Young-onset dementia: A systematic review of the psychological and social impact on relatives

Authors

Maddison Wiggins*[1], Alison McEwen[1], Adrienne Sexton [1,2,3]

[1]Graduate School of Health, University of Technology Sydney, Ultimo NSW Australia

[2]Genomic Medicine Department, The Royal Melbourne Hospital, Parkville VIC Australia

[3]Department of Medicine Royal Melbourne Hospital, The University of Melbourne, Parkville VIC Australia

*Corresponding author at:

Level 1 Pritchard Building, St. George Hospital, Gray St Kogarah NSW 2217,

E: Maddison.wiggins@health.nsw.gov.au or maddiwiggins03@gmail.com
Ph: +61 451 836 489
Fax: (02) 9113 3635

ABSTRACT

Objective

Young-onset dementia (YOD) has significant impact for the affected person, but also has far-reaching effects on the family. Additionally, biological relatives have an increased genetic risk of developing the condition themselves. This review aimed to identify the psychological and social impacts of YOD in the family, for asymptomatic relatives.

Methods

A systematic review of key databases for empirical studies about the lived experience of biological relatives at risk for YOD was performed. Data was collated and interpreted via narrative synthesis.

Results

The majority of the nineteen included studies were qualitative and explored the experiences of children with a parent with YOD. Five themes were developed: (1) Onset of YOD disrupts family functioning (2) Emotional impact is significant and varied (3) Uncertain future (due to uncertainty of diagnosis, care-giving responsibilities, and their own increased genetic risk) (4) Lack of visibility in health care and society (5) Coping strategies include physical/cognitive distancing, and emotion-focused coping.

Conclusion

Our findings demonstrate a diagnosis of YOD significantly impacts the lives of relatives, yet their experiences and needs often go unnoticed.

Practice Implications

We present a practical framework of questions and strategies for care of relatives, mapped to the self-regulation model of genetic counselling.

1. Introduction

Young-onset dementia (YOD) refers to onset of symptoms under 65 years old. A recent meta-analysis indicated a prevalence of more than 1 in 1000 people, equating to 3.9 million people worldwide living with YOD[1]. YOD disorders include early-onset Alzheimer's disease, frontotemporal dementia (FTD) and rarer types of dementia [2, 3]. In YOD, the potential for a dominant genetic cause is increased (Table 1). In addition to underlying monogenic causes, moderate risk genotypes such as *APOE* common variants (Table 1) and polygenic risk (combinations of many small additive background genetic factors) contribute to increased heritability in YOD, compared with older onset dementia which is usually multi-factorial and unlikely to have a major genetic aetiology [4]. Genetic testing can be performed for young-onset dementia genes and may identify an exact cause (familial pathogenic DNA variant), however it is not possible to identify all genetic causes with currently available methods. This can be further complicated by the introduction of variants of unknown significance [4]. Some families have a strong history where a parent and grandparent in previous generations also had YOD, while in other families the extended family history may be unknown, or a DNA problem can occur for the first time (*de novo*) in genes such as *PSEN1* for early onset Alzheimer's disease (for review of genetic testing and counselling for YOD see [5] and [6]). If a genetic cause has been identified in a family, then predictive genetic testing becomes an option for relatives. However, choosing to find out whether one has inherited the genetic variant and will almost certainly develop YOD in the future is a challenging decision and uptake ranges from 5-30% [7].

Although little data is available, it is anticipated that the psychological and social relevance may vary at different life-stages. Relevant life-stages include forming long-term relationship, family planning, and approaching the average age of onset, although this often is highly variable (age of onset of symptoms can range from age 20s onwards, depending on the gene (Table 1)). Genetic counselling for support with decisional dilemmas, preparation, coping, timing, relationships, reproductive options, psychological support, and family communication is recommended (for review see Crook, Williams [7]).

Table 1. Genetic causes for young onset dementias^

Condition#	**Genes**	**Inheritance pattern**	**Average age of onset (range)**
Early onset Alzheimer disease	*PSEN1, PSEN2, APP*	Autosomal dominant (1 in 2 chance of inheriting from a parent who has gene and will develop YOD)	40s-50s (20s-70s)
Prion disease including Creutzfeldt-Jakob disease	*PRNP*	Autosomal dominant	40s-50s (teens – 70s)
Frontotemporal dementia with or without ALS*	*C9ORF72*	Autosomal dominant	58 (30s-80s)

Frontotemporal dementia	*MAPT,* *GRN*	Autosomal dominant	49 (late teens-80s) 61 (30s-80s)
CADASIL	*NOTCH3*	Autosomal dominant	40s-50s (30s-80s)
CARASIL	*HTRA1*	Autosomal recessive (1 in 4 chance of inheriting, when both parents are healthy genetic carriers)	30s (20s-50s)
Late onset Alzheimer disease	*APOEe4*	Major risk factor, combines with polygenic risk (Common genotype; difficult to quantify risk)	40s-90s. Confers risk of younger onset when *APOEe4*/e4 genotype is present

^ For the purposes of this review, Huntington disease has not been included under the YOD umbrella, due to the specific nature of the condition and a separate body of literature.

This list is not exhaustive – other rare genetic causes of young onset dementia exist.

*ALS, amyotrophic lateral sclerosis/motor neuron disease

The social context of YOD creates extra psychological and social challenges: those living with YOD may still be part of the workforce, have children living at home and be caregiving for their own parents, and may experience symptoms including personality change, psychosis, lack of empathy, disinhibition, aggression, irritability, and apathy [2, 3]. The impact of these symptoms may also lead to changes in lifestyle and intra-family

relationships for which families are unlikely to be prepared [8]. In addition to asymptomatic family members being involved in the care of the affected person, they may also be trying to understand their own genetic risk for disease development. Studies with genetic conditions have shown that children can begin to understand concepts about hereditary from age eight onwards, and prefer to learn gradually about genetic risk during childhood, whilst parents find conversations about genetic risk very painful and may withhold the information until older to avoid distressing conversations [9]. Little is known about how adult relatives or child relatives in families with YOD learn that there is a chance of a genetic cause, although in some families a strong history in close relatives may make this obvious.

The many complex aspects of YOD and the gaps in the literature highlight that consideration of the needs and support of the affected person must be extended to the family unit [10-13]. Further insight into this will be relevant for counselling and other healthcare interactions with relatives at any age from childhood to mid-adulthood. This systematic review addresses the question "How does having a family history of young-onset dementia impact the psychological and social experiences of asymptomatic relatives?"

2. Methods

The methods and reporting for this review are based on the quality standards outlined by the Cochrane Database of Systematic Reviews for qualitative syntheses[14] and the Preferred Reporting Items for Systematic Reviews and Meta-Analysis (PRISMA) criteria[15].

2.1 Search Strategy

A systematic search of MEDLINE, CINAHL, PsychINFO and EMBASE was conducted from inception to May 10th, 2022. Reference lists and citations of included studies were also searched. The following keywords were used in various combinations: family history, young-onset dementia, psychological/social impact and terms capturing various relatives and biological relationships (see supplementary Box S1 for further details).

2.2 Eligibility Criteria

Inclusion/exclusion criteria were based on the PICO (population, intervention, comparison and outcome) framework[16] (Table 2). Studies reporting on the experiences of asymptomatic relatives of a person with YOD, where dementia symptom onset was under the age of 65, were included. As there are few studies on this topic, experiences of all ages (child, teen and adult relatives) were included. There was not enough data to distinguish between those with a known genetic cause in the family from those where genetic testing had not been performed or where genetic testing was uninformative. Details of inclusion/exclusion criteria are provided in Table 2.

2.3 Study Selection

The study selection process was carried out by two independent researchers. Inter-rater reliability was determined using Cohen's Kappa, ensuring a score of at least 0.7 was met, indicating substantial agreement[17]. Disagreements were resolved through discussion. The PRISMA flow diagram in Figure 1 summarises the process of systematic review.

Table 2. Inclusion and Exclusion Criteria

Inclusion	Exclusion
Population	*Population*
Asymptomatic relative of a person with young onset dementia (YOD)	Caregivers or spouses not at potential genetic risk
AND onset of YOD in the affected person before age of 65	Onset of dementia in affected family member after the age of 65
	Included population not differentiated from excluded population (e.g if results about spouses and children are not distinguished)
	Age of onset of the affected person is not specified
Intervention	*Intervention*
Family member with YOD onset before age 65	Family member diagnosed with late-onset dementia
Comparison N.A	*Comparison* N.A
Outcome	*Outcome*
Articles presenting data on the experiences, needs, challenges, social or psychological impact of having a relative diagnosed with YOD	Articles that do not differentiate the experiences of asymptomatic family members and spouses
	Articles which do not include information on the experience or psychological or social impact of having a family member with YOD
Study design	*Study design*
Empirical peer reviewed studies (qualitative, quantitative & mixed methods)	Reviews, editorials or opinion pieces
Articles written in English	Unpublished research or poster presentations
No date restrictions were imposed	Empirical studies that are not peer reviewed or have been retracted
	Articles not in English

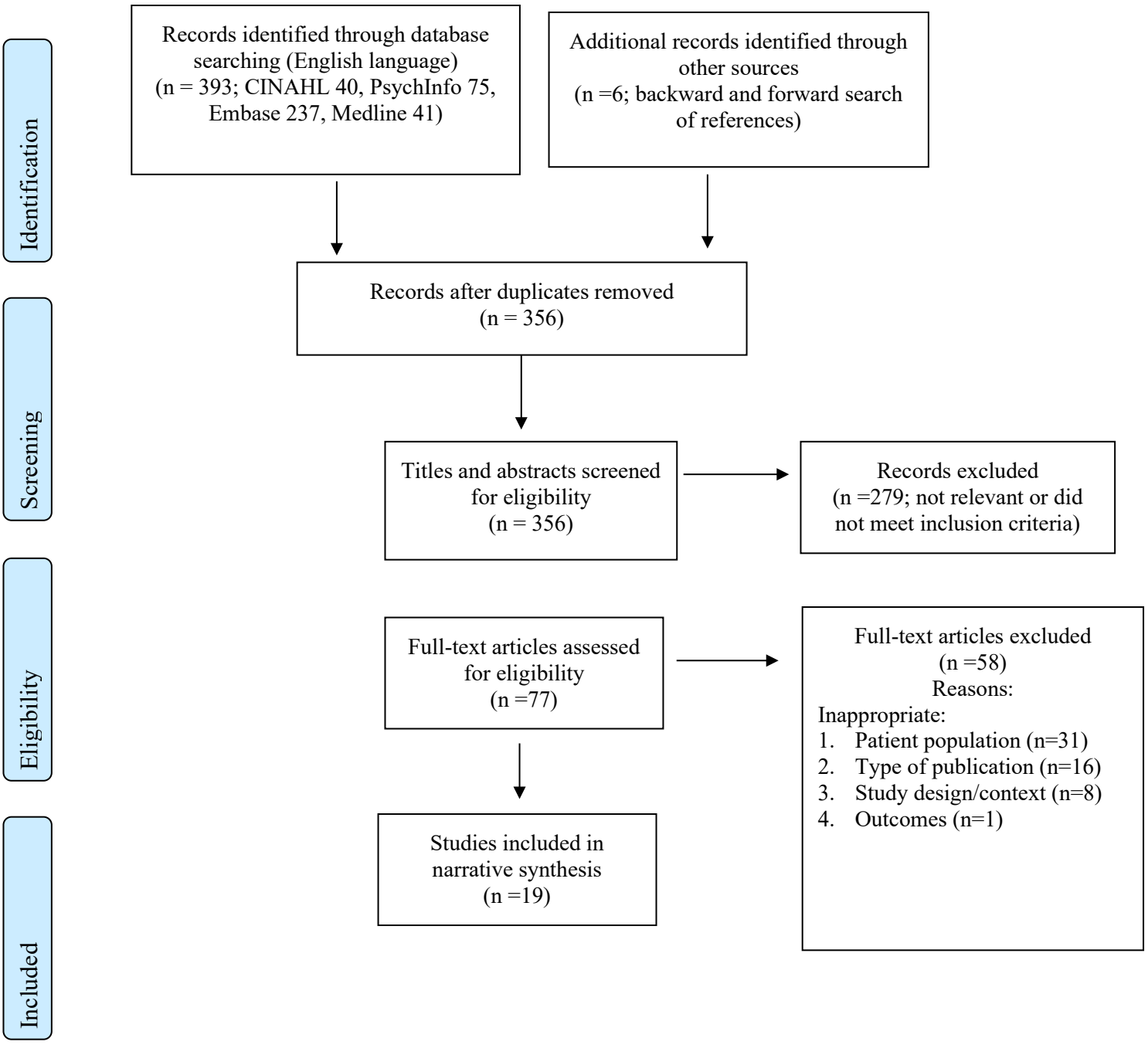

Figure 1. PRISMA flow diagram summarising the systematic selection process

Quality Assessment

The QualSyst tool is a validated checklist and was used to assess the quality and risk of bias for all qualitative studies[18]. The QualSyst tool has ten items relating to study design, analysis and reporting, each with score of 0-2. An average per-item score is calculated after scoring each article. All studies from the screening process met the minimum required score of 0.55 or higher. The Mixed Methods Appraisal Tool (MMAT) was used to assess the quality and risk of bias for the included mixed methods study[19].

4.3 Data Extraction and Analysis

Data were extracted based on three categories: general data, individual study characteristics, and outcome measures. The general data extracted included the author, title, source and year of publication. The extracted study characteristics included study design, participant characteristics, methodology, and main findings. The outcome measures extracted included support needs and/or psychological, emotional, behavioural, cognitive, and social changes due to having a family history of YOD. This was represented descriptively, or by scales, when questionnaires were used. An inductive narrative synthesis, as described by Popay, Roberts [20], was conducted to identify similar themes across the studies which met the inclusion criteria.

3. Results

Nineteen papers representing ten qualitative studies and one mixed methods study were included (Table 3), with one study presented across six papers and three studies presented across two papers. The majority of studies reported the experiences of youth with a parent with a diagnosis of YOD[21-37]. One study explored the experiences of at-risk family members

including children, siblings, nieces/nephews at genetic risk for YOD, as well as spouses of a person with YOD who are at genetic risk due to consanguinity [38]. This is the only paper in which all families had a known dominant genetic cause for YOD in the relative with YOD (a pathogenic variant in the early onset Alzheimer disease gene *PSEN1*), although the asymptomatic participants did not report having had predictive genetic testing for themselves [38]. One study used mixed methods and investigated the perspective of siblings of a person with early onset Alzheimer disease[39], and is the only study in which participants (three of 24 total) have had genetic testing, two of which were on a research basis only with unknown results, and one who had negative results for a known dominant early onset Alzheimer disease gene. Although this paper does not state the family history, 25% of participants had one or more first degree relatives with Alzheimer's disease in addition to their affected sibling. No other studies state the family history apart from one first-degree relative with YOD. The main themes and subthemes developed from the narrative synthesis are discussed below.

Table 3. Summary of included papers on experiences of relatives of a person with young-onset dementia:

Study	Country	Study Design	Methods	Participant Total	Participant Age	Relation to Affected Person	Risk of Bias*
Aslett, Huws [21]	UK	Qualitative	Semi-structured Interview	5	23-36	Child	0.8
Allen, Oyebode [22]	UK	Qualitative	Semi-structured Interview	12	12-24	Child	0.9
Barca, Thorsen [23]	Norway	Qualitative	Semi-structured Interview	14	20-37	Child	0.85
García-Toro, Sánchez-Gómez [38]	Colombia	Qualitative	Semi-structured Interview	27	18-60	Child, Sibling, Niece, spouses at genetic risk	0.8
Gelman and Rhames [24]	United States	Qualitative	Semi-structured Interview	12	13-20	Child	0.65
Gelman and Rhames [25]	United States	Qualitative	Semi-structured Interview	12	13-20	Child	0.8
Hall and Sikes [26]	UK	Qualitative	Unstructured interview	22	6-31	Child	0.65

Study	Country	Study Design	Methods	Participant Total	Participant Age	Relation to Affected Person	Risk of Bias*
Hall and Sikes [29]	UK	Qualitative	Unstructured interview	22	7-31	Child	0.65
Hall and Sikes [31]	UK	Qualitative	Unstructured interview	22	6-31	Child	0.65
Sikes and Hall [27]	UK	Qualitative	Unstructured interview	22	6-31	Child	0.55
Sikes and Hall [28]	UK	Qualitative	Unstructured interview	24	6-31	Child	0.7
Sikes and Hall [30]	UK	Qualitative	Unstructured interview	19	8-31	Child	0.6
Hutchinson, Roberts [32]	Australia	Qualitative	Semi-structured Interview	12	10-33	Child	0.8
Hutchinson, Roberts [33]	Australia	Qualitative	Semi-structured Interview	12	10-33	Child	0.8
Johannessen, Engedal [34]	Norway	Qualitative	Semi-structured Interview	14	18-30	Child	0.9
Johannessen, Engedal [35]	Norway	Qualitative	Semi-structured Interview	14	18-30	Child	0.9

Study	Country	Study Design	Methods	Participant Total	Participant Age	Relation to Affected Person	Risk of Bias*
Millenaar, Bakker [40]	Netherlands	Qualitative	Semi-structured Interview	15	15-27	Child	0.85
Svanberg, Stott [37]	UK	Qualitative	Semi-structured Interview and questionnaires	12	11-17	Child	0.7
Wain, Uhlmann [39]	United States	Mixed Methods	Semi-structured Interview and questionnaires	25	37-83	Sibling	N/A

Note: The term “child” is used to describe the offspring of an affected person, at any age.

*Risk of bias score for all qualitative studies calculated using the Qualsyst tool, where a score greater than 0.55 is sufficient for inclusion [18],. The Mixed methods assessment tool does not provide an overall score [19]

3.1 Onset of YOD Disrupt Family Functioning

All included papers report that asymptomatic family members take on caretaking responsibilities[21-39]. Role reversal was common for children of a parent with YOD, taking on responsibilities the affected parent had prior to the onset of symptoms, such as ensuring safety of their affected parent, completing household tasks and providing personal/intimate care[21-25, 31, 33-37]. Factors which are hypothesised to influence the roles/responsibilities of the child include their age, gender, whether they reside at home, the severity of the illness and the rate of progression[22-24].

Two studies describe the experience of adult siblings[38, 39] and caring responsibilities are portrayed. This includes providing personal care, supporting family members, preparing meals and coordinating living and medical arrangements.

Asymptomatic family members experience a sense of responsibility to take on the additional roles[22, 32, 35, 36], and children also feel a responsibility for the well-being of the unaffected parent[22, 25, 36, 37]. Guilt results when the asymptomatic family member feels they have not properly fulfilled this responsibility. This has been reported in cases where the young person physically distances themselves by moving out of the family home[32], when making the decision to place the affected person in residential care[21, 34], when a family member feels they are not doing enough[21, 35, 37], or when feeling resentment or anger towards their new responsibilities or towards the affected persons behaviour[23, 30, 38].

Disruption of the usual day to day functioning and activities is commonly reported as taking on additional responsibilities and often results in less time for the individual to take part in activities they enjoy, such as socialising with peers, and an increase in absences from school[22, 23, 28, 37].

3.2 Emotional Impact is Significant and Varied

In addition to guilt, discussed above, grief is one of the most commonly reported emotional responses. The natural progression of YOD results in gradual and continuous losses for the affected family member, often causing the unaffected family members to grieve the loss of a person who is still physically present[23, 24, 27, 28, 32, 34]. Unaffected family members may also experience grief from the loss of the relationship and role that the affected person once played in their life[21-25, 31, 37-39], or grief for the future they once imagined for themselves and their affected relative[22, 26, 28, 32].

Many asymptomatic family members reported a decline in mental health, anxiety and/or depression, or a mental illness such as obsessive-compulsive disorder, psychosis or alcohol/substance additions[22, 27, 28, 32-34, 37-39]. Four studies reported self-harm or suicidal thoughts/attempts in children of a parent with YOD[22, 24, 26, 32].

Positive emotional responses were also experienced when providing care for an affected family member, including a sense of pride, resilience, purpose and direction[33, 37, 38]. Increased self-efficacy was described by two qualitative studies, investigating the experiences of young people with a parent with YOD in Australia and caregivers at genetic risk of EOAD in Colombia, which was related to increased self-growth, self-belief and self-worth [33, 38].

3.3 Uncertain Future

The progression of YOD is often unpredictable, which can lead to a level of uncertainty for family members who are not sure how the disease will affect their loved one, or their caring responsibilities[26, 28, 29, 36, 38]. Initial uncertainty, sometimes over a long period, resulted

from delays in diagnosis. Three studies report that teen and adult family members put plans on hold as they felt their future is uncertain due to their caregiving responsibilities and the unknown progression of their relative's disease [26, 29, 36, 38].

Four studies report that uncertainty is associated with not knowing one's own genetic status, and the risk for developing YOD themselves [26, 29, 41, 42]. None of the studies differentiate the impact this uncertainty has on the unaffected relatives based on age. However, in a study with 25 adult siblings, Wain, Uhlmann [39] found that for some participants, the potentially increased genetic risk of early Alzheimer disease did not impact decision-making or future plans, whilst for others the ability to plan ahead was impeded. Additionally, although genetic status was not reported, Aslett, Huws [21] identified that some adult children with a parent with YOD question their mortality, knowing there is a potentially increased risk for personally developing YOD, while Hutchinson, Roberts [43] report that young carers have fears about who will take care of them, should they also develop YOD in the future.

As a result of layers of uncertainty reported above, many asymptomatic family members report putting their life "on hold"[21, 26, 29, 32, 36, 38, 39]. This includes altering or changing future plans, including education, career and family planning. Examples of this includes turning down scholarship opportunities due to caregiving responsibility [43], postponing milestones such as moving out of the home [42] or relatives neglecting their own health and fertility [44]. The results of the studies do not distinguish facets of uncertainty in much detail, and therefore future work on this topic is needed to inform counselling.

3.4 Lack of Visibility in Health Care and Society

Relatives often noted a lack of recognition from health professionals about the impact of YOD on family members. Although asymptomatic family members provide care to their

affected relative, they are often not the primary caregiver, as this title is frequently linked with the spouse. Many family members felt unsupported or dismissed by health care professionals[23, 32, 34], and often resources or formal supports for this population were either not offered, or when provided, not appropriate[21, 35-37, 39].

As a result of a lack of awareness and visibility of YOD in society, asymptomatic family members experience a sense of shame or guilt related to their relative being stigmatised or marginalised within the community[22, 24, 26-28, 31-33]. Additionally, isolation was reported in nine studies, where asymptomatic family members felt their personal experiences, or the behaviours of their relatives are not understood or accepted by others[21-24, 26, 28, 30, 32-34, 37, 38]. Adding complexity, an interview and questionnaire study by Svanberg, Stott [37] reported that although children take on caregiving responsibilities, the label of "young carer" sometimes induced feelings of guilt for not doing enough for their parent.

3.5 Asymptomatic Family Members Implement Coping Strategies

A common finding was the coping strategy of distractions, such as going to school, extracurricular activities or connecting with peers, to provide a sense of normalcy in one's life[22, 24, 28, 32, 33, 35-37]. Physical, cognitive and emotional distancing as a coping strategy is reported by six studies[27, 33, 35-38]. Four studies, in which participants aged 6 to 31 years old, with a parent with YOD, report emotion-focused coping, such as drinking, smoking or self-harming[22, 24, 26, 32]. Three studies report finding humour, staying positive and helping others assists family members' meaning-making and adaptation to their situation[36-38].

Coping strategies used by those experiencing uncertainty about their own potential risk for developing YOD include living in the moment, denial, avoidance of information and

behaviour or lifestyle changes[21, 38, 39]. An interview study by Johannessen, Engedal [35] explored the coping efforts of 14 children with a parent with YOD and found that when various coping strategies were combined, participants described an improvement in their lives at one-year follow-up.

4. Discussion and Conclusion

4.1 Discussion

Five main themes highlight the significant psychological and social effects of YOD on relatives, including disruption to family functioning, varying emotional responses including guilt and grief as well as increased resilience and sense of purpose, uncertainty about the future, a perceived lack of visibility or acknowledgment of their needs, and the use of specific coping strategies in response to these challenges. Other than uncertainty regarding future planning, specific aspect of concern in relation to heredity were not reported, and this is an important gap in current knowledge.

The results of this study highlight that asymptomatic family members take on roles and responsibilities previously allocated to the affected person. Role reversal, also referred to as parentification[45], is consistent with family systems theory, where the illness of a parent disrupts parental boundaries and role reversal occurs to maintain equilibrium [46, 47]. This finding is well supported in the literature of chronic illness in families [45, 48], and in families with YOD [49, 50]. However, no studies in this review analysed the long-term impact of role reversal on children with a parent with YOD, and this is an important area for further research. In other situations, parentification in childhood impacts many areas of adult life and has been associated with attachment issues, risk of mental illness, psychological distress and substance abuse [51-54]. Positive outcomes, including adaptative coping skills, higher levels of empathy and resilience have also been identified in families with chronic illness [55, 56].

This review reveals that unaffected family members experience a type of grief known as ambiguous loss, which is the response to an uncertain loss, such as the changing conditions of a significant relationship, or the loss of dreams for the future. It is common for individuals to experience this type of grief when a person is physically present, but psychologically absent [57-59]. Family members may experience a wide range of consecutive losses as YOD progresses. This finding is supported by the literature for other neurodegenerative diseases and suggests that this type of loss may lead to difficulties in grief adaptation [59-61].

This study raises the important role a definitive diagnosis can make in a family, as delays or misdiagnoses can cause multiple layers of uncertainty for the affected person, as well as their family members. Not having a clear diagnosis may result in unrealistic expectations of the affected person, difficulties obtaining appropriate support and feelings of anxiety and helplessness for family members [62, 63].

In most families where YOD is present, the spouse of the affected person is typically considered the primary caregiver [62, 64, 65], and this is supported by the results of this systematic review, where at-risk family members take on caregiving responsibilities but often experience a lack of visibility in the health care setting, and in society, when the impacts of YOD are not understood by others. The social category of "young carers" is relatively recent and although this has increased representation and advocacy for this population, there are many areas in which they are underserved [66]. Often, young carers are considered "hidden" as they do not disclose, or view themselves as carers [66, 67].

The most commonly reported coping strategy utilised by children with a parent with YOD is an emotion-focussed strategy through use of distractions to provide a sense of normalcy. This strategy has been reported in other chronic illness literature [68-70]. Other emotion-focused

coping strategies included humour, positive re-framing, avoidance or alcohol use [71-75]. In contrast, undergoing presymptomatic genetic testing could be considered a problem-focused coping strategy [72-74]. In other chronic illnesses, coping is most successful when young people implement a mixture of problem-focused and emotion-focused coping strategies. [73, 76]

This review used broad search strategies to capture relevant literature and was conducted in a rigorous manner. Ten studies received a Qualsyst score above 0.8, and all included studies had a score of 0.55 or above, indicating the studies included were of high methodological quality. The limitations are that resources did not enable inclusion of papers written in languages other than English. Additionally, there is limited diversity in terms of study design, population, culture and country or origin. The majority of papers explored the experience of children of a parent with YOD, meaning the results of this review may not be generalisable to all asymptomatic individuals in a family with YOD. YOD are reported as a whole, and therefore condition specific outcomes are not reported.

4.2 Conclusion

The results of this systematic review highlight the complex psychological and social impacts of belonging to a family where YOD is present. Little data is available on the added layer of complexity that the increased genetic risk in YOD adds for asymptomatic relatives. Our findings demonstrate a diagnosis of YOD directly impacts the lives of at-risk family members, yet their experiences and needs often go unnoticed. Health professionals may utilise the self-regulation model of genetic counselling (**outlined in section 4.3**) as a guide for exploring the **psychological and social** needs of at-risk family members, which may promote facilitation of more personalised care and support.

4.3 Practice Implications

This systematic review brings to light the psychological and social effects of YOD in families, where the younger onset implies a potential genetic risk in addition to the impact of experiencing the illness in their relative. For further insight we have mapped the themes onto the self-regulation model for people at risk of a genetic condition[77] (Fig. 2).

This theory suggests that individuals develop personal representations of illness through their experiences, emotions and cognitions[77]. Lived experience of illness together with factors such as perceived genetic risk, perceived personal control and options, tolerance for ambiguity, family influences, values, and access to health care will inform decision-making and ability to cope[77].

Here we utilise the outcomes of this systematic review to adapt the model for YOD and provide a range of questions that general practitioners and other health professionals could consider when meeting with a person with a family history of YOD. These questions may assist the practitioner to gain an understanding of the illness representations and needs of clients who are experiencing YOD in a relative and may have concerns about their own genetic risk. Proactively attending to the needs of family members, facilitating referral for genetic counselling, and/or introducing targeted support groups could help to minimise feelings of invisibility, stigma, isolation and shame[21, 23, 24]. As demonstrated by the studies included in this review, the experiences of children of a person with YOD have been investigated, however, no papers have differentiated their findings for different age groups among their participants. Therefore, future research using a life-span perspective to investigate the long-term impacts of being a child of a parent with YOD will be important. Approaches focused on narrative construction, as shown in a recent study by Hoppe [78], will be very useful in understanding how

relatives make meaning from their experiences**.** Additionally, insight into how the perspectives and coping strategies of relatives are shaped by the potentially increased genetic risk of YOD for themselves will be another important focus for future research.

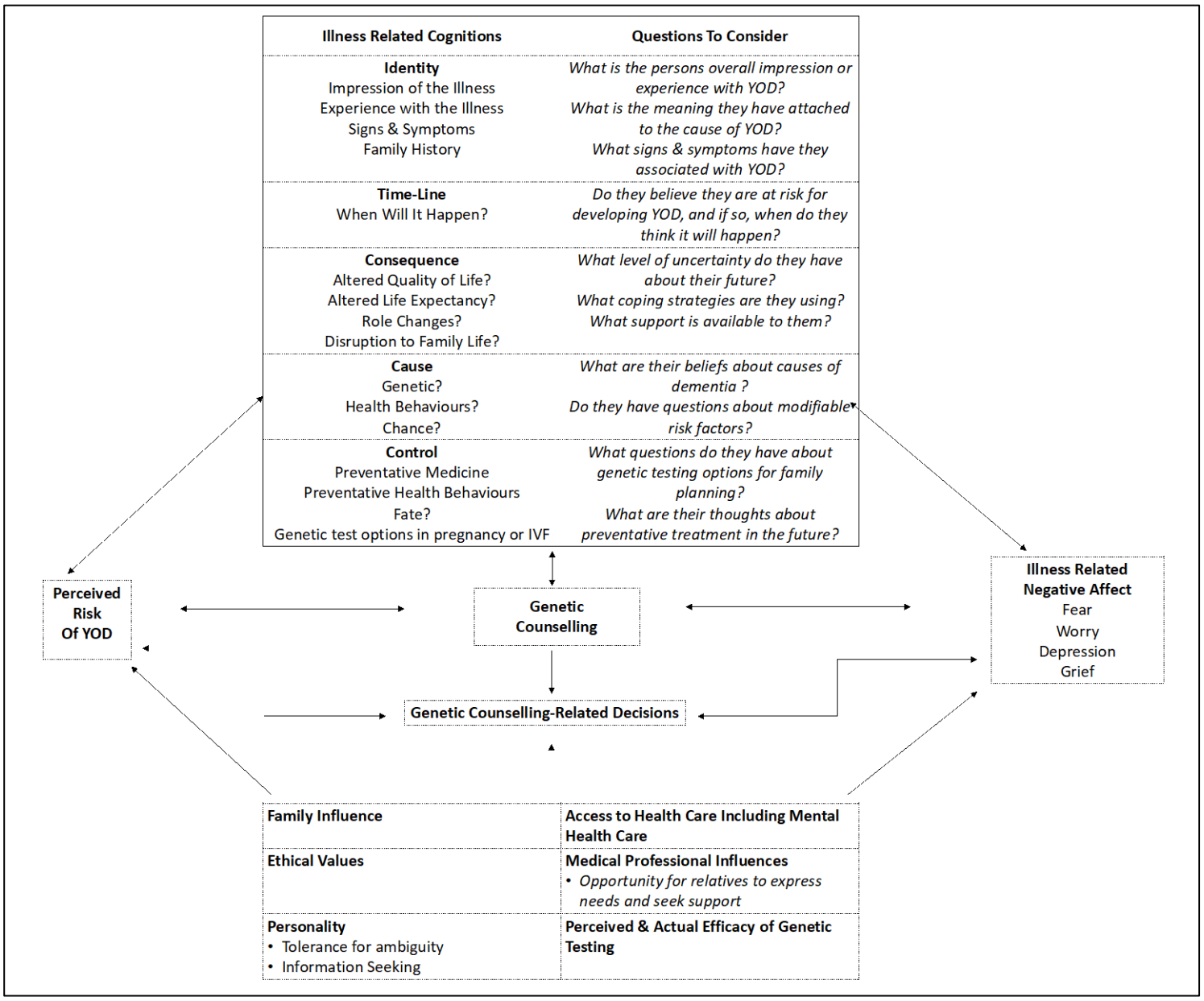

Fig.2. Suggested questions for primary health care professionals to facilitate care for clients with a relative who has YOD, mapped to the self-regulation model of genetic counselling. Diagram adapted from Shiloh [77]

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Competing interests.

Maddison Wiggins completed this study as part of a Master of Genetic Counselling qualification. AS and AM have no competing interests to declare.

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