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Clinical genetics (accepted)

**DOING THE RIGHT THING FOR ONE'S CHILDREN:
DECIDING WHETHER TO TAKE THE GENETIC TEST FOR
HUNTINGTON'S DISEASE AS A MORAL DILEMMA**

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Abstract

This is a qualitative examination of candidates’ decision-making in relation to the genetic test for Huntington’s disease (HD). Semi-structured interviews were conducted with nine participants who were asked about factors influencing their decision whether to take up predictive genetic testing. Transcripts of interviews were subjected to interpretative phenomenological analysis to elicit emergent themes. A key factor for participants was to do the right thing for their children. Interestingly this factor presents a moral dilemma to participants and can direct them either towards or away from testing. This paper offers a detailed examination of how participants think through this dilemma.

Keywords

Children; decision-making; genetic test; Huntington’s disease; qualitative; moral dilemma

Introduction

Huntington's disease is a dominantly-inherited, progressive, neurodegenerative disorder, usually of adult onset, characterised by motor disability, affective disturbance, and cognitive impairment. A predictive genetic test is available which, in the overwhelming majority of cases, is unequivocal; a positive test result means the individual will develop HD at some time in the future, unless they die of another cause earlier. There is currently no cure and time of onset is unclear. A positive test will also change the risk status of any children they have from 25% to 50%. How does an individual decide whether to undergo testing?

A survey of motives of at-risk individuals seeking testing found an important reason was 'to clarify the risk for their existing children' (1). In a questionnaire study of at-risk individuals choosing not to test, the biggest factor given was 'if my risk goes up so does that of my children' (2).

Qualitative methods are helpful in illuminating such tantalizing findings. We analyzed participants' accounts of the decision-making process in HD and found strong affective and interpersonal components of the process (3). That paper reports the *process* of decision-making. Preliminary analysis of the *content* of the decision-making was also conducted but not completed. We have now had the opportunity to complete this analysis, in part prompted by our recent work on family-communication of genetic test results in hereditary breast and ovarian cancer (HBOC) (4). We interviewed patients who had taken the genetic test for HBOC and biological relatives with whom patients had discussed results. Participants showed a moral obligation to take the test, not primarily for oneself but for the benefit of other family members. As our preliminary analysis of the HD data suggested

something similar was happening, we decided to look again at that data-set with this more specific focus.

The moral issues experienced by at-risk individuals have not been extensively studied (5). Taylor's research points to some of the moral dimensions of predictive genetic testing for HD. She offers an insightful analysis of one participant's ambivalence over telling a potential partner about his risk status (5). Another study provides three detailed qualitative case-studies of moral issues involved in prenatal genetic testing in HD (6).

Methods

The study employed interpretative phenomenological analysis (IPA) (7). IPA is concerned with a participant's personal experience but recognises that the researcher's interpretations are required to make sense of that other personal world. IPA offers detailed idiographic analysis and therefore requires relatively small samples.

The study received NHS ethical approval. All participants were at 50% risk of HD and recruited through a medical genetics centre. There were 9 participants- 6 women and 3 men. All had children. All names have been changed to protect confidentiality.

Semi-structured interviews on perception of genetic testing were conducted with participants in their home. Important issues arising were probed further. Interviews were audio-recorded and transcribed verbatim. Transcripts were coded for the

presence of important themes and connections made between themes within and cross-case.

We present here the analysis of one important theme emerging during analysis. In the results section, we begin with a summary of how prevalent the theme is for subgroups within our set of participants. The primary aim of this paper is to provide a thorough and nuanced analysis of how individuals are thinking through the decision-making process. As part of this, we need to explore how participant circumstances, perceptions and psychological profile contribute to this. Therefore the bulk of the results section is given to a detailed account of three participants for whom this theme is significant.

Results

This paper reports one specific theme: 'Doing the right thing for one's children's reproductive decision-making'. In our clinical experience, this is commonly given as a reason by individuals wanting to take the genetic test for HD. We focus on this factor because participants in this study actually employed it as a reason which was leading either towards or *away* from testing.

Most participants want to do the right thing to assist their children's reproductive decision-making. Four participants wish to take the test and 3 of these claim a major reason is to assist with their children's reproductive decision-making. Two participants are currently opposed to taking the test and concern about their children's reproductive decision-making is a key factor in their decision. Three

participants are undecided. Two of them explicitly describe a major consideration is their children's reproductive decision-making.

We now present an analysis of three participants for whom this theme is significant.

Eleanor

Eleanor is 30 years old and has two daughters aged 12 and 9. She wants to take the test, 'for me kids...I need to give them all the information they can possibly have.'

HD has affected a high proportion of women in her family and Eleanor is fatalistic about the risk for her and her children:

I'll be devastated [if tested negative] cos I think I've psyched up that much for it to be positive... It's gunna cop for one of mine or both of mine.

Eleanor has witnessed the debilitating effects of HD at close quarters, seeing her grandmother become ill when Eleanor was 20 and this strongly colours her perception:

If I do get that bad...I'd take me own life cos I can't see the point of being like me Nana is.

Belief in a high rate of transmission and its debilitating impact seem to inform Eleanor's moral imperative to do what she can to halt the progression of HD, 'I can stop it dead.' Consistent with this, If she had known she was at-risk before getting pregnant, she, 'wouldn't have had the test and I wouldn't have had kids'.

Eleanor considers a hypothetical future where she doesn't test, becomes ill with HD at 60 and meanwhile her daughter has had four children. She would feel personal

responsibility because, 'I have just carried four more on'. And she can imagine her daughter saying:

“Why didn't you have that test? You could have stopped me getting pregnant...you could have stopped it in our line”.

Reinforcing her decision to test is Eleanor's conviction of late onset, believing it would start at or around 60. So Eleanor believes that even if she did test positive she would have a good life before onset, 'I've got another 30 years if I'm positive, I could do all sorts'.

Eleanor sees testing as part of a continuing process to keep her children informed:

I could stop it. I can say to my kids when they are old enough, “look this is what is running through the family, you've seen your Nana....There are tests.” But it's their decision.

She subsequently repeats that her daughters would have to make their own decisions, 'It's their decision, but they will have everything there.'

However the 'everything there' being offered to the children is pretty loaded by Eleanor's perceptions of HD and what is the right thing to do:

Hopefully with my two if they're old enough to understand, then it does stop with my line, it stops with my two girls.

The children will be making a decision within a context which includes Eleanor's powerful belief that the right thing is to do whatever is necessary to stop the continuation of HD in the family.

Angela

Angela is in her fifties and has a daughter of 28 with children and a son of 30 who does not yet have children. Angela is as adamant as Eleanor that the primary reason for testing is to help her children. Her experience of HD is very different to Eleanor's. Angela's father died recently and was only diagnosed with HD posthumously. Angela's demeanor and cognitive style are also very different to Eleanor:

It's always the positive things that I think about...I may be the lucky one.

So Angela approaches the testing decision in a very different frame to Eleanor. Eleanor has years of experience of the devastating impact of HD and believes she has the mutation. Angela has only recently been exposed to HD and is optimistic about the risk and its implications. However, just like Eleanor, Angela feels obliged to take the test to give her children the relevant information.

Angela explicitly introduces something not mentioned by Eleanor. Eleanor is so concerned with taking the opportunity of stopping a dreadful disease that she does not refer to the concomitant cost- potentially stopping the family line. For Angela, however, this is a real concern which she could see as a reason pushing her away from testing:

One of the biggest cons was... if my son decided not to have any children... and if my daughter's children decided, if we were ones that did have the defect, our family could actually stop now.

However that for Angela is counteracted by a more important concern:

If you're given the knowledge that you have got a choice whereas...we didn't have any knowledge, we didn't have any choice....If I didn't take it, I would be then taking on the whole of that power onto me...it's not allowing other people any choice...I think that's quite selfish.

Compare the two women's accounts of agency. For Eleanor, testing is an imperative assertion of power, 'I'm in the position, I can stop it dead'. For Angela however, not testing would be an equivalently singular decision but one she would not be comfortable with. So while Eleanor needs to test as part of cutting down options, Angela needs to test to open options up. While Eleanor was clear that the right course of action, if one tests positive, is to do what is necessary to stop the continuation of HD, Angela is equally clear that there isn't an obvious right course of action:

But it's still a dilemma if he finds out that he has got the genetic default. Does he have the children or doesn't he? ... He could be thinking "well I want a child whatever" and that would be his decision... If the only way to eradicate it is not to have any children... it is quite a powerful decision and a choice to make, to end a line in that way.

William

William is 30 and has a daughter aged 5. He found out his father had HD when his daughter was 1 and his father died a year ago. William's attitude to testing has changed over time. His first reaction was to want the test but he then realized he hadn't thought through the consequences of a positive test result, 'I wanted to be told that I hadn't got it'.

William has currently decided not to take the test. Perhaps because a decision not to take the test is not irrevocable, William isn't as single-minded as Eleanor and Angela and offers a number of reasons for not testing. William is concerned he'd become demotivated if he got a positive test result. While William is the same age as Eleanor, he has a very different conception of expected age of onset, believing it could start soon:

They say 30s...I'm now at the bottom edge it could start now... It's a strange feeling.

Thus William does not have the psychological luxury of being able to test while believing a positive test would not have physical consequences for 30 years.

William then discusses his wish to protect his daughter. He feels it would be in her interests to remain at 25% risk for as long as possible. William links this to her projected agency and reproductive decision-making. Like Eleanor, he envisages a future conversation with his child. In this case, she might specifically ask him not to take the test:

When she'd say, "...We would like to start a family one day obviously. We've got this worry over us...I don't want you to have a test Dad because that might, if it's positive, it could really put the kibosh on things".

William believes having children deepens life experience. Without his daughter, 'I think we'd have a very shallow existence.' Nevertheless, had he known he was at-risk earlier, he:

Wouldn't have wanted to get involved in a deep relationship...I don't think I would have had the test and I don't think I would have, well I wouldn't have children.

And since discovering he is at-risk, he has had a vasectomy:

We felt as a couple that it was the responsible thing to do not to put another child through it because it's the only way to totally eradicate Huntington's is not to have children.

William wishes to preserve a degree of freedom of choice for his daughter. Drawing on his own experience, he feels that freedom would be compromised by a 50% rather than 25% risk, so he is choosing to avoid an action which could lead to that enhanced risk for his daughter.

Comparing the three cases

Starting from the same wish to do the right thing by their children, participants are faced with a powerful and potentially conflicting set of moral imperatives. Individual characteristics mean they see these imperatives differently and end up with divergent decision trajectories (see Table 1).

Table 1 here

Eleanor and William feel impelled to try to stop HD. Angela and William highlight the right of their children to choose to have their own family. It is these two positions which can cause a moral tension.

Eleanor's gloomy fatalism and strong desire to stop HD cast a shadow over her children's reproductive decision-making and lead her, unequivocally, to decide to test. For her the moral dilemma is not manifest.

Angela has less distressing experience of HD and is less pessimistic than Eleanor about its impact on her family. She recognizes the potential moral dilemma that her testing generates for her descendants, realizing that stopping HD could be at the expense of stopping the family line. However Angela's optimism, commitment to the importance of informed choice, and belief that someone testing positive can still have a child means she is also driven to taking the test.

William most explicitly addresses the dilemma himself as he feels an equal commitment to the two imperatives which potentially collide- stopping HD and having children. Like Eleanor, he stresses the importance of trying to stop HD. However William, like Angela and unlike Eleanor, talks of the great value of having children and of the importance of having that choice to decide to have a family. William sees the value in not compromising that freedom by gaining potentially damaging information. Deciding against testing, for the moment, helps to reconcile the opposed moral forces and, in one sense, frees his daughter from the shackle of information which Eleanor sees as beneficial.

Discussion

Our paper complements previous qualitative work on moral aspects of genetic testing (5,6). One study describes some of the moral issues for individuals considering genetic testing for HD (e.g. the range of interpersonal commitments which can be felt) alongside some powerful idiographic analysis (5). A second paper presents three case-studies of individuals' own reproductive decision-making (6).

Our paper shows how individual parents differ in their reading of components in a moral obligation to do the right thing for their children, and how this can lead to different decisions.

We can also compare our results with qualitative research on moral aspects of genetic testing for HBOC (4,8,9). Women at-risk for HBOC emphasize they are testing more for the benefit of other biological relatives than themselves, and sometimes women feel pushed into acting in this way. One paper shows a small number of at-risk individuals deciding not to test, in-part because of the stress a test result would put on others (9).

Positive test results for HD and HBOC have different implications. With HD, the recipient knows they will get the disease and there is no cure. In HBOC, it offers an enhanced risk of getting the disease and risk-reducing options are available. The value of testing can be seen to be higher in HBOC and hence the predominance of a discourse of a need to test in the relevant participants. The issue represents a starker moral dilemma in the case of HD because testing offers no medical advantage. Therefore participants' thinking is more directed to existential issues around stopping a disease versus stopping a family line.

It is suggested that most philosophers have been sceptical of the existence of moral dilemmas, arguing that apparent moral dilemmas involve conceptual confusion (10). One philosopher who did believe there are real moral dilemmas is Sartre (11) who describes the predicament of a student in France during the Second World War, whose brother has been killed by the German occupiers. The student has to choose whether to stay in France and look after his grieving mother or go to England to join the resistance. Stressing the inherent moral dilemma, Sartre tells him:

‘You are free, therefore choose...No rule of general morality can show you what you ought to do.’(p38)

Reflecting on the case, Sartre states:

We cannot say that this man, in choosing to remain with his mother... would be making an irresponsible choice, nor could we do so if he preferred the sacrifice of going away to England.(p50)

We believe the same argument applies to our participants. They each face a difficult choice and ultimately need to make their choice themselves. We are not in a position to judge a choice as either right or wrong.

There is also a familial nexus for HD. While participants consider they are doing the right thing for their children, those children may in turn eventually face their own moral dilemma over testing. And some participants may discover that, well-meaning though it was, their children disagree with the decision the parent made.

Genetic counselling aims to promote understanding of medical facts and inheritance, achieve informed consent, facilitate decision-making, manage psychological distress, restore feelings of personal control, and help individuals to adapt to a genetic condition (12). The focus of genetic counselling is on the process of decision-making rather than on the decision that is reached (13). The practice of genetic counselling is complex and there is much variation in content and approach (14) but a non-directive approach is key (15) and there are clear protocols for the genetic counselling of patients considering genetic testing for HD (16). A range of counselling skills, such as active listening, reflective questioning and presentation of alternative scenarios, may be employed within genetic counselling to help patients

reflect on the consequences of their decisions in the context of their family and to consider the possible outcomes and implications of their decisions (17).

We hope our results may be useful to those offering genetic counselling in showing how different individuals can conceptualize the apparently same factor in different ways. Our results also speak to possible tactics for working with patients considering genetic testing for HD. This is analogous to the implications offered for clinical practice in (6). Candidates for testing often cite wishing to do what is right for their children. Our results offer possible material for engaging in dialogue with patients. Examples of how such tactics could be fleshed out are presented in Table2.

Table 2 here

The interviews were conducted at a time when exclusion testing in pregnancy was available but before pre-implantation genetic diagnosis (PGD) for HD was as available. PGD is an option which is now discussed in genetic counselling and for a few couples this can be their first reproductive option. We recognise this limitation of our study. However we believe our results still have resonance and relevance.

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