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Original Research

What Do We Tell the Children? Contrasting the Disclosure Choices of Two HD Families Regarding Risk Status and Predictive Genetic Testing

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Above all else, predictive genetic testing provides information. Gaining insight into the psychosocial effects of this information is a primary goal of genetic counseling. For individuals utilizing predictive genetic testing, the acquisition of genetic information requires choices regarding disclosure within the family. This study uses a phenomenological methodology to explore the contrasting choices of two sets of HD parents regarding the disclosure of genetic risk status to their children. Additionally, the children (now adults) discuss their lived experience growing up with contrasting disclosure dynamics, and their current views regarding the use of predictive genetic testing for themselves. The primary finding of this study is that all of the adult children now express preference for early disclosure of genetic risk and an open/supportive communication style regarding HD. This finding has value for clinicians working with HD families who must make decisions regarding disclosure issues related to predictive genetic testing.

KEY WORDS: communication; disclosure; family communication; genetic counseling; genetic testing; Huntington's Disease; qualitative; predictive genetic testing; risk statusIntroduction.

INTRODUCTION

Above all else, predictive genetic testing provides knowledge. As a result, one of the most significant goals within the field of genetic counseling is to gain increasing insight into the psychological effects of having specific knowledge of genetic status through predictive genetic testing. This goal is especially important for predictive genetic testing concerning Huntington's Disease (HD), which holds the potential to identify an individual with the gene mutation while they are asymptomatic (MacMillan and Snell, 1993). Therefore, choosing to utilize predictive genetic testing for HD is a life changing event. In such cases, the client is encouraged to consider not only the objective information that will be made available as a result of the test, but also the psychosocial issues that may present themselves after the test results have been revealed (Agan and Gregg, 2002; Broadstock *et al.*, 2000; Sarangi *et al.*, 2004; Williams and Schutte, 2000). For example, one of the critical issues associated with the use of predictive genetic testing is the decision concerning the specific genetic information to be disclosed to family members (d'Agincourt-Canning, 2001). For parents, in particular, the issue of disclosure can be very difficult when considering what, if anything, to tell their children about their genetic risk status (Forrest *et al.*, 2003).

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BACKGROUND

Predictive Genetic Testing Related to HD

Although linkage analysis has been available for HD since 1987, the first predictive genetic test in the form of direct mutation analysis became available in 1993 when the specific gene mutation responsible for HD was identified (MacMillan and Snell, 1993). First described by Dr. George Huntington in 1872, HD occurs at the rate of approximately 1 in 10,000 people, with estimates that 30,000 people in the United States currently have been diagnosed with HD while another 200,000 remain undiagnosed and at risk (HDSA, 2005). With no treatment and no intervention recommendations, HD is "an untreatable progressive neuropsychiatric disorder characterized by involuntary movements, neuropsychological defects, and personality changes" (Evers-Kiebooms *et al.*, 2002, p. 168). Although HD is classified as a late onset disease, the timing of symptom presentation has been known to vary from as young as 2-years-old to as late as 90 years, with 40-years-old being the average age of onset (Young, 2003).

Predictive genetic testing for HD is able to identify the presence of the gene mutation while an individual is asymptomatic. Most importantly, the presence of the gene mutation indicates that the individual will develop the disease at some point in time (MacMillan and Snell, 1993). Once this information is known, the decision arises whether to disclose or not disclose the information to family members.

Predictive Genetic Testing and Family Communication

An important factor relevant to the use of predictive genetic testing, and subsequent disclosure decisions, is an understanding of the dynamics involved with family communication patterns. These patterns provide the context for important processes related to predictive genetic testing such as the communication of family history as well as the manner in which risk status is perceived and managed (Bowen *et al.*, 2004; Croyle and Lerman, 1999).

Family communication patterns have been researched through a number of theoretical perspectives. For example, in the 1970s, Chaffee and McLeod (1972) proposed that families could be identified primarily through two communication styles: *socio-oriented* and *concept-oriented*. Socio-oriented families placed the highest priority on maintaining harmony between family members and, as a result, limited any communication that would hold potential for causing disruptions to the family system. In contrast, concept-oriented families encouraged the free communication of ideas and emotions, even if it came at the expense of harmony within the family group.

This theory was revised by Ritchie (1991) with the designation of family communication patterns as either *conformity oriented* or *conversation oriented*. In conformity oriented families, the parents are perceived as powerful and the children are expected to conform to the parents' perspectives. In this system, communication is limited in terms of acceptable topics, the expression of emotions is discouraged, and the avoidance of conflict with parents is of prime importance. In contrast, conversation oriented families encourage the expression of ideas and emotions in an open manner, even if such expressions cause contention within the family.

Research specifically related to family communication patterns as they are impacted by chronic illness and/or genetic testing have produced several additional theories. For example, Bury (1991) identified two basic styles of communication related to chronic illness: (1) *accomodation*, characterized with open discussion regarding the disease and associated emotions, and (2) *active denial*, which restricts the topics that can be discussed regarding the disease. More specifically, Kenen, Ardern-Jones, and Eeles (2004) further expanded the categorization of family communication styles regarding disclosure of genetic risk factors related to breast/ovarian cancer (HBOC). In this study, five communication styles were identified: (1) *open/supportive*, in which one is free to discuss anything and everything regarding the disease and the experience, (2) *directly blocked*, in which strong boundaries are overtly drawn around acceptable and unacceptable topics, (3) *blocked indirectly,* in which the boundaries defining acceptable and unacceptable topics are subtly drawn through primarily non-verbal means such as silence, (4) *self-censored*, in which one consciously limits what is said due to sensitivity regarding the comfort level of another person, and (5) *third party*, in which an intermediary is selected to conduct the discussion on behalf of another. Furthermore, the self-censored style was found to consist of two sub-types. The *reactive self-censored* style would choose to limit what one says in a discussion due to the perception of discomfort in another person as the conversation is taking place, while the *proactive self-censored* style would choose not to enter into any

discussion of an issue out of anticipatory concern that it would cause anxiety in another person.

Additionally, previous research has suggested that there are unique communication dynamics surrounding the issue of disclosure. For example, in their meta-analysis of literature related to the disclosure of genetic information, Wilson *et al.* (2004) described disclosure as more than a single event in which there is a transfer of information. Rather, it is conceptualized "in the context of complex individual, familial and sociocultural beliefs, behaviours and (often) tensions" (Wilson *et al.*, 2004, p. 21). This process may be affected by several factors including the nature of the disease, whether or not preventive measures were available, the overall pattern of family communication, and the individual coping styles of family members. The process may also be impeded by feelings of guilt, denial, rationalization, and/or the desire to protect others.

Additionally, the manner in which families communicate may directly impact the extent of disclosure regarding genetic risk factors to children. While research has shown that many families believe it is the responsibility of the parents to disclose genetic information to their children (Forrest *et al.*, 2003; Wilson *et al.*, 2004), the parents may find themselves torn between wanting to protect the children for "as long as possible, but also knowing that they needed to be told in time to make key life decisions" (Forrest *et al.*, 2003, p. 324). Forrest *et al.* (2003) note that parents in HD families, in particular, may struggle with this delicate timing due to the nature of the gene mutation and its 100% penetrance rate. As a result, Wilson *et al.* (2004) note that the choice to delay disclosure may be perceived as a preferable option as parents seek to protect their children while providing a temporary sense of control. This may be especially true of those choosing a proactive self-censored style which was found by Kenen *et al.* (2004) to be "usually, but not always" (p. 341) associated with mothers considering disclosure to their children.

Finally, it is important to note that the dynamics of disclosure have been found to be influenced by issues of gender in families (d'Agincourt-Canning, 2001; Forrest *et al.*, 2003; Wilson *et al.*, 2004). In particular, it has been suggested that it is the women in many families who not only serve as the primary keepers of health information (d'Agincourt-Canning, 2001; Wilson *et al.*, 2004) but it is also they who understand and implement "implicit rules about who gets told, or not, and who take precedence in being 'allowed' to tell" (Forrest *et al.*, 2003, p. 324).

PURPOSE OF THE STUDY

The purpose of this specific study is to explore the contrasting choices of two sets of HD parents regarding disclosure of genetic risk status to their children. Additionally, the children (now adults) discuss their lived experience growing up with contrasting disclosure dynamics, and their current views regarding the use of predictive genetic testing for themselves. The goal of this study is to shed light upon the complicated process of disclosure to children, especially in the context of predictive genetic testing related to HD.

METHODS AND PROCEDURES

Methodology

A phenomenological methodology was chosen for this study, specifically Interpretative Phenomenological Analysis (IPA), which has gained increasing acceptance as a methodology suitable for research in areas of health related psychology and the new genetics (Chapman and Smith, 2002; Smith, 2004). In general, phenomenology seeks to facilitate the exploration of an individual's experience as they are living it, and as they perceive it (Smith, 2004; Smith and Osborn, 2003). In particular, IPA places an emphasis upon understanding the meaning that the individual has attached to the particular experience in question (Smith, 2004; Smith and Osborn, 2003). However, in contrast to other phenomenological methodologies, IPA places a greater emphasis upon the interpretative role of the researcher, who is expected to bring his/her own conceptualizations and understandings to the process (Smith, 2004; Smith and Osborn, 2003). In doing so, the researcher provides the primary means of interpretative work through seeking themes and concepts that will serve to interpret the participant's primary perceptions. As Smith (2004) states, "The researcher is trying to make sense of the participant trying to make sense of their personal and social world" (p. 40).

Procedures

Semi-structured interviews were conducted individually with each participant at a location of their choosing. All of the participants chose to be interviewed in their homes. The interviews were

approximately 90 min in length and audio-taped. Verbatim transcripts were completed from each taped interview and analysis was guided by the reductionistic techniques of phenomenology and IPA. The transcripts were analyzed for key words or phrases which were then clustered around themes that identified the essence of the experience (Moustakas, 1994; Smith and Osborn, 2003). For each participant, the themed clusters were incorporated into a written summary of the experience (Smith and Osborn, 2003). The final list of themes for each participant was then compared across the participant group to look for commonalities and variations (Chapman and Smith, 2002; Smith and Osborn, 2003). The result of this cross-comparision resulted in a list of superordinate themes which were then incorporated into narrative summaries of the experience (Smith and Osborn, 2003).

IRB approval for this study was administrated and granted by the Harold Abel School of Psychology at Capella University, Minneapolis, MN.

Participants

The number of participants was purposely limited by the study's choice of phenomenological methodology, with two families (eight individuals) participating. Eligibility criteria for participation included (a) a family history of HD, (b) at least one at risk family member had to be actively considering the use of predictive genetic testing for themselves at the time of the interview, and (c) the families had to live within reasonable travel distance to allow for face-to-face interviews. As a convenience sample, the families were recruited individually through personal contact, with Family One volunteering in response to a letter of inquiry regarding interest in research participation, and Family Two volunteering as the result of a discussion of the research study at an HD convention.

Family One consisted of five members, four of whom participated in the study. "Dad" (age 47) was symptomatic for HD, while "Mom" (age 45) was not at-risk for the mutation. Two biological daughters were each at 50% risk. Eldest Daughter (age 19) was considering the use of predictive genetic testing and was eligible to do so. Middle Daughter (age 16) was also considering testing and would be eligible shortly (at age 18). Youngest Daughter (age 10), while being too young to be included in the study, was adopted and was not at risk. The recalled family history with HD went back four generations and, with each generation, the age of symptom presentation had decreased. Great-great grandfather became symptomatic in old age, approximately in his 90s. Great-grandfather, too, had symptom onset in older years, approximately 70s. Grandfather's onset was in his 50s, and Dad's was in his 30s.

Family Two consisted of four members, all of whom participated in the study. "Dad" (age 49) was symptomatic with HD. "Mom" (age 47) was not at-risk for the mutation. Both "Son" (age 21) and "Daughter" (age 19) were at 50% risk, considering predictive genetic testing, and were eligible to do so. The recalled family history with HD included four generations on Dad's side, with each generation experiencing symptoms at a younger age: Great-grandfather at 60 years of age, Grandfather in his 50s, Dad in his 30s, and a paternal nephew who had recently undergone predictive genetic testing and had tested positive in his early 20s, although he was not symptomatic. For these reasons, both Son and Daughter were considering predictive genetic testing for themselves, especially now that they knew HD had entered into their generation of the family.

RESULTS

Family One: Choosing Disclosure

In Family One, there was a deliberate decision to disclose the children's risk status for HD while they were very young. This decision was the direct result of Dad's experience growing up in an HD family with a blocked communication style regarding the disease. Dad does not remember a time when his own father was not symptomatic with HD, however the symptoms were never explained to Dad as a child. Dad recalls:

> I didn't have that much concept of what was going on [with father] as far as Huntington's Disease was concerned. I mean I thought that he was hurt from some old injury 'cause he'd been in World War II, and I figured it was a war injury. Or maybe it had been something that I had done to him.

Dad finally learned that his father's condition had a name when pamphlets explaining HD suddenly appeared in the family home. Dad could never determine if it had been intended for him to find the pamphlets as a substitute for talking about the situation,

but he strongly resented learning about HD in this indirect manner:

> We [Dad and siblings] just didn't know what it [father's condition] was until Mom went to a meeting and brought home little pamphlets from the meeting and I had to read it in my bedroom. They'd been placed there, like a stack of letters. That's when I knew it wasn't anything that I'd done, it wasn't a war injury, it was Huntington's Disease. I'm just never sure [if I was intended to find the pamphlets or not]. With my Mom, you never know, and it's kind of like the "elephant in the living room." I don't know if you've ever heard that story. But, for my family, a lot of times, Huntington's Disease was the "elephant in the living room." She wouldn't talk about it. And, you know, it's kind of one of those things that a lot of kids did, a lot of kids' parents did. So, if you had personal problems, you just kind of hid that. So, you don't talk about it.

Determined to fight against ignorance concerning HD, Dad became very open in his communication regarding the condition and his own risk status:

> I began to be open with my dating relationships and stuff. I started telling everybody, and if they weren't able to handle that then I just didn't figure that they were worth having . . . I just felt like everything can be worked through and worked upon as long as you are open about it.

As the culmination of his own life experience, predictive genetic testing is now seen by Dad as a tool available to the next generation that will enable them to make the best decisions possible based upon knowledge and information. This knowledge is highly valued by Dad as he continues to fight against ignorance regarding HD, especially within his own family:

> I don't think [my Mother made the right decision not to talk about HD]. I think she made the wrong decision because ever since then, you know, you're just kind of wondering what the deal is . . . We [my wife and kids] went together to the HD convention and I was about as open as you could be. It's when they see other people at different stages, with each trying to fight their own battle, some people with a lot of heavy chorea movements, a lot of shaking and stuff. Some people grieve their kids who have died. That's pretty tough. But nevertheless, it's pretty important to realize that it's OK to have Huntington's Disease.

When Dad married Mom, she supported his decision for early disclosure to their children regarding the presence of HD in the family and the risk status each child faced. Mom explains:

> In my husband's family, no one talked about HD, no one. And we referred to it as "the elephant in the living room" because it is so big and so obvious, that no one can possibly miss seeing it, however, everyone tip-toes around it and pretends it doesn't exist. And, somehow, that is their coping mechanism, I guess. And we found that to be very unsatisfying and, really, very unhealthy. So, from very, very early on, my husband was open with the Huntington's Disease . . . It was very important to him to be honest about it and open, and that's how we've always been with our children, including our eldest daughter. I do think that our openness in dealing with the whole Huntington's Disease situation has influenced her, and is in part responsible for her attitude about it [predictive genetic testing] and her wanting to be responsible about the future.

Yet, Mom readily acknowledges that choosing disclosure does not lessen the psychological and emotional pain of living with a condition like HD. However, Mom hopes that the decision to place an emphasis upon open/supportive communication will, ultimately, prove empowering to the family in the midst of their struggles:

> To live with that burden of knowing that it [HD] will come one day, sooner or later, that is very, very difficult . . . It's like living with a death sentence that is passed on from generation to generation to generation. And not knowing for sure if your children are also under this death sentence or not . . . It's watching your soul-mate die inch by inch, losing function, losing the ability to do all of the, you know, drive, swallow, talk, walk . . . Yet there comes a point where you just need to face reality as it is and decide, "Where do we go from here? We need to make plans for the future" and so forth. It was time that we needed to face the Huntington's Disease squarely and to make plans for our family . . . I think that the way our family has dealt with things, it's kind of like you can handle anything if you just know what you're dealing with.

The decision for early disclosure, in combination with the open/supportive communication style in this family, has resulted in the two biological daughters growing up with the knowledge that they are at risk for HD. Family meetings were the central forum for these discussions, as Middle Daughter recalls:

> I remember Mom would have family meetings and she would sit all us kids down on the couch and she would sit Dad down and then she would sit down. Then we would talk about almost everything . . . sometimes we would talk about HD . . . The way my Dad found out about HD, and what my grandfather had had, was he found a pamphlet on the table. I mean, his parents didn't talk to him about it at all. And, my parents were determined not to make the same mistake with us. So, they told us, like, from

> "day one," you know, that there was a possibility that Dad would get HD.

Yet from the perspective of a child growing up with this ongoing discussion, the information initially caused ambivalence regarding the value of the knowledge:

> We were well aware of it, but we never really understood what it meant. And then, one time, the time I really remember was the time . . . Mom was telling us that our cousins didn't know about HD because their parents hadn't told them yet and we weren't to talk about it when we were in their house. I guess it's like knowing that Santa Claus isn't real when every other kid believes that he is. I mean, when we were there, I wanted so much to have their ignorance because they didn't seem worried at all whereas, every time Dad fell, I was kind of like, you know, out of the corner of my eye and then just a second thought, like "Maybe he has HD." And in other ways, I felt sorry for them because . . . I didn't want them finding out from a pamphlet on the coffee table . . . I was always really, really startled whenever Mom and Dad told me that story about Dad finding that pamphlet on the coffee table because our parents were always so open with us . . . and the fact that they had been so honest with us really helped me accept, you know, HD as just another thing that they were being honest about.

Now, at age 16, Middle Daughter struggles with depression and the reality of what HD is doing to her family. Yet, she believes that her parents made the right decision in choosing early disclosure and an open/supportive communication style about the disease:

> I would rather know about it and disregard it and still know about it than never know about it and find my Dad is really, really sick and have no idea why and find out, you know, from a pamphlet on the coffee table. I would absolutely hate that! And I think that their way is the best way, you know? I mean, we may disregard it, but that's our fault, not theirs.

Eldest Daughter is eligible for predictive genetic testing and is actively considering its use to determine her own genetic status. She, too, believes that her parents made the right decision in choosing early disclosure and an open/supportive communication about HD:

> Mom and Dad always wanted us to know about the possibility just in case. I really appreciate that . . . I mean, knowing the possibility made it so much easier than, if we'd never ever been told . . . I remember at least a couple of family meetings like that where we could just ask questions . . . I could ask questions and, although it's always uncomfortable, it's not exactly a fun topic of conversation, but it was not something that I felt they were ashamed about.

Eldest Daughter is also able to identify the influence of her parents' viewpoint upon her thinking regarding predictive genetic testing:

> My Dad was the one who really said, "It's important for them [the children] to know about it" and [Mom] was supportive. She understood why . . . Dad had learned what his father had by picking up a brochure on the table and it hadn't been talked about, he hadn't spoke with his family much, it just hadn't been discussed at all. And he had found that very difficult to adjust to, especially with, well, it's not so much the knowing or not knowing, it's the not talking. Then it makes it really hard to talk about anything. I know my mother really wished that my grandmother on my father's side had felt more able to talk with her because she had had the experience of living with a Huntington's Disease husband and my Mom really wanted to learn from her, but she didn't want to talk about it . . . It was private and she didn't want to talk about it even within the family . . . Because of the way that my father and my mother talked with us, and because of the way that my father acted about it, that may be a major influence in my wanting to be, to have it open, I mean, to know.

Most of all, the open/supportive communication pattern has been influential in Eldest Daughter's desire to pursue predictive genetic testing in order to know the truth:

> It's about the knowing and not knowing. They say it's easier not knowing. I've never found it easy. I just want to know . . . It's almost a relief. I'm *doing* something! And it's not like if I didn't know it would be any different. Whether I know or not, my genes are the way they are . . . I just want to know.

Eldest Daughter views the knowledge to be gained from predictive genetic testing as a tool that will empower her in a variety of ways regarding her health status and her future planning:

> I know there are reasons [why someone would not choose predictive genetic testing], but I really have a difficult time seeing why people would not want to know. I mean . . . I know that if it's positive, then there's still that uncertainty of when [the disease will become symptomatic], but you can sign up for the research, and take the medicine and stuff, and know what to look for, and know that things might not just be your imagination. And, if you're negative, then there's not much uncertainty about that anymore. That's not an issue in your behavior anymore . . . If I am positive, then I want to make a difference somehow. If I'm negative, I want to do that too, but if I'm positive, it would give me a little bit more limited timeframe.

Summary of Family One: Choosing Disclosure

To summarize the communication dynamics of Family One, they have an open/supportive communication style which has been coupled with the deliberate choice for early disclosure regarding the family's history of HD and the children's risk status. As a result of this decision, the two biological daughters have grown up with the knowledge that HD is part of their family history and that they, too, are at risk. Although individual family members continue to struggle with episodes of distress and depression from the progression of HD within their family, the family consensus regarding predictive genetic testing is that it can provide knowledge, thereby enabling the family members to be active in their united fight against the disease.

Family Two: Choosing Non-Disclosure

In contrast, in Family Two there was a deliberate decision not to disclose to the young children the family's history of HD and their own risk status. Instead, it was decided that disclosure would only be made, if Dad should become symptomatic and receive a definitive diagnosis of HD. Only at that time would the information be shared.

Eventually, Dad did become symptomatic, as was his father before him. When Dad received a definitive diagnosis of HD via genetic testing, disclosure was made to the children, who were adolescents at the time.

Dad characterizes the communication style in his family of origin as being "real open" regarding his father's condition. As a result, Dad values knowledge related to HD and is now supportive of genetic testing because of its potential for providing definitive diagnosis when HD symptoms are suspected:

> It's a good thing, you know. You can find the truth of the matter, and you need to find the truth of the matter. You have to know the truth about what you do. You know, you can take, get over, anything if you know the truth.

Although Dad is supportive of genetic testing to verify a suspected diagnosis as a result of symptom presentation, he does not necessarily endorse the use of *predictive* genetic testing while an individual is in an asymptomatic state. Mom explains that Dad, himself, only chose genetic testing when he was already showing signs of HD and wanted confirmation of their suspicions:

> He [Dad] had always been against genetic counseling or testing, the whole time we were dating, and even married. He just felt it was wrong, and so for him to finally just say he wants to go take the test, I was very surprised, but I think in your life you get to the point, you're on the fence and you can't move either way, you're paralyzed until you know. And I think he probably already knew, [and] I already knew before he went, that he probably had it.

This caveat on genetic testing has played a significant role in the parents' decision to choose non-disclosure within the family regarding the presence of HD and the children's risk status. This decision was centered in the desire to protect the psychological and emotional health of the family as a whole. Mom explains that, unless someone is already symptomatic at the time of testing, the knowledge offered by predictive genetic testing only causes emotional turmoil, especially when it is related to HD, a disease for which there is no cure:

> I think [predictive] genetic testing is great as long as, if they can find out that you have it, it can be treated and cured or something, but it's very devastating when there's not a treatment, there's not a cure. So, for other people, I think it could be great. I guess for breast cancer, I think it's a very positive thing because if you have [the marker for breast cancer], you could start watching for it more closely, taking tests. You can cure breast cancer. So, to me, if it was a matter of our family had breast cancer, would I take the marker test? Yes, I would. And there's no question I my mind, I would! With HD, there is no treatment, there is no cure. So what, in some ways other than marriage and children, what are the advantages to knowing? So I guess for HD, I'm not very positive about it [predictive genetic testing] . . . I'm not ready to find out if my kids have it, but I'm not going to be ready 10 years from now to find out if they have it, so, I don't think it's ever going to be easy either way . . . The thought of knowing that my kids will get sick is very, very traumatic . . . It's very devastating when there's not a treatment, there's not a cure . . . I feel like there is a sense of no hope.

The preservation of hope for the children is an important reason that non-disclosure was chosen and the main reason that *predictive* testing is not embraced. Mom explains this view in relation to a nephew who tested positive for HD although he is asymptomatic:

> Basically the overall thing [predictive genetic testing], I think I'm kind of negative on, kind of negative, I guess, because, to me, I feel like there is a sense of no hope. Before you take the test, there

> could always be a portion that there is some hope, that maybe you will not get it. Like for my nephew [who used predictive genetic testing]. You know, if he had waited 10 years from now, he still would have that hope that he's not going to get it [HD]. Well, now, to me, I feel that hope has been taken away.

In order to foster this sense of hope, the children were not given the facts regarding the family's history of HD, even when Dad's sister was the first in his generation to be diagnosed with HD. It was not until several years later, when their Dad was diagnosed, that the children were told of their own risk status. Mom explains the difficulty of the choice to disclose at that time:

> I guess I really didn't want to concern them [the children] with that [knowledge] or stress them out, but then when it finally came to telling them that their Dad was sick, they were, they were devastated ... My daughter said we should've said something sooner, you know, that there was this disease and their Dad could get it. So, in some ways, I thought some of it had been communicated, but maybe not clearly ... I mean, I don't know what age you would tell your kids this. I guess I just thought, in a way, why tell them their Dad's at risk or they could get this or that if we don't even know if he has it?

Dad, too, shares concern about the predictive aspect of predictive genetic testing and its possible psychological impact upon the one being tested:

> There's a psychological aspect to it [predictive genetic testing]. That is, people definitely, if they know they're getting sick, they believe ... you see what I mean? If they take the test, there can be a "downhill" to it in that ... some people get sicker or stay weller [sic] depending on the psychology of the disease and whether they know or not.

The preservation of hope has played a central role in the parents' coping strategy ever since the time Dad was asymptomatic and he married Mom. She explains that, as the years went by, the sustaining power of hope ultimately provided the foundation for their non-disclosure choices:

> I knew, before me and my husband got married, I knew it was hereditary, so I knew he had a chance of getting it. At that particular time, there was no test to know, "Will he get it?" You don't know. And when we had children ... there still was no test to find out if you should or should not have children ... so [we] just took the chance and hoped for the best.

Recently, Son has begun to show signs of HD. At this point, Mom is supportive of his consideration of genetic testing because she no longer views it as *predictive:*

> For my son, with him thinking he's already showing signs, I think he needs, I think he needs to know. I think it's eating away at him not knowing because he sees so many signs that could be it. So, yeah, I would encourage him to do it ... We haven't really kind of thought through what are the pros and cons and how would we use that or what would we do with that [information]. I think with my son ... we have already, in our minds, see signs in my son. So, I think for us, it's not so much how is he going to use this information. I think it's just to give us verification of what we already think or what we already think we know.

As for Daughter, who is asymptomatic, Mom states that she is willing to support Daughter's own decisions regarding predictive genetic testing, but Daughter explains her impression of Mom's position on the matter:

> It's mostly my decision [but] I know my Mom doesn't want me to get tested 'cause I know she's not, I mean my Mom isn't ready for anyone to know that anyone has HD or is going to get HD, you know? I'm not ready to know that my cousin has HD, or the gene for it. I'm not ready to know that. And I know my Mom's not, let alone me. I couldn't deal with it and I know my Mom couldn't deal with it ... I don't want to get tested. I see no need for it.

Although the children have grown into adulthood and they now know their genetic risk status, Mom describes the family's current lack of communication regarding HD:

> We really don't talk about it a whole lot ... But we're all in counseling, so maybe they deal with some of the issues in counseling. We still don't really talk about it a whole lot. I mean, I think they know they can come to me. I think we're at a point, or we were, especially after the diagnosis, I don't think they wanted to say anything and upset me and I didn't want to say anything to upset them. So you don't say anything ... Me and my son have talked about it ... He may have talked to his Dad, but the four of us have not sat down together and talked about it ... Me and my son have talked about it, me and my daughter have talked about it, regarding her brother. Me and my husband have talked about it regarding the son, but the four of us have not sat down and talked about it.

The children, now adults, clearly remember when they first learned of their family history of

HD and its implications for their immediate family. Daughter, 19, recalls:

> I had never heard of Huntington's Disease until my aunt was diagnosed . . . And I didn't think my Dad had it and I remember when my Dad got diagnosed . . . and it ended up being HD and I was surprised. And then, I don't know, it didn't really sink in until a few years later.

As an adult, Daughter does not feel that her parents made the right decision regarding disclosure and communication about HD. Instead, she believes that their choice for non-disclosure has only complicated the task that she now faces in adjusting to the truth:

> I didn't think it was fair for my parents not to tell me. 'Cause then, when you find out, you're just hurt and angry that they didn't tell you before. Plus you have to deal with the fact that HD is in your life . . . I mean I see why they didn't tell me, but I think they should have just not kept it a secret at least, you know? . . . I would tell my children 'cause I think they need to know what could happen.

However, when Daughter thinks about predictive genetic testing, she echoes the position of her Mom, fearing that having knowledge about her genetic status would directly affect the presence of hope in her life:

> I'm just going to assume that I have it [HD], but I don't want to actually know . . . Like I want some hope that I don't have it, but I'm just going to assume and live my life like I'm going to get HD . . . If you don't test, even though you want to live like you have HD, you want to have that hope. . .Yeah, 'cause with HD there is no hope . . . but I still have hope in the back of my head that I might not have it. I couldn't live without that hope, you know? . . . I never want to be tested unless I'm showing symptoms.

Son, also, has distinct memories of learning about the presence of HD in his family:

> My Dad had been showing signs for a long time and it was really hard when he got tested and stuff. We didn't really know what it was, me and my sister. We sort of knew what it was a little bit, we knew the name and stuff, "Huntington's Disease," "it's a disease that runs in our family," and "it's incurable." That's basically what we knew . . . But I remember when, you know, Dad got tested and he came back positive. I cried . . . I remember it like it was yesterday.

Additionally, he did not realize that he was at-risk for the disease until his Dad was diagnosed. It was only at that time that his parents told him and Daughter of their risk status for HD. That was approximately 3 years ago, and yet Son has only recently had it "sink in" that this could be happening to him:

> When I was in high school . . . I did a research paper on it [HD]. And it was kind of hard because I started seeing things, you know, for the first time and stuff and I guess it was then that I started thinking that maybe, maybe I should get tested.

Because of the blocked communication in the family, it was only by accident that Son learned that members of his family were concerned that they were beginning to see signs of HD in his own behavior. He learned of their suspicions one day when he logged onto the family computer after Daughter had been writing an e-mail and she had forgotten to close out the service:

> I could still see what my sister was writing and stuff and she was writing to a friend of hers about me having it now, you know, that there's a possibility that I have it now. And, so I think maybe my family's been really upset about me having it now . . . I think some of my best friends that have been with me for a very long time think I have it now . . . they're worried and stuff.

Son states that his family's lack of communication is a frustration to him when it comes to HD:

> We don't really talk about it . . . They do kind of, you know, they tiptoe around it and stuff. A little bit. I mean, they haven't told me, you know, for sure that they think I have it and stuff . . . Sometimes, I wish they would be more up front with it because, you know, they know a lot more about it than I do . . . I would like to know what they think.

Now, Son is considering predictive genetic testing because of his own suspicions that he is beginning to exhibit signs and symptoms of HD. Yet, he wants to be protective of Mom. Therefore, he does not try to discuss his situation with her. Additionally, Son does not believe that his sister should consider predictive genetic testing because she is not symptomatic and he knows Mom does not want to know the genetic status of her children unless they are displaying symptoms. He realizes how much of a toll the family disease has taken on Mom:

> My mom is very, very strong. And she's always, you know, been the bread winner here. She's the backbone of the family. And she's very, very family centered. She's very, very, very smart . . . She's very tough . . . I know that she cries a lot, but she never cries in front of us. At least me, I've seen her cry and stuff actually, but she's trying to keep tough . . . I probably would say [my sister should not use

> predictive genetic testing] right now, because, for my Mom's sake 'cause that'd kill her because, to be tested early and know, that'd be really bad.

At the same time, while Son is considerate of his Mom's position, he does not share it. He does not believe that having knowledge of his genetic status necessarily means a loss of hope in his life. Instead, Son has sought to establish communication with his Dad and his aunt, both symptomatic with HD, in order to gain perspective on his own situation. Son states:

> Well, my Dad and my aunt have become, like, huge inspirations and stuff, you know, to me and to everybody they come in contact with. And I kind of just hope I can become like them. My Dad has become more like a saint, he's very focused on God . . . He uses God in everything that he does and he's always, you know, praying and reading the Bible and stuff. And he's very, very others focused. Very, very evangelical . . . So my Dad is, you know, just very, very saint-like and he's kind of just, you know, an angel without the wings yet, you know what I mean? Like *It's A Wonderful Life.*

Summary of Family Two: Choosing Non-Disclosure

To summarize the choice of Family Two, the parents opted for non-disclosure to their young children regarding the family history of HD and their risk status. Disclosure was only made when Dad was diagnosed himself, and the children were adolescents. The decision for non-disclosure was made in an effort to protect the children psychologically and emotionally while preserving the hope that HD would not be a part of their lives. Therefore, the two biological children have grown up without the knowledge that HD is a part of their family history and that they, too, are at risk. The choice for non-disclosure has caused the family to discourage communication regarding HD and to draw a sharp distinction between genetic testing and *predictive* genetic testing. While the knowledge provided by predictive genetic testing is viewed as threatening, particularly to hope, the knowledge provided by genetic testing is accepted if symptomatic presentation has progressed to the point where the knowledge has become self-evident and cannot be denied. In such a case, the hope that HD will not affect the individual is no longer valid and does not need to be preserved.

For the children, now grown and eligible for predictive genetic testing, there is the added complication of family loyalty and the desire to protect Mom. As a result, varying attitudes regarding communication have occurred in the family regarding predictive genetic testing as Daughter (now asymptomatic) follows Mom's lead in avoiding genetic knowledge while Son (now symptomatic) has sought to establish communication with Dad in order to deal with his own possible HD status.

DISCUSSION

The findings of this specific study support the suggestion of previous research that individuals considering predictive genetic testing for themselves are influenced by the dynamics of the family unit, when it comes to the decision-making process regarding predictive genetic testing (Brouwer-DudokdeWit *et al.*, 2002; Chapman, 2002; Coyne *et al.*, 2000; Forrest *et al.*, 2003; Hagoel *et al.*, 2000; Sanders *et al.*, 2003; Sobel and Brookes Cowan, 2000; Sobel and Brookes Cowan, 2003). All four of the adult children display evidence of direct familial influence on their current thinking regarding predictive genetic testing. For the children from Family One, predictive genetic testing represents empowerment in the form of information, a position which is expressed by the parents. In Family Two, the children share their parents' view that genetic testing is useful only if, one is presenting with symptoms, as Dad was when he was tested. In the predictive form, genetic testing is viewed with skepticism and hesitancy in the belief that it may ultimately represent the loss of hope. This is true even for Son who accepts his own use of predictive genetic testing, but only as a non-predictive tool due to his suspected symptom presentation. Aside from symptom presentation, he would not encourage predictive genetic testing for his sister, who is now asymptomatic.

Additionally, this study supports previous research in identifying the desire to protect as a central issue when non-disclosure is chosen (Forrest *et al.*, 2003; Wilson *et al.*, 2004). In Family Two, this desire was articulated by Mom in relation to the children as the rationale for her choice in the matter. Furthermore, the work of Kenen *et al.* (2004) was supported by the identification of proactive self-censoring as the primary communication style between Mom and the children in Family Two. As Kenen *et al.* (2004) noted, this style is most concerned with an anticipation that a discussion of the topic would cause anxiety in the other person, resulting in avoidance of the topic altogether. In Family Two, Mom indicated that the anticipation of anxiety inhibited the family's communication as a

whole, beginning with her desire not to upset the children with disclosure. However, it is interesting to note that the proactive self-censoring style was not only adopted by Mom toward the children, but also later by the children toward Mom, as evidenced by the statements of the two children in Family Two regarding their desire to protect Mom from feelings of distress.

Finally, this study had a mixed result regarding the issue of gender as an influence upon disclosure decisions as suggested by previous research (d'Agincourt-Canning, 2001; Forrest *et al.*, 2003; Wilson *et al.*, 2004). In Family One, the primary influence in choosing disclosure was the conviction of Dad who had grown up in a family system of non-disclosure. In Family Two, however, the primary influence in choosing non-disclosure was Mom, although her decision was ultimately based upon the coping style that had developed jointly between her and Dad over the years. It is unknown how this finding would be affected by variation in the gender of family members symptomatic with HD. In this study, the symptomatic member in both families was male.

While this study demonstrates that the parental decision regarding the question of disclosure is complicated and heartfelt, with both families seeking to provide the best options for their children, this study also moves the discussion forward by providing insight into the lived experience of the individuals who grew up with contrasting disclosure choices. As such, this study offers the opportunity to hear directly from these children, now grown, and to consider their stated disclosure preferences as adults. All 4 of the adult children expressed a preference to learn of their genetic history and risk status early in life through disclosure from their parents. Additionally, all four of the adult children expressed a preference for an open/supportive style of communication regarding HD within the family.

This finding can provide insight to genetic counselors, and other healthcare providers, who work with HD families facing questions regarding the use of predictive genetic testing. Incorporating analysis of family communication patterns into clinical assessment may prove beneficial not only to the families, but also to the clinicians for whom issues of disclosure and non-disclosure may have ethical implications. Additionally, an awareness of communication patterns within a family can equip the clinician to be alert for signs of ineffective communication or indicators of risk associated with HD such as depression and suicidal ideation (Dawson *et al.*, 2004; Kent, 2004; Wood *et al.*, 2002).

It is also important for clinicians to note that research has demonstrated that family communication patterns can be altered and improved through training in communication skills (Riesch *et al.*, 2003). This is especially true as children approach adolescence. Riesch *et al.* (2003) note, "Participation in communication training by families who are on the cusp of adolescence may improve family relationships and communication, which, in turn, may promote optimal developmental outcomes among adolescents" (p. 164). As Forrest *et al.* (2003) have indicated, this information may be particularly helpful to HD families struggling with disclosure issues as the children approach adolescence and the time is coming for "key life decisions" (p. 324). Understanding and addressing family communication dynamics can be a critical aspect of assisting a client to move toward a greater level of informed consent as they consider predictive genetic testing. Phenomenological studies such as this help genetic counselors, and other healthcare providers, to understand the lived experience to a greater extent.

LIMITATIONS AND ASSUMPTIONS

There are several limitations to this study. First, the sample size is small with eight individuals in two family units. Therefore, the findings of this study must be recognized as being specific to this group, and the results must not be misapplied for purposes of generalization. To facilitate appropriate generalization in this area, larger samples and supplemental data (such as quantitative measures) may be necessary.

Secondly, one of the significant aspects of this study is that it focuses upon the broader context of the family unit, as opposed to having an exclusive focus upon the individual test candidate. However, it must be noted that this study was conducted by means of individual interviews with each family member. Therefore, there is a limitation in regard to the direct observance of family dynamics in a group setting. Perhaps valuable insights could be gained by conducting future research within family groups in addition to individual interviews.

Thirdly, this study may be limited by the fact that the symptomatic member in both families was male. How the findings of the study would be altered with variation in the affected family members is unknown.

Finally, it must be noted that this study was conducted with a convenience sample of volunteers and, as such, this group may have characteristics that would differentiate it from a random sample of HD family groups. For instance, convenience samples may exclude important characteristics found in a larger, random participant group. Therefore, the possibility of both the inclusion of unique attributes within these specific volunteer families, and the exclusion of important attributes of a random sample, must be considered a limitation of this study.

Two basic assumptions serve as foundational elements for this study. The first assumption is that it is possible to gather significant data from a small sample within the framework of phenomenologically based methodologies. The value of phenomenological methodology is to gain a greater understanding of the lived experience of individuals in a given situation.

Secondly, although the results of this study are specific to the participants, it is assumed that there are underlying commonalities in HD families eligible for predictive genetic testing and, as such, there is scientific benefit in investigating the experience of individual family units such as these with histories of HD.

FUTURE DIRECTIONS

Although predictive genetic testing is currently available for a limited number of genetic diseases, rapid test development is anticipated, with the expectation that predictive genetic testing will eventually become a commonly accepted tool in primary care. As a result, there is a continuing need for studies exploring issues associated with genetic testing in general. Two specific areas for future research have been identified as (1) family communication patterns and (2) the decision making process, both as they are related to the disclosure/non-disclosure of genetic information. These topics have implications not only for individuals and families considering the use of genetic testing in general, but also for clinicians who are hold ethical obligations implicated by disclosure issues. Therefore, the continuing development of effective interventions related to disclosure and family communication patterns is an important area of future research and discussion.

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