility of DNA banking/testing for palliative patients, and their families
- ☐ Lack of resources
- ☐ Distress of the patient or family members
- ☐ Complex family dynamics
- ☐ Concern that a discussion about genetics could damage the therapeutic relationship
- ☐ Under-referral of palliative patients to genetics services
- ☐ In my experience DNA storage or genetic/ genomic testing has not ever been considered
- ☐ Other

(Please choose only up to 3 responses)

Please explain.

---

10. Please indicate your level of confidence about the following:

| | | Not at all confident | Fairly unconfident | Neither confident or unconfident | Fairly confident | Confident |
|---|---|---|---|---|---|---|
| a. | Communicating with patients at the end of life | ○ | ○ | ○ | ○ | ○ |
| b. | Communicating with the families of patients who are at the end of life | ○ | ○ | ○ | ○ | ○ |
| c. | Discussing DNA banking with patients or their families in the palliative care setting | ○ | ○ | ○ | ○ | ○ |
| d. | Discussing DNA testing with patients or their families in the palliative care setting | ○ | ○ | ○ | ○ | ○ |
| e. | Facilitating collection of a DNA sample from a patient receiving palliative care | ○ | ○ | ○ | ○ | ○ |
| f. | Disclosing genetic/genomic test results to palliative care patients | ○ | ○ | ○ | ○ | ○ |
| g. | Disclosing genetic/genomic test results to bereaved families | ○ | ○ | ○ | ○ | ○ |
| h. | Knowing my legal responsibilities when sharing health information with family members when a patient is terminal or after they have died | ○ | ○ | ○ | ○ | ○ |

11. What resources or tools have you found helpful when communicating with patients receiving palliative care and/or their families? (Please indicate all that are relevant.)

- ☐ Web-based risk assessment tool
- ☐ Smart phone App
- ☐ Support from a palliative care colleague
- ☐ Support from a genetics colleague
- ☐ Educational brochures
- ☐ Telephone information hotline
- ☐ Face to face education
- ☐ Online education
- ☐ Clinical decision-making algorithm
- ☐ Clinical practice guidelines
- ☐ I have not found any resources or tools helpful
- ☐ Other

How frequently did you receive "face to face education" for communicating with patients receiving palliative care and/or their families? _____

Which clinical practice guidelines did you find useful? _____

Please explain.

_____

12. What additional resources or tools would be helpful, if any?

13. Please indicate to what extent you agree or disagree with the following statements:

| | | Strongly disagree | Disagree | Neither agree nor disagree | Agree | Strongly agree |
|---|---|---|---|---|---|---|
| a. | Discussing DNA banking/testing with people receiving palliative care undermines the central ethos of palliative care in providing comfort and support at an emotionally vulnerable time. | ○ | ○ | ○ | ○ | ○ |
| b. | Patients may experience positive emotional benefits from being able to give a sample for DNA banking/testing for the possible future benefit of their relatives. | ○ | ○ | ○ | ○ | ○ |
| c. | Discussing DNA banking/testing may cause distress to the families by making them assume/fear their fate is pre-determined. | ○ | ○ | ○ | ○ | ○ |
| d. | DNA banking/testing will have been discussed by other health professionals before the patient is referred to palliative care. | ○ | ○ | ○ | ○ | ○ |
| e. | The priority in palliative care is to improve quality of life and relieve suffering and therefore it is not an appropriate time to discuss DNA banking/testing. | ○ | ○ | ○ | ○ | ○ |
| f. | DNA banking/testing of the patient may be important for the surviving relatives. | ○ | ○ | ○ | ○ | ○ |
| g. | It is not the responsibility of health professionals in palliative care to discuss DNA banking/testing. | ○ | ○ | ○ | ○ | ○ |
| h. | DNA banking/testing is not appropriate for people receiving palliative care because it will not help the patient. | ○ | ○ | ○ | ○ | ○ |

14. Please indicate to what extent you agree or disagree with the following statements:

| | | Strongly disagree | Disagree | Neither agree nor disagree | Agree | Strongly agree |
|---|---|---|---|---|---|---|
| a. | Palliative care health professionals are well placed to have discussions about DNA banking/testing with the family members of a palliative patient | ○ | ○ | ○ | ○ | ○ |
| b. | Concerns about genetic discrimination (eg. insurance and employment discrimination) make discussions about DNA banking/testing with palliative patients and their families difficult | ○ | ○ | ○ | ○ | ○ |
| c. | Families generally appreciate being told genetic information that is relevant to their health | ○ | ○ | ○ | ○ | ○ |
| d. | Discussions about DNA banking/testing need to be individualised to the palliative patient, and their families | ○ | ○ | ○ | ○ | ○ |
| e. | As time goes on, genetic health information will inevitably become part of the palliative care health professionals' scope of practice | ○ | ○ | ○ | ○ | ○ |
| f. | If a patient declines a discussion about DNA banking/testing , palliative care health professionals should revisit this discussion with palliative patients at a later date to see if they've changed their mind | ○ | ○ | ○ | ○ | ○ |
| g. | The family of a palliative patient have a right to know if they are at risk of developing a genetic disease, regardless of the palliative patient's wishes | ○ | ○ | ○ | ○ | ○ |
| h. | The right of a palliative patient to decline a discussion about DNA banking/testing are to be respected, regardless of the family's wishes | ○ | ○ | ○ | ○ | ○ |

15. In your opinion, which of these would most help genetics health professionals to discuss and/or facilitate DNA banking/testing with palliative patients, and their families? (Please choose up to 3 responses)

*Please note: These response options have all been identified as facilitators in prior studies. For the purpose of identifying priority areas, we are requesting you nominate your top three facilitators

- ☐ Physically co-locating palliative care and genetics health professionals within a hospital or organisation
- ☐ Developing a specific referral template for palliative care patients to the genetics service that includes relevant family member details
- ☐ Embedding a genetic counsellor in the palliative care team
- ☐ Having both palliative care and genetics health professionals attending the same multidisciplinary team meetings
- ☐ Fostering a closer working relationships between palliative care and genetics health professionals
- ☐ Delivering genetics education to palliative care health professionals, including ways of sensitively communicating with patients and families about genetics
- ☐ Policy guidance detailing how and when to discuss DNA banking/testing with palliative patients and their families
- ☐ Empowering palliative patients, and their families, to seek out DNA banking/testing for themselves
- ☐ Speaking directly to the palliative care health professional about the palliative patient
- ☐ Improving the capability of electronic medical records to share relevant information between health professionals
- ☐ Collaborating with palliative care health professionals to facilitate collection of a DNA sample from a palliative patient
- ☐ We shouldn't be discussing DNA banking/testing with palliative patients, or their families
- ☐ Other

Please explain

_____

16. Previous research suggests that patients who are suitable for DNA testing and close to end of life should be offered DNA banking by palliative care health professionals, rather than referring them to the genetics service for DNA testing.

We would like to understand your views about how this would work in practice.

Please indicate your comfort with palliative care health professionals performing the following actions:

| | | Very uncomfortable | Somewhat uncomfortable | Neutral | Somewhat comfortable | Very comfortable |
|---|---|---|---|---|---|---|
| a. | Introduce the idea of DNA banking with the patient and/or the family | ○ | ○ | ○ | ○ | ○ |
| b. | Obtain consent for DNA banking from the patient, or appointed representative | ○ | ○ | ○ | ○ | ○ |
| c. | | | | | | |

| | | | | | | |
|---|---|---|---|---|---|---|
| | Facilitate collection of DNA sample | ○ | ○ | ○ | ○ | ○ |
| d. | Organise for the DNA sample to be banked | ○ | ○ | ○ | ○ | ○ |
| e. | Instruct the family how to follow up with the genetics service | ○ | ○ | ○ | ○ | ○ |
| f. | Communicate family follow up plan to the genetics service | ○ | ○ | ○ | ○ | ○ |

Please describe any further thoughts you have about palliative care health professionals performing these actions

_____

**Thank you. We will now ask you some demographic questions.**

17. What is your gender?
- ◯ Female
- ◯ Male
- ◯ Self-described (specify below)
- ◯ Prefer not to say

Please describe

---

18. What is your age group?
- ◯ < 20
- ◯ 20-24
- ◯ 25-34
- ◯ 35-44
- ◯ 45-54
- ◯ 55-64
- ◯ 65 or older
- ◯ Prefer to not say

19. What is your country of birth?
- ◯ Australia
- ◯ New Zealand
- ◯ England
- ◯ India
- ◯ Philippines
- ◯ Vietnam
- ◯ Italy
- ◯ South Africa
- ◯ Malaysia
- ◯ Scotland
- ◯ Other (please specify)
- ◯ Prefer not to say

Please specify

---

20. What cultural background or ethnicity do you identify with? (Select all that apply)
- ☐ None
- ☐ Australian
- ☐ New Zealand
- ☐ English
- ☐ Irish
- ☐ Scottish
- ☐ Chinese
- ☐ Italian
- ☐ Indian
- ☐ German
- ☐ Greek
- ☐ Vietnamese
- ☐ Other (please specify)
- ☐ Prefer not to say

(You may select more than one)

Please describe your cultural background or ethnicity

---

| | | |
|---|---|---|
| 21. | Which language do you mainly speak at home? (If more than one language, indicate the one that is spoken most often) | ◯ English<br>◯ Mandarin<br>◯ Arabic<br>◯ Cantonese<br>◯ Vietnamese<br>◯ Italian<br>◯ Greek<br>◯ Hindi<br>◯ Spanish<br>◯ Punjabi<br>◯ Other (please specify)<br>◯ Prefer not to say |
| | Please describe the language you mostly use at home | _____ |
| 22. | What is your primary profession? | ◯ Medical<br>◯ Nursing<br>◯ Genetic counselling<br>◯ Other |
| | Please explain. | _____ |
| 23. | How many years has it been since you qualified in your current profession? | ◯ Less than 2 years<br>◯ 2 to 5 years<br>◯ 6 to 10 years<br>◯ 11 to 15 years<br>◯ More than 15 years |
| 24. | What is your highest academic achievement? | ◯ PhD<br>◯ Master degree<br>◯ Bachelor degree<br>◯ Diploma<br>◯ Professional qualification<br>◯ Other |
| | Please explain. | _____ |
| 25. | What is your primary area of specialty? | ◯ Clinical genetics<br>◯ Cancer genetics<br>◯ Other |
| | Please explain. | _____ |

| | | |
|---|---|---|
| 26. | What is your job title? | ◯ Genetic Counsellor (not yet Registered/Certified)<br>◯ Genetic Counsellor (Member of the HGSA)<br>◯ Genetic Counsellor (Fellow of the HGSA)<br>◯ Genetic Counsellor (GCRB Registered)<br>◯ Registered nurse<br>◯ Senior registered nurse (including nurse practitioner)<br>◯ Junior doctor (eg intern, resident or equivalent)<br>◯ Senior doctor (eg registrar, fellow or equivalent)<br>◯ Consultant doctor (including staff specialist or equivalent)<br>◯ Other |
| | Please explain. | _____ |
| 27. | How long have you worked in your current specialist area? | ◯ Less than 2 years<br>◯ 2 to 5 years<br>◯ 6 to 10 years<br>◯ 11 to 15 years<br>◯ More than 15 years |
| 28. | Which country do you work in? | ◯ Australia<br>◯ New Zealand<br>◯ England<br>◯ Scotland<br>◯ Northern Ireland<br>◯ Wales<br>◯ Other |
| | Please explain. | _____ |
| 29. | Which sector do you usually work in? | ◯ Public<br>◯ Private<br>◯ Public and private<br>◯ Other |
| | Please explain. | _____ |
| 30. | What location do you usually work in? | ◯ City/metropolitan/urban<br>◯ Regional<br>◯ Rural<br>◯ Other |
| | Please explain. | _____ |
| 31. | What setting do you usually work in? | ◯ Hospital<br>◯ Independent clinic<br>◯ Other |
| | Please explain. | _____ |

**Many thanks for your participation in this survey.**

**If you have any further comments, you may write them below.**

**When you are finished, please press submit.**

**Have a great day!**

32. Please add any further comments about facilitating DNA banking/testing with people receiving palliative care or their families.

33. Please tell us if you have any further comments about this survey or study.

SUPPLEMENTARY FILE E6: SURVEY FOR PALLIATIVE CARE HEALTH PROFESSIONALS



# Barriers and facilitators to genetic testing in palliative care: Survey for Palliative Care health professionals

| | | |
|---|---|---|
| 1. | Are you now working, or have you previously worked, in a clinical area? | ◯ Yes<br>◯ No |
| 2. | Which organisation sent you the link to the survey that you are completing? | ◯ Australian and New Zealand Society for Palliative Medicine (ANZSPM)<br>◯ Palliative Care Nurses Australia (PCNA)<br>◯ Palliative Care Nurses New Zealand (PCNNZ)<br>◯ Association for Palliative Medicine (APM)<br>◯ Royal College of Nursing (RCN)<br>◯ Palliative Care Forum<br>◯ Other |
| | Please explain. | _____ |

The following questions are to understand your experiences and views about discussing DNA banking/testing with patients receiving palliative care, and their families.

We will ask you some demographic questions at the end of the survey.

Please note:

DNA banking, as opposed to DNA testing, is the process of obtaining a DNA sample (usually blood, saliva or buccal) and storing this sample in a laboratory, without performing any DNA testing As you answer these questions, please consider DNA banking for CLINICAL use only This means the DNA would be used for future DNA testing to help understand genetic risk for relatives and NOT used for research purposes.

| | |
|---|---|
| Have you ever received any training in family history risk assessment, genetic testing and/or genomic testing? | ◯ Yes<br>◯ No |
| What area(s) did you study? (Please indicate all that are relevant) | ☐ Family history risk assessment<br>☐ Genetics and/or genetic testing<br>☐ Genomics and/or genomic testing<br>☐ Other |
| Please explain. | _____ |
| What type of training have you received in family history risk assessment, genetic testing and/or genomic testing? (Please indicate all that are relevant) | ☐ Degree/diploma<br>☐ Short course/module over at least two sessions<br>☐ One-off lecture/seminar/workshop<br>☐ On-line short course or Massive Online Open Course (MOOC)<br>☐ Courses/sessions on genetics/genomics as part of another course or study day<br>☐ Private study (e.g. reading papers)<br>☐ Other |
| Please explain. | _____ |
| How long has it been since you last received training or education in family history risk assessment, genetic testing and/or genomic testing? | ◯ Less than 12 months<br>◯ 1 to 2 years<br>◯ 3 to 5 years<br>◯ 6 to 10 years<br>◯ More than 10 years |
| Are you interested in receiving training in family history risk assessment and genetic/genomic testing? | ◯ Yes<br>◯ No |
| What type of training would you like to receive in family history risk assessment, genetic testing and/or genomic testing? (Please indicate all that are relevant) | ☐ Degree/diploma<br>☐ Short course/module over at least two sessions<br>☐ One-off lecture/seminar/workshop<br>☐ On-line short course or Massive Online Open Course (MOOC)<br>☐ Courses/sessions on genetics/genomics as part of another course or study day<br>☐ Private study (e.g. reading papers)<br>☐ Other |
| Please explain | _____ |

What is the main reason you are not interested in receiving training?

- ◯ Lack of time to receive training
- ◯ I have other education/training priorities
- ◯ I already know a lot about genetics/genomics
- ◯ Genetics/genomics is not relevant to my work
- ◯ Other

Please explain

_____

Please add any comments about the training you have received or would find helpful about genetics/genomics.

_____

4. Please indicate how often you are involved in the following activities in your current practice.

| | | Never | Occasionally | Sometimes | Usually | Always |
|---|---|---|---|---|---|---|
| a. | Taking a family health history | ◯ | ◯ | ◯ | ◯ | ◯ |
| b. | Drawing a three-generation family tree (pedigree) | ◯ | ◯ | ◯ | ◯ | ◯ |
| c. | Making a genetic risk assessment | ◯ | ◯ | ◯ | ◯ | ◯ |

5. In your experience, at what point in the patient's trajectory is a family health history usually taken?

- ◯ When the patient commences palliative care
- ◯ When the patient is close to death
- ◯ After the patient has died
- ◯ Family health history is not taken
- ◯ Not sure
- ◯ Other

Please explain

_____

6. In your experience, who usually initiates requests for DNA testing?

- ◯ Patients
- ◯ Family members
- ◯ Palliative care health professionals
- ◯ Genetics health professionals
- ◯ Oncology health professionals
- ◯ It has never been raised in my experience
- ◯ Not sure
- ◯ Other

Please explain.

_____

7a. Please indicate approximately how often you have been involved in the following activities in the last 12 months.

| | | Zero | Once or twice | Three to five times | Six to 10 times | More than 10 times | I have not worked clinically in the last 12 months |
|---|---|---|---|---|---|---|---|
| a. | | | | | | | |

| | | | | | | | |
|---|---|---|---|---|---|---|---|
| | Identifying a patient receiving palliative care who is eligible for DNA banking/testing. | ○ | ○ | ○ | ○ | ○ | ○ |
| b. | Initiating a discussion about DNA banking/testing with a patient receiving palliative care or their relative. | ○ | ○ | ○ | ○ | ○ | ○ |
| c. | Seeking advice from a genetics health professional about one of your palliative patients | ○ | ○ | ○ | ○ | ○ | ○ |
| d. | Referring a patient receiving palliative care to specialist genetics services. | ○ | ○ | ○ | ○ | ○ | ○ |
| e. | Taking consent for DNA banking/testing from a patient receiving palliative care. | ○ | ○ | ○ | ○ | ○ | ○ |
| f. | Taking consent for DNA banking/testing from a relative of a patient receiving palliative care. | ○ | ○ | ○ | ○ | ○ | ○ |

7b. Please indicate approximately how often you have been involved in the following activities in the last 12 months.

| | | Zero | Once or twice | Three to five times | Six to 10 times | More than 10 times | I have not worked clinically in the last 12 months |
|---|---|---|---|---|---|---|---|
| g. | Facilitating collection of a DNA sample from a patient receiving palliative care | ○ | ○ | ○ | ○ | ○ | ○ |
| h. | Disclosing genetic/genomic test results to a patient receiving palliative care | ○ | ○ | ○ | ○ | ○ | ○ |
| i. | Disclosing a deceased patient's genetic/genomic test results to bereaved relatives | ○ | ○ | ○ | ○ | ○ | ○ |
| j. | Being aware that I was caring for a palliative patient with an underlying genetic condition | ○ | ○ | ○ | ○ | ○ | ○ |
| k. | Checking if my palliative patient (or their relatives) had already had an opportunity to discuss genetics before coming into my care | ○ | ○ | ○ | ○ | ○ | ○ |

| | | |
|---|---|---|
| 8. | Which scenario best describes the availability of specialist genetics services for your current clinical area? | ◯ Specialist genetics services and palliative care services are embedded within the same hospital or group of hospitals.<br>◯ Specialist genetics services and palliative care services are not embedded within the hospital or group but are accessible to each other.<br>◯ Specialist genetics services and palliative care services are NOT accessible to each other.<br>◯ Palliative care services have access to private genetics services only<br>◯ Other<br>◯ Not sure |
| | Please explain | _____ |
| 9. | In your experience how is consent for DNA banking or testing obtained and documented? | ◯ Verbal consent only (undocumented)<br>◯ Verbal consent (documented in the patient's clinical records)<br>◯ Written consent using locally available paperwork (but not a formal genetics consent form)<br>◯ Written consent using a formal genetics consent form<br>◯ In my experience consent has not been taken<br>◯ Other |
| | Please explain. | _____ |

10. In your experience, what have been the main challenges for palliative care health professionals in facilitating DNA banking/testing? (Please choose up to 3 responses.)

* Please note: These response options have all been identified as challenges in prior studies. For the purpose of identifying priority areas, we are requesting you nominate your top three challenges

- ☐ Identifying eligible patients
- ☐ Urgency of the situation/referral
- ☐ Conflicting priorities between providing palliative care and facilitating genetic testing
- ☐ Obtaining informed consent
- ☐ Discomfort with initiating discussions about DNA banking/testing with patients or families
- ☐ Palliative care health professionals' lack of knowledge about DNA banking/testing or procedures
- ☐ Genetics health professionals' lack of knowledge of the procedure for consent and DNA banking
- ☐ The views or expectations of the family
- ☐ Lack of availability of specialist genetics services
- ☐ Communication difficulties between genetics and palliative care services
- ☐ Conflicting views within the palliative care team about the utility of DNA banking/testing for palliative patients, and their families
- ☐ Lack of resources
- ☐ Distress of the patient or family members
- ☐ Complex family dynamics
- ☐ Concern that a discussion about genetics could damage the therapeutic relationship
- ☐ Under-referral of palliative patients to genetics services
- ☐ In my experience DNA banking or genetic/genomic testing has not ever been considered
- ☐ Other

(You can choose up to 3 responses)

Please explain.

_____

11. Please indicate your level of confidence about the following:

| | | Not at all confident | Fairly unconfident | Neither confident or unconfident | Fairly confident | Confident |
|---|---|---|---|---|---|---|
| a. | Identifying patients who may be eligible for DNA banking/testing | ○ | ○ | ○ | ○ | ○ |
| b. | Discussing DNA banking with patients or their families | ○ | ○ | ○ | ○ | ○ |
| c. | Discussing DNA testing with patients or their families | ○ | ○ | ○ | ○ | ○ |
| d. | Contacting my local genetics service | ○ | ○ | ○ | ○ | ○ |
| e. | Taking a DNA sample for banking or testing | ○ | ○ | ○ | ○ | ○ |
| f. | Disclosing genetic/genomic test results to palliative care patients | ○ | ○ | ○ | ○ | ○ |
| g. | | | | | | |

| | | | | | | |
|---|---|---|---|---|---|---|
| | Disclosing genetic/genomic test results to bereaved families | ○ | ○ | ○ | ○ | ○ |
| h. | Knowing how to respond if a family member asks me about their genetic risk | ○ | ○ | ○ | ○ | ○ |
| i. | Knowing my legal responsibilities when sharing health information with family members when a patient is terminal or after they have died | ○ | ○ | ○ | ○ | ○ |
| j. | Assessing an appropriate time to broach a discussion about genetics | ○ | ○ | ○ | ○ | ○ |

12. What resources or tools have you found helpful when facilitating DNA banking/testing in the palliative care setting? (Please indicate all that apply)

- ☐ Web-based risk assessment tool
- ☐ Smart phone App
- ☐ Support from a palliative care colleague
- ☐ Contact with specialist genetics services
- ☐ Educational brochures
- ☐ Telephone information hotline,
- ☐ Face to face education
- ☐ Online education
- ☐ Clinical decision-making algorithm
- ☐ Clinical practice guidelines
- ☐ I have not found any resources or tools helpful
- ☐ Other

How frequently did you receive "face to face education" for DNA banking/testing in the palliative care setting?

_____

Which clinical practice guidelines did you find useful?

_____

Please explain.

_____

13. What additional resources or tools would be helpful, if any?

_____

14. Please indicate to what extent you agree or disagree with the following statements:

| | Strongly disagree | Disagree | Neither agree nor disagree | Agree | Strongly agree |
|---|---|---|---|---|---|
| a. | | | | | |

| | | | | | | |
|---|---|---|---|---|---|---|
| | Discussing DNA banking/testing with people receiving palliative care undermines the central ethos of palliative care in providing comfort and support at an emotionally vulnerable time. | ○ | ○ | ○ | ○ | ○ |
| b. | Patients may experience positive emotional benefits from being able to give a sample for DNA banking/testing for the possible future benefit of their relatives. | ○ | ○ | ○ | ○ | ○ |
| c. | Discussing DNA banking/testing may cause distress to the families by making them assume/fear their fate is pre-determined. | ○ | ○ | ○ | ○ | ○ |
| d. | DNA banking/testing will have been discussed by other health professionals before the patient is referred to palliative care. | ○ | ○ | ○ | ○ | ○ |
| e. | The priority in palliative care is to improve quality of life and relieve suffering and therefore it is not an appropriate time to discuss DNA banking/testing. | ○ | ○ | ○ | ○ | ○ |
| f. | DNA banking/testing of the patient may be important for the surviving relatives. | ○ | ○ | ○ | ○ | ○ |
| g. | It is not the responsibility of health professionals in palliative care to discuss DNA banking/testing. | ○ | ○ | ○ | ○ | ○ |
| h. | DNA banking/testing is not appropriate for people receiving palliative care because it will not help the patient. | ○ | ○ | ○ | ○ | ○ |

15. Please indicate to what extent you agree or disagree with the following statements:

| | | Strongly disagree | Disagree | Neither agree nor disagree | Agree | Strongly agree |
|---|---|---|---|---|---|---|
| a. | Palliative care health professionals are well placed to have discussions about DNA banking/testing with the family members of a palliative patient | ○ | ○ | ○ | ○ | ○ |
| b. | | | | | | |

| | | | | | | |
|---|---|---|---|---|---|---|
| | Concerns about genetic discrimination (eg. insurance and employment discrimination) make discussions about DNA banking/testing with palliative patients and their families difficult | ○ | ○ | ○ | ○ | ○ |
| c. | Families generally appreciate being told genetic information that is relevant to their health | ○ | ○ | ○ | ○ | ○ |
| d. | Discussions about DNA banking/testing need to be individualised to the palliative patient, and their families | ○ | ○ | ○ | ○ | ○ |
| e. | As time goes on, genetic health information will inevitably become part of the palliative care health professionals' scope of practice | ○ | ○ | ○ | ○ | ○ |
| f. | If a patient declines a discussion about DNA banking/testing, palliative care health professionals should revisit this discussion with palliative patients at a later date to see if they've changed their mind | ○ | ○ | ○ | ○ | ○ |
| g. | The family of a palliative patient have a right to know if they are at risk of developing a genetic disease, regardless of the palliative patient's wishes | ○ | ○ | ○ | ○ | ○ |
| h. | The right of a palliative patient to decline a discussion about DNA banking/testing are to be respected, regardless of the family's wishes | ○ | ○ | ○ | ○ | ○ |

16. In your opinion, which of these would most help palliative care health professionals to discuss and/or facilitate DNA banking/testing with palliative patients, and their families? (Please choose up to 3 responses)

* Please note: These response options have all been identified as facilitators in prior studies. For the purpose of identifying priority areas, we are requesting you nominate your top three facilitators

- ☐ Physically co-locating palliative care and genetics health professionals within a hospital or organisation
- ☐ Developing a specific referral template for palliative care patients to the genetics service that includes relevant family member details
- ☐ Embedding a genetic counsellor in the palliative care team
- ☐ Having both palliative care and genetics health professionals attending the same multidisciplinary team meetings
- ☐ Fostering a closer working relationships between palliative care and genetics health professionals
- ☐ Receiving genetics education from genetics health professionals, including ways of sensitively communicating with patients and families about genetics
- ☐ Policy guidance detailing how and when to discuss DNA banking/testing with palliative patients and their families
- ☐ Empowering palliative patients, and their families, to seek out DNA banking/testing for themselves
- ☐ Speaking directly to the genetics health professional about the palliative patient
- ☐ Improving the capability of electronic medical records to share relevant information between health professionals
- ☐ Collaborating with genetics health professionals to facilitate collection of a DNA sample from a palliative patient
- ☐ We shouldn't be discussing DNA banking/testing with palliative patients, or their families
- ☐ Other

Please explain

_____

17. Previous research suggests that patients who are suitable for DNA testing and close to end of life should be offered DNA banking by palliative care health professionals, rather than referring them to the genetics service for DNA testing.

We would like to understand your views about how this would work in practice.

Please indicate your comfort with palliative care health professionals performing the following actions:

| | | Very uncomfortable | Somewhat uncomfortable | Neutral | Somewhat comfortable | Very comfortable |
|---|---|---|---|---|---|---|
| a. | Introduce the idea of DNA banking with the patient and/or the family | ○ | ○ | ○ | ○ | ○ |
| b. | Obtain consent for DNA banking from the patient, or appointed representative | ○ | ○ | ○ | ○ | ○ |
| c. | | | | | | |

| | | | | | | |
|---|---|---|---|---|---|---|
| | Facilitate collection of DNA sample | ○ | ○ | ○ | ○ | ○ |
| d. | Organise for the DNA sample to be banked | ○ | ○ | ○ | ○ | ○ |
| e. | Instruct the family how to follow up with the genetics service | ○ | ○ | ○ | ○ | ○ |
| f. | Communicate family follow up plan to the genetics service | ○ | ○ | ○ | ○ | ○ |

Please describe any further thoughts you have about palliative care health professionals performing these actions

---

Thank you. We will now ask you some demographic questions.

18. What is your gender?

- ○ Female
- ○ Male
- ○ Self-described (please specify below)
- ○ Prefer not to say

3a Please describe your gender

---

19. What is your age group?

- ○ < 20
- ○ 20-24
- ○ 25-34
- ○ 35-44
- ○ 45-54
- ○ 55-64
- ○ 65 or older
- ○ Prefer to not say

20. What is your country of birth?

- ○ Australia
- ○ New Zealand
- ○ England
- ○ India
- ○ Phillipines
- ○ Vietnam
- ○ Italy
- ○ South Africa
- ○ Malaysia
- ○ Scotland
- ○ Other (please specify)
- ○ Prefer not to say

Please specify your country of birth

---

| | | |
|---|---|---|
| 21. | What cultural background or ethnicity do you identify with? (Select all that apply) | ☐ None<br>☐ Australian<br>☐ New Zealand<br>☐ English<br>☐ Irish<br>☐ Scottish<br>☐ Chinese<br>☐ Italian<br>☐ Indian<br>☐ German<br>☐ Greek<br>☐ Vietnamese<br>☐ Other (please specify)<br>☐ Prefer not to say<br>(You may select more than one) |
| | Please describe your cultural background or ethnicity | _____ |
| 22. | Which language do you mainly speak at home? (If more than one language, indicate the one that is spoken most often) | ◯ English<br>◯ Mandarin<br>◯ Arabic<br>◯ Cantonese<br>◯ Vietnamese<br>◯ Italian<br>◯ Greek<br>◯ Hindi<br>◯ Spanish<br>◯ Punjabi<br>◯ Other (please specify)<br>◯ Prefer not to say |
| | Please describe the language you mostly speak at home | _____ |
| 23. | What is your primary profession? | ◯ Medical<br>◯ Nursing<br>◯ Other |
| | Please explain. | _____ |
| 24. | How many years has it been since you qualified in your current profession? | ◯ Less than 2 years<br>◯ 2 to 5 years<br>◯ 6 to 10 years<br>◯ 11 to 15 years<br>◯ More than 15 years |
| 25. | What is your highest academic achievement? | ◯ PhD<br>◯ Master degree<br>◯ Bachelor degree<br>◯ Diploma<br>◯ Professional qualification<br>◯ Other |
| | Please explain. | _____ |

| | | |
|---|---|---|
| 26. | What is your primary area of specialty? | ◯ Oncology<br>◯ Palliative care<br>◯ Primary care<br>◯ Aged care<br>◯ Other |
| | Please explain. | _____ |
| 27. | Which best describes your job title? | ◯ Registered nurse<br>◯ Senior registered nurse (including nurse practitioner)<br>◯ Junior doctor (eg intern, resident or equivalent)<br>◯ Senior doctor (eg registrar, fellow or equivalent)<br>◯ Consultant doctor (including staff specialist or equivalent)<br>◯ Other (please specify) |
| | Please explain. | _____ |
| 28. | How long have you worked in your current specialist area? | ◯ Less than 2 years<br>◯ 2 to 5 years<br>◯ 6 to 10 years<br>◯ 11 to 15 years<br>◯ More than 15 years |
| 29. | Which country do you work in? | ◯ Australia<br>◯ New Zealand<br>◯ England<br>◯ Scotland<br>◯ Northern Ireland<br>◯ Wales<br>◯ Other |
| | Please explain. | _____ |
| 30. | Which sector do you usually work in? | ◯ Public<br>◯ Private<br>◯ Public and private<br>◯ Other |
| | Please explain. | _____ |
| 31. | What location do you usually work in? | ◯ City/metropolitan/urban<br>◯ Regional<br>◯ Rural<br>◯ Other |
| | Please explain. | _____ |

32. What setting do you usually work in?

- ◯ Hospital
- ◯ Hospice
- ◯ Community clinic
- ◯ Home care
- ◯ General Practice
- ◯ Other

Please explain.

_____

**Many thanks for your participation in this survey.**

**If you have any further comments, you may write them below.**

**When you are finished, please press submit.**

**Have a great day!**

33. Please add any further comments about facilitating DNA banking/testing with people receiving palliative care or their families.

34. Please tell us if you have any further comments about this survey or study.

## SUPPLEMENTARY FILE E7: DATA ANALYSIS PLAN

| PALLIATIVE CARE SURVEY ANALYSIS PLAN | | | | | |
|---|---|---|---|---|---|
| **QN #** | **QUESTION TYPE** | **QUESTION DESCRIPTION** | **STATISTICAL PLAN** | **JUSTIFICATION** | **SPECIFIC TESTS PLAN** |
| 1 - 2 | Radio buttons | Previously worked in clinical area and organisation that sent the survey | No analysis | For eligibility only | NA |
| 3. + branched questions | Radio buttons and check boxes (nominal data) | Genetics/genomics training received, areas studied, training type received, time since training, interest in receiving training, training type interested in, reason for not being interested | Descriptive statistics | Describe PC HPs genetics training received, including their interest in receiving training, how this would be delivered and understanding why they may not be interested in further training. | Raw numbers & percentages |
| 4. | Likert scale (ordinal) | Please indicate how often you are involved in the following activities in your current practice (Taking a family health history, Drawing a three-generation family tree (pedigree), Making a genetic risk assessment) | Descriptive statistics | Describe the frequency at which PC HPs are involved in the described activities | Raw numbers & percentages |
| 5. | Radio button (nominal data) | In your experience, at what point in the patient's trajectory is a family health history usually taken? | Descriptive statistics | Describe PC HPs perceptions of when a family health history is obtained in their practice | Raw numbers & percentages |
| 6. | Radio button (nominal data) | In your experience, who usually initiates requests for genetic testing? | Descriptive and inferential statistics | Describe PC HPs perceptions of who initiates requests for genetic testing and compare this response to the genetics health professionals to see if there is a difference in their experience | Chi-square test comparing all PC & genetics health professionals. Report raw numbers, percentages & 95% CI for overall groups, and report raw numbers, percentages & 95% CI between specialties |
| 7a & 7b | Likert scale (interval data) | Please indicate approximately how often you have been | Descriptive statistics | Describe the frequency at which PC HPs are involved in the described activities | Mean, median and standard deviation |

| | | | | | |
|---|---|---|---|---|---|
| | | involved in the following activities in the last 12 months (11 activities related to integrating genetics into their practice) | | | |
| 8. | Radio button (nominal data) | Which scenario best describes the availability of specialist genetics services for your current clinical area? | Descriptive statistics | Describe the availability of genetics services to PC HPs | Raw numbers and percentages/proportions |
| 9. | Radio button (nominal data) | In your experience, how is consent for DNA banking or testing obtained and documented? | Descriptive and inferential statistics | Describe the experience of PC HPs obtaining consent for DNA banking/testing and compare this experience to genetics HPs to see if there is a practice difference | Chi-square test & raw numbers, percentages/proportions |
| 10. | Check boxes (nominal data) | In your experience, what have been the main challenges for palliative care health professionals in facilitating DNA banking/testing? (Please choose up to 3 responses.)<br><br>*There are 18 listed options that participants can choose from* | Descriptive and inferential statistics | Describe PC HPs perception of the main challenges and compare these to genetics HPs to understand if there is a difference in views | For the 18 binary variables, do a chi-square test for each, or assess the most frequently selected options and do chi-square for these only.<br><br>Report a 'rate difference' (e.g. % of GCs reporting option - % of PC physicians) with a 95% confidence interval<br><br>Note: There is the risk that of multiplicity with 18 options – but so long as this is kept in mind when interpreting the results this is ok. For example if you would expect to probably see 1 'difference' just by chance, so if you end up only finding evidence of a difference for one of these options you would make the interpretation that your data is consistent with no important differences between professions. |
| 11. | Likert scale (ordinal data) | Please indicate your level of confidence about the following (10 scenarios related to | Descriptive statistics | Describe the level of confidence PC HPs have related to undertaking a variety of genetics-related activities. These activities | Frequency & proportions (perhaps mean?) |

| | | | | | |
|---|---|---|---|---|---|
| | | integrating genetics into practice) | | are not identical to the genetics HPs survey so will not compare this question. | |
| 12. | Checkboxes (nominal data) | What resources or tools have you found helpful when facilitating DNA banking or genetic/genomic testing in the palliative care setting? (Please indicate all that apply) | Descriptive statistics | Describe the resources PC HPs have found useful in their practice | Assess the most commonly selected options and report raw numbers, percentages |
| Not numbered (branched) | Free text box | How frequently did you receive "face to face education" for DNA banking/testing in the palliative care setting? | Content analysis | Collate and analyse responses to understand the frequency of which this training was received | Assess text response |
| Not numbered (branched) | Free text box | Which clinical practice guideline did you find useful? | Content analysis | Collate and analyse responses to understand the which clinical practice guidelines are useful and how often these are mentioned | Assess text response |
| 13. | Free text box | What additional resources or tools would be helpful, if any? | Content analysis | Collate any free text responses to understand whether there are other tools or resources PC HPs would find useful | Assess text response |
| 14 & 15 | Likert scale | Please indicate to what extent you agree or disagree with the following statements (8 x 2 statements that relate to views about the appropriateness of genetics in PC) | Descriptive and inferential statistics | Describe the views of PC HPs regarding the appropriateness of genetics in PC and compare these responses to the genetics HPs to understand whether their views align or diverge | Ordinal logistic regression (the OR is the odds of a 'shift' from one category (e.g.. very comfortable) to the next (e.g. somewhat comfortable). |
| 16. | Checkboxes (nominal data) | In your opinion, which of these would most help palliative care health professionals to discuss and/or facilitate DNA banking/testing with patients, and their families? (Please choose up to 3 responses) | Descriptive and inferential statistics | Describe the views of PC HPs about which facilitators would most help them to integrate genetics into their practice and compare these responses with genetics HPs to see if their views align or diverge | For the 13 binary variables, do a chi-square test for each, or assess the most frequently selected options and do chi-square for these only.<br><br>Report a 'rate difference' (e.g. % of GCs reporting option - % of PC physicians) with a 95% confidence interval<br><br>Note: There is the risk that of multiplicity with 13 options – but so long |

| | | | | | |
|---|---|---|---|---|---|
| | | | | | as this is kept in mind when interpreting the results this is ok. For example if you would expect to probably see 1 'difference' just by chance, so if you end up only finding evidence of a difference for one of these options you would make the interpretation that your data is consistent with no important differences between professions. |
| 17. | Likert scale (ordinal data) | Previous studies with genetics health professionals have suggested that they believe patients close to end of life should be offered DNA banking by palliative care health professionals, rather than referring them to the genetics service for DNA testing. We would like to understand your views about how this would work in practice. Please indicate your comfort with palliative care health professionals performing the following actions: 6 actions that represent the model of care described by genetics HPs in their qual study) | Descriptive and inferential statistics | Describe the views of PC HPs regarding the actions in this question to understand whether it is a feasible model of care. Compare these responses to genetics HPs to understand whether these views align or diverge. | Ordinal logistic regression |
| Not numbered | Free text box | Please describe any further thoughts you have about palliative care health professionals performing these actions | Content analysis | Collate and analyse any free text responses here to provide further details about PC HPs views related to this model of care | Assess text response |
| 18 - 32 | Radio buttons and check boxes | Demographic questions | Descriptive statistics | Describe population and compare to available data from palliative care organisations that distribute survey | Raw numbers & percentages. Using a table, compare matched baseline demographics between participants and |

| | | | | | |
|---|---|---|---|---|---|
| | (nominal, ordinal & interval data) | | | | data provided by PCNA, PCNNZ & ANZSPM. Formal inferential approach not recommended |
| 33. | Free text box | Please add any further comments about facilitating DNA banking/testing with people receiving palliative care or their families. | Content analysis | Collate responses related to a. views and b. experiences AND/OR a. barriers and b. facilitators and analyse to provide further info about views/experiences of PC HPs in facilitating DNA banking/testing | Assess text response |
| 34. | Free text box | Please tell us if you have any further comments about this survey or study. | Content analysis | Collate and analyse free text options here | Assess text response |
| GENETICS HEALTH PROFESSIONALS SURVEY DATA ANALYSIS PLAN | | | | | |
| **QN #** | **QUESTION TYPE** | **QUESTION DESCRIPTION** | **STATISTICAL PLAN** | **JUSTIFICATION** | **SPECIFIC TESTS PLAN** |
| 1 - 2 | Radio buttons | Previously worked in clinical area and organisation that sent the survey | No analysis | For eligibility only | NA |
| 3 + branched questions | Radio buttons and check boxes (nominal data) | Training received, areas studied, training type received, time since training, interest in receiving training, training type interested in, reason for not being interested | Descriptive statistics | Describe genetics HPs experiences in training received around communication at end of life, including their interest in receiving training, how this would be delivered and understanding why they may not be interested in further training. | Raw numbers & percentages |
| 4. | Radio button (nominal data) | Have you ever been involved in facilitating DNA banking/testing for people receiving palliative care? (yes, no, not sure) | Descriptive statistics | Report simple description about whether genetics HPs are involved in facilitating DNA banking/testing | Raw numbers & percentages/proportions |
| 4a. (branched from 17) | Radio button (nominal data) | In your experience, at what point did you usually become involved? | Descriptive statistics | Report proportion of genetics HPs who become involved at different time points | Raw numbers & percentages/proportions |
| 5. | Radio button (nominal data) | In your experience, who usually initiates requests for genetic testing? | Descriptive and inferential statistics | Describe genetics HPs perceptions of who initiates requests for genetic testing and compare this response to the PC health professionals to see if there is a difference in their experience | Chi-square test comparing all PC & genetics health professionals. Report raw numbers, percentages & 95% CI for overall groups, and report raw numbers, percentages & 95% CI for each professional group (eg. GC, clinical |

| | | | | | |
|---|---|---|---|---|---|
| | | | | | geneticist etc). Convert job_titlev1 variables into combined groups first |
| 6a & 6b | Likert scale (interval data) | Please indicate approximately how often you have been involved in the following activities in the last 12 months (10 activities related to integrating genetics into their practice) | Descriptive statistics | Describe the frequency at which genetics HPs are involved in the described activities | Mean, median, standard deviation |
| 7. | Radio button (nominal data) | Which scenario best describes the availability of specialist genetics services for people receiving palliative care in your area? | Descriptive statistics | Describe genetics HPs understanding of the availability of genetics services to people receiving palliative care | Raw numbers & proportions/percentages |
| 8. | Radio button (nominal data) | In your experience, how is consent for DNA banking or testing obtained and documented? | Descriptive and inferential statistics | Describe the experience of genetics HPs obtaining consent for DNA banking/testing and compare this experience to PC HPs to see if there is a practice difference | Chi-square test & raw numbers, percentages/proportions |
| 9. | Check boxes (nominal data) | In your experience, what have been the main challenges for palliative care health professionals in facilitating DNA banking/testing? (Please choose up to 3 responses.)<br><br>*There are 18 listed options that participants can choose from* | Descriptive and inferential statistics | Describe genetics HPs perception of the main challenges and compare these to PC HPs to understand if there is a difference in views | For the 18 binary variables, do a chi-square test for each, or assess the most frequently selected options and do chi-square for these only.<br><br>Report a 'rate difference' (e.g. % of GCs reporting option - % of PC physicians) with a 95% confidence interval<br><br>Note: There is the risk that of multiplicity with 18 options – but so long as this is kept in mind when interpreting the results this is ok. For example if you would expect to probably see 1 'difference' just by chance, so if you end up only finding evidence of a difference for one of these options you would make the interpretation that your data |

| | | | | | |
|---|---|---|---|---|---|
| | | | | | is consistent with no important differences between professions. |
| 10. | Likert scale (ordinal data) | Please indicate your level of confidence about the following (10 scenarios related to communicating with patients about genetics at end of life) | Descriptive statistics | Describe the level of confidence genetics HPs have related to communicating with PC patients and undertaking a variety of genetics-related activities. These activities are not identical between the two surveys so will not compare this question. | Frequency & proportion/percentages |
| 11. | Checkboxes (nominal data) | What resources or tools have you found helpful when facilitating DNA banking or genetic/genomic testing in the palliative care setting? (Please indicate all that apply) | Descriptive statistics | Describe the resources genetics HPs have found useful in their practice | Assess the most commonly selected options and report raw numbers, percentages |
| Not numbered. (branched from Q11) | Free text box | How frequently did you receive "face to face education" for communicating with patients receiving palliative care and/or their families? | Content analysis | Collate and analyse responses to understand the frequency of which this training was received | Assess text response |
| Not numbered. (branched from Q11) | Free text box | Which clinical practice guideline did you find useful? | Content analysis | Collate and analyse responses to understand the which clinical practice guidelines are useful and how often these are mentioned | Assess text response |
| 12. | Free text box | What additional resources or tools would be helpful, if any? | Content analysis | Collate any free text responses to understand whether there are other tools or resources PC HPs would find useful | Assess text response |
| 13 & 14 | Likert scale (ordinal data) | Please indicate to what extent you agree or disagree with the following statements (8 x 2 statements that relate to views about the appropriateness of genetics in PC) | Descriptive and inferential statistics | Describe the views of genetics HPs regarding the appropriateness of genetics in PC and compare these responses to the PC HPs to understand whether their views align or diverge | Ordinal logistic regression (the OR is the odds of a 'shift' from one category (e.g.. very comfortable) to the next (e.g. somewhat comfortable). |
| 15. | Checkboxes (nominal data) | In your opinion, which of these would most help genetics health professionals to discuss and/or facilitate DNA | Descriptive and inferential statistics | Describe the views of genetics HPs about which facilitators would most help them to integrate genetics into their practice and | For the 13 binary variables, do a chi-square test for each, or assess the most frequently selected options and do chi-square for these only. |

| | | | | | |
|---|---|---|---|---|---|
| | | banking/testing with palliative patients, and their families? (Please choose up to 3 responses) | | compare these responses with genetics HPs to see if their views align or diverge | Report a 'rate difference' (e.g. % of GCs reporting option - % of PC physicians) with a 95% confidence interval<br><br>Note: There is the risk that of multiplicity with 13 options – but so long as this is kept in mind when interpreting the results this is ok. For example if you would expect to probably see 1 'difference' just by chance, so if you end up only finding evidence of a difference for one of these options you would make the interpretation that your data is consistent with no important differences between professions. |
| 16. | Likert scale (ordinal data) | Previous studies with genetics health professionals have suggested that they believe patients close to end of life should be offered DNA banking by palliative care health professionals, rather than referring them to the genetics service for DNA testing. We would like to understand your views about how this would work in practice. Please indicate your comfort with palliative care health professionals performing the following actions: 6 actions that represent the model of care described by genetics HPs in their qual study) | Descriptive and inferential statistics | Describe the views of genetics HPs regarding the actions in this question to understand whether it is a feasible model of care. Compare these responses to PC HPs to understand whether these views align or diverge. | Ordinal logistic regression |

| | | | | | |
|---|---|---|---|---|---|
| Not numbered | Free text box | Please describe any further thoughts you have about palliative care health professionals performing these actions | Content analysis | Collate and analyse any free text responses here to provide further details about genetics HPs views related to this model of care | Assess text response |
| 17 - 31 | Radio buttons and check boxes (nominal and ordinal) | Demographic questions | Descriptive statistics (& inferential statistics??) | Describe population and compare to available data from genetics organisations that distribute survey | Raw numbers & percentages. Using a table, compare matched baseline demographics between participants and data provided by HGSA. Formal inferential approach not recommended |
| 32. | Free text box | Please add any further comments about facilitating DNA banking/testing with people receiving palliative care or their families. | Content analysis | Collate responses related to a. views and b. experiences AND/OR a. barriers and b. facilitators and analyse to provide further info about views/experiences of genetics HPs in facilitating DNA banking/testing with palliative patients | Assess text response |
| 33. | Free text box | Please tell us if you have any further comments about this survey or study. | Content analysis | Collate and analyse free text options here | Assess text response |

## SUPPLEMENTARY FILE E8: FULL LIST OF BARRIERS, FACILITATORS, AND RESOURCES/TOOLS

**Table A. Participants were asked to select their top 3 challenges (barriers) to integrating genetics into the care of people with palliative care needs and their families.**

| **Challenge descriptions** | TOTAL (*n*=72) | | G-HP (*n*=29) | | PC-HP (*n*=43) | | p-value |
|---|---|---|---|---|---|---|---|
| | n | % | n | % | n | % | |
| Palliative care HPs' lack of knowledge | 32 | 44 | 13 | 45 | 19 | 44 | 1 |
| Identifying eligible patients* | 19 | 26 | 4 | 14 | 15 | 35 | 0.046# |
| Conflicting priorities between providing palliative care and genetic testing | 15 | 21 | 6 | 21 | 9 | 21 | 1 |
| Under-referral of palliative patients to genetics* | 15 | 21 | 12 | 41 | 3 | 7 | <0.001# |
| Urgency of the situation/referral | 13 | 18 | 8 | 28 | 5 | 12 | 0.119 |
| In my experience DNA storage or testing has not been considered* | 11 | 15 | 1 | 3 | 10 | 23 | 0.041# |
| Discomfort with initiating DNA storage/testing discussions | 10 | 14 | 6 | 21 | 4 | 9 | 0.187 |
| Obtaining informed consent | 6 | 8 | 4 | 14 | 2 | 5 | 0.212 |
| Conflicting views within the palliative care team about DNA banking/testing* | 6 | 8 | 5 | 17 | 1 | 2 | 0.036# |
| The views or expectations of the family | 5 | 7 | 4 | 14 | 1 | 2 | 0.15 |
| Communication between genetics and palliative care services* | 5 | 7 | 5 | 5 | 0 | 0 | 0.008# |
| Distress of the patient or family members | 5 | 7 | 2 | 7 | 3 | 7 | 1 |
| Lack of specialist genetics services | 4 | 6 | 0 | 0 | 4 | 9 | 0.143 |
| Lack of resources | 4 | 6 | 2 | 7 | 2 | 5 | 1 |
| Complex family dynamics | 4 | 6 | 0 | 0 | 4 | 9 | 0.143 |
| Other | 3 | 4 | 1 | 3 | 2 | 5 | 1 |
| Concerns about harming the therapeutic relationship | 1 | 1 | 0 | 0 | 1 | 2 | 1 |
| Genetics HPs' lack of knowledge | 0 | 0 | 0 | 0 | 0 | 0 | NA |

| **Table B. Participants were asked to select their top 3 facilitators to integrating genetics into the care of people with palliative care needs and their families.** | | | | | | | |
|---|---|---|---|---|---|---|---|
| **Facilitator description** | TOTAL (*n*=72) | | G-HPs (*n*=29) | | PC-HPs (*n*=43) | | p-value |
| | **n** | **%** | **n** | **%** | **n** | **%** | |
| Developing a specific genetic referral template for palliative care patients | 31 | 43 | 9 | 31 | 22 | 51 | 0.145 |
| Fostering closer working relationships between palliative care & genetics health professional | 27 | 38 | 15 | 52 | 12 | 28 | 0.05 |
| Genetics health professionals deliver education to palliative care health professionals | 25 | 35 | 11 | 38 | 14 | 33 | 0.801 |
| Embedding a genetic counsellor in the palliative care team | 17 | 24 | 8 | 28 | 9 | 21 | 0.578 |
| Palliative care & genetics health professional attend the same multidisciplinary team meetings* | 15 | 21 | 11 | 38 | 4 | 9 | 0.006 |
| Policy guidance for discussing DNA banking/testing | 12 | 17 | 6 | 21 | 6 | 14 | 0.455 |
| Physically co-locating palliative care and genetics services | 8 | 11 | 2 | 7 | 6 | 14 | 0.461 |
| Empowering palliative patients and families to seek out DNA banking/testing for themselves | 7 | 10 | 1 | 3 | 6 | 14 | 0.23 |
| Speaking directly to the palliative care health professional about the palliative patient | 7 | 10 | 2 | 7 | 5 | 12 | 0.694 |
| Collaborating with genetics health professional to facilitate collection of a DNA sample | 7 | 10 | 5 | 17 | 2 | 5 | 0.11 |
| Improving electronic medical record capabilities | 2 | 3 | 1 | 3 | 1 | 2 | 1 |
| Other | 2 | 3 | 0 | 0 | 2 | 5 | 0.512 |
| We shouldn't be discussing DNA banking / testing with palliative patients, or their families | 1 | 1 | 0 | 0 | 1 | 2 | 1 |

| **Table C. Participants were asked to select the most useful resources or tools to support them to integrate genetics into the care of people with palliative care needs and their families.** | | | | | | |
|---|---|---|---|---|---|---|
| **Resources or tools to support HPs to facilitate DNA banking or testing** | TOTAL (n=72) | | G-HP (n=29) | | PC-HP (n=43) | |
| | **n** | **%** | **n** | **%** | **n** | **%** |
| Support from a specialist genetics service or colleague | 33 | 46 | 19 | 66 | 14 | 33 |
| Support from a palliative care colleague | 15 | 21 | 9 | 31 | 6 | 14 |
| I have not found any resources or tools helpful | 10 | 14 | 3 | 10 | 7 | 16 |
| Other/no experience | 10 | 14 | 1 | 3 | 9 | 21 |
| Clinical decision-making algorithm or guideline | 9 | 13 | 3 | 10 | 6 | 14 |
| Web-based risk assessment tool | 7 | 10 | 4 | 14 | 3 | 7 |
| Educational brochures | 6 | 8 | 2 | 7 | 4 | 9 |
| Face to face or online education | 5 | 7 | 2 | 7 | 3 | 7 |
| Telephone information hotline | 3 | 4 | 1 | 3 | 2 | 5 |
| Smart phone App | 1 | 1 | 0 | 0 | 1 | 2 |

## SUPPLEMENTARY FILE E9: STROBE STATEMENT

| Checklist of items that should be included in reports of cross-sectional studies. Adapted from von Elm E, Altman DG, Egger M, Pocock SJ, Gøtzsche PC, Vandenbroucke JP. The strengthening the reporting of observational studies in epidemiology (STROBE) statement: guidelines for reporting observational studies. Epidemiology. 2007;18(6):800-4. | | | |
|---|---|---|---|
| **Item** | **Item** | **Recommendation** | **Page number** |
| Title and abstract | 1 | (a) Indicate the study's design with a commonly used term in the title or the abstract | 110 |
| | | (b) Provide in the abstract an informative and balanced summary of what was done and what was found | 110 |
| Introduction | | | |
| Background/rationale | 2 | Explain the scientific background and rationale for the investigation being reported | 110-112 |
| Objectives | 3 | State specific objectives, including any prespecified hypotheses | 112 |
| Methods | | | |
| Study design | 4 | Present key elements of study design early in the paper | 112 |
| Setting | 5 | Describe the setting, locations, and relevant dates, including periods of recruitment, exposure, follow-up, and data collection | 112-113 |
| Participants | 6 | (a) Give the eligibility criteria, and the sources and methods of selection of participants | 112-113 |
| Variables | 7 | Clearly define all outcomes, exposures, predictors, potential confounders, and effect modifiers. Give diagnostic criteria, if applicable | 113-114<br>Appendix E5 & E6 |
| Data sources/ measurement | 8* | For each variable of interest, give sources of data and details of methods of assessment (measurement). Describe comparability of assessment methods if there is more than one group | 114 |
| Bias | 9 | Describe any efforts to address potential sources of bias | 125-126 |
| Study size | 10 | Explain how the study size was arrived at | 112 |
| Quantitative variables | 11 | Explain how quantitative variables were handled in the analyses. If applicable, describe which groupings were chosen and why | 114 |
| Statistical methods | 12 | (a) Describe all statistical methods, including those used to control for confounding | 114 |

| | | | |
|---|---|---|---|
| | | (b) Describe any methods used to examine subgroups and interactions | |
| | | (c) Explain how missing data were addressed | |
| | | (d) If applicable, describe analytical methods taking account of sampling strategy | NA |
| | | (e) Describe any sensitivity analyses | NA |
| Results | | | |
| Participants | 13* | (a) Report numbers of individuals at each stage of study—eg numbers potentially eligible, examined for eligibility, confirmed eligible, included in the study, completing follow-up, and analysed | 114-116 |
| | | (b) Give reasons for non-participation at each stage | NA |
| | | (c) Consider use of a flow diagram | NA |
| Descriptive data | 14* | (a) Give characteristics of study participants (eg demographic, clinical, social) and information on exposures and potential confounders | 114-116 |
| | | (b) Indicate number of participants with missing data for each variable of interest | 114-122 |
| Outcome data | 15* | Report numbers of outcome events or summary measures | 114-122 |
| Main results | 16 | (a) Give unadjusted estimates and, if applicable, confounder-adjusted estimates and their precision (eg, 95% confidence interval). Make clear which confounders were adjusted for and why they were included | 114-122 |
| | | (b) Report category boundaries when continuous variables were categorized | NA |
| | | (c) If relevant, consider translating estimates of relative risk into absolute risk for a meaningful time period | NA |
| Other analyses | 17 | Report other analyses done—eg analyses of subgroups and interactions, and sensitivity analyses | NA |
| Discussion | | | |
| Key results | 18 | Summarise key results with reference to study objectives | 123-125 |
| Limitations | 19 | Discuss limitations of the study, taking into account sources of potential bias or imprecision. Discuss both direction and magnitude of any potential bias | 125-126 |
| Interpretation | 20 | Give a cautious overall interpretation of results considering objectives, limitations, multiplicity of analyses, results from similar studies, and other relevant evidence | 123-125 |
| Generalisability | 21 | Discuss the generalisability (external validity) of the study results | 125-126 |

| | | | |
|---|---|---|---|
| Other information | | | |
| Funding | 22 | Give the source of funding and the role of the funders for the present study and, if applicable, for the original study on which the present article is based | Not presented in chapter 5. Funding provided in the form of a PhD stipend by the Translational Cancer Research Network. |