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Living at Risk: Concealing Risk and Preserving Hope in Huntington Disease

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Abstract

Much of the qualitative research on Huntington disease has focused on the genetic testing aspects of HD. The overall purpose of this qualitative study was to gather information about the everyday experience of living with the risk of developing Huntington disease in a sample of individuals at risk for HD who have chosen not to pursue genetic testing. Data for this article was obtained from unstructured, open-ended qualitative interviews of a sample of people participating in the PHAROS study. PHAROS, the Prospective Huntington At-Risk Observational Study, is a multi-site study that aims to establish whether experienced clinicians can reliably determine the earliest clinical symptoms of Huntington disease in individuals at 50% risk for HD who have chosen not to undergo genetic testing. Interviews were conducted at six PHAROS research sites across the United States. In this paper, the research team used qualitative description to construct and explore two main themes: (1) careful concealment of risk as an act of self-preservation and (2) preserving hope.

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On behalf of the Huntington Study Group PHAROS investigators. A full list of PHAROS investigators and coordinators can be found in Shoulson I. *et al.* (2006). At risk for Huntington disease: The PHAROS (Prospective Huntington at risk observational study) cohort enrolled. *Archives of Neurology*, 63, 991–998.

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Disclosure; Genetic disease; Genetic testing; Huntington disease; Risk

Introduction

Huntington Disease and Genetic Testing

Huntington disease (HD) is the most common inherited neurological disorder, with a prevalence ranging from 4.1–7.5 cases per 100,000 in the Caucasian population (Folstein 1989). HD is inherited in an autosomal dominant manner such that each child of an affected parent has a 50:50 chance of having inherited the HD gene. Men and women are equally likely to be affected. Penetrance, the likelihood of showing symptoms of the disease if the associated genetic mutation is present, is virtually 100%. The characteristic symptoms of HD, (abnormal movements, intellectual deterioration leading to dementia, and a variety of psychiatric disturbances most notably depression), usually begin around the age of forty although the disease onset has been seen in children as young as 2 and adults as old as 70 (Hayden 1981). Death normally occurs between ten and 17 years after onset (Harper 1991).

In 1983, HD became the first disease to be mapped to a previously unknown genetic location on chromosome 4 through the use of restriction enzymes (Gusella *et al.* 1983) which divide deoxyribonucleic acid (DNA) at sequence specific sites. Inherited variations of these DNA sequences could then be used as markers to trace the inheritance of HD within families. Based on this technology, it became possible to inform some individuals at risk for HD whether or not they would develop the disease in the future. In 1993, the discovery that HD is caused by an expanded trinucleotide repeat of cytosine-adenine-guanine (CAG_n) on chromosome 4 (Huntington Disease Collaborative Research Group 1993) made testing readily available and less expensive for anyone at risk who wished to be tested. Despite early estimates of a high uptake of predictive testing in individuals at risk for HD, 80–85% of at risk individuals to date have elected not to undertake the predictive test for HD (Creighton *et al.* 2003).

Studies in Huntington Disease

At the present time, treatment for HD is entirely symptomatic, with no treatment or cure that affects or slows the underlying disease process. Research, especially since the discovery of the HD gene, is progressing rapidly and the prospect of a treatment for HD that may delay the onset or slow the progression of this disorder is now on the horizon. In preparation for future clinical trials, the Huntington Study Group, a consortium of basic and clinical HD researchers, undertook to conduct a study of individuals at risk for HD. This study, the Prospective Huntington at Risk Observational Study (PHAROS) is a longitudinal study designed to establish whether experienced clinicians can reliably identify emerging clinical symptoms that indicate the earliest onset of HD in a cohort of 1,001 clinically unaffected individuals at 50% risk for carrying the HD gene but who have not had genetic testing. Participation requires individuals to return for a research visit consisting of a neurological examination, several paper-and-pencil questionnaires, medical history, and neuropsychological testing every nine months. The overall purpose of this qualitative study was to gather information about the everyday experience of living with the risk of HD in a subset of PHAROS participants.

Only a small number of qualitative studies have explored the meaning, significance, experience and communication of hereditary risk. The earliest study, conducted by a woman who is, herself, at risk for HD was published before the advent of genetic testing (Wexler 1979). Since then, much of the literature has focused on genetic testing and its

consequences. Most researchers agree that complex social, biological and temporal factors affect the understanding of hereditary risk and that the relevance of that risk fluctuates throughout the course of life as a function of life choices made at any given time. (Cox and McKellin 1999). Qualitative studies described the psychosocial impact and process of coping with a negative genetic test result with a focus on the process of redefining oneself as a person no longer at risk (Williams *et al.* 2000). Forrest *et al.* (2003) explored barriers and facilitators in family communication about genetic risk in patients with hereditary breast and ovarian cancer (HBOC) and Huntington disease. They concluded that telling family members about genetic risk was generally seen as a family responsibility, but whether and how to communicate this risk varied as a function of family structure, dynamics, and rules. More importantly for our purposes, these researchers found that the level of certainty felt by a person in relation to his or her own risk influenced what he or she could tell other family members. They also looked at the communication of genetic risk in HBOC and HD and identified two broad categories of information sharing which they termed 'pragmatic' and 'prevarication.' Pragmatic respondents tended to talk about disclosure in fairly active practical terms while prevaricators described a process, which might take years, of looking for opportunities within the normal processes of family life to find the 'right moment' to share genetic information. Hallowell *et al.* (2003) completed a retrospective interview study of 30 women who had undergone testing for HBOC and found that when describing their motivations for undergoing DNA testing and their experiences in disclosing genetic information within their families, they used care-based ethical justifications for their actions. They saw their role in generating genetic information for their relatives as the responsible and right thing to do. In the communication of their genetic test information to the family, these women often found that their desire to benefit their families was in direct conflict with the potential burden that this information might place on family members. Taylor (2005) used an exploratory qualitative approach to investigate the experiences and process of predictive test decision-making for individuals at risk for HD. She described four positions that could be taken with regard to predictive testing: (1) the decision to be tested, (2) the decision not to be tested, (3) the decision not to be tested now, and (4) indecision. She concluded that the decision whether or not to be tested was fluid, dynamic, and complex, and that the process of making that decision occurred over time and in response to life events.

Each of these studies examines an element in the process of testing, not testing, or communication about genetic risk. While they add to our understanding of decision-making, they do not address the broader experience of living at risk. Therefore the purpose of this study was to explore the everyday experience of being at risk for Huntington disease.

Materials and Methods

Design and Procedures

We developed an unstructured interview guideline and conducted five pilot interviews in the homes of PHAROS participants from the Indiana site. Then we invited PHAROS investigators and coordinators from the sites enrolling the largest number of participants to volunteer as interviewers. We selected five interviewers who traveled to our research site for a two day training session in interview techniques. Training focused on active listening, minimal direction for answers, conversational initiators, and summarizing techniques. Interviewers practiced during this training and received feedback on their approaches. We were confident that interviewers understood the unstructured interview strategies and could conduct interviews successfully and dependably.

We asked these interviewers to identify individuals from their PHAROS cohorts whom they thought would be "narratively active" and to invite them to participate in the interview

process. The study was approved by the Institutional Review Board of Indiana University and the Institutional Review Boards of the institutions of the five interviewers.

Interviews occurred either in a place convenient for the participant or at the PHAROS research site, usually at the time of the scheduled PHAROS research visit. The open-ended, conversational interviews lasted one hour or more. Interviewers encouraged participants to describe freely their personal experiences with HD, their relationships with affected family members, their involvement in care-giving, their attitudes toward telling others about their HD risk, and their motivation to participate in research. Open-ended questions encouraged responses during an interview. Common opening questions used by the various interviewers were, “How did you first learn you were at risk for Huntington’s disease?” and “Tell me about your experience of being at risk.” Interviewers audio-taped the conversations for later transcription. The authors checked each transcript by listening to the tape while reading through the transcript. In addition to enhancing the quality of the data, listening allowed members of the research team to hear the “voices” of the respondents more clearly. This strategy enhanced the ability of the researchers to understand and interpret the meanings intended by the participants.

PHAROS Participants and Subset Characteristics

The purposive sample of 55 PHAROS participants, (including the 5 pilot interviews) consisted of 38 women and 17 men, reflecting the 2:1 ratio of women to men in the larger PHAROS study. Their average age was 42 (SD=7.6) with an education level of 15.6 (SD=2.2) the equivalent of over 3 years of college. Participants were 87% White, 7% Black, 4% Hispanic and 2% Other. Seventy-eight percent were married, 16% were single and 5% were divorced. Thirty had affected mothers and 25 had fathers affected with HD. The average age of parental onset was 47.6 (SD=8.9) for affected fathers and 48.7 for affected mothers. The subset of 37 participants who are the focus for this paper did not differ demographically from the larger group of 55 interviewees.

We compared this group of 55 participants to the larger PHAROS cohort and found no significant differences in the distribution of our sample by gender, marital status, or the sex of their affected parent. There was a significant difference in the percentages of Black and Hispanic participants among the 55 interviewees (when compared to the larger cohort) due to our deliberate over-sampling of minority participants to interview.

Thematic Analysis

We used qualitative descriptive research methods as the foundation for the analysis in this study. Qualitative description uses naturalistically acquired data¹ such as unstructured, open-ended interviews to increase the understanding and explanation of a social situation by vividly and conceptually describing it (Sandelowski 2000). The analysis aims at the identification and discussion of themes and patterns to enable presentation of complicated and extensive situations.

Quality Criteria

Trustworthiness and fairness are quality criteria addressed in the steps of our data analysis:

1. Interviewer training regarding the use of unstructured approaches and qualitative paradigm;
2. Using open-ended, unstructured interviews;

¹Naturalistic inquiry is used when situations are unique or complex, when the level of uncertainty about the questions to ask is high and when there is little or no theory to direct the researcher (MDRC 2004).

3. Conducting these interviews in a familiar and comfortable setting (not a laboratory setting);
4. Allowing participants to direct the flow of the conversation;
5. Prolonged engagement with participants (over a year devoted to interviews and analysis);
6. The research team engaged in peer debriefing and discussion on a regular basis throughout the analysis;
7. Negative case analysis resulted in our choosing to focus on the 37 interviews that clearly demonstrated an identifiable theme (the other 18 interviews did not demonstrate these themes as clearly);
8. Liberal and extensive use of direct quotations to support discussion of identified themes.

The three authors engaged in an organized procedure to analyze the transcript data, as follows:

1. Careful transcription of the audio tapes by a professional transcriber;
2. Audio review of the tapes to ensure accuracy of the transcription;
3. Reading and re-reading the transcripts, paying special attention to shared and contrasting concerns of participants;
4. Discussion among investigators regarding key elements of respondents' perceptions of living with HD risk. During analytic meetings, the researchers discussed and applied more abstract and metaphorical labels, which facilitated a joint development of sub-themes and larger themes across all 55 transcripts;
5. Description and elaboration of predominant thematic ideas.
6. Decision to focus on conceptual themes related to concealment and hope for this paper. We are presenting this paper on the analysis of 37 of the 55 transcripts that most clearly demonstrated those two themes.. This subset of 37 did not differ demographically from the original 55 transcripts.
7. Checking to ensure that strong and sufficient transcript data supported each theme; and
8. Re-reading the transcripts to select relevant verbatim support for proposed thematic constructs.

Results

Living at risk was the overarching pattern demonstrated in these data. We developed it from the two foundational themes: (1) careful concealment as an act of self-preservation. (2) preserving hope. We present quotations from the respondent interviews to warrant and support these themes, based on representativeness, salience, and vividness.

Careful Concealment

We think those at risk for HD perhaps bear a greater burden regarding concealment and disclosure, comparatively speaking, than do people at risk for other chronic or life-threatening conditions. As an example, women with a high genetic risk for breast cancer escape the additional factors of psychological stigma, lack of treatment options, or the high probability of mental deterioration.

We titled this theme *concealment*, rather than disclosure, because it more accurately describes what is happening in our interviews. It is remarkable that most of the literature focuses on the concept of disclosure, when the *absence* of disclosure is more relevant. Concealment means to hide, or cover, to keep, or to preserve (Buck 1949). What these stories demonstrate is careful concealment, uncovering the HD risk only to those who are safe, and at times and places within the control of each participant. The act of disclosure in people at risk for Huntington Disease is not a single act, but a never-ending process. It is a process of balancing fear and hope in the same moment, and having to decide every day whether or not to tell. It is not an analytical accounting of pros and cons, or a weighing of probabilities of occurrence of one outcome over another.

Concealment does not happen with only one person. People at risk for HD must consider whether (or when) to tell their employers, their potential spouses, their friends, their health care providers, their insurance carriers, their attorneys, and perhaps most importantly, their children about their risk of HD. And while some individuals are very open about their risk, others take great pains to conceal the fact that HD is in their family.

For example, Monica (names are disguised) had few qualms about discussing HD.

I think you know I am right now I am up front with it. I don't hide it from anybody. I work for the State. They can't fire me for it. They got so many other people there with so many other disabilities that it's not even funny. They got several good years left in me. So I am up front with it.

Joy, too, was willing to share her genetic status with her co-workers because she trusted them to maintain her secret and to protect her "back" from being stigmatized by others.

You know, I don't mind them knowing and I told two people at work, which one is my co-worker that I work with every day. She's great. And the other is my supervisor and she's got my back. I mean I am so lucky I work where I do. She'll never, she'll never tell anyone though.

However, she was so reluctant to share this information with a wider audience that she consciously limited her involvement with the HD community.

I have felt very bad about the fact that I don't involve myself with the Huntington's community and which is kind of ironic, because, for one thing, I work in a system where people do a lot of nonprofit work. And we are very involved in fundraisers and very supportive. But I don't want to be known [as someone involved with HD]. I also work in a very small community and I don't want to and I have a very distinctive last name. I kept my name at work...and if, if I was very public about that, anyone with that name would be publicly recognized. And I have a very visible job. So I have purposely, unfortunately, had to distance myself from that. And I told my mother that she cannot use her last name in Huntington's stuff because people will recognize her. And I'm a little sorry about it but I am just not ready to deal with that yet.

Although there is little reliable data to document the extent of employment or insurance discrimination based on genetic information, three of our participants cited specific examples. In one case, Megan was in the process of completing her education and planning, ultimately, to be ordained into the church.

I belong to what we call an ordination process, and the committee was saying that that it may be a factor in my being ordained, because I have this Huntington's in my family and because I guess the process is so different now. But that's not fair.

In the second case, Emily's supervisor learned of her risk with very negative consequences.

My supervisor actually found out about the HD because he was aware [of HD] because he knew my mom. He knows my mom. And he saw kind of this progression and he knew that my uncle had killed himself. My supervisor and I had gone to college together and so that was kind of like “this thing,” and he was just aware that there was something going on. He didn’t know the name for it. And then, sure enough, he found out it was Huntington’s. So it was kind of difficult because here he was this kind of acquaintance friend and then also he’s my supervisor. And then he broke my confidentiality and told some other people about the HD without really knowing what he had done. Didn’t really know the ramifications of that. So there was a promotion that I was going up for, and the person who was in charge of that actually knew about the Huntington’s and outright said, well, I don’t feel comfortable doing this promotion because we don’t know where Emily’s going to be in the next ten years because of this whole HD thing.

Frances had a negative experience with an insurance company after her at-risk mother was injured in a car accident. Concealment in her case was thwarted by the revelation of HD by her mother’s doctor, a person she counted on for protection from the stigma of the disease.

Mother was doing pretty well with HD until the [motor vehicle] accident. Unfortunately the accident left her incontinent, among other things. And you know it was very hard and long process [filing for the insurance] but she got a portion of the settlement, monetary wise. And the insurance company agreed to pay for her to have a home health aid and pay for her medical treatment in regard to what was caused by the car accident, everything. Everything was going fine. Then finally, it seemed like seven, eight months later, once they got the neurologist records that said she was tested for Huntington’s. He said that she had the gene for Huntington’s, and then the insurance company stopped everything. They stopped payment for everything. Literally everything. They didn’t pay her doctors. They said point blank this is what her attorney said to me: her injuries were not from the accident, they were from the Huntington’s. So they wouldn’t pay.

Other participants spontaneously mentioned their concerns about the possible discrimination based on information about their genetic risk, even in the context of confidential research records. As Eric stated:

And I know we talk about the forms [to sign]. And whenever we sign a form, I worry that, you know, people go through other people’s trash and someone could find one of those forms. Not that anyone’s out to get me specifically, I wouldn’t think that; it’s just that I wouldn’t want to lose my job or my health insurance or anything because they found out I had it.

Our participants discussed the difficulty of bringing up the topic of HD in the context of relationships. Emily felt that she had been “discriminated against” in relationships.

That there are people that I’ve you know dated and we’re flirting along and everything’s great and they’re all in love and blah, blah, blah, and then we start talking. And then I say Huntington’s, and they don’t really know what that means. And then we talk a little more and then they’re like, not so sure about that, you know. Like maybe they can’t handle that, and to be honest, I’d much rather they say it now and move on. And so there were a few people I dated that really were [bothered by HD] and there was such a marked difference between ‘oh, you’re so wonderful and I could see me spending the rest of my life with you’ and ‘oh, I don’t think I can handle this HD thing’.

Douglas, too, maintained concealment in a dating situation until he found out that the woman he was dating would continue to “be there” for him despite the HD risk. His concerns related as much to his worry about passing on the gene defect for HD to potential children as it did with the possible dissolution of the relationship. Unfortunately for Douglas, that relationship ultimately did not last, and it failed because his HD risk was revealed.

I went through a short stage at the time I was dating a person and it was hard telling her about me being possibly at risk. But she had told me that she would totally support me and be there for me no matter what goes on. But I went through a short stage of telling myself, and possibly opening up to others, how I wouldn't want to have kids because I wouldn't want to risk that 50/50 chance of passing it on to another generation. I would rather wait until they come up with a cure or find a cure for the disease or possibly have some way of testing prior to having a child.

These interviews confirmed that disclosure of genetic risk within relationships is a complex task (Forrest *et al.* 2003). Most of our respondents discussed, in detail, the circumstances of telling, or not telling, their own children about their risk for HD. In what seems, on the surface, to be an act of supreme denial or withholding, participants revealed their extreme ambivalence in the ways they chose to tell, or not to tell, their children about this risk. This silence is astonishing, considering the risk their adult children carry as they have children of their own. As found in other studies of genetic disclosure for HD, people who did consider telling children first assessed the family member's vulnerability and receptivity to the information (Hamilton *et al.* 2005).

Christine echoes this idea when she describes the paradox of concealment, even though the children might already know about HD and possibly even know about its risk.

That's one area [telling the children] where we've made some really bad decisions. We haven't really had too many discussions about it. Partly because the last couple of years have been hard on them. They've lost three uncles in my husband's family because all three of his brothers got divorced like right at the same time. And that's been kind of devastating for my kids.... They know that my brother [who has HD], comes to visit usually twice a year for a week. They know that he, you know, that he has problems with his balance and they can't jump on him and play. And there's certain things that they can't do with him, and they know that he doesn't work because of his balance and stuff like that. They know that my father died of Huntington's disease. They know that my brothers and my sister were sick. They don't know that I'm at risk for Huntington's disease. And they know the name of it and we've, we have been I guess a little bit evasive about it because we're, I'm afraid that as soon as we say Huntington's disease they're going to think “death” you know. They're going to jump to that and I don't think my youngest son could understand that. He's very young....

She balances the need to talk with her children about HD with their ages, and with all of the other things going on in their lives right now. But her main reason seems to be this notion that talking too much about the disease will encourage the children to become too focused on it, especially on the terminal and hopeless nature of the disease.

Arthur's children are older, and have children of their own. When asked about any discussion he and his wife have had with the children, he responded,

I don't know if we've ever sat down and said it, you know.... They know they're at risk and they know that I'm at risk and we've had some emotional discussions from time to time. I'm sure my son has read up on it some. My daughter, she's probably

pretty much where I was. Neither one of them has had a parent that's went through it or a grandparent. Of course my brother, they know what it did to him. But I don't know if they've ever considered testing or anything. I guess we've really never had those type of conversations. Probably the conversations we've had have been emotional in you know with family loss and this is what you're up against and you know all kinds of discussions with them about blaming my drinking and other things on HD at times and, but really as far as sitting down and have a clinical discussion and you know sitting down and looking them in the eye and saying you don't know and I don't know but you may have a fifty-fifty chance of getting this and passing it on. We've never had a discussion like that.

The silence maintained by these two participants is not complete; they are silent only about the meaning of being at risk. Arthur's family had emotional discussions about their losses as a family due to HD, but these never progressed to talking about what risk really meant to him or to them. Christine has explained some things to her young children, but resisted going further because she wanted to protect them from reaching the wrong conclusions about her own life and death, or theirs. This approach coincides with the conclusions drawn in a study by Gallo *et al.* (2005) regarding parents sharing information with their children about genetic conditions. We found that parents do assess their children's development in making these decisions, and act in ways that protect their children from emotional distress associated with the knowledge of risk. Arthur, however, has maintained this silence throughout his children's lives, even past the point when his children began to have children of their own. He assumes that they will discover what they need to know the same way he did: by reading about it (his son), or by simply going on with life (his daughter). Both Christine and Arthur want their children to have normal lives, and this hope guides their concealment more than any other factor. The paradox, of course, is that none of them can have a normal life unless they are tested, and the test result is negative.

Some participants were children themselves when they first learned about HD, usually because a parent became symptomatic. Kathy was on the receiving end of parental silence in this stark example

When I got a little bit older, Dad put mom in a nursing home when I was fifteen years old. And I remember this being, now, as very emotional for me to talk about because when we came home from school one day my father had put my mother in a nursing home but he didn't tell us. So we came home and she was just gone. And to this day it's very emotional. But you know I guess I feel like my dad was just doing the best that he could do under the circumstances....

She also points out that she never really got many details on her family history of HD from anyone in her extended family, including her father. So in this case, the family silence was pervasive and complete.

Like Kathy's father, Rachel also tries to protect her children from knowledge of their at-risk status, and this began when the family first learned that her mother had HD. Her first response was just to protect them and to keep them from being overwhelmed:

It was very hard when my mother got sick. It was a very harsh reality to come upon the whole family and now we're in front of our children. And we're trying to pull it together enough for our children so that they're not so overwhelmed and anxious if I tell them what's going on. And so we did not tell them what was going on. I don't know how I'm going to introduce it to my children future-wise. I don't know. I don't know. And I guess maybe you know counseling will help me out because I think it's a very devastating thing for someone so young to decide whether they might want children or not. Whether they should go through genetic counseling or

testing and all that stuff. It's still very, you know they're young. We're talking about waiting until after they get their master's degrees because that's what I tell them. You ain't doing nothing until after you get your master's degree. I still don't know how to introduce it to them.

Julie, on the other hand, does not hesitate to include her children in visits to their grandmother with HD, and also talks with them about the disease openly.

Whenever they'd see their grandma do something outrageous you'd explain to them what's going on. I mean I remember riding in the car. My kids and I were in the back seat of my parents' car and one time even my mom started hitting my dad. You know and that was pretty hard for them to see. But I tell them that that's not their grandma, that's the disease. And they've seen her go downhill.

Matthew also talked with his children about his mother's disease, though he hasn't spoken of his, or their, risk for the disease. Like Christine's family, his children are still young and he thinks they are not yet ready for that conversation.

My daughter knows there's a worry here. We've discussed it with her. We've said grandma has Huntington's Disease, a neurological disease that affects your personality and motor skills. And I tell her this isn't her, you know. What you see, your grandma, is not the same person I grew up with. She is not the same woman. So my daughter understands it. My son is a little too young to fully understand it. But we tell him too, and every time we're there at the nursing home, we have them tell their stories and relate what their week was like and give her hugs and kisses and everything else, even though it is hard for grandma to give hugs and kisses.

Douglas, who had no children of his own, neatly summed up the parental dilemma regarding concealment. He pointed out that parents may not control all the information available to their children, even if they would wish to. Others may un-conceal the truth about HD and the potential risk of developing the disease.

At a young age I don't think children would understand. Possibly under ten. I don't know if they would actually be able to understand the impacts of the disease if they were told that they are at risk. But then if you wait too late, then you know, it's a problem. And if they've dealt with it for a long time in their life they might be more comfortable with it and depending on the situation you could tell them earlier as long as, you know, they've been around it for awhile. I think it's best to know more about the disease, but of course the hard part is keeping it from them. If you know that, if their parent is positive or has the disease, it would be hard to keep that information from them. And if they're involved with others in the HD community it might come about that way too. So parents might not even be the ones to tell them. It might be through reading information or so on so forth.

It is clearly a struggle for parents who are themselves at risk to talk with their children about this aspect of HD. They, for the most part, do not shield their children from seeing the effects of HD on their grandparents or aunts and uncles. Many work to help their children understand that what they see is the horrible disease and not the person. As in the Gallo *et al.* (2005) study, parents in our research cohort were more likely to talk about the disease itself rather than about the genetic risk for it.

Preserving Hope

Hope is a crucial factor in the dynamic of living at risk. Participants sustain hope that they will not have HD, or that their risk decreases with age, as a means of keeping open the possibilities of a disease-free future for themselves and their children.

Participants' reasons for not being tested seem to rest in this kind of hope. Christine's reasoning incorporates hope that she does not have the disease at all, or, at the very least, that her risk is decreasing with age. In both instances, testing would gain her little.

My family has a history of early onset so I guess in some ways I'm starting to feel like that risk is diminished considerably. So even though I know it's not entirely out of the picture, I guess I feel more comfortable now than I have in the past, as far as my at-risk status. I just don't see anything positive coming out of testing.

In this example, not being tested is not passive inaction, but rather a positive step taken to maintain hope through uncertainty. Even when testing would give children important knowledge about their own hereditary status, parents resist. By doing so, they can keep hope alive regarding both their own and their children's risk for the disease.

Christine's active maintenance of uncertainty about her status allows her to preserve hope that she has escaped the disease. Interestingly, when she did go for a simple neurological exam during the time her brothers, sister, and father were sick, it proved to be too frightening for her.

It was incredibly scary. It was so frightening. I remember I would walk over to the doctor's office and sit outside in the chair and then I wanted to say to myself, "You just go in there, you just let them look at you and you'll know—in five minutes' time you'll know." And I couldn't do it. I couldn't bring myself to get up and go in there for that neuro test. Now eventually I did, but it took a lot of courage. I guess I just don't feel like there's anything in it for me. So that's why I don't get tested for the gene for HD.

In her mind, the risk that a neurological examination could result in a diagnosis of HD was far more frightening than not knowing her risk status.

Kathy echoes this point when she says:

What would be the reason? If I got tested and I found out I had it, is that going to make me feel any better? I don't think so. Is there anything they can do about it? No. So what's the point? I can't imagine coming to the realization that you have it, knowing that there's no hope. No hope of there being a cure. I'm going to die eventually, so that's not the point. It's the point of the way I'm going to live the rest of my life. And really, just being alive [is not living]. I'm terrified of it right now but I can't imagine not having any hope, because right now, I have hope. I have hope that it's not going to be me.

While this response may look like denial, it reflects both women's experience with the disease itself in other family members. Knowing that you are positive diminishes hope because it also ends uncertainty. Sometimes hope is situated in the potential for cure, or on the possibility that children would be lucky enough not to be affected.

Other participants believe that a cure or effective treatment will be found by the time their children or other family members have to face their own risk. Barbara was enthusiastic about her own hope:

So I mean I think it's remarkable the genome and the coding of the marker and things that they have found for genetics. I think that there is a wide, wonderful wild world out there that is going to be uncovered in the next twenty years with science. I really hope that it comes in time for my siblings. It's most exciting that Huntington's can possibly be reversed, new neurons might grow if they were to do any of the stem cell type work. So I'm hopeful.

Reuben uses the story of Schrödinger's cat to illustrate the story of his own uncertainty and its importance to hope in his every day life.

Basically you got a box with no holes in it. You put a cat in there and cover the box. Days later, is the cat alive or dead? There's two possibilities you know. Before you open the box, there's that moment of uncertainty. At the moment you open the box, reality splits, and in one reality the cat is alive and is allowed to live and eat and breathe and have children. But the other reality is there's a dead cat. You won't know till you open the box and then it's too late. Now I see that moment of uncertainty before my opening up the box is that genetic test.

He hopes to live and eat and breathe and have children, but it also means he must live in that perpetual moment of uncertainty in order to do so.

The intertwining of hope and uncertainty is different from denial. These participants understand HD in its everyday consequences. They know what HD looks like, sounds like, acts like, and they know how it ends. They have observed and cared for, been abused by, and have lost numerous family members with the disease. They harbor no illusions about life and death with HD. Monica's fears derive from her years of taking care of her mother who had HD:

But seeing, you know, either way that it could happen. I try to put myself in both situations. And it just terrifies me just thinking that it could be me laying in that bed like a vegetable. Wondering if I get to the point where I don't even have a thought running through my head. That all I'm doing is just laying there and breathing and that's it. That's what scares me most.

She may be afraid of having the disease, but she clearly is not in denial about what that future could bring. Contrast this with her family's response whenever she tries to discuss it:

I don't know if they're saying it just to make me feel better or if it's to make them feel better or what. But you know, if I even just mention HD, you know, that it could be me getting it. They say, "Oh, well, you're not gonna have it you know. There's just no way in earth that you're gonna have it. My husband does it, my father does it, my brother does it, my husband's family does it. Oh, well, they say, you're gonna be fine. It's not gonna be you."

This family talk serves to sustain their hopes for her, while also limiting her ability to talk about it with them.

Shade (2001) discusses how hope structures responses to threatening situations and keeps possibilities open, even in the face of catastrophe:

...consider those all too common and frightful instances when we discover a loved one has been involved in an accident whose consequences remain unknown...our first hope is typically that our loved one survived or at least died a relatively painless death. In either case, we are generally unable to do anything to directly affect the outcome.... Hope can still give us the strength to support one another, or to prepare to help the loved one should he or she survive. It can keep us open and oriented to the various *possible* goods still available to us, even if the worst of our fears should prove true. (p. 48).

It is not difficult to substitute HD for a frightful accident in his example. Participants expressed their hope for a meaningful life for as long as possible, even if they succumbed to the disease. Reuben offers this as his definition of the good life:

Good food, creativity in the room. That's all I want. You know and have enough money to pay my bills, have a spot or a place or an apartment where I could do so and not worry about where my next paycheck is coming in or how I'm going to pay this next bill. That is the good life. Even if I do have HD and all my friends are in the hospital with me in the room. Even then.

Mark echoes this hope when he talks about the life he would choose if he develops the disease:

Well, there's just too much to fight for. I mean I'm not saying that I wouldn't be disappointed or sad or upset. I would be, but I would also know that my wife is supporting me and my children care for me and support me and there would be a lot to do. And today I can't tell you how disappointed I'd be if I came back positive. I mean that would be impossible for me to comment on.... I would be actively seeking the answer and then there would be some point in there where there would be a great deal of disappointment and sadness. But then you know once you grieve, that's not a good word, but once you deal with that, then I'm all for moving ahead and fighting it as best I can. My hope is that if it is positive that my doctor will have some way to fight it. You know something that I can do. Either through medication or through therapy or something to help fight it.

Participants are not demoralized by their fear of this possible future, in part because they create and use uncertainty to sustain hope. This is not to claim that all people at risk for HD are able to hold open a future of uncertain possibilities. In many cases, the parents or siblings of these participants lived an opposite kind of life, in fear of a certain future. Emily's uncle emphasized the importance of hope in his suicide note.

Something that my uncle said, that I think really stuck with me, is he wrote a suicide note. He said that there's such a big difference between living with hope and living with knowledge. And that he would take the living with hope any day. And so he really did not think we needed to know, one way or another.

Barbara believes her brother committed suicide because of his fear of HD, and that her other siblings live in terror.

But in his letter, his suicide letter he also mentioned you know, that it was a struggle for him, this girl broke his heart, but also even you know coming to terms with Huntington's and running in the family with no hope also caused him to take his own life. I think my siblings don't know a lot about this (research). They are terrified and just go to the deepest darkest depression when they think about it, you know. I don't know if it's isolation, abandonment, I just really don't know what it is that terrifies them the most about the disease. I know that some people at risk, I know how my brothers and sisters have been terrified their whole life of it and had planned their whole lives, saying "I'm not having kids..."

Discussion

Significance and Implications

This research begins to illuminate the experiences of people choosing to live at risk for Huntington Disease in an era when genetic testing is possible. Baréma (2005) refers to HD as "the beast" or "that bitch"—his risk, his hope, and his uncertainty infuse every aspect of his life and consciousness. He eloquently captures this way of being-in- the- world at risk, yet hoping. His uncertainty gave him hope.

A few weeks later, we started our first child. And then we made a second one, and then a third. And we went on living. Happily. With passion for life despite the

risks. Because you have to live and defy the bitch [HD]...until the day you go down (p 26).

That outlook was demonstrated by the participants in this study. They have achieved a remarkable balance between living at risk and living with hope in their lives. Our analysis of these interviews suggests several implications for research and practice.

Disclosure of genetic risk, as we came to understand it, is not a single decision. It is a decision made every day, and in relation to various individuals and groups: oneself, one's children, potential spouses, employers, insurers, and the world at large. Schneider and Conrad (1980) addressed this issue in their well known article on epilepsy and disclosure, using the metaphor "in the closet with epilepsy." (p. 37). Persons with epilepsy use "strategies of selective concealment" in deciding when to reveal and when to conceal their condition. They are never entirely in or out of the closet. The closet door revolves, just as it does for people at risk for HD. The participants in this study made the same kind of complex decisions about disclosing or concealing their at-risk status. As told, sometimes these decisions resulted in very negative consequences, diminution of job prospects, refusal of medical benefits, the loss of a valued relationship, consequences that might make further disclosure either less likely or, at the very least, more anxiety provoking. And if disclosure of genetic risk is problematic, one might infer that a positive genetic test might cause even more problems down the road. Unless we, as a nation, come to grips with the very real threat of genetic discrimination and take positive steps to remove that threat, the use of genetic testing is likely to remain low and fraught with anxiety.

The discussion of genetic risk within the family, especially between parents and their children, is especially problematic. Nearly every participant with children experienced terrible difficulty in talking to their children about their risk, even when the children were grown. We infer from this difficulty that practitioners could, and should, find ways to help people at risk develop plans for educating their children at an appropriate age. We envision such plans to be developmentally based, geared to answering questions at the child's level, as well as being persistent and gradual in the presentation of the issues of importance. Parents need help in moving beyond descriptions of the disease to discussions of its meaning in their own and their children's lives. This would assist them in having a more realistic approach to disclosure, versus just waiting until the children are contemplating marriage (which could be years after they had already initiated sexual activity).

It may be that this difficulty is a direct result of the fact that these individuals have not been tested. As found in Forrest *et al.* (2003) what is conveyed to family members is influenced by the level of certainty regarding one's own genetic risk. To initiate a conversation explicit in the details of the inherent genetic risk for HD might logically lead to the issue of testing and why the parent has chosen not to be tested. Many parents would feel a reasonable amount of pressure to be tested in light of their children's risk, especially if their children voiced a desire to clarify this risk.

Participants live in a world where their hope for their children is anchored in their hope for themselves. Nunn (2005) describes hope as always connected to uncertainty. She states "... we can garner two important points about hope. First, uncertainty about the future is the human condition and hoping for possible states of affairs is one stance that human beings can take towards this uncertainty. Certainty creates no space for hope: uncertainty creates space for hope" (p. 67). It is only through maintaining uncertainty (and thus hope) about their own HD status that they can sustain that hope for their children as well. It creates a life where hyper-vigilance and silence exist in uneasy *détente*.

Choosing to be tested is, in a way, a decision to disclose one's real risk to oneself. Participants' choosing not to be tested is not denial but a positive way to preserve both hope and their identities as people with a future. The central role that uncertainty plays in preserving hope for the future for these individuals underscores a central tenet of genetic testing for Huntington disease: that each individual at risk needs to make the decision about testing for him or herself freely and without coercion. This role also provides further support for testing guidelines that argue against testing children at their parents' request, guidelines crafted with the intent of preserving the autonomy of the child to decide for him or herself whether or not to be tested. Anecdotal evidence from our interviews indicates that the absence of such hope may prove fatal (i.e. through suicide) and suggests continued caution in the genetic testing of conditions for which there is neither treatment nor cure. Our findings suggest parallels between HD and other chronic and stigmatizing conditions where there is a possibility of advance knowledge of one's probable fate. (One of our participants described this situation as not wanting to know the "timeline of my own death.") Researchers should investigate the broader relevance of our work to other stigmatizing and/or life threatening conditions, such as cancer, epilepsy, and mental illness.

Clinicians also need to reflect on their own beliefs and biases about genetic testing, and to examine the extent to which those beliefs and biases present themselves in their care for people at risk for HD. Primary care health professionals need to be cognizant of the fact that just because a test can be done does not mean that it should be done. What these men and women are telling us is that it is not safe to assume that genetic testing for incurable diseases will necessarily provide information that is wanted, or needed, by those at risk and that testing may have a significant negative impact on the lives of their patients. Practitioners can support their client's desire to say no to genetic testing. "Knowledge is considered an asset, a necessity, even a virtue in a cultural tradition of enlightenment." (Hunniche 2003, p. 66). But knowledge in the case of HD may serve to destroy hope. Clinicians need to be vigilant about promoting an attitude that privileges knowing over not knowing.

Limitations

Unstructured interviews can be challenging, in terms of telling a complicated story that unfolds over long periods of time. Training interviewers requires time and practice, and more of both would always be beneficial. In addition, our participants were selected on the basis of their narrative abilities and willingness to tell their story of living at risk. We believe it is equally important to listen to the voices missing from this subgroup, and we hope to access this subset in subsequent studies. In addition, we acknowledge that relying on words alone to reveal the nature of such complex issues is itself a limitation. Future research should make use of additional methods, such as observation, to construct a more complete picture of this complicated communication.

Conclusion

This research opens a window into the meaning of genetic risk in everyday life. Further research could focus on spouses and adult children of those at risk for HD, from the perspective of their own experiences and decisions about testing, reproduction, and living at risk. Another possibility for future research might focus on the implications of verbal communication versus other forms of communication, in both concealment and disclosure. (For example, unstructured interviews versus direct observation of family interactions.)

It is noteworthy that several participants said the interview for this study was their first opportunity to talk about the emotional side of HD, despite their years of experience with neurological, cognitive, and psychomotor testing as part of PHAROS. Qualitative interviewing may actually have therapeutic and diagnostic implications (Shamai 2003). We

think that unstructured interviews might actually change the views and actions of the participants with respect to their careful concealment of risk and their preservation of hope.

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References

- Barema, J. The test: Living in the shadow of Huntington's disease. New York: Franklin Square; 2005.
- Buck, CD. A dictionary of selected synonyms in the principal Indo-European languages. Chicago: University of Chicago Press; 1949.
- Cox S, McKellin W. There's this thing in our family: Predictive testing and the construction of risk for Huntington's disease. *Sociology of Health and Illness*. 1999; 21:622–646.
- Creighton S, Almqvist EW, MacGregor D, Fernandez B, Hogg H, Beis, et al. Predictive, pre-natal and diagnostic genetic testing for Huntington disease: The experience in Canada from 1987 to 2000. *Clinical Genetics*. 2003; 63:462–475. [PubMed: 12786753]
- Folstein, SE. Huntington's disease: a disorder of families. Baltimore, MD: The Johns Hopkins University Press; 1989.
- Forrest K, Simpson SA, Wilson BJ, van Teijlingen ER, McKee L, Haite N, Matthews E. To tell or not to tell: barriers and facilitators in family communication about genetic risk. *Clinical Genetics*. 2003; 64:317–326. [PubMed: 12974737]
- Gallo AM, Angst D, Knafl KA, Hadley E, Smith C. Parents sharing information with their children about genetic conditions. *Journal of Pediatric Health Care*. 2005; 19:267–275. [PubMed: 16202834]
- Gusella J, Wexler N, Conneally P, Naylor S, Anderson M, Tanzi R, et al. A polymorphic DNA marker linked to Huntington's Disease. *Nature*. 1983; 306:234–238. [PubMed: 6316146]
- Hallowell N, Foster C, Eeles R, Ardern-Jones A, Murday V, Watson M. Balancing autonomy and responsibility: the ethics of generating and disclosing genetic information. *Journal of Medical Ethics*. 2003; 29:74–79. [PubMed: 12672886]
- Hamilton RJ, Bowers BJ, Williams JK. Disclosing genetic test results to family members. *Journal of Nursing Scholarship*. 2005; 37(1):18–24. [PubMed: 15813582]
- Harper, PS. Huntington's disease. London: Saunders; 1991.
- Hayden, MR., editor. Huntington's chorea. Berlin: Springer; 1981.
- Hunich L. Learning from the voiceless. *New Genetics and Society*. 2003; 22:257–269. [PubMed: 15115026]
- Huntington Disease Collaborative research Group. A novel gene containing a trinucleotide repeat that is expanded and unstable on Huntington's disease chromosomes. *Cell*. 1993; 72:971–983. [PubMed: 8458085]
- MDRC. Management research in VA. 2004. Retrieved December 13, 2004 from: http://www.mdrc.research.med.va.gov/mgt_research/typical_methods.htm
- Nunn, BV. Getting clear what hope is. In: Elliott, JA., editor. *Interdisciplinary perspectives on hope*. New York: Nova Science; 2005. p. 63-77.
- Sandelowski M. Whatever happened to qualitative description? *Research in Nursing & Health*. 2000; 23:334–s340. [PubMed: 10940958]
- Schneider JW, Conrad P. In the closet with illness: epilepsy, stigma potential and information control. *Social Problems*. 1980; 28(1):32–44.
- Shade, P. *Habits of hope: a pragmatic theory*. Nashville, TN: Vanderbilt University Press; 2001.
- Shamai M. Therapeutic effects of qualitative research: reconstructing the experience of treatment as a by-product of qualitative evaluation. *Social Service Review*. 2003; 77:455–467.
- Taylor SD. Predictive genetic test decisions for Huntington disease: Elucidating the test/no test dichotomy. *Journal of Health Psychology*. 2005; 10:597–612. [PubMed: 16014395]

- Wexler, NS. Genetic Russian roulette: The experience of being 'at risk' for Huntington disease. In: Kessler, S., editor. Genetic Counseling: Psychological Dimensions. London: Academic; 1979. p. 199-220.
- Williams JK, Schutte DL, Evers C, Holkup PA. Redefinition: Coping with normal results from predictive testing for neurodegenerative disorders. Research in Nursing and Health. 2000; 23:260–269. [PubMed: 10940951]