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Family communication and results disclosure after germline sequencing: A mixed methods study

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ABSTRACT

Objective: Research on family communication of germline genome sequencing (GS) results (versus of genetic results after targeted genetic testing) is still emerging, yet potentially complex results increase the importance of communicating risk to relatives. Promoting equity by ensuring patients have sufficient health literacy to interpret results is important in this context. This study aimed to identify cancer patients' perceived importance of result disclosure, predictors of perceptions, and perspectives on family communication.

Methods: This explanatory-sequential, cross-sectional mixed-methods study involved participants (n = 246) completing a questionnaire and (n = 20) a semi-structured interview. Ordinal logistic regressions determined associations between potential predictors and perceived importance of result disclosure. Interview transcripts were analysed thematically using a constant-comparative approach.

Results: More participants intended disclosing to nuclear (77.4%) than to extended family (42.7%). More than half (59.3%) felt results were family information; 62.7% believed it was important to disclose results to family members. Nuclear and extended family communication scores and education level were significantly positively associated with perceived importance of disclosure ($p < 0.05$). Six qualitative themes were identified: i) Responsibility to inform, ii) Choice, iii) Autonomy, iv) Family Communication, v) Significance of results, and vi) Health professional role.

Conclusion: Low health literacy and family conflict can complicate communication of GS results. Patients seek clear, interpretable information in a format they can easily communicate.

Practice implications: Healthcare professionals can facilitate discussion of GS results by offering written information, encouraging disclosure, exploring existing family dynamics and communication patterns, and offering strategies to improve family communication. Centralised genetic communication offices and chatbots can also be helpful

1. Introduction

Germline genome sequencing (GS) maps the DNA sequence of the whole genome and has potential to generate multiple results with varying health implications for both the individual tested and biological relatives [1,2]. Cancer patients who undergo GS may receive information that is relevant to the target cancer or other cancers or diseases (secondary findings) or are of unknown significance.

Family communication about GS results to relatives is important as it can trigger confirmatory testing in relatives and subsequent risk management. However, only around 30% of at-risk relatives undergo genetic testing [3], possibly due to inadequate family communication [4].

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Systematic reviews [5,6] have noted that probands may worry about potential effects of disclosure on relatives and family relationships and have difficulty deciding what and when to disclose. The latter finding is consistent with Family Systems Theory (FST) [7], which suggests that family relationships, function, and the broader social context in which families exist can impact individual family member's behaviour. Rolland [8] has also suggested that open versus closed communication and emotional distance in families may influence genomic communication.

A number of studies have confirmed a link between general family communication and sharing of health information amongst families. Hovick et al. [9] found that, in concordance with FST, families with more open communication and greater parental control were more likely to process health history messages, and in turn, to intend to seek health information from family members. Similarly, Campbelle-Salome et al. [10] found that open family communication encouraged collection of family health history, particularly amongst women, while families in which hierarchy and homogeneity of beliefs were stressed were less open in communication and collection of family health history. However, these variables have not been explored widely in relation to communication of GS results specifically.

Another potential predictor of willingness to disclose GS results to relatives is low health literacy [11]. Individuals with poor health literacy lack skills in understanding and applying information about health issues, which may erode their confidence in communicating genetic results [12]. To promote equitable uptake and implementation of GS, while avoiding deepening health disparities between disadvantaged groups, it is critical to explore the impact of health literacy [13] on family communication of GS results. Kaphinski et al. [14] found that individuals with low health literacy perceived family health history to be less important, but no research has focused specifically on the impact of health literacy on intended family communication of GS results.

While family communication intentions have been explored extensively with regards to cancer genetic results, intentions in the context of GS, where unexpected and non-cancer-related genes may be discovered, are less well understood. Further, cancer patients' perceptions of the role of health professionals in facilitating this process have not been well explored. In many countries (including Australia and the US), health professionals are not legally allowed to directly provide genetic results to family members, however they can support the proband in communicating with family. Thus, in this study, we aimed to investigate cancer patients'**: i) perceived importance of disclosing GS results to relatives; ii) intended communication of positive GS results to both first-degree relatives (immediate family members including a spouse or de facto partner, child, parent, grandparent, grandchild or sibling) and more distant relatives (such as cousins);** iii) family and personal characteristics associated with willingness to disclose GS results; iv) perceived facilitators and barriers to family disclosure; and v) patients' perspectives of the role of healthcare professionals (HPs) in promoting communication of germline GS results within families.

2. Methods

2.1. Study setting and design

This study, the Psychosocial Issues in Genomic Oncology (PiGeOn) study, [15] is a sub-study of the Genetic Cancer Risk in the Young (RisC) study. The RisC study (ongoing) offers GS to participants with a cancer of likely genetic origin to identify the origin of their cancer and/or determine risk of another cancer/illness. Participants indicate whether they would like to be informed if they have a pathogenic variant that increases the likelihood of cancer and/or are found to have a secondary finding that may be important to their health. If participants are found to carry a pathogenic gene variant and have chosen to be informed, they are referred to a genetic counsellor. If the variant is cancer-related, participants could be eligible to enroll in a related cancer surveillance trial. No results had been given at the time of this study. PiGeOn investigated psychosocial issues in RisC participants [15] using a longitudinal mixed methods sequential design. PiGeOn examined a wide range of psychosocial issues, many of which have been published elsewhere e.g. [16–19]. The current paper is based on a second wave of PiGeOn qualitative and quantitative data collection including more targeted questions about result disclosure and family communication.

This study was approved by the St Vincent's Hospital Human Research Ethics Committee (HREC/16/SVH/24) and the University of Technology Human Research Ethics Committee (ETH21–6522).

2.2. Participants

Eligibility criteria for enrolment into the RisC study included: i) a histologically confirmed malignancy, ii) aged 16–40 years at diagnosis OR having > 1 primary cancer diagnosed < 50 years of age OR having > 2 primary cancers at any age. Exclusion criteria were: i) non- melanoma skin cancer diagnosis and ii) inability to meet study requirements. Recruitment to RisC took place at the centre in which participants received clinical care, or they were referred by a consumer advocacy organisation (Rare Cancers Australia), clinical geneticists, or genetic counsellors. The RisC coordinator provided information and gained written consent to both RisC and PiGeOn. Eligibility criteria for PiGeOn included eligibility for RisC, as well as sufficient English to complete questionnaires and interviews.

2.3. Procedure

All participants completed a questionnaire after consent to RiSC/PiGeON and before GS, and a subset (of those who completed the questionnaire) completed a subsequent semi-structured interview a few days after their questionnaire was completed. The interview explored participants' questionnaire responses in greater detail, with rationales and further detail about their questionnaire responses elicited. Note, no participant received results within the study time-frame, so questions about disclosure were hypothetical.

2.4. Measures

2.4.1. Quantitative

The primary outcome was a 5-point Likert scale measuring the degree to which participants agreed with the statement *"It is important for me to disclose my whole genome sequencing results to my relatives"* (higher scores indicate greater agreement). The result type was not specified in this question, but participants had previously consented (or not) to receipt of results indicating a variant of significance to their cancer or health more broadly. Other data collected included demographic and cancer characteristics, genome knowledge (Knowledge of Genome Sequencing [KOGS] questionnaire [20]: scores range from 0 to 9, higher scores indicate better knowledge, health literacy (Short Test of Functional Health Literacy in Adults [S-TOFHLA]) [21]: scores were dichotomised: scores > 10 show low health literacy, quality of family communication (Family Communication Scale [FCS] [22]: scores range from 1 to 25, high scores indicate better communication, perceived results disclosure responsibility (adapted from Gilbar & Barnoy [23]: scores 1–4 with higher scores indicating greater patient responsibility, family communication motivation and barriers (adapted from Kohut et al. [24]), and HP communication assistance, motivation and barriers (adapted from McGivern et al. [25]). For more detail on measures, see [15].

2.4.2. Qualitative

Interview questions asked respondents to further expand on questionnaire responses regarding intended family communication about GS (see Supplementary File 2) to explore these issues in-depth. Phone interviews lasting on average 30 min were conducted by an experienced, qualitative Psycho-Oncology researcher (NB or SV), audio-recorded and

transcribed verbatim.

2.5. Sample size

Sample size (a minimum of n = 245) for quantitative data was calculated to allow exploration of up to ten predictors of perceived importance of disclosing results to relatives (the primary outcome) (with at least 20 responses per predictor). Interviewees were purposively sampled in terms of age and gender to ensure sample heterogeneity. Recruitment continued until data saturation was reached, i.e., the point at which no new information emerged after three consecutive interviews, which was achieved at $N = 20$.

2.5.1. Data analysis: quantitative

Following testing for statistical assumptions, univariate and multivariate ordinal logistic regressions were conducted in the Statistical Package for Social Sciences (SPSS) to determine associations between the primary outcome and potential predictors, including the nuclear and extended Family Communication Score (FCS) as well as age, sex, ethnicity, number of biological children, health literacy score, previous family cancer clinic experience, incidence of cancer, and family 'closeness'. Univariate results are presented in Supplementary File 1. A threshold of $p < 0.30$ was set to determine inclusion in the multivariate regression. Variables included were nuclear ($p < 0.001$) and extended ($p < 0.001$) FCS, age, sex, cancer incidence, number of children, and education. As responses to the primary outcome were skewed, we conducted logistic regressions, with the variable dichotomised as very important or important, versus other responses.

2.5.2. Data analysis: qualitative

Interview transcripts were analysed thematically, with emerging themes generated using the constant comparative approach [26]. Initial familiarisation with the data was followed by generating codes, searching for themes, reviewing and refining candidate themes, and defining and naming themes. Line-by-line coding was independently carried out on nine transcripts by the research team. Two coders (CH and NB) coded the remaining transcripts and illustrative quotes were extracted. Any discrepancies were resolved through discussion with the entire team. While analysing themes, we looked for, but did not detect, systematic differences according to the age and gender of participants.

Qualitative and quantitative data were then integrated by comparing and contrasting questionnaire responses with related qualitative responses, in accordance with mixed method procedures [27].

3. Results

A total of 264 participants completed the questionnaire. See Table 1 for demographic characteristics. Mean age of participants was 41.6 years, with 61% female. Mean health literacy was classified in the 'sufficient' category, with a score of 9.71 out of 12. All participants elected to receive GS results that had implications for their health.

3.1. Participants' perspectives on GS result disclosure to relatives

Over half of participants (62.7%) perceived it was important to disclose GS results to relatives, believing results to be more family information (59.3%) than personal private information (41.3%). More participants believed the responsibility for disclosing results to relatives was mostly or all the patient's responsibility (52.3%) versus to be equally shared between patient and HP (38.3%), or all the HP's responsibility (9.5%). The majority of the sample (75.5%) reported they would not have difficulty disclosing results to relatives, but more participants were likely to share *all* information with their first-degree relatives (77.4%) compared with their extended family members (42.7%). Most participants indicated they would share results with relatives if they were at increased risk of treatable/preventable (83.2%) conditions as opposed to *early* (77.1%) or *late* (73.6%) onset non-treatable conditions or if no pathogenic changes were found (67.9%).

Table 1
Participant Demographic Characteristics (n = 264).

Characteristic	Participants, n (%)
Sex at Birth	
Female	160 (60.6)
Male	104 (39.4)
Age in Years	
<30	39 (14.8)
30–45	151 (57.2)
46–60	37 (14.0)
61–75	32 (12.1)
> 75	5 (1.9)
Marital Status	
Married	137 (51.9)
Single	65 (24.6)
De facto	36 (13.6)
Widowed	5 (1.9)
Separated	5 (1.9)
Divorced	16 (6.0)
Highest Level of Education	
Primary School	0 (0)
Secondary school – year 7–10	27 (10.2)
Secondary school – year 11 or 12	43 (16.3)
Vocational training	57 (21.6)
University – did not graduate	21 (7.9)
University - graduated	115 (43.6)
Don't Know	0 (0)
Number of Biological Children	
None	105 (39.8)
1–6	159 (60.2)
Socioeconomic Status (1–10)	
1–4	59 (22.3)
5–7	84 (31.8)
8–10	121 (45.8)
Cancer Incidence	
Common	77 (29.2)
Less common	10 (3.8)
Rare	177 (67.0)
Medical or Science Occupation	
Yes	23 (8.7)
No	241 (91.3)
Previous Genetic Counselling appointment	
Yes	108 (40.9)
No	146 (55.3)
Don't Know	8 (3.0)
Health Literacy Score (HL)	
n (n missing)	261(3)
Mean (*SD*)	9.71(*2.55*)

3.2. Family communication

Scores on the FCS were high and indicated significantly better communication for the nuclear (m = 39.9 ± 0.86) versus extended (m = 35.2 ± 1.01) family, $p < 0.001$.

3.3. Factors associated with perceived importance of disclosing GS results to relatives

A multivariate ordinal logistic regression (see Table 2) indicated FCS nuclear family [OR= 1.08 (1.05, 1.12, $p = 0.007$)], FCS extended family [OR= 1.07 (1.04, 1.10, $p = 0.03$)] and education [OR= 1.56 (1.04, 2.07, $p = 0.03$)] were significantly associated with perceived importance of disclosing GS results to relatives.

3.4. Qualitative results

Six overarching themes were identified from qualitative analysis, discussed below. For additional supporting quotes, please see Supplementary file 2.

Table 2
Ordinal Logistic Regression Results.

Variable	Odds Ratio, OR (95%CI)	*p* value (3 sf.)	*t* value
FCS nuclear	1.082 (1.049, 1.115)	0.00653 * *	53.62
FCS extended	1.067 (1.036, 1.097)	0.0272 *	57.65
Education	1.557 (1.040, 2.074)	0.0359 *	6.53
Cancer Incidence	0.708 (0.242, 1.174)	0.0970	2.51
Age	1.022 (1.005, 1.038)	0.102	98.08
Number of Children	1.287 (1.104, 1.470)	0.324	9.94
GC Appt	0.796 (0.338, 1.253)	0.329	3.41
Med/Sci Occupation	0.825 (0.052, 1.598)	0.624	2.10
Sex	0.755 (0.308, 1.207)	0.780	3.77
Health Literacy	0.995 (0.908, 1.083)	0.918	22.5

* $p < 0.05$
* * $p < 0.01$

3.4.1. Responsibility for, and mutual benefit from, informing family

Most participants considered it their responsibility to inform relatives of GS results. Several participants felt it was their moral obligation to inform relatives of results due to shared ownership of genetic information about which relatives had a right to know and so that relatives could optimise their health management or explain the cause of their own cancer.

> *"So it's an ethical thing…it's that moral decision…to let them know if you have the information" (ID7)*

> *"I have my own DNA, but I also share that with someone…they're all linked because you're family…that information also impacts on this person. And how they might live their life."(ID19)*

Several participants explained they assumed relatives who had been emotionally invested in their cancer journey to date would want this information, based on a sense of reciprocity and mutuality.

> *"…if they were the patient and it was the other way around, I would also hope they would tell me if it concerns me as well" (ID9)*

> *"I feel like I should tell them because they've had a lot of. worry when it comes to my health" (ID20).*

Participants also felt informing relatives could benefit themselves. Some found relatives helpful in decoding information and providing emotional, financial and risk management support and advice. Other participants felt relatives were not qualified to provide such advice and support.

"They… both spend a lot of time looking things up… And clarifying things for me." (ID10). .

> *"I don't see how you can get information and risk management from your family, they don't know anything" (ID13).*

3.4.2. The importance of choice

Participants felt it was important they were given the choice of who, what and how to inform. They also valued choice in what they and their relatives did with this information. Thus, they wished to carefully consider who and how they would tell, and did not want this mandated in any way by health professionals. Nor did they feel responsible for influencing their relatives' response to results.

> *"I just thought it was up the patient to decide on who they can inform" (ID17)*

> *"I think my family and extended family would also have the right to do whatever they want with that information" (ID9).*

3.4.3. Personal disclosure optimal

Most participants said a personal approach to disclosing GS results to relatives was preferred, at least initially.

> *"Breaking the news and speaking to them initially, it would be better coming from me" (ID2).*

Participants felt they knew their family members and dynamics best. They could, therefore, tailor communication to the individual's comprehension, language ability and likely response and lessen the impact of any potentially shocking results. Participants felt they could get consent from relatives for contact with a HP, if needed.

> *"I think that when it comes as a cold hard fact from a healthcare worker, it can be a bit impersonal and scary" (ID2).*

Some participants felt that under-resourced health professionals should not be burdened with disclosing GS results and might not be able to put the time and care into this task that patients could.

> *"I wouldn't have that expectation that they… disseminate the information. I wouldn't want that extra burden on a health professional because… that's just another box that they have to tick" (ID3).*

However, some participants preferred sharing the responsibility of disclosure with HPs or having the HP take on this role entirely due to their greater expertise and ability to provide an accurate explanation.

> *"I think you need the genuine sincerity of the family member saying 'I think you need to see this' but you also need the accurate information from a healthcare professional." (ID5)*

3.4.4. Family Communication is a complex and idiosyncratic process

Participants noted the importance of careful communication, considering emotions, understanding, and family characteristics. Some felt it would be helpful to separate emotions from facts to prevent extra confusion and overwhelming those receiving results.

> *"If you communicate emotionally then you know. that could invoke panic or… incorrect facts" (ID20).*

Most participants discussed not wanting to say the wrong thing, wanting to give relatives all the information, and being able to answer questions fully.

> *"Instead of dribs and drabs - better having the full responses to give them before you pass anything on" (ID4).*

However, there was also an assumption (not necessarily correct) among some participants that their results would be clear, personalised, and actionable, which would facilitate disclosure:

> *"I assume when all the results come out there'll be an extensive report written about. what those results actually mean… that should give me all the information I need." (ID9)*

3.4.4.1. The Bush Telegraph. Many participants noted that GS results would pass from one person to another and get shared with all eventually, according to well-established communication lines.

> *"I would get them to pass the word around rather than ring each one individually. They call it the bush telegraph." (ID10).*

Often, the main conduit of information was maternal. Where a biological mother was deceased or absent, participants might choose a female relative or someone they were confident would do the job well. In some families the father would tell his side of the family and the mother hers.

> *"Our mother died when we were very young. So, she [an aunt] was kind of like a mother figure, so I would pick [her] and then get [her] to pass the word around" (ID10).*

> *"[Dad] communicates with [his side of the family] over there because of the language barrier. He speaks good English, so he passes on all the information" (ID15).*

Technology such as phone calls, group video calls and family conversations on social platforms was often used to facilitate communication.

"A lot of my extended family are on a group chat. I think I would probably use that as a way of communicating that information. everyone is constantly in communication with one another" (ID9).

3.4.4.2. Family characteristics. Family characteristics impacted the complexity of communication and participants' decisions regarding who and how to tell. These included family size, immediate versus extended family, physical distance of relatives, closeness of relationships, side of the family, general openness of communication, previous cancer experience/family history, vulnerability, and interest in science or medicine. Smaller family size appeared to facilitate communication and create a greater sense of urgency to protect family members.

"I've only got a fairly small family. so I'll definitely let them know" (ID1).

Participants often preferred to disclose to nuclear before extended family but were open to eventual disclosure to all. However, some respondents preferred not to disclose to extended family, due to physical or emotional distance. Family feuds could exacerbate this. Generally, openness of communication in the family was seen to facilitate result disclosure and vice-versa.

"I wouldn't tell anyone [in extended family] anything. It's just I don't talk to them" (ID12).

"It's very good in my immediate family. we're very open with regards to communication especially when it's concerning something as important as this" (ID9).

Participants with a higher burden of cancer in the family and previous experience communicating difficult medical information felt more comfortable disclosing results. Participants often judged those with a cancer family history or with an interest in science and/or a medical occupation to be likely more interested in and needing results.

"Recently one of my uncles. passed away from cancer, so that kind of brought everyone together…So I feel like that would. improve their communication" (ID9).

"My cousin on my father's side. it was her mother that died in her 40s and I'd want that information available to her" (ID19).

"…my son's education is in science, so he's really interested in it" (ID2).

Some participants acknowledged the vulnerability of some family members due to young or old age or mental unwellness and tried to protect them by potentially withholding information.

"I do have a mentally unwell father so it's hard to talk to him about things and he doesn't process things. so I probably may not tell my dad" (ID5).

3.4.5. Significance of Results

Participants reported that significant or actionable results were more likely to be disclosed. Contrastingly, some participants felt they would share everything, having nothing to hide.

"I'd definitely share if there was some kind of result that might affect them. if they need to look out for anything in themselves" (ID20).

"I'll tell anyone who asked. Even… the anonymous person on the bus next to me. I'm not private like that" (ID6).

3.4.6. HPs have a role in supporting family communication

Participants felt that HP support facilitated family communication, especially with difficult or distant relatives. The forms of support most appreciated were prompts for thinking about who is in the family, providing accurate information and answers to patient questions, and preparing a written report of results. The decision around which HPs to involve was influenced by trust and closeness, with GPs often preferred.

"I'd get a doctor or someone involved to explain it… rather than me. Because they know more about it than I would, and they'd be able to explain it better" (ID13).

"She's a very thorough GP, so I'd like to keep her in the loop with everything that's happening" (ID4).

4. Discussion

This mixed-methods study explored factors associated with perceived importance of disclosing GS results to relatives, patient perspectives on disclosure, and perceived barriers and facilitators of disclosure. As noted above, while factors associated with perceived importance of disclosing genetic results to relatives have been well explored in the single gene setting, whether findings from this setting will be similar or different to those found in the context of GS testing, remains an empirical question. Quantitative and qualitative results were highly convergent. Our key findings indicate that most patients felt a moral obligation to disclose and valued autonomy during the result-sharing process. In both the nuclear and extended family, pre-existing patterns of communication influenced this process. More educated patients found potentially communicating results a less daunting task. Families had their own unique paths by which information gets disseminated. Finally, health professionals were thought to play an important role in facilitating this process. **These findings are highly consistent with those from the single gene setting.**

Participants generally perceived high importance in disclosing GS results to relatives, intended to do so, and saw disclosure as a moral obligation, as has been reported in the single gene setting. For example, Etchegary and Fowler [28] found a sense of 'genetic responsibility' to disclose in participants receiving risk results for Huntington's Disease, while moral obligation was found to be a major motivator for disclosure in healthy US participants if tested for alpha-1 antitrypsin deficiency results [29].

Participants perceived benefit from disclosure not only for relatives but also for themselves, through relatives providing emotional and financial support, helping to decode information, and giving risk management advice. This conforms with the family-centred model for sharing genetic risk proposed by Daly et al. [30], which emphasises the importance of harnessing family support for the proband as well as educating and providing support to family members.

Importantly, both our quantitative and qualitative findings suggest that pre-existing patterns and quality of communication and relationships within the family influence the perceived importance of disclosing GS results, intent to disclose and ease of disclosure. This supports previous studies from the single gene setting, which have found the openness of family communication to be a significant predictor of genetic disclosure [9,10]. Previous practice in communicating about serious health issues (as occurred after a cancer diagnosis) also facilitated this process. High-quality communication and close relationships were significantly more apparent in the immediate family circle than the extended family, leaving the latter vulnerable to non-disclosure, as has been reported previously in the single gene setting [31]. Qualitatively, the 'Bush Telegraph' emerged as a process often employed to reach extended family. Patients delegated the burden of disclosure responsibility to key family members (often women, as found previously [32,33]), who dispersed the information using existing diverse lines of family communication. Technology also emerged as helpful tool to overcome barriers of distance and multiple disclosures. Using social platforms, such as video calls or family group conversations reduces the effort, time and burden on others to communicate to all relatives

individually [34]. However, while these strategies may be effective in some families (such as large Irish families [35,36]), it may be less effective in others that are less interconnected. Systematic reviews have shown that communication of genetic risk is often a highly selective process, in terms of what and to whom results are disclosed [5,37]. Thus, exploration of family dynamics may be critical to ensure those who need to know GS results are told.

Patients with a higher education level were more likely to feel they should disclose results to relatives, as was found by Lafrenière [38] when looking at family communication of *BRCA1/2* results. Surprisingly, health literacy was not associated with perceived importance of disclosure, perhaps due to limited variability on this measure. However, findings in the single gene setting have shown that people perceive risk differently depending on their knowledge of genetic information [39]. As many of our participants intended to communicate results only if they conferred high risk (as has been previously reported [6]), and if they were confident in their ability to accurately communicate results, clear communication of health and genetic information to patients remains critical.

Most patients expressed a preference for managing disclosure of GS results to families themselves, echoing genetic health professionals' perceptions when considering their own responsibility to communicate results to family members [40]. In focus groups, HPs argued that patients should be afforded the autonomy to decide when and how to pass on results, that patients knew their relatives best and could thus better tailor the message, and that clinic contact might undermine family relationships and harm patient-health-professional relationships, trust and confidentiality [40].

However, despite seeking control over the process, many of our participants expressed a desire for health professional support for results disclosure to family members, as has been previously reported in the single gene setting [41]. Our participants specifically desired provision of accurate, written information and answers to their questions. This was associated with the (potentially unrealistic) expectation that results would be clear, actionable and personalised, as has also been reported in the single gene setting [42]. Consent and information documents that set realistic expectations and raise the issue of results disclosure upfront might be helpful [43]. Genetic clinics, with the patient's permission, could send a family letter summarising results. The family letter has, however, been criticised as placing pressure on patients to prepare family members for its receipt, being difficult to word to ensure accurate understanding, and failing to respect family preferences for communicating and receiving this information [44]. Furthermore, it has not proven particularly successful [43]. Other strategies to support family communication include providing psychoeducational guidance and strategies for communication [39], logistically structuring appointments to account for family dynamics, and offering follow-up appointments for patients and relatives if required [45,46].

Limitations of this study include its cross-sectional design, limiting exploration of causation. Furthermore, this study gained insight only into how patients view the importance of family communication and cannot inform how patients communicate in practice. We chose to measure perceived importance of disclosure (rather than intention to disclose), which may be seen as a more indirect measure of intended behaviour. Future studies could measure intent, or even better, actual behaviour as a primary outcome. Although our sample was heterogeneous across age, sex, and cancer type, education level was generally high and likely not representative of the population as a whole. While age was not found to be a significant predictor of communication intention, in future studies it would be useful to purposively compare qualitative responses of different age groups. Education and health literacy were unsurprisingly moderately correlated in our sample, and this may have impacted our results; for example, health literacy may have been significantly related to the primary outcome had it been assessed on its own. Finally, as our participants were recruited because they had cancers indicative of a pathogenic variant, their intentions to disclose cancer-related results may have been higher than patients receiving GS results who had not experienced an illness related to their GS result. However, this limitation does not extend to non-cancer-related GS results. This study also had many strengths, including a large sample and use of a mixed-methods study design, allowing for data triangulation.

5. Conclusions

Our findings show that patients feel GS results belong to family too. There is long-standing controversy in genetic counselling as to whether results disclosure should be patient-centric and confidential to the patient only or involve the whole family [47,48]. As genetic information is increasingly considered to be family information [49], medical ethicists' viewpoints on this issue are shifting. Our findings support expansion of the role of HPs in facilitating family communication, particularly to support probands with low education who may lack confidence in communicating and whose families are dispersed and without clear pre-existing communication channels.

Practical implications

A family-centred approach to genetic healthcare [47] that respects patients' autonomy and desire to protect relatives and consider family dynamics but also supports them in this process is needed. Genetic health professionals can provide support by ensuring patients are fully informed so they can make informed choices regarding if, who and how to inform. They can offer written information, encourage disclosure, explore existing family dynamics and patterns of communication, offer strategies to improve family communication and provide the opportunity to roleplay communication. The successful Dutch model for cascade screening could provide a useful model: this involved healthcare workers gaining consent from familial hypercholesterolemia probands to provide their contact details to a centralised government-funded coordinating office [50]. This office educated probands on the importance of screening and how to talk about screening with their first-degree relatives. Family history was gathered, and after initial proband contact, the office directly contacted first degree relatives to arrange screening, with 90% of probands having family members screened. This model was successfully adapted to the US context also [51]. Chatbots and other digital tools provided to family members of probands may prove helpful also. For example, the chatbot employed by Schmidlen et al., [52], described the proband's result, associated disease risks and recommended management, and also captured whether the person using the chatbot was a blood relative or caregiver, sex, and relationship to the proband. However, our study emphasised that attention to family dynamics and patterns of communication is key in this context. It it is likely that effective models of care will incorporate some personalised discussion, probably one-on-one between a genetic counsellor and proband, to understand and develop strategies to overcome any family communication barriers to discussion of genetic results.

Practice implications

Healthcare professionals can facilitate discussion of GS results by offering written information, encouraging disclosure, exploring existing family dynamics and communication patterns, and offering strategies to improve family communication. Centralised genetic communication offices and chatbots can also be helpful.

CRediT authorship contribution statement

Camelia Harrison: Writing – original draft, Formal analysis, Data curation. **Nicci Bartley:** Writing - review & editing, Supervision, Project adminisrtation, Formal analysis, Conceptualization. **Chris Jacobs:** Writing – review & editing, Supervision, Formal analysis, Conceptualization. **Megan Best:** Writing – review & editing, Project administration,

Conceptualization. **Sabina Vatter:** Writing – review & editing, Data curation. **Bettina Meiser:** Writing – review & editing, Investigation. **Mandy Ballinger:** Writing - review & editing, Project administration. **David Thomas:** Writing - review and editing. **Phyllis Butow:** Writing review and editing, Supervision, Formal analysis, Conceptualization. **Ballinger Mandy:** Writing – review & editing, Data curation, Conceptualization. **Butow Phyllis Noemi:** Writing – review & editing, Supervision, Formal analysis, Conceptualization.

Declaration of Competing Interest

BM has a remunerated consultant role with the company Astrazeneca with respect to an unrelated project. The other authors declare no competing interests.

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Appendix A. Supporting information

Supplementary data associated with this article can be found in the online version at doi:10.1016/j.pec.2023.107800.

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