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Genetic testing for Huntington's disease: a thematic analysis of online support community messages

Author names and affiliations.

Richard M. Smedley [a], Neil S. Coulson [a]

[a] Division of Rehabilitation and Ageing, Queen's Medical Centre, Nottingham, UK

Corresponding author.

Dr Neil S. Coulson

Room B127 Medical School

Division of Rehabilitation & Ageing

Queen's Medical Centre

University of Nottingham

Nottingham

NG7 2UH

UK

+44 115 846 6642

Abstract

Huntington's disease is a fatal late-onset genetic illness that causes motor, cognitive and psychiatric disorders. Individuals considering genetic testing may benefit from online social support. This study investigates how genetic testing is discussed within health forums. 337 messages written by 58 individuals were analysed using deductive thematic analysis. Discussions examined three themes: deciding to be tested (enquiring about symptoms, starting a new family), preparing for the test (information seeking, attending appointments), and receiving the results (positive results, negative results). Forums can reduce the uncertainty of ambiguous symptoms, and provide ongoing personalized support before, during and after a genetic test.

Keywords: Huntington's disease; online support community; thematic analysis; genetic testing; genetic counselling.

Introduction

Huntington's disease

Huntington's disease (HD) is a fatal, hereditary, degenerative disease caused by an expanded CAG repeat on the HTT gene; CAG counts of 6-26 are normal, 27-35 are unstable and could expand in future generations, 36-39 may cause HD, and 40+ always cause HD (Dayalu and Albin, 2015; Imarisio et al., 2008). It is a relatively rare illness with an estimated prevalence of 6.68 per 100,000 in the United Kingdom (Rawlins et al., 2016). Males and females are equally at risk (Frank, 2014), the onset of symptoms can occur at any time between infancy and old age (Adams et al., 1988; Roos et al., 1991; Walker, 2007), the mean age of onset is 40 years (Roos et al., 1991), and the median illness duration is 16.2 years (Roos et al., 1993). When an individual becomes symptomatic, HD is always fatal unless the individual dies of an unrelated cause such as cancer before dying of HD (Solberg et al., 2018; Sørensen and Fenger, 1992).

Symptoms consist of motor, cognitive and psychiatric disorders (Dayalu and Albin, 2015; Roos, 2010). Motor disorders include muscle twitching, unsteady gait and difficulty speaking. Cognitive

disorders affect concentration, memory and attention, and eventually lead to dementia. Psychiatric disorders can include depression, anxiety, apathy and obsessive-compulsive behaviours, and psychosis in the later stages.

Genetic testing

Individuals have a 50% risk of inheriting HD if one of their parents has the faulty gene, and those with untested parents have a 25% risk if one of their grandparents has the faulty gene (Craufurd, 1996). Undergoing a genetic test can lead to ethical dilemmas for at-risk individuals, particularly if other family members prefer not to know their disease status (Taylor, 2004). For example, a pregnant woman might want to have her fetus tested to ensure the next generation is disease-free even though her at-risk male partner does not want to know his disease status. If the fetus tests negative then her partner's disease status will remain unknown, but if the fetus tests positive then it will confirm that her partner carries the faulty gene. This can lead to difficulties concerning if, when, how and to whom the results should be disclosed (Tassicker et al., 2003).

Some at-risk individuals also find it difficult to interpret how factual information about HD applies to their own circumstances. For example, Smith et al. (2013) reported that untested individuals who already have children are sometimes reluctant to undergo testing because they mistakenly believe that a positive test result would increase the probability of each child having the faulty gene. As explained above, the children of parents who have tested positive have a 50% risk of inheriting the gene and the children of untested parents have a 25% risk. Some untested parents believe that if they undergo testing and receive a positive result then this will increase each child's risk from 25% to 50%, and they understandably do not want to do anything that might put their children at increased risk. However this misconception arises from a misunderstanding of genetic risk and, in reality, each child was born either with or without the faulty gene. Undergoing testing after having children does not change the existing genetic status of each child.

For individuals and families who are at-risk of developing HD, there are a number of possible genetic tests that can be used. These fall into three broad categories: prenatal tests, predictive tests and diagnostic tests. First, prenatal tests are used to determine if a fetus carries the faulty gene, and there are a number of options for doing this. Direct testing reveals the disease status of both fetus and parent (Simpson and Harper, 2001), exclusion testing determines if a fetus has a low/high risk of HD without revealing the parents' status but could lead to a healthy fetus being terminated if a high risk fetus does not have the faulty gene (Simpson and Harper, 2001; Tyler et al., 1990), and pre-implantation genetic diagnosis (PGD) transfers disease-free embryos into the womb without revealing the parents' status (Sermon et al., 2002). Second, predictive tests determine if a symptom-free individual has inherited the faulty gene that will cause them to develop HD at some point in the future (Baig et al., 2016). Third, diagnostic tests are used on individuals who have already developed HD-related symptoms to confirm if they have got HD (Craufurd et al., 2015). It is estimated that 0.9% of HD genetic tests are prenatal, 62.3% are predictive and 36.8% are diagnostic (Creighton et al., 2003).

The testing process comprises three broad stages: deciding to be tested, preparing for the test, and receiving the results. Deciding to be tested is a major decision for those who are at-risk (Taylor, 2004). Individuals have a right not to know their genetic status (Andorno, 2004; Asscher and Koops, 2010), a right that is strongly defended by some at-risk individuals (Taylor, 2004). Choosing not to be tested lacks the finality of being tested; an untested individual can undergo testing at any future point, whereas a tested individual cannot unlearn the knowledge of their disease status (Cox, 2003). Deciding to undergo testing is often seen as a moral dilemma where individuals must balance their perceived level of risk, the possible consequences of knowing their HD status, and responsibilities towards their partner, relatives and the next generation (Cox, 2003; Klitzman et al., 2007; Smith et al., 2002).

Preparing for the test involves undergoing genetic counselling. This ensures the individual understands the molecular genetics of HD, explores their motivations for being tested, and considers possible consequences that could arise (Walsh, 1999). Counselling sessions typically provide

biomedical or educational information rather than exploring the individual's psychosocial needs (Meiser et al., 2008). Counselling requires multiple appointments taking several weeks or months to prepare the individual and ensure that follow-up care is provided if they are symptomatic (Hawkins et al., 2011). The individual then gives a final confirmation about completing the test, which may produce a positive, negative or indeterminate result (Sarangi et al., 2004).

Receiving the results is an important transition point in the life of an at-risk individual (Tibben, 2007). Both carriers and non-carriers experience increased distress after receiving their results (Meiser and Dunn, 2000), with some individuals maintaining the appearance of coping while struggling to adjust to knowing their status (DudokdeWit et al., 1998). They may need to re-evaluate their future plans and decide if, when and how to disclose their test results, particularly if they test positive and have already got children (Andersson et al., 2012).

Few studies have examined genetic counselling outcomes in terms of helpfulness or satisfaction ratings by individuals who are undergoing a HD genetic test. However, a study by Jones and Macleod (2014) reported that 93% of individuals undergoing genetic counselling for HD had a 'good' or 'very good' relationship with their counsellor and 84% thought the number of counselling sessions was 'about right', while 32% indicated that the testing process took too long and 11% had difficulty engaging with the process. In addition, some individuals reported that they would have appreciated a more tailored approach together with access to information resources such as videos of people discussing the advantages and disadvantages of testing. Therefore, for some individuals who are undergoing the testing process, it appears that their informational and psychosocial needs are not being fully met through genetic counselling. Furthermore, there is a lack of ongoing professional support for individuals who have undergone testing but are not yet symptomatic (Andersson et al., 2016).

Taken together, this suggests that many individuals do not have their psychosocial needs adequately met in the testing process. Additional sources of information and support may benefit individuals at

all stages of the testing process, including those who have learned that they are at-risk of HD and struggle to correctly interpret how this might apply to their personal circumstances, those who are considering a genetic test or undergoing the testing process, and those who have been tested but are not yet symptomatic. Consequently, online support communities might be a useful resource for individuals during all stages of the HD testing process. This would provide individuals with a way to communicate with others who have been through similar testing-related decisions and dilemmas, to obtain information, advice and support about all aspects of the testing process.

Online support communities

Online support communities, herein called forums for brevity, are used to exchange messages with others who have experienced similar health-related issues. Forums can be used at any time of the day or night to ask questions, obtain information and access support as-and-when required (Coulson and Knibb, 2007; Malik and Coulson, 2008). The individual might not know anybody else who is affected by HD, and forums provide direct access to others who are in a similar situation and facing similar challenges (Coulson et al., 2007). Members often adopt an anonymous online persona (Brady et al., 2016), which may aid discussing sensitive or distressing topics (Buchanan and Coulson, 2007; Coulson and Knibb, 2007). Individuals affected by HD are sometimes reluctant to attend face-to-face support groups because they are uncomfortable meeting others with an advanced stage of the illness (Dawson et al., 2004), so the anonymity of forums may be particularly beneficial.

Discussions within forums take the form of 'threads', where each thread is a self-contained conversation consisting of messages written by one or more members (Smedley and Coulson, 2018). An individual typically starts a new discussion by posting an initial message, and others post replies that are closely tailored to the individual's needs and circumstances (Coulson et al., 2016; Liang and Scammon, 2011).

HD forums can provide a wide range of information, advice and support (Coulson et al., 2007). Individuals who are considering a HD genetic test, undergoing genetic counselling, or adjusting to

receiving a positive or negative test result may benefit from the tailored assistance provided through forums, but little is known about how HD genetic testing is discussed within forums. Further research is needed to understand how forums can make a positive contribution to the wellbeing of individuals affected by HD, and to also provide valuable information to healthcare professionals.

Aims

The aim of this study was to investigate how the process of genetic testing is discussed within HD forums.

Methods

Data collection

Data was analysed from the asynchronous forum of a UK-based organisation supporting individuals affected by HD. Their forum provided multiple discussion boards covering various aspects of living with HD, including a dedicated board for testing-related issues. This was used by at-risk individuals who are considering being tested, those undergoing testing, and members who have previously been tested. The researchers selected this specific forum as a data source because they were already familiar with the forum and they had used it in previous research.

All messages posted on the testing board during the 13-month period from April 2012 to May 2013 were selected for analysis, producing an initial dataset of 43 threads (371 messages). Off-topic threads not directly related to genetic testing were excluded, leading to the removal of 6 threads (34 messages). The final dataset comprised 37 threads (337 messages). Threads were downloaded and saved reproducing each message verbatim including the original formatting, layout and emoticons (Smedley and Coulson, 2018). The analysis was carried out immediately after data collection was completed and an initial draft was written straight away, but time constraints on other projects meant that the paper was not prepared for publication until 2018.

Participants

58 unique usernames were identified in the dataset. Demographic information was limited because forum members often maintain an anonymous online persona (Brady et al., 2016), however some information was obtained by examining the content of their messages. Gender was identified for 43 participants (11 males, 32 females). Health status was identified for 48 participants (35 individuals were diagnosed/at-risk of HD, 13 were the partner of somebody diagnosed/at-risk).

Analysis

Messages were analysed using deductive thematic analysis (Braun and Clarke, 2006). Thematic analysis was used because this technique examines the patterns (or themes) that are present within textual data, and a deductive approach was employed to analyse the data in the context of three aspects of the testing process: deciding to be tested, preparing for the test, and receiving the results. Both authors have extensive experience in conducting this type of analysis with online data. First, each thread was read multiple times to gain familiarity with the data and identify the broad discussion topics related to these themes within that thread. Initial ideas and observations were noted during this stage. Second, constant comparison was used to refine and develop these notes by moving back and forth across the dataset comparing threads that discussed these issues. After further discussion, both authors identified illustrative extracts and reviewed the themes for coherence. It is unclear whether data saturation was achieved. It can be difficult to achieve data saturation when analysing forum messages because there is always a possibility that downloading more messages may have yielded additional insights (Im and Chee, 2006).

Ethical considerations

Ethical approval for this study was granted by the University of Nottingham, and the study complied with British Psychological Society guidelines for internet-mediated research (British Psychological Society, 2017).

Informed consent and anonymity can pose ethical dilemmas in forum studies (Smedley and Coulson, 2018). Consent requests can trigger feelings of anger and distrust among members (Eysenbach and

Till, 2001; Hudson and Bruckman, 2004), and some members may be impossible to contact or reluctant to reveal personal information needed for consent (Roberts, 2015). Consequently data was collected from a publically-viewable forum where anybody can read messages without needing to register or obtain a password. It is sometimes unclear whether the members of a public forum regard it as being a public or private place (Bond et al., 2013; Lomborg, 2013), however using a public forum removed the need to obtain consent because attempting to obtain consent may have caused more harm than the protection it is intended to provide (Robinson, 2001). In addition, the use of public domain data sources, such as the identified forum, is considered acceptable by the British Psychological Society provided appropriate safeguards are put in place to ensure that the forum is not named and no personally identifiable information is reported (British Psychological Society, 2017). Therefore, in order to ensure the anonymity of forum members whose postings had been included in our dataset, we paraphrased extracts such that the original meaning was retained but the quote was not traceable to the originating forum (Roberts, 2015).

Results

Three aspects of the HD testing process were examined: deciding to be tested, preparing for the test, and receiving the results.

Aspect 1: Deciding to be tested

Common initiating events for considering a HD genetic test were noticing symptoms that might be HD-related and wanting to start a family.

Enquiring about symptoms

There were several threads where at-risk individuals described symptoms they were currently experiencing, and enquired if this might indicate they have inherited the faulty gene. The following quote is a typical example of this type of enquiry:

> *My mother had eye twitches for many years before developing full-blown HD. I have recently noticed twitches in my eyes and fingers, particularly when I am relaxed, reading or watching television. I am concerned it might mean I have got HD. Should I get tested?*

Replies to these messages were broadly grouped into two subcategories. In the first subcategory, individuals responded by relating the original query to their own concerns and experiences. Sometimes others would comment that they had joined the forum because they too were worried about symptoms ("*I joined because I had exactly the same concerns as you*"). In-groups and out-groups were occasionally used to emphasize these shared concerns and distinguish them from the experiences of those who are not at-risk ("*Outsiders will never understand how stressful it is worrying about symptoms and struggling with the guilt that I may have passed HD on to my children, while simultaneously feeling too scared to get tested*"). More commonly, respondents talked about their own symptoms and, where possible, they tried to compare those symptoms with what their affected parent experienced when they became symptomatic ("*I wish I could remember the symptoms my mother had when she first developed HD. I get twitches, stumble and trip over things. Is it nothing, or could I have HD?*").

The second subcategory of reply provided factual and educational information about symptoms. Individuals often explained that it is normal for at-risk individuals to worry about symptoms ("*Symptom hunting is common among people who are at-risk, I often do it too*"). They discussed the age when individuals typically become symptomatic and used this to assess whether their symptoms were likely to be caused by HD ("*Your mother was diagnosed a few years ago and you are only thirty, so it will be a long time before you develop any symptoms*"). There were frequent discussions about alternative explanations for what might be causing their symptoms ("*Many different things could cause the symptoms you described. Just feeling anxious about HD could be causing your twitches. Being tested is the only way to be certain*").

Starting a new family

Another common reason for deciding whether to be tested was starting a family. Discussions on this topic were grouped into two subcategories: unplanned pregnancies and family planning.

The first subcategory was concerned with unplanned pregnancies. Threads would typically begin with an individual discovering that they are pregnant and describing their emotions or asking for advice, as illustrated by the following quote:

> *I have just found out that I am 10 weeks pregnant with my first child. My partner immediately got tested and has just received a positive result for HD. We have arranged a CVS test but I feel petrified about what might happen next…*

Replies to this form of enquiry would explain the full range of available options. These included leaving things to chance and carrying the baby regardless ("*I don't want to know my status and I don't believe in abortions, so we let nature decide*") and having the fetus tested ("*For us, testing was the only option. We would prefer to be childless than inflict HD on another generation*"). Respondents emphasized how important it was for each individual to make their own decision about testing and what to do if the results are positive ("*IF the test is positive then YOU alone must decide whether to keep it. I don't have the right answer. Morally it's a minefield and others may try to sway you, don't let anyone tell you what to do*"). In the thread quoted above the original message contributor had already decided to have the fetus tested, which prompted several messages offering emotional support ("*I'm sorry to hear what you're going through, it is understandable that you are finding it so difficult*").

The second subcategory was concerned with family planning. These discussions typically focussed on the queries and dilemmas that individuals faced when considering starting a family, and the consequences of having children. The following quote is typical of the quandaries associated with this:

> *I'm unsure about being tested. An important consideration is the possibility of starting a family. If a prenatal test comes back positive then I must have HD too. Would that mean I'm*

> *using my unborn child to test me as well? Is it irresponsible having children without being tested first?*

It was evident from reading the replies to such posts that members had a range of responses. Like with unplanned pregnancies, some individuals advocated having children without any form of genetic testing, arguing that leaving things to chance would give their children the same opportunity to live that they themselves had ("*we were given the chance to live, so why shouldn't our children?*"). This was sometimes accompanied by the hope that better treatments might become available before their children become symptomatic ("*by the time my children grow up, there might be a way to treat HD or even a cure*"). For some individuals, this could produce terrible feelings of guilt:

> *That is how my mother-in-law felt, believing the cure would be here before it affected her son. She now has survivor's guilt, watching her husband fade away while monitoring her son for any tiny hint of HD.*

Others described how they had the genetic test in adulthood and discovered that HD had been naturally eliminated from their family. Even though older relatives were still affected, they could have children without worrying about future generations inheriting the disease ("*My sister and I both tested negative, so HD has stopped with our father. We are now free but HD still continues with him*").

More frequently, individuals considering starting a family were advised to use pre-implantation genetic diagnosis (PGD), where only disease-free embryos are implanted into the womb. This technique can still be used if the individual prefers not to know their own disease status ("*Having the gene doesn't stop you having children. You can use PGD to ensure the baby is HD-free. Don't lose hope, there is always a workaround*"). Genetic testing was seen by some individuals as a way to eliminate the faulty HD gene from families without needing a cure ("*We are all responsible for the next generation. We should all ensure our children are HD free and eliminate the disease without needing a cure*").

Aspect 2: Preparing for the test

After deciding to be tested, members described how they wanted to learn more about genetic testing and the genetic counselling that forms a central part of this process.

Information seeking

A range of information and advice was requested through the messages posted to this board of the HD online community. For some, there was a need to ask about practical aspects of the process, such as how to initiate a genetic test ("*How do I arrange to be tested? Do I just make a GP appointment and they contact the required people*?"). More commonly, individuals wanted to learn about counselling and testing to prepare for their first appointment after being referred to a specialist ("*I'm going to the genetics department this week for my first appointment. Any advice on what to expect?*"). Replies used informational support to explain what the counsellor was likely to discuss during these sessions in addition to describing the emotional aspects of preparing for the outcome ("*The counsellor will explain about HD and CAG repeats, and make sure you are planning for the future. It can be devastating, people who test negative can still struggle if other relatives test positive. The impact of various outcomes is hard to put into words*").

Several messages discussed how long it takes before receiving the results. The number of appointments could vary according to personal needs and circumstances, with some individuals needing more sessions than others:

> *The timeline is roughly this: 1st appointment with genetic counsellor. 2nd appointment 2-3 months later, with genetic counsellor and a consultant. First blood sample taken and stored. 3rd appointment with the consultant, second blood sample taken, consent forms signed. 4th appointment to get the results.*

Some individuals questioned the amount of time needed to go through the testing process and asked whether genetic counselling was genuinely necessary ("*I know I want to be tested so it seems silly having counselling before getting the results. Why can't I just have the test and get it over with? If the*

results are negative then waiting adds to the stress"). Frustration with the need to attend counselling sessions led some individuals to ask if tests can be conducted privately ("*Does anyone know if you can be privately tested? How much does this cost, and how do you go about it?*"). The replies to these specific messages typically used informational support to discourage individuals from rushing through the testing process, and reminded them to spend time considering how the results might impact upon the rest of their lives so that they make the right decision and do not later regret being tested:

> *I started off thinking men don't do counselling. They insisted I attend all the sessions, and they were right. It gave me time to think things through and reflect on the implications for my family. I did the right thing, after all the results will affect the rest of my life.*

Attending appointments

Several individuals described their feelings about attending their first appointment. It was common to report experiencing stress, anxiety and trepidation about being tested ("*Went for my first appointment this morning, and the anxiety nearly got me. It felt like I was on death row, I was terrified and wanted to go home*"). Emotional support was often provided in response to these types of messages ("*I totally understand how you felt, and I hope it all goes well*"). Perceptions of the counsellors were nearly always positive with individuals describing their appreciation for the information and support that was provided ("*I was very stressed going to the hospital. After a short wait, the consultant was amazing. She was very warm and gave me lots of helpful information*").

Despite the complex emotional dilemmas associated with being tested and preparing for news that will affect the rest of their life, some individuals pointed out that being tested does not change whether or not they have the faulty gene:

> *I wish I could say 'good luck' but it isn't about luck. If you have the gene then you were born with it. It's a part of you that cannot be changed. The only thing that will change is knowing about it.*

Aspect 3: Receiving the results

The anxiety and emotional strain of receiving the test results was evident across many of the messages posted. These messages illustrated the complexities around 'making sense' of the outcome and what it might mean for individuals, couples and families.

A number of individuals described the emotional impact of waiting to receive their test results. This appeared to be a particularly stressful time, with individuals preparing themselves for a possible positive result ("*My results are due tomorrow. I'm struggling with nerves, and my arms are shaking now that the answer is almost here. I'm preparing for the worst possible outcome*"). Replies used informational support to provide useful suggestions to prepare for receiving their results, to help individuals prepare themselves for what could be potentially life-changing information ("*Take time off work both before and after getting the results, and consider going away to keep yourself busy. Make sure you have the support that you need. Sleeping pills may help with nerves*"). Other replies used emotional support to provide encouragement during what was an inherently stressful time ("*Good luck, we're all thinking of you. Tell us how it goes*").

Positive results

Each individual responded differently to receiving their results. For some, a positive result came as a devastating blow despite all the preparations ("*Believe me, learning I was positive felt like being slammed into a wall. I thought I was prepared but I wasn't*"). Others felt ambivalent or relieved after receiving a positive result because it explained their symptoms ("*The consultant did a neurological test at the same time as taking the blood sample, and said it was likely to be HD. It was a relief to finally know what was wrong. The uncertainty was worse*"). Years after receiving a positive result, some individuals still struggled to cope but nonetheless explained the importance of maintaining an optimistic outlook and continuing to live in a meaningful way, as demonstrated by this quote:

> *I wouldn't say I was coping but I'm still symptom free. I go to work, pay the rent, and try to make the best of my circumstances. You have to find a way to live positively. It isn't a death sentence, and there are worse illnesses to have.*

Some individuals underwent prenatal testing following an unexpected pregnancy. Unfortunately, this did not always produce the results that members hoped for ("*The CVS produced a positive result so I'm booked in for a surgical termination. Words cannot describe how I now feel*"). When this happened, respondents would again provide emotional support ("*My thoughts are with you. Keep trying, and one day you will have a healthy baby*").

Negative results

Some individuals who tested negative described feelings of shock and disbelief upon learning they do not have the faulty gene ("*I can't believe it! My results came back negative with a CAG count of 18. I was convinced it would be positive because my brother's results were negative*"). Others responded with relief ("*Just got my results, my CAG count is 16. It feels like a dream, and a big relief after all the anxiety*"). Those with a negative result still sometimes faced ongoing challenges, particularly if others in their family were at-risk or tested positive. HD was still part of their lives, and some individuals felt a responsibility to encourage other family members to be tested ("M*y results were negative but my sister came back positive. What happens next? I had a lucky escape but HD is still part of my life, and I need to convince others to get tested as well*").

A small number of messages questioned the reliability of test results. One individual said that her husband had tested negative and he now wondered if his results could be wrong ("*My husband's results came back negative in [year]. His mother is still symptom-free, yet he is concerned that the results might have been wrong. How accurate is the test?*"). Others replied to these messages by providing assurance about the accuracy of the results ("*At my hospital, they took two blood samples and needed four signatures verifying the results. It is very unlikely that his results were wrong, and his CAG count is well below the threshold for HD*").

Discussion

This study investigated how HD genetic testing is discussed within online support forums, examining three key aspects of the process: deciding to be tested, preparing for the test, and receiving the results.

The first aspect, deciding to be tested, was associated with wanting to know if a symptom might be an early sign of the onset of HD, and wanting to start a family. Enquiring about symptoms was common within the forum, with members describing ambiguous symptoms that may or may not be representative of HD. This appeared to put members in a state of uncertainty, consistent with uncertainty in illness theory (Mishel, 1990). Research indicates that health uncertainty can have a range of causes including experiencing ambiguous symptoms that could be associated with multiple underlying illnesses, and a lack of knowledge regarding the probable onset and development of a health condition (Han et al., 2011). Uncertainty is strongly and positively associated with anxiety (Kuang and Wilson, 2017), indicating that greater levels of uncertainty lead to heightened anxiety. In the present study, individuals may have experienced health uncertainty due to not knowing their genetic status combined with the ambiguous nature of symptoms that might potentially be indicative of HD. The perception that HD is a fatal illness may have further increased the resulting feelings of anxiety.

The lack of treatment options for HD combined with a high probability of mental deterioration can give HD a psychological stigma that may lead some individuals to carefully conceal their status from others (Quaid et al., 2008). The anonymous online personas associated with forums (Brady et al., 2016) may make it easier to discuss challenging, sensitive or stigmatised topics that individuals might not otherwise be able to talk about (Coulson and Knibb, 2007; White and Dorman, 2001). Taken together, health uncertainty combined with psychological stigma may have prompted individuals to join the forum to seek further guidance, information and support. Members in this situation discussed whether it was the right time to consider undergoing genetic testing to reduce or eliminate this uncertainty by confirming if they have got the faulty gene.

Wanting to start a family involved discussing a diverse range of issues with a particular focus on prenatal and predictive testing, whether to prevent their children from inheriting the faulty gene or leave things to chance, and the dilemmas and feelings of guilt arising from these decisions. Similar

findings were obtained by a study interviewing at-risk individuals about reproductive decisions, where participants talked about having children, reproductive options such as PGD, and feelings of guilt (Klitzman et al., 2007). This suggests the current results may broadly reflect the reproductive concerns faced by individuals affected by HD. Whether or not individuals undergo genetic testing, they subsequently tend to feel happy with their decision (Nance, 2017). When at-risk individuals consider starting a family, forums may be an important source of information and guidance to help with reproductive decisions. Indeed, none of the messages indicated that becoming a member had negatively impacted their wellbeing or that they had found the forum unhelpful. That said, it is possible that some members found the forum unhelpful but those individuals may have been less likely to post messages about their experiences (Amichai-Hamburger et al., 2016). For individuals who are comfortable with being tested despite the emotional dilemmas that can arise, using genetic testing to aid reproductive decisions was sometimes perceived as an effective strategy for eradicating the faulty gene from future generations of their family (Turner and Willoughby, 1990).

The second and third aspects of testing were preparing for the test and receiving the results. Individuals discussed the testing process, what happens during counselling sessions, and expressed their emotions about being tested. Replies used informational support to provide factual information and advice, and emotional support when discussing their fears and anxieties. Many threads described the experience of receiving their results, with both positive and negative test results having long-term consequences. Informational support was used to provide advice and coping strategies when preparing for the results, while emotional support helped them adjust to knowing if they had the faulty gene.

These results are consistent with optimal matching theory (Cutrona and Suhr, 1994). This theory argues that individuals will find different types of social support effective depending on what kind of health-related problem they are experiencing, the controllability of that problem, and how it impacts on other important aspects of their life such as personal or professional relationships. In particular, optimal matching theory distinguishes between two support categories: action-facilitating support such as informational support helps the individual by providing the resources needed to solve or

overcome a problem, while nurturant support such as emotional support helps them to cope with their circumstances by providing reassurance and reducing stress.

Informational and emotional support were both commonly used in the forum, supporting the findings of other studies investigating both HD forums and optimal matching theory. Coulson et al. (2007) examined the prevalence of these support categories within a HD forum, reporting that informational support was present in 56.2% of messages and emotional support was present in 51.9% of messages. Similarly, in their meta-analysis looking at support types in relation to different health-related stressors, Rains et al. (2015) found that nurturant support is associated with health conditions that are likely to impact on personal relationships and conditions that might potentially result in death, while informational support is common in conditions with a longer duration. HD is a late-onset disorder (Roos et al., 1991) that can adversely affect relationships (Andersson et al., 2016) and culminates in death (Solberg et al., 2018), thus offering an explanation as to the types of support that were observed in the present study.

The long-term availability of support through forums appears to be especially important. Professional support is typically only provided to individuals while they are undergoing genetic counselling to prepare for being tested, whereas online peer support is available through all stages of the testing process and beyond. Forums provide access to information and support to help individuals decide whether to undergo testing, they act as a supplemental source of support while preparing for the test, and they are a source of long-term help that individuals can continue to access after receiving their results. Individuals who continue using the forum may derive additional benefits as a consequence of providing help to other members, such as those who have not yet undergone testing. This is consistent with the helper-therapy principle, which argues that individuals who help others also help themselves by taking on valued social roles, developing their own coping skills and abilities, and focusing on other people's concerns instead of dwelling upon their own issues (Riessman, 1997).

Forum replies were personalised to each individual's unique needs (Coulson et al., 2016). This may be particularly beneficial to individuals who would like additional information and support that is more closely tailored to their own circumstances (Jones and Macleod, 2014). Genetic counselling is provided by healthcare professionals who might not have personally experienced HD, whereas online support is provided by individuals who have experienced similar challenges and difficulties (Coulson et al., 2007). Interacting with and receiving support from peers who have been through similar experiences may be particularly helpful because they often have a similar perspective about the illness and can relate to what a person is experiencing (Coulson, 2013; Coulson et al., 2016).

Many forums are used by laypeople and their carers for peer support, however some forums are also used by healthcare professionals (O'Grady et al., 2010; van der Eijk et al., 2013). Few studies have examined self-help forums that are used by both peers and healthcare professionals, but it appears that individuals derive additional benefits when healthcare professionals join forums. For example, Vennik et al. (2014) reported that individuals appreciated receiving 'expert knowledge' from professionals alongside the 'experiential knowledge' provided by peers. Experts provided specialist information about medications, treatment options and new findings, while peers discussed ideas and suggestions that might not be evidence-based but which other individuals found helpful for self-managing their health condition. Having both sources of information available side-by-side helped individuals to achieve a deeper understanding of their personal health.

Healthcare professionals are sometimes reluctant to engage with forums and other online resources. For example they might be worried about loss-of-control in consultations or feel that their professional expertise is undervalued (Ahluwalia et al., 2010), or they might not have the time, knowledge, training or computer skills required to recommend or use forums (Anderson, 2008). Despite such reservations, it appears that there is potential for healthcare professionals to play a range of important roles in forums. They could make face-to-face patients aware of the potential benefits of using forums, join forums themselves to provide expert knowledge, and also use forums to assess

whether current clinical practice guidelines are successfully addressing the psychosocial needs of patients.

There are a number of limitations to this study which should be considered. Firstly, the present study analysed all relevant messages posted within a 13-month period. However, this yielded only 337 messages generated by 58 unique community members. As a consequence of this modest dataset, the extent to which the findings can be generalised may be limited. Secondly, whilst males and females have an equal risk of HD (Frank, 2014) and females are slightly more likely to undergo predictive testing (Baig et al., 2016) the data used in this study were from predominantly females, which is consistent with the findings of other forum studies (Coulson et al., 2016; Smedley and Coulson, 2017). The reasons for this gender imbalance are not clearly understood. It may be an oversimplification to assume that females are more sensitive to health issues and/or more likely to adopt caring roles (Manierre, 2015), so further research is needed to fully understand the underlying cause of these gender differences.

Future research could build upon these findings by examining the following issues. First, these results indicate that forum membership may play an important role in all aspects of the HD genetic testing process, but little is known about how forum participation contributes to decision making. Qualitative research could be used to investigate this area in greater detail, perhaps by adapting a longitudinal case study design that follows individuals through all aspects of the testing process and beyond. Second, it is unknown how the experience of undergoing genetic testing differs among individuals who use forums compared with those who do not use them. For example, individuals who receive additional support from online forums may fare better, worse or just the same as those who do not use forums. Third, there is an urgent need for research examining the factual accuracy of postings within HD forums. If individuals are using forums to assist them with genetic testing decisions, aided by postings that may or may not contain potentially misleading information, then this may be of concern for both individuals and healthcare professionals.

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Conflicts of interest

The Authors declare that there is no conflict of interest.

References

Adams P, Falek A and Arnold J. (1988) Huntington disease in Georgia: age at onset. *American Journal of Human Genetics* 43: 695-704.

Ahluwalia S, Murray E, Stevenson F, et al. (2010) 'A heartbeat moment': qualitative study of GP views of patients bringing health information from the internet to a consultation. *British Journal of General Practice* 60: 88.

Amichai-Hamburger Y, Gazit T, Bar-Ilan J, et al. (2016) Psychological factors behind the lack of participation in online discussions. *Computers in Human Behavior* 55, Part A: 268-277.

Anderson AS. (2008) The Internet: friend or foe when providing patient education? *Clinical journal of oncology nursing* 12: 55.

Andersson PL, Juth N, Petersén Å, et al. (2012) Ethical aspects of undergoing a predictive genetic testing for Huntington's disease. *Nursing Ethics* 20: 189-199.

Andersson PL, Petersén Å, Graff C, et al. (2016) Ethical aspects of a predictive test for Huntington's Disease: A long term perspective. *Nursing Ethics* 23: 565-575.

Andorno R. (2004) The right not to know: an autonomy based approach. *Journal of Medical Ethics* 30: 435-440.

Asscher E and Koops BJ. (2010) The right not to know and preimplantation genetic diagnosis for Huntington's disease. *Journal of Medical Ethics* 36: 30-33.

Baig SS, Strong M, Rosser E, et al. (2016) 22 Years of predictive testing for Huntington's disease: the experience of the UK Huntington's Prediction Consortium. *European Journal of Human Genetics* 24: 1396-1402.

Bond SC, Ahmed HO, Hind M, et al. (2013) The Conceptual and Practical Ethical Dilemmas of Using Health Discussion Board Posts as Research Data. *Journal of Medical Internet Research* 15: e112.

Brady E, Segar J and Sanders C. (2016) "I Always Vet Things": Navigating Privacy and the Presentation of Self on Health Discussion Boards Among Individuals with Long-Term Conditions. *Journal of Medical Internet Research* 18: e274.

Braun V and Clarke V. (2006) Using thematic analysis in psychology. *Qualitative Research in Psychology* 3: 77-101.

British Psychological Society. (2017) Ethics Guidelines for Internet-mediated Research. (accessed July 13, 2017).

Buchanan H and Coulson NS. (2007) Accessing dental anxiety online support groups: An exploratory qualitative study of motives and experiences. *Patient Education and Counseling* 66: 263-269.

Coulson NS. (2013) How do online patient support communities affect the experience of inflammatory bowel disease? An online survey. *JRSM Short Reports* 4: 1-8.

Coulson NS, Buchanan H and Aubeeluck A. (2007) Social support in cyberspace: A content analysis of communication within a Huntington's disease online support group. *Patient Education and Counseling* 68: 173-178.

Coulson NS and Knibb RC. (2007) Coping with food allergy: Exploring the role of the online support group. *CyberPsychology & Behavior* 10: 145-148.

Coulson NS, Smedley R, Bostock S, et al. (2016) The Pros and Cons of Getting Engaged in an Online Social Community Embedded Within Digital Cognitive Behavioral Therapy for Insomnia: Survey Among Users. *Journal of Medical Internet Research* 18: e88.

Cox SM. (2003) Stories in Decisions: How At-Risk Individuals Decide to Request Predictive Testing for Huntington Disease. *Qualitative Sociology* 26: 257-280.

Craufurd D. (1996) Huntington's disease. *Prenatal Diagnosis* 16: 1237-1245.

Craufurd D, MacLeod R, Frontali M, et al. (2015) Diagnostic genetic testing for Huntington's disease. *Practical Neurology* 15: 80-84.

Creighton S, Almqvist EW, MacGregor D, et al. (2003) Predictive, pre-natal and diagnostic genetic testing for Huntington's disease: the experience in Canada from 1987 to 2000. *Clinical Genetics* 63: 462-475.

Cutrona CE and Suhr JA. (1994) Social Support Communication in the Context of Marriage: An Analysis of Couples' Supportive Interactions. In: Burleson BR, Albrecht TL and Sarason IG (eds) *Communication of Social Support: Messages, Interactions, Relationships, and Community.* Thousand Oaks: Sage Publications, 113-135.

Dawson S, Kristjanson LJ, Toye CM, et al. (2004) Living with Huntington's disease: Need for supportive care. *Nursing & Health Sciences* 6: 123-130.

Dayalu P and Albin RL. (2015) Huntington Disease: Pathogenesis and Treatment. *Neurologic Clinics* 33: 101-114.

DudokdeWit AC, Tibben A, Duivenvoorden HJ, et al. (1998) Predicting adaptation to presymptomatic DNA testing for late onset disorders: who will experience distress? Rotterdam Leiden Genetics Workgroup. *Journal of Medical Genetics* 35: 745-754.

Eysenbach G and Till JE. (2001) Ethical issues in qualitative research on internet communities. *British Medical Journal* 323: 1103-1105.

Frank S. (2014) Treatment of Huntington's Disease. *Neurotherapeutics* 11: 153-160.

Han PKJ, Klein WMP and Arora NK. (2011) Varieties of Uncertainty in Health Care: A Conceptual Taxonomy. *Medical Decision Making* 31: 828-838.

Hawkins AK, Ho A and Hayden MR. (2011) Lessons from predictive testing for Huntington disease: 25 years on. *Journal of Medical Genetics* 48: 649-650.

Hudson JM and Bruckman A. (2004) “Go Away”: Participant Objections to Being Studied and the Ethics of Chatroom Research. *The Information Society* 20: 127-139.

Im E-O and Chee W. (2006) An online forum as a qualitative research method. *Nursing Research* 55: 267-273.

Imarisio S, Carmichael J, Korolchuk V, et al. (2008) Huntington's disease: from pathology and genetics to potential therapies. *Biochemical Journal* 412: 191-209.

Jones M and Macleod R. (2014) K01 Patient Views On The Delivery Of Predictive Test Counselling Services For Huntington's Disease. *Journal of Neurology, Neurosurgery & Psychiatry* 85: A79.

Klitzman R, Thorne D, Williamson J, et al. (2007) Decision-Making About Reproductive Choices Among Individuals At-Risk for Huntington's Disease. *Journal of Genetic Counseling* 16: 347-362.

Kuang K and Wilson SR. (2017) A meta-analysis of uncertainty and information management in illness contexts. *Journal of Communication* 67: 378-401.

Liang B and Scammon DL. (2011) E-Word-of-Mouth on health social networking sites: An opportunity for tailored health communication. *Journal of Consumer Behaviour* 10: 322-331.

Lomborg S. (2013) Personal internet archives and ethics. *Research Ethics* 9: 20-31.

Malik SH and Coulson NS. (2008) Computer-mediated infertility support groups: An exploratory study of online experiences. *Patient Education and Counseling* 73: 105-113.

Manierre MJ. (2015) Gaps in knowledge: Tracking and explaining gender differences in health information seeking. *Social Science & Medicine* 128: 151-158.

Meiser B and Dunn S. (2000) Psychological impact of genetic testing for Huntington's disease: an update of the literature. *Journal of Neurology, Neurosurgery & Psychiatry* 69: 574-578.

Meiser B, Irle J, Lobb E, et al. (2008) Assessment of the Content and Process of Genetic Counseling: A Critical Review of Empirical Studies. *Journal of Genetic Counseling* 17: 434-451.

Mishel MH. (1990) Reconceptualization of the Uncertainty in Illness Theory. *Journal of Nursing Scholarship* 22: 256-262.

Nance MA. (2017) Genetic counseling and testing for Huntington's disease: A historical review. *American Journal of Medical Genetics Part B: Neuropsychiatric Genetics* 174: 75-92.

O'Grady L, Bender J, Urowitz S, et al. (2010) Promoting and participating in online health forums: A guide to facilitation and evaluation for health professionals. *Journal of Communication in Healthcare* 3: 246-257.

Quaid KA, Sims SL, Swenson MM, et al. (2008) Living at Risk: Concealing Risk and Preserving Hope in Huntington Disease. *Journal of Genetic Counseling* 17: 117-128.

Rains SA, Peterson EB and Wright KB. (2015) Communicating Social Support in Computer-mediated Contexts: A Meta-analytic Review of Content Analyses Examining Support Messages Shared Online among Individuals Coping with Illness. *Communication Monographs* 82: 403-430.

Rawlins MD, Wexler NS, Wexler AR, et al. (2016) The Prevalence of Huntington's Disease. *Neuroepidemiology* 46: 144-153.

Riessman F. (1997) Ten self-help principles. *Social Policy* 27: 6-11.

Roberts LD. (2015) Ethical Issues in Conducting Qualitative Research in Online Communities. *Qualitative Research in Psychology* 12: 314-325.

Robinson KM. (2001) Unsolicited Narratives from the Internet: A Rich Source of Qualitative Data. *Qualitative Health Research* 11: 706-714.

Roos RA. (2010) Huntington's disease: a clinical review. *Orphanet Journal of Rare Diseases* 5: 40.

Roos RA, Hermans J, Vegter-van der Vlis M, et al. (1993) Duration of illness in Huntington's disease is not related to age at onset. *Journal of Neurology, Neurosurgery, and Psychiatry* 56: 98-100.

Roos RA, Vegter-van der Vlis M, Hermans J, et al. (1991) Age at onset in Huntington's disease: effect of line of inheritance and patient's sex. *Journal of Medical Genetics* 28: 515-519.

Sarangi S, Bennert K, Howell L, et al. (2004) Initiation of Reflective Frames in Counseling for Huntingtons Disease Predictive Testing. *Journal of Genetic Counseling* 13: 135-155.

Sermon K, De Rijcke M, Lissens W, et al. (2002) Preimplantation genetic diagnosis for Huntington's disease with exclusion testing. *European Journal of Human Genetics* 10: 591-598.

Simpson SA and Harper PS. (2001) Prenatal testing for Huntington's disease: experience within the UK 1994-1998. *Journal of Medical Genetics* 38: 333-335.

Smedley RM and Coulson NS. (2017) A thematic analysis of messages posted by moderators within health-related asynchronous online support forums. *Patient Education and Counseling* 100: 1688-1693.

Smedley RM and Coulson NS. (2018) A practical guide to analysing online support forums. *Qualitative Research in Psychology*: 1-28.

Smith J, A., Michie S, Stephenson M, et al. (2002) Risk Perception and Decision-making Processes in Candidates for Genetic Testing for Huntington's Disease: An Interpretative Phenomenological Analysis. *Journal of Health Psychology* 7: 131-144.

Smith JA, Stephenson M, Jacobs C, et al. (2013) Doing the right thing for one's children: deciding whether to take the genetic test for Huntington's disease as a moral dilemma. *Clinical Genetics* 83: 417-421.

Solberg OK, Filkuková P, Frich JC, et al. (2018) Age at Death and Causes of Death in Patients with Huntington Disease in Norway in 1986–2015. *Journal of Huntington's Disease* 7: 77-86.

Sørensen SA and Fenger K. (1992) Causes of death in patients with Huntington's disease and in unaffected first degree relatives. *Journal of Medical Genetics* 29: 911-914.

Tassicker R, Savulescu J, Skene L, et al. (2003) Prenatal diagnosis requests for Huntington's disease when the father is at risk and does not want to know his genetic status: clinical, legal, and ethical viewpoints. *British Medical Journal* 326: 331-333.

Taylor SD. (2004) Predictive genetic test decisions for Huntington's disease: context, appraisal and new moral imperatives. *Social Science & Medicine* 58: 137-149.

Tibben A. (2007) Predictive testing for Huntington's disease. *Brain Research Bulletin* 72: 165-171.

Turner DR and Willoughby JO. (1990) Ethical issues in Huntington disease presymptomatic testing. *Australian and New Zealand Journal of Medicine* 20: 545-547.

Tyler A, Quarrell OW, Lazarou LP, et al. (1990) Exclusion testing in pregnancy for Huntington's disease. *Journal of Medical Genetics* 27: 488-495.

van der Eijk M, Faber MJ, Aarts JWM, et al. (2013) Using Online Health Communities to Deliver Patient-Centered Care to People With Chronic Conditions. *Journal of Medical Internet Research* 15: e115.

Vennik FD, Adams SA, Faber MJ, et al. (2014) Expert and experiential knowledge in the same place: Patients' experiences with online communities connecting patients and health professionals. *Patient Education and Counseling* 95: 265-270.

Walker FO. (2007) Huntington's disease. *The Lancet* 369: 218-228.

Walsh A. (1999) Presymptomatic Testing for Huntington's Disease: The Role of Genetic Counseling. *Medicine and Health* 82: 168-170.

White M and Dorman SM. (2001) Receiving social support online: implications for health education. *Health Education Research* 16: 693-707.