

Predictive testing for Huntington's disease: an exploration of the
partner's role in decision-making

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by
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ABSTRACT

Predictive testing for Huntington's disease: an exploration of the partner's role in decision-making

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Program in Genetic Counseling

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Huntington's disease (HD) is a progressive neurodegenerative disease that causes motor and cognitive impairment. Symptom onset is typically during adulthood and HD is inherited in an autosomal dominant manner. Predictive testing is available for at-risk individuals who desire to learn whether they have inherited the HD gene mutation. Factors contributing to the decision regarding predictive testing have been studied many times in the at-risk population. Few studies, however, have focused on the partners of these individuals to explore their views and opinions about predictive testing. In this study, we interviewed the spouses of six individuals who underwent presymptomatic predictive testing to better understand their thoughts, feelings, and specific roles throughout the decision-making process. Several important findings emerged from the interviews. First, we found that spouses preferred earlier predictive testing than their at-risk partner, but remained extremely supportive of their partner's decision to delay testing. Second, we identified a lack of understanding regarding the full purpose and benefits of pre-test counseling sessions. Two of the six couples opted out of pre-test counseling through an HD

center because ‘they already knew what HD was,’ but ended up receiving inadequate information and support. Finally, we provide further evidence that spouses’ needs for additional support following a positive test result for HD are often overlooked. Genetic counselors possess the necessary training to assist couples through the decision-making process, educate them regarding pertinent information during a pre-test counseling session, and provide support as well as resources to these patients and their families.

Key words: Genetic counseling, Huntington’s disease, HD, predictive testing, spouses perspective, pre-test counseling, spousal support, forgotten persons

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Introduction

Huntington's disease (HD) is a progressive neurodegenerative disease with an autosomal dominant pattern of inheritance. The molecular cause of HD is an expansion of at least 36 (CAG) trinucleotide repeats in the 5'-coding region of the IT15 gene. The disease is fully penetrant with a prevalence in the United States of approximately 1/10,000. Onset of symptoms can occur at any age, but commonly begins between the ages of 35-50 (Ross and Tabrizi, 2011). There is an inverse relationship between the number of CAG repeats and the age of onset, so individuals with earlier onset ages typically have more repeats.

Huntington's disease symptoms are broadly described as a triad consisting of motor impairment, psychiatric disturbance, and cognitive decline. Symptoms manifest in unique and unpredictable combinations (Zinzi et. al, 2007). Motor disruption, the predominant initial symptom, presents as choreiform movements coupled with a progressive loss of voluntary motor coordination. Individuals affected with HD can experience a multitude of psychological or psychiatric symptoms ranging from depression and anxiety, to mania and personality changes (Duff et. al, 2007). Hubers et. al (2012) report that the suicide rate among HD patients is over four times that of the general population. Cognitive declines are gradual and culminate with profound dementia. Individuals can also experience deterioration in their memory and communication skills. Huntington's disease generally results in death from complications within 15-20 years after the onset of symptoms.

Direct predictive testing for Huntington's disease became available in 1993 subsequent to the identification of the IT15 gene. The availability of this test allows at-risk individuals to learn whether they carry the expanded IT15 gene, which is diagnostic for Huntington's disease. Surveys of at-risk individuals prior to 1993 suggested approximately 60% of people would pursue testing once it became available (Evers-Kiebooms et al., 1989; Tibben, 2007; Keenan et al., 2013). However, the documented uptake of predictive testing for HD since it became available ranges from 5%-20% of the at-risk HD population (Evers-Kiebooms and Decruyenaere, 1998; Wahlin, 2006, Walker, 2007).

Individuals living at-risk of developing HD have identified many factors that influence their decision to pursue or not pursue predictive testing. Common reasons in favor of testing include: to have certainty, to revise their children's risks of developing HD, to inform future reproductive decisions, and to plan for the future with regard to marriage, career, and finances. Individuals are more inclined to pursue testing when beginning to plan for and make decisions about their future (Evers-Kiebooms et. al, 1989; Richards, 2004). The reasons at-risk individuals provide for refraining from testing include: worry about being unable to cope with a positive result, the desire to hold out hope, the absence of a cure, and the fear of discrimination. Recurring themes in the literature continue to reinforce that the decision is very personal and individualized (Tibben, 2007).

The partners and spouses of individuals who are at-risk experience as much or even more psychological distress following a positive test result as the HD carriers themselves (Decruyenaere et. al, 2005; Williams et. al, 2000). Partners were observed to express higher levels of depression, anger, worry, and anxiety compared to the carriers. One analysis examining participants' coping strategies found that partners intuitively utilized

more passive coping, such as withdrawal behaviors, which are generally correlated with higher levels of long-term distress. When asked why they never sought out more psychological support, partners frequently said they did not feel they had a right to grieve and/or burden the carrier (Lowit and van Teijlingen, 2005).

The depth and diversity of partner's opinions regarding predictive testing is an area not yet fully explored. Evers-Kiebooms et. al (1989) studied the specific motivations for testing in at-risk individuals and their partners. Predictive testing was not readily available at that point, so the questions and subsequent analysis were purely hypothetical. However, when asked if they intended to pursue testing, 66% of at-risk individuals said 'yes' compared to 74% of their partners. Factors identified most often from both at-risk individuals and their partners in favor of predictive testing were the need 'to have certainty' and 'to make arrangements for the future'. The factors contributing to both groups' hesitancy toward predictive testing included 'because there is no treatment', 'because I prefer uncertainty', and 'because a bad result might be too difficult to handle' (Evers-Kiebooms et. al, 1989). Richards (2004), with HD testing a reality, analyzed decision making in regard to predictive testing within eleven couples. The results showed the majority of couples were in accord, however, 3 of the 11 couples disagreed about the decision to undergo predictive testing (Richards, 2004).

The primary goal of this study is to further explore the attitudes regarding predictive testing of individuals whose partners were known to be at-risk for Huntington's disease. We interviewed the spouses to elicit information about their opinions of predictive testing, the factors that informed these opinions, and their experience of going through the predictive testing process with their partners. We also asked the spouses for

advice about the predictive testing process to assist future couples in collaborative decision-making for such testing, as well as for professionals who work with at-risk individuals and couples. Data analysis revealed themes that strengthen our understanding of the decision-making process and provide insight for how healthcare professionals can better support couples considering and undergoing predictive testing to ensure they are making an informed and well thought out decision.

Methods

The Brandeis University Institutional Review Board approved the following study protocol.

Sampling Methods

We recruited participants for this study through Karen Baker, LICSW, the New England Regional Social Worker for the Huntington's Disease Society of America (HDSA). Participants were also recruited through Boston Medical Center and Albany Medical Center movement disorder clinics. We distributed a recruitment notice (Appendix A) to individuals outlining the study and eligibility criteria. Subject inclusion criteria included the following:

- All participants must be a partner/spouse of an individual that underwent predictive testing for Huntington's disease within the last 10 years
- All participants must have been in a relationship with their at-risk partner at the time of the testing
- The predictive testing must have been sought out pre-symptomatically
- All participants must speak English and be at least 18 years old

Six respondents who expressed interest were eligible for study participation and telephone interviews were scheduled at their convenience. We emailed a copy of the informed consent document (Appendix B) to each participant prior to the interview. The participants were asked to verbally consent on the audiotape prior to the start of the

interview. All six interviews were conducted over the phone and each participant received a \$25.00 gift card for their participation in the study.

Interview Design

We designed a semi-structured interview guide (Appendix C) with open-ended interview questions to facilitate a conversation about the participants' experiences with HD predictive testing. The interview design was based on previous literature as well as the committee members' previous personal and professional experience. Prior to the start of the study, we pilot-tested the interview guide with a committee member who is the spouse of another committee member who presymptomatically sought out predictive testing for HD. These responses were not included in the data sample. The first author interviewed all participants using the interview guide. The interviews focused on the decision to pursue presymptomatic predictive testing and understanding what factors contributed to that decision. We explored the participants' experiences from when they first learned of their partner's family history of Huntington's disease through the testing process and beyond. Though the same interview guide was used for each participant and all the questions were asked to all participants, some questions were adapted based on the thoughts and experiences expressed by the interviewee.

Data Collection and Analysis

Interviews lasted between 30-45 minutes and all interviews were audiotaped. The audiotaped interviews were transcribed, any potentially identifying information was removed, and the transcript was labeled with an identification number prior to coding and

analysis. All documents containing identifying information were stored in a password-protected database.

Atlas.ti qualitative analysis software was used to code the transcripts and organize the data into themes. Open coding was used to identify significant topics mentioned by the participants. Codes and key quotations were then grouped into themes. Participants' responses were then categorized into subthemes.

Results

Participant Demographics

We interviewed six spouses, three men and three women. The spouses were all from the Midwest or Northeast United States. Their ages ranged from 27-51, with a median age of 38.2 years. The spouses reported the length of their relationships ranged from 5-31 years, with a median relationship length of 16.8 years. Five of the spouses said the predictive testing occurred within the last four years, and the sixth spouse stated their testing occurred almost nine years ago. Demographics for the spouses and their partner's test results are listed in Table 1.

Table 1: Demographics of Study Participants

Gender of Participant	Age of Participant	Length of Relationship	Year of predictive test	Spouse's test result
Female	42 years old	21 years	2005	Positive
Female	51 years old	31 years	2010	Negative
Male	27 years old	10 years	2010	Positive
Male	44 years old	26 years	2012	Positive
Female	30 years old	5 years	2012	Positive
Male	37 years old	8 years	2012	Negative

Three main themes emerged from the interviews: spouse's thoughts and feelings regarding the family history of Huntington's disease, spouse's opinions about predictive testing, and spouse's responses to test results. Each of these themes encompasses several subthemes, outlined below in Table 2.

Table 2: Outline of themes and subthemes

Themes	Subthemes
Spouse’s thoughts and feelings regarding the family history of HD	Positive outlook
	Under-informed but accepting
	Fatalistic outlook
	Fearful outlook
	Was not discussed
Spouse’s opinions about predictive testing	Reasons in favor of testing
	Reasons against testing
	Desired testing before at-risk spouse did
	Supported spouse’s decision
	Opted out of pre-test counseling
Spouse’s responses to test results	Concern for family/children
	Staying positive after a positive result
	Spouses need more support
	Making reproductive decisions
	Getting involved in the HD community

THEME: SPOUSE’S THOUGHTS AND FEELINGS REGARDING THE FAMILY HISTORY OF HD

The first section of each interview focused on how the spouse learned of their partner’s family history of HD and how they felt upon learning this information. Responses portrayed a broad range of emotions both within the spouse and between spouses.

Subtheme: Positive outlook

All six spouses described some positive thinking when learning about the disease or contemplating their futures. Several found that the 50/50 risk and typical adult-onset of symptoms were reassuring and allowed them to think more positively. One husband commented:

“50/50, they’re not bad odds, I mean it’s better than a 90/10 chance or a 100% chance. You know, it’s a 50/50 chance that she has it or she doesn’t have it. So again, we’re trying to stay positive, as opposed to dwelling on the negatives of this horrible disease.”

Another husband drew on his knowledge about gene expansion likelihoods as a source of positivity:

“For us, with the idea that when it’s passed from the mother the gene is more stable, we’d still be looking at some really good years ahead, and we probably would live life to its fullest, so to speak, more so than if you know that you might have more time than that.”

Other spouses also found hope in the possibility of future technological and medical advances providing a cure or more effective treatment options.

Subtheme: Under-informed but accepting

Five spouses discussed their acceptance of the disease risks and implications without having a comprehensive understanding of HD. These spouses described their thoughts and feelings when they first learned of their partner’s family history of HD, which for most spouses was in the early stages of their relationships. One wife said:

“He told me his dad passed away from something called Huntington’s disease, and I had absolutely no idea what that was at the time. A few months later, he told me there was a 50% chance that he would also get Huntington’s, and since our relationship was getting more serious he wanted to let me know. And I said okay, no big deal. Again, I really didn’t know anything about it.”

One husband reflected back to when he was first told about the history of HD:

“I knew it was a big deal, but I don’t think I realized how big of a deal it was. I was just like, “Yeah okay whatever. It’s Huntington’s disease- it’s just another disease.” I guess I didn’t full comprehend it at all. I don’t want to say I wanted to deny that she could have it. It just didn’t seem like that big of a deal to me at the time.”

Two other spouses also focused on the fact that HD is typically adult-onset, which lessened their initial perception of Huntington’s severity.

Subtheme: Fatalistic outlook

Two spouses expressed feelings of fatalism. They were informed of the disease implications, but felt it was unproductive to dwell on the risks because they could not change the test result or disease course. One husband said:

“Well, it was just one of those things where, you know, what can you do about it? I mean there’s no cure, and you have a 50/50 chance of having it. So there was nothing we could do.”

One wife was accepting of future implications, but still desired to focus more on living in the present moment, saying:

“We just deal with things as they come up. But, I think the most important thing is to not let something kind of take over your life. I mean, just live your life every day, and whatever is going to happen is going to happen, and once it does then we’ll just take care of it then.”

Subtheme: Fearful outlook

All six spouses disclosed that they held fears about Huntington’s disease and the direct implications for their families prior to testing. Three spouses stated that Huntington’s disease is ‘scary.’ One husband discussed how involved the disease is and said:

“The disease is absolutely horrible. It comes at you in many different facets. It’s not just, you know, “Oh I have the disease and it deals with my health.” It deals with your life as a whole. And it deals with your family as a whole. So it’s not just one person suffering from the disease. We’ll all suffer as a family.”

Another husband expressed his frustrations about being unable to do or fix anything in terms of the Huntington’s risks in his wife, stating:

“One of the hardest things that I’ve had to deal with is that this is something I can’t fix. I can’t make it go away. I can’t help her or bring her to a doctor to fix her. There’s no amount of money that can make a cure for this right now, today. So there’s nothing I can do to fix what’s potentially going to go on with my wife.”

Subtheme: HD wasn’t discussed

Two spouses stated Huntington’s disease was not openly discussed between them and their at-risk spouse. Besides the initial conversation that revealed a family history of HD, there were no other discussions. These spouses were not upset by the lack of

communication, but instead respected the partners' apparent desire to forgo engaging in these conversations. One wife said:

“He keeps his feelings to himself a lot. So I never really pushed him to talk about it, and I figured, personally, if I want to discuss something, especially something so upsetting and personal, I'll bring it up. So I kind of give other people that same benefit.”

THEME: SPOUSE'S OPINIONS ABOUT PREDICTIVE TESTING

During the second part of the interview, we asked spouses to discuss how they felt about predictive testing: whether they were in favor of or against it, what factors contributed to their views, and to describe their perception of how their at-risk spouse felt regarding predictive testing prior to being tested.

Subtheme: Reasons in favor of testing

The spouses discussed four main reasons that contributed to their decision to pursue predictive testing as a couple. These are shown in Table 3.

Table 3: Reasons spouses gave in favor of pursuing predictive testing

Reasons given in favor of pursuing testing	Number of spouses that discussed the reason
To gain information regarding their future	6
Desiring certainty about the result	5
To make decisions for family planning	3
To revise their current children's risks	2

All six spouses indicated the test result would provide beneficial information as they made major life decisions. They discussed advantages as being able to recognize symptoms earlier, make responsible financial decisions, as well as live fuller lives. One wife said:

“I mean, you're still not symptomatic, but you just know something, and you have

more information now so you can go about things differently.”

Five spouses stated they ‘just wanted to know.’ One spouse discussed the question of uncertainty being a ‘huge weight.’ Another husband stated succinctly:

“Well, we wanted to know. That’s not a question we wanted to keep lingering over our heads.”

Three of the four spouses who had not had children prior to learning about their partners family history of HD stated family planning was the biggest contributing factor to pursuing testing as a couple. They placed great importance on pursuing testing in the at-risk spouse in order to determine what reproductive route was necessary to have children without the risk of Huntington’s. One wife said:

“I would never be able to forgive myself if we just had children the old-fashioned way and never had any genetic testing done, knowing [predictive testing] was available to us and knowing we passed the risks on to our children.”

Subtheme: Reasons against testing

Spouses discussed four main reasons contributing to the decision to delay predictive testing as a couple. These are shown in Table 4.

Table 4: Reasons spouses gave that made them hesitant to pursue testing.

Reasons given against predictive testing	Number of spouses that discussed the reason
Not being ready to know the result	4
Fear of insurance discrimination	3
Because there is no cure	3
Fear of a significant negative emotional reaction	2

Four spouses stated their partners were initially unprepared to learn the test result.

Predictive testing is a significant decision and couples wanted to make sure there was no doubt that they were prepared for the result. One spouse stated his wife said:

“Do I really want to dread the next 25 years of my life knowing it’s coming?”

Half of the spouses had concerns regarding genetic discrimination. One spouse stated his wife even pursued testing using a pseudonym to avoid potential negative repercussions.

These spouses mentioned past concerns about health insurance and employment discrimination that were problematic prior to the initiation of the Genetic Information Nondiscrimination Act (GINA) in 2008. The Affordable Care Act continues to provide more protection against insurance discrimination for individuals with a pre-existing condition.

However, some still have concerns about genetic discrimination. One wife said:

“You have to have your affairs in order before you go get testing, everything from disability, life insurance, long-term care, and nursing home insurance. You have to have that in order before you make the decision to even go talk to anyone about being tested. Because otherwise, you’re uninsurable and then what?”

Two of the three female spouses were personally hesitant for their at-risk husbands to pursue testing due to a fear of their husband’s possible significant negative emotional reaction. None of the male spouses expressed similar concerns. One wife said:

“The scariest part for me was [my husband’s] dad ended up taking his own life within a couple years of learning his diagnosis. And so, I didn’t want my husband to go into that same mindset if the test came back positive.”

Subtheme: Desired testing before at-risk spouse

Four spouses indicated that they desired predictive testing for their at-risk partners as soon as they learned about the HD history but their at-risk partners were not yet ready

to pursue testing. The decision appeared to be an easier one for the partner. One husband said:

“My initial reaction was, ‘We should get tested.’ I wanted to know one way or the other. That is like having the question up in the air, well is she or isn’t she?”

Another spouse reflected back on a long relationship and remembered she desired predictive testing from the beginning. When her at-risk husband decided he was ready to pursue testing for himself, she said:

“I had waited a good 20 years for him to come to the place I’ve been at. So when he said he wanted to do it, I was like, ‘Okay, you want to do this. Great.’ Inside I was doing cartwheels but I was trying to stay calm.”

Subtheme: Supported spouses decision

Four spouses felt strongly that the decision regarding predictive testing was ultimately their at-risk spouses’ decision. These spouses provided full support despite their personal preference for testing earlier in life. One wife said:

“In a marriage most things are a joint decision. But going to get tested for a disease that has no cure and will kill you eventually, that’s probably the one thing in marriage I never, ever forced- I said my opinion but I could not force that on my husband because that’s the one thing that had to be up to him.”

These spouses also recounted reassuring their at-risk partners that a positive test result would not weaken or change the status of their relationship. One wife said:

“I had already decided whatever the outcome is, I’m going to be by his side absolutely, no matter what. Like I told him time and time again, the results weren’t going to affect our relationship. “

Subtheme: Opted out of pre-test counseling

Two of the spouses shared that they and their at-risk partner decided to opt out of pre-test counseling and the formal testing protocol through an HD testing center. Instead, these two couples asked their primary care physicians to order the testing. We asked the

spouses to discuss why they made the choice to opt out of specialized counseling, and one wife responded by saying:

“The biggest thing was we’d been living and breathing HD for 20 years so there just wasn’t anything that somebody was going to tell us that we hadn’t discussed a gazillion times. I mean it took my husband 20 years to make that decision. Obviously, he wasn’t making it rashly.”

The second of these spouses, a husband, stated:

“Going through the pre-genetic counseling, and all the other stuff, was not going to change the result. It was not going to change the outlook of what’s going on. My wife is just like, ‘Give it to me straight, and we’ll deal with it.’”

THEME: SPOUSE’S RESPONSES TO TEST RESULTS

During the third part of the interviews, we asked partners about their reactions after receiving the test results and to discuss how they have moved forward with their lives since learning the result. The spouse’s responses were separated into five different subthemes.

Subtheme: Concern for family/children

Five of the spouses discussed having immediate concerns regarding their family and children. The responses demonstrated a deep sense of love and protection for their families. One husband said:

“I mean as a parent, you live and you die by your children. And the initial shock was now there’s a possibility that [our children] have it. It was tough, both of our initial thoughts were for our children.”

One wife discussed the joys of receiving a negative result with respect to their children:

“I mean my husband was thrilled because he was free. And more importantly, he kept saying all he cared about was the kids. He didn’t care about himself.”

Another spouse, who has not yet had children, expressed his wife’s worries of wanting children soon so that she could be a part of their lives as long as possible. He said:

“I know my wife is getting antsy about it, just because she knows she has HD, so that clock is ticking, where someday she is going to show symptoms and she doesn’t want to wait too long and then her kid grows up and by the time our kid is having their own kids, she is out of the loop and already in a nursing home or something.”

According to their spouse, the two individuals who tested negative experienced survivor’s guilt, especially with respect to siblings that were positive or still living at-risk.

Subtheme: Staying positive after a positive result

All four spouses who received a positive result stated that the positive result was valuable information that allowed them to plan for the future. One husband said:

“Now we can set up so it’s not a financial hardship to the family, and that there’s money set aside for the kids, and that [my wife] does enough things with the kids that they remember her as this person, and not the person she’s going to become.”

These four spouses also spoke of the relief they received by having an answer even though the positive result was devastating. One wife stated:

“Even when the results were yes, I can’t exactly speak for him by saying it was some sort of relief. Being positive is not a relief by any means, but knowing is no longer that constant uncertainty.”

One husband recounted another benefit the positive test result has brought to their lives:

“Now we just take more vacations and try to do more things as a family.”

None regretted undergoing predictive testing.

Subtheme: Spouses need more support

One spouse felt like there was a lack of support for him following the positive result. He felt like his gene-positive wife received the appropriate amount of love and support, but he did not. This husband stated:

“When you go to the HD conventions, there are all these support groups but no real support group that deals with the other spouse, people like me that have been thrust into the position and hey now I have to deal with this too, even though I’m not symptomatic.”

This husband reflected back on the initial years following his wife's positive result and discussed how he might have advocated more for himself to get the support he needed, saying:

“When I was first thrown into all of this, I just wanted to sit in the back and not talk to anybody and just keep my mouth shut. Now that I've been into it for four years, I've learned you've got to talk to people about it, you've got to be open about the way you feel so you can get help and better deal with it.”

Subtheme: Making reproductive decisions

All three couples who pursued predictive testing in order to determine how to proceed with having children tested positive for HD. These three couples then explored their reproductive options to continue with the best option for them. They considered PGD, adoption, and not having children. None were willing to gamble on having a child with HD.

One husband said:

“Well, if she's positive we can't have children the normal way, just because that's a 50/50 chance, so we would have to go through in vitro or adoption.”

Two of the couples utilized PGD. Only one resulted in a successful pregnancy. The wife who was unsuccessful with PGD said:

“It was hard because we went through PGD for two years and it came back that all the embryos were affected. We still didn't have a child and we couldn't afford going through it again. It was hard making the decision that we weren't going to have kids. Probably harder for me than it was for him.”

The wife who is currently pregnant after utilizing PGD discussed how her pregnancy has helped them cope and move beyond the positive test result. She said:

“We're pretty lucky that we are now pregnant, so it's like a constant something to look forward to opposed to a negative, 'Well, this is now what our life is going to become.' It's turned something that could potentially be negative into a blessing.”

Subtheme: Getting involved in the HD community

Four spouses said they got involved in the HD community immediately after receiving their partner's test result, regardless of the result. They identified meeting people, learning about options, participating in studies, and supporting others as reasons for their continued participation. One husband said:

“She got involved right after we found out about the result. She wanted to get involved as much as she could. We are both very involved with HDSA, she goes down to the seminar in Florida, and we're involved in a study too.”

Both of the participants whose spouses were negative for HD still discussed getting involved in the HD community. One husband said:

“She's very active in a local support group for Huntington's patients and families. You know, we haven't necessarily talked about the negative test result in a while, so it's kind of shifted into, 'How can I support other families? How can I support mom? What are some things I can do?' And, she's still really worried about her two sisters that are still at-risk.”

Discussion

In this study, we interviewed the spouses of six individuals who underwent presymptomatic predictive testing for Huntington's disease in order to gain an understanding of the spouse's unique views and role in the decision-making process. We identified a number of recurring themes, some of which highlight specific ways genetic counselors and other medical providers can better inform and assist couples throughout the predictive testing process.

The decision to pursue or not pursue predictive testing is made based on a variety of factors unique to each individual and couple. In our study, the contributory factors in favor of testing were primarily to have information regarding their futures, to gain certainty, for family planning reasons, and to revise their children's risks. The contributory factors against predictive testing were not being ready to know, a fear of discrimination, the absence of a cure, and a fear of a negative emotional reaction following a positive test result. All of the above factors have previously been identified as contributory for both the at-risk population and their spouses (Evers-Kiebooms et. al, 1989).

Spouses in our study, however, reported fewer hesitations with regards to predictive testing compared to their at-risk partners. They were also predominately in favor of undergoing predictive testing immediately, while their at-risk partners needed more time contemplating the decision. Spouses generally felt the test result would only be informative for their futures and did not express concerns for any personal repercussions.

At-risk individuals may be more apprehensive about pursuing testing because their health and future would be compromised by a positive result.

The spouses in our study demonstrated unconditional support to their at-risk partners and reported they refrained from pressuring their partner into testing when they expressed uncertainty. Several spouses also reassured their at-risk partner that the test result would not alter their commitment to the relationship while simultaneously understanding that the result might change the planned trajectory of their lives. The support exhibited by the spouses in this study allowed the at-risk partners to comfortably postpone testing until they were ready. The fact that spouses reported no major adverse reactions after receiving the test results may demonstrate the preparedness of each couple as they ultimately pursued testing.

Receiving a positive test result for HD can be a devastating event with enumerable life-altering implications. Professionals are usually cognizant of the severity of these scenarios and provide ample support and appropriate referrals for the HD carriers. In the past, spouses of at-risk individuals and HD carriers were so consistently overlooked that they were retroactively identified as the 'forgotten persons' (Decruyenaere et. al, 2005). Spouses are now receiving increased attention in the literature, with researchers examining the specific ways a positive result influences their lives. Decruyenaere et. al (2005) first studied spouse's various psychological responses to a positive test result. They found that spouses experience at least as much psychological distress, and sometimes more, than the carrier themselves.

In our study, we did not directly ask spouses about how they coped with the positive result or for details about the type, quantity, and quality of support they received. However,

one husband did voice feeling like he received insufficient support. He discussed the lack of opportunities for the spouse in general, especially considering their immediate and long-term distress. Since the need for more spousal support is replicated and reinforced by other studies, it is important for professionals to have an increased awareness to the possibility of spousal suffering and consider referring spouses for more individualized support when appropriate.

Pre-test counseling is an integral component to all genetic testing. The pre-test counseling session provides genetic counselors and other healthcare professionals the opportunity to engage individuals in a discussion about the specific test including information on the disease, the testing methodology, the format of possible results, and general benefits and limitations of genetic testing. The United States Huntington's Disease Genetic Testing Group supports the importance of pre-test counseling in their published guidelines outlining recommended steps in predictive testing for HD (Nance et. al, 2003).

In our study, two spouses reported their at-risk partners decided to opt out of pre-test counseling and relied instead on their primary care physicians to order the genetic testing. The at-risk partner's decision to opt out stemmed from them already possessing a thorough understanding of HD as a result of having affected family members. At-risk partners felt they made the decision to pursue testing carefully and were fully informed in their personal choice. They therefore viewed the pre-test counseling session as an inconvenient redundancy and also a source of delay in their desire to learn the result.

We asked the spouses to describe the results session and their level of understanding based on the information provided by their primary care physician. One wife expressed significant disappointment in the careless way the results were delivered in

their physician's office, saying, "these results are supposed to be received in a proper way- it was not done in a proper way." She described feelings of anxiety, shock, anger, and sadness despite receiving a negative result for HD. This wife also discussed their confusion with the format of the test result. More specifically, they were unaware the result was returned as a repeat number and had trouble understanding the significance of the various ranges. The ramifications of this avoidable confusion were compounded further because this couple also had adult children who were impacted by his result. This wife further discussed that their physician was unfamiliar with HD and also confused by the test result, further preventing her and her husband from fully comprehending the implications of their test result.

The confusion and anxiety identified above would have likely been avoided if this couple attended a pre and post-test counseling session through an HD testing center. During the pre-test session, the couple would have been informed about the way results are returned and the format of results being broken down into a specific repeat number. It is important for any individual considering predictive testing to understand the specific purpose of pre-test counseling sessions to limit opt outs resulting from misinformation. Ideally, sufficient information and proper education regarding the benefits of pre-test counseling can be communicated through HD testing centers, support groups, and even physician's offices.

Clinical Implications

The spouses of individuals contemplating predictive testing for Huntington's disease fill a unique and challenging role in their respective relationships. This study demonstrates how healthcare providers can better collaborate with couples to facilitate informed decision-making both before and after predictive testing.

The pre-test counseling session is an integral part of the predictive testing process. In the absence of a pre-test session, genetic counselors are unable to perform their role or offer help to at-risk individuals and their families. Education throughout the HD community via support groups, online resources, and testing centers can highlight the importance and purpose of the pre-test counseling session from knowledgeable health professionals. Additionally, primary care physicians should be educated about the availability of predictive testing at an HD center including pre-test counseling to better ensure they can appropriately refer patients.

During pre-test counseling sessions, healthcare providers should also discuss the possibility of psychological distress following a positive test result not only in the at-risk individual but in their partner as well. Initiating an open discussion before testing may allow the partners to seek out support earlier and lessen the need to subjugate their own feelings in support of their carrier partner. Professionals trained in counseling can initiate a discussion about each individual's emotional well-being by checking in directly and expressing genuine concern. Normalizing an individual's reaction can engender a sense of

validation and relief. This could be achieved by saying, “Many people feel upset or angry following a positive result” or “Many spouses also feel a significant burden or disappointment, it’s normal to feel that way.” Once someone’s immediate distress is acknowledged, more effective support and referrals can be made for both partners.

Study Limitations

The major limitations of this study include the small sample size and diverse demographics; couples were of differing ages, in different stages of their lives, and had predictive testing at different times. Furthermore, all interviewed spouses were still in successful relationships with their partner throughout and following predictive testing. Their approaches to decision-making and predictive testing were likely more supportive and collaborative than in couples that did not stay together following testing. The attitudes of those willing to participate in this study may not be generalizable to the broader population of partners of those at-risk for HD due to their self-selection bias.

Future Research

Future studies might serve to gain the perspective of spouses no longer with their at-risk partners to understand why these relationships failed and to what extent HD contributed. Studies with a more specific demographic window and larger sample size could help validate our findings and expand on information relevant to clinicians.

Conclusions

We interviewed six spouses of individuals who underwent presymptomatic predictive testing for Huntington's disease. Two findings provide important insights for healthcare providers on how to better understand and assist unaffected spouses and couples as a whole.

First, we found spouses are not always receiving the support they desire following a positive test result. Given that a positive test result for HD also has significant implications for the spouse, their psychological well-being following testing should not be overlooked. Professionals can better support both individuals if each of their needs are addressed with appropriate counseling and support referrals.

Second, we found that two out of the six couples opted out of integral pre-test counseling sessions because of an under-informed interpretation of the purpose of pre-test session. Both individuals reported opting out of the pre-test protocol through an HD center because they 'already knew what HD was.' Healthcare professionals within testing centers, as well as professionals in the HD community, should continue to educate individuals considering predictive testing about the benefits of pre-test counseling given their knowledge of HDSA pre-test guidelines. Clarifying the purpose of pre-test counseling may help encourage individuals to pursue predictive testing through an experienced and recommended HD testing center, which could help to limit mistakes and confusion.

Furthermore, primary care physicians could be better educated about the availability and benefits of HD testing centers, thus encouraging them to refer appropriate patients.

Genetic counselors possess the necessary training to effectively assist these patients and their spouses in all facets by disseminating up to date information, ascertaining patient needs, providing support, and introducing applicable resources.

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Are you a partner of someone who has had predictive testing for Huntington disease within the last 10 years?

I am a graduate student in the Genetic Counseling Program at Brandeis University and I am seeking volunteers to participate in a research project for my Master's Thesis. The goal of my study is to learn what factors contributed to your opinions about HD predictive testing and how you and your partner made the decision to pursue testing. I hope that this information will give genetic counselors a clearer understanding of the predictive testing process from the partner's perspective to enhance our services and support for future couples undergoing predictive testing.

Who is eligible?

- You must have been in a relationship with your at-risk partner at the time of HD predictive testing
- At the time of their predictive testing, your partner must have been asymptomatic
- Participants must speak English and be at least 18 years old

What will study participation involve?

- A 35 minute audiotaped telephone interview
- All participants will receive a **\$25 amazon gift card** to thank you for your time and participation

If you are interested in participating in this study, please contact **Shelley Towner** at sct26@brandeis.edu by February 28th, 2014.

Please note participation is completely voluntary and confidential. No identifying information will be used and all audiotapes will be destroyed after I receive the transcripts.

I look forward to hearing from you!

Sincerely,
Shelley Towner, Genetic Counseling Student
Brandeis University, Waltham, MA

**Brandeis University
Genetic Counseling Graduate Program**

Informed Consent to Participate in Research

**Decision making about Huntington Disease predictive testing: an
exploration of the partner's role and perspective for those who decided
to be tested**

Principal Investigator: Judith Tsipis, Ph.D.
Student Researcher: Shelley Towner, B.S.

Dear Participant,

Thank you for your interest in my research study. Through talking with you, I hope to learn what factors contribute to your opinion about HD predictive testing and how you and your partner made the decision to pursue testing. Please take a moment to read the enclosed informed consent form. When I call you for the interview, I will first discuss the consent form with you. If you decide to participate in my study, I will have you consent verbally over the phone on the audiotape.

INTRODUCTION

Shelley Towner is a student at Brandeis University in the Genetic Counseling Master's degree program. Judith Tsipis is the Director of the Genetic Counseling Program at Brandeis University. We are conducting a research study as my (Shelley's) Master's Thesis to learn more about partners' opinions about HD predictive testing and to gain insights into the decision to be tested. You are being asked to participate because you are a partner of an individual that pursued HD predictive testing within the last ten years.

Taking part in this research study is voluntary. You should not feel any pressure to participate. You can decide to stop taking part in this research at any time for any reason. You can also choose to skip any question that you prefer not to answer. Please read all of the following information carefully. Please ask any questions that you have about this research study or your participation.

If you decide to take part in this research study, you will be asked to consent verbally over the phone. You should keep this copy for your records. It has information, including important names and telephone numbers, to which you may wish to refer in the future.

PURPOSE OF STUDY

The purpose of this study is to obtain an in-depth understanding of the experiences and emotions of individuals who were romantically involved with their partner at the time of testing for HD. Your partner must have been asymptomatic at the time of testing in order to participate in this study. It is our hope that the experiences shared by participants in this study will be useful in helping genetic counselors and other health professionals further understand the decision making process to provide more support to couples considering HD predictive testing.

PROCEDURES TO BE FOLLOWED

You will be asked to participate in an audiotaped telephone interview lasting approximately 30-45 minutes. During this interview you will be asked questions about your experiences with decision making for HD predictive testing. You will also be asked to reflect on how they differed, if at all, from the experiences of your partner. The questions will also inquire about how you made a decision as a couple to pursue testing.

RISKS

Participation in this study presents no more than minimal risk. However, it is possible that by taking part in this interview, you may experience thoughts or feelings that are upsetting to you. Should that occur, Dr. David Rintell, a licensed psychologist, is available to speak with you. He can be contacted on his cell phone at 617-734-6778.

BENEFITS

Participants will receive no direct benefit for participating in this study. We hope that in the future, information obtained in this study will help genetic counselors to gain a better understanding of the decision making process for HD predictive testing in couples where one member is at-risk for HD.

ALTERNATIVES

An alternative is to not participate in this research study.

PRIVACY AND CONFIDENTIALITY

During this study all records containing any identifying information will be kept strictly confidential. All study related materials (including interview transcripts) will be kept in a secure location accessible to only the student researcher and PI. Upon enrollment in the study you will receive an ID number, which will be used to code transcripts, interview notes, and audiotapes. If you are quoted or referred to in a written or oral report, you and your partner will both be referred to by pseudonyms. You, or your partner, will never be referred to by your real names. No other identifying information obtained from this interview will be disclosed.

PAYMENT

You will receive a \$25 gift certificate to Amazon.com for participation in the research study as a gesture of appreciation for your time and feedback. The certificate will be emailed to you after the interview is completed.

COST

There will be no cost to you to participate in the study, other than the time it takes to be interviewed.

WHOM TO CONTACT

If you encounter any problems related to study participation or have questions about the study, you may contact the Student Researcher, Shelley Towner, at sct26@brandeis.edu or (801) 597-1702.

You may also contact the Principal Investigator for this project, Judith Tsipis, at tsipis@brandeis.edu.

If you have questions about your rights as a research study subject, contact the Brandeis Committee for Protection of Human Subjects by email at irb@brandeis.edu, or by phone at (781) 736-8133.

Appendix C

Shelley Towner
Interview Guide
Huntington's Research

Interview Guide

Hi (name), thank you very much for agreeing to speak with me today. I really appreciate your willingness to share your story about predictive testing for HD. You are the expert on this subject and I want to learn about this experience from you. I hope the information I gather can help genetic counselors better assist individuals like you and your partner in the future.

I will be recording our interview so I don't miss anything you say. Your name will not be associated with any of the results of this interview and the audiotape will be destroyed after the transcription is completed. I will be asking you a series of questions. There are no right or wrong answers. If for any reason you feel uncomfortable answering any of the questions, please let me know and I will move on. The interview should last 30-45 minutes. Do you have any questions before we begin?

Did you receive and look over the informed consent form that I sent you? If so, great! I will now be turning the audiotaping on. Do you understand everything that was discussed on the informed consent? Do you voluntarily consent to being a participant in this study?

Great, now let's begin!

- 1) Tell me a bit about yourself and your partner
 - a. Demographics (How old are you?, M/F?, Where are you from?)
 - b. Relationship status/title (i.e. Married, Engaged, Living together)
 - c. Length of the relationship (How long have you been together?, How long have you been _____ (married, engaged etc))
- 2) Can you describe how you learned about your partner's family history of HD?
 - a. When/how did you find out?
 - b. Can you describe your feelings after learning this information?
- 3) How did you learn about predictive testing for HD? (i.e. partner, internet, professional etc.)
- 4) Can you walk me through your initial reactions when learning about the availability of predictive testing for HD? (probe: relieved, upset, hopeful, nervous etc.)
 - a. Were you in favor of or against it? Why?
 - b. If in favor, when did you want your partner to get tested? Immediately? Before marriage? Before having children? Etc.
- 5) Can you describe your perceptions of your partner's initial thoughts about predictive testing when you first discussed it with him/her
 - a. What were his or her reasons for or against testing?
 - b. If he or she were in favor of testing, when did he or she want to get the testing done?

- c. Did he or she always feel that way? Only since getting involved in a serious relationship?
 - d. If he or she were against testing, how would that have made you feel?
- 6) Can you describe the process for how you reached your decision as a couple to get tested
- a. How long did it take to reach the decision? How much time passed between the first discussion and getting tested?
 - b. If you and your partner's opinions varied initially, who made the final decision to be tested? (i.e. My partner decided, we decided together, my partner agreed to be tested because s/he knew it was important to me etc.)
 - i. If you decided together, how did you work it out?
- 7) Did either of you seek advice from others when making this decision? Tell me about that
- a. From whom did you seek advice?
 - b. What was the advice?
 - c. Did the advice influence your opinions?
 - d. Did you ever talk to a genetic counselor? If so, how did that conversation contribute to the decision to get tested, if at all?
- 8) When did your partner get tested?
- a. Why did you two choose to pursue testing at this particular time?
- 9) What was your partner's test result?
- a. What were your initial reactions to the result? What about your partner?
 - b. How are you both doing now?
- 10) What advice do you have for future couples faced with this decision?
- 11) What advice do you have for professionals working with couples that are trying to make this decision together?

We have now reached the end of the interview. Thank you again for your time and wonderful insight. If I had a follow-up question, would it be okay for me to contact you again? I wanted to remind you that David Rintell, a clinical psychologist, is available as a resource if you experience any emotional distress following this interview. Within the next few weeks, I will email you the \$25 amazon.com gift certificate. Thanks again, and please feel free to contact me with any questions or concerns.