

**The experience of a sample of individuals in the United Kingdom living in the pre-manifest stage of Huntington's disease; an interpretative phenomenological analysis**

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### **Abstract**

This paper explores the experience of ten individuals living in the UK who were in the pre-manifest stage of Huntington's disease (HD), a genetic neurodegenerative condition. Data were gathered using semi-structured interviews and analysed using interpretative phenomenological analysis. Three themes were reported from the data: 'feeling limited by time', 'the perception of stalling time' and 'making the most of time', all highlighting the way in which time holds significant meaning when living in the pre-manifest stage of HD. This study has highlighted the difficulties experienced by individuals when adjusting to the pre-manifest stage of HD. Feeling able to manage their anxieties and dealing with ongoing uncertainty related to future deterioration was key to supporting their wellbeing, facilitated by factors such as positivity and hope. Individuals may benefit from counselling which supports them to develop proactive coping strategies to manage their anxieties and acceptance of an uncertain future.

**Keywords:** Huntington's disease; chronic illness; lived experience; decision-making; predictive genetic testing

#### **What is known about this topic:**

HD-related difficulties across the motor, cognitive and psychological domains are likely to occur gradually and may be present more than a decade before the actual diagnosis. Many individuals report anxiety related to potential symptom onset, often many years before diagnosis.

#### **What this paper adds to the topic:**

Little is known regarding how people who are living in the pre-manifest stage of HD cope with the knowledge that they will develop the disease. This paper adds in-depth exploration of the lived experience of pre-manifest individuals and the longer-term implications of having tested positive for the gene expansion.

## **Introduction**

Huntington's disease (HD) is a progressive neurodegenerative condition which is caused by an expanded CAG (cytosine-adenine-guanine) trinucleotide repeat in the gene encoding the huntingtin protein. HD is inherited in an autosomal dominant pattern, meaning a child of a parent with the HD gene expansion has a 50% chance of inheriting the disease. The age of onset differs across individuals, but signs of the condition and clinical diagnosis typically occur around age 30-50 years old (Carlozzi et al., 2016). The expansion number of the CAG repeat shows a very strong negative correlation with age at onset of motor signs, with a larger expansion number leading to earlier onset (Lee et al., 2012), but this is still hugely variable from person to person. Individuals testing positive for the HD gene expansion will almost certainly develop symptoms. Predictive genetic testing has allowed individuals who are at risk of HD (because a family member has been identified as having the HD gene expansion) to find out whether they will develop the disease, prior to showing motor symptoms. If they test positive, they are described as having "pre-manifest" or "pre-symptomatic" HD. Although the predictive test has allowed researchers to study the natural course of the disease and provide opportunities for early intervention, there are significant ethical issues regarding the potentially life-changing consequences which living with the test result entails (Lilani, 2005). By offering the predictive test, health services have an ethical and moral responsibility to understand the impact of genetic testing and to support individuals with the long-term consequences (Andersson et al., 2016).

HD symptoms occur across three domains: motor, cognitive and psychological. Motor difficulties include problems with swallowing, balance and mobility as well as chorea and bradykinesia (Ho & Hocaoglu, 2011). Cognitive difficulties include problems with memory, executive function and attention. Communication ability is impacted by dysarthria, linguistic

difficulties and behavioural changes, leading to negative effects on social interaction and diminished quality of life (Nuzzi, 2018). Psychological difficulties are also common, including apathy, depression, irritability and anger, aggression, suicidal ideation, obsessional compulsive traits, anxiety and psychosis (Eddy et al., 2016; Wetzel et al., 2011; van Duijn et al., 2008). Motor and cognitive difficulties typically worsen with advancing disease progression, resulting in reduced health related quality of life (Carlozzi & Tulskey, 2013). In advanced stages, individuals are likely to require 24-hour care and become totally dependent on others for their most basic needs, a level of need which is very frequently not met by healthcare and social support services (van Walsem et al., 2015).

In order to receive a diagnosis of manifest HD, an individual must experience the onset of formal motor symptoms (Reilman et al., 2014). However, difficulties across all three domains are likely to occur gradually and may be present more than a decade before the actual diagnosis (Paulsen, 2010). Furthermore, many individuals report anxiety related to potential symptom onset, often many years before showing motor difficulties (Konrad, 2003). There is no way of preventing the onset of the disease, currently no cure, and HD is generally fatal within 15-20 years of clinical diagnosis (Carlozzi et al., 2016). Nevertheless, the advances in the understanding of HD has led to numerous therapeutic strategies which aim to target the pathogenic protein, many now in clinical trial stages (Dash et al., 2020), offering hope to the HD community.

Although much research has been done identifying psychological consequences of a positive test result, the focus is generally on clinical symptoms such as anxiety and depression or on adverse events such as family breakdown or suicide (McCusker & Loy, 2017). Furthermore, the focus is generally on the short-term (typically under one year since testing) psychological

implications of predictive testing (Almqvist et al., 2003). These include symptoms of depression, obsessive compulsiveness, anxiety, and psychosis, as documented by large scale quantitative studies (Berrios et al., 2002; Duff et al., 2007; Crozier et al., 2014). There is little knowledge about the in-depth experiences of these individuals from a qualitative perspective, including the longer-term experience after having tested positive. For instance, there has been little research into how a person who has tested positive for the HD gene expansion but is not yet symptomatic perceives their status as ‘embodied risk’ (Kavanagh & Broom, 1997). Rather than being risks caused by lifestyle or an individual’s environment, embodied risks are located in the person said to be ‘at risk’ and thus say something about the person’s identity (Kavanagh & Broom, 1997). The uncertainty about when individuals will develop the physical symptoms (and thus receive a diagnosis) is likely to compound any psychological difficulties. This may be particularly so given the difficulties inherent in deciding whether symptoms in pre-manifest individuals constitute a clinical diagnosis (McCusker & Loy, 2017).

Exploring the meaning of the pre-manifest experience is fundamental in order to better understand variations in psychological outcomes across individuals and how to provide effective psychological support to those experiencing difficulties. Previous qualitative literature on the impact of genetic testing in HD has highlighted some of the ethical dilemmas involved in testing and explored how individuals integrate the knowledge of their positive HD status into their lives. In one study exploring the impact of genetic testing on reproductive decision making, all of the study participants stated that the test had had a positive effect on their lives or no impact at all (Chapman 2002). Where individuals did not feel their HD status had changed the way they think about themselves or others, this was often because of not having symptoms and thus individuals were able to push the implications to one side and consider it as part of their future, not part of

their present. This suggests that the potential impact of genetic knowledge in affecting an individual's life depends upon how salient the knowledge is for them at the time. If the knowledge is just one part of many other competing factors in an individual's life, the impact may be more minimal. This is in keeping with the work of Schutz (1946), who described zones of relevance depending upon the degree of knowledge, certainty and action when an individual is faced with a situation or difficulty (Etchegary, 2011). Genetic risk may have high relevance at significant life points (such as reproductive decision making) but at other times may have low relevance and thus not be accorded much attention or have much impact on an individual's life (Parsons & Atkinson, 1992; Cox & McKellin, 1999). In a study of individuals living at risk of HD but prior to predictive testing, it was observed that risk saliency interacted with biological disruption (Etchegary, 2011). Where an individual's genetic risk was more salient because of certain life junctures (e.g. decisions about reproduction), an individual was likely to experience more disruption to their identity, their plans for the future and their social relationships (Etchegary, 2011). Risk saliency was high at particular life stages such as family planning, marriage etc.; age at which their parent developed the disease; other disease-related events in the family (e.g. the receipt of test results or the death of an affected family member) and other life events specific to an individual (Etchegary, 2011). It may thus be anticipated that living in the pre-manifest stage is a complex and nuanced experience, in which the impact of being 'at risk' is dynamic and temporal according to the individual's context and the degree of relevance their 'at risk' status holds.

It seems clear that understanding the meaning an individual ascribes to their experience and a focus on the pre-manifest stage beyond the impact of the genetic test requires further exploration. Genetic counsellors and individuals who support people considering genetic testing

may benefit from this understanding of how people experience the pre-manifest stage. The individuals at risk of HD who do decide to undergo testing may have ways of coping with their experience which can be of value to all those exploring genetic testing. Thus, this study aimed to answer the question: ‘how do individuals experience the pre-manifest stage of HD’? With considerations relating to the importance of individual context and meaning, interpretative phenomenological analysis (IPA) appeared the most appropriate methodology (Smith et al., 2009). In contrast, an alternative approach such as grounded theory or thematic analysis focuses on the emerging patterns within the data or superordinate theories in its analysis, rather than the way in which an experience is embodied and given meaning by the individual’s relationship to their experience. IPA enables the patterns in the data to generate salience or meaning for the researcher, but crucially, an individual’s voice in those patterns is not lost, thus enriching our understanding of prominent issues in healthcare (Peat et al. 2019).

## **Method**

### **Ethical approval**

The study received ethical approvals from the UK Health Research Authority Tayside Research Ethics Committee (reference: 19/ES/0105) and approval to conduct the study from the Research and Development Department at the host National Health Service (NHS) trust.

### **Design**

The study design followed guidelines for IPA research (Smith et al., 2009). As a hermeneutic approach it gives precedence to the individual’s personal interpretation of, and meaning given, to their experience. As an idiographic approach, IPA provides a framework for thorough analysis of individual experiences (Smith et al., 2009), which felt to be important to

capture the complexity of living with a chronic illness (Brocki & Wearden, 2006). Furthermore, IPA recognizes the important role of the researcher in making sense of an individual's account of their experience (Smith & Osborn, 2003). This awareness felt crucial to the present study in order to continually reflect on the ways in which the researcher's active involvement in the participant's narrative might shape the research outcome (Pringle et al., 2011) and thus the importance of constructive self-reflection in the research process. IPA enables the researcher to more fully explore the way the participant's experience influences the meaning that they give to being human (Engward & Goldspink, 2020).

### **Reflexivity**

Reflexivity is a core component within IPA, given the need to continually reflect on the meaning which we generate from the data in the analysis process (Shaw, 2010). The analysis was conducted by the first author, at the time a trainee clinical psychologist, with supervision from the second and third authors. Although the first author has no direct experience of HD, family experience of multiple sclerosis shaped her understanding of the importance of psychological mindset in adjusting to a long-term neurological condition. The second author is a clinical psychologist and researcher who works in an HD service, and the third author is a researcher in neurodegenerative conditions. Regular supervision and the use of a reflective diary enabled the first author to reflect on her shared and divergent experiences with each participant and consider how these experiences may have contributed to her interpretation of the data. The second and third authors also reflected on the knowledge they brought to the analysis and in particular the second author's experience of working with individuals with HD was acknowledged, looking for similarities and differences in the data to that experience. Consideration was also given to the way in which the authors' interpretations may have been

limited given our understanding of each individual's experience was only generated through the participant's own account of it (Smith et al., 2009).

## **Participants**

To be included in the study, participants were required to be between 18 and 65 and to have tested positive for the HD gene expansion at least one year prior to the interview. It was felt that interviewing participants at least one year following the test would encourage participants to focus on their experiences of living with their genetic status rather than those of the testing period. Participants self-reported that they had the gene expansion for HD as confirmed through genetic testing. They also confirmed that they had not been diagnosed with the disease. Participants were excluded if they had a comorbid significant illness. Two participants were recruited through an HD clinic at an NHS Trust and eight were recruited through a Facebook advertisement on the Huntington's Disease Association's webpage; all provided written informed consent prior to interview commencement. Participant recruitment was terminated at 10 participants in order to obtain a small, well defined sample required for IPA (Smith et al., 2009). Pseudonyms were used in all tables, transcripts and subsequent analyses and any other identifying information was changed. In brief, there were 3 male and 7 female participants, mean age was 38 years (range 25-50) and mean time since their genetic test was 5 years (range 2-10). All were White British. There were 6 individuals who were married or in a relationship with one or more children, 2 married with no children and 2 single with no children. Most (8) individuals were in paid employment. The demographics of participants are shown in Table 1.

### **Recruitment and data collection**

Participants were invited to take part through letters of invitation provided by research nurses at the host NHS Trust, or by responding to the Facebook advertisement. The first author's contact details were included, and potential participants were asked to contact her if they were interested in participating. After answering any questions and confirming they were happy to proceed, a mutually convenient time was arranged with each participant for the interview. Consent forms had to be completed in advance of the interview and emailed or posted back to the researcher; at the beginning of this meeting further questions were clarified and participants were asked to reaffirm consent.

Semi-structured interviews were used to ensure the focus was on the pre-manifest stage but were also guided by participants' responses. Interviews were held between January and March 2020 and were conducted via telephone due to differing geographical location of participants. Interviews lasted between 52 and 80 minutes. Arguably, telephone interviews are an effective option for qualitative data collection due to it complementing the style of semi-structured interview schedules (Cachia & Millward, 2011) and have been used previously in IPA research (Swift & Wilson, 2001).

### **Data analysis**

Data analysis followed guidance from Smith et al. (2009). There were four stages to the analysis. The first stage involved multiple readings of an individual participant's transcript with comments noted around points of interest. These comments were closely grounded in the raw data. The second stage involved re-reading of the transcript, with emergent themes noted in the right-hand column which attempted to describe the participant's experiences. In the third stage of

the analysis, emergent themes were listed, and similar/connecting themes were grouped into superordinate themes. In the fourth stage, analysis was conducted across all the transcripts and superordinate themes for the entire data sample were generated by looking for concordance across participants. Researcher assumptions were reflected upon and incorporated into the analysis using a reflective journal and regular supervision. Supervision was a space for analytic interaction, highlighting which interpretations belonged to the researchers and which belonged to the participants, strengthening the rigour and transparency of the interpretations (Engward & Goldspink, 2020). Sensitivity to context is equally important to high quality research (Yardley, 2008) and this was enhanced through regular discussion with the second author who works in a HD clinic.

## **Results**

The findings indicated that the way in which individuals adjust to living in the pre-manifest stage of HD is underpinned through their representation of time. The results span past, present and future and are depicted by three main themes: ‘Feeling limited by time’, ‘the perception of stalling time’ and ‘Making the most of time’. Each of these themes and the sub-themes are described in the following discussion.

### **Theme 1: “It’s like a trigger for a countdown”- Feeling limited by time**

The theme “feeling limited by time” captured participants’ reflections of the way in which receiving their genetic test results felt to limit the time they had left. This theme incorporated two main sub-themes: the countdown to symptom onset and HD dictating the present.

### **Countdown to symptom onset.**

One recurring theme across participants' accounts was the way in which receiving their test results felt to initiate a process of counting down to symptom onset. The acuity of the countdown period was maintained by a hypervigilance towards any possible signs which might suggest symptom onset. The overwhelming fear of possible symptom onset was described by most participants and was alluded to by Daisy:

And if you do things like you forget things or names or you put your purse in the fridge or whatever, it only takes a couple of things to happen over a period of time for you to start panicking and think 'oh my god, is this it, is it starting?'

Many individuals were able to question their fears and think through alternative explanations given the context of their experiences. As Sharon pointed out: "you know, it could well be it's a symptom, or it could just be that you're very, very stressed at that point and actually, the kinds of things that happened to the brain when it's stress [ed] is very similar." Despite this sense of rationality, symptom questioning was pervasive, resulting in the countdown to symptom onset being an ever-present feature in the background of the participants' lives. Paul summed this up neatly: "is it Huntington's starting? I'll always have that"

The idea of the HD status being an ever-present feature in an individual's life was echoed by others, in which HD became part of "my forever radar rather than my temporary radar" (Daisy) and "an unresolved threat just lurking in the background" (Angie). For Steve, there was a period at which "it settle(d) into the background noise and has kind of remained fairly constant ever since". Alongside "threat", "radar" and "noise", the contextualized nature of "background" becomes apparent. It would seem as though the word 'background' reflects the underlying and

insidious backdrop of HD in the participants' lives, always reminding them of the countdown which commenced following the test result.

The countdown to symptom onset was often made more pertinent by seeing the course of parental disease. As Catherine described, seeing the way her mum experienced symptoms: "can be upsetting sometimes because it is a window into your own future almost". This window into the future is the lens through which many individuals described seeing their own onset. For the participants, the experience of parental symptoms often enhanced the clarity about the nature of what their own symptoms might look like.

### **HD dictating the present.**

The ways in which an individual's HD status dominated their thought processes highlights how participants often felt HD was dictating their present. As Joanna explained with regards to her decision to change careers from events marketing to teaching following the test results:

Honestly it was the Huntington's gene. Originally, I trained in events, it's a very active job and you are up and down all the time running around different venues and it's not something I would be able to do long term.

The personification of the Huntington's gene depicted Joanna's sense that the gene had control over her decision making, where she felt forced to re-train because she knew she may not be able to continue in her preferred career once motor difficulties started. The negotiations individuals made with regard to their HD status is echoed in Angie's account when speaking about navigating the dating world with HD, where she described that she had to "negotiate that with a ball and chain around your foot which is how I view Huntington's." The ball and chain metaphor depicts an image of a criminal being physically restrained and held back; perhaps

Angie felt a similar sense of restraint in having tested positive for the HD gene expansion; “if you don't get tested everything remains a possibility for you and then when you test positive doors start closing.”

It seemed the test results had prompted urgency about what one could realistically aim to do long term. A similar sense of urgency is reflected in Carol’s comments: “I've always been sort of a straightforward person, but it made me re-evaluate a lot of things. We moved to a new house kind of because I knew that I needed to do certain things within a certain timeframe.” Here Carol described feeling “made” to re-evaluate things and move house due to the sense of urgency which came about from having the test. This sense of urgency is triggered by the knowledge that one has the HD gene expansion, such that one felt the result: “is a trigger for a countdown if you like.” (Paul)

Interestingly, the word “trigger” was used by several participants, highlighting the sense felt by participants of the results setting off something, whether that is forced decision making or emotional rollercoasters.

## **Theme 2: “I have no intention of becoming symptomatic”- The perception of stalling time**

The theme “perception of stalling time” encompassed the way in which individuals gained some sense of agency over the unpredictable and uncertain nature of living in the pre-manifest stage. Two sub themes accounted for the ways in which individuals were able to do this: distancing self from HD and holding onto hope.

### **Distancing self from HD.**

The perception of stalling time reflected participants’ beliefs that they were able to delay symptom onset by distancing themselves from the reality of their situation, or by viewing it as

very much a concern for the future. This differed from holding onto hope, which emerged as a way of directing their focus to research and drug trials. Instead, distancing from HD reflected a determination to disengage with HD related thoughts and concerns, such as by viewing their future as free of symptoms:

So this time in 10 years I fully intend to be part of, if not a drug trials programmes, certainly using whatever it is that's going to delay onset for as long it needs, so that's where I'm coming from when I say I've no intention of becoming symptomatic. (Steve)

Not only did Steve describe how he still expects to be asymptomatic in 10 years' time, but also that he had no intention of becoming symptomatic. This felt to be more than just an expression of hope, and more of a deliberate determination not to engage with the idea that he too might develop symptoms. He distanced himself from present concerns about symptom onset by reinforcing his belief that it would not happen for at least another 10 years.

Most participants seemed to refer to family patterns of the disease in order to maintain the belief that symptom onset was very much for the future. Hannah explained how "nobody in the family [have shown any] symptoms till 60 plus so I think I'm very lucky in that sense and I think I use that as sort of a crutch like 'oh I won't be sick until I'm 60.'"

Similar certainty about onset was described by Daisy, who was working on an "18-year timeframe", Catherine, who used her CAG count to "give (her) at least 20 healthy years" and Paul, who thought "there is a good chance that (he's) got another 5 to 10 years". For most participants, their own predictions of symptom onset seemed to be heavily influenced by parental experience of symptom onset, providing a sense of certainty that one had a defined period prior to showing symptoms.

An alternative way that parental experience of the disease helped participants to distance themselves from their own experience of HD was by providing a focus for their time and energies. As Joanna described when asked how she felt about knowing she would develop symptoms, “honestly at the moment it doesn’t bother me as I’m normally focused on my mum”. Similarly, Sharon described how “going through the fact that obviously we were dealing with my mum made coping with my own experience easier”. While these quotes by Sharon and Joanna highlighted how focusing on their parents with the disease enabled them to create distance from thoughts about their own symptom onset, this contrasted with Catherine’s point earlier (as discussed under the theme of ‘countdown to symptom onset’) whereby seeing one’s family members with clinical HD could also be upsetting and present a window to the future.

### **Holding onto hope.**

A closely related concept which linked with an individual’s perception of stalling time was their ability to hold onto hope. Many of the participants described hope that current drug trials might eradicate or delay symptom onset. Carol described the hope she shared with her children when telling them that she had tested positive for HD: “...‘by the time mummy is 70 there will definitely be a medicine out there’...so to look at it from that point of view, so the future isn’t all dark and bleak there is hope, there is hope there.” Here Carol described how she holds onto hope that future medical trials will alleviate or prevent symptom onset, much in the same way that Steve described his hope for future drugs being developed prior to his symptom onset. Carol’s use of the word “mummy” in the opening of this statement indicated the significance of hope for her; it is as though she needed to remain hopeful as part of her responsibilities as a parent to her children.

The idea that hope is almost necessary in order to manage life in the present is echoed by Catherine when responding to why hope was important: “because if you don't have hope then what do you have?” Hope was not around initially for Catherine. As she described: “there is a massive adjustment that you go through when you are told you've got the gene and rather than think about hope and stuff you're kind of adjusting to the news that you've got this defective gene”. This adjustment period was echoed by Steve, who described that “once you find out, it's quite an emotional roller coaster”, Jake, who thought: “it does take a period of adjustment”, Angie who had “a period of time where I was really anxious about them and upset by them” and Paul who mentioned that “initially I didn't see that there was a lot of hope and I guess I was approaching it from a terminal illness point of view.” It was almost against these backdrops however that hope emerged as even more significant in the participants' lives. Paul summed up the importance echoed by many of having hope in their life:

I guess it stood a chance of almost paralysing me knowing that it was happening or would happen...But I kind of overcame that knowing that, well you could waste a lot of time which is bad for everyone around you...I want to live my life with the view that I'm free from Huntington's.

Thus, despite the very real difficulties which HD presented to individuals and their families, hope was a very tangible force for participants in preventing them from being paralysed by the fear of HD and allowing them to live their lives.

**Theme 3: “I could be hit by a bus tomorrow”- Making the most of time**

The theme “making the most of time” described an individual’s acknowledgement that, despite symptom onset being a future reality, their time in the present was precious. This is reflected in two sub themes: staying positive and choosing valued connections.

**Staying positive.**

For most participants, the need to stay positive was significant in helping them to make the most of their present. As Carol described: “I need to put all my time and energy into positive things and positive people and doing good things and making a positive impact”. The importance which Carol attached to remaining positive is reflected in its reiteration three times within this statement. It was by staying positive that she was able to maximize her time and energy and really make the most of the time she has, implicitly conveying her caution that she might not be able to make a positive impact for as long as she might like. Similar descriptions of the need to be positive were conveyed by other participants. For example, in relation to the test result, Daisy explained “I’m quite positive about it to be honest...I’m focused on the here and now”. Jake also described how “trying to have a positive impact towards [the result] to some extent tends to help me”, while Joanna similarly expressed determination to keep positive until the symptoms started. Furthermore, Steve talked about “trying to make a positive action now to something that is quite negative”. It is thus through positivity that individuals seemed able to make the most of the time they had symptom free and prevent themselves from dwelling on their potential future.

Catherine for instance avoided dwelling on future fears by focusing on the things she wanted to do with her life and reflecting on the ways it might not be HD which limited her life. As she explained: “I could dwell on it and worry myself sick about it and not do things that I

want to do and then go out and get hit by a bus...” Interestingly, the phrase “I could get hit by a bus” was echoed in several participants’ accounts. It highlighted that, for many of the participants, there was comfort to be gained from knowing that it is impossible to predict what can happen in the future, regardless of having the HD gene expansion. An individual can either waste time worrying about something which may never transpire, or instead choose to live for today and be positive about what they currently have. In reflecting on the relative benefits of focusing on symptoms versus living their life with a positive mindset, many of the participants were able to “push it to the back of [their] mind and carry on living life” (Daisy).

### **Choosing valued connections.**

Making the most of time was further reflected in the way that individuals made conscious decisions about who to spend their time with and which relationships were no longer worth engaging with. For many, spending time with family helped them to make the most of a symptom free present. As Sharon described: “The family is very supportive and it’s [HD] not something that’s not talked about”. For Sharon, it felt very important to talk about and share her concerns with family in order to move on from her worries and embrace her time in the present. This was echoed in others’ accounts: “But I guess I felt a lot better after I talked to Jane [partner] about my concerns in that my anxiety almost completely melted away” (Paul).

In sharing concerns with family members, Sharon felt able to reduce the power of HD from being that which was hidden and stigmatized in the past to something which could be dropped into casual conversation, similar to Carol who valued talking with her family about her HD concerns even if it was not “deep and meaningful”.

Being selective with whom one spent time with extended to being less tolerant of people who did not share their perspective about life. As Carol described when she discussed some of the implications of the HD result: “And I think I’ve become less, not patient, I can’t think of the word for it, less tolerant of idiots and people that are moaning about something completely ridiculous and shallow.” By discarding negative relationships, individuals were able to focus on the positive relations in their life, and even used their HD status as an impetus for coming together as a family. Joanna summed this up nicely when describing how HD had brought the family closer: “life cannot get in the way anymore. You can’t be putting stuff off thinking I’ll do it later I’m too busy because there might not be a later.”

### **Discussion**

The results of this study describe the experience of living in the pre-manifest stage of HD. Embodied in participants’ accounts and reflected in each of the three themes is the salience of time; its ability to take on new meaning and value once an individual is aware of their test result. The participants’ experience of knowing they will develop a chronic illness induced different relationships to time than are experienced by an otherwise healthy person (Jowsey, 2016), where expectations for the future changed, the meaning of their present altered and their past relationships to others also adjusted. For the present participants, this was reflected in their alterations of future plans, their discarding of negative past relationships and their determination to see gratitude in their present. The salience of time for individuals with chronic illness is not new and its origins can be found in the work of Bury (1982), who suggested that a chronic illness can disrupt a person’s previously accustomed experience of time and their expectations of living within established time-frames of childhood through to old age. That work focused on the experience of individuals with rheumatoid arthritis and has been applied to many chronic

illnesses (e.g. Faircloth et al., 2004; Harris, 2009; Carricaburu & Pierret, 1995) and differs from the experience of individuals in the present study who are not yet experiencing their condition. Nevertheless, the account of “biographical disruption” appears to mirror the account of the participants, in which their understanding of time and their life within it felt to be disrupted upon receiving the genetic test results. Similarly, Charmaz, in their longitudinal work on chronic illness describes how an individual’s struggle to control their illness is really an effort to control time and to preserve the defining image of the self (Charmaz, 1993). The experience of chronic illness presents ambiguity for an individual over who they are in the world; in order to resolve this ambiguity, the individual uses time as a framework to understand and make sense of their experiences (Ellingsen et al., 2014). Such ambiguity and unpredictability are reflected in the participants’ accounts as they described their initial struggle when finding out they had tested positive. Thus, the participants’ way of adjusting to the results was to gain back some control over their present and to exercise some agency in decision making for an uncertain future. This was also true in a study of 18-35-year olds who had tested positive for the gene expansion, where individuals described the importance of taking action following the test results (Gong et al., 2016). In that study, individuals described how the awareness that their time without symptoms was limited prompted the need to gain back some agency by expediting career and relationship choices and plans. The urgency with which they embarked on this process was not negative however but involved pursuing their career interests with more determination and choosing partners who were more mature and serious about settling down (Gong et al., 2016). The uniquely certain entity in our life world, which is time, becomes unpredictable and uncertain when knowing you will experience chronic illness in the future. It seemed that managing the

uncertainty and unpredictability of time in the pre-symptomatic stage involved individuals taking back control over key decisions and their pathway in life.

As indicated in theme 2, one way that participants managed the unpredictability of living in the pre-manifest stage was to distance themselves from the reality of HD. Distancing oneself from HD seemed to be an antidote to the biological disruption described above, where participants actively chose to see themselves as someone without the HD gene expansion. Often this was because they did not have symptoms and could therefore push any potential implications to the future and focus on their symptom free present. In a study which identified individuals' perceived risk of developing the condition both before and after predictive testing, many individuals did not show the expected increase in risk perception following a positive test result (despite being told what the positive test result meant), suggesting a number of factors influence whether risk is perceived to be salient for the individual (Stuttgen et al., 2018). In the study by Stuttgen et al. (2018), factors such as hope for a cure, optimism that one might not develop HD even after a positive result, doubting the accuracy of the result and an inability to accept a positive result were all important components of reducing risk perception for participants. The participants in the study by Stuttgen et al. (2018) were interviewed in the immediate aftermath of the results, which may have influenced their responses, unlike the participants in the current study who were interviewed at least one year following the test. Indeed, none of the participants in the present study questioned the accuracy of the test or totally denied the positive result; instead, denial functioned as a coping strategy which enabled them to live as if they were free of the HD gene expansion. Nevertheless, similar parallels can be seen across the studies; hope for a cure and optimism that they might not develop symptoms were also prominent themes in the participants' accounts in the current study.

The complexity of risk perception is also evidenced in a study of individuals who had not yet experienced predictive testing but were at risk because of family history of HD, where numerous factors were identified as influencing an individual's perceived risk status i.e. how relevant the fact they were at risk for HD was in their lives (Etchegary, 2011). Interestingly, in the study by Etchegary, aspects of an individual's family history of HD (such as age of onset and disease progression) were particularly important in influencing perceived risk status and were noted as a primary zone of relevance in the study. This is similar to the present study, in which parental age of symptom onset and family history of the disease were important factors in enabling participants to distance themselves from their own symptom onset. The present sample differs from the sample in the Etchegary study, some of whom had tested negative for the disease or had not received test results; thus, caution should be warranted in drawing parallels with this research. Nevertheless, the results of Stuttgen et al. (2018) suggest that an individual's risk perception is complex and influenced by a variety of factors, unlikely to only be related to the genetic test results. Indeed, where participants had a living relative with disease, a person's own risk after testing positive for the condition may not be the priority (Sanders et al., 2003) and many of the participants in the present study described focusing on their relative's illness and helping them to cope, rather than their own pre-manifest status. Risk saliency is clearly influenced by a diverse range of social and personal circumstances which go beyond one's individual assessment of risk; when risk saliency is low, an individual may be able to manage the uncertainty of the pre-manifest stage by distancing themselves from their own genetic status. This distancing is evidenced too in a study of pre-symptomatic individuals (Hagberg et al., 2011), where many participants admitted that they avoid thinking about the onset in order to cope, instead holding onto thoughts that they would not develop HD or would not become ill.

Some individuals even described avoiding all contact with a physician and HD organisations because they did not want reminders about being a mutation carrier or visualise what might be in store for them. It would seem that distancing oneself from HD allows individuals in the pre-manifest stage to redirect their anxieties about an uncertain future, instead focusing on more positive or productive means.

An alternative way in which individuals managed the uncertainty was by holding onto hope. By holding onto hope for future medical advances which might prolong or even eliminate symptom onset, participants described how hope allowed them to live each day as if they did not have the gene expansion. Hope is an internal response which develops over time and helps individuals to deal with difficult and unexpected circumstances (Rustøen & Moum, 1997). In holding onto hope, an individual can visualise a positive outcome to their situation and believe in their ability to overcome the challenges presented to them (Acquaye et al., 2016). For individuals diagnosed with a life-threatening illness, with limited opportunities to improve the expected outcome of their condition, hope offers a means to gain some agency over the situation and results in meaningful outcomes for the individual (Acquave et al., 2016). With drug therapies which aim to slow disease progression being developed and evaluated for the first time (Dash et al., 2020), there is cause for hope in HD populations and hope supported participants in the present study to maintain a healthy coping response despite the adversity they faced.

Closely related to hope and the management of uncertainty was the ability to maintain a positive outlook, discussed in theme 3, which allowed individuals to embrace their present and lead a meaningful life. Embracing the present is reflected in another study of pre-manifest individuals, where individuals described how having a positive gene test had encouraged them to re-evaluate the things in their life which they found meaningful and engage with valued activities

(Chapman, 2002). Similarly, in a study by Hagberg et al. (2011), which explored the longer term experience of having the gene expansion, many individuals described some of the positive aspects about taking the test. In that study, participants described using the opportunity to live their best lives whilst they were still healthy and some also described how the test results had brought their family closer together. The test results prompted those participants to recognize how precious life was and the benefits which had come about following the test results. Similarly, gratitude and benefit finding were also important components of positivity in the present study, where individuals described being grateful both for their symptom free functioning and for family and friends. Gratitude is typically considered to be an important factor in an individual's ability to find benefit after a traumatic experience (Peterson & Seligman, 2003) and may act as a protective factor in buffering the effects of adversity on a person's wellbeing (Tugade & Fredrickson, 2007). In focusing on the present rather than being focused on their genetic status individuals may be able to direct their focus to meaningful activities and interests, experiencing renewed energy as a result (Cordova et al., 2001). This ability to find greater appreciation of life and a renewed focus on the present is not just limited to older participants but has also been identified in young people's experience of living in the pre-manifest stage (Gong et al., 2016). In the study by Gong et al., none of the participants regretted their decision to take the test, and the majority were able to identify positive impacts of the result, such as identifying clearer priorities for life, living in the moment and letting go of trivial worries (Gong et al., 2016). Embracing the present and finding meaning is one way to find hope for the future and holding a positive outlook in life worked well for many of the participants.

Many of the participants in the present study had chosen testing as a way to gain certainty of their situation; yet, the test results prompted a different form of uncertainty which participants

had to learn to accept. The desire to resolve uncertainty was also true of the participants in the study by Hagberg et al. (2011), where the majority of the participants had chosen testing in order to resolve uncertainty. Interestingly, in that study, six of the ten participants felt relieved following the test results and more secure about what to expect from the future, thus suggesting their motivations for testing had been successful in that regard. Nevertheless, most of the participants in that study also reported the difficulties they experienced in coping with the positive test result, especially in the first two years. They also had to cope with many obstacles even after accepting the positive test result and successfully adapting required an ongoing period of negotiating uncertainty. This understanding could be helpful for genetic counsellors in supporting those considering genetic testing to recognise that testing does not always offer the certainty individuals and their families seek. Genetic counsellors could provide a valuable role in the provision of information exploring testing, and the possible implications of a positive result, recognising that there may be a diversity of factors which influence an individual's ability to cope with a positive result (Stuttgen et al., 2018). In taking the test, individuals seek to have the power to predict and to make the prediction itself more manageable-to give an identity to what otherwise remains elusive and unmanageable (Chapman, 2002). Such motivations for testing may not always recognise the implications of testing positive for HD on an individual's identity, their relationship to their body, their relationships with others and the uncertainty which the positive gene status creates. It is essential therefore that genetic counsellors provide individuals with the space and time to adjust to the understanding that they are at risk, to reflect on the nature of the disease itself, and to fully explore what it might mean to test positive. Only then can professionals fully prepare individuals for the potential implications of the results and sustain hope and optimism in the face of uncertainty.

### **Clinical implications**

It is important for genetic counsellors to appreciate that salience of genetic risk is likely to differ according to zones of relevance over the course of an individual's life, and to provide follow up support in accordance with this. Genetic counsellors should use this understanding in order to support people proactively, helping individuals to consider the points in their life when they may experience more distress from a positive test result. Although much of the research on predictive testing for HD suggests that most people cope well with the results and do not experience psychological distress, distress may occur later or at certain time points in an individual's journey with HD (Timman et al., 2004). Supporting both those who are undergoing testing, as well as providing follow up support where necessary, should be an important component of psychological support. Given the aforementioned benefits of diminished risk saliency for those in the pre-manifest stage, support which helps individuals to reduce the relevance of their risk or heightens the relevance of other aspects of their lives, may be of value. Techniques derived from acceptance and commitment therapy (ACT) such as cognitive defusion (a process which teaches individuals to take a step back from their thoughts, recognising that their thoughts do not need to dominate their behaviour) can help individuals to manage difficult thoughts and anxiety and refocus their attention to alternative aspects of their experience (Harris, 2009). Supporting individuals to cognitively defuse may help individuals in the pre-manifest stage to manage heightened risk saliency and to focus on the positive aspects of their illness experience.

Many of the participants attributed subtle changes in functioning to HD, and those who engaged in symptom watching were likely to experience heightened risk saliency and increased distress. Similar findings were reported by Cox and McKellin (1999) in their interviews with

people at risk for HD and in the findings of Etchegary (2011). Additionally, in the study by Hagberg et al. (2011), individuals used their own experience of affected relatives to predict what their own symptoms might look like and described more anxiety about what the future might hold, in particular, when and how the disease will begin. One of the participants in the study by Hagberg et al. also described how they felt more psychologically unstable because of the inevitable fate of HD. So too in the present study, where risk saliency was high, many experienced anxieties related to future deterioration. The process of managing this anxiety often involved sharing their concerns with close family members and friends. Genetic counsellors could play an important role in supporting the sharing of individual concerns. In a recent study by Stopford et al. (2020), a genetic counsellor and clinical psychologist delivered group based one off sessions using collective narrative practices for individuals who had received a positive test result, in which individuals were supported to share their experience of testing with other pre-manifest individuals. Participants' experience of the group was overwhelmingly positive, where they described how narrative storytelling has supported them to re-discover their strengths and resilience, find connection with others and embrace their identity as having pre-manifest HD. Implementing group narrative interventions may be an important part of the predictive test counselling support offered for Huntington's disease and could also be adapted for use in other genetic conditions (Stopford et al., 2020).

### **Limitations**

Some limitations were present in this study. Participants were largely recruited from a flyer advertised on the HDA website, which may have biased the sample to individuals who were already engaged with the HD community and thus more motivated to increasing awareness and understanding of HD. As a result, their attitude toward living in the pre-manifest stage may not

generalise to others in the pre-manifest stage of HD. Secondly, as participants were required to proactively express their interest to participate, it is likely that the participants felt able to talk about their experiences and thus may have been coping better mentally and physically than the general population of HD gene expansion carriers. Indeed, some participants were recruited from a clinic where active psychology involvement is offered for all those who have tested positive for the HD gene expansion, which is unusual in the UK. Nevertheless, some of the participants were deliberately recruited from the HDA community as an alternative to the NHS clinic. Whilst again individuals who participate in the HDA community may be those who are coping better with the impact of the test results, such individuals had not received the same level of psychology input, suggesting the results may not just be applicable to those receiving psychological support.

It was notable that the participants in the study appeared to represent a subset of people whose level of positivity and hope was comparatively high compared to individuals in studies assessing the consequences of testing positive for the gene expansion, where high levels of anxiety and depression post-testing have been observed (Berrios et al., 2002; Almqvist et al., 2003). Although the relatively high levels of optimism and hope could be explained by multiple factors it may be that, in self-selecting to participate, individuals were functioning better than many of those living in the pre-manifest stage.

IPA research focuses on the examination of a small number of participants in depth, which limits the generalizability of the results. The aim of this study was not to explore the experience of all those living in the pre-manifest stage of HD, but to gain a richer understanding of the experience of a small number of individuals in the UK as situated within their social context. Nevertheless, drawing upon the current findings, the existing literature and author

reflections on the research can help to accumulate a body of knowledge which allows for theoretical generalizability to be achieved (Smith & Osborn, 2003).

### **Future research**

It is noteworthy that many of the participants seemed to focus on the success of advancing medical research and hopes for the drug trials, privileging knowledge which allowed them to remain optimistic, despite the inevitability of their prognosis and the limited success of current research. For the participants in the current study, this optimism seemed to serve to cultivate hope and embrace their present as symptom free, despite their awareness of their genetic status and the likelihood that they would develop symptoms. Researching the longitudinal consequences of risk perception which focuses on optimistic outcomes at the expense of consideration of an inevitable prognosis would be useful to better understand the potential value of this approach. It has been suggested that high levels of optimism may prevent people from feeling the need to self-manage their condition or take action to minimize health related deterioration (deRidder et al., 2000). Nevertheless, optimism has been shown to be beneficial in helping to protect against the effect of negative illness perceptions (Hurt et al., 2014), highlighting that some level of optimism is important. It seemed that for the participants in the current study, optimism worked well to support their wellbeing, although it may be anticipated that such optimism may be more challenging the closer an individual gets to manifest HD. Future research needs to better understand the relationship between levels of optimism and clinical outcomes for those undergoing genetic testing for HD. This understanding might prompt consideration of how optimism might contribute to wellbeing in other neurodegenerative conditions, including those in which the genetics are less certain regarding disease development,

but in which disease progression and prognosis are similarly uncertain (e.g. familial dementias and amyotrophic lateral sclerosis).

### **Conclusion**

This study has highlighted the difficulties experienced by individuals when adjusting to the pre-manifest stage of HD. The results found that the experience of living in the pre-manifest stage of HD involves navigating the difficulties of feeling limited by the time they have left symptom free by holding onto hope for future treatments and in making the most of their symptom free present. An individual's perceptions of their genetic risk status, and the saliency of living as 'pre-manifest', are both complex factors which require a nuanced understanding of the individual's context. In developing our understanding of what it means for an individual to live as 'pre-manifest' and the ways in which they maintain their wellbeing, we may be better equipped to support those who show more difficulty in managing their experiences.

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Table 1: Participants' demographics

<b>Participant</b>	<b>Gender</b>	<b>Age Range</b>	<b>Marital status</b>	<b>Children</b>	<b>Time since genetic test</b>
<b>Paul</b>	Male	40-44	Married	One	3 yrs
<b>Angie</b>	Female	35-39	Single	None	4 yrs
<b>Daisy</b>	Female	30-34	Married	Two	5 yrs
<b>Catherine</b>	Female	35-39	Single	None	6 yrs
<b>Steve</b>	Male	45-49	Co-habiting	One	3 yrs
<b>Joanna</b>	Female	25-29	Married	Two	2 yrs
<b>Carol</b>	Female	45-49	Married	Two	4 yrs
<b>Hannah</b>	Female	25-29	Cohabiting	One	4 yrs
<b>Sharon</b>	Female	35-39	Married	None	8 yrs
<b>Jake</b>	Male	40-44	Married	None	10 yrs