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Doctoral Thesis

Experiences of living with Huntington's disease

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Statement of total word count

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	Main Text	Appendices (inc. tables, figures, references)	Total
Thesis Abstract	298	-	298
Literature Review	7941	11,717	19, 658
Research Paper	8000	11,775	19,775
Critical Appraisal	3879	963	4,842
Ethics Section	4,851	3,410	8,261
Total	24, 969	27, 865	52, 536

Thesis abstract

This thesis explores aspects of the lived experience with Huntington's disease (HD). The first section presents a meta-ethnography, with the review question: 'what is the experience of young people growing up in a family with HD?' A systematic search was conducted which resulted in 13 papers for inclusion. Following the synthesis of the qualitative papers, 4 interrelated themes were formed: (1) thief of relationships, (2) thief of self, (3) thief of transparency and (4) search for reclamation. The findings contribute to understanding the complex impact of HD on the lives of young people and their experience of growing up in a family with HD. Further research could focus on psychological assessment to identify the needs of young people in HD families and, following this research, could assess the provision of services and the formation of accessible support.

The second section presents the empirical research which sought to answer the question 'what is the experience of maintaining psychological wellbeing when living at risk of HD?' Twelve individuals living at risk of HD were interviewed using semi structured interviews. Interpretative Phenomenological Analysis (Smith et al., 2021) revealed three themes: (1) 'you're constantly in limbo': living in two worlds, (2) "I have to live, just bloody live": possibility of a time limited lifespan and (3) "is that who I am, is that what I am?": the exhausting quest to be seen as an individual first. The findings explore how individuals managed their wellbeing and the complexities of living at risk. Further research could focus on the lack of support experienced by individuals and poor professional knowledge of HD.

The third section presents a critical appraisal in which the similarities and differences of the meta-ethnography and empirical paper are discussed. Clinical implications are discussed further, with researcher reflections also presented.

Declaration

This thesis was undertaken between June 2021 and May 2023 as part requirement of the Lancaster University Doctorate in Clinical Psychology. The work documented here is my own except where due reference has been made in the text. This thesis has not been submitted for an award of a higher degree elsewhere.

Signature: *Hollie Cooper*

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Date: 12th May 2023

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I would like to thank the participants who took part in the empirical study. Without you, there would be no story to tell, no research to write. Thank you for your openness, honesty and for allowing me into your world. I hope I have shared your experience in a way that you are pleased with.

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(Excluding title page, references, figures, tables and appendix)

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Abstract

Huntington's disease is a genetic neurodegenerative condition with wide physical and psychological impacts. Children of a parent with the condition have a 50% chance of carrying the gene expansion and developing the condition themselves. This systematic review and meta-ethnography presents a synthesis of the qualitative research on the experiences of young people growing up in a family with Huntington's disease. The MEDLINE, PsycINFO, and CINAHL databases were systematically searched and 13 papers met the inclusion criteria. Through the process of meta-ethnography, four themes were identified highlighting aspects of childhood that were stolen and fought for: thief of relationships, thief of self, thief of transparency and search for reclamation. Within the themes the complex challenges young people faced when growing up in a HD family were explored such as the impact of adverse childhood experiences and the possible effects HD on attachment and social relationships. Clinical implications are considered, and recommendations made for future research.

Keywords: Huntington's disease, lived experience, qualitative, young people, meta-ethnography, family

What is known about this topic: Young people often become carers of a parent and are required to fulfil typical adult roles when living in a HD family. Little is known about the experience of young people growing up in a family affected by Huntington's disease.

What this paper adds to the topic: This review synthesises, from 13 qualitative studies, the experience of young people growing up in a family affected by HD. It explores the complexities young people face, the impact of this on their lives and highlights the difficulties young people have in accessing support.

Experiences of young people growing up in a family with Huntington's disease: a meta-ethnography of qualitative research

Huntington's disease (HD) is a genetic condition in which psychological distress is common. Due to its genetic nature, a child of an individual with the expanded HD gene will have a 50% chance of inheriting and subsequently developing the condition. HD is incurable and progressive, with increasing problems with movement and cognitive impairment leading to dementia. The global prevalence is approximately 2.7 per 100,000 (Pringsheim et al., 2012) but is higher in some countries, for example 10 per 100,000 in the UK (Furby et al., 2022). Predictive genetic testing can be carried out internationally usually at the age of 18 to determine if an individual carries the expanded gene and, if positive, the individual will go on to develop the disease.

Individuals are not diagnosed with HD until they become symptomatic with motor difficulties e.g., chorea or loss of balance (Novak & Tabrizi, 2010). Therefore those with the gene expansion and no motor symptoms are known as presymptomatic or premanifest and those who have not sought genetic testing but have a biological parent (50% risk) or grandparent (25% risk) with HD are known as being at risk (Novak & Tabrizi, 2010). Usual onset of symptoms occurs between the ages of 30 and 50 (McColgan & Tabrizi, 2018), though with wide variation (Frich et al., 2014). Based on median values reported by Rodrigues et al. (2017), in Europe individuals with HD die 24 years after a HD motor diagnosis and 35 years after symptoms started. Currently no medications slow the progression of the disease, though help with symptom control is available for some elements, such as involuntary movements (Venuto et al., 2012).

In families where HD is present, many complex challenges need to be navigated that extend past the physical and neurological effects of the disease, for example, decisions around

genetic testing. As HD is inherited, one positive genetic test may result in multiple other family members being identified as at risk. Indeed, Sobel and Cowan (2000) conclude that genetic testing is a family issue therefore members should discuss genetic testing decisions together.

Caring can also have negative effects on care providers. For example, McGarva (2001) reported that family caregivers experienced compromised health and lifestyle changes during the progression of their loved one's HD. This was due to the age of onset of HD being at a time in the lifespan where partners, siblings and children were likely to be in paid employment or education and have significant life commitments of their own (Aubeeluck & Buchanan, 2007). A review on the impact of caring for a person with HD on family carers' quality of life and caregiver burden (Domaradzki, 2016) found that, despite similarities with other progressive conditions (such as Parkinson's or Alzheimer's), specific factors were more burdensome for HD families. This included age of onset, the severity of symptoms, the progression of the disease and length of time of the disease, the hereditary nature of the disease and lack of knowledge of HD socially and medically.

Moreover, complex challenges are experienced by families with HD such as communication, conflict and being able to be understood by others. For example, Vamos et al. (2007) found low levels of family cohesion and verbal expression, alongside higher levels of conflict compared to non-HD families. A qualitative study, including parent/adult child dyads, explored the impact of the condition on the family throughout HD progression, concluding that understanding the needs of families experiencing HD is required (Maxted et al., 2014).

Furthermore, the challenges of living in an HD family may differ according to an individual's role within the family. One voice that needs to be amplified through research on HD within families is that of young people. It is important to understand the perspective of young people as, due to the typical age of onset of HD, parents often present with symptoms when their children are entering the life stage of becoming a young person (Driessnack et al.,

2012). Moreover, although research has highlighted the impact of HD on the family unit in relation to attachment (Van der Meer et al., 2006), family life (Vamos et al., 2007), family caring roles (Williams et al., 2009), family morals in the context of genetic awareness (Huniche, 2011) and genetic testing (Sobel & Cowan, 2000), only a limited amount of studies present the explicit voice of young people as a unique voice. This is despite research indicating as early as 1983 that 48% of children from a HD family experienced psychological distress (Folstein et al., 1983).

In a review on carers' experiences in HD by Parekh et al. (2018), of the twelve studies included, two focused on teen carers (Sparbel et al., 2008; Williams et al., 2009). The themes specific to the teens centred around the level of wisdom and insight young people developed, high level of emotional distress experienced, multiple forms of social restriction and financial worries that teens experienced. They also highlighted the lack of authority in the home and difficulties accessing and using services, including the conflict this brought to teens' decision-making around caring for a loved one conflicting with their own plans and desires. However, the findings specific to young people were drawn from only two studies and the focus of the review was specifically on the caring role. Thus, the experience of being a young person in an HD family has not been fully reviewed.

Consequently, the following review aims to answer the question: 'what is the experience of young people who have grown up in a family with HD?' Although several studies exist which have incorporated a young person's perspective, a review has not yet been conducted. Given the focus of the research question and its emphasis on lived experience, only qualitative studies were included. A meta-ethnography was chosen as this can bring new understanding (Campbell et al., 2011); it does not simply aggregate findings (Noblit et al., 1988) but can produce new evidence on experience (Campbell et al., 2011) grounded in the primary studies' data (France et al., 2019). Findings from such syntheses have the potential not

only to inform future research but also the development of appropriate interventions and services (France et al., 2019).

Method

Meta-ethnography is inductive and interpretative in its approach and has been widely used in health-related research (Hannes & Macaitis, 2012). Meta-ethnography was chosen as the method for synthesis as the aim of the review was to expand conceptual knowledge concerning the experiences of young people in HD families (Sattar et al., 2021).

Conceptualisation of data varies across methodologies. In this review, and according to the theoretical position of meta-ethnography, data were perceived as both authors' interpretations and participant quotes. In the process of meta-ethnography, data are considered on three levels: level one focuses on the participants' interpretations, understanding and experiences; this is known as first order constructs. Level two relates to the authors' (of the original paper) interpretation and understanding of what their participants shared; this is known as second order constructs. Level three is the meta-ethnographer's understanding and interpretation of the meaning of the first two levels; this is known as third order or higher order constructs.

The approach allows reciprocal (similar) or refutational (opposing) understanding to be drawn from the combination of papers. In reciprocal relationships the papers are translated into each other to offer an overarching explanation from the gathered concepts or metaphors. In refutational relationships, opposing explanations, metaphors or concepts from the papers are explored. The final stage in the meta-ethnography is to create a line of argument, indicating how such concepts relate to each other.

The synthesis was conducted following Noblit et al. (1988) seven steps of meta-ethnography which include getting started, deciding what is relevant, reading the studies,

finding how the studies are related, translation of studies into each other, synthesis of the translation and finally the expression of the synthesis.

Identifying relevant papers

Scoping searches identified a body of existing literature on HD and young people's experiences in an HD family and enabled the review question to be formed. With the support of an academic librarian, three databases were identified as the most suitable to search: MEDLINE, CINAHL, and PsycINFO and search terms focused on two concepts: HD and qualitative designs. An example of free text terms and subject headings can be seen in Table 1 -A. No data restrictions were applied.

[Table 1 - A about here]

Inclusion/exclusion criteria

To be included papers had to:

1. Include qualitative data collected via qualitative methods with participants' quotes so that beliefs and experiences could be understood.
2. Be published in a peer-reviewed journal.
3. Focus on the experience of a contemporaneous account or include retrospective accounts of childhood by an adult who grew up in a family with HD.
4. Have data relevant to the research question easily identifiable and extractable.
5. Be in English.

Papers were excluded when they contained:

1. Comparative or dyadic studies where the experience of the young person in a HD family could not be clearly extracted or where such distinction could not be clearly made (Duncan et al., 2008; MacLeod et al., 2014; Mand et al., 2013; Stuttgen et al., 2021).
2. Mixed methods studies with no in-depth qualitative work (Chase et al., 2022; Kavanaugh, 2014; Kavanaugh et al., 2016; Lewit-Mendes et al., 2018; Williams et al., 2013).

Search results

In the initial search 1663 articles were identified of which 816 were duplicates, 35 were not in English and 104 were not peer reviewed and were therefore removed. The remaining 708 were appraised using the title and abstract to establish relevance. Of these, 686 were excluded due to irrelevance to the research question or not meeting the inclusion criteria. Of the final 22 papers which were read in full, five were excluded either because they were quantitative or mixed methods and four because the data on young people in HD families were not clearly extractable. This resulted in 13 papers from nine studies being included in the review. A PRISMA diagram (Page et al., 2021) to illustrate this process can be seen in Figure 1.

[Figure 1 about here]

Study characteristics

Study characteristics are shown in Table 1 - B. Studies were carried out between 2007 and 2022 in four countries i.e., USA (*n*=5), Norway (*n*=3), Scotland (*n*=3), Australia (*n* = 2). The following papers used the same participant sample to address differing research aims:

- Forrest Keenan et al. (2009) and Forrest Keenan et al. (2007).
- Kjoelaas, Jensen, et al. (2022), Kjoelaas, Feragen, et al. (2022) and Kjoelaas et al. (2020).

- Williams et al. (2009) and Sparbel et al. (2008)

Thus the 13 papers for inclusion represented data from nine original studies. While some reviews exclude multiple papers from the same study, this was not the approach selected within this review due to the papers' different questions and therefore differing selection of data. In total, data were gathered from 199 individuals who formed sample sizes of between five and 40 of varied HD status at the point of recruitment (160 at risk, 22 gene positive, 16 gene negative, 1 diagnosed). Participants were aged between eight and 65 years old, resulting in current and retrospective accounts of childhood and adolescent experiences of growing up in a family with either a HD parent or close relative. Four of the studies presented a mixture of current and retrospective accounts (Gong et al., 2016b; Kjoelaas, Feragen, et al., 2022; Kjoelaas, Jensen, et al., 2022; Kjoelaas et al., 2020). The decision to include such an age variance was informed by the wider existing literature on young carers which includes ages 5-18 (e.g., Baumann et al., 2006; Gates & Lackey, 1998) and the United Nations' (2003) definition of young people being between 15 – 24. However, research focusing on young people in this age bracket (5-24) in HD families is limited. Consequently, retrospective accounts of growing up in an HD family where the participant was over 25 and reflecting on their experience of being a young person were included.

[Table 1 - B about here]

Quality of the selected studies

Due to study selection in meta-ethnography being guided by what is available (Noblit et al., 1988), and to ensure conceptually rich papers were included, studies were not excluded based on methodological quality. However, quality assessment of included studies was still considered important as the quality of the meta-ethnography is dependent upon this, although it is important to note that appraisal tools have a focus on methodological strength and not conceptual strength (Long et al., 2020; Toye et al., 2014).

To understand the quality of the included studies, the Critical Appraisal Skills Programme (CASP) checklist (Burls, 2014) was used as it has been endorsed for qualitative studies by Cochrane (a source of influential advice on the conduct of reviews) and the World Health Organization (Noyes et al., 2018). The CASP provides a framework for exploring 10 questions with answers of 'yes', 'can't tell' and 'no' to appraise quality of the study. The CASP rating for each study can be seen in Table 1 - C and shows a relatively strong collection of papers. The CASP highlighted the main weakness across papers to be lack of description of the relationship between author and participants in terms of researchers examining their roles and influence during the research.

[Table 1 - C about here]

Analysis

Following the seven steps advocated by Noblit et al. (1988) and meta-ethnography guidance (Sattar et al., 2021), papers were first read and re-read with each read resulting in detailed annotation. This enabled key concepts to be drawn from each paper which were placed into a table (appendix 1 - A).

Concepts were transferred into a second table during translation demonstrating which concept appeared in which paper (appendix 1- B). Through this process, the four third order constructs (i.e., final themes) were formed. Discussion with supervisors throughout this process aided in maintaining the quality of the findings and methodological rigour (Atkins et al., 2008). To improve rigour, two randomly selected papers were coded by a colleague external to the research team with no knowledge of HD. Concepts generated by this individual were compared to those created by the primary author (HC) for the two papers and indicated a high level of similarity. The same colleague also completed a CASP for two papers and generated the same ratings as the primary author.

Findings

Through synthesising the 13 papers, four interrelated constructs were identified: thief of relationships, thief of self, thief of transparency and search for reclamation.

Thief of relationships

HD stole young people's relationships with their parents, family members and friends and contributed to their lack of social support networks. Within this theme were factors that influenced the taking of relationships such as parental absence, grief and loss, loneliness, and confusion. The effects of this were also evident in terms of the difficulties experienced by the young people in their own social relationships.

Studies reported the negative effects of parental absence; this could be due either to parents separating (Forrest Keenan et al., 2007; Kjoelaas, Jensen, et al., 2022) or the parent with HD becoming unavailable due to admission to hospital or a care home. The non-HD parent was also sometimes unavailable due to their caring role or need to work (Forrest Keenan et al., 2007; Williams et al., 2009). This created difficulty in achieving a joint approach or joint understanding: *'my mum (is) working two jobs…it's kind of hard for my dad to understand her stresses and it's hard for her to understand my dad's stresses…it just (kind of) creates an irritable mess'* (Sparbel et al., 2008, p. 331).

The absence of parents often left young people feeling alone and isolated with an intense sense of loss for both parents. This was often accompanied by the awareness of their parents' own losses*: 'my dad has been without a job ever since my mum got sick. So, in a way he got sick too. He doesn't seem to live any more either'* (Kjoelaas et al., 2020, p. 133).

Some young people shared their realisation how, in retrospect, HD had stolen their parent for years before a diagnosis was given. Finding out about the presence of HD caused participants to question their childhood experiences and feelings towards their parents. This was particularly the case for young people whose HD diagnosed family member had been

aggressive, violent or abusive and inconsistent in their boundaries, emotional responses and discipline (Forrest Keenan et al., 2007; Keenan et al., 2009; Kjoelaas, Jensen, et al., 2022; Kjoelaas et al., 2020; Mand et al., 2015). For example: *'my dad is really passive and gentle and friendly, he's very calm, and never says a bad word about anyone, and all of a sudden he was belting my sister and belting the dog, really cutting himself off from the family, really detached, really angry'* (Mand et al., 2015, p. 211). Prior to knowing about HD, some participants were confused by their parents' behaviour, often attributing it to being drunk (Duncan et al., 2007). Finding out about HD created turmoil, questioning whether their parent was hostile and aggressive because of HD or because of who they were prior to HD (Forrest Keenan et al., 2007; Mand et al., 2015).

For those who had knowledge of the parent's HD diagnosis, young people spoke about the difficulties of managing the disease progression. They faced repeated losses with the changes in the parent's personality, as HD seemed to become more dominant, rendering them unable to maintain their parent/child relationship (Kjoelaas, Jensen, et al., 2022; Kjoelaas et al., 2020; Sparbel et al., 2008) or move on in their grief. There was an air of desperation in some reports of the need to grasp the last parts of life with the HD parent and value those moments together, before time ran out: *'trying to, you know, enjoy the last poofs of my mum before something really bad happens'* (Williams et al., 2009, p. 282).

The sense of responsibility young people expressed in having to protect those around them, especially if the young person was a carer, was high (Keenan et al., 2009; Kjoelaas, Jensen, et al., 2022). This sense of responsibility often resulted in young people leaving education and taking on typically adult roles in managing the home or becoming carers (Forrest Keenan et al., 2007; Sparbel et al., 2008). Young people also experienced not being able to sustain friendships and feeling misunderstood by friends when they chose caring over friendship (Kavanaugh et al., 2015b). Some young people were too embarrassed or

apprehensive to bring friends home (Kjoelaas et al., 2020) or reported that friends were avoidant of visiting (Sparbel et al., 2008), meaning that social relationships were difficult to both start and maintain.

Some young people tried to find positive ways to cope, such as keeping distance between the disease and their parents' identity which helped them make sense of difficult behaviour and physical care needs. The refusal to place blame on their HD parent for their behaviour was evident: *'I know that she doesn't mean to, if she wasn't sick, she wouldn't be this way'* (Mand et al., 2015, p. 212).

Feelings were also not always negative as sometimes, despite the loss, participants viewed the non-HD parent providing care with admiration and respect (Sparbel et al., 2008) and described them as a *'hero'* (Keenan et al., 2009) or *'lightening rod'* (Kjoelaas, Jensen, et al., 2022) as they protected others from the impact of HD. This admiration seemed to be expressed when young people witnessed family members' difficult journey when caring for a partner with HD, suggesting the young people in the home were aware of what was occurring despite their relatively young age. Ultimately growing up with an awareness of HD had a significant impact on young people's relationships.

Thief of self

HD stole the young person's sense of self. Within this theme is the impact of HD on a variety of areas in the young people's lives that removed aspects of their identity. This was stolen by HD triggering distressing thoughts about areas such as genetic status and testing, life choices and decision making which seemed to have a negative impact on mental health and esteem. Young people found it difficult to identify and state what their needs were in the face of such dominant other family needs.

Living with inconsistent behaviour from parents affected young people's understanding of themselves and their mental health, resulting in low self-esteem and loss of security and

stability (Kjoelaas, Jensen, et al., 2022): *'I became very insecure…unsure of what I had done wrong'* (Kjoelaas, Jensen, et al., 2022, p. 217). Changes in parental boundaries (what was acceptable childhood behaviour one day may not have been acceptable behaviour the next) was described as *'…an absurdity you cannot understand as a child'* (Kjoelaas et al., 2020, p. 133), resulting in anxiety and apprehension.

Caring roles of the child often extended past the immediate family home where other members of the family were HD positive (Forrest Keenan et al., 2007) such as supporting aunts, uncles or cousins in a HD positive household; this required them to put themselves and their needs on a *'back burner'* (Kavanaugh et al., 2015b). In taking on this responsibility, the identity of the child was lost, with the child often taking a parent role in their relationships (Dondanville et al., 2019). No study documented that the child wanted to take on, or reject, such roles; rather it was viewed as a duty to take care of the ones they loved (Dondanville et al., 2019).

Young people shared how being a child in the HD family felt as though they were unseen (Kavanaugh et al., 2015b; Kjoelaas et al., 2020) and left without protection or provision (Kjoelaas, Jensen, et al., 2022): *'…the kids always seem to get ignored, I mean you were there but you're not listened to..'* (Keenan et al., 2009, p. 1895). Some explained how they protected themselves by becoming non-existent: *'I sort of erased myself...'* (Kjoelaas, Jensen, et al., 2022, p. 219). Thus, young people were not acknowledged in their family homes amid the chaos of the disease. Despite this chaos some participants were able to find a different part of their identity in sports, clubs and with other relatives, which helped increase their sense of belonging, inclusion and being loved (Kjoelaas, Feragen, et al., 2022), although this seemed rare.

Some young people wanted to know their genetic status as they believed this knowledge would give them control over their lives and a sense of a confirmed identity (Mand et al., 2015). Knowing that their test result was positive, and life could potentially be short, some participants described how they chose to live differently, engaging in more meaningful ways to spend their

time and gaining a different perspective to life problems: *'knowing that time is limited makes things that would otherwise seem like bigger deals really seem like not a deal at all'* (Gong et al., 2016a, p. 1190). However, some also experienced distress on hearing the test result, regardless of the outcome (Duncan et al., 2007).

Some young people at risk described themselves as living as though they were gene positive so that they self-prepared for symptoms or a positive result if they were to get tested (Duncan et al., 2007; Forrest Keenan et al., 2015). Similarly to those who tested positive, they described self-monitoring for symptoms (Forrest Keenan et al., 2007) and high levels of anxiety were reported if they noticed a trait in themselves that was like that of their family member with HD (Forrest Keenan et al., 2007). Living at risk was described as *'... having a noose around your neck constantly. You don't know if it's there or not and that's what makes it so much harder. I have a 50/50 (chance of a) death sentence'* (Kjoelaas et al., 2020, p. 135). The impact on self of test-taking was viewed differently by different participants; one participant expressed how growing up gene positive would be a *'weird'* experience (Mand et al., 2015) but living at risk was somehow easier to understand and *'mould yourself around'* (Mand et al., 2015, p. 213).

The management of the news of being gene positive was described as a process for some; the knowledge became less salient as time progressed *'and then one day, it just goes to the back of your mind'* (Forrest Keenan et al., 2007, p. 125). However, for others, being gene positive influenced decisions not to have a family or take time investing in careers and instead they lived life with the motto of *'work harder, achieve it faster'* (Gong et al., 2016a, p. 1190), living within a window of allocated time. This expanded into decisions around partners and ensuring that, for those who wanted to have a family, they searched for an older partner with the priority of having children (Gong et al., 2016a) rather than choosing a partner for other reasons. Some reported settling down with someone who knew about their genetic status

through fear that no-one else would accept them (Gong et al., 2016a). Thus, HD continued to dictate life choices and family life.

Thief of transparency

HD stole transparency and instigated secret keeping. Within this theme is lack of transparency concerning disclosure. Some families kept the presence of HD to themselves only allowing a select few to know about its existence. Being able to keep HD a secret also seemed to improve aspects of life for some young people such as protecting family and enabling young people to be the same as their peers.

Some families did not discuss HD with each other despite HD being present and family members being able to see the effects of the disease (Forrest Keenan et al., 2015; Kjoelaas et al., 2020; Mand et al., 2015). Keeping the presence of HD a secret in these ways resulted in the absence of internal familial and external social support (Forrest Keenan et al., 2015; Mand et al., 2015). Explanations for the secrecy were that families viewed HD as shameful (Mand et al., 2015), feared judgement from friends, were concerned about the impact on employment and insurance cover (Gong et al., 2016a) or were unsure of when the right time would be to tell their children (Kjoelaas et al., 2020).

Some families did not speak about HD until the effects of the disease were becoming unavoidable, despite the young people in the home being aware of the stage of progression (Forrest Keenan et al., 2009). Then sharing the news of HD and its effects came as a shock to the young people (Kjoelaas et al., 2020). Parents who were in denial about their own status and symptoms were reported to feed into the idea of keeping the disease a secret (Kjoelaas, Feragen, et al., 2022). This had a negative effect on young people who wanted support or help but felt unable to pursue it: *'it's the most difficult part about this whole thing when you are a young*

carer who wants help, but you are not getting anywhere because your parent is denying that they have a disease' (Kjoelaas, Feragen, et al., 2022, p. 665).

For those who did speak about HD, family support was not sought by some young people who expressed a preference to withhold their worries and concerns from family to protect them from the effects of their parents' genetic history (Dondanville et al., 2019; Kjoelaas, Feragen, et al., 2022). However, young people who knew the family HD history and felt they had a family member who truly understood them and their experience felt close to that person (Forrest Keenan et al., 2009). The supporting person (e.g., non-HD parent) could then interject and support the child during unreasonable challenges from the HD positive parent, indicating the supportive power of being transparent (Kjoelaas, Jensen, et al., 2022).

Others shared how, within their immediate family, HD would be spoken about and discussed but outside of the family system it was not discussed or shared (Forrest Keenan et al., 2007; Kjoelaas et al., 2020; Mand et al., 2015). The opposite of this also occurred, with immediate family not discussing the disease but those closely linked, such as cousins, sharing the news and genetic risk of the disease with the young people (Duncan et al., 2007), resulting in negative familial impacts such as lack of trust. Despite how they were told, it appears that young people who were told about HD coped better with the impact of the disease and making life adjustments than those who were not told. Those living in families where HD was openly discussed had a strong awareness of how the disease shortened life expectancy for their parent and the possibility that HD could shorten their lives too. This brought an appreciation for life and awareness of how much time they may have left (Forrest Keenan et al., 2015).

In terms of how transparency affected social relationships, some young people reported that they wanted to be a normal child and not be treated as any different from their peers. To achieve this, they kept the knowledge of HD in their family a secret from their peers when they could: *'I didn't want them to treat me differently. I just wanted to be the same as everyone else'*

(Kjoelaas, Feragen, et al., 2022, p. 668). Some young people had a desire to be able to exist as an individual outside of the disease, to be viewed as a normal person with their peers and have a life that was independent of HD, and this removed the ability of the young people to be able to choose to be transparent.

Search for reclamation

Young people seemed to be searching for reclamation of things stolen from them. Within this theme are ways that young people tried to reclaim aspects of their lives, such as creating a sense of normality, searching for understanding and validation, information seeking and seeking professional support and being on a journey to acceptance. For some this search was successful at times, though it appeared hard and arduous for all young people.

Young people used varied methods to cope with their situation. One such method was an attempt to bring a sense of normality to their situation. Some young people held the view that they were a normal family despite HD and that they should be treated as such (Kavanaugh et al., 2015b), which brought a sense of resilience and self-preservation to their approach. Normalisation of HD as a family illness or family problem or *'quirk'* seemed to help families adjust and cope (Forrest Keenan et al., 2007; Forrest Keenan et al., 2009; Kavanaugh et al., 2015b).

Recognition of the difficulties experienced and support through these was desired but was not always available. Some young people wanted family and friends to accept that they could not understand the young person's situation but nevertheless could offer recognition of how difficult it was (Forrest Keenan et al., 2015; Gong et al., 2016a; Kavanaugh et al., 2015b; Kjoelaas, Feragen, et al., 2022; Sparbel et al., 2008; Williams et al., 2009). However, others felt such recognition and support was impossible: *'I don't think they understand that they don't have the full picture and that I don't feel like I can talk to them at all. Even though I know they*

are only trying to be supportive (and) they want me to talk to them and want to help me, it's like….it only makes it worse'… (Kjoelaas, Feragen, et al., 2022, p. 666).

Some young people actively sought information for example, via the TV, internet, or through taking part in research, to find out about the disease and what was happening in their family (Forrest Keenan et al., 2009). Some reported being shocked to learn of their 50% risk (Forrest Keenan et al., 2007; Forrest Keenan et al., 2009) having not realised the 'family problem' was a hereditary genetic condition. As well as aiding transparency (theme 3), having a name to the problem and explanation of what was happening helped young people accept and deal with the disease and not see it as overwhelming (Forrest Keenan et al., 2009): *'…the more information I got, the safer I felt. In terms of the possibility that I could become ill one day too, I learned that everyone with HD is not the same. That was really good to know.'* (Kjoelaas, Feragen, et al., 2022, p. 664). This seemed to be in line with young people's experiences of professional support regarding gaining information.

Young people wanted support with finances and planning for future caring needs (Kavanaugh et al., 2015a) but professional support was sparse (Keenan et al., 2009; Kjoelaas, Feragen, et al., 2022; Williams et al., 2009). In particular, young people wanted support with the emotional aspects of their journey (Forrest Keenan et al., 2015), as this was not routinely provided by genetic counsellors or other sources even after finding out their results (Gong et al., 2016a). Professionals gaining a true understanding of the situation was described as a key factor for young people feeling supported and connected to others (Kjoelaas, Feragen, et al., 2022). However, young people were also aware of negative effects some support could have. For example, they worried that a parent may be assessed as not able to provide safe care or needing additional professional care. Such fear often resulted in avoidance of support seeking.

Young people who had formed meaningful connections with another family member (aunt, uncle, grandparent), a social group or a professional service seemed able to cope better

due to the sense of connection and belonging such relationships gave them. This dynamic support was not available to most young people and to those who did have access to such support, many challenges were experienced. These challenges included the desire to self-protect so that the young person did not feel overwhelmed, the desire to protect the family from intrusion and judgement and the young person feeling misunderstood due to professionals' lack of HD knowledge and withdrawing from support as a result (Kjoelaas, Feragen, et al., 2022).

Self-acceptance was a key factor in being able to move on and hold HD in a place in the mind that enabled people to continue with life (Dondanville et al., 2019; Duncan et al., 2007; Forrest Keenan et al., 2007). Having a positive attitude and making plans (Dondanville et al., 2019; Forrest Keenan et al., 2007), as well as finding out their own HD status and a sense of hope, aided taking control of life (Duncan et al., 2007) and feeling more able to adjust, cope and make decisions.

Discussion

This review explored the experience of young people growing up in a family with HD. Through synthesising 13 papers, four interrelated constructs were identified: thief of relationships, thief of self, thief of transparency and search for reclamation. HD stole different aspects of the young people's childhood, which the young people tried their best to reclaim. In terms of the line of argument, i.e., how the themes are interrelated, growing up in a family with HD can be a multifaceted and challenging experience. The complex interplay between the removing of relationships, the sense of self and the ability to be open and transparent often resulted in susceptibility to mental health challenges such as low mood, low confidence, low esteem and feelings of isolation. In efforts to reclaim what was stolen and control some of the impact on their lives, young people had to face their fears and their own feelings about themselves, worries about their family members, fears of becoming like their parent and what

that might mean for them. Young people sought support; however this was sparse and inconsistent in quality and availability.

The review highlights the significant level of distress experienced by young people in families with HD. Similarly, quantitative studies have reported that young people in HD families experience high levels of emotional, social and practical burden (Lewit-Mendes et al., 2018) and that HD families experience high levels of dysfunction, including sub-optimal parenting (control and abuse) from both the HD and non-HD parent (Vamos et al., 2007). The qualitative approach of this review adds the experiential voice of young people and a detailed understanding of the contributing factors that have resulted in such distress through the descriptions of HD as a thief and young people's resulting need to try and reclaim aspects of themselves and their lives.

In theme 1 (thief of relationships) young people expressed how the unavailability of their parents was difficult to manage. Such absence may have influenced parent/child attachment. Attachment refers to being psychologically connected to other people (Bowlby, 1979) and attachment theory explains the life-long importance of the bond between children and their primary caregivers. The theory argues that it is important that parents are attuned to their children's needs (Howe, 2011) for children to develop secure, safe and meaningful relationships. A number of attachment styles have been proposed (Levy et al., 2011) but all agree that secure attachment is most important in terms of supporting optimal development both in childhood and further into adulthood. Indeed, Van der Meer et al. (2006) found that HD families experienced higher levels of preoccupied and unresolved/disorganised attachments and lower levels of secure attachment styles when compared to a non-HD population using the Adult Attachment Interview (George et al., 1996). The lack of a secure parent figure or secure base providing comfort and support could result in the young people developing a negative attachment style (Bowlby, 1988) which could cause difficulty relating

to the self and other people resulting in psychological and emotional distress that extends into future relationships (McCarthy & Taylor, 1999). This is supported by research findings that young people with a HD parent display lower levels of secure adult attachment and higher levels of unresolved or disorganised attachment when compared to families without a HD parent (Van der Meer et al., 2006)

Social relationships were also affected, with young people feeling unable to let peers fully into their lives. Having social connection with peers and communities is important as people with such connections are generally happier, have better physical and mental health and live longer (Holt-Lunstad et al., 2010). Peer to peer relationships are important as they form an essential part of development concerning cognition, social interaction (Rubin et al., 2011), prosocial behaviours (Eisenberg et al., 2019), emotional regulation and adjustment (Contreras & Kerns, 2000) and reciprocal roles, which are essential in forming healthy relationships and a sense of identity and belonging (Parker et al., 2006).

Theme 2 (thief of self) showed how HD interfered with the identity that young people wanted to form and negatively affected their mental health. Traumatic events experienced in childhood (known as ACEs – adverse childhood experiences) have been found to be associated with psychological distress and poor physical health outcomes in non-HD populations (Boullier & Blair, 2018; Hughes et al., 2017). Young people in HD families have often experienced multiple ACEs (for example, anticipatory mourning, loss of a member of their family unit, possible domestic violence and aggression in their home) placing them at higher risk of future physical and psychological health complications (Monnat & Chandler, 2015). As ACEs originate mostly though not always from parent-child relationships that experience disruption, young people may have a negative view of the self and poor social skills resulting in compromised identity development (Wong et al., 2019). This is important to hold in mind

when working with young people from HD families who may experience such disruption in their relationships with their parents.

Another finding of this review is the negative effect of secrecy (theme 3: thief of transparency). A study with young people living with chronic illness found the main factor influencing illness disclosure with peers was that the peer had a shared experience with illness (Kaushansky et al., 2017). When disclosure did not occur, similarly to the young people in this review, this was due to fear of rejection, pity or being viewed as different (Kaushansky et al., 2017). HD research with adults documents various examples of secrecy such as hidden coping strategies, including substance misuse (Aubeeluck & Moskowitz, 2008), recognising but disguising personality or health changes in family members (Hayes, 1992), the bidirectional nature of secret keeping between the member with HD, spouse and other family members (Kessler, 1993) and the impact of secret keeping in couple relationships (Forrest Keenan et al., 2013). These examples, excluding couples' relationships, were identified within the papers in this review which focused solely on young people.

Despite the challenges that living in a family with HD brought, young people fought to live their own lives (theme 4: search for reclamation). One way reclamation was sought was through acceptance of HD as being a normal part of family life. Studies on chronic illness document the positive effect of acceptance on a person's ability to adapt to living with illness (Livneh et al., 2006; Telford et al., 2006). Other studies have also noted that in families where HD was openly discussed and the presence of the genetic risk and disease accepted, positive changes were reported such as a change in perspective and an ability to focus on enjoyable experiences (Maxted et al., 2014).

Clinical Implications

Within theme four, young people spoke about the importance of feeling understood, both by their friends and family, but also by professionals. While information provision was

valued, emotional support was frequently highlighted as lacking. Well placed to meet this need are professionals such as psychologists and mental health specialists.

When providing support to young people and their families, both the systems surrounding a young person and the young person as an individual need to be considered. Biopsychosocial models adapted for families with genetic illnesses, such as the Family Systems Genetic Illness Model (Rolland & Williams, 2005), suggest a family focused method to enable effective communication of genetic risk and then understand the impact of such risk on the family unit (Miller et al., 2006).

The Family Systems Genetic Illness model is created with a focus on the sharing of genetic risk and effects of genetic illness on the family system, rather than considering wider impacts on children and young people. Therefore, this model may be helpful to understand the family and the stages of their HD journey by offering a structured framework of progression within which to work. Family therapy approaches may also be helpful. For example, using the family life cycle model could provide a framework for helping families progress through life stages while acknowledging and addressing their challenges and narrating their own HD story (Brouwer-DudokdeWit et al., 2002).

A resilience framework, such as the strength-based cognitive therapy four step model (Padesky & Mooney, 2012), may also be useful to explore with young people in HD families and may add a preventative aspect. Such a model focuses on strengths and brings these into awareness using imagery and metaphor. The goals of this are to increase resilience rather than reach resolution. While such a singular approach may be useful, there is also value in exploring family focused interventions that may assist young people. Aspects from the Child Illness and Resilience Program (CHiRP) (Hamall et al., 2014) could be adapted for use with HD families. The programme, based on family resilience theory, aims to provide intervention to improve resilience of families experiencing childhood illness. It focuses on the strengths of the family,

forming strategies and processes to improve resilience (function, coping, utilising resources). Utilising family-based interventions as a preventative tool may be useful. Family-based interventions in chronic disease and illness management have shown promising outcomes concerning family relationships and cohesion (Fisher & Weihs, 2000).

Future research

Two avenues for future research should be considered. Firstly, as there is currently no routine support for this group in the UK, it would be beneficial for research to explore ways of improving the assessment and formulation of young people in HD families and the need for psychological support. A recent formulation model for use with individuals affected by HD has been proposed (Dale et al., 2022). The model offers prompts for individual components that require consideration within the lives of those affected by HD (e.g., HD narrative, HD triad of symptoms) and offers guidance for professionals to facilitate an effective HD formulation. This could be adapted for young people in HD families to provide guidance for professionals with little knowledge of HD concerning what areas need to be discussed. It could also enable client-led education (Hatcher et al., 2012) for the professional using it as the professional would gain knowledge of HD and the young person's experience of HD during such conversations. If the tool is successful in its adaptation, it may be able to inform the production of other tools such as HD focused interview schedules which could inform professionals how to begin conversations about HD within their clinical settings. In terms of psychological support, it is accepted that intervention research in this area is minimal (Zarotti et al., 2020). However, a feasibility study is currently being trialled to examine the use of a particular psychological approach, e.g., guided self-help for anxiety, for people with HD (Dale et al., 2023). Such psychological intervention could be adapted for use with young people. Exploring resilience-based approaches previously mentioned on an individual and family level, may also be useful

considering the lack of good evidence and relative lack of any evidence concerning psychological interventions for HD (Zarotti et al., 2020).

Secondly, this review highlights that most research on young people and HD draws out the negative aspects of living in a HD family. It may be beneficial to focus on the positives that young people can experience, adopting a more positive psychology perspective (Seligman & Csikszentmihalyi, 2000) to understand further the life experience of this group and progress away from the persistent pathological-laden narrative (Barak & Achiron, 2009).

Limitations

Although 13 papers were included, these represent only 9 studies, and this may have impacted the representation of second order constructs. All studies were conducted in western countries and therefore the review does not represent the experiences of young people who do not have access to healthcare and support systems present in emerging economies. The CASP tool revealed that few papers described the researchers' possible influence on data analysis therefore future research may benefit from explicit statements concerning reflexivity.

Conclusion

Young people in HD families endure emotional, social, and practical burden as well as dealing with their own genetic risk and legal limits to independent decision making. This review contributes to a fuller picture of some of the complexity of these interlinked issues and the challenges young people face living in HD families.

Author Contributions

Authors Hollie Cooper, Professor Jane Simpson, Dr Maria Dale and Dr Fiona Eccles contributed to this work. Hollie Cooper made substantial contributions to the acquisition, analysis, and interpretation of data for the work which was supported by Professor Jane Simpson, Dr Maria Dale and Dr Fiona Eccles. All authors made substantial contributions drafting the work and revising it critically for important intellectual content.

Conflict of interest statement

Authors Hollie Cooper, Professor Jane Simpson, Dr Maria Dale, Dr Fiona Eccles declare that they have no conflict of interest.

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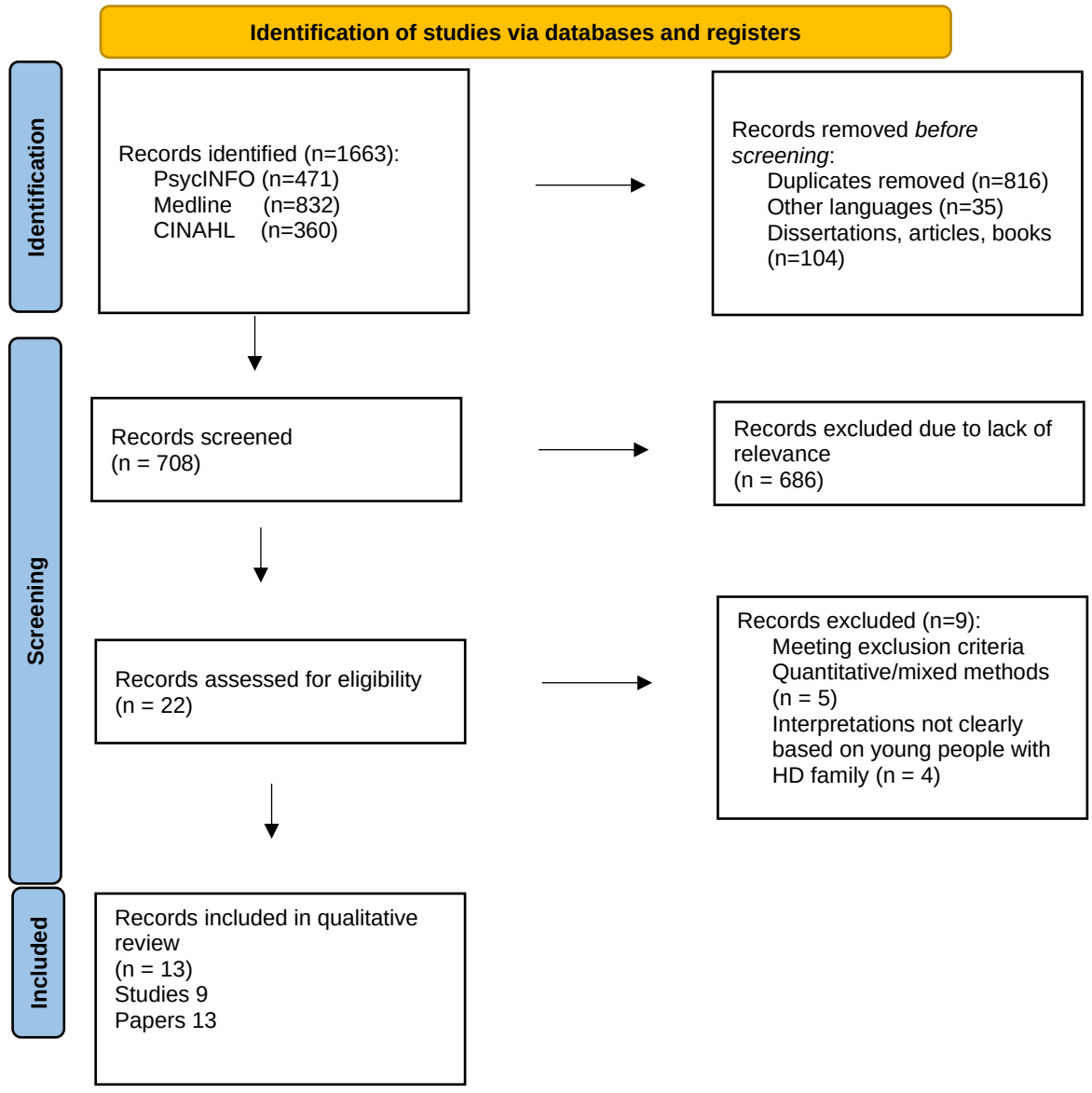
Figure 1

A PRISMA Flow Diagram to Illustrate Study Identification via databases.



From: Page MJ, McKenzie JE, Bossuyt PM, Boutron I, Hoffmann TC, Mulrow CD, et al. The PRISMA 2020 statement: an updated guideline for reporting systematic reviews. BMJ 2021;372:n71. doi: 10.1136/bmj.n71

For more information, visit: http://www.prisma-statement.org/

Table 1 - A

Systematic Review Search Strategy example (PsycINFO)

1 (qualitative research)	([1]DE "Qualitative Measures" OR DE "Questionnaires" OR DE "General Health Questionnaire" OR DE "Interviewing" OR DE "Interviewers" OR DE "Attitudes" OR DE "Abortion (Attitudes Toward)" OR DE "Adolescent Attitudes" OR DE "Adult Attitudes" OR DE "Aged (Attitudes Toward)" OR DE "Aging (Attitudes Toward)" OR DE "Attitude Change" OR DE "Attitude Formation" OR DE "Attitude Similarity" OR DE "Child Attitudes" OR DE "Childrearing Attitudes" OR DE "Client Attitudes" OR DE "Community Attitudes" OR DE "Computer Attitudes" OR DE "Consumer Attitudes" OR DE "Counselor Attitudes" OR DE "Cultural Attitudes" OR DE "Death Attitudes" OR DE "Disabled (Attitudes Toward)" OR DE "Drug Usage Attitudes" OR DE "Eating Attitudes" OR DE "Educational Employee Attitudes" OR DE "Employee Attitudes" OR DE "Employer Attitudes" OR DE "Environmental Attitudes" OR DE "Explicit Attitudes" OR DE "Family Planning Attitudes" OR DE "Female Attitudes" OR DE "Gender Role Attitudes" OR DE "Health Attitudes" OR DE "Health Personnel Attitudes" OR DE "Ideology" OR DE "Implicit Attitudes" OR DE "Job Applicant Attitudes" OR DE "Law Enforcement Employee Attitudes" OR DE "Male Attitudes" OR DE "Marriage Attitudes" OR DE "Obesity (Attitudes Toward)" OR DE "Occupational Attitudes" OR DE "Parental Attitudes" OR DE "Paternalism" OR DE "Political Attitudes" OR DE "Preferences" OR DE "Psychologist Attitudes" OR DE "Public Opinion" OR DE "Racial and Ethnic Attitudes" OR DE "Sex Role Attitudes" OR DE "Sexual Attitudes" OR DE "Socioeconomic Class Attitudes"

[1] DE refers to descriptors. These are specific subject terms that control how subjects are searched and have been created by the American Psychological Association.

	OR DE "Sports (Attitudes Toward)" OR DE "Stereotyped Attitudes" OR DE "Student Attitudes" OR DE "Teacher Attitudes" OR DE "Work (Attitudes Toward)" OR DE "World View" OR DE "Ethnology" OR DE "Phenomenology" OR DE "Discourse Analysis" OR DE "Observation Methods" OR DE "Direct Observation" OR DE "Participant Observation" OR DE "Qualitative Methods" OR DE "Focus Group" OR DE "Grounded Theory" OR DE "Interpretative Phenomenological Analysis" OR DE "Narrative Analysis" OR DE "Semi-Structured Interview" OR DE "Thematic Analysis") OR TI (mixedmethod* OR "mixed method*" OR mixed-method* OR qualitative OR interview* OR experience* OR "focus group∗" OR ethnograph∗ OR fieldwork OR “field work” OR “key informant” OR ((“semi-structured” OR semistructured OR unstructured OR informal OR “in-depth” OR indepth OR “face-to-face” OR structured OR guide) N3 (interview∗ OR discussion∗ OR questionnaire∗))) OR AB (mixedmethod* OR "mixed method*" OR mixed-method* OR qualitative OR interview* OR experience* OR "focus group∗" OR ethnograph∗ OR fieldwork OR “field work” OR “key informant” OR ((“semi-structured” OR semistructured OR unstructured OR informal OR “in-depth” OR indepth OR “face-to-face” OR structured OR guide) N3 (interview∗ OR discussion∗ OR questionnaire∗)))
2 (Huntington’s Disease)	DE "Huntingtons Disease" OR TI huntington* OR AB huntingto
3	#1 AND #2

Table 1 - B

Study characteristics

Author	Year	Title	Country	Participants	HD status	Aims	Method of data collection	Method of analysis	Findings
Dondanville et al. (2019)	2018	"This could be me": exploring the impact of genetic risk for Huntington's disease young caregivers.	USA	13 individuals, 15-25yr old recruited from local youth groups/support groups 11 female 2 male	At risk	Understand the interaction between young caregivers' perception of risk, the caregiving experience and their thoughts about predictive testing.	Interviews	Thematic analysis	Caregiving is affected by genetic risk which evokes feelings about the future and possible diagnosis and affects plans for testing. Knowledge for genetic counsellors and support needed for young people.
Duncan et al. (2007)	2007	"Holding your breath": interviews with young people who have undergone predictive genetic testing for Huntington disease.	Australia	15-24yrs, 8 young people who had undergone testing, 4 male 4 female	x2 gene positive, x6 gene negative	Explore the experience of predictive testing from the young person's perspective, document the impact of testing on young person's lives	Interviews	Thematic analysis	Three themes: living as though gene positive Risk behaviours, complex pasts. Themes after testing: identity, living again. All reported on testing in the wider context of growing up in a HD family. No one regretted testing.
Forrest Keenan et al. (2015)	2015	Help or hindrance: young people's experiences of predictive testing for Huntington's disease.	Scotland	17-26yrs, 12 participants recruited via genetic services	All at risk at recruitment	Explore young people's experiences of predictive testing, the impact of the result and gaps in support	Interviews	Thematic analysis	3 testing experiences regardless of result: empowerment, ambivalent, poor experience. Changes in family dynamics made the post-test stage difficult.
Forrest Keenan et al. (2009)	2009	How young people find out about their family history of Huntington's Disease	Scotland	33 Young people aged 8-28 X21 female X12 male	X26 at risk X1 gene positive X5 gene negative X1 diagnosed	Explore young people's experiences of finding out about the presence of HD in the family	Interviews	Thematic Analysis	Four main themes: always been told, gradually told, kept a secret, new diagnosis
Forrest Keenan et al. (2007)	2007	Young people's experiences of growing up in a family affected by HD	Scotland	33 Young people aged 8-28 X21 female X12 male	X26 at risk X1 gene positive X5 gene negative X1 diagnosed	To describe the experience of young people growing up in HD families	Interviews	Thematic analysis	Four main themes: young people as carers, worried well, those who cope, those in need
Gong et al. (2016a)	2016	Impact of Huntington's Disease Gene-positive status on pre-symptomatic young		15 Young people aged 20-33 12 female 3 male	All carriers	To explore how a positive result affects the attainment of	Interviews	Grounded theory and thematic analysis	Three main categories: changes in attitude and approach to life, influences on milestones of adulthood, suggestions for genetic counsellors – new finding that young

		adults and recommendations for genetic counsellors				milestones in education, relationships etc			adults have awareness that healthy years are limited.
Kavanaugh et al. (2015b)	2015	"It'd be nice if someone asked me how I was doing. Like,'cause I will have an answer": Exploring support needs of young carers of a parent with Huntington's disease	USA	40 participants between 12 -20with a family member with HD	At risk	To understand the roles of young care givers and their support needs	Interviews	Content analysis	Themes: instrumental support, emotional support, personal needs – implications for social work and health care professionals in deigning support programs
Kjoelaas, Jensen, et al. (2022)	2022a	'I knew it wasn't normal, I just didn't know what to do about it': adversity and caregiver support when growing up in a family with HD	Norway	36 participants aged 13-65 yrs old	At risk	Explore ACEs of children who grew up in HD families and understand their perception of caregiver support	Semi-structured interviews	Interpretative Phenomenological Analysis	Participants with support tolerate adversity better than those without support who feel overwhelmed
Kjoelaas, Feragen, et al. (2022)	2022b	Social support experiences when growing up with a parent with Huntington's disease	Norway	36 participants aged 13-65 yrs old	At risk	Explore young peoples experience of accessing social support outside of the parent-child relationship in HD families	Semi-structured interviews	Thematic Analysis	Social support can help when there is a lack of support at home though there are barriers to be addressed.
Kjoelaas et al. (2020)	2020	The ripple effect: a qualitative overview of challenges when growing up in families affected by HD	Norway	36 young people and adults 13-65yrs old	At risk	Explore the challenges of growing up in a HD family	Semi-structured interviews	Thematic analysis	Four main themes: family functioning, emotional and reactions, social functioning, public and care services.
Mand et al. (2015)	2015	"I'm scared of being like mum": the experience of adolescents living in families with Huntington disease	Australia	10 young people, 13-20 years old 9 under 18 at the time, non-requested a test. Purposive sample	At risk	Explore psychosocial context of young people in HD families and understand their experiences and challenges they face	Semi-structured interview	Thematic analysis	Young people in HD families face greater responsibilities and stressors
Sparbel et al. (2008)	2008	Experiences of teens living in the shadow of HD	Canada	32 young people 14-18 yrs old	X27 at risk X5 negative	Explore the experience of teens living in HD families	Focus groups	Content analysis	Data showed a complex and often painful family environment. Watching and waiting Living in the shadow Alone in the midst of others Family life is hard Having to be an adult
Williams et al. (2009)	2009	Caregiving by teens for family members	Canada	32 young people 14-18 yrs old	As above	Explore the experiences of	Focus groups	Content analysis and	Two themes consistent with adult caregiving literature: tasks and responsibilities and

		with Huntington's Disease				teens caring for family members with HD		narrative synthesis	subjective burden. Two these specific to young care givers: caregiving in the context of personal risk and decisional responsibility.

Table 1 - C

Critical Appraisal Skills Programme

CASP	Dondanville et al., (2018)	Duncan et al., (2007)	Forrest - Keenan et al., (2007)	Forrest Keenan et al., (2009)	Forrest Keenan et al., (2015)	Gong et al., (2016)	Kavanaugh et al., (2015)	Kjoelaas et al., (2022a)	Kjoelaas et al., (2022b)	Kjoelaas et al., (2020)	Mand et al., (2015)	Sparbel et al., (2008)	Williams et al., (2009)
Was there a clear statement of the aims of the research?	Y	Y	Y	Y	Y	Y	Y	Y	Y	Y	Y	Y	Y
Is qualitative methodology appropriate?	Y	Y	Y	Y	Y	Y	Y	Y	Y	Y	Y	Y	Y
Was the research design appropriate to address the aims of the research?	U	U	Y	Y	Y	Y	Y	Y	Y	Y	U	Y	Y
Was the recruitment strategy appropriate to the aims of the research?	Y	Y	Y	Y	Y	Y	Y	Y	Y	Y	U	Y	Y
Was the data collected in a way that addressed the	Y	Y	Y	Y	Y	Y	Y	Y	Y	Y	Y	Y	Y

research issue?													
Has the relationship between researchers and participants been adequately considered	N	U	U	U	U	Y	U	Y	U	U	N	U	U
Have ethical issues been taken into consideration ?	U	U	U	U	U	U	U	Y	Y	Y	U	Y	Y
Was the data analysis sufficiently rigorous?	U	Y	Y	Y	Y	Y	Y	Y	Y	Y	U	Y	Y
Is there a clear statement of findings?	Y	Y	Y	Y	Y	Y	Y	Y	Y	Y	Y	Y	Y
How valuable is the research?	Y	Y	Y	Y	Y	Y	Y	Y	Y	Y	Y	Y	Y

Y – yes: the study fulfilled majority of the prompts per question
N – no: the study did not fulfil many of the prompts per question
U – unclear: difficult to establish a definitive yes or no in response to prompts per question

Appendix 1 - A

Example to illustrate data extraction to form constructs

Study: Forrest Keenan, K. F., van Teijlingen, E., McKee, L., Miedzybrodzka, Z., & Simpson, S. A. (2009). How young people find out about their family history of Huntington's disease. *Social Science & Medicine*, *68*(10), 1892-1900.

1st order: Participants	2nd order: primary authors	3rd order: meta ethnographer	3rd order construct title contributed to
'..all I can remember about my granddad is this grumpy man sitting in a wheelchair….he couldn't move and his speech was very poor…I would hear grandad screaming and shouting… so you see things and you hear things'	Always been told about HD – this helped young people adapt to the presence of HD and young people who had known for a longer time seemed to cope better with eh impact and losses HD caused.	Symptoms affect the ability of the person to have relationships and engage in them shell of a person – aspects that made the person who they were have been removed by HD, abilities to be active in a two way relationship are compromised and deteriorating, what made them who they were was disappearing – YP reminded themselves of what made the person who they were to remember the person above the disease recurrent loss – as HD progresses more of the person has gone at each stage and this caused high levels of distress stolen parent – the disease took over the parent so that they still looked like the parent to an extent (HD ravished the body also) but personality wise were very different. awareness – unable to escape from the deterioration and loss of the parent/child interaction	Thief of relationships search for reclamation
'My gran used to cry for hours with me sat on her lap, giving me a cuddle'		Symptoms – became apathetic, loss of empathy and caring nature Shell of a person – aspects that made the family member who they were were disappearing/being taken Demands of time – awareness that as the disease progresses	Thief of relationships Thief of self

		death approaches/loss of the person approaches Split roles – the child often because the parents of had to replace some of the adult roles yet still be able to function as a child in school/with friends. Unwell family members were still Gran/Mum etc but also an ill patient. Admiration for continuing care – desire to not let the disease win and to still love and care for family Shared experience – not alone, have support, help, love Connection – still feel connected to people through memories, looking for the person above the disease	search for reclamation

Appendix 1 - B

Table to illustrate constructs in included papers.

	Dondanville et al., (2018)	Duncan et al., (2007)	Forrest Keenan et al., (2007)	Forrest Keenan et al., (2009)	Forrest Keenan et al., (2015)	Gong et al., (2016)	Kavanaugh et al., (2015)	Kjoelaas et al., (2020)	Kjoelaas et al., (2022a)	Kjoelaas et al., (2022b)	Mand et al., (2015)	Sparbel et al., (2008)	Williams et al., (2009)
Person vs HD	X	X	X	X	X	X	NA	X	X	NA	X	X	X
Relationships & connection	X	X	X	X	X	X	X	X	X	X	X	X	X
Relationships removed by HD	X	X	X	X	X		X	X	X	X	X	X	X
Education	X	X	X	X	X	X	NA	X	NA	NA	X	X	X
Childhood	X	X	X	X	X	X	X	X	X	X	X	X	X
Mental health	X	X	X	X	X	X	X	X	X	X	X	X	X
Identity	X	X	X	X	X	X	X	X	X	X	X	X	X
Choice/control	X	X	X	X	X	X	X	X	X	X	X	X	X
Age of testing, age to know about HD	X	NA	X	X	NA	X	NA	X	NA	NA	X	NA	NA
Understanding & information seeking	X	X	X	X	X	X	X	X	X	X	X	X	X
Professionals	X	X	X	X	X	X	X	X	X	X	X	X	X
Keeping secrets, denial	NA	X	X	X	X	X	NA	X	NA	X	X	NA	NA
Coping	X	X	X	X	X	X	NA	X	X	X	X	X	X

Appendix 1 - C

Journal of Genetic Counselling Author Guidelines

AUTHOR GUIDELINES

SECTIONS

1. AIMS AND SCOPE

The Journal of Genetic Counseling (JOURNAL), published for the National Society of Genetic Counselors, is a timely, international forum addressing all aspects of the discipline and practice of genetic counseling. The JOURNAL focuses on the critical questions and challenges that arise at the interface between rapid advances in genetics and technology and the impact on individuals and communities at genetic risk. The publication provides genetic counselors, other clinicians and health educators, laboratory geneticists, bioethicists, legal scholars, social scientists, and other researchers with a premier resource on genetic counseling topics in national, international, and cross-national contexts.

As a crucial resource for genetic counselors and associated professionals, the JOURNAL'S primary purpose is to report original research in the following areas:

- **Genetic Counseling Theory, Methods, and Practice:** addresses theory development and evaluation, methods development and evaluation, current practice, and/or outcomes research relevant to the discipline and practice of genetic counseling in clinical or non-clinical settings;
- **Public Health, Public Policy, and Access and Genetics Service Delivery:** addresses public health genomics, health behaviors, policy aspects related to genetic counseling and genetic testing, precision medicine, models of genetics services delivery;
- **Education and Genetics Professional Workforce Issues:** addresses educational training, professional development, and workforce topics related to genetic counseling;
- **Ethical, Legal, Psychological, and Social Issues:** addresses ethical, legal, psychological, and/or social issues related to genetic counseling, genetic testing, genetic services, and/or genetic information regarding individuals, communities, and the public
- **Risk Assessment:** addresses algorithms, theoretical models, or empirical data for use in genetic counseling risk assessment
- **Minority and Health Disparities:** addresses diversity, equity, and inclusion topics relevant to the practice and discipline of genetic counseling

Note: The Journal of Genetic Counseling does not publish research involving non-human animals.

Return to Guideline Sections

2. SUBMISSION

Once the submission materials have been prepared in accordance with the Author Guidelines, manuscripts should be submitted via the JOURNAL'S Editorial Manager site: **https://www.editorialmanager.com/jogc/default.aspx**. More details on how to use Editorial Manager are also available at **https://www.editorialmanager.com/jogc/default.aspx**.

Need assistance? For help with submissions, please contact the Editorial Office at JOGC@Wiley.com. When necessary, the Editorial Office staff may refer questions to the Editor-in-Chief.

A manuscript is considered for review and possible publication on the condition that it is submitted solely to the JOURNAL, and that the manuscript or a substantial portion of it is not under consideration elsewhere. Preprints and/or presentation of the content at meetings prior to submission are acceptable. However, authors should kindly note that submission implies that the content has not been published or submitted for publication elsewhere except as a brief abstract in the proceedings of a scientific meeting or symposium or as a preprint. Note, the JOURNAL uses iThenticate's CrossCheck software to detect instances of overlapping and similar text in submitted manuscripts.

The submission system will prompt the author to use an ORCiD ID (a unique author identifier) to help distinguish their work from that of other researchers. **Click here** to find out more.

Data Protection Statement: By submitting or reviewing a manuscript for the JOURNAL, your name, email address, and affiliation, and other contact details the JOURNAL might require, will be used for the regular operations of the JOURNAL, including, when necessary, sharing with the publisher (Wiley) and partners for production and publication. The JOURNAL and the publisher recognize the importance of protecting the personal information collected from users and have practices in place to maintain the security, integrity, and privacy of the personal data collected and processed. You can learn more **here**.

Return to Guideline Sections

3. MANUSCRIPT TYPES AND GENERAL REQUIREMENTS

MANUSCRIPT TYPES (see Table for additional details)

Original Article: Manuscript reporting original quantitative, qualitative or mixed methods research using a form of systematic study or inquiry to address a question relevant to the discipline and practice of genetic counseling. Systematic reviews included in this category.

Brief Report: Manuscript reporting an observation that adds to the knowledge of the discipline and practice of genetic counseling.

Case Study: Manuscript addressing issues relevant to discipline and practice of genetic counseling by demonstrating and stimulating thought about a difficult ethical, counseling, or genetic testing situation the author has encountered.

Professional Issue: Manuscript reporting reflections by the author(s) on the discipline and practice of genetic counseling.

Review: Manuscript summarizing the literature on a focused topic. Authors should contact the Editor-in-Chief prior to submission.

Commentary: Manuscript addressing matters of interest or controversy to the readership. Generally commissioned by the Editor-in-Chief.

Correspondence: Manuscript addressing work previously published in the JOURNAL

Practice Resource, Practice Guideline, Focused Revision: Manuscripts addressing specific areas of genetic counseling practice. Commissioned by the National Society of Genetic Counselors' Practice Guidelines Committee.

Book Review: Manuscript that reviews a book of relevance to the JOURNAL scope.

Conference Report: Manuscript with executive summary of important conference and/or select conference abstracts. Authors should contact Editor-in-Chief prior to submission.

Corrigenda and Errata: Manuscript describing corrections to a work previously published in the JOURNAL.

GENERAL REQUIREMENTS

Format

- double-spaced
- 1 inch margins
- 12 point font (Arial or Times New Roman).
- Footnotes should be avoided in the main text. When their use is absolutely necessary, footnotes should be numbered consecutively using Arabic numerals and should be typed at the bottom of the page to which they refer. Place a line above the footnote, so it is set off from the text. Use the appropriate superscript numeral for citation in the text.

English Language

Manuscripts must be submitted in grammatically correct American English. Manuscripts that do not meet this standard cannot be reviewed. Authors for whom English is a second language should consult an English-speaking colleague or consider having their manuscript professionally edited before submission to improve the English. A list of independent suppliers of editing services can be found at **https://wileyeditingservices.com/en/**. All services are paid for and arranged by the author, and use of one of these services does not guarantee acceptance or preference for publication.

Inclusive Language

An important element of the Journal of Genetic Counseling's commitment to diversity, equity, and inclusion is fostering authors' use of inclusive language in their manuscripts. To facilitate use of inclusive language we have compiled two excellent and comprehensive resources for authors' use:

CDC guidelines - **https://ehe.jhu.edu/DEI/Health_Equity_Style_Guide_CDC_Reducing_Stigma.pdf**

APA guidelines - **https://apastyle.apa.org/style-grammar-guidelines/bias-free-language**

Specific guidance:

1. Participant-reported demographic variables should be ascertained/used when reporting demographic information on participants.
2. Authors should explain the use of person-first or non-person-first language in the manuscript. For guidance, see https://apastyle.apa.org/style-grammar-guidelines/bias-free-language
3. For guidance on reducing stigma through use of terminology describing groups, see https://ehe.jhu.edu/DEI/Health_Equity_Style_Guide_CDC_Reducing_Stigma.pdf, and https://apastyle.apa.org/style-grammar-guidelines/bias-free-language
4. For guidance on use of terminology for sex and gender, see https://apastyle.apa.org/style-grammar-guidelines/bias-free-language

General Style Points

- **Abbreviations:** In general, terms should not be abbreviated unless they are used repeatedly and the abbreviation is helpful to the reader. Initially, use the word in full, followed by the abbreviation in parentheses. Thereafter use the abbreviation only.
- **Units of measurement:** Measurements should be given in SI or SI-derived units. Visit the Bureau International des Poids et Mesures (BIPM) website for more information about SI units.
- **Numbers and p-values:** numbers under 10 should be spelled out, except for: measurements with a unit (8 mmol/L); age (6 weeks old), or lists with other numbers (11 cousins, 9 aunts, 4uncles). Numerical figures (excluding p-values) should not exceed 2 decimal places. For p-values less than 0.001, report as $p < 0.001$. For p values greater than 0.001, use 2-3 decimal places. Do not use a leading zero for statistics that cannot exceed 1.0.
- **Genomic Terminology and Nomenclature:** Please use the following terms:
    - *genome sequencing* instead of *whole genome sequencing*
    - *exome sequencing* instead of *whole exome sequencing*
    - *pathogenic variant* instead of *mutation*
    - *secondary finding* instead of *incidental finding*

Use of italics

- italicize gene names (e.g. *FBN1*)
- do not italicize protein names (e.g. fibrillin)

Sequence variants

- Sequence variants should be described in the text and tables using both DNA and protein designations whenever appropriate.
- Sequence variant nomenclature must follow the current HGVS guidelines; see hgvs.org, where examples of acceptable nomenclature are provided.
- Human gene nomenclature should follow the standards of the HUGO Gene Nomenclature Committee (HGNC), see https://www.genenames.org/.

- **Pedigrees:** Pedigrees should follow the recommendations for standardized nomenclature accepted by the National Society of Genetic Counselors. Authors should consult the following references for these recommendations:
    - Bennett, R. L. , Steinhaus, K. A., Uhrich, S. B., O' Sullivan, C. K., Resta, R. G. , Lochner-Doyle, D., Markel, D. S., Vincent, V., & Hamanishi, J. (1995). Recommendations for Standardized Human Pedigree Nomenclature. Journal of Genetic Counseling, 4, 267-279.

    - Bennett, R. L., Steinhaus French, K., Resta, R. G., & Lochner Doyle, D. (2008). Standardized Human Pedigree Nomenclature: Update and Assessment of the Recommendations of the National Society of Genetic Counselors. Journal of Genetic Counseling, 17, 424-433.
- **Trade Names:** Chemical substances should be referred to by the generic name only. Trade names should not be used. Drugs should be referred to by their generic names. If proprietary drugs have been used in the study, refer to these by their generic name, mentioning the proprietary name and the name and location of the manufacturer in parentheses.

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4. PREPARING THE SUBMISSION

Specific Requirements for each manuscript type are summarized in this **Table.**

Parts of the Manuscript

The manuscript parts should be uploaded as separate files:

- cover letter
- main text file
- tables
- figures
- supplementary information files

Cover Letter

- include a statement that the work presented in the manuscript has not been published elsewhere and is not currently under review elsewhere. Include a statement that the work is available as a preprint or was presented at a conference, if relevant.
- If the study includes original data, at least one author must confirm in the cover letter that he or she had full access to all the data in the study and takes responsibility for the integrity of the data and the accuracy of the data analysis.

Main Text File

The **main text file** should be presented in the following order in one document (as appropriate for manuscript type):

1. Title Page
2. Abstract and keywords
3. 1-2 sentence responses to:

    1. What is known about this topic:
    2. What this paper adds to the topic:
4. Main body of paper
5. Author Contributions
6. Acknowledgements
7. Compliance with Ethical Standards
    1. Conflict of Interest
    2. Human Studies and Informed Consent
    3. Animal Studies
    4. Data Availability Statement
8. References
9. Tables
10. Figure legends
11. Figures

Title Page

The title page should include (in this order):

- title of the article. Authors may benefit from referring to Wiley's best practice tips on Writing for Search Engine Optimization.
- authors' names (no degrees) in the order to be published. Please denote cases of equal authorship with a superscript and footnote, e.g., for joint first or joint senior authorship, the footnote should say 'X and Y should be considered joint first author' or 'X and Y should be considered joint senior author', respectively.
- authors' institutional affiliation(s) where the work was conducted, and the author's present affiliation if different from where the work was conducted. The affiliation should comprise the department, institution (usually university or company), city, and state (or nation) and should be noted with numbered superscript to the author's name and footnote.
- Telephone number and e-mail address of the one author designated to review proofs (the corresponding author).
- Suggested running head. The suggested running head should be less than 80 characters (including spaces) and should comprise the article title or an abbreviated version thereof

Abstract

- unstructured, i.e., no main headings or subheadings
- maximum 300 words
- contains major keywords summarizing the work
- If reporting on a clinical trial, include the name of the trial register and the clinical trial registration number at the end of the Abstract.
- See Table for manuscript category-specific information

Keywords

- 3 to 6 keywords
- **Please include 2-3 keywords from** this list.

What is known about this topic

- 1-2 sentences

What this paper adds to the topic

- 1-2 sentences

Main Body of Paper

- **Please refer to this Table for specific manuscript type requirements**.
- For all research involving human participants, please include a statement in the **Methods** section confirming that the study was reviewed by an institutional review board/human investigations committee/ethics committee (include name of committee and IRB protocol number) and approved or waived as human subjects research.

Author Contributions

Prior to submitting the manuscript all authors fulfilling the International Committee of Medical Journal Editors (ICMJE) criteria for authorship should be identified and should agree on the order in which their names will be listed in the manuscript.

ICMJE criteria are:

- Substantial contributions to the conception or design of the work; or the acquisition, analysis, or interpretation of data for the work; AND
- Drafting the work or revising it critically for important intellectual content; AND
- Final approval of the version to be published; AND
- Agreement to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved.

If the study includes original data, at least one author must confirm that he or she had full access to all the data in the study and takes responsibility for the integrity of the data and the accuracy of the data analysis.

Please insert the following statements in the Author Contributions section, specifically identifying the relevant author(s):

Authors X and Y confirm that they had full access to all the data in the study and take responsibility for the integrity of the data and the accuracy of the data analysis. All of the authors gave final approval of this version to be published and agree to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved.

During the submission process you will be asked to select the contribution(s) made by each author from a pre-specified drop down menu.

Acknowledgements

Contributions from anyone who does not meet the ICMJE criteria for authorship should be listed in an Acknowledgements section. Financial and material support should also be mentioned in this section. Authors should list all funding sources and are responsible for the accuracy of their funder designation. If in doubt, please check the Open Funder Registry for the correct nomenclature: **www.crossref.org/services/funder-registry**.

If this paper is to be considered for the **Journal of Genetic Counseling Best Trainee Paper award**, please include a statement indicating that the research presented in the paper was conducted while the first author was in training or to fulfill a degree requirement of the first author. See the **Best Trainee Paper Award** tab on the JOURNAL website for more information about this award. Thanks to anonymous reviewers is not considered appropriate to include in Acknowledgements.

Compliance with Ethical Standards

- ***Conflict of Interest Statement***

The JOURNAL requires that all authors disclose any potential sources of conflict of interest. Any interest or relationship, financial or otherwise, which might be perceived as influencing an author's objectivity is considered a potential source of conflict of interest. These must be disclosed when directly relevant or directly related to the work that the authors describe in their manuscript. Potential sources of conflict of interest include, but are not limited to, patent or stock ownership, membership of a company board of directors, membership of an advisory board or committee for a company, and consultancy for or receipt of speaker's fees from a company. The existence of a conflict of interest does not necessarily preclude publication in the JOURNAL.

If the authors have no conflict of interest to declare, they must also state this in the manuscript. It is the responsibility of the corresponding author to review this policy with all authors and collectively to list in the manuscript under the subheading “Conflict of Interest” *all* pertinent commercial and other relationships.

The above policies are in accordance with the Uniform Requirements for Manuscripts Submitted to Biomedical Journals produced by the International Committee of Medical Journal Editors (**http://www.icmje.org/**).

The Conflict of Interest Statement should **mention each author separately by name.**

Recommended wording.

Author W declares that she has no conflict of interest.

Author X has received research grants from Drug Company A.

Author Y has received a speaker honorarium from Genetic Testing Company B and owns stock in Genetic Testing Company C.

Author Z is founder and CEO of Genetic Education Company D.

If multiple authors declare no conflict, this can be done in one sentence:

Author X, Author Y and Author Z declare that they have no conflict of interest.

Submitting authors should ensure they liaise with all co-authors to confirm agreement with the final statement.

- ***Human Studies and Informed Consent***

The JOURNAL requires that all appropriate steps were taken to obtain informed consent of all human subjects participating in the research reported in the manuscript submitted for review and possible publication, and statements to this effect must be included under this subheading. Participant anonymity should be preserved and all identifying information should be excluded in the manuscript unless the information is essential for scientific purposes and the study participants or patients (or parents or guardians) have provided written informed consent for publication. Images and information from individual participants will only be published where the authors have obtained the individual's free prior informed consent. Photographs need to be cropped sufficiently to prevent human subjects being recognized (an eye bar must not be used because of insufficient de-identification).

The editors reserve the right to reject manuscripts that do not comply with these requirements. The author will be held responsible for false statements or failure to fulfill these requirements.

For manuscripts reporting studies that involve human participants, a statement identifying the ethics committee that approved the study and confirmation that the study conforms to recognized standards is required, for example: **Declaration of Helsinki**; **US Federal Policy for the Protection of Human Subjects**; or **European Medicines Agency Guidelines for Good Clinical Practice**. It should also state clearly in the text that all persons gave their informed consent prior to their inclusion in the study.

Examples (not exhaustive)

Approval to conduct this human subjects research was obtained by the [name of institutional review board or ethics committee]. All procedures followed were in accordance with the ethical standards of the responsible committee on human experimentation (institutional and national) and with the Helsinki Declaration of 1975, as revised in 2000. Informed consent was obtained from all patients for being included in the study.

The study was approved by the [name of institutional review board or ethics committee]. No informed consent was required from subjects as data were anonymously extracted from the [name of system]. All procedures followed were in accordance with US Federal Policy for the Protection of Human Subjects.

This study was approved by and conducted according to the ethical standards of the [name of institutional review board or ethics committee]. All applicable international, national, and/or institutional guidelines were followed. This study was approved by the IRB after expedited review and was granted an informed consent waiver.

This study was conducted in accordance with all guidelines set forth by the [name of institutional review board or ethics committee]. Informed consent for genetic testing was obtained from all individuals undergoing testing, and [name of institutional review board or ethics committee] waived authorization for use of de-identified aggregate data. Individuals or institutions who opted out of this type of data use were excluded.

This study was reviewed and granted an exemption by the [name of institutional review board or ethics committee]. All procedures followed were in accordance with the ethical standards of the responsible committee on human experimentation (institutional and national) and with the Helsinki Declaration of 1975, as revised in 2000. Implied informed consent was obtained for individuals who voluntarily completed the online survey and submitted their responses.

Approval to conduct this human subjects research was obtained by the [name of institutional review board or ethics committee]. All procedures followed were in accordance with the ethical standards of the responsible committee on human experimentation (institutional and national) and with the Helsinki Declaration of 1975, as revised in 2000. Informed consent was obtained from all patients for being included in the study.

The study was approved by the [name of institutional review board or ethics committee]. No informed consent was required from subjects as data were anonymously extracted from the [name of system]. All procedures followed were in accordance with US Federal Policy for the Protection of Human Subjects.

This study was approved by and conducted according to the ethical standards of the [name of institutional review board or ethics committee]. All applicable international, national, and/or institutional guidelines were followed. This study was approved by the IRB after expedited review and was granted an informed consent waiver.

This study was conducted in accordance with all guidelines set forth by the [name of institutional review board or ethics committee]. Informed consent for genetic testing was obtained from all individuals undergoing testing, and [name of institutional review board or ethics committee] waived authorization for use of de-identified aggregate data. Individuals or institutions who opted out of this type of data use were excluded.

This study was reviewed and granted an exemption by the [name of institutional review board or ethics committee]. All procedures followed were in accordance with the ethical standards of the responsible committee on human experimentation (institutional and national) and with the Helsinki Declaration of 1975, as revised in 2000. Implied informed consent was obtained for individuals who voluntarily completed the online survey and submitted their responses.

If any identifying information about participants is included in the article, the following sentence should also be included:

Informed consent was obtained from all participants for which identifying information is included in this article.

Authors do not need to provide a copy of the consent form to the publisher; however, in signing the author license to publish, authors are required to confirm that consent has been obtained. Wiley has a **standard patient consent form available** for use.

- ***Animal Studies***

The JOURNAL does not publish non-human animal studies. To affirm that this is the case for your submission, please include the following sentence under this subheading in the manuscript:

No non-human animal studies were carried out by the authors for this article

- ***Data Availability Statement***

Authors are required to provide a data availability statement to describe the availability or the absence of shared data. When data have been shared/are available in a repository, authors are required to include in their data availability statement a link to the repository they have used/created, and to cite the data they have shared. Whenever possible the scripts and other artefacts used to generate the analyses presented in the paper should also be publicly archived. If sharing data compromises ethical standards or legal requirements then authors are not expected to share it.

Sample statements are **available here from Wiley Author Services**.

Example with data repository

Data Availability Statement

Data collected for this study are available through the publicly available repository MINDS@Uw. The data file is available at ***http://digital.library.wisc.edu/1793/78637***

Citation in Reference List

Donahue, A., Hall, A., & Petty, E. (2018). Genetic counselor information needs & current library services for genetic counselor. [Data file] Retrieved from ***http://digital.library.wisc.edu/1793/78637***

Although it would be rare for a paper submitted to the JOURNAL to report novel nucleotide sequence data, should that be the case, the novel nucleotide sequence data including genetic mutations must be submitted to a public database prior to publication and a sentence naming the database should be included in the manuscript.

References

The accuracy of references is the responsibility of the authors. The JOURNAL **strongly prefers references that have undergone peer review** and are not conference abstracts, unpublished masters' theses, unpublished dissertations, unpublished data in manuscripts, etc. However, if essential to the manuscript, they should be cited and referenced appropriately.

References should be prepared according to the *Publication Manual of the American Psychological Association* (6th edition). The APA website includes a range of **resources for authors** learning to write in APA style, including **an overview** of the manual, **free tutorials** on APA Style basics, and an **APA Style Blog**. For more information about APA referencing style, please also refer to the **APA FAQ**.

EndNote users can download the style **here**.

In-text citation

- follow the author-date method whereby the author's last name and the year of publication for the source should appear in the text, for example, (Jones, 1998)
    - publications with no authors, use brief phrase to describe publication and year of the publication, for example, ("Report of 1979 Business Meeting", 1979, p. 1) or ("NSGC Professional Status Survey: Executive Summary", 2020)
    - publications with no dates, use n.d., for example, ("American board of Genetic Counseling, Mission and History", n.d.)
- Multiple citations should be listed alphabetically by author's last name

Personal communications

- cite within the text as (Name of person providing the communication, personal communication, Date of communication), for example (Jane Doe, ABCD Executive Director, personal communication, September 2019)
- do not include in the reference list
- permission in writing from the communicator is required. Submit written permission with manuscript.

Reference List

- alphabetical by last name of first author
- the reference list is not numbered

General Comments

- Digital Object Identifier (DOI) should be provided for all references where available
- For journal articles, issue numbers are not included unless each issue in the volume begins with page one.
- For references with more than seven author names list first six with three dots and then last author name.

Reference examples

- *Journal article with 7 or fewer authors*
  - Beers, S. R., & De Bellis, M. D. (2002). Neuropsychological function in children with maltreatment-related posttraumatic stress disorder. *The American Journal of Psychiatry, 159*, 483–486. *doi:*1176/appi.ajp.159.3.483

- *Journal article with more than 7 authors*
  - Reuter, C. M., Kohler, J. N., Bonner, D., Zastrow, D., Fernandez, L., Dries, A., ... Wheeler, M. T. (2019). Yield of whole exome sequencing in undiagnosed patients facing insurance coverage barriers to genetic testing. *Journal of Genetic Counseling, 28*, 1107-1118. doi: 10.1002/jgc4.1161

- *Book with 7 or fewer authors*
  - Bradley-Johnson, S. (1994). *Psychoeducational assessment of students who are visually impaired or blind: Infancy through high school* (2nd ed.). Austin, TX: Pro-ed.

- *Book with 8 or more authors*
  - Gilbert, J. R., Smith, J. D., Johnson, R. S., Anderson, A., Plath, S., Martin, G., . . . White, N. (2014*). Choosing a title* (2nd ed.). New York, NY: Unnamed Publishing.

- *Internet Document*
  - Norton, R. (2006, November 4). How to train a cat to operate a light switch [Video file]. Retrieved 4/20/2020 from http://www.youtube.com/watch?v=Vja83KLQXZs
  - Note: only include the retrieval date if the content is likely to change

- *Publicly available data repository*
  - Donahue, A., Hall, A., & Petty, E. (2018). Genetic counselor information needs & current library services for genetic counselor. [Data file] Retrieved from http://digital.library.wisc.edu/1793/78637

- *Newsletter/newspaper, no author*
  - Report of 1979 Business Meeting of the NSGC. (1979). *Perspectives in Genetic Counseling*, 1(4), 1. Retrieved from nsgc.org [members only access]

- *Newsletter/newspaper, author*
  - Smith, A.C.M. (1982). "The sermon on the amount:" The status of NSGC finances. *Perspectives in Genetic Counseling*, 4(1), 2. Retrieved from nsgc.org [members only access]

- NSGC Professional Status Survey
  - NSGC Professional Status Survey: Executive Summary (2020). p. 2. Retrieved from https://www.nsgc.org/page/whoaregeneticcounselors

Figure Legends

Every figure must have a legend that includes the figure number and figure title. Legends should be concise but comprehensive – the figure and its legend must be understandable without reference to the text. The figure legend should Include definitions of any symbols used and define/explain all abbreviations and units of measurement.

Additional Files

Tables

Tables should be self-contained and complement, not duplicate, information contained in the text. The table must be understandable without reference to the text.

Tables should:

- be numbered and referred to by number in the text.
- have a brief explanatory title,
- have a concise but comprehensive legend in the case where additional explanation is provided using footnotes. Footnotes should be indicated by superscript lowercase letters
- define all abbreviations in the legend.
- be supplied as editable files, not pasted as images
- be uploaded as separate file(s)

Figures

Authors are encouraged to send the highest quality figures possible. Line art should be exported at 600 dpi or higher, and halftone images should be exported at 300 dpi or higher.

Figures should:

- be numbered and referred to by number in the text
- be clearly labeled
- be uploaded as separate file(s)

Color figures. Color figures may be published online free of charge.

Supporting Information

Supporting information is information that is not essential to the article, but provides greater depth and background. It is hosted online and appears without editing or typesetting. It may include copies of surveys or interview questions, consent forms, tables, figures, videos, datasets, etc.

Click here for Wiley's FAQs on Supporting Information.

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5. EDITORIAL POLICIES AND ETHICAL CONSIDERATIONS

Peer Review and Acceptance

The acceptance criteria for all papers are the quality and originality of the research and its significance to JOURNAL readership and the practice and discipline of genetic counseling. Papers will only be sent to review if the Editors determine that the paper meets the appropriate quality and relevance requirements.

Except where otherwise stated, manuscripts are single-blind peer reviewed. Wiley's policy on the confidentiality of the review process is **available here.**

Pre-Print Policy

The JOURNAL will consider for review articles previously available as preprints. Authors may also post the **submitted version** of a manuscript to a preprint server at any time. Authors are requested to update any pre-publication versions with a link to the final published article.

Revisions

When submitting a revised manuscript please also submit:

- Response to reviewer comments in the form of a table with reviewers' suggestions on the left-side and edits made/how addressed on the right-side
- A marked version (tracked, highlighted, etc.) and unmarked version of revised manuscript

Changes in Authorship

Prior to submitting the manuscript all authors should agree on the order in which their names will be listed in the manuscript. Any changes to authorship, including adding, removing or rearranging the authorship list, must be made before the manuscript has been accepted and only with approval from the Editor. The corresponding author is expected to write to the Editor with the reason for the change and will complete an Authorship Change Request form with written agreement from ALL authors, including those affected by any change. Only in exceptional circumstances will the Editor consider changes in authorship after a paper has been accepted. If a request for change in authorship is submitted after acceptance but before publication, the article will be suspended from proceeding to publication until the authorship change request has been resolved. Once an article has been published in an online issue, a completed Authorship Change Request form and a corrigendum will need to apply to reflect any allowed changes in authorship.

Decision Appeals

Appeals should be filed within 28 days of notification of the decision. The appeal should be in the form of a letter addressed and submitted to the Journal of Genetic Counseling Editorial Office at JOGC@wiley.com. The letter should include clear and concise grounds for the appeal, including specific points of concern. The appeal will then be assessed by the Journal of Genetic Counseling management team, led by the Editor-in-Chief, and informed by the subsequent editorial communications. You will be informed of the outcome of the appeal in writing, normally within 28 days. The decision will be final.

Clinical Trial Registration

The JOURNAL requires that clinical trials are prospectively registered in a publicly accessible database and clinical trial registration numbers are included in all papers that report their results. Authors are asked to include the name of the trial register and the clinical trial registration number at the end of the Abstract. If the trial is not registered, or was registered retrospectively, the reasons for this should be explained.

Research Reporting Guidelines

Accurate and complete reporting enables readers to fully appraise research, replicate it, and use it. Authors are encouraged to adhere to recognized research reporting standards. The EQUATOR Network collects more than 370 reporting guidelines for many study types, including for:

- Randomized trials: CONSORT
- Observational studies: STROBE
- Systematic reviews: PRISMA
- Qualitative research: COREQ
- Quality improvement studies: SQUIRE
- Study protocols: SPIRIT
- **Studies reporting on genetic counseling as an intervention should follow the reporting standards found here:** Standards for the Reporting of Genetic Counseling Interventions in Research and Other Studies (GCIRS)

Publication Ethics

The JOURNAL is a member of the **Committee on Publication Ethics (COPE)**. Read Wiley's Top 10 Publishing Ethics Tips for Authors **here**. Wiley's Publication Ethics Guidelines can be found **here**.

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6. AUTHOR LICENSING

If a paper is accepted for publication, the author identified as the formal corresponding author will receive an email prompting them to log in to **Author Services**, where via the Wiley Author Licensing Service (WALS) they will be required to complete a copyright license agreement on behalf of all authors of the paper. This email will be issued within a few days of paper acceptance.

- For authors signing the copyright transfer agreement

If the Hybrid Open Access option [requires payment] is not selected the corresponding author will be presented with the copyright transfer agreement (CTA) to sign. The terms and conditions of the CTA can be previewed in the samples associated with the **Copyright FAQs**.

- For authors choosing Hybrid Open Access

If the Hybrid Open Access option is selected the corresponding author will have a choice of the following Creative Commons License Open Access Agreements (OAA):

Creative Commons Attribution License (CC-BY) OAA

Creative Commons Attribution Non-Commercial License (CC-BY-NC) OAA

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2 Section Two: Research Paper

The experience of maintaining psychological wellbeing when living at-risk of Huntington's Disease: An Interpretative Phenomenological Analysis

Word count - 8000

(Excluding title page, references, figures, tables and appendix)

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Abstract

Living at-risk of a genetically inherited disease can be a challenging experience causing psychological distress as well as physical health problems. Huntington's disease (HD) is a genetic, neurodegenerative disease. It causes motor dysfunction, cognitive decline and during the progression of the disease psychological conditions are common. Twelve participants living at-risk of HD were interviewed and interpretative phenomenological analysis methodology was used to understand their experiences of maintaining psychological wellbeing. This resulted in three themes: (1) 'you're constantly in limbo': living in two worlds, (2) "I have to live, just bloody live": managing the possibility of a time-limited lifespan and (3) "Is that who I am, is that what I am?": the exhausting quest to be seen as an individual first. The findings indicated a need for improved knowledge within professional settings, provision of accessible support and implementation of systemic interventions. Future research could contribute to the formation of such knowledge and provision of HD aligned services to help support the psychological wellbeing of people living at-risk of Huntington's disease.

Keywords: psychological wellbeing, lived experience, Huntington's disease, interpretative phenomenological analysis, qualitative, meta-ethnography, systematic review.

What is known about this topic: There is currently limited knowledge on the psychological aspects of Huntington's disease outside of the test taking process. Little is known about the experience of managing psychological wellbeing while living at-risk of Huntington's disease.

What this paper adds to the topic: This paper explores the experience of managing psychological wellbeing while living at-risk of HD. It presents insights into the complex skills and strategies used by participants to manage psychological wellbeing such as living in two worlds, living life as well as possible and the exhaustion created by living at-risk of HD.

Introduction

Genetic diseases often have profound, progressive and irreversible physical effects on individuals. Where such a condition is not present at birth, but develops later in the person's lifetime, evidence also suggests that even before any physical changes, individuals can experience considerable psychological challenges. Nonetheless, genetic testing and diagnosis can help form treatment and management plans to minimise distress caused by, for example, the potential physical implications of such conditions (Walter & Emery, 2012) and threats to identity (Klitzman, 2009). However, less is known about the well-being of those 'at risk' individuals, i.e., those who choose not to partake in seeking testing or diagnosis despite a high risk of being affected. As they are not in contact with services, they have been largely ignored in terms of the effects of their 'at risk' status on their well-being. An example of one such condition where many at risk individuals chose not to pursue genetic testing (Baig et al., 2016) is Huntington's disease (HD).

HD is a genetically inherited neurodegenerative disease caused by CAG triple repeat expansion mutation in the HD gene (MacDonald et al., 1993). A recent systematic review of global incidence and prevalence estimated 4 per 100,000 people worldwide are affected by HD (Medina et al., 2022). HD symptoms progress over time, for approximately 10-20 years (Ready et al., 2011; Roos et al., 1991), causing progressive motor dysfunction, cognitive decline (leading to dementia) and distress (Heiberg, 2008). The disease is extremely debilitating in the later stages, leaving individuals unable to speak, wheelchair-bound and experiencing severe involuntary movements (Klein et al., 2014). At present there is no cure for HD though treatments for some effects of the condition, such as involuntary jerking (chorea), are available (Dash & Mestre, 2020).

As HD is genetic, children of a parent with the expanded gene have a 50% chance of inheriting the condition (Rivera-Navarro et al., 2015). Offspring who are informed of their 50%

risk can experience negative emotions (Folstein et al., 1983) similar to the grief process experienced following a traumatic event (Evers-Kiebooms & Decruyenaere, 1998). If the genetic expansion is inherited the person will develop HD, usually between 30 and 50 years of age (Di Maio et al., 1993; Roos et al., 1991) when the person moves from a premanifest (movement symptom free) state to experiencing symptoms of the disease.

Individuals 'at-risk' (i.e., those with a biological parent or grandparent with HD) who are aged 18 and above can, in most countries, request a genetic test to see if they have the HD gene expansion before symptoms occur. Research indicates that uptake in genetic testing for HD is low, with over 80% of those at-risk choosing not to take the test (Baig et al., 2016).

Hawkins et al. (2013) argue that individuals who decide to take the test are often met with barriers to testing that are not only practical, e.g., regarding finances, but also psychological e.g., increased distress. Research into the psychological needs of those who have not pursued testing is limited.

HD services are largely inaccessible to individuals who are not engaged in the route to testing and do not have either the symptoms or a diagnosis of HD (Etchegary, 2011) meaning that those living at-risk of HD are unable to access specialist support though the NHS. When non-specialist services, such as the primary care service and local counselling services, are accessed by those living at-risk, health care providers' knowledge of HD is reported as being too poor to enable appropriate support (Skirton et al., 2010).

Further research focusing on those living at-risk, the Prospective Huntington At-risk Observational Study (PHAROS) in the US (Quaid et al., 2017), reported that people living at-risk carried a significant burden around disclosure and concealment of their at-risk knowledge. While further analysis from this study highlights some of the experience of day-to-day experiences of living at-risk (Quaid et al., 2008), it does not offer understanding of the

psychological or emotional experience outside of the study's themes focused on risk (disclosure or concealment) and living in hope of being negative. Further quantitative research (Chisholm et al., 2013) has found that HD has a complex impact on well-being. Alongside other findings, the study suggests no significant difference on psychological measures concerning well-being between those at-risk and those who had pursued genetic testing. The study suggests that greater focus is required to understand the impact of HD on wellbeing.

Several health-related models found in the chronic illness literature have been used to understand aspects of living with HD and aid the recommendation of appropriate interventions and support. One such model is the stress and coping model (Lazarus & Folkman, 1984). This model suggests that how well a person copes with a health threat is dependent on how confident they feel about managing the threat, i.e., the individual's belief in their ability to cope with a stressful situation begins at the point of cognitive appraisal. In the context of chronic illness, appraisals of stressful situations are in line with the person's beliefs about their condition and can inform adaptive coping strategies (e.g., the belief that pain can either be controlled or is not chronic may inform more positive strategies than the belief that pain cannot be controlled) and have a positive impact on psychological wellbeing. Cognitive appraisal, however, can also involve an array of emotional responses (anxiety, anger, low mood) which have been found to contribute to the formation of maladaptive coping strategies and therefore likely to have a negative impact on psychological wellbeing. Research examining the role of coping in wellbeing for those experiencing chronic illness indicates that generally coping strategies that are problem-focused and acceptance and adjustment directed influence a better level of functioning. In contrast, individuals with more avoidant strategies experience worse physical health and psychological adjustment and increased psychological distress (Helder et al, 2002; Kershaw et al., 2004).

Developments from the stress and coping model established the link between stress and coping and emotional regulation. It was found that individuals who were able to regulate their emotions successfully formed more effective coping strategies. This led to the development of Leventhal's self-regulation model (SRM; Leventhal et al., 2001). The SRM offers a theoretical framework applied in chronic illness research that suggests that how we conceptualise an illness influences wellbeing (Hagger & Orbell, 2003; Kaptein et al, 2003). In particular, perceptions of control and chronicity (how long the illness may last) have been linked to negative impacts on wellbeing across conditions (Hagger & Orbell, 2003; Kaptein et al., 2003).

When the SRM has been applied to research in HD, results (e.g., Arran et al., 2013; Helder et al., 2002) have emphasised the biopsychosocial nature of the predictors which influence psychological wellbeing. The issue remains, however, that, despite such models and adaptations, most psychologically focused research remains within the context of genetic testing (Quaid, 2017; Sarason et al., 1978; Taylor, 2004) or is quantitative in nature. While useful, this does not offer an understanding of the experience of managing wellbeing while living at-risk of HD.

Consequently, the current study aims to explore the experience of maintaining psychological wellbeing for those living at-risk of HD. Understanding such experiences will offer insight needed to develop psychological care provision for supporting those living at-risk, which is currently minimal (Zarotti, et al., 2020). The research question to guide this exploration is: What is the experience of maintaining psychological wellbeing for those living at-risk of HD?

Method

Methodology

Interpretative phenomenological analysis (IPA) was used as a qualitative method with an idiographic focus that allows the consideration of individual lived experiences in informing meaning (Smith et al., 2021). IPA explores human experience (phenomenology) and how such experience is understood (hermeneutics) and was originally designed to understand the experiences of people with chronic health conditions (Smith et al., 1999). The theoretical underpinnings of IPA (phenomenology, hermeneutics, idiography) recognise that experience needs to be examined closely in research, i.e., initially at the individual level, as opposed to a more broad or distant approach. The flexibility offered through the use of IPA in its focus on lived experience and interpretation, presents an in-depth method of meaning making concerning the experience of maintaining psychological wellbeing when living at risk of HD.

Data were approached from the perspective that parts of reality can exist independent of a person's awareness and knowledge, which is consistent with the critical realist ontological view. Also consistent with critical realism is the relativist epistemological position, which acknowledges that context enables the development of knowledge and that there is no objective or universal truth, with knowledge dependent on cultural and historical context. The impact of this approach on research is that conclusions are fallible as the true context and knowledge of another cannot be fully understood by the researcher.

Participants

Participants were recruited via two pathways within which a digital poster was shared with contact details for those interested in taking part. The first was via an on-line HD social media support group, the second via the social media account of a HD charity, both UK based.

The inclusion criteria ensured that the purposive and homogeneous sample required for IPA was achieved (Smith et al., 1999). Smith et al. (2021), suggest that an ideal number of participants for IPA is between four and ten individuals as smaller samples allow for deeper understanding to be achieved. To allow for potential withdrawal and to ensure the gathering of sufficient data, 18 participants were recruited from 112 initial expressions of interest in order of response time. Six participants did not attend interviews or respond to follow up contact. This resulted in 12 completed interviews (four via the HD social media support group and eight via the HD charity).

To be included in the research, participants were required to be over 18, aware of their at-risk status and able to communicate where their risk originated allowing level of risk (25% via grandparent, 50% via parent) to be established. Guidance suggests individuals need around six months for adjustment to this knowledge, and a further six-months for the experience of living with this knowledge (Tibben et al., 1997). Therefore, participants needed to have knowledge of their at-risk status for a minimum of 12 months. In line with the inclusion criteria, participants were symptom free, not tested for HD, spoke English without any communication difficulties and were able to participate in an interview (or two if needed) of up to 90 minutes in length. Participation was voluntary and participants could withdraw at any point up to two weeks after the completion of their interview. At this point data analysis had begun, meaning extracting individual data would be difficult. Demographics of the participants are presented in Table 2 - A. All names are pseudonyms to preserve confidentiality.

[Table 2 – A about here]

Procedures

The first author's academic institution granted ethical approval (FHMREC20188). Participants were sent an explanation of the study, a consent form explaining audio consent and what would happen with recordings, transcribed interviews and results. Following informed consent, participants were interviewed using Microsoft Teams due to COVID-19 restrictions and to ensure a varied sample from across the UK. Interviews were transcribed verbatim by the primary author.

Data Collection

Data were collected using semi-structured individual interviews. The semi-structured interview schedule was developed using existing literature for guidance on creating topic guides (Busetto et al., 2020; Murray & Wilde, 2020; Pietkiewicz & Smith, 2014) and topic guides shared within existing qualitative HD research (Quinn et al., 2010; Stopford et al., 2020). Input was also sought from supervisors and a clinical psychologist working within the HD field. The topic guide was finalised with experts by experience. This helped establish areas for focus (such as experiences of distress, the causes and how these were managed, strategies people use to stay psychologically well, how people made sense of HD and risk in their own lives), exploration and the phrasing of questions in a curious and explorative way suited to a phenomenological approach (Smith et al., 2021).

Data Analysis

Participants' narratives were transcribed verbatim with identifying details (e.g., places, names) removed. The most recent IPA guidelines were followed for analysis, creation of themes and use of terminology (Smith et al., 2021). The analysis involved the idiographic approach of reading each transcript individually and following the seven steps described by Smith et al. (2021). The first of these involved reading and re-reading the transcripts to enter

the participants' world during this process the author was required to keep an open mind (Smith et al., 2021). The second involved making of exploratory notes; during reading the transcript anything of interest or possible significance is noted in the text (an example of this process can be seen in image 2 – A). The third involved the formation of Experiential Statements. These statements are directly related to the participants' experience. The fourth involved the formation of Personal Experiential Themes (PETS) created from commonalities across the experiential statements (an example of this process can be seen in table 2 - B). The fifth involved naming the PETs and placing them in a table. The sixth involved the repeating of steps one to five for each participant. The seventh and final step involved analysing PETS across participants and grouping similarities to form Group Experiential Themes (GETS). An example of this process can be seen in table 2 - C. At this stage researcher interpretation is integral to the formation of themes.

[Image 2-A about here]

[Table 2-B about here]

[Table 2-C about here]

The primary author generated the GETs and the interpretation of such themes. As this is an intersubjective dynamic, reflexivity (Finlay & Gough, 2008) was addressed through the keeping of a reflexive journal so the author could remain aware of the lens through which data were being explored. The primary author had no lived experience that aligned with participants in this research; therefore, it is important to be aware that social and cultural beliefs may have affected her perceptions of people living at-risk of HD. Furthermore, to ensure the findings were rooted in the participant data and improve the trustworthiness of interpretations, data analysis was supported by three co-authors through discussion of understanding participants' experiences throughout the IPA process. This ensured that the four benchmarks of quality in

qualitative research, which are sensitivity to context, transparency, coherence and importance, (Yardley, 2000) were also considered.

Findings

Through analysing the transcripts, three themes were generated: (1) "you're constantly in limbo": living in two worlds, (2) "I have to live, just bloody live": managing the possibility of a time-limited lifespan and (3) "Is that who I am, is that what I am?": the exhausting quest to be seen as an individual first. Where quotations have been shortened below, this is indicated with a bracketed ellipsis (...).

"You're constantly in limbo": living in two worlds.

This theme explores how participants managed psychological wellbeing by moving between HD and non-HD-dominated worlds. Living in the HD world involved an awareness of being at-risk and of the experiences around HD that were constantly present and causing distress. Living in the non-HD world explores the reprieve participants experienced when they moved out of the HD world.

Some participants ensured they had a group of friends or an environment in which no one knew about HD. These participants had genuine, invested relationships with separate groups of friends (those who knew about HD and those who did not) but would choose who to socialise with depending upon how they were feeling about their risk. Being able to spend time with those with no knowledge of HD offered participants an identity independent of HD and a period of respite from having an awareness of the disease. For some participants this social separation seemed to enable a recharging of resources. For example, for Ian, it was important to be able to keep HD out of his working life but have the flexibility to speak to people he chose to about HD:

"…it's still a secret, but it's a controlled secret now, it's not, it's not taken over my life..." (Ian).

Similarly, other participants described how they were able to wear a 'mask' that eliminated HD when they wanted to be in the non-HD world. The 'mask' was used in work, within social circles or for their children and family. This gave a feeling of accomplishment and control, i.e., that they could silence HD when they needed to and be fully present as themselves, making a choice to remove the 'mask' when they were ready to enter the HD world.

As well as choosing when to access the non-HD world, participants could also choose to dwell in the HD world. Some participants' experiences suggested that choosing to spend time in the HD world had a beneficial outcome amid their distress. Spending time with people who knew about HD provided a shared experience that offered a sense of belonging, acceptance and understanding that brought connection and a release*:*

"…it just feels good inside knowing that other people (know about HD), so that's why at the same time seeing more people and talking to more people helps as well as like, as a coping mechanism as opposed to just keeping it locked up inside, cause that ain't good for no-one, that" (Ian).

Being able to discuss and explore HD openly with people who understood provided an outlet for stress and worry and relief for participants as they did not have to embark on the tiresome repetition of explaining their relationship with HD.

However, as well as being able to choose to live in one world or the other, participants could be forced from one to the other. This resulted in higher levels of distress and participants fought to regain control. For example, to control distress triggered by worries around onset of symptoms, which would force them into the HD world, participants described tests they would do. The aim of these tests was to evidence that the onset of HD had not occurred. This

reassurance enabled participants to reduce distress and move between HD worlds again. For some, this testing process would be conducted multiple times throughout the day. Some participants described tests as cognitive tasks such as puzzles, reciting names, places and details of songs whereas other participants might use a physical test:

"I have a habit of when I'm in bed, of rubbing my feet together and I, like, try and hold, stop doing it to see how long I can hold it, so (….) I can really discover whether they're involuntary or not" (Lisa).

Participants fought to regain control by reminding themselves that they had arranged an HD plan for the future (for example, where they would be cared for, who would provide that care, whether they would pursue euthanasia, or make end of life pacts with siblings). Having a plan helped people reassure themselves that if they did develop HD, they had a plan to manage their situation. Interestingly the idea of euthanasia and end of life pacts brought reassurance to participants who spoke about their end-of-life preferences. This seemed positive; however, such plans were only considered in an effort to avoid the "horrific" suffering that would come if they were to develop HD.

Participants also used perspective to control the pull into the hopelessness of the HD world by comparing their lives to the lives of others. This brought short-lived relief that they were *'lucky (…) it's (HD) nowhere near as bad'* (Marie). This could be in comparison to tragedy reported on the news or, more specifically, other people suffering with HD:

"…reading other people's (HD) stories and I think it makes me, in a really horrible way, makes me feel better about my own situation because, you see younger people like suffering, oh my gosh I'm stressing about something I might get when I'm 60" (Lucy).

This was temporary and the realisation that this did not change their situation pushed them back into the HD world and the overwhelming awareness of the *'disgusting disease' (Lisa).*

"....trying to think it could be so much worse but it's, it's quite difficult to maintain that, I think you think it for a few minutes and 'yeah, you're lucky it could be worse', and then it sort of hits you like a sack of shit afterwards that you're still at-risk" (Lisa).

Ultimately it seemed that being able to step into the non-HD world brought temporary respite and a period of recuperation before the awareness of the at-risk status and their HD related life experiences hit them again. For all participants, time was required to be spent in both worlds to maintain a sense of wellbeing and balance.

"I have to live, just bloody live": managing the possibility of a time-limited lifespan.

This theme describes the experience of living in a HD-free period and awareness that being HD-free may end. It explores the pressures of a possible time-limited life span, the need to be worthy of an HD-free life, and the battle to gain control over onset of symptoms. While this pressure had many negatives, there appeared to be positive factors involved with the idea of living within a pre-determined lifespan.

Participants spoke of the pressure to live their lives within an allocated window of HD-free time that was largely based on their family narrative concerning age of onset. For some this was the idea of a non-HD life until their 70s due to previous family members' age of onset and, for others, this was non-HD life until their 30s*:*

"…all the time, every day it is a constant, 'oh well, I'm losing time' I know, I know everyone's got an expiry, but with this it feels like it's just that's it, personally I thought for the past seven years that as soon as I hit thirty I'm gone." (Ian).

Regardless of their predicted potential age of onset, participants strived to live a *'full life' (Cath)* before HD, with intense sense of pressure to fit in as much as they could. Participants seemed to use this pressure to give them energy and motivation to live life to the full. For example, creating a list of goals or accomplishments to be achieved prior to symptom onset (for example, buying a house, having children, learning to drive) were presented as a *'motivating force to keep going' (Cath)*. However, it seemed that the experience of doing this in such a time pressured way removed some of the joy of the experience and the celebration of the achievement. There was no time to celebrate and reflect on the achievement as the next goal was waiting.

For most, despite their efforts to live a full life, the threat of the future arrival of HD stole long-term goals and dreams. For example, one participant shared how their dream to relocate to a rural location had been abandoned due to the possible need to access health care. Others described how they felt that life was constantly making them aware of the possible impact of HD on their future when they witnessed their in-laws enjoying their retirement, grand-children or travelling. This led to an intense sadness, grief, envy, and anger that they may *'not get the chance'* (Sarah).

Some participants explained that they had a fear that HD would cause their ability to empathise and be kind to people to deteriorate. To control the level of deterioration experienced, participants spoke of starting at a higher point:

"*...and I think I made a decision that I would be really nice to everybody, all the time, because then, if it got me as I'd have, if my starting place was at really one end of the spectrum when I was really such a lovely, nice person, then perhaps I'd die before I became evil, which is magical thinking isn't it?"* (Jane).

The idea of being able to start at a higher point or having a scale of effort was continually referred to when participants spoke of ways they tried to limit the impact of onset should they have the genetic expansion. Knowing that such effort had been made seemed to bring a sense of control over onset but also a prediction of peace if onset did begin. For example, some participants aimed to care for the body and mind as well as they could through diet, exercise and tasks designed to keep the brain active:

"...the longer I could keep my brain active, and you know, keep going, I think the healthier connections will stay and the longer my neurology can, yeah, I can keep exercising my brain for as long as possible, hopefully it will protect itself a little bit. I mean that's wishful thinking, there's no evidence that will happen, but I think for me that what I want and, so, yeah, I can't let myself shut my brain down and like stop thinking. I've got to try and keep, keep going." (Sarah).

Participants knew there was no evidence to suggest such techniques had an impact on onset, though this did not remove the importance of active efforts to feel like onset was in their control. By doing this hard work, participants were able to reduce their anxiety with the self-reassurance such behaviours brought. If they developed symptoms, they would be more peaceful in their suffering knowing they had done everything in their power to prevent onset and lived a good life for as long as they could.

Ultimately, it seemed even when participants aimed to live life to the full, HD stole from their experiences with added pressure, a sense of having to rush and the acknowledgement that they could still lose life experiences related to old age. No matter how fast they lived, this area of life remained out of reach.

"Is that who I am, is that what I am?": the exhausting quest to be seen as an individual first.

Within this theme the participants' experience of being perpetually exhausted by living at-risk is explored. Exhaustion came from many areas, such as the worries about HD taking their physical body, the challenge of finding their own identity independent of HD, family related stressors and inconsistent access to information and poor support from professionals.

Participants spoke about the difficulties they experienced in being able to keep HD in the *"back of their head"* (Sarah) so they could be themselves and how unexpected observations, such as seeing a particular gait of an individual, would move HD to the front of their mind.

"It's hard. I do think about it most days, not dark, not in a dark way, I just it's there and you know, I mean it's there and I know it's there and sometimes when I'm in the street and I see somebody they might be struggling and I could tell it's neurological, I think maybe they've got Huntington's disease and I often think 'I hope you got some support. I hope you're not on your own" (James).

It seemed that for participants, trying to find their own identity outside of HD was an exhausting task overshadowed by fear of losing oneself. Losing the mind and the parts of themselves that made them who they were and not being able to do the activities they enjoyed, as well as being met with societal judgement, were the greatest fears expressed about

developing HD. The biological manifestation of the disease and its progression was difficult though manageable, but the loss of the self was described as *'terrifying'* (Lisa). This fear seemed to be driven by the idea of losing their identity, the things they loved (for example being able to read, revisit memories, laugh) and experiences with unhelpful cultural narratives. Narratives that called HD the *'devil's disease'* (Lisa) and those who experienced it *'devil's spawn'* (Jane) or *'drunk'* (Daniel) were intensely distressing and affected self-perception:

"How can I be loveable when it's called the devil's disease?" (Jane).

Participants who experienced public shaming of their relatives were deeply hurt. Such experiences resulted in a tiring psychological process of preparing for social outings followed by a needed recovery period to process the stigma encountered. It was apparent such distressing social interactions took away the small victories participants tried to implement for their symptomatic family members, leading to a sense of hopelessness:

"…that's the kind of situations you face sometimes, when you're trying to do the best for your family who are suffering, and you're trying to make them have as much joy as what you have, and then I guess they just get put in a corner sometimes" (James).

Another source of exhaustion could be seen when participants described thinking about their family history and relationships. It seemed important to participants to distinguish between the person and the disease and recall the positive traits and memories of the person prior to HD and retain them throughout their HD deterioration. This often brought a circular questioning which resulted in a *'Pandora's box'* (Gill) of questions concerning whether they were parented by a person or a disease:

"...I've never really known him without symptoms, but in my head, I try and sort of picture what he was like, and kind of look at the photos and like tell myself that it was HD that made him do those things, and not, it wasn't my Dad kind of thing..." (Karen).

Feelings of helplessness experienced by participants seemed to drain their ability to remain in control of their distress. As participants were forced to watch their family members experience years of suffering, it seemed as though they were unable to provide relief for their loved ones or their own suffering for any lengthy period. This experience would often feed back into their own worries about the possibility they were witnessing their own future. This triggered the need for more strategies to be put into action to cope, using more of their energy resources.

All participants described challenging interactions with health professionals which made living at-risk more difficult, stressful and tiring. This seemed to add to their levels of exhaustion and reinforced feelings of being alone, disconnected and unseen*:*

"..I speak to my therapist. She's not specifically trained in Huntington's disease (...) I don't think she understands it very much (....). She said that she doesn't think that my mum's got Huntington's and she said that she thinks that I know deep down that I don't have Huntington's (....). Saying stuff like that isn't helpful, because, I'm trying to come to terms with the fact that I am at-risk (....) and she's saying things like, well, I don't think you have it" (Evelyn).

Ultimately, when reflecting on experiences with professionals, all participants spoke of how they felt unheard and questioned *'where are our voices?'* (Jane). This resulted in feelings of isolation and exclusion that led to intense sadness, low mood, anger and loss of hope that

their situation could change, and an unwanted idea, presented by professionals and current health care systems, that whatever they did, success was unlikely.

Discussion

This analysis of individuals' experiences of maintaining psychological wellbeing while living at-risk of HD resulted in three themes. The themes suggest that maintaining psychological wellbeing was a complex challenge that required the implementation of multiple strategies.

In the first theme, *'you're constantly in limbo': living in two worlds*, participants spoke about the movement between the HD and non-HD world. This is similar to experiences reported by people affected by HD in Sweden (including individuals at-risk, negative, and pre-symptomatic, and members of the family affected by HD) (Hagen, 2018) and Canada (at-risk or a family member not themselves at-risk) (Etchegary, 2009). Such findings are in line with earlier research which have concluded that living with HD is a fluid and dynamic experience and awareness of risk has a fluctuating prominence over life for those living at-risk and their families (Cox & McKellin, 1999).

Participants needed specific skills to make movement between the HD and non-HD world consistently available to them. Participants spoke of the importance of having separate groups of friends who may or may not know about HD or periods of time where they did not need to think about HD in their current conversation or environment. Participants had a varied age range and length of knowing about HD and risk, though this did not seem to impact their ability to move between the two worlds. Other factors seemed to be influential such as having a reliable social support network and being able to control one's own thoughts about their risk. Being within a positive mind-set and understanding risk seemed to have a more significant impact than age or length of knowing. For all participants, this movement was not an attempt to deny the existence of HD in their lives but a provision of respite from the distress the

awareness of the disease brought and an opportunity to be themselves independent of HD. A way of understanding this dilemma could be explained by the Sense of Coherence Theory (Kvåle & Synnes, 2013). The term sense of coherence (SoC) refers to the ability of an individual to approach life with a balance of optimism and control, referred to as a dispositional orientation (Antonovsky, 1993). Though participants did not openly discuss optimism, determination to live and live well was evident in the data. This is important when considering the application of such theory. The SoC has been shown to provide an understanding of the psychological processes involved in living with a chronic illness and that a strong sense of coherence, or understanding, directly relates to a better quality of life (Galletta et al., 2019). Rather than a pathological approach to illness, the sense of coherence theory brings a salutogenic approach (Antonovsky, 1987). That is, it focuses on the person's ability to stay healthy and maintain wellbeing when faced with adversity rather than focusing on factors that cause ill health.

Three main aspects form SoC, the first is comprehensibility: this refers to how understandable life events are to a person and the degree to which these events make sense. It is thought that the more understanding a person has concerning what is happening to them, the more able they are to face difficult situations. The second is manageability: this refers to what resources are available to meet the person's needs both from an internal (e.g., cognitive, behavioural and emotional strategies) and external (e.g., social support, relationships, culture) perspective. It is thought that having such resources increases a sense of control which increases coping. Participants did this in many ways such as deciding which friends should and should not know about their risk. Moreover, making decisions about with whom to share their HD risk is a technique found to be effective in other HD research to assist coping (Quaid et al., 2008). The third is meaningfulness: this refers to where the person's motivation comes from. If life has emotional meaning to the person, they are more likely to view problems as challenges

as opposed to hindrances being able to allocate meaning to events helps create motivation to increase effort to face the situation. To summarise: '…SoC is an overall orientation that conveys a feeling of trust because stressors are predictable, that resources to face challenges are available and that the challenges are with the individual effort because they have meaning' (Galletta et al., 2019, p. 2).

Participants in this study responded with various strategies to manage their wellbeing while living at-risk which can be understood through the application of this theory. For example, using this approach helps bring an understanding to the experience of the participants and their movement between HD worlds as they decided which world they were capable of interacting with when they were emotionally able to and had the resources to dwell within that particular place.

Participants also sought information, social support, a sense of control over onset and rationalisation which have a direct link to the comprehensive and manageability aspect of the SoC theory as participants tried to interact with the world around them and respond to the demands of living at-risk.

The second theme, *"I have to live, just bloody live": managing the possibility of a time-limited lifespan*, presents an awareness of time. Other chronic illness research suggests that individuals who know they will develop a chronic illness experience time in a different way from those who do not have such susceptibility (Jowsey, 2016). Participants often changed their plans which enabled them to prioritise their commitments and keep a focus on gaining as much enjoyment and achievement from life as they could. Participants identified the turmoil their attempts to control time caused and the effects of this time pressure and resulting distress on moving between the two HD worlds and the relevant HD identities. Understanding such difficulty is also supported by longitudinal grounded theory work on chronic illness (Charmaz, 1990, 1991), the results of which suggest that the difficulty people face is the balance between

the efforts to control time while preserving their self-identity. All participants expressed an awareness of time and time pressure to live well that seemed independent of their biological age and family narrative concerning age of onset; participants who were older (e.g., Jane and Marie) did not view their risk as diminishing as they aged, and it seemed their window of time moved with them.

Socioemotional Selectivity Theory (SST; Carstensen, 2021) seeks to explore the concept of time on life-span development and suggests that endings, whatever type (aging, relocating, illness), trigger motivational change. As a result of this motivational change, the person replaces existing goals with more emotionally meaningful goals. The theory has been explored in the context of breast cancer and concluded that SST can provide a useful way to understand motivation and psychological adjustment in people experiencing a time-limited future due to illness (Sullivan-Singh et al., 2015). The different factor with the population in this study remains the at-risk status and the desire to not engage with information in terms of a definitive answer from a genetic test that will place a putative end date in participants' thinking. Without such an end in mind, theoretically this should eliminate the time parameters within which they live, however this is not the case for participants in this study. Participants in this study did not experience a belief in an expanded lifespan. The at-risk status seemed to limit the lifespan in the same way as in other future limiting diseases.

Theme three, *"Is that who I am, is that what I am?": the exhausting quest to be seen as an individual first,* explored the tiring work involved in maintaining a sense of wellbeing while living at-risk of HD. The Illness Trajectory Framework (Corbin & Strauss, 1991), initially formed through grounded theory, focused on the work of people with chronic illness that is unseen by professionals (Star, 1995). The framework presents three reciprocally interactive types of work and distinguishes between biographical, everyday life and illness related work. The work involved in the context of people living at-risk of HD refers to the strategies and

coping techniques they utilised to remain psychologically and physically well. For this group, as there are currently no symptoms to manage, illness related work included taking physical care of themselves in terms of diet, exercise, and activity. The term biographical work within this study can be used to describe the participants' aims to form an identity independent of HD. The term everyday life work can be used to describe usual life commitments such as caring for their family (externally focused) and managing distress (internally focused). All participants consistently engaged in all areas of work. The intensity of this work did not diminish as age progressed and participants did not view their risk reducing as they aged. All participants hoped to reach retirement and have considerable time to enjoy that experience, only then did it seem that peace would be theirs and the hard work could stop, once all areas of the life cycle had been lived in.

Study Limitations

There are several limitations in the current study. Firstly, IPA relies on the ability of participants to articulate their experiences, complex thoughts and feelings clearly although this has been stated to be difficult (Willig, 2008). The limitations of language and articulation are acknowledged within IPA and overcome through the researcher understanding how participants make sense of their experiences (Smith, 2011). IPA relies on the interpretations of the researcher and while transparency can be evidenced (e.g., through the sharing of transcripts, procedures, and involvement of co-authors) recognition of the researcher's interpretations and possible alternatives requires consideration.

Secondly, as participants were recruited via HD specific support groups on social media platforms, this could have led to selection bias. People involved in such groups may be more proactive in maintaining their psychological wellbeing than those living at-risk who do not embark in such involvement. All participants identified as white British; therefore, future

research focused on non-western participants may be useful to explore a more diverse representation.

Thirdly, the participants in this research were living at-risk in the UK, therefore, their experiences of their own cultural context and interactions with the NHS healthcare system needs to be considered. Cultural and social context and experiences of accessing medical care or support can influence individual's perception of illness, such as HD (Arran et al., 2014), which influences the emotional response and coping strategies used to manage illness (Leventhal et al., 1980). Such influences need to be considered when exploring the transferability of the findings to other people living at-risk.

Research recommendations

Studies evaluating the effectiveness of psychological interventions for people affected by HD remain limited (Zarotti et al., 2020), therefore research is required to develop and evaluate the use of such approaches with those living at-risk of HD. The issues raised in this study highlight, as well as the unmet individual needs of this group, the wider systemic issues concerning the impact of social stigma, lack of knowledgeable professionals and services.

Given participants' experience of stigma, awareness campaigns could help raise knowledge and reduce such negative interactions. It has been suggested that making society aware of the negative and harmful impact such stigma has on people with genetic diseases, such as HD, could reduce its occurrence (The Lancet, 2010; Wexler, 2010). It is important that campaigns that focus on raising awareness are delivered effectively and with clarity and that they have substantial funding provision to create accessible and attractive resources, as insufficiencies in such areas have been shown to make such campaigns ineffective (Wakefield et al., 2010). Recommendations from this included auditing of the materials and a planned, targeted and more comprehensive approach focusing on reaching the public in GP waiting rooms and consultations (McNulty et al., 2010).

The knowledge of professionals concerning HD would need to be targeted specifically using specific clinical resources such as continued professional development courses or clinical updates. There is also the consideration that such campaigns and educational approaches will not alter the current lack of service provision for those living at-risk of HD. While the theoretical models presented may provide a starting point by presenting ways for professionals to increase their knowledge of the impact of living at-risk of HD, without further research, funding and implementation, no effective change will emerge for this group.

Practice implications

The findings of this study have relevance for clinical psychology and wider health systems due to the complex interactions between living at-risk of a genetic disease and levels of experienced distress. Provisional research has found that psychoeducation in the form of forums was well received by people who were pre-symptomatic (Gluyas et al., 2023). It may be useful to explore whether such an intervention would be useful for the at-risk population.

When living at-risk, findings show there was a need for emotional regulation skills. Emotional regulation is the ability to experience, make sense of and modulate emotions (Gross, 1998) which is a useful skill to place within the SoC in terms of understanding one's identity and the illness trajectory framework in terms of the level of work needed to stay well. It is thought that the ability to self-regulate when experiencing a chronic health condition is important when considering adjustment to illness (de Ridder et al., 2008).

Self-management techniques were also evident throughout all themes. In chronic illness literature (Dunbar-Jacob & Mortimer-Stephens, 2001; Riemsma et al., 2004) good self-management (e.g., regarding diet, exercise, medication compliance) has been consistently evidenced as having positive impacts on physical health due to the care taken to provide wellness for the physical body. The findings in this study highlight participants' engagement in

self-management techniques before any advice, testing or diagnosis is even given. There is a need for professionals to support such explorations and implementation of interventions and strategies.

Participants explored their varied experiences of accessing psychological support. Professionals knowledge (psychologists, psychiatrists and therapists) of HD was important to participants with poor knowledge having negative impact on participants. Although specialist HD service provision is sparse, it may be beneficial for existing HD services to routinely employ Clinical Psychologists who have knowledge of HD to provide specialist psychological support for those at-risk as well as those with HD.

In terms of improving knowledge within first contact settings, primary care health settings in the UK often employ the Chronic Care Model (CCM). The CCM offers a framework for health care providers to be supportive in patient provider interactions resulting in productive outcomes and is often used when working with chronic illness, such as diabetes (Wagner et al., 2001). The model focuses on the parts individuals and professionals facilitate in chronic illness management. While the model incorporates self-management for the physical and psychological aspects of illness, it is often delivered within health care settings as instructions with which a patient must comply (Bower et al., 2012). This is an example of how current health care providers may focus on the biomedical aspects of health conditions (managing the disease) and not the individual perspective of managing the impact of the condition on the person (Leys, 2010). The CCM has been adapted for use with families with a person with HD, focusing on the implementation of the keyworker role to understand the specific needs and therefore provide the required individualised support (Wilson et al., 2014). While the model is flexible, it does not incorporate support for those living at-risk as it appears to be diagnosis dependent. Indeed, access to such interventions and support plans is only available if a diagnosis of chronic disease/illness has been given and so those living at-risk are often

excluded by services due to their choice not to embark on genetic testing. Even if at-risk individuals had received a positive test result, they would still be unable to access most chronic illness support until an actual HD diagnosis is given.

Conclusion

This IPA study explored the experience of 12 participants maintaining their psychological wellbeing while living at-risk of HD. Participants spoke about the complex day to day navigation between HD and non-HD worlds, the drive to live the best life they could in the time they have and the exhausting impact of living at-risk of HD. The findings highlight the need for improved support for people living at-risk of HD and theoretical models that may support this direction have been discussed. Further research to create a model that encapsulates the needs of those living at-risk of HD in its fullness is required, alongside changes to health care systems that exclude those in need either by the diagnostic eligibility criterion and/or lack of HD knowledge in health settings.

Author Contributions

Authors Hollie Cooper, Professor Jane Simpson, Dr Maria Dale, Dr Fiona Eccles contributed to this work. Hollie Cooper made substantial contributions to the acquisition, analysis, and interpretation of data for the work which was supported by Professor Jane Simpson, Dr Maria Dale and Dr Fiona Eccles. All authors made substantial contributions drafting the work and revising it critically for important intellectual content.

Authors Hollie Cooper and Dr. Fiona Eccles confirm that they had full access to all the data in the study and take responsibility for the integrity of the data and the accuracy of the data analysis. All of the authors gave final approval of this version to be published and

agree to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved.

Conflict of interest statement

Authors Hollie Cooper, Professor Jane Simpson, Dr Maria Dale, Dr Fiona Eccles declare that they have no conflict of interest.

Human studies consent

Ethical approval was granted by the Lancaster Faculty of Health and Medicine Research Committee. Further information regarding ethics can be seen in the Ethics section.

Data availability Statement

The author elects not to share data.

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Table 2 – A

Participant demographics

Pseudonym	Age	Gender	Relationships status	Biological children	Employment status	Family member with HD	Length of time aware of HD risk	Likelihood to have the expansion (%)	Accessed psychological support
Sarah	43	female	married	2	Full time	Father Grandmother Great Uncle	Long known history of HD – unsure of when knowledge of risk was gained	50	Y
Jane	60	female	widowed	0	Self employed	Father Grandmother Aunt Uncle	Long known history of HD – unsure of when knowledge of risk was gained	50	Y
Lucy	37	female	married	1	Full time	Mother	Long known history of HD – unsure of when knowledge of risk was gained	50	Y
Marie	66	female	married	0	Retired	Sister Mother x2 Uncle Grandad	Long known history of HD – unsure of when knowledge of risk was gained	50	N
Daniel	45	male	Long term relationship	0	Full time	Mother Sister	9 years	50	N
Lisa	33	female	married	1	Full time	Mother Grandad	13 years	50	Y
Ian	24	male	Long term relationship	0	Full time	Father	8 years	50	Y
Gill	53	female	Married	1	Self-employed	Mother	18 years	50	Y
Karen	19	female	Single	0	Unemployed	Father	12 years known of HD, understood risk later though unsure when	50	Y
James	43	male	married	1	Full time	x2 brothers cousin	14 years	50	Y
Cath	40	female	married	0	Full time	Mother	32 years known of HD, Understood risk later though unsure when	50	Y
Evelyn	23	female	single	0	Full time	Grandfather	4 years	25	Y

Image 1

Making exploratory notes on a participant transcript

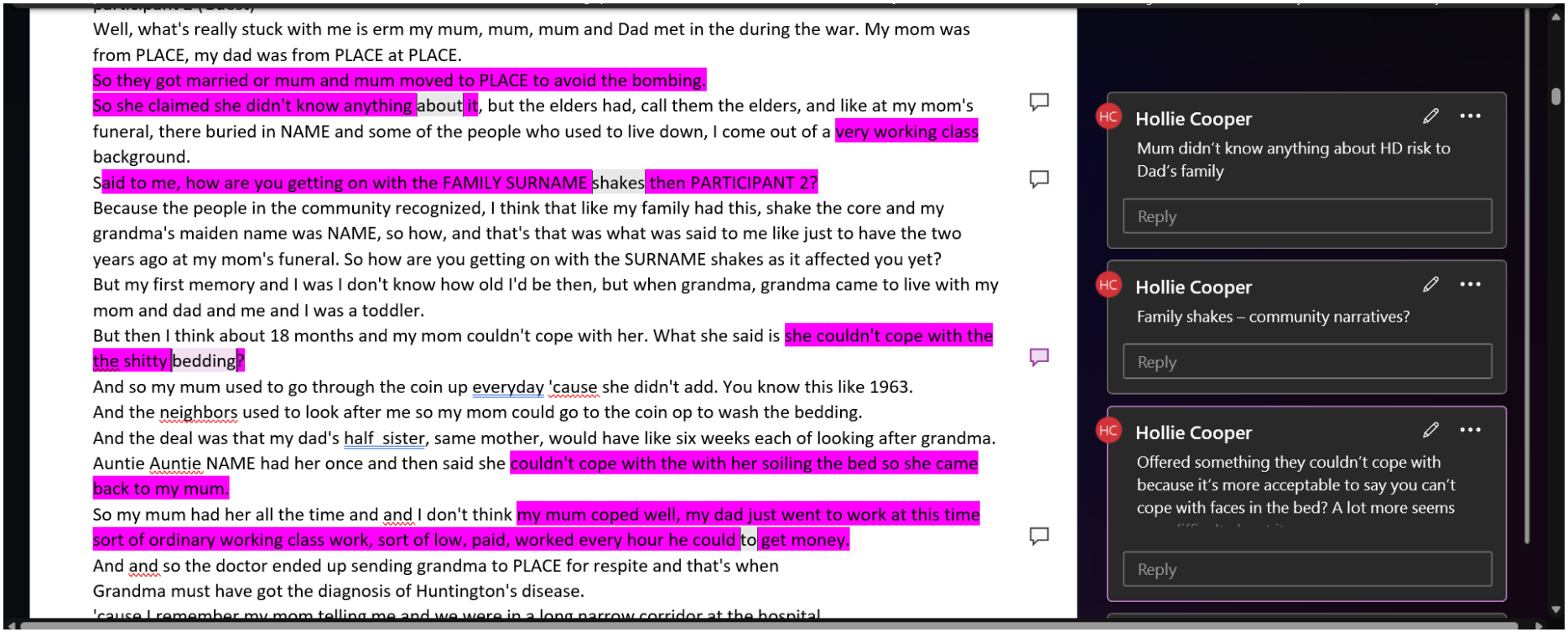

Table 2 - B

Formation of experiential statements into personal experiential themes for Lucy and Marie

Participant	Experiential statements	Personal experiential themes	Name of theme
Lucy	Starting at the super nice end of the spectrum so that if I get symptoms I have a long way to go before I am nasty If I am a good person I might suffer less Having more patient for people Understanding when people struggle and recognising that before they say it	HD makes me a better person I am more attuned to others because of my experience	HD makes me a better person/HD isn't all bad?
	Who raised me and for how long – mum vs HD Suicide is a solution 25 risk is not important, 50 risk is the end of your life Parent is an unstable alcoholic Tells people you are nasty and terrible	Makes you question your life Overwrites the truths you try to hold on to Clouds perspective Brings in judgement from those observing and gives them permission to speak in to your life	HD is a liar
	Betrayed by your own body and you can't chop it off to fix it or treat it like breast cancer		Betrayal

	Late-onset in my family (60s) Family need to agree on testing before anyone tests Depression is normal in our family Being nasty and hostile is a family trait You will suffer – medicine can do nothing so just suck it up and get on with it	Family narratives can be hindering and difficult to manage	Narratives have power to help or hinder
	No one knows anything about it There's no easy way to get accurate information No one wants to talk about the wider life impact on mortgages, insurance, adoption, IVF (testing implications) Was only able to access knowledge and help because of husbands degree and professional friends in medicine Social media does not help – full of people crying out for help and no-on e responding but us We are not heard	Lack of information resulting in lack of support Unheard and unsupported	Where are our voices – who will hear us

Participant	Experiential statements	Personal experiential themes	Name of theme
Marie	Abandoned by medical professionals – diagnosed with HD via a letter in the post after a admission unrelated to HD – first family knew of HD No support Unable to access anything unless getting tested or had been tested and was positive No leaflets at any of the neuro clinics only Parkinson's and Alzheimer's No support for the family for a disease that is a family disease – you drop a bombshell and are then no-where to be seen	Professionals don't know what they're doing Communication is poor There is no information from medical professionals that is helpful	Professional support is poor
	Some of my friends know some don't – it's nice being able to pick which category to spend time with Being busy is important in staying well Doing things I value and get a s ense of satisfaction from are important – if I don't enjoy it, I don't do it Keeping it in the back of the mind where you can without ignoring it's a factor	Being able to cope is a process that has ups and downs Coping is difficult most of the time Knowing yourself and where HD sits for you is important	Coping comes in peaks and troughs Keeping HD in its own space helps – two worlds?

	Allowing some symptoms watching helps – is that HD, no, I'm ok lets her place it to the back of her mind Social media can help but most of the time doesn't – the connection and perspective it can give is temporary, your own story takes over quickly when you come off it Staying healthy and active and knowing I have looked after my body is important – I have done all I can to stop it, eventhough it doesn't work like that	Being able to offer self reassurance in important	
	You have to decide to live and not dwell on it all the time Make a decision to live each day and give it your all Live each day as if it was the last – no regrets Your decision if you want to test – no one else's Decisions are a process and they can change and that's ok	Making decisions that are true to you and your self is important Living a meaningful life in the time you have and deciding to do so rather than being pushed into it	Making decisions gives a sense of control
	Later onset Affects all the men in my family until mum Family denied mum had HD and said she was a drunk because it only affects men Family shakes	Family narratives help and hinder – false sense of hope, very negative	Narratives have power

Table 2 -C

Example of a section of Grouped Experiential Themes, interpretations and supporting quotes

Group experiential theme (GET)	**My thoughts & interpretations** Text in bold indicates my recurrent thoughts/interpretations. These were collated to form the final themes. Not all bold text was included in themes.	**Participants**	**Supporting quotes**
techniques to help/maintain wellbeing		All	
Determination to live **living a good life** **living life to the full**	All participants had things they did to stay well. The main aspect of this was to be able to accept that there will be rough times where you can't get away from HD as your life is written to be prolonged suffering and death like the family members before you. No choice, no option, imprisoned in a future you don't want, you want to get away from but have to accept – caught **between the two, forced to exist in both.** Seeking knowledge and planning was the most common way people stepped back out of this zone. Almost as if there **are zones participants can access and**	All	You've got to bloody live, you get one life (pt 2 pg 8) Live your life, anything can get you (pt 2, pg 1) I live my life very fully, try and treat everyday as if it's the last and I think that's the only approach that you can take whether you have HD or anything else sort of hanging over you (pt 11 pg 3) Pure dedication and drive to live a better life than my brother then just to get the out of the most out of life (pt 10 pg 9)

	move through but no order, fluid, dynamic? Some people avoided professionals because their lack of knowledge forced them into a state of anxiety around HD which their coping skills were less effective against as it made them question where they would get support and felt increasing sense of isolation. Alone. **Also had social groups that they could feel apart from but within. Does this relate to the zones above?**		I've got the drive every morning to get up and think you know this is another opportunity to try and be the best version of me possible (pt 11 pg 14)
Setting tests to evidence no symptoms/onset to control anxiety	Aware of my own thoughts and experiences of health care interactions and the emotive response I am having to reading and re-reading in the search for quotes. I feel pressure to make sure the work I produce does the participants experience justice. Tests helped participants remain level, put HD on the back burner, back of their minds, not in the	2 3 4 5 7 8 10 11 12	I'd have to learn songs and have to rehearse them in my head and I'd have to know the full title…when it came out..the band as well so I used to test myself (pt 7 pg 18) I have a habit when I'm in bed of rubbing my feet together and I like trying hold to stop doing it to

	immediate sight for the day. Using the test enabled life? **Live in a illness free space?**		se how long I can hold it so that it's I can really discover whether they're involuntary or not (pt 6 pg 5) I try to box it off and I live in the awareness that at the moment I'm symptom free, that I know that if I start to display symptoms I have a plan ahead of me (pt 1 pg 2)
Having a good example of care and support for HD+ family, having future plans agreed and care agreed	Those who saw good examples of care and support seemed to have a more positive view of their future if they were to develop HD and less fear, not so overwhelmed, the **biggest fear was losing themselves, identity**, what makes them them not the physical deterioration. Loss of self outweighs loss of physicality?	1 3 4 5 6 7 10 11 12	My mum she's been amazing like she's been by my Dad's side the whole time, she still visits him every week and like fights for him to have the best care and everything (pt 9, pg 16) Having such a positive and supportive father was reassuring for us that you know there are people out there that want to genuinely help and be there for people you know, society won't necessarily just turn its back on you (pt 11 pg 2)

			He (dad) also said to me you know who are you to say to somebody that they shouldn't have the opportunity to go through that you know who are you to turn around and say that you know somebody shouldn't be your future carer or whatever it might be, it's not your choice, it's their (pt 11 pg 7) Me and my sister have spoken about it (euthanasia) and we both said that if we can afford it, then as soon as we are symptomatic we want to sort of go there so that we're not suffering for decades…..we're going to look after each other if either one of us is positive…I guess that gives me a bit of like hope for the future that there is that kind of option (pt 9 pg 11). I don't ever want to be a burden to my husband…put me with the professionals…I have to make him aware (pt 3, pg 9)

Practical skills & tools Exercise Eating well Active Friends Boundaries Talking Managing stress Turning it into a positive Knowing how you feel and what you think about HD Knowing your warning signs	**Self-management** skills – reminds me of the five ways to wellbeing as though using techniques and methods that are general offer a sense of control or achievement over the disease **Acceptance** that there is an illness and something needs to be done, a taking of control, a taking of action and direct involvement as if that may changer the direction of the course of positive vs negative? Taking control and power back from HD, not allowing it to take over – resilience, determination, **hard work**, strategies	1 2 4 5 6 11 12	If I'm in a crisis or ….I can't think rationally I have to do like things like exercise or I have a cold shower to like clear my head or I do a lot of yoga…then more general things like trying to reach out to people like my family….therapy every week….obviously medication is a big part of me being stable (pt 9 pg 19). You've got to manage those things that are causing stress and try taking them out of the frame (pt 5 pg 11) I know my boundaries, I know what I need to keep me focused and to keep me fit and healthy (pt 10 pg 13) I might go into work and not want talk to anyone or I might

			not go the gym or I might not want to cook (pt 7 pg 19) My resilience come from within being able to talk…I can openly talk to people about it and it helps me when I talk so its just its almost like its cleansing yourself in some way you are able to get that lift that weighty off your shoulder slightly (pt 10 pg 10). It's a motivating force (HD) but will there ever be a point at which I sit back and say you know what I'm happy I'm happy with where I am now, I don't know (pt 11 pg 14)…..it's not something you would actively choose (being at-risk)…I turn it into the positive because that's the best way for me to handle it and motivate myself (pt 11, pg20) I suppose I know in my own mind how I'm feeling about Huntington's and how I'm feeling about whether I'm going to get it or not and I know how

Magical thinking: Starting at the good person end of the spectrum as far you as you can so HD takes longer to get you Living the best life you can	**Working hard** to remain a good person, to keep a part of themselves, to prevent HD from 'winning' and 'taking' them. **Have to work** to know you have done everything you can to stay as well as you can for as long as you can. Know you've done all you can so if you do get it you can be peaceful knowing you've lived a good life There will be a cure in time for my children Thinking you can delay onset by keeping active, healthy etc as above	1 2 4 5 6 12	emotional I'm feeling about it (pt 5 pg 23) I made a decision that I would be really nice to everybody all the time, because then if it got me as I'd have, if my starting place was sort of really at one end of the spectrum when I was really really such a lovely, nice person, then perhaps I'd die before I became evil. Which is magical thinking isn't it? Pt 2 pg 5. I want to keep my cognitive levels ticking over (pt 6 pg 17)
Having correct knowledge: Understanding what your risk is and what it means Decisions on testing Accepting that the journey has ups and downs	Important of knowledge, the impact and effect of that knowledge, whether to share it and who to share it with and the impact of this – the zones seem affected by who knowledge is shared with? Is it like there are two worlds as oppose to zones? Moving between two worlds, a well world and a ill/disease world? **Or a HD and non-HD world?**	3 5 7 9	I'm educated and I'm aware of the biology and I know what the risk is and sometimes it's hard (pt 1 pg 9) At the time I had 25% risk, so to me I had 75% risk of not having

Understanding what HD is in your life Accepting that there are no definitive answers Other ways to achieve goals			it so…so 75 versus 25 I'm kind of, why would I worry about the 25% pt 3 pg 1) Within the family there's no history of Huntington's so when mum got diagnosed that came as quite a shock because my understanding was that it was genetic disease (pt 5, pg 2) I can't have kids, now I've learnt the ways that you can then its another relief type of thing (pt 7 pg 12)
Spirituality	Being connected to people and the world, relational connection, higher being, best friends, true self, sense of purpose – connected to things other than HD, purpose outside of HD. **New zone?**	1 4 6 11	I would say it's a peace of God…..where's that peace come from….Having a faith definitely helps (pt 1 pg 9) My entire ambition really just to live a good virtuous life connected with nature with all people and so you can't really have any fuller life than feeling as if you're working towards that, there isn't anything you could say

			that would be more fulfilling to me than that (pt 11 pg 4) I need a sense of purpose..something that at the end of the day I feel satisfied with what I am doing (pt 4 pg 17) I have got a job, I've got friends who are fantastic my husband is amazing my son is just the absolute be all and end all of everything…we are a really close family (pt 6 pg 19) I need people around me (pt 4 pg 15) My mental wellness comes from my friends pt 4 pg 18)
Hope	People are interested More research More treatments A cure Planning for the future, the future exists but is out of reach, the future tempts, jokes, teases?		I have hope that I'm negative at the moment…while we are at this stage I'm feeling hopeful that we might just miss this (pt 1 pg 11) When I saw your thing and I thought bloody hell someone here is doing a professional thing,

			how do they know about it, I never knew about it…why are they interested, well if they're trying to find information about it I need to support it I need to at least help them (pt 7 pg 7) I'm hopeful that in 20 years' time that there's kind of, I'm doing this research for you cause I think anything that I can help with that might help…or we can do to help future generations (pt 3 pg 9)

Appendix 2 - A

Journal of Genetic Counselling Author Guidance

AUTHOR GUIDELINES

SECTIONS

1. AIMS AND SCOPE

The Journal of Genetic Counseling (JOURNAL), published for the National Society of Genetic Counselors, is a timely, international forum addressing all aspects of the discipline and practice of genetic counseling. The JOURNAL focuses on the critical questions and challenges that arise at the interface between rapid advances in genetics and technology and the impact on individuals and communities at genetic risk. The publication provides genetic counselors, other clinicians and health educators, laboratory geneticists, bioethicists, legal scholars, social scientists, and other researchers with a premier resource on genetic counseling topics in national, international, and cross-national contexts.

As a crucial resource for genetic counselors and associated professionals, the JOURNAL'S primary purpose is to report original research in the following areas:

- **Genetic Counseling Theory, Methods, and Practice:** addresses theory development and evaluation, methods development and evaluation, current practice, and/or outcomes research relevant to the discipline and practice of genetic counseling in clinical or non-clinical settings;
- **Public Health, Public Policy, and Access and Genetics Service Delivery:** addresses public health genomics, health behaviors, policy aspects related to genetic counseling and genetic testing, precision medicine, models of genetics services delivery;
- **Education and Genetics Professional Workforce Issues:** addresses educational training, professional development, and workforce topics related to genetic counseling;
- **Ethical, Legal, Psychological, and Social Issues:** addresses ethical, legal, psychological, and/or social issues related to genetic counseling, genetic testing, genetic services, and/or genetic information regarding individuals, communities, and the public
- **Risk Assessment:** addresses algorithms, theoretical models, or empirical data for use in genetic counseling risk assessment

- **Minority and Health Disparities:** addresses diversity, equity, and inclusion topics relevant to the practice and discipline of genetic counseling

Note: The Journal of Genetic Counseling does not publish research involving non-human animals.

Return to Guideline Sections

2. SUBMISSION

Once the submission materials have been prepared in accordance with the Author Guidelines, manuscripts should be submitted via the JOURNAL'S Editorial Manager site: **https://www.editorialmanager.com/jogc/default.aspx**. More details on how to use Editorial Manager are also available at **https://www.editorialmanager.com/jogc/default.aspx**.

Need assistance? For help with submissions, please contact the Editorial Office at JOGC@Wiley.com. When necessary, the Editorial Office staff may refer questions to the Editor-in-Chief.

A manuscript is considered for review and possible publication on the condition that it is submitted solely to the JOURNAL, and that the manuscript or a substantial portion of it is not under consideration elsewhere. Preprints and/or presentation of the content at meetings prior to submission are acceptable. However, authors should kindly note that submission implies that the content has not been published or submitted for publication elsewhere except as a brief abstract in the proceedings of a scientific meeting or symposium or as a preprint. Note, the JOURNAL uses iThenticate's CrossCheck software to detect instances of overlapping and similar text in submitted manuscripts.

The submission system will prompt the author to use an ORCiD ID (a unique author identifier) to help distinguish their work from that of other researchers. **Click here** to find out more.

Data Protection Statement: By submitting or reviewing a manuscript for the JOURNAL, your name, email address, and affiliation, and other contact details the JOURNAL might require, will be used for the regular operations of the JOURNAL, including, when necessary, sharing with the publisher (Wiley) and partners for production and publication. The JOURNAL and the publisher recognize the importance of protecting the personal information collected from users and have practices in place to maintain the security, integrity, and privacy of the personal data collected and processed. You can learn more **here**.

Return to Guideline Sections

3. MANUSCRIPT TYPES AND GENERAL REQUIREMENTS

MANUSCRIPT TYPES (see Table for additional details)

Original Article: Manuscript reporting original quantitative, qualitative or mixed methods research using a form of systematic study or inquiry to address a question relevant to the discipline and practice of genetic counseling. Systematic reviews included in this category.

Brief Report: Manuscript reporting an observation that adds to the knowledge of the discipline and practice of genetic counseling.

Case Study: Manuscript addressing issues relevant to discipline and practice of genetic counseling by demonstrating and stimulating thought about a difficult ethical, counseling, or genetic testing situation the author has encountered.

Professional Issue: Manuscript reporting reflections by the author(s) on the discipline and practice of genetic counseling.

Review: Manuscript summarizing the literature on a focused topic. Authors should contact the Editor-in-Chief prior to submission.

Commentary: Manuscript addressing matters of interest or controversy to the readership. Generally commissioned by the Editor-in-Chief.

Correspondence: Manuscript addressing work previously published in the JOURNAL

Practice Resource, Practice Guideline, Focused Revision: Manuscripts addressing specific areas of genetic counseling practice. Commissioned by the National Society of Genetic Counselors' Practice Guidelines Committee.

Book Review: Manuscript that reviews a book of relevance to the JOURNAL scope.

Conference Report: Manuscript with executive summary of important conference and/or select conference abstracts. Authors should contact Editor-in-Chief prior to submission.

Corrigenda and Errata: Manuscript describing corrections to a work previously published in the JOURNAL.

GENERAL REQUIREMENTS

Format

- double-spaced
- 1 inch margins
- 12 point font (Arial or Times New Roman).

- Footnotes should be avoided in the main text. When their use is absolutely necessary, footnotes should be numbered consecutively using Arabic numerals and should be typed at the bottom of the page to which they refer. Place a line above the footnote, so it is set off from the text. Use the appropriate superscript numeral for citation in the text.

English Language

Manuscripts must be submitted in grammatically correct American English. Manuscripts that do not meet this standard cannot be reviewed. Authors for whom English is a second language should consult an English-speaking colleague or consider having their manuscript professionally edited before submission to improve the English. A list of independent suppliers of editing services can be found at **https://wileyeditingservices.com/en/**. All services are paid for and arranged by the author, and use of one of these services does not guarantee acceptance or preference for publication.

Inclusive Language

An important element of the Journal of Genetic Counseling's commitment to diversity, equity, and inclusion is fostering authors' use of inclusive language in their manuscripts. To facilitate use of inclusive language we have compiled two excellent and comprehensive resources for authors' use:

CDC guidelines
- **https://ehe.jhu.edu/DEI/Health_Equity_Style_Guide_CDC_Reducing_Stigma.pdf**

APA guidelines - **https://apastyle.apa.org/style-grammar-guidelines/bias-free-language**

Specific guidance:

1. Participant-reported demographic variables should be ascertained/used when reporting demographic information on participants.
2. Authors should explain the use of person-first or non-person-first language in the manuscript. For guidance, see https://apastyle.apa.org/style-grammar-guidelines/bias-free-language
3. For guidance on reducing stigma through use of terminology describing groups, see https://ehe.jhu.edu/DEI/Health_Equity_Style_Guide_CDC_Reducing_Stigma.pdf, and https://apastyle.apa.org/style-grammar-guidelines/bias-free-language
4. For guidance on use of terminology for sex and gender, see https://apastyle.apa.org/style-grammar-guidelines/bias-free-language

General Style Points

- **Abbreviations:** In general, terms should not be abbreviated unless they are used repeatedly and the abbreviation is helpful to the reader. Initially, use the word in full, followed by the abbreviation in parentheses. Thereafter use the abbreviation only.
- **Units of measurement:** Measurements should be given in SI or SI-derived units. Visit the Bureau International des Poids et Mesures (BIPM) website for more information about SI units.

- **Numbers and p-values:** numbers under 10 should be spelled out, except for: measurements with a unit (8 mmol/L); age (6 weeks old), or lists with other numbers (11 cousins, 9 aunts, 4uncles). Numerical figures (excluding p-values) should not exceed 2 decimal places. For p-values less than 0.001, report as $p < 0.001$. For p values greater than 0.001, use 2-3 decimal places. Do not use a leading zero for statistics that cannot exceed 1.0.
- **Genomic Terminology and Nomenclature:** Please use the following terms:
    - *genome sequencing* instead of *whole genome sequencing*
    - *exome sequencing* instead of *whole exome sequencing*
    - *pathogenic variant* instead of *mutation*
    - *secondary finding* instead of *incidental finding*

Use of italics

- italicize gene names (e.g. *FBN1*)
- do not italicize protein names (e.g. fibrillin)

Sequence variants

- Sequence variants should be described in the text and tables using both DNA and protein designations whenever appropriate.
- Sequence variant nomenclature must follow the current HGVS guidelines; see hgvs.org, where examples of acceptable nomenclature are provided.
- Human gene nomenclature should follow the standards of the HUGO Gene Nomenclature Committee (HGNC), see https://www.genenames.org/.

- **Pedigrees:** Pedigrees should follow the recommendations for standardized nomenclature accepted by the National Society of Genetic Counselors. Authors should consult the following references for these recommendations:
    - Bennett, R. L. , Steinhaus, K. A., Uhrich, S. B., O' Sullivan, C. K., Resta, R. G. , Lochner-Doyle, D., Markel, D. S., Vincent, V., & Hamanishi, J. (1995). Recommendations for Standardized Human Pedigree Nomenclature. Journal of Genetic Counseling, 4, 267-279.
    - Bennett, R. L., Steinhaus French, K., Resta, R. G., & Lochner Doyle, D. (2008). Standardized Human Pedigree Nomenclature: Update and Assessment of the Recommendations of the National Society of Genetic Counselors. Journal of Genetic Counseling, 17, 424-433.
- **Trade Names:** Chemical substances should be referred to by the generic name only. Trade names should not be used. Drugs should be referred to by their generic names. If proprietary drugs have been used in the study, refer to these by their generic name, mentioning the proprietary name and the name and location of the manufacturer in parentheses.

Return to Guideline Sections

4. PREPARING THE SUBMISSION

Specific Requirements for each manuscript type are summarized in this **Table.**

Parts of the Manuscript

The manuscript parts should be uploaded as separate files:

- cover letter
- main text file
- tables
- figures
- supplementary information files

Cover Letter

- include a statement that the work presented in the manuscript has not been published elsewhere and is not currently under review elsewhere. Include a statement that the work is available as a preprint or was presented at a conference, if relevant.
- If the study includes original data, at least one author must confirm in the cover letter that he or she had full access to all the data in the study and takes responsibility for the integrity of the data and the accuracy of the data analysis.

Main Text File

The **main text file** should be presented in the following order in one document (as appropriate for manuscript type):

1. Title Page
2. Abstract and keywords
3. 1-2 sentence responses to:
    1. What is known about this topic:
    2. What this paper adds to the topic:
4. Main body of paper
5. Author Contributions
6. Acknowledgements
7. Compliance with Ethical Standards
    1. Conflict of Interest
    2. Human Studies and Informed Consent
    3. Animal Studies

4. Data Availability Statement
8. References
9. Tables
10. Figure legends
11. Figures

Title Page

The title page should include (in this order):

- title of the article. Authors may benefit from referring to Wiley's best practice tips on Writing for Search Engine Optimization.
- authors' names (no degrees) in the order to be published. Please denote cases of equal authorship with a superscript and footnote, e.g., for joint first or joint senior authorship, the footnote should say 'X and Y should be considered joint first author' or 'X and Y should be considered joint senior author', respectively.
- authors' institutional affiliation(s) where the work was conducted, and the author's present affiliation if different from where the work was conducted. The affiliation should comprise the department, institution (usually university or company), city, and state (or nation) and should be noted with numbered superscript to the author's name and footnote.
- Telephone number and e-mail address of the one author designated to review proofs (the corresponding author).
- Suggested running head. The suggested running head should be less than 80 characters (including spaces) and should comprise the article title or an abbreviated version thereof

Abstract

- unstructured, i.e., no main headings or subheadings
- maximum 300 words
- contains major keywords summarizing the work
- If reporting on a clinical trial, include the name of the trial register and the clinical trial registration number at the end of the Abstract.
- See Table for manuscript category-specific information

Keywords

- 3 to 6 keywords
- **Please include 2-3 keywords from** this list.

What is known about this topic

- 1-2 sentences

What this paper adds to the topic

- 1-2 sentences

Main Body of Paper

- **Please refer to this Table for specific manuscript type requirements**.
- For all research involving human participants, please include a statement in the **Methods** section confirming that the study was reviewed by an institutional review board/human investigations committee/ethics committee (include name of committee and IRB protocol number) and approved or waived as human subjects research.

Author Contributions

Prior to submitting the manuscript all authors fulfilling the International Committee of Medical Journal Editors (ICMJE) criteria for authorship should be identified and should agree on the order in which their names will be listed in the manuscript.

ICMJE criteria are:

- Substantial contributions to the conception or design of the work; or the acquisition, analysis, or interpretation of data for the work; AND
- Drafting the work or revising it critically for important intellectual content; AND
- Final approval of the version to be published; AND
- Agreement to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved.

If the study includes original data, at least one author must confirm that he or she had full access to all the data in the study and takes responsibility for the integrity of the data and the accuracy of the data analysis.

Please insert the following statements in the Author Contributions section, specifically identifying the relevant author(s):

Authors X and Y confirm that they had full access to all the data in the study and take responsibility for the integrity of the data and the accuracy of the data analysis. All of the authors gave final approval of this version to be published and agree to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved.

During the submission process you will be asked to select the contribution(s) made by each author from a pre-specified drop down menu.

Acknowledgements

Contributions from anyone who does not meet the ICMJE criteria for authorship should be listed in an Acknowledgements section. Financial and material support should also be mentioned in this section. Authors should list all funding sources and are responsible for the accuracy of their funder designation. If in doubt, please check the Open Funder Registry for the correct nomenclature: **www.crossref.org/services/funder-registry**.

If this paper is to be considered for the **Journal of Genetic Counseling Best Trainee Paper award**, please include a statement indicating that the research presented in the paper was conducted while the first author was in training or to fulfill a degree requirement of the first author. See the **Best Trainee Paper Award** tab on the JOURNAL website for more information about this award. Thanks to anonymous reviewers is not considered appropriate to include in Acknowledgements.

Compliance with Ethical Standards

- ***Conflict of Interest Statement***

The JOURNAL requires that all authors disclose any potential sources of conflict of interest. Any interest or relationship, financial or otherwise, which might be perceived as influencing an author's objectivity is considered a potential source of conflict of interest. These must be disclosed when directly relevant or directly related to the work that the authors describe in their manuscript. Potential sources of conflict of interest include, but are not limited to, patent or stock ownership, membership of a company board of directors, membership of an advisory board or committee for a company, and consultancy for or receipt of speaker's fees from a company. The existence of a conflict of interest does not necessarily preclude publication in the JOURNAL.

If the authors have no conflict of interest to declare, they must also state this in the manuscript. It is the responsibility of the corresponding author to review this policy with all authors and collectively to list in the manuscript under the subheading “Conflict of Interest” *all* pertinent commercial and other relationships.

The above policies are in accordance with the Uniform Requirements for Manuscripts Submitted to Biomedical Journals produced by the International Committee of Medical Journal Editors (**http://www.icmje.org/**).

The Conflict of Interest Statement should **mention each author separately by name.**

Recommended wording

Author W declares that she has no conflict of interest.

Author X has received research grants from Drug Company A.

Author Y has received a speaker honorarium from Genetic Testing Company B and owns stock in Genetic Testing Company C.

Author Z is founder and CEO of Genetic Education Company D.

If multiple authors declare no conflict, this can be done in one sentence:

Author X, Author Y and Author Z declare that they have no conflict of interest.

Submitting authors should ensure they liaise with all co-authors to confirm agreement with the final statement.

- ***Human Studies and Informed Consent***

The JOURNAL requires that all appropriate steps were taken to obtain informed consent of all human subjects participating in the research reported in the manuscript submitted for review and possible publication, and statements to this effect must be included under this subheading. Participant anonymity should be preserved and all identifying information should be excluded in the manuscript unless the information is essential for scientific purposes and the study participants or patients (or parents or guardians) have provided written informed consent for publication. Images and information from individual participants will only be published where the authors have obtained the individual's free prior informed consent. Photographs need to be cropped sufficiently to prevent human subjects being recognized (an eye bar must not be used because of insufficient de-identification).

The editors reserve the right to reject manuscripts that do not comply with these requirements. The author will be held responsible for false statements or failure to fulfill these requirements.

For manuscripts reporting studies that involve human participants, a statement identifying the ethics committee that approved the study and confirmation that the study conforms to recognized standards is required, for example: **Declaration of Helsinki**; **US Federal Policy for the Protection of Human Subjects**; or **European Medicines Agency Guidelines for Good Clinical Practice**. It should also state clearly in the text that all persons gave their informed consent prior to their inclusion in the study.

Examples (not exhaustive)

Approval to conduct this human subjects research was obtained by the [name of institutional review board or ethics committee]. All procedures followed were in accordance with the ethical standards of the responsible committee on human experimentation (institutional and national) and with the Helsinki Declaration of 1975, as revised in 2000. Informed consent was obtained from all patients for being included in the study.

The study was approved by the [name of institutional review board or ethics committee]. No informed consent was required from subjects as data were anonymously extracted from the [name of system]. All procedures followed were in accordance with US Federal Policy for the Protection of Human Subjects.

This study was approved by and conducted according to the ethical standards of the [name of institutional review board or ethics committee]. All applicable international, national, and/or institutional guidelines were followed. This study was approved by the IRB after expedited review and was granted an informed consent waiver.

This study was conducted in accordance with all guidelines set forth by the [name of institutional review board or ethics committee]. Informed consent for genetic testing was obtained from all individuals undergoing testing, and [name of institutional review board or ethics committee] waived authorization for use of de-identified aggregate data. Individuals or institutions who opted out of this type of data use were excluded.

This study was reviewed and granted an exemption by the [name of institutional review board or ethics committee]. All procedures followed were in accordance with the ethical standards of the responsible committee on human experimentation (institutional and national) and with the Helsinki Declaration of 1975, as revised in 2000. Implied informed consent was obtained for individuals who voluntarily completed the online survey and submitted their responses.

Approval to conduct this human subjects research was obtained by the [name of institutional review board or ethics committee]. All procedures followed were in accordance with the ethical standards of the responsible committee on human experimentation (institutional and national) and with the Helsinki Declaration of 1975, as revised in 2000. Informed consent was obtained from all patients for being included in the study.

The study was approved by the [name of institutional review board or ethics committee]. No informed consent was required from subjects as data were anonymously extracted from the [name of system]. All procedures followed were in accordance with US Federal Policy for the Protection of Human Subjects.

This study was approved by and conducted according to the ethical standards of the [name of institutional review board or ethics committee]. All applicable international, national, and/or

institutional guidelines were followed. This study was approved by the IRB after expedited review and was granted an informed consent waiver.

This study was conducted in accordance with all guidelines set forth by the [name of institutional review board or ethics committee]. Informed consent for genetic testing was obtained from all individuals undergoing testing, and [name of institutional review board or ethics committee] waived authorization for use of de-identified aggregate data. Individuals or institutions who opted out of this type of data use were excluded.

This study was reviewed and granted an exemption by the [name of institutional review board or ethics committee]. All procedures followed were in accordance with the ethical standards of the responsible committee on human experimentation (institutional and national) and with the Helsinki Declaration of 1975, as revised in 2000. Implied informed consent was obtained for individuals who voluntarily completed the online survey and submitted their responses.

If any identifying information about participants is included in the article, the following sentence should also be included:

Informed consent was obtained from all participants for which identifying information is included in this article.

Authors do not need to provide a copy of the consent form to the publisher; however, in signing the author license to publish, authors are required to confirm that consent has been obtained. Wiley has a **standard patient consent form available** for use.

- ***Animal Studies***

The JOURNAL does not publish non-human animal studies. To affirm that this is the case for your submission, please include the following sentence under this subheading in the manuscript:

No non-human animal studies were carried out by the authors for this article

- ***Data Availability Statement***

Authors are required to provide a data availability statement to describe the availability or the absence of shared data. When data have been shared/are available in a repository, authors are required to include in their data availability statement a link to the repository they have used/created, and to cite the data they have shared. Whenever possible the scripts and other artefacts used to generate the analyses presented in the paper should also be publicly

archived. If sharing data compromises ethical standards or legal requirements then authors are not expected to share it.

Sample statements are **available here from Wiley Author Services**.

Example with data repository

Data Availability Statement

*Data collected for this study are available through the publicly available repository MINDS@Uw. The data file is available at **http://digital.library.wisc.edu/1793/78637***

Citation in Reference List

*Donahue, A., Hall, A., & Petty, E. (2018). Genetic counselor information needs & current library services for genetic counselor. [Data file] Retrieved from **http://digital.library.wisc.edu/1793/78637***

Although it would be rare for a paper submitted to the JOURNAL to report novel nucleotide sequence data, should that be the case, the novel nucleotide sequence data including genetic mutations must be submitted to a public database prior to publication and a sentence naming the database should be included in the manuscript.

References

The accuracy of references is the responsibility of the authors. The JOURNAL **strongly prefers references that have undergone peer review** and are not conference abstracts, unpublished masters' theses, unpublished dissertations, unpublished data in manuscripts, etc. However, if essential to the manuscript, they should be cited and referenced appropriately.

References should be prepared according to the *Publication Manual of the American Psychological Association* (6th edition). The APA website includes a range of **resources for authors** learning to write in APA style, including **an overview** of the manual, **free tutorials** on APA Style basics, and an **APA Style Blog**. For more information about APA referencing style, please also refer to the **APA FAQ**.

EndNote users can download the style **here**.

In-text citation

- follow the author-date method whereby the author's last name and the year of publication for the source should appear in the text, for example, (Jones, 1998)
    - publications with no authors, use brief phrase to describe publication and year of the publication, for example, ("Report of 1979 Business Meeting", 1979, p. 1) or ("NSGC Professional Status Survey: Executive Summary", 2020)
    - publications with no dates, use n.d., for example, ("American board of Genetic Counseling, Mission and History", n.d.)
- Multiple citations should be listed alphabetically by author's last name

Personal communications

- cite within the text as (Name of person providing the communication, personal communication, Date of communication), for example (Jane Doe, ABCD Executive Director, personal communication, September 2019)
- do not include in the reference list
- permission in writing from the communicator is required. Submit written permission with manuscript.

Reference List

- alphabetical by last name of first author
- the reference list is not numbered

General Comments

- Digital Object Identifier (DOI) should be provided for all references where available
- For journal articles, issue numbers are not included unless each issue in the volume begins with page one.
- For references with more than seven author names list first six with three dots and then last author name.

Reference examples

- *Journal article with 7 or fewer authors*

    - Beers, S. R., & De Bellis, M. D. (2002). Neuropsychological function in children with maltreatment-related posttraumatic stress disorder. *The American Journal of Psychiatry, 159,* 483–486. *doi:*1176/appi.ajp.159.3.483

- *Journal article with more than 7 authors*
    - Reuter, C. M., Kohler, J. N., Bonner, D., Zastrow, D., Fernandez, L., Dries, A., ... Wheeler, M. T. (2019). Yield of whole exome sequencing in undiagnosed patients facing insurance coverage barriers to genetic testing. *Journal of Genetic Counseling, 28,* 1107-1118. doi: 10.1002/jgc4.1161

- *Book with 7 or fewer authors*
    - Bradley-Johnson, S. (1994). *Psychoeducational assessment of students who are visually impaired or blind: Infancy through high school* (2nd ed.). Austin, TX: Pro-ed.

- *Book with 8 or more authors*
    - Gilbert, J. R., Smith, J. D., Johnson, R. S., Anderson, A., Plath, S., Martin, G., . . . White, N. (2014*). Choosing a title* (2nd ed.). New York, NY: Unnamed Publishing.

- *Internet Document*
    - Norton, R. (2006, November 4). How to train a cat to operate a light switch [Video file]. Retrieved 4/20/2020 from http://www.youtube.com/watch?v=Vja83KLQXZs
    - Note: only include the retrieval date if the content is likely to change

- *Publicly available data repository*
    - Donahue, A., Hall, A., & Petty, E. (2018). Genetic counselor information needs & current library services for genetic counselor. [Data file] Retrieved from http://digital.library.wisc.edu/1793/78637

- *Newsletter/newspaper, no author*
    - Report of 1979 Business Meeting of the NSGC. (1979). *Perspectives in Genetic Counseling*, 1(4), 1. Retrieved from nsgc.org [members only access]

- *Newsletter/newspaper, author*
    - Smith, A.C.M. (1982). "The sermon on the amount:" The status of NSGC finances. *Perspectives in Genetic Counseling*, 4(1), 2. Retrieved from nsgc.org [members only access]

- NSGC Professional Status Survey
    - NSGC Professional Status Survey: Executive Summary (2020). p. 2. Retrieved from https://www.nsgc.org/page/whoaregeneticcounselors

Figure Legends

Every figure must have a legend that includes the figure number and figure title. Legends should be concise but comprehensive – the figure and its legend must be understandable without reference to the text. The figure legend should Include definitions of any symbols used and define/explain all abbreviations and units of measurement.

Additional Files

Tables

Tables should be self-contained and complement, not duplicate, information contained in the text. The table must be understandable without reference to the text.

Tables should:

- be numbered and referred to by number in the text.
- have a brief explanatory title,
- have a concise but comprehensive legend in the case where additional explanation is provided using footnotes. Footnotes should be indicated by superscript lowercase letters
- define all abbreviations in the legend.
- be supplied as editable files, not pasted as images
- be uploaded as separate file(s)

Figures

Authors are encouraged to send the highest quality figures possible. Line art should be exported at 600 dpi or higher, and halftone images should be exported at 300 dpi or higher.

Figures should:

- be numbered and referred to by number in the text
- be clearly labeled
- be uploaded as separate file(s)

Color figures. Color figures may be published online free of charge.

Supporting Information

Supporting information is information that is not essential to the article, but provides greater depth and background. It is hosted online and appears without editing or typesetting. It may include copies of surveys or interview questions, consent forms, tables, figures, videos, datasets, etc.

Click here for Wiley's FAQs on Supporting Information.

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5. EDITORIAL POLICIES AND ETHICAL CONSIDERATIONS

Peer Review and Acceptance

The acceptance criteria for all papers are the quality and originality of the research and its significance to JOURNAL readership and the practice and discipline of genetic counseling. Papers will only be sent to review if the Editors determine that the paper meets the appropriate quality and relevance requirements.

Except where otherwise stated, manuscripts are single-blind peer reviewed. Wiley's policy on the confidentiality of the review process is **available here.**

Pre-Print Policy

The JOURNAL will consider for review articles previously available as preprints. Authors may also post the **submitted version** of a manuscript to a preprint server at any time. Authors are requested to update any pre-publication versions with a link to the final published article.

Revisions

When submitting a revised manuscript please also submit:

- Response to reviewer comments in the form of a table with reviewers' suggestions on the left-side and edits made/how addressed on the right-side
- A marked version (tracked, highlighted, etc.) and unmarked version of revised manuscript

Changes in Authorship

Prior to submitting the manuscript all authors should agree on the order in which their names will be listed in the manuscript. Any changes to authorship, including adding, removing or rearranging the authorship list, must be made before the manuscript has been accepted and only with approval from the Editor. The corresponding author is expected to write to the Editor with the reason for the change and will complete an Authorship Change Request form with written agreement from ALL authors, including those affected by any change. Only in exceptional circumstances will the Editor consider changes in authorship after a paper has been accepted. If a request for change in authorship is submitted after acceptance but before publication, the article will be suspended from proceeding to publication until the authorship

change request has been resolved. Once an article has been published in an online issue, a completed Authorship Change Request form and a corrigendum will need to apply to reflect any allowed changes in authorship.

Decision Appeals

Appeals should be filed within 28 days of notification of the decision. The appeal should be in the form of a letter addressed and submitted to the Journal of Genetic Counseling Editorial Office at JOGC@wiley.com. The letter should include clear and concise grounds for the appeal, including specific points of concern. The appeal will then be assessed by the Journal of Genetic Counseling management team, led by the Editor-in-Chief, and informed by the subsequent editorial communications. You will be informed of the outcome of the appeal in writing, normally within 28 days. The decision will be final.

Clinical Trial Registration

The JOURNAL requires that clinical trials are prospectively registered in a publicly accessible database and clinical trial registration numbers are included in all papers that report their results. Authors are asked to include the name of the trial register and the clinical trial registration number at the end of the Abstract. If the trial is not registered, or was registered retrospectively, the reasons for this should be explained.

Research Reporting Guidelines

Accurate and complete reporting enables readers to fully appraise research, replicate it, and use it. Authors are encouraged to adhere to recognized research reporting standards. The EQUATOR Network collects more than 370 reporting guidelines for many study types, including for:

- Randomized trials: CONSORT
- Observational studies: STROBE
- Systematic reviews: PRISMA
- Qualitative research: COREQ
- Quality improvement studies: SQUIRE
- Study protocols: SPIRIT
- **Studies reporting on genetic counseling as an intervention should follow the reporting standards found here:** Standards for the Reporting of Genetic Counseling Interventions in Research and Other Studies (GCIRS)

Publication Ethics

The JOURNAL is a member of the **Committee on Publication Ethics (COPE)**. Read Wiley's Top 10 Publishing Ethics Tips for Authors **here**. Wiley's Publication Ethics Guidelines can be found **here**.

Return to Guideline Sections

6. AUTHOR LICENSING

If a paper is accepted for publication, the author identified as the formal corresponding author will receive an email prompting them to log in to **Author Services**, where via the Wiley Author Licensing Service (WALS) they will be required to complete a copyright license agreement on behalf of all authors of the paper. This email will be issued within a few days of paper acceptance.

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If the Hybrid Open Access option [requires payment] is not selected the corresponding author will be presented with the copyright transfer agreement (CTA) to sign. The terms and conditions of the CTA can be previewed in the samples associated with the **Copyright FAQs**.

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Note to NIH, The Wellcome Trust and the Research Councils UK Grantees

Pursuant to NIH mandate, Wiley will post the accepted version of contributions authored by NIH grant-holders to PubMed Central upon acceptance. This accepted version will be made publicly available 12 months after publication. Please click **here** for further information. If you select the Hybrid Open Access option and your research is funded by The Wellcome Trust or the Research Councils UK (RCUK) you will be given the opportunity to publish your article under a CC-BY license supporting you in compliance with The Wellcome Trust and Research Councils UK requirements.

- *Self-Archiving Definitions and Policies*

Note that the JOURNAL'S standard copyright agreement allows for self-archiving of different versions of the article under specific conditions. Please click **here** for more detailed information about self-archiving definitions and policies.

Return to Guideline Sections

7. PUBLICATION PROCESS AFTER ACCEPTANCE

Accepted Articles

All accepted manuscripts are subject to editing. Authors have final approval of changes prior to publication.

Proofs

Once the paper is typeset, the author will receive an email notification with full instructions on how to provide proof corrections.

Please note that the author is responsible for all statements made in their work, including changes made during the editorial process – authors should check proofs carefully. Note that proofs should be returned within 48 hours from receipt of first proof.

Publication Charges. There are no publication charges for the Journal of Genetic Counseling.

Color figures. Color figures may be published online free of charge.

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8. POST PUBLICATION

Access and Sharing

Early View

The JOURNAL offers rapid publication via Wiley's Early View service. **Early View** (Online Version of Record) articles are published on Wiley Online Library before inclusion in an issue. Note that there may be a delay after corrections are received before your article appears online, as Editors also need to review proofs. Once your article is published on Early View no further changes to your article are possible. Your Early View article is fully citable and carries an online publication date and DOI for citations.

When the article is published online:

- The author receives an email alert (if requested).
- The link to the published article can be shared through social media.
- The author will have free access to the paper (after accepting the Terms & Conditions of use, they can view the article).
- The corresponding author and co-authors can nominate up to ten colleagues to receive a publication alert and free online access to the article.

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To find out how to best promote an article, click **here**.

Measuring the Impact of an Article

Wiley also helps our authors measure the impact of their research through specialist partnerships with **Kudos**) and **Altmetric.**

Return to Guideline Sections

9. WILEY AUTHOR RESOURCES

Wiley Author Resources

- *Manuscript Preparation Tips*

Wiley has a range of resources for authors preparing manuscripts for submission available **here**. In particular, authors may benefit from referring to Wiley's best practice tips on **Writing for Search Engine Optimization**.

- *Editing, Translation, and Formatting Support*

Wiley Editing Services can greatly improve the chances of a manuscript being accepted. Offering expert help in English language editing, translation, manuscript formatting, and figure preparation, Wiley Editing Services ensures that the manuscript is ready for submission.

- *Video Abstracts*

A video abstract can be a quick way to make the message of your research accessible to a much larger audience. Wiley and its partner Research Square offer a service of professionally produced video abstracts, available to authors of articles accepted in this JOURNAL. You can learn more about it by **clicking here**. If you have any questions, please direct them to videoabstracts@wiley.com.

10. EDITORIAL OFFICE CONTACT DETAILS

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3 Section Three: Critical Appraisal

Word count – 3999

(Excluding title page., references, figures, tables and appendix)

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This critical appraisal explores how the findings of the systematic literature review and the research paper relate to each other and in doing so illuminates the lived experience of growing up in a family with HD and the experience of maintaining psychological wellbeing for those living at-risk. I also explore quality appraisal, clinical implications, the value of reflexivity, offer some personal reflections on the research process and analysis methods used and end with suggestions for future research.

Overview of the findings

When considering the findings of the review and the research paper, the review presents four interrelated themes, three that name HD as a thief (thief of relationships, thief of self, thief of transparency) and the fourth that refers to the ways people tried to reclaim life from HD (search for reclamation). The research paper presented three interrelated themes: (1) 'you're constantly in limbo': living in two worlds, (2) "I have to live, just bloody live": managing the possibility of a time limited lifespan and (3) "Is that who I am, is that what I am?": the exhausting quest to be seen as an individual first. Within the research paper interesting parallels emerged in the narratives of participants with the findings of the review in terms of HD being a thief, however, the idea of HD as a thief was not discussed within the research paper due to the focus being on the challenges in maintaining psychological wellbeing. Participants spoke about the confusion of who had parented them in the research paper, whether it was HD or their parent and what behaviour they could assign to the person or the disease. This was also found within the thief of relationships theme in the review. The main difference between the young people in the review and the participants in the research paper in the context of HD stealing relationships was that young people reported a feeling of lacking a social network whereas eleven of the twelve participants in the research paper reported having an established social support network of friends and family. This may be due to the participants of the research paper being independent adults and the young people within the review being in the family home and

education at the time of life that they were discussing. Another explanation could be that most participants in the research paper described a late onset within their family compared to the young people in the review being aware of onset as young people.

Another similarity could be seen between the review and the empirical paper regarding the secrecy involved with HD. In the review HD stole transparency and instigated secret keeping where families would decide to keep their knowledge of HD to themselves. Some young people chose not to tell their friends or some of their friends and this was seen in the empirical paper where adults chose to have separate groups of friends to spend time with, i.e., those who knew about HD and those who did not. In the empirical paper this enabled a movement between the HD and non-HD world whereas for the participants in the review, this secrecy seemed driven by a different motive, perhaps self-protection and the desire to be seen as having a disease independent identity.

Quality Appraisal

The thirteen papers included within the literature review were quality appraised using the Critical Appraisal Skills Programme (CASP), a generic quality appraisal tool. Although the academic community has an agreed broad consensus concerning what makes qualitative research high quality, there is not a singular agreed nor established tool for such use (Sandelowski & Barroso, 2002). The CASP was selected due to the guidance produced by Cochrane on producing reliable systematic reviews (Harris, 2011) and its recommendation for use within social and health care related fields (Hannes & Macaitis, 2012). The length of the tool and ease of utilisation were additional contributory factors. As the review themes were present in each of the thirteen papers assessed as being of reliable quality, this suggests the findings have not been compromised by my interpretation. While validity is a debated word/concept within approaches to ensuring quality in qualitative research (Leung, 2015), further support of the findings could be formed through the consideration of each study's

theoretical, ontological and epistemological framework as suggested by Long et al. (2020). As the CASP is the least sensitive tool on interpretative and theoretical validity, when compared to the Evaluation Tool for Qualitative Studies (Long & Godfrey, 2004) for example, it would be useful if more sensitive tools were considered for use in future qualitative syntheses.

Clinical implications and Future research

The review and research paper contribute to the understanding of psychological distress experienced by young people in HD families and adults living at-risk of HD. Both the review and the research paper highlight the need for support for these groups. The findings suggest that there is not only a need for individual support to be developed but also for family support. For families, there is a need for all areas of the HD journey to be explored within professional settings including the impact of the disease on familial relationships, communication of genetic risk and external sources of support. For individuals there is a need for knowledgeable and accessible information and intervention provision for people affected by HD. This is important considering that current research surrounding psychological interventions for those with the expanded HD gene is limited (Zarotti et al., 2020). Both the review and the research paper add knowledge and suggestions to this area.

Throughout the review and the empirical paper there seemed to be a weight of negativity when HD and its impact was discussed. Indeed, when reading the literature on HD and the lack of cure or effective treatments, coupled with the devastating losses and psychological distress associated with the disease, it appears there is not much space for positivity. When considering HD, it is often the physical aspects that receive most focus and the psychological are widely ignored (Zarotti et al., 2020). This, however, must change in conditions such as HD where the medical interventions are limited and the psychological impact significant. What was apparent through my exploration of the literature and lived experience of HD was the creation of positivity and balance in the approaches by the young

people in the review and the participants in the empirical paper. In a systematic review (Ghosh & Deb, 2017) exploring the impact of positive psychological interventions in chronic illness, it was found that such interventions are accepted by people with health conditions but that the usefulness of these is inconclusive. Following the recommendations from the review to explore this potential usefulness further, a narrative review (Duncan et al., 2021) concluded that hope theory can add to positive change and quality of life for people with chronic illness. The review suggests that when professionals explore the person's hope and understand what their hope is and where it comes from, empathic connections and overcoming barriers could be easier.

People within the review and the empirical paper spoke of their hope for their lives and that of their families and the ways that such hope helped them look to a future and progress day by day. Using an approach such as hope theory would assist in the move away from traditional clinical psychology and psychiatry approaches that function on/re-enforce DSM labels and progress towards a focus on psychological qualities that bring growth. The PERMA (Seligman, 2011) model may be of use in locating hope in people experiencing the impact of HD.

The model suggests that five areas contribute to psychological wellbeing: positive emotion, engagement, relationships, meaning, accomplishments. While this offers a good basis for gaining an understanding of hope in the HD population, the model does not consider constructs that some participants in the empirical specifically mentioned such as religious beliefs, spirituality or security which need to be considered. It is important to note that the PERMA model within chronic illness literature has been used to focus on improving patient self-management and illness management strategies (Nikrahan et al., 2016; Schrank et al., 2012). This would not be as effective with HD and therefore the scope of the use of the model would need to be widened to explore the impact on the areas within day-to-day life outside of a management focus. A recent study has explored the use of audio-visual positive psychology

resources for parents of a child with epileptic encephalopathy (Nevin et al., 2022). Participants described that they found the resources normalised their emotional responses, offered ways to manage social and personal relationships and support seeking as well as accepting help from other people. The resources brought feelings of connectedness, reassurance, comfort and hope which are similar to those feelings desired by participants in the empirical paper and the young people in the review. It could be that producing such a resource specifically focused on life experiences with HD would be beneficial. Such a resource may also provide a useful tool while professionals increase their knowledge and support is developed.

Formulation may also offer a useful tool. Formulation is an in-depth clinical assessment approach used to understand psychological difficulties (Baird et al., 2017). Formulations are used to guide interventions by creating an understanding of life experiences, triggers for distress and how distress is maintained. This information can guide possible interventions (Sim et al., 2005). The process of formulation is collaborative and progressive and creates a dynamic understanding of a person as an individual and potentially the world around them that can be reformulated as understanding increases or situations change (Christofides et al., 2012).

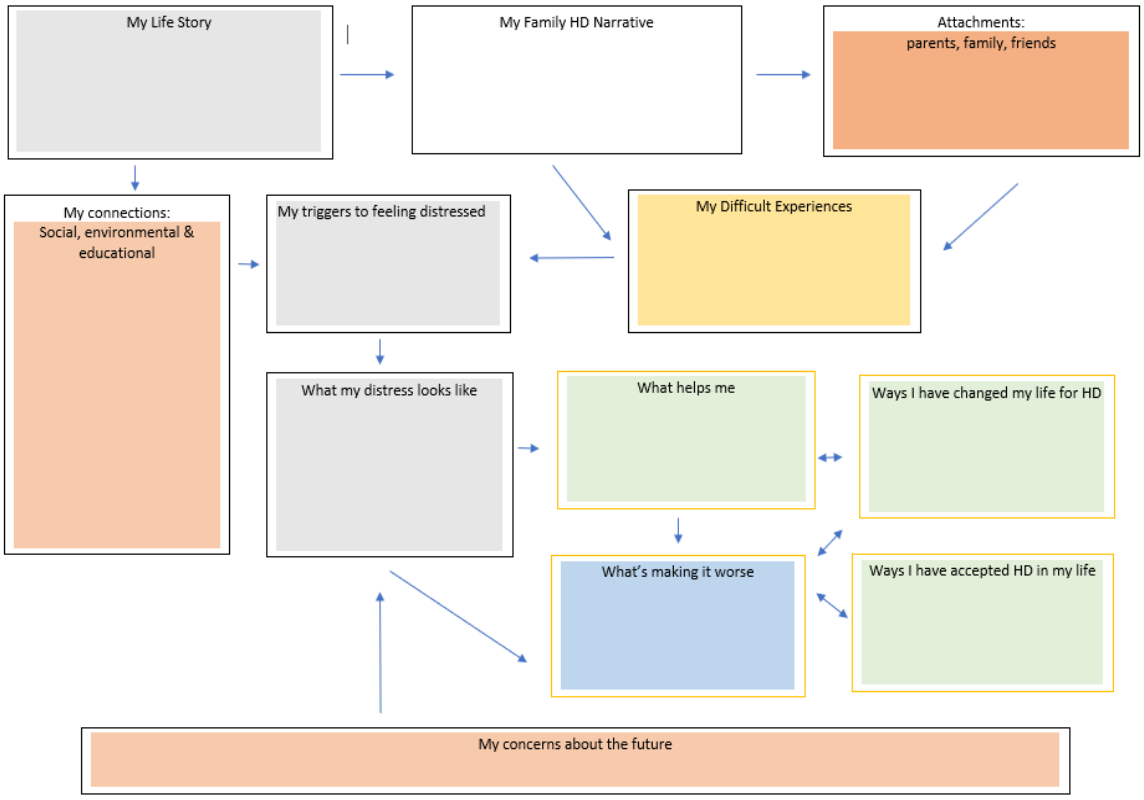
Using a clinical formulation model specifically created for understanding distress in the context of HD could be highly valuable in encapsulating the complexities the disease brings. Dale et al. (2022) recently presented such a tool for use with individuals with HD. The model explores experience, social impact, triggers, presentations of distress, HD narratives, symptoms and concerns about the future as separate yet interlinked sections.

This current model is aimed at use with adults, however this could be adapted as a formulation model for HD for young people. Figure 3 - A shows how this could be adapted for use with young people in HD families by altering the focus of sections of the model in line with the presented findings of this review. This approach would result in a person-centred, rather than family centred, intervention strategy. The use of the guided tool may also help professionals

with more limited HD knowledge learn about the disease from the perspective of those living with the effects of HD. This may also provide evidence for further intervention developments, such as peer support, which have been discussed as being lacking. The model would need further focus and refinement before being evaluated to discover what the next steps may be in development of effective interventions (Campbell et al., 2000; Craig et al., 2008).

Figure 3 - A

Clinical formulation to understand impact of HD on young people.



Reflexivity, Reflection and Journaling

The quality and validity of interpretative phenomenological analysis has been argued to be a matter of judgement (Smith, 2011) although reflexivity, reflection and journalling (Vicary et al., 2017) are three important components in ensuring quality is achieved. While there is no widely accepted single definition of reflexivity, it can be explained as the awareness

and examination of the researcher's beliefs, judgements and assumptions while conducting research and an awareness of how these may influence their research and interpretations. This means that the researcher needs to have an awareness of their position within the research and that the research and the researcher are in mutual existence with one another (D'cruz et al., 2007).

Reflexivity and awareness of the relationship between the research and researcher is significant for research that is qualitative or ethnographic (Davies, 2008). Though reflexivity is a crucial part of the IPA process it is not as simple as the definition would suggest and is often referred to in the literature as a way of being (Engward & Goldspink, 2020). On reading the guidance by Smith et al. (2021), I assumed that the process of IPA would be linear and progressive however as I became more practised at the method I realised that in taking such a linear and literal stance on the guidance, the hermeneutic and phenomenological point of IPA would be missed. This meant that using a journal to take notes throughout the process and notice the interlinks in my interpretations of participants' words and meanings was important within my own context so that I could lay my assumptions aside. To use IPA in a way that offers validity, credibility and transparency, it is important for researchers to be able to step outside of their 'environing world' (Heidegger & Gregory, 1998). This is the ability to step away from assumptions from cultural and historical environments so that they do not affect the exploration and subsequent interpretation of the participant's world.

I also found the creative and imagination aspects of IPA challenging. I am a creative person, but I felt a pressure to offer Grouped Experiential Themes that were totally reflective of the data and not to my own creative thinking. This resulted in a fear that allowing my own creative use of language may have brought my own assumptions into the data when I had tried so hard not to allow this seepage. I found the process of understanding and interpreting the data to take significantly longer and be far more complex than I thought. To maintain the quality

of the research and remain reflexive I employed several strategies to assist me. I discussed the formation of themes with my supervisors who were aware of the content of interviews and the process of formation from exploratory notes through to grouped experiential themes. In relation to sensitivity to context, the field supervisor brought extensive knowledge from her experiences in the HD field. As I used a journal throughout the research, I was able to explore and appreciate my own influence in how I collected and interpreted the data in both pieces.

An example from the reflective journal in response to the empirical research is below:

I am finding this journey far more emotional than I thought it would be. The absolute horror some of these people have experienced through people in the public, people like me, shaming them and judging them has deeply affected me. I am struck by how unjust their situation is. We as professionals have abandoned them, we as people judge them. I hope I have never made anyone feel the way these people have been made to feel by other humans. I feel such a deep pull to ensure that I present every aspect of their story, but I fear that the idea of this being a research piece with a word count will prevent such expression.

Reflection on recruitment of participants

Recruitment posters were initially shared via a social media HD support group ran by the experts by experience who aided the development of all information circulated to participants. Following the recruitment of 6 individuals via this pathway over a period, I decided to pursue the second recruitment option approved in the ethics application which was to recruit via a HD charity's social media page. Although recruitment was via two pathways I believe that sample homogeneity, as a key aspect of IPA research (Smith, 2017), was maintained due to both pathways to recruitment being social media and HD specific support groups and the specificity of the research question and inclusion criteria (Murray & Wilde,

2020). I chose to recruit via social media pathways due to feedback from the experts by experience that there were not many support services with psychological or support need awareness for people with experience of HD, a reflection which I found to be true in my own research (Dawson et al., 2004; Zarotti et al., 2020).

A total of 112 individuals responded to the recruitment poster. To recruit fairly I began working through the responses aligning with eligibility criteria in date order of who had responded first. I continued this process until I had recruited 18 participants who agreed to be interviewed. Of the people who expressed an interest that I had reviewed, 8 were too young to participate and 15 were not within the 12 months of knowing about their at-risk status.

I decided that 18 participants would be sufficient enough to allow for withdrawals and non-attenders and hoped that I would be able to interview between 10 and 12 individuals. Smith et al. (2021) suggest that smaller samples would offer more depth to understanding the explored phenomenon however there are no definitive numbers regarding participants for IPA (Smith & Eatough, 2007). I found that having 12 interviews supplied me with an overwhelming amount of data that provided much more experiential information than I expected. I could have used a smaller sample size which would have allowed a more individual focus on participants in this work (Collins & Nicolson, 2002). Despite this, I feel at peace with myself that I have been able to present the 12 participants' experiences of psychological wellbeing equally and fairly.

Reflections on philosophical underpinnings and methodology

My ontological stance requires clarification as this affects the decision concerning the methodology I used and the ways I gathered data, though IPA addresses both methodology and method. While I am undecided on a firm position, I would name my current ontological position as being within the critical realist paradigm. I state this as I seek understanding of phenomena by exploring what people experience within their context, that is I am interested in

the experience, the events and the underlying deeper level mechanisms (Fletcher, 2017). This stance is in line with IPA which openly acknowledges the involvement of the researcher and their active role in knowledge discovery, interpretation, and understanding (Smith et al., 1999). I am of the belief that through this research I can gain an understanding of people's experiences from a subjective standpoint, and this is line with the double hermeneutic engagement seen in IPA. I adopted a reflexive approach in which I could be present and responsive in the moment and value naturalistic methods such as semi-structured interviews; however, I acknowledge I cannot gain a full understanding of the participants' experiences. I also acknowledge that there are many ways of viewing and experiencing being in the world around oneself.

I chose to conduct a meta-ethnography for the review. This was due to my preference of working within guidance and being able to present the voice of the participants involved in research. While I followed Noblit and Hare (1988) I also used recent guidance on how to use meta-ethnography for the purpose of synthesising literature (Sattar et al., 2021). The guidelines provide clear step by step instructions with examples and follow the seven steps outlined by Noblit and Hare (1988) and this aided my understanding of how to work through the steps involved.

To report the meta-ethnography appropriately, I followed the eMERGe reporting guidance for meta-ethnography (France et al., 2019). The eMERGe guidance also offered instruction on the synthesis process which other guidance, such as the ENTREQ (Tong et al., 2012), did not seem to do. Another reason I wanted to conduct a meta-ethnography is that I wanted to produce a review with an outcome in the form of a new model. While I did not manage to accomplish this within the review, the review brought information that is needed in terms of understanding and being able to form such models to support young people living in HD families. I did not find in the review any refutational translations therefore this was not discussed though if this had been identified it would have been important to include.

I choose to work with IPA methodology in the research paper as I wanted to ensure that the voices of the participants were narrated clearly to present their experience. IPA can bring each participant's experience as it is idiographic and as the experience of this group is limited in research, IPA presented a wise choice. IPA offers a double hermeneutic approach meaning that I as the researcher am trying to make sense of how participants are making sense of their own experiences. This differs from other approaches such as discourse analysis, for example, which seek to learn how participants construct accounts of what is happening to them. IPA also offers guidance on the procedures and question formation (Larkin et al., 2006) of the research piece and I valued having such guidance to follow throughout the empirical research. I also found the examples given within the text on how to conduct IPA extremely helpful in aiding my understanding of the processes involved. This was the main reason I chose to follow IPA guidance from Smith et al. (2021) rather than other authors such as Murray and Wilde (2020).

I had considered the analysis method of thematic analysis as I had used this before and felt comfortable with the stages involved. However, I further reflected that thematic analysis would not bring the voice to the participants in the way that IPA would even though it is likely that the themes produced could have been very similar, though perhaps more descriptive and less interpretative than in IPA. In my view, the thematic approach to aggregate findings would miss important experiential details that can be discovered and explored within IPA.

IPA acknowledges that interpretating what participants have brought to interviews could be susceptible to being viewed through the experiences of the researcher responsible for providing such interpretation. I valued IPA's recognition of the researcher's position and how open the approach is to exploring this.

I feel that both IPA and meta-ethnography fit with my own approach to knowledge and the value I place on understanding people's experience being crucial to exploring their needs and offering the right support. I am comfortable knowing that there is much I do not know and

that I can never fully understand the experience of another person. The way I learn is via other people's experiences of knowledge and the sense they make of those experiences. I find this helps me understand a person more than any historical chronology in most instances.

Conclusion

This thesis explored the lived experience of young people growing up in a family with HD and the experience of managing psychological wellbeing of adults living at-risk of HD. There were similarities in the themes presented within the review and the empirical paper as discussed. The use of a reflective journal helped to manage my thoughts as I noticed these similarities. The review and research found that professional understanding and support for young people in HD families and adults living at-risk of HD is lacking despite these experiences leading to high levels of distress and difficulty. Both groups had formed their own strategies to live life to the best of their ability when professionals were perceived to have failed them. Clinical psychologists and professionals are required to bring their knowledge, skills and willingness to explore how access to support and provision for people affected by HD can be improved. People experiencing such a traumatic disease with wide systemic impact are currently being failed by our care system; this cannot continue.

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4 Section Four: Ethics

Word count – 4851

(Excluding title page, references, and appendix)

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Prepared for: Journal of Genetic Counselling

Faculty of Health and Medicine Research Ethics Committee (FHMREC)

Lancaster University

Application for Ethical Approval for Research

Title of Project: Challenges in maintaining psychological wellbeing when living at risk of Huntington's Disease

Name of applicant/researcher: Hollie Cooper

ACP ID number (if applicable)*: **Funding source (if applicable)**

Grant code (if applicable):

***If your project has *not* been costed on ACP, you will also need to complete the Governance Checklist [link].**

Type of study

☐ Involves existing documents/data only, or the evaluation of an existing project with no direct contact with human participants. **Complete sections one, *two* and four of this form**

☒ Includes *direct* involvement by human subjects. **Complete sections one, *three* and four of this form**

SECTION ONE

1. Appointment/position held by applicant and Division within FHM Trainee Clinical Psychologist, DClinPsy

2. Contact information for applicant:

E-mail: h.cooper3@lancaster.ac.uk **Telephone**: 07771542664

Address: Trainee Clinical Psychologist, Lancaster University, Lancaster, LA1 4YT

3. **Names and appointments of all members of the research team (including degree where applicable)**

Hollie Cooper Trainee Clinical Psychologist

Dr Fiona Eccles, Research Supervisor, Lecturer, Lancaster University

Professor Jane Simpson, Field Supervisor, Professor, Lancaster University

Dr Maria Dale, Field Supervisor, Clinical Psychologist, Leicester NHS

3. If this is a student project, please indicate what type of project by marking the relevant box/deleting as appropriate: (please note that UG and taught masters projects should complete **FHMREC form UG-tPG**, following the procedures set out on the FHMREC website

PG Diploma ☐ Masters by research ☐ PhD Thesis ☐ PhD Pall. Care ☐

PhD Pub. Health ☐ PhD Org. Health & Well Being ☐ PhD Mental Health ☐ MD ☐

DClinPsy SRP ☐ [if SRP Service Evaluation, please also indicate here: ☐] DClinPsy Thesis ☒

4. Project supervisor(s), if different from applicant: Dr Fiona Eccles, Dr Maria Dale, Professor Jane Simpson

5. **Appointment held by supervisor(s) and institution(s) where based (if applicable)**:

Dr Fiona Eccles, Research Supervisor, Lecturer, Lancaster University

Professor Jane Simpson, Field Supervisor, Professor, Lancaster University

Dr Maria Dale, Field Supervisor, Clinical Psychologist, Leicester NHS

SECTION TWO

Complete this section if your project involves existing documents/data only, or the evaluation of an existing project with no direct contact with human participants

1. Anticipated project dates (month and year)

Start date: End date:

2. Please state the aims and objectives of the project (no more than 150 words, in lay-person's language):

Data Management

For additional guidance on data management, please go to Research Data Management *webpage, or email the RDM support email:* rdm@lancaster.ac.uk

3. Please describe briefly the data or records to be studied, or the evaluation to be undertaken.

4a. How will any data or records be obtained?

4b. Will you be gathering data from websites, discussion forums and on-line ‘chat-rooms’ n o

4c. If yes, where relevant has permission / agreement been secured from the website moderator? n o

4d. If you are only using those sites that are open access and do not require registration, have you made your intentions clear to other site users? n o

4e. If no, please give your reasons

5. What plans are in place for the storage, back-up, security and documentation of data (electronic, digital, paper, etc)? Note who will be responsible for deleting the data at the end of the storage period. Please ensure that your plans comply with General Data Protection Regulation (GDPR) and the (UK) Data Protection Act 2018.

6a. Is the secondary data you will be using in the public domain? n o

6b. If NO, please indicate the original purpose for which the data was collected, and comment on whether consent was gathered for additional later use of the data.

Please answer the following question *only* if you have not completed a Data Management Plan for an external funder

7a. How will you share and preserve the data underpinning your publications for at least 10 years e.g. PURE?

7b. Are there any restrictions on sharing your data?

8. Confidentiality and Anonymity

a. Will you take the necessary steps to assure the anonymity of subjects, including in subsequent publications? yes

b. How will the confidentiality and anonymity of participants who provided the original data be maintained?

9. What are the plans for dissemination of findings from the research?

10. What other ethical considerations (if any), not previously noted on this application, do you think there are in the proposed study? How will these issues be addressed?

SECTION THREE

Complete this section if your project includes *direct* involvement by human subjects

1. Summary of research protocol in lay terms (indicative maximum length 150 words):

This thesis aims to explore the challenges faced in keeping psychologically well by people who are at risk of the genetically inherited, neurodegenerative disease, Huntington's disease (HD). Being at risk means that they are aware of HD in their family and may inherit HD but have not had the genetic test themselves and currently have no symptoms.

Within current research there is a focus on people who have been tested for HD and have received a positive test result but little focus on those people who are undiagnosed or known as living at risk. Little is known about the needs of this group, especially in the areas relevant to this thesis (how they stay well, what challenges they face and what support they need)).

It is hoped that this piece of qualitative work will bring some understanding of the needs of this group and their coping strategies, self-management techniques and other things they may do to maintain psychological wellbeing as well as challenges faced. This can add direction to what psychologists and health care clinicians/practitioners can provide.

2. **Anticipated project dates (month and year only**)

Start date: October 2021 End date: March 2023

Data Collection and Management

For additional guidance on data management, please go to Research Data Management *webpage, or email the RDM support email:* rdm@lancaster.ac.uk

3. Please describe the sample of participants to be studied (including maximum & minimum number, age, gender):

To be included in the research participants will need to be aged 18 and over (adult in the UK). Participants will need to be aware of their at-risk status and be able to communicate where their risk comes from in their biological family so that level of risk can be established (25 or 50%). Participants will need to have knowledge of their at-risk status for a minimum of 12 months. This is due to the suggested adjustment stage for a return of psychological consequences of this knowledge to return to baseline being 6 months (Tibben et al,. 1997) and the experience of living with the knowledge of being at risk for a further 6 months.

Participants must be motor symptom free, not have been tested for HD, not have any other significant health problems and able to withstand an interview for up to 90 minutes or an interview that spans over two separate occasions to total up to 90 minutes.

Participation will be voluntary, and participants may choose to withdraw at any time without reason up to two weeks following the time of the interview. At this point data analysis has begun. Once analysis has begun it would be difficult to deduct individuals' responses from the emerging themes.

Smith et al., (2009) recommend an ideal number of participants for IPA to be between four and ten. Having between four and ten participants will allow an in-depth exploration of each

narrative enabling the highlighting of experiences and comparison. In order to be equipped for individuals who may decide to withdraw I will recruit between eight and twelve individuals.

4. How will participants be recruited and from where? Be as specific as possible. Ensure that you provide the *full versions* of all recruitment materials you intend to use with this application (eg adverts, flyers, posters).

Participants will be recruited via a social media groups on Facebook, EXTRACTED, ran by the involved experts by experience. A digital poster will be shared within the group by the administrator of the group asking people who meet the inclusion criteria and how would like to know more to contact me directly using the Lancaster university email address: h.cooper3@lancaster.ac.uk. Participants will then be emailed the information pack and given time to make an informed decision regarding participation in the research.

If enough participants cannot be recruited via social media (Facebook), then the EXTRACTED will be contacted for recruitment via their website. Contact methods will remain the same for both avenues. As a third recruitment option, if the first two avenues do not result in the required participants, a bespoke Twitter account will be created to aid recruitment.

5. Briefly describe your data collection and analysis methods, and the rationale for their use.

Interpretative Phenomenological Analysis (IPA) will be used as it is an established qualitative method with an idiographic focus that allows the consideration of individual lived experiences in informing meaning (Smith, Flowers & Larkin, 2009).The two main aims of IPA are to

examine in detail how an individual has made sense of their experiences and to offer an interpretation of the experience to aid understanding of it. The main advantage of using IPA is that it enables the data to provide insight into complicated interrelationships and patterns within experience in a specific context. The research will be qualitative and collected via semi-structured interviews with participants. Data will be collected that shares the participants experiences of maintaining their own psychological wellbeing once they had learned of and adjusted to (12 months post) their at-risk status. Following the completion of each interview the narrative will be transcribed and any identifying factors removed from transcript where possible to do so without affecting the information. IPA guidelines by Smith et al., (2009) and Nizza et al., (2021) will be followed for analysis and creation of categories. Each transcript will be analysed line by line, keeping awareness of similarities and differences and any emerging themes. The transcripts will be read individually and coded, reread multiple times before collating themes that are consistent across the narrative. Any quotes that are to be used within the writing up of the research will be anonymised though basic demographic information may be included.

6. What plan is in place for the storage, back-up, security, and documentation of data (electronic, digital, paper, etc.)? Note who will be responsible for deleting the data at the end of the storage period. Please ensure that your plans comply with General Data Protection Regulation (GDPR) and the (UK) Data Protection Act 2018.

Participants will be informed of what will happen with their information and how it will be stored. Any information shared with the researcher and any interviews conducted will remain confidential unless risk is disclosed. Interviews, transcripts, and notes will be encrypted and

stored on the researchers Lancaster University H drive and OneDrive (both of which are password protected) on the researcher's password protected computer.

Participants will be made aware that Lancaster University's DClinPsy Research Coordinator will keep a copy of consent forms, audio consent recordings and transcriptions for 10 years following the completion of the research. The responsibility of the research in this location is overseen by Dr Fiona Eccles, Research Supervisor.

Microsoft teams will be used for interviews which will be recorded. Microsoft Teams has a detailed security and compliance section stating that data is encrypted at transit and rest and is accessed via two-factor authentication (https://docs.microsoft.com/en-us/microsoftteams/security-compliance-overview). Participants may choose to have cameras on or off for the recording.

7. Will audio or video recording take place? ☐ no ☒ audio ☒ video

a. Please confirm that portable devices (laptop, USB drive etc) will be encrypted where they are used for identifiable data. If it is not possible to encrypt your portable devices, please comment on the steps you will take to protect the data.

All saved data of any kind will be stored using encryption via the Lancaster University OneDrive access via authentication and password protected computer.

b What arrangements have been made for audio/video data storage? At what point in the research will tapes/digital recordings/files be destroyed?

Participants will be made aware that Lancaster University will keep a copy of consent forms and audio consent recordings and transcriptions for up to 10 years following the completion of the research. Original analysis of transcripts may also be kept in case of benefit in aiding future

publications. The responsibility of the research in this location is overseen by Dr Fiona Eccles, research supervisor. All recordings will be destroyed once the thesis has been examined.

Please answer the following questions *only* if you have not completed a Data Management Plan for an external funder

8a. How will you share and preserve the data underpinning your publications for at least 10 years e.g. PURE?

It is not intended that the raw data from this research is shared. The transcripts produced from participants interviews will be in the narrative style of the participant and will be anonymised as much as possible. Despite this it is possible that when the transcript is read in its entirety, confidentiality could be compromised.

8b. Are there any restrictions on sharing your data ?

Raw data will not be available to people not working on the proposed study.

9. Consent

a. Will you take all necessary steps to obtain the voluntary and informed consent of the prospective participant(s) or, in the case of individual(s) not capable of giving informed consent, the permission of a legally authorised representative in accordance with applicable law? yes

b. Detail the procedure you will use for obtaining consent?

Participants who respond to the social media digital poster by emailing h.cooper3@lancaster.ac.uk will be emailed an information pack. Participants will then be given time to read the information and make an informed choice about whether they wish to continue. Participants will then be invited to discuss the interview process and the study and express any

worries and ask any questions. If participants wish to continue consent will be discussed. At this point participants will be asked to sign a consent form and email it back or send an email from their address stating that they have read and agreed to the statements on the consent form or alternatively asked permission to record audio consent. Participants will be given time to ask any questions and discuss any concerns about taking part in the research. Participants will be able to withdraw at any point without question up to the point of analysis of transcripts begins, which will be 2 weeks post interview. To ensure a transparent process, communication between the researcher and participants will be recorded in a table and saved on OneDrive and the universities H drive in an anonymous manner using encryption and a password protected computer until the study ends. Participants will also be asked at the start of each interview to confirm they understand the consent form, that they give consent and are happy to continue to the interview questions.

10. What discomfort (including psychological eg distressing or sensitive topics), inconvenience or danger could be caused by participation in the project? Please indicate plans to address these potential risks. State the timescales within which participants may withdraw from the study, noting your reasons.

Due to the nature of the research question in asking participants to discuss their own current at-risk status, there is potential for distress to be experienced during the interviews. Attention will be brought to this possibility in the consent procedure and in the information pack volunteers receive. Participants will be asked if they have a source of support that they could draw on if needed following the interview and numbers of organisations will be provided. Participants will be informed that they may withdraw at any point (up to two weeks post interview when analysis begins) and that they may have a break or re-arrange their interview if preferred. The researcher will keep in mind awareness around distress and signs of upset in participants and manage this by checking with the participant whether they wish to continue,

take a break, rearrange or cancel participation. If there is immediate risk the local emergency services would be contacted. The researcher would seek advice from the project supervisors where needed.

11. What potential risks may exist for the researcher(s)? Please indicate plans to address such risks (for example, noting the support available to you; counselling considerations arising from the sensitive or distressing nature of the research/topic; details of the lone worker plan you will follow, and the steps you will take).

As interviews will be conducted via Microsoft Teams, there are no physical risks to the researcher. Contact with participants will be via email using the Lancaster University email address.

12. Whilst we do not generally expect direct benefits to participants as a result of this research, please state here any that result from completion of the study.

It is not thought that there would be any direct benefits to participants. It may be helpful for participants to know that the information they share will help inform knowledge and awareness in the provision of wellbeing whilst living at risk of HD. This information can inform therapeutic models by providing the current skills and areas of support needed.

13. Details of any incentives/payments (including out-of-pocket expenses) made to participants:

There are no incentives for participation

14. Confidentiality and Anonymity

a. Will you take the necessary steps to assure the anonymity of subjects, including in subsequent publications? yes

b. Please include details of how the confidentiality and anonymity of participants will be ensured, and the limits to confidentiality.

Participants will be informed of their right to withdraw at any stage in the interview and that the internet is not a secure source of communication, though all that can be done to protect participants identify and information will be done.

The researcher will be responsible for the transcription of interviews and these transcripts will be anonymised and have identifiable information removed as much as possible without affecting the analysis. Quotations that are used will be sections of the transcript and will contain only demographics alongside a pseudonym. All collected data will be stored on the university's secure server with access for the researcher and academic supervisor.

Personal information will remain confidential and anonymous unless there is an expression of risk. If risk is detected in the interviews advice will be sought from the supervisors or in the case of immediate risk, emergency services.

15. If relevant, describe the involvement of your target participant group in the *design and conduct* of your research.

There are two experts by experience involved in the study. The researcher has sought their input in terms of design, recruitment methods and poster and interview schedule construction. Feedback has also been given on the consent forms and information pack that participants will receive via email. Due to the feedback that the language in the information pack and consent form appeared clinical, changes were made to make the language appear warm without compromising the information that needed to be shared.

16. What are the plans for dissemination of findings from the research? If you are a student, include here your thesis.

The study will be written up to form part of the doctorate in clinical psychology thesis. Findings will be shared with participants who have declared that they would like to receive them. It is hoped that the findings will also be submitted to a peer reviewed journal and shared with the Leicestershire NHS HD service and presented at Lancaster University thesis presentation days. It is possible that findings will be shared at academic conferences, special interest groups and training events.

17. What particular ethical considerations, not previously noted on this application, do you think there are in the proposed study? Are there any matters about which you wish to seek guidance from the FHMREC?

None

SECTION FOUR: signature

Applicant electronic signature: Hollie Cooper Date

13.07.2021

Student applicants: please tick to confirm that your supervisor has reviewed your application, and that they are happy for the application to proceed to ethical review ☒

Project Supervisor name (if applicable): Dr Fiona Eccles Date application discussed

13.07.2021

Research Protocol for the project: Challenges in maintaining psychological wellbeing when living at risk of Huntington's disease

Applicants

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Introduction

Huntington's disease (HD) is a genetically inherited disease which is neurodegenerative caused by CAG triple repeat expansion mutation in the HD gene (MacDonald et al., 1993). HD was approximated as 1-4 cases per million (Harper, 1992) meaning the disease was classed as rare however more recent research has found there is an estimated 5700 people in the UK with HD (Evans et al., 2013) . HD progresses over time, causing motor dysfunction, cognitive decline (dementia), and psychological symptoms (Heiberg, 2008). Symptoms include impaired fine motor coordination, difficulty swallowing, abnormal physical movement, reduced language clarity, reduced word recall, and reduced speed of speech (Walker, 2007). The disease is debilitating in the later stages often leaving individuals unable to speak, wheelchair-bound, and experiencing severe involuntary movements (Roos, 2010). The most common cause of death is infections, mainly of the respiratory system, such as pneumonia (Di Maio et al., 1993). At present there is no cure for HD though disease modifying treatments are available (Dash & Mestre, 2020).

HD is an autosomal dominant disease meaning that children of a parent with the expanded gene have a 50% chance of inheriting the expanded gene (Rivera-Navarro et al., 2015). If the gene is inherited the person will experience HD, with onset usually between 30 and 50 years of age (Di Maio et al., 1993; Roos et al., 1991) when the person moves from a premanifest (movement symptom free) state to experiencing symptoms of the disease. The disease has a longevity of

approximately 10-20 years (Ready et al., 2011; Roos et al., 1991) meaning that once symptoms appear there is approximately 10-20 years until the individual dies (Harper, 1996).

Individuals at risk (those with a biological relative with HD, e.g., parent, grandparent, sibling) who are aged 18 and above, can request a genetic test to see if they have the HD gene expansion before symptoms occur. The test has been available via linkage since 1986 (Kremer et al., 1994) though it is now used to identify the specific CAG triple-expansion mutation which has been available since 1993 (Bernhardt et al. 2009), The predictive genetic test for HD is not a diagnostic test or assessment and is undertaken pre-manifest (carried out before movement symptoms occur). A diagnostic genetic test can be carried out post symptom discovery (Craufurd et al., 2015). Testing allows the at-risk status to be abandoned if a negative result is received.

Research indicates that uptake in genetic testing for HD is low, with over 80% of those at-risk choosing not to take the test (Baig et al., 2016). There are international guidelines for the support of individuals considering predictive genetic testing for HD, including providing genetic counselling, psychosocial assessment, and evaluations to receive psychological support in deciding to have the test (MacLeod et al., 2013). This non-directive approach aims to aid individuals in deciding whether genetic testing for HD is a procedure they wish to opt for (Elwyn et al., 2000). However, the provision of this service and ability to meet the guidelines for psychological support remain inconsistent due to funding and access to qualified genetic counsellors, meaning that individuals who opt in for testing are often unsupported and misunderstood (Hawkins et al., 2013). Furthermore, research into the area of psychological needs of those who opt out of testing is limited and HD services are usually inaccessible to individuals who opt out of testing and do not have symptoms or diagnosis of HD (Etchegary, 2011). When regular clinical services, such as the GP and local counselling services, are

accessed knowledge that health care providers have of HD is too poor to enable appropriate support (Skirton et al., 2010).

Research on the decision-making processes around test taking has indicated that individuals considering testing are more likely to experience low self-esteem, irritability and increased apathy than those not considering test-taking (Quaid et al., 2017). One study reported that some individuals had carried out a self-analysis of their own makeup to decide whether they perceive themselves to be a 'strong person' with ability to cope with the test results. Interestingly, not all those who concluded they could cope took the test, and not all those who decided that they were not strong enough chose not to take the test (Taylor, 2004). It was found that those who decided not to take the test used more avoidant coping skills than those who requested testing (Binedell et al., 1998) and that the tolerance of uncertainty was directly linked to ability to cope for non-testers.

Whilst this research holds significant value, it does not offer depth or understanding of the at-risk group and their experiences of psychological wellbeing outside of the decision-making process in test taking. Research by Chisholm et al. (2013) compared individuals who were at-risk, pre-manifest, and symptomatic for HD in the USA. Findings showed that those in the symptomatic category experienced worse psychological health, lower levels of life satisfaction and quality of life than individuals who were at risk or pre-symptomatic. Those living at risk and those who were pre-symptomatic showed similarities in negative psychological symptoms and psychological assets (that is a decline in coping skills and resilience). Chisholm et al. (2013) concluded from their research that further focus was needed in the area of psychological assets, that is the ability manage information and interact with it and others in a positive way. To understand how to enhance and support resilience and coping for individuals living at risk, more research is required.

One piece of research that does exist explored the experiences of everyday life for the at-risk group taking participants from the Prospective At-Risk Observational Study in the USA (Quaid et al., 2008). The study reported that people living at risk carry a greater burden around disclosure and concealment of their at-risk knowledge when compared to other chronic diseases. It also concluded that having hope is a crucial factor in living well whilst living at risk and this enables coping ability for the future. Whilst this research highlights some of the experience of day to day life living at risk it does not offer a complete understanding of psychological need or emotional experience. Furthermore, it was conducted some time ago and only in one country. There is little research into the experiences of this specific group within HD which examines psychological wellbeing of this group.

Future research is needed on the common challenges faced among people at-risk of HD, in addition to psychological symptoms and ways of maintaining well-being. Those living at-risk of HD face difficult decisions (life insurance, starting a family of their own, or embarking on a career) and it is unclear how these and other possible challenges affect psychological well-being.

Consequently, the current study will aim to explore the challenges in maintaining psychological wellbeing that individuals living at risk of HD in the UK face. Having this understanding will equip us with the insight needed to start the initial stages of psychological care provision for supporting those living at risk.

Given the aim is to explore experiences in depth, a qualitative approach will be adopted. Interpretative phenomenological analysis (IPA) will be used as this qualitative analysis method allows the researcher to make sense of experiences and the perspectives within these

experiences (Larkin et al., 2021). This research will therefore address the question: what are the challenges of maintaining psychological wellbeing while living at risk of HD in the UK.

Method

Design

IPA will be used as it is an established qualitative method with an idiographic focus that allows the consideration of individual lived experiences in informing meaning (Larkin et al., 2021). The two main aims of IPA are to examine in detail how an individual has made sense of their experiences and to offer an interpretation of the experience to aid understanding of it. The main advantage of using IPA is that it enables the data to provide insight into complicated interrelationships and patterns within experience in a specific context (Larkin et al., 2021). The research will be qualitative and collect data using semi-structured interviews with participants. Data will be collected on the participants' experiences of maintaining their own psychological wellbeing once they had found out and adjusted to their 'at risk' status.

The semi-structured interview topic areas have been discussed fully with experts by experience. This helped to establish areas for focus and exploration and the phrasing of questions. Experts by experience also helped with the creation of the recruitment advertisement, participant information and consent form to ensure that the research description is clear yet inviting and that the materials are easy to understand.

Participants

To be included in the research participants will need to be over the age of 18 (adult in the UK). The participants will need to be aware of their at-risk status and able to communicate where their risk stems from in their biological family so that level of risk (25% (via grandparent) or 50% (via parent) can be established. Participants will need to have had knowledge of their at-risk status for a minimum of 12 months. This is due to the suggested time for adjustment to this knowledge being 6 months (Tibben et al., 1997) and the experience of living with the knowledge for a further 6-month period. It is hoped that interviewing participants who have lived with their at-risk status for at least a year will allow the retrospective capture of the adjustment stage and challenges that may have been experienced in terms of maintaining psychological wellbeing in this time, as well as their current status.

Participants must be symptom free, not have been tested for HD, speak English without any communication difficulties and able to withstand an interview of up to 90 minutes in length (or be able to commit to an interview spanning over two separate dates and times).

Participation will be voluntary, and participants may decide to withdraw at any point up to two weeks after the completion of their interview. At this point data analysis will have begun. Once analysis has begun it would be very difficult to deduct an individual response to interview questions. There is no requirement for a reason to be offered for a participant's withdrawal from the research.

Creswell (2012) suggest that an ideal number of participants for IPA is between two and twenty-five individuals. To allow for potential withdrawal and to ensure the gathering of sufficient data, between eight and twelve participants will be recruited. Having between eight and twelve participants will allow an in-depth exploration of each narrative which will highlight experiences and comparisons.

Procedure

Participants will be recruited via a social media group which is on Facebook called EXTRACTED, run by the experts by experience who have contributed to the study design. This group is for people who have any link to Huntington's disease in the UK. This could mean for example that they have a family member in the UK who has HD, they may be at risk of HD or they may have HD at any stage themselves. A digital poster will be shared within the group (by the group administration, not my personal Facebook account) asking members who meet the inclusion criteria and are potentially willing to participate to contact me directly using the Lancaster University email address: h.cooper3@lancaster.ac.uk.

If enough participants cannot be recruited via social media, EXTRACTED will be contacted for recruitment via their website. If there is a problem recruiting via Facebook and the EXTRACTED, then a bespoke Twitter account will be created to aid recruitment.

On first contact, participants will be sent an explanation of the study. This will include information on confidentiality, inclusion and exclusion criteria and commitment information. If the individual remains willing to participate, a consent form explaining audio consent and what will happen with recordings, transcribed interviews and results will be sent to the participant (via email).

Opportunity to discuss the information and raise any queries or worries will be offered to potential participants prior to scheduling an interview. Those who wish to participate will be checked against the inclusion and exclusion criteria. Participants will have at least a 24-hour time period between agreeing to participate and the interview date to ensure they have had time to consider the implications of taking part in research for themselves.

Consent will be taken in audio format (via Microsoft Teams) at the start of the interview which will be recorded and securely stored on the researcher's Lancaster University OneDrive or H drive. Once consent has been received participants will be offered an interview via Microsoft Teams for up to 90 minutes which will also be recorded using Microsoft Teams. Following the completion of each interview on Microsoft Teams, the software will auto generate a transcript. This will be a basic transcription and will need to be carefully read and edited to ensure it is correct.

Recordings will be uploaded immediately to a secure location (e.g. main researcher's H drive or One Drive).

Recordings of the interviews will be kept on the researcher's H Drive or One Drive until the thesis is complete, at which point audio recordings (excluding consent) stored in this location can be destroyed.

Analysis

The narrative will be transcribed verbatim and any identifying factors removed. IPA guidelines by (Larkin et al., 2021) will be followed for analysis and creation of themes. The transcripts will be read individually and coded and reread multiple times before collating the themes for each participant. Themes will then be analysed across participants. Any quotes that are to be used within the writing up of the research will be anonymised, though basic demographic information, such as sex, age and ethnicity, may be included.

Practical issues

As recruitment is carried out primarily through a social media group and possibly the EXTRACTED, participants could be from anywhere within the UK. The use of Microsoft Teams alleviates any practical issues and costs this could incur.

Ethical concerns

As the topic of discussion around psychological wellbeing is in the context of currently living at risk it is possible that some participants may find the conversation upsetting. The researcher will keep in mind that distress may be more difficult to notice in participants who are being interviewed online compared to the experience of interviewing participants face to face. If this is identified in the interview the participant will be offered a break and asked if they would like to continue. Participants will be given the option to complete the research on another day. They will be provided with details of support lines (Huntington's Disease Association and The Samaritans) and if needed encouraged to talk to some they trust such as a GP or family member. If necessary, direction to A&E may also be offered.

At the end of each interview the researcher will conduct a check-in with each participant to establish how the individual is feeling before the interview is ended.

Participants will be informed of what will happen with their information and how it will be stored. Interviews, transcripts and notes will be encrypted and stored on the researcher's Lancaster University H drive or One Drive (or equivalent storage approved by the university - all password protected) on the researchers' password protected computer. Participants will be made aware that Lancaster University will keep a copy of consent recordings and transcriptions for 10 years following the completion of the research. Storage will be by the Research Co-ordinator of the DClinPsy, overseen by Dr Fiona Eccles, Research Supervisor.

Microsoft teams will be used for interviews and interviews will be recorded. Microsoft state that: ‘Teams enforces team-wide and organization-wide two-factor authentication, single sign-on through Active Directory, and encryption of data in transit and at rest. Files are stored in SharePoint and are backed by SharePoint encryption. Notes are stored in OneNote and are backed by OneNote encryption.’ At the end of each interview recording will be downloaded and deleted from Microsoft Teams. https://docs.microsoft.com/en-us/microsoftteams/security-compliance-overview#:~:text=Teams%20enforces%20team%2Dwide%20and,are%20backed%20by%20OneNote%20encryption.

Timescale

Submit ethics – August 2021

Gain ethics approval - October 2021

Recruitment and interviews throughout October – January 2021

Start transcribing interviews and analyse as appropriate – January - March 2022

Complete analysis– March - April 2022

Create a complete draft of the thesis – May - July 2022

Work on suggested changes and edits August – October 2022

Submit thesis - March 2023

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Appendix 4 – A

Ethics Approval Letter

Applicant: Hollie Cooper

Supervisor: Dr Fiona Eccles

Department: DHR

FHMREC Reference: FHMREC20188

08 October 2021

Re: FHMREC20188

What are the challenges in maintaining psychological wellbeing for people living at risk of Huntington's disease?

Dear Hollie,

Thank you for submitting your research ethics application for the above project for review by the **Faculty of Health and Medicine Research Ethics Committee (FHMREC)**. The application was recommended for approval by FHMREC, and on behalf of the Chair of the Committee, I can confirm that approval has been granted for this research project.

As principal investigator your responsibilities include:

- ensuring that (where applicable) all the necessary legal and regulatory requirements in order to conduct the research are met, and the necessary licenses and approvals have been obtained;

- reporting any ethics-related issues that occur during the course of the research or arising from the research to the Research Ethics Officer at the email address below (e.g. unforeseen ethical issues, complaints about the conduct of the research, adverse reactions such as extreme distress);

- submitting details of proposed substantive amendments to the protocol to the Research Ethics Officer for approval.

Please contact me if you have any queries or require further information.

Email: fhmresearchsupport@lancaster.ac.uk

Yours sincerely,

Tom Morley, Research Ethics Officer, Secretary to FHMREC.

Appendix 4 – B

Digital Recruitment Poster

I am Hollie Cooper and I am looking for participants (8 -12 required)

to help answer the following question for my doctoral thesis in Clinical Psychology:

What are the challenges in maintaining psychological wellbeing for people living at risk of Huntington's disease?

- If you have a parent or grandparent with HD and are NOT tested yourself
- If you are not experiencing any of the possible symptoms of HD and do not have any other significant physical health problems
- If you are willing to take part in an interview of up to 90 minutes duration

I would really value hearing about how you maintain or experience psychological wellbeing whilst living in the knowledge of being at risk of HD

For further information (with no obligation to take part) please contact Hollie Cooper: h.cooper3@lancaster.ac.uk or university supervisor Dr Fiona Eccles: f.eccles@lancaster.ac.uk

Appendix 4 – C

Consent script for online video consent

Project Title: What are the challenges in maintaining psychological wellbeing for people living at risk of HD?

Please note, the questions below are just for information. You do not need to return this form. Your consent will be taken in an audio recording where I will read the below form to you and request your responses. The recording will then be stored securely in line with GDPR for 10 years by Lancaster University.

Name of researcher: Hollie Cooper

I am asking if you would like to take part in a research project exploring how individual's living at risk of Huntington's disease maintain their psychological wellbeing.

Before you consent to participating in the study, I ask that you read the participant information sheet. After this, I will ask you to verbally respond to the statements below in an audio recording. If you have any questions or queries before giving your consent please speak to me, Hollie Cooper, the principal investigator.

1. I confirm that I have read the information sheet and fully understand what is expected of me within this study

2. I confirm that I have had the opportunity to ask any questions and to have them answered.

3. I understand that my interview will be recorded using Microsoft Teams and then made into an anonymised written transcript.

4. I understand that recordings will be kept until the research project has been examined.

5. I understand that my participation is voluntary and that I am free to withdraw at any time without giving any reason, without my medical care or legal rights being affected.

6. I understand that I may withdraw my data at any point up to the ending of the two-week period following my interview. I understand that once my data has been anonymised and incorporated into themes it will not be possible for it to be withdrawn.

7. I understand that the information from my interview will be pooled with other participants' responses, anonymised and may be published.

8. I consent to information and quotations from my interview being used in reports, conferences and training events. Any information and quotations used will be anonymised

9. I understand that the researcher will discuss data with their supervisors as needed.

10. I understand that any information I give will remain confidential and anonymous unless it is thought that there is a risk of harm to myself or others, in which case Hollie Cooper will need to share this information with her research supervisors, Dr Fiona Eccles, Dr Maria Dale and Professor Jane Simpson.

11. I consent to Lancaster University keeping written transcriptions of the interview, and my recorded audio consent for 10 years after the study has finished

12. I consent to take part in the above study

Appendix 4 – D

Participant Information pack

Created: 06.08.2020

Participant Information Sheet

Title of Study

What are the challenges in maintaining psychological wellbeing for people living at risk of Huntington's disease?

For further information about how Lancaster University processes personal data for research purposes and your data rights please visit our webpage:

www.lancaster.ac.uk/research/data-protection

I am Hollie Cooper, and I am currently a trainee at Lancaster University on the Clinical Psychology Doctorate programme. This information sheet aims to provide everything you need to know about the study I am conducting for my thesis.

What is the purpose of this study?

- To discover the challenges faced by people living at risk of Huntington's disease (HD) when aiming to stay psychologically well
- To establish what challenges people living at risk of HD face when they try to maintain their psychological wellbeing
- To help inform future psychological support for people living at risk
- To share important findings about techniques and struggles and the experiences of people living at risk and their psychological wellbeing

Do I have to take part?

No. I greatly appreciate your expression of interest in the study. Expressing an interest does not mean that you have to take part. It's completely up to you to decide whether or not you take part after you have received all the information and had chance to consider it.

Unfortunately, you will not be eligible to participate if:

- you experience communication difficulties
- do not speak English
- have received a genetic test for HD as this would mean you would no longer be living at risk
- have motor symptoms of HD as this would suggest that you would no longer be living at risk of HD
- do not have a biological parent or grandparent with HD as this would mean your risk is not known or you are unlikely to be at risk of HD
- only found out about your at-risk status within the past year
- Under 18

What will I be asked to do if I take part?

If you decide to participate, we will arrange an interview at a time that suits you. The interview will be conducted using Microsoft teams and will be recorded and will last for approximately an hour, but could continue to 90 minutes if beneficial. Questions I ask will be focused on your psychological wellbeing and how you have tried to, or how you have maintained psychological wellbeing whilst living in the knowledge that you are at risk of HD.

You will not be required to talk about anything you do not wish to speak about and you may stop the interview at any time.

Example Questions

1. How do you feel knowing you are at risk of HD?
2. What thoughts go through your mind about HD?
3. Do these feelings/thoughts affect your life on a day to day basis?
4. How do you manage these thoughts/feelings?

Will my data be identifiable?

For the study to take place, I will need to record interviews. This will be done using Microsoft teams. The internet cannot be guaranteed to be completely secure and confidential. There will be a number of measures in place to ensure the information you share will be stored safely and kept as secure as possible. The interview may be recorded with the camera on, which would make your face and surrounding area visible, or camera off, which would mean only audio would be recorded. On completion the recording will be downloaded from Microsoft teams and deleted from Microsoft teams. Microsoft teams will automatically provide a transcription of our conversation which I will download and remove from Microsoft teams. I will then, solely re-watch (and listen to) the recording and ensure an accurate transcript is produced. All personal information you share will be confidential unless you disclose information regarding harm to yourself or others. If this occurred, we would discuss this and the action that would be necessary in order to ensure your safety and that of others.

What happens to the research?

- The research will form part of my thesis in doctoral training.

- The research is likely to be submitted for publication in a journal article.
- This research may also be presented at conferences
- Recordings of the main interview will be destroyed once the thesis has been examined
- Lancaster University will keep copies of the interview transcriptions and verbal consent recordings electronically for 10 years after the study has finished or 10 years from publication, whichever is longer. At the end of this time, they will be securely destroyed.
- Transcripts will have as much identifiable information removed as possible.
- Files stored on my computer will be encrypted (that is no-one other than me will be able to access them) and the computer is password protected.
- I will discuss the research and transcripts with my supervisors – this will be anonymous.
- You can choose to withdraw your data that you have shared anytime up to the end of the two weeks after your interview. At this point data analysis will have begun, at which point it will not be possible.
- Direct quotations may be included in the written thesis. These will be anonymous.

What will happen to the results?

The results will be summarised and reported in my thesis and may be submitted for publication in an academic or professional journal and presented at conferences, meetings and training events. A copy of the results will also be available to you at your request.

Are there any risks?

There are no risks anticipated with participating in this research though there may be some sensitive areas of discussion. If you experience any distress following participation, I encourage you to let me know so that I can direct you to appropriate support. I will endeavour to minimise any distress you are experiencing and you or I can stop the interview at any point if needed. It may be helpful to inform someone from your support network (friend, sibling, family member) that you are taking part in this research so that you can discuss anything that comes up for you if needed

Are there any benefits to taking part?

There are no direct benefits in taking part in this research. I hope that gaining a better understanding of how people maintain their psychological wellbeing and knowledge of the tools and techniques used will help others who are living at risk and also help to inform services of needs and service provision.

Who has reviewed the project?

This study has been reviewed and approved by the Faculty of Health and Medicine Research Ethics Committee at Lancaster University.

Where can I obtain further information about the study if I need it?

If you have any questions about the study or think you might like to take part, please feel free to contact me via email: h.cooper3@lancaster.ac.uk

You can also contact:

Dr Fiona Eccles (academic supervisor) on f.eccles@lancaster.ac.uk or 01524 592807

Dr Maria Dale (field supervisor) on Maria.Dale@leicspart.nhs.uk

Complaints

If you wish to make a complaint or raise concerns about any aspect of this study and do not want to speak to the researcher, you can contact:

Ian Smith

Research Director

Lancaster University,

Tel: +44 (0)1524 592981

Email: i.smith@lancaster.ac.uk

If you wish to speak to someone outside of the Doctorate Programme, you may also contact:

Dr Laura Machin Tel: +44 (0)1524 594973

Chair of FHM REC Email: l.machin@lancaster.ac.uk

Faculty of Health and Medicine

(Lancaster Medical School)

Lancaster University

Lancaster

LA1 4YG

Thank you for taking the time to read this information sheet.

Resources in the event of distress

Should you feel distressed either as a result of taking part, or in the future, the following resources may be of assistance:

Huntingtons Disease Association. Home | Huntington's Disease Association (hda.org.uk)

Telephone: 0151 331 5444

Huntingtons Disease Youth Organisation. Huntington's Disease Youth Organization (hdyo.org). E-mail: info@hdyo.org or support@hdyo.org

The Samaritans. Samaritans | Every life lost to suicide is a tragedy | Here to listen

Telephone: 116 123

The GP Practice at which you are a registered patient

Appendix 4 – E

Interview schedule and topic guide

It is thought that within IPA, interview schedules should be short, beginning with broad questions which will enable the participant to set the boundaries of the topic. This will prevent the researchers from imposing their own understanding on the participant's responses (Smith et al., 2009). With this in mind the purpose of this interview schedule is to offer some suggestions should it be required to help the researcher and participant feel at ease until a natural conversation can be engaged with.

Introduction

Introduce myself. Revisit the research question 'what are the challenges in maintaining psychological wellbeing for people living at risk of HD?

Recap the purpose of the interview and any relevant sections of the participant information sheet.

Remind participants of the limits of confidentiality involving disclosure of risk.

Collect demographic and other basic information: age, gender, ethnic group, partnership status, living status (who they live with), employment status, who has HD in their family

Reflecting on knowledge of living at risk

In this section I will seek information about how long it has been since the individual found out that they have been at risk of HD

This section may include their initial reactions and their expectations for the future in the context of wellbeing. This will not remain the focus but will provide context to their adjustment response.

Example questions:

When did you find out about HD in your family?

When did you understand that you yourself were at risk of HD?

Can you reflect on how this affected you at the time in terms of your wellbeing?

How do you make sense of your at-risk status?

Are there any reasons why you have not gone for a test?

Do you think you might get a test at some point?

What do you think the likelihood is that you may have the gene expansion? Why do you think this?

To answer the research question more specific topics will be raised. Examples of these questions may be:

Topic 1: Background and understanding.

What do you understand your risk to be?

What is it like knowing you at risk of developing HD?

Does this knowledge impact your interactions with others at home? At work? With friends/socially?

Topic 2: wellbeing

What does wellbeing mean to you?

How do you keep yourself psychologically well?

Do you think that knowing you are at risk affects your wellbeing?

What are your experiences of wellbeing?

What are the challenges to maintain your wellbeing?

Topic 3: Support

Have you ever sought support for psychological distress (e.g. from a GP, counsellor, psychologist etc?) If yes, what was this like?

How does it make you feel to explore your knowledge of HD in this way?

Conclusion

The participant will be thanked for taking part and I will ask how the participant is feeling. I will discuss any lasting concerns or worries with the participant. If the participant has been distressed by the interview, I will direct the participant to sources of support on the participant information sheet.