Journal of Genetic Counseling, Vol. 15, No. 2, April 2006 (© 2006)
DOI: 10.1007/s10897-006-9018-7

Original Paper

Discovering the Family History of Huntington Disease (HD)

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Published Online: 16 March 2006

A considerable body of research has explored both predictive genetic test decisions for Huntington disease (HD) and the impact of receiving a test result. Extant research reveals little, however, about how and when at risk persons first discover their family history of HD. Drawing upon 24 semi-structured interviews with at risk persons and their family members, this study explored initial discovery of HD in the family. Qualitative data analysis revealed four different, though sometimes related, trajectories of discovery: (1) something is wrong, (2) out of the blue, (3) knowing, but dismissing, and 4) growing up with HD. These pathways highlighted the importance of the temporal and historical contexts in which genetic risk for HD was discovered. Notably, ignorance about HD was the most salient feature shaping participants' narratives of discovery. Implications for research and clinical practice are discussed.

KEY WORDS: Huntington disease (HD); initial discovery; family history; genetic risk.

We knew there was something wrong, but no one seemed to know what it was.—Serena, at risk for Huntington disease.

INTRODUCTION

Huntington disease (HD) is an incurable genetic illness that manifests around mid-life. It is a progressive, neuro-degenerative disease whose symptoms include a movement disorder (e.g., chorea), personality changes and cognitive decline (Harper, 1996). HD affects both men and women, and each child of a HD parent has a 50:50 chance of inheriting the mutation and is said to be at risk for the disorder. The genetic defect is the result of a CAG repeat expansion in the IT15 gene on chromosome 4 (Potter *et al.*, 2004). With few exceptions, mutation carriers *will* manifest the disease in their lifetime, excepting death from some other cause before the disease manifests (Potter *et al.*, 2004).

Extant literature is replete with studies and reviews that describe reasons for and against predictive genetic testing for HD (e.g., Binedell *et al.*, 1998; Evers-Kiebooms *et al.*, 2000; Meiser and Dunn, 2000). The actual uptake of testing has been much lower than indicated by earlier studies of test intention (Evers-Kiebooms and Decruyenaere, 1998). The most common reasons cited for testing included a desire for certainty, planning for the future and to inform children. Common reasons for declining were related to the emotional and psychological consequences of coping with a positive test result.

Similarly, the psychological impact of receiving a predictive test result for HD has been well studied (e.g., Dudokdewit *et al.*, 2002; van't Spijker and ten Kroode, 1997). This research was generally carried out as part of predictive testing protocols in specialized genetics centres. The majority of these studies used validated psychological instruments to assess psychological morbidity in tested and non-tested people prior to, and following, test

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disclosure. An early review (van't Spijker and ten Kroode, 1997) found that many mutation carriers experienced short-term emotional reactions such as numbness, sadness, anxiety or anger. However, levels generally returned to normal one year following test disclosure. Increased feelings of hopelessness were observed in mutation carriers, while reduced scores were recorded for non-carriers (Tibben *et al.*, 1994). Within 6 months, however, unwanted intrusive thoughts about HD decreased for both groups. After 3 years, there were no differences in intrusive or avoidant thoughts about HD or in hopelessness between mutation carriers and non-carriers (Tibben *et al.*, 1997). Further, non-carriers did not always experience immediate, uncomplicated relief; rather, some experienced survivor guilt and had difficulty adjusting to a new identity (Dudokdewit *et al.*, 2002; Sobel and Cowan, 2000).

Short-term impact of the predictive test result (positive or negative) is fairly good. Anticipated psychiatric problems (e.g., suicide) have rarely materialized. However, a recent longitudinal study suggested that research to date could have underestimated the real impact of a positive test result. Timman *et al.* (2004) found increased levels of hopelessness in mutation carriers over the study's seven to 10-year follow-up.

A significant portion of existing research on predictive testing for HD focused on the individual psychological aspects of the clinical experience (Cox, 1999). Test result was the main independent variable, and the focus was on elucidating causal relationships between test result and various clinical outcomes. This approach is valuable as it can help identify subgroups of the testing population who may experience the most distress following result disclosure and who may need additional counseling. However, this approach provides minimal insight into the primary *focus* of anxiety (e.g., what are people anxious about and why?) and the lived experience of HD in the family. Lived experience is notable since family experiences of genetic risk and illness influenced predictive testing decisions for HD (Etchegary, 2005a,b; Taylor, 2004, 2005) and may influence post-test adjustment for some inherited cancers (McAllister, 2002).

How (and When) Do People Discover They are at Risk?

Cox (1999) cogently argued that initial discovery of HD in the family has been overlooked in clinical studies of predictive testing for HD. Clinical research on the psychological effects of testing choose an arbitrary baseline (e.g., 1 month prior to the test) from which to measure morbidity and to determine the impact of test results. However, this practice fails to explore the perceived significance of individual or family disease-related events that may precede such baseline measures. For example, whether awareness of the family history was abrupt or gradual or whether at-risk persons cared for an affected relative may have implications for psychological well-being or predictive testing decisions (Etchegary, 2005a,b; McAllister, 2002; Taylor, 2004). Interviews with people from families with a clinical diagnosis of hereditary nonpolyposis colorectal cancer (HNPCC) revealed several social factors—many related to the family experience of disease—that either facilitated or blocked the process of engaging with cancer risk (McAllister, 2002). For example, personal experience with the family history of cancer and family talk about cancer were identified as causal conditions that influenced engagement with cancer risk. Further, ignorance of the family history and impersonal knowledge of the family history were identified as intervening conditions that blocked the process of engagement. Engagement with genetic risk is an important concept since it may have implications for psychological morbidity subsequent to test results. For example, McAllister (2002) suggested that those who were only partially engaged with their cancer risk might have poorer post-test adjustment than those who were intensely engaged at the time of the test.

Family history and experience with HD also influenced predictive testing appraisals and decisions (Cox, 1999; Etchegary, 2005a,b; Taylor, 2004, 2005). Taylor (2004) noted that the decision-making contexts and time frames for individuals with only *recent* knowledge of their family history of HD appeared to be quite different to those who had long-standing knowledge about the family history and their own risk. For the former, there was *sudden* knowledge of potential fatal illness in the future, bringing with it the attendant psychological complexities in marriage, reproductive and career choices (Taylor, 2004). Such individuals might have unique informational or emotional needs, having implications for genetic counselors who must prepare at risk individuals as comprehensively as possible for predictive testing (Taylor, 2004).

Thus, family history and experience of disease were part of the multiple contexts within which genetic test decisions were taken and test results

incorporated and managed (Cox, 1999). Given the relative lack of data on initial discovery of the family history of HD, the purpose of this study was to explore how and when participants discovered their family history and to document any differences in pathways to discovery.

METHODS

This study used semi-structured interviews to explore participants' initial awareness of their family history of HD. The study received full ethical approval from Memorial University's Human Investigation Committee (HIC).

Participant Recruitment

Participants were recruited from the provincial medical genetics clinic and from two HD support groups in St. John's, Newfoundland and Labrador (NL), from November 2003 to February 2004. Decisions about who should be informed about the study were taken by genetic counselors and a social worker with the Huntington Society of Canada (HSC). Those deemed as too vulnerable to participate (e.g., because of recent family death or cognitive impairment) were not invited to participate in the study. This practice was consistent with the local ethics committee policy. The HSC also included an insert about the study in its newsletter, *Horizon*.

Unfortunately, response rate for the current study could not be accurately determined (Etchegary, 2005a). In an effort to reduce the amount of time spent on recruitment, it was agreed that clinic representatives would contact only one person in HD families and ask him/her to inform other family members about the research. This practice, while useful for reducing time and effort spent on recruitment, could have attenuated participation as there was no way of knowing if the contacted individual actually informed other family members about the study. A total of 115 people had been offered genetic testing for HD in NL at the time of the study, and 14 families were contacted by the genetics clinic about the research (M. Crowley, personal communication, 2004). However, it was not known how many families were represented by the 115 individuals, making it difficult to estimate response rate. Seven participants learned of the study through HD support groups. Five participated, while two others arranged interviews on several occasions but eventually did not have time to complete the interview. In total, 24 people participated in the study, representing 10 different families affected by HD. There were only two refusals to participate, both owing to deaths in these families at the time of recruitment.

Table I. Risk Status of Interview Participants

Tested positive	3
Tested negative	5
Tested, intermediate gene	2
Tested, did not receive results	2
Tested, now affected with HD	2
Family history, never tested (i.e., at risk)	6
Family member, not at risk	4
Total	24

Participants

Thirteen participants had undergone predictive genetic testing and one had undergone diagnostic testing, resulting in a variety of test outcomes (see Table I). Six participants had not been tested and were at risk for HD. Four family members, not at risk themselves, also participated. For 11 of 20 tested or at-risk persons, the family history of HD originated on the maternal side; nine traced the history to the paternal side. The affected parent of most participants was deceased, and almost all had a sibling at risk for (or already diagnosed with) HD.

The mean age of all participants was approximately 46 years ($SD = 11.3$; *Range* 21–73), and for those who had been tested, an average of 6.5 years had passed since testing at the time of the interview. Table II summarizes other participant demographic information. Three-quarters of the participants were female and most were married or co-habitating at the time of the interview. Almost all participants had children. Participants were fairly well-educated; all but two had completed high school and most went on to complete college diplomas or university degrees, two at the graduate level.

The Interviews

Interviews were conducted between January and June 2004. Most interviews were conducted in participants' homes, with some in the researcher's office and some by telephone. Telephone interviews

Table II. Participant Demographic Information ($N = 24$)

Mean age (*SD*)	46.2 years (11.3)
Gender	
Male	6
Female	18
Children	
Yes	21
No	3
Employed	
Yes	15
No	9
Marital status	
Single	3
Married/common law	17
Separated/divorced	2
Widowed	2
Education	
Less than high school	2
High school	3
Partial college	4
College graduate	15
Residence[a]	
Urban newfoundland	13
Rural newfoundland	11

[a]Urban Newfoundland was defined as any community with a population of 7, 000 or greater; Rural Newfoundland consisted of any community with less than 7, 000 residents.

were conducted only when this was expedient and convenient for both the participant and researcher, normally when participants resided more than 4 hr away from the study site. There were no notable differences between face-to-face and telephone interviews. For example, the mean single-spaced page length of typed transcripts for telephone interviews was 17; for face-to-face interviews, 18. Nor were there any obvious differences in the richness and depth of the stories related during the interviews. Finally, similar themes arose in all participant accounts, regardless of interview mode.

It is acknowledged, however, that telephone and face-to-face interviews are different interview modes. In particular, body language and other nonverbal cues were difficult to record in telephone interviews. As such, extra vigilance was paid to sighs, pauses or other hesitations in speech (e.g., crying) during telephone interviews.

With participants' permission, interviews were tape-recorded and transcribed verbatim. All participants were provided with a copy of the interview transcript, except one who wanted only a summary report of research findings.

Interviews lasted from 1–3 hr and averaged about an hour and 15 min. Interviews were semi-structured in that questions were not confined to a specific order, and participants were actively encouraged to discuss any other issues they felt were important. Despite a slightly different order of administration or deviations from original wording, all questions were asked to each participant. A detailed question guide facilitated the tracking of all questions asked during each interview. Additionally, participants received a general interview guide prior to the interview outlining the broad topics (e.g., test decisions or family history) to be discussed in the interview. Despite some different wording that emerged naturally during interviews, each interview encompassed the same content.

Interviews covered a core set of topics such as family history and emergence of HD, test decisions, stigma associated with HD and healthcare concerns. The current analysis focuses on the emergence of HD in the family. Questions were chosen following a wide reading of the literature in diverse fields including health and social psychology, medical sociology and anthropology and medical and clinical genetics. Discussion with key informants, including representatives from the local and national HSC and the provincial medical genetics clinic, including genetic counselors and geneticists, helped refine the interview guide.

Data Analysis—Interpretative Phenomenological Analysis (IPA)

IPA (Smith *et al.*, 1997, 1999) is a qualitative methodology having its roots in phenomenology and symbolic interactionism. It emerged in the last decade as a distinct approach to empirical research in psychology and may be a particularly valuable approach to issues surrounding the new genetics (Chapman and Smith, 2002). IPA aims to achieve a detailed exploration of how people make sense of their experiences, recognizing that the researcher's own perceptions are needed in order to make sense of the personal world being studied. While the researcher attempts to understand the participant's personal world, this cannot be done directly or completely. Rather, "Access is both dependant on, and complicated by, the researcher's own conceptions, which are required in order to make sense of that other personal world through a process of interpretative activity" (Smith, 1996, p. 70).

IPA follows a case-by-case approach to analysis, beginning with an initial transcript and slowly working up to more general categorization or theory (Smith *et al*., 1999). Each interview transcript was read and examined several times. Meaningful groupings of data were identified in each transcript that generated clusters of themes. For example, themes of ''looking back at past behavior'' and ''memories of affected relatives'' seemed to cluster together in a meaningful way to describe one pathway to the initial discovery of HD. Clusters of themes were compared across all transcripts until a final set of shared themes was identified. Thus, ''initial discovery of HD'' was a shared theme in all interview transcripts; however, four clusters of themes emerged that described four different pathways to initial discovery. Following this analysis of interviews, a detailed summary report of findings/themes was fed back to participants for their review. No errors or misinterpretations were reported by participants.

RESULTS

Initial discovery of HD in the current study could be categorized into four (sometimes interrelated) themes: (1) Something is wrong; (2) Out of the blue; (3) Knowing, but dismissing; and (4) Growing up with HD. Routes to discovery were neither mutually exclusive, nor exhaustive, but were the central themes which organized participants' narrative accounts of how they discovered that HD was part of their family.

Something is Wrong

For nine participants, the family history of HD was unknown until a relative, usually a parent, began to manifest symptoms of the illness. The unknown family history usually meant that neither participants, nor their family physicians, initially suspected HD. Odd behavior in a family member or a general sense of something being wrong motivated a search for answers, beginning with visits to family physicians. While the odd behavior was worrisome, it was often initially attributed to a benign origin. For example, Michelle[3] explained why she did not originally worry about her parent's twitching movements:

> . . .ever since I can possibly remember, Mom always had this twitching, and I had gone to the doctor with my Mom, and I asked him what it was, and he said that it was the twitching nerve syndrome; it wasn't anything too serious. (. . .) Huntington's didn't mean much more to me at that time.—Michelle, at risk

The physician's assurance that her mother's twitching was nothing ''too serious,'' coupled with an unknown family history, obviated the need for Michelle to seek out information about HD. It was only when her mother's mysterious illness progressed that other medical investigations were initiated and the diagnosis of HD eventually confirmed. Prior to the diagnosis, Michelle had never heard of HD:

Actually, we had to ask what it was because we had never heard of it.

Similarly, Serena recalled how she first heard the words ''Huntington disease:''

> I only remember it as a nightmare, that part of it. My Mom was to several doctors and we all knew something wasn't right. We knew there was something wrong with her, but no one seemed to know what it was. So, the doctor finally referred her to a neurologist. He diagnosed her right away.—Serena, at risk

Serena confirmed that neither she, nor her siblings, had ever heard of HD prior to their mother's diagnosis. Note also she described the process of initial discovery as ''a nightmare.'' The family began to search for information about the illness; what they found was both frightening and devastating:

> We didn't know what Huntington disease was. It was the first time we had ever heard of it. Then, [names partner] went to the library and when he came back, it was even more devastating. (. . .) We found out about us, and our kids and heredity.—Serena, at risk

Note that a diagnosis of HD in her parent did not *immediately* translate into Serena's understanding of her own risk and that of her children's. It was only subsequent to researching HD that an awareness of the implications for herself and her family emerged.

Another participant recounted the misdiagnosis of her relative and her suspicion that the initial diagnosis was incorrect:

> I knew there was something neurologically wrong for a long time, but I didn't know anything and he got misdiagnosed, which happens with Huntington's a lot. For a time, he was diagnosed as Alzheimer's. (. . .) When he was diagnosed, I said, ''No, that's not it.'' But I didn't know.

[3]Names and significant social details (e.g., number of children or siblings, exact ages) have been changed to protect participant anonymity.

When asked if she questioned the physician, she replied:

> Yeah. He wasn't listening. But I didn't really know, I didn't really have a face or a name. When they said Huntington's, and I began to educate myself about Huntington's, it was, "yes, yes, yes, yes."

Shirley explained how she began to recognize symptoms of HD in her relative:

> I could go down the line. If there was 20 items, I could tick off 16 or 17. At first, I didn't really clue in on the neurological part, but I just knew there was something amiss.—Shirley, caregiver

These stories illustrated that there was often limited knowledge about HD in some participants at the time of initial discovery. Most participants who had attended genetic counseling sessions were very satisfied with the experience, perceiving counselors as very good and caring people. Despite this perception, however, difficulties in risk comprehension were evident. One participant, for example, suggested the genetic counselors were very good; however, the technical information imparted during counseling was hard to understand:

> They [genetic counselors] were very good. It was useful, but not knowing what it was about and trying to soak it all in. Still, it was like you were stupid, and I didn't know what they were saying. Sometimes, doctors have a way of putting words in there…I'm not a smart person.

Out of the Blue

Three participants narrated the initial discovery of their family history of HD as emerging out of the blue. Each participant was unaware of their family history. The initial reaction to such news was normally (but not always) one of shock, confusion and disbelief. This trajectory of discovery contained elements of *something is wrong* (e.g., unknown family history of HD and odd behavior(s) in a family member). However, it was distinguished by social and/or geographic distance from a family member diagnosed with HD (cf. Cox, 1999).

Gerald described how a distant relative died with (what was later discovered to be) HD:

> I don't remember anything about my uncle who had the disease, other than the fact I went to their funeral when I was about 8. And that's all I remember. No one talked about it then. As far as what we knew about it? Nothing.—Gerald, tested negative

It was many years before Gerald's immediate family realized the cause of death and the implications for them. Gerald recounted a relative's description of his affected uncle:

> She had said that he was really strange for the last couple of years, he was very abusive. (…) Then there was all the twitching and jumping. It was years after his death, or more, before some guy — he was a young doctor, out of practice a couple of years— said, "Yes, we did a thing on that sometime in class." He sent him on to a geneticist who started the whole ball rolling…

Gerald's reaction to the discovery of his family history of HD was different from other participants who became aware of HD *out of the blue*:

> Dad's onset, I think he was in his early 70s when we noticed it. At that stage, [neurologist] said it was not likely going to kill you, because it's so late onsetting. It might be a contributing factor, but it is not likely going to be the cause of anything. And it wasn't really. But, from there, we just learned to live with it, knew what to expect to come up because Dad was so late.—Gerald, tested negative

Gerald's account of discovery was informative because it highlighted the importance of the temporal context of discovery in his reaction to his own genetic risk. Gerald suggested that because his father's illness was very late onset, he could learn "to live with it" and "knew what to expect." When asked if he remembered feeling distressed about having the genetic test, there was no evidence this was a particularly stressful time for him:

> As far as I was concerned, it was a done deal basically. If I had it and passed it on, the kids had it anyway, so there was nothing to be done there.

Additionally, the fact that Gerald tested negative for the HD mutation might partly explain why his reflections on discovering his risk were less emotionally charged than those who were still at risk or who had tested positive.

For example, Lori had only recently discovered her parent had HD. Adding to her shock and disbelief at discovery, Lori's unaffected parent had recently died. She recalled her initial reaction to discovering the family history of HD:

> I had no idea. I didn't even know what it was. I had just lost my [unaffected parent], and when I did find out what it was, I thought, "My God, how can this happen to both my parents?" (…) When I found out the risk I was at with the Huntington's [pause]…I'm

> struggling. —Lori, tested, waiting for results at the time of the interview[4]

The context of Lori's life contributed to the difficulty of discovering her family history of HD. She had just cared for a parent through a horrific illness and now had to acknowledge her own risk for HD. As with the theme of *something is wrong*, discovery initiated an information search and contact with healthcare professionals. Lori said:

> That night, I got into it up to my elbows, and I've been into it up to my neck ever since.

Knowing, but Dismissing

For four participants in the current study, the initial discovery of HD could be described as *knowing, but dismissing*. There had always been knowledge that a distant family member had HD, and it was this element that distinguished this route of discovery from *out of the blue*. However, geographic and/or social distance precluded day-to-day exposure to the illness. Additionally, the immediate parent had never shown any symptoms of HD. Therefore, while the family had knowledge that the illness was in the family, there was no direct experience with HD in immediate family members. This theme, therefore, contains elements of *something is wrong* since participants knew a distant family member was not well, but the illness was not originally diagnosed as HD. Marjorie recalled:

> [Names deceased family member], we thought she had bad nerves. We were *told* that was her problem. I remember she had all these movements, but we thought it was nerves. No one knew about Huntington disease. No doctors knew Huntington disease.—Marjorie, caregiver, emphasis in original

It was only when a second family member was diagnosed with HD that the family could, in hindsight, see similarities in the two relatives. Jackie described her memories of finding out about HD:

> Really early on, my uncle was sick. We knew that there was something wrong with him and it was neurological or whatever. But there had never been a name put to it. Then we found out that my aunt who lived away in [names place] was diagnosed with HD.—Jackie, tested negative

Jackie had a vague recollection that HD was part of the family; however, the first affected relative had died when she was fairly young and a second affected relative lived quite some distance away. Social and geographic distance kept the experience of HD from her immediate family, and her parent was not exhibiting any signs of HD. In this way, HD was regarded as irrelevant to the immediate family. "We thought we might have escaped."

It was not until an immediate family member began showing signs of HD that the family began to realize they had not escaped and there were implications for others. Julie remembered:

> [Names affected family member] was living away and we didn't see; there wasn't any sign of anything until he came home. . .but then we noticed the coordination thing and my sister and I are on the Internet and the whole time you are thinking that you don't want it to be because you know what it is going to mean for you.—Julie, at risk

The affected family member returned home as the illness progressed, and diagnostic testing eventually confirmed HD. In the theme *knowing, but dismissing*, participants had known for some time about their family history of HD; however, geographic and/or social distance from affected relatives, coupled with an asymptomatic parent, allowed them to dismiss the relevance of HD for their own lives. Thus, this theme also contained elements of *out of the blue*, since the return of a visibly sick family member did occur out of the blue. It was distinguished from *out of the blue*, however, since participants were aware of their family history of HD. Despite this awareness, it was only with visible signs of HD in an immediate family member that participants began to acknowledge the implications for themselves.

Growing up with HD

Finally, eight participants grew up knowing about the family history of HD, and many had vivid memories of their affected relative. Some had cared for an affected parent or sibling. In this theme, the family history of HD was not deliberately hidden from participants. However, in some cases, there was a period of time when something was wrong with a family member, but this something was not immediately attributed to HD. This was in the context of 30 or 40 years ago when HD was more obscure than it is today, highlighting the historical context in which discovery took place. Brenda, for example, recalled her early experience with HD in this way:

[4]Lori has since received a negative test result.

> Well, my father died from the disease. In my family, it seems to have manifested in the late twenties/early thirties, whereas normally, from what I understand, people are in their sixties even before the disease starts to manifest. But in my family, it seems young. Actually, when I was growing up, we never really knew what my father had. We didn't know what it was, but he used to have these rages. I thought he was crazy because he used to take these fits.—Brenda, at risk

It was not until her parent's death that the illness was officially pronounced to be HD:

> My father died on [day], and we went to the hospital and this doctor had come from [place] and had experience with Huntington patients. He was the one who diagnosed it.

It was not until Brenda was in her mid-teens that she heard the words ''Huntington disease'' attributed to her father's crazy behavior. She didn't remember when she realized the implications for herself, but admitted certain life decisions (e.g., reproductive decisions) may have been affected by knowledge of her genetic risk:

> Myself, I don't know if it's there sub-consciously, but I was 36 before I had [names children]. And, I don't know if that's subconscious because the onset is always in the late twenties, early thirties. I feel today that I'm past that and I probably won't get it.—Brenda, at risk

Similarly to Brenda, Dorothy had vivid memories of her mother's illness, without immediately knowing it was HD. By the time she was entering her teenage years, she knew her mother was sick, but it was not until her mid-teens that a label was attached to the illness:

> I didn't [understand], not for a long time. And it wasn't called Huntington's then, it was called St. Vitus Dance. That's what they said she had, but it was years and years after that before they said it was Huntington's disease.—Dorothy, tested, intermediate gene

Brenda and Dorothy, both in their fifties, grew up with affected parents, but neither knew the illness was HD until their mid-late teens. It must be recalled that the timing of their parent's illness would have been the 1960s. As both noted, there was very little communication about HD at the time (publicly or privately). There was no evidence in either narrative, however, of the deliberate concealment of the family history.

Other participants grew up with HD in full recognition of the illness. These participants were in their twenties and thirties at the time of the interview, and in general, there was no question that their relative was affected with HD:

> I remember being terrified of her, so I couldn't have been more than 5 or 6 I guess. That was quite a few years before she died, at that time we called it Huntington's Chorea of course. I have always been aware of the name, you know, I have always known that it could be passed on and so on, but not knowing everything, like neurological and everything like that. -Stacey, tested negative

Following a parent's test result, Stacey decided to take the genetic test:

> . . .we found that she had an intermediate and a normal. Still not quite sure how this was working, but I was like, ''Intermediate, that's not a normal gene, right?'' So, it was only a short time after that that I called [doctor] and asked her if I could get it done, just to be certain. If Mom had the intermediate, I just wanted to make sure that I didn't have either of them.

Cheryl discovered her relative was affected with HD in her early teens. While geographic distance precluded everyday exposure to her relative, Cheryl recalled changes evident during family trips and visits:

> So, grade 6 we found out, but she had been sick. I can remember seeing a change since I was a little kid. (. . .) I never knew about it at that time. I was so young. Mom talked about it in bits and pieces. (. . .) Still there were changes, and I knew things weren't the same as they used to be. –Cheryl, at risk

Cheryl's narrative suggested that awareness of HD in the family was a gradual process for her. As a child, she remembered being excited about her relative coming to visit, but as childhood progressed, ''there were changes.'' Despite her youth at the time of initial awareness, Cheryl knew the implications of the family history for herself:

> Well, my Mom was really good about it. She really started to investigate it and get a lot of information. And she never kept it from me. As soon as there was a concern raised, me and [siblings] were informed. So we always knew about it. Pretty much, I knew what it was as soon as I was told. Mom explained it to us. She didn't try to go around the fact that my Mom might have it. I understood what was going on. Nothing was uncertain for me. I knew exactly what was going on. –Cheryl, at risk

As this quote illustrates, Cheryl was confident in her knowledge about HD and the implications of her parent's risk for herself. Her experience highlighted

the importance of family communication about the illness: Cheryl's parent gathered and distributed information to her children, and as such, Cheryl "knew exactly what was going on."

DISCUSSION

Alice Wexler, herself at risk for HD, offered a powerful description of the emergence of HD in families:

> First there is the grandfather who has died of "nervous trouble" on the back ward of a state hospital, the uncle who attracts whispers and stares from the neighbors as he staggers down the street, the doctor who says, "Women do not get it." (. . .) Divorce, arrests, abandonment, suicide punctuate the action. There is always a moment of discovery, when the protagonists finally learn the truth, usually after having several children. In the end, the characters all come to resemble one another, and the action winds down to a predictably gruesome close, with no resolution or release and always the promise of more performances to come. (Wexler, 1995, p. xi).

Her depiction was echoed, though only in part, by some participants in the current study. Many recalled relatives who were diagnosed with "bad nerves," and all suggested HD was a never-ending disease. Importantly, however, most narratives did not evince a singular moment of discovery. Rather, there was often only gradual awareness of HD in the family, sometimes after months or years of odd behavior in relatives.

The narratives from the study interviews illustrated four inter-related (but distinguishable) pathways of discovering the family history of HD. Some stories exemplified the type of discovery referred to as *something is wrong*. In this experience, the family history of HD was unknown. Without a known family history, HD is often misdiagnosed as another dementia disorder (O'Shea, 1997). Following the protracted odd behavior of a relative or the uneasy feeling that something was wrong, a search was initiated to discover the source of the behavior. The family member was eventually diagnosed with HD. This was the most common route to discovery in the current study. Given the lack of awareness of a family history of HD in many families and the low public profile of the illness, this type of discovery is likely to be common in the HD community (Cox, 1999).

For others, initial discovery of the family history was *out of the blue*. The family history of HD was unknown, and initial discovery was often marked by disbelief and shock. Unlike the stories referred to in *something is wrong*, there was no protracted period of odd behavior in relatives. Rather, this pathway to discovery was distinguished by social and/or geographic distance from an affected family member.

In *knowing, but dismissing*, there was usually vague knowledge that a distant relative had HD; however, social and/or geographic distance precluded day-to-day exposure to the illness. For members of these families, there was a sense of having escaped HD, especially when a parent reached an advanced stage in life symptom-free. As in *out of the blue*, there was usually upheaval in the family when an immediate family member became symptomatic.

Finally, some participants' initial awareness of their family history was the result of *growing up with HD*. This route to discovery was marked by living in close social/geographic proximity to an affected relative. Awareness of the illness was usually gradual, and the family history of HD was not deliberately hidden from family members. This route to discovery in particular highlighted the importance of the historical context in which an initial awareness of the family illness emerged.

While these moments of discovery are clearly related, they can be contrasted according to several dimensions. These include: the temporal context of discovery (e.g., childhood or adulthood), the geographic context of discovery (e.g., close proximity or distance), the process by which awareness of the family history emerged (e.g., immediate or gradual), the process of recognizing the implications for self (e.g., immediate or gradual), and prior knowledge of HD (knowledge may mean simply having heard of the illness, without necessarily understanding it). These dimensions are summarized in Table III.

While Table III is useful as a summary device, it does not highlight that the initial discovery of the family history of HD was only one complex element in participants' personal biographies (Cox, 1999). The construction of these biographies may not be an individual enterprise, as suggested by Beck's (1992) ''risk society'' perspective. Rather, genetic risk was situated within the context of family life and memories of affected relatives. In this study, an overarching theme within all four trajectories of discovery was the temporal context of genetic risk and illness. Families have a past and a future, and genetic narratives, in particular, vividly illustrate this point. Remembering the quirks or odd behavior of long-deceased relatives and speculating on the fate of children or grandchildren of the future, gave the sense that the family

Table III. Four Tajectories of Discovering the Family History of Huntington Disease (HD)

Trajectory of discovery	"Something" is wrong	Out of the blue	Knowing, but dismissing	Growing up with HD
Timing	Adulthood	Adulthood	Childhood and adolescence, but sometimes adulthood	Childhood and adolescence
Social orGeographic Distance	Close; but, no one is aware of family history of HD	Usually distant	Usually distant	Close; may or may not initially be aware illness is HD
Process of awareness of family history	Gradual	Abrupt	Abrupt; usually when an immediate family member becomes sick	Gradual
Process of awareness for self	Gradual	Gradual	Gradual	Gradual
Knowledge of HD prior to discovery	None	None	Some; but, usually in a distant family member	Some have full knowledge of HD; some can only label HD at later stage of the illness

history of HD extends infinitely backward and forward in time (Cox, 1999).

How at-risk individuals initially discovered their family history of HD has been relatively unexplored in the literature with the exception of Cox (1999). However, an appreciation of how people first discover their family history is significant for several reasons.

No participant recalled hearing about HD in any other context prior to discovering his/her own family history. Ignorance about HD was, by far, the most salient feature shaping narratives of discovery in the current study and in Cox (1999). Misdiagnosis and/or an undocumented family history were prominent in many participant stories and appeared to be significant factors shaping participants' awareness of HD. An obvious implication of these findings is that some people who attend genetic counseling sessions may arrive with very limited knowledge about HD and the implications of predictive genetic testing. Knowledge brought to counseling might be quite different, however, for individuals who grew up with the illness. Cheryl's experience in the current study highlighted how growing up with HD influenced knowledge about the illness and the availability of predictive genetic testing. Thus, a different kind or amount of information about HD and the predictive test may be needed by at risk individuals depending on their family history and experience with the illness.

Clinical research on the impact of predictive testing begins with an arbitrary baseline (e.g., 1 month prior to the test) from which to measure morbidity and to determine the impact of genetic test results. However, this approach fails to explore the significance of disease-related events in families or individuals that may precede these measures. For example, McAllister (2002) suggested that the degree of engagement with HNPCC risk may change with the passing of time and cancer-related events in family life, having implications for cancer testing decisions and possibly for post-test adjustment. Similarly, Taylor (2004, 2005) found that HD-related events in the family, such as the diagnosis or death of a parent or the test results of a sibling prompted a re-thinking of predictive genetic testing for HD, particularly for previous no-test decisions. Such findings underscore the need for a thorough exploration of the family experience of illness during counseling sessions, including initial discovery of the family history.

While some research has explored the effect of age and duration of awareness on genetic-test decisions (Quaid and Morris, 1993; van der Steenstraten *et al.*, 1994), little attention has been given to the *meaning* of initial awareness. For example, findings from the current study suggest that being aware of the family history of HD may *not* translate into immediate awareness of the implications for oneself or for one's children. This has implications for researchers, clinicians and counselors working with HD families. It implies that awareness of the family history is not a dichotomous variable, and discovering the family history of HD does not always unfold in a linear fashion. Instead, the significance of the temporal and historical contexts in which discovery took place was highlighted in the current study. Younger participants, for example, had always known about their family history of HD. They escaped much of the confusion, uncertainty and upheaval faced by

their parents and grandparents. New genetic technologies and other medical advancements such as new medications are changing the phenomenological experience of living at risk for a genetic disorder. It is important for clinicians and researchers to take account of the temporal and historical contexts within which an awareness of genetic risk for HD emerges. This suggests that exploring the family history and experience of HD may help to elucidate the process of deciding about genetic testing for HD (Etchegary, 2005a,b; Taylor, 2004, 2005). This exploration might also highlight gaps in genetic counseling clients' knowledge about HD. In the current study, for example, Cheryl's experience of her family history was distinguished from some of the other participants who grew up with HD by her knowledge of the family history at a relatively young age (early teens). Her story underscored the importance of the historical context of discovery, particularly when compared with Brenda's and Dorothy's experiences. Both discovered their family history in the 1960s, and each remarked that no one knew or talked about HD during that time.

How an awareness of the family history of genetic risk emerges may also have implications for post-test adjustment (McAllister, 2002) and psychological well-being. In the current study, the pathways to discovery illustrated in *something is wrong* and *out of the blue* suggested that the initial discovery of the family history of HD may be emotionally distressing, generating disbelief, confusion, shock and fear. These findings highlighted the distressed pretest state in which at least some test candidates might find themselves and raise questions about the ability to absorb and integrate complex information imparted during counseling sessions. Janis and Mann's (1977) decisional conflict theory suggested that stress interfered with the ability to consider the salient features of a decision situation and to deliberate carefully about the pros and cons of alternative options. Lerman *et al.* (1995), for example, found an inverse relationship between risk comprehension (for breast cancer) and levels of distress, suggesting that distress interfered with information processing. Counselors and clinicians working with individuals at risk for HD might explore their clients' experience of discovering the family history in an effort to identify potentially stressful pathways of discovery.

It is notable, however, that family experience with HD may alter the degree of distress experienced upon discovery, even for those who discover their family history out of the blue. In the current study, Gerald's story was illustrative of how disease-related events in the family could mitigate the distress associated with initial discovery. Since his parent did not manifest signs of HD until his early 70s, Gerald's initial discovery was not marked by significant emotional distress. In contrast, Lori had only recently discovered her family history of HD. She did not live in the same community as her parent, and this geographic distance limited her proximity to signs and symptoms of the illness. Unlike Gerald, she suggested that her initial discovery was marked by significant emotional distress. These findings highlight, once again, the importance of thoroughly exploring an at risk person's discovery of their family history of HD in an effort to understand, and hopefully mitigate, potential psychological distress.

CONCLUDING COMMENTS

Study Limitations

Findings of the current study are not generalizable. Participants were self-selected into the study, and it employed a relatively small sample of people at risk for an incurable genetic disease. While the size of the sample was in line with many qualitative studies, findings might not generalize to persons living at risk for other genetic disorders or even to others at risk for HD. Concerns about generalizability are alleviated somewhat, however, since generalizability is not the goal of research utilizing IPA. Rather, the goal is to capture how particular people perceive and respond to their experiences, highlighting the value of each particular case. Finally, the lack of a second analyzer of interview transcripts may appear to threaten the validity of the study findings. Throughout the study, however, rigorous research approaches to data collection, validation and analysis were adopted (e.g., constant comparison between and within interview transcripts, respondent validation, see Etchegary, 2005a).

The current study has shown that not all people at risk for HD discovered their family history in the same way or time. Pathways to discovery may have implications for predictive test decisions for HD, psychological well-being or the level of knowledge brought to counseling sessions. Future research could explore how risk for other genetic illnesses is discovered, providing valuable information for healthcare professionals who work with families fac-

ing genetic risk. Genetic risk for HD is notably different from genetic risk for multi-factorial diseases (e.g., inherited cancers); however, predictive testing for HD, in particular, has raised numerous clinical and ethical issues. As genetic tests become available for a variety of other adult-onset disorders, it will be important to know how those most closely affected by new genetic technologies understand and manage genetic risk information. An appreciation for how genetic risk is discovered may help address these issues.

ACKNOWLEDGMENTS

I am grateful to participants for their patience and trust. Without their generosity, such research would simply not be possible. I also thank represe ntatives from the provincial medical genetics clinic and the Huntington Society of Canada for their help with recruitment. The research presented in this article was generously supported by a doctoral fellowship from the Newfoundland and Labrador Centre for Applied Health Research and the Department of Psychology, Memorial University, both in St. John's, NL. This research was part of the author's larger doctoral thesis on living with genetic risk for HD. During the writing of this article, the author was supported by a Postdoctoral Assistantship from the GeneSens research group, Department of Epidemiology and Community Medicine and the Gap Santé research group, National Institute of Population Health, both at the University of Ottawa, Ottawa, ON. The author thanks postdoctoral supervisors Dr. Brenda J. Wilson and Dr. Louise Lemyre for their support. I am also indebted to the editor and two anonymous reviewers for their excellent comments and suggestions which markedly improved earlier drafts of this article.

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