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## **Working with Patients at risk for Hereditary Degenerative Brain Disorders**

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Working with Patients at risk for Hereditary Degenerative Brain Disorders

by

Stephen John White

Bachelor of Science  
Iowa State University, 2010

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Submitted in Partial Fulfillment of the Requirements

For the Degree of Master of Science in

Genetic Counseling

School of Medicine

University of South Carolina

2013

Accepted by:

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## **Dedication**

To my grandmother, Marilyn White, and my late grandfather, Clifford White. Both developed Alzheimer disease, and have since become my inspiration toward the study of hereditary degenerative brain disorders. They lived and loved with no cure. May the next generation take courage from their examples and continue to hope for the day Alzheimer disease is cured.

## **Acknowledgements**

Thank you to Jill Goldman for being the first to listen to my ideas about this project back when I was just an unknown student in another state. Thank you for your time, patience, and for giving me the feedback to make this thesis work.

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Thank you to Peggy Walker for aiding me in this process of creating a thesis. I could always tell you cared about me and my successful completion of this project despite all your other duties. Thank you.

## **Abstract**

Hereditary degenerative brain disorders (HDBDs) are a unique class of genetic conditions that result in progressive loss of function within the nervous system, many of which present during adulthood. Given this, the diagnosis of a HDBD can be daunting for both the patient and the genetic counselor assisting in medical care. The purpose of this study was to find themes among genetic counselors who see this patient population and help provide a framework to counselors entering this field by recognizing verified methods of HDBD counseling. Sixteen genetic counselors who routinely see patients at-risk for HDBDs were interviewed concerning how they prepared for and engaged with their patients, how they handled the complex emotions frequently associated with presymptomatic HDBD counseling, and how they perceived the stress levels and coping mechanisms of their patients. The results of this study showed that genetic counselors who see patients at-risk for HDBDs utilize agendas and/or protocols in preparing for and directing sessions. Additionally, these genetic counselors perceive their HDBD patients as capable of coping with their own presymptomatic testing results. There was not enough evidence from this study to determine if genetic counselors who see patients at risk for HDBDs are at any greater risk for compassion fatigue than other genetic counselors. Overall, the genetic counselors who see patients at risk for HDBDs are more similar to than different from genetic counselors who do not see this patient population. Given that a significant number of people are living at risk for HDBDs, experiences in

case observation and counseling alongside expert HDBD counselors could benefit genetic counselors and the patients they serve.

*Keywords:* Hereditary degenerative brain disorders, Huntington disease, Alzheimer disease, adult-onset neurodegenerative disorders, presymptomatic genetic testing, genetic counseling

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## List of Abbreviations

AD .....	Alzheimer disease
CAG .....	Cytosine-Adenine-Guanine
EOAD .....	Early-onset Alzheimer disease
HD.....	Huntington disease
HDBD .....	Hereditary degenerative brain disorder
<i>HTT</i> .....	Huntingtin gene
NSGC.....	National Society of Genetic Counselors
REVEAL.....	Risk Evaluation and Education for Alzheimer’s Disease
SSB .....	Symptom-seeking behavior

## **Chapter 1: Background**

Hereditary degenerative brain disorders (HDBDs) are a unique class of genetic conditions that result in progressive loss of function within the nervous system. In many of these disorders, such as Alzheimer disease (AD), amyotrophic lateral sclerosis, or frontotemporal dementia, the affected individuals experience physical and/or mental regression as the brain degenerates during adulthood. Given this, the diagnosis of a HDBD can be daunting for both the patient and the genetic counselor assisting in medical care. Huntington disease (HD) is a HDBD and an ideal model genetic disease for this study as the guidelines for the molecular genetics predictive test in Huntington's disease (IHA & WFN, 1994) provide a widely accepted model for presymptomatic testing for other adult-onset disorders (Goldman et al., 2011; Guimarães, Sequeiros, Skirton, & Paneque, 2013).

### **1.1 Huntington Disease**

Huntington disease was first known as Huntington's disease; that is, it was named after George Huntington who identified the symptoms of HD as well as documented the disease as being heritable (Shoulson & Young, 2011). Currently, HD affects less than or equal to 1 in 10,000 people (Ha & Fung, 2012; Shoulson & Young, 2011). Though HD usually affects adults, symptoms can appear as early as infancy depending on the number of trinucleotide Cytosine-Adenine-Guanine (CAG) repeats (Ha & Fung, 2012; Koutsis, Karadima, Kladi, & Panas, 2013).

The natural history and progression of HD has been associated with the trinucleotide repeat number of the Huntingtin gene (*HTT*) (Ha & Fung, 2012; Semaka, Balneaves, & Hayden, 2012; Shoulson & Young, 2011). A higher repeat number can often mean an earlier onset of symptoms (Ha & Fung, 2012; Semaka et al., 2012; Shoulson & Young, 2011). Even so, no accurate estimate for age of onset can currently be made by counting the repeat number (Ha & Fung, 2012; Semaka et al., 2012). It is suspected that other factors, whether genetic or environmental, may contribute to the age of onset for HD (Semaka et al., 2012; Shoulson & Young, 2011).

Ha and Fung (2012) have reported that signs of HD can be present during the presymptomatic, or prodromal, stage of this disease years before a clinical diagnosis is confirmed. MRI scans have shown in some studies that presymptomatic individuals have brain atrophy before the diagnosis and this progresses throughout the course of HD. Tests measuring skills in visuomotor performance, emotion recognition tasks, and working memory can separate presymptomatic HD patients who are closer to their expected age of onset from control groups. Studies have shown dysfunction in self-paced timing tasks, oculomotor function, tongue protrusion forces, and finger tapping tasks among prodromal individuals.

The age of onset of HD can be quite variable among individuals and is frequently family specific, with the mean age of onset about at 40 years (Ha & Fung, 2012; Heemskerk & Roos, 2012; Myers, Madden, Teague, & Falek, 1982; Shoulson & Young, 2011). Onset of clinical symptoms is different for each individual with some having primarily chorea with little cognitive dysfunction as measured by cognitive testing and other individuals having cognitive dysfunction with little motor involvement, yet

symptoms progressively become worse for all (R. Abramson & E. Frank, personal communication, 04/06/2013). Chorea may start in the distal limbs and in the facial muscles (Ha & Fung, 2012). Over time, chorea may decrease, and increased rigidity and dystonia become more pronounced (Ha & Fung, 2012). Throughout the course of the disease, features such as bradykinesia, ideomotor apraxia, motor impersistence, dysarthria, and dysphasia may be present (Ha & Fung, 2012; Heemskerk & Roos, 2012). Progressive loss of executive functioning, attention, visuospatial and construction skills can occur from dementia (Ha & Fung, 2012). Behavioral changes such as irritability, aggression, psychosis, and sometimes suicidal ideation may be present at any time during the disease (Ha & Fung, 2012).

HD patients often need extensive help with activities of daily living and medical aid at the end-stages of the disease. The leading cause of death for persons with HD is aspiration pneumonia, secondary to dysphagia (Heemskerk & Roos, 2012). Loss of motor control and balance can contribute to frequent falls, some of which have led to early demise (Ha & Fung, 2012). Death tends to occur between the first and second decade after the onset of HD (Heemskerk & Roos, 2012; Keenan, Simpson, Miedzybrodzka, Alexander, & Semper, 2013; Semaka et al., 2012).

A variety of non-curative treatments have been developed for symptoms of HD (Nance, 2012). These include medications for symptoms such as chorea, psychosis, and aggressive behavior; management strategies such as speech, augmentative communication, physical, and occupational therapies (Ferm, Sahlin, Sundin, & Hartelius, 2010); counseling for psychological and family concerns; and lifestyle changes such as exercises and environmental modifications (Ha & Fung, 2012; Nance, 2012). Though

there is no cure for HD, Nance (2012) insists that when a patient receives a diagnosis for HD, healthcare providers should emphasize, “There is never ‘nothing we can do’ for a person with HD (p. 359).”

A clinical diagnosis can be made when chorea is found in an adult patient along with changes in his or her behavior, cognitive capabilities, and/or mood (Nance, 2012). A family history of HD can strengthen the diagnosis, but a genetic test can provide undeniable evidence for this disease (Ha & Fung, 2012; Nance, 2012). Genetic testing is especially helpful in situations involving non-paternity, *de novo* mutations, or masked symptoms (Nance, 2012). For example, changes in behavior may be a normal part of puberty even if the adolescent carries a HD mutation (Koutsis et al., 2013; Nance, 2012). However, these behavioral changes may instead be masked symptoms of HD for some prodromal individuals during young adulthood (Nance, 2012). Genetic testing may help distinguish between the two possibilities, though it is recommended that testing for juvenile HD only be pursued if the symptomatic adolescent has a positive family history for HD (Koutsis et al., 2013).

HD is an autosomal dominant disorder that arises from having a CAG trinucleotide expansion in the *HTT* gene on chromosome 4 (Ha & Fung, 2012; Heemskerk & Roos, 2012; Koutsis et al., 2013; MacDonald et al., 1993; Shoulson & Young, 2011). The resulting huntingtin protein has too many glutamines, changing protein folding and disrupting several cellular processes (Bugg, Isas, Fischer, Patterson, & Langen, 2012; Ha & Fung, 2012; MacDonald et al., 1993). Most individuals in the general population have fewer than 26 CAG repeats in *HTT* and will not display the disease (Semaka et al., 2012; Shoulson & Young, 2011). A premutation for HD is

classified as having between 27 and 35 CAG repeats (Semaka et al., 2012; Shoulson & Young, 2011). Individuals with this premutation are not at risk for developing HD, but children of fathers carrying a CAG repeat size from 27 to 35 repeats are at risk for inheriting an expanded CAG repeat sequence (Semaka et al., 2012). This phenomenon known as anticipation, which is most often paternally derived in HD, can increase the CAG repeat number in the children and, therefore, increase their risk of HD (Semaka et al., 2012; Shoulson & Young, 2011). Individuals who have between 35 to 39 CAG repeats will display reduced penetrance of symptoms, but individuals with 40 or more CAG repeats have complete penetrance of Huntington disease (Ha & Fung, 2012; Roze, Bonnet, Betuing, & Caboche, 2010; Semaka et al., 2012; Shoulson & Young, 2011). Those with greater than 60 to 80 CAG repeats may experience juvenile HD (Koutsis et al., 2013; Semaka et al., 2012; Shoulson & Young, 2011).

## **1.2 Genetic Counselors**

Genetic counselors are healthcare professionals trained to help patients understand the role of inheritance in genetic conditions and guide them through decisions on genetic testing as well as supporting them through the complexities of diagnosis, management, and coping with these medical disorders. Genetic counseling is the service given to patients and their families that pertains to the specific genetic conditions they are experiencing or for which they are at risk. As health professionals with training and experience in the personal and emotional situations affecting the livelihood of their patients, genetic counselors acquire a skill set that includes the ability to promote the resilience and endurance of their patients in the face of new diagnoses and extreme hardship.

This definition for genetic counseling best describes the career of a clinical genetic counselor; that is, a counselor who works in a clinical setting as opposed to in another environment, such as a laboratory or a research institution. Furthermore, clinical genetic counselors often refine their roles when they choose from a variety of medical specialties such as prenatal counseling, cancer genetics, or neurogenetics. Thus, individual genetic counselors are likely to routinely have certain counseling experiences based on which patient populations they see. Clinical genetic counselors must develop the ability to cope with various patient and counselor emotions, based on what disorders they counsel for in clinic and how severe the outcomes may be for their patients.

### **1.3 Preparing for and Engaging with Patients**

Genetic counseling for the at-risk HDBD patient population can be significantly different from counseling other patient populations because the symptoms of dementia or psychiatric disturbances may prevent the patients from understanding and directing their own medical care appropriately (Goldman et al., 2011). As recommended by professional practice guidelines written by Goldman et al. (2011), a family member or legal guardian should be requested to accompany a HDBD patient to genetic counseling sessions. In fact, encouraging the presence of a support person for the presymptomatic testing process is part of the Guidelines for Genetic Testing for Huntington Disease published by The Huntington Disease Society of America (Goldman et al., 2011; IHA & WFN, 1994). These guidelines, written shortly after the identification of *HTT* as the gene responsible for HD, are often used by genetic counselors when testing for other adult-onset genetic disorders to help minimize the risk for severe psychological distress (Butler et al., 2011; Goldman et al., 2011; IHA & WFN, 1994; Rahman et al., 2012).

In addition to bringing a support person, the guidelines by IHA and WFN (1994) describe a list of other recommendations. Many of these recommendations follow well-established genetic counseling practices such as respecting autonomy and providing sufficient information for individuals to make informed decisions. Other recommendations are more specific to HD testing, such as the need for counselors to be in a multidisciplinary team, the necessary number of sessions, and the minimal time interval required between them.

The authoring committee of these guidelines emphasized that "...the different sections of these recommendations are inextricable parts of a whole." These guidelines, written with input from HD family organizations, are designed to safeguard patients at risk for HD during presymptomatic testing. However, many genetic counselors may find strict adherence to this set of instructions too burdensome for their presymptomatic HD sessions, let alone for patients at risk for other HDBDs. This thesis will expound on the different techniques that genetic counselors use with their patients at risk for HD as well as other HDBDs.

#### **1.4 Compassion Fatigue**

Compassion fatigue is the state of being unable to act on behalf of a client to the best of one's abilities because of a drain on the medical professional's empathy (Benoit, Veach, & LeRoy, 2007). Compassion fatigue can prevent genetic counselors from best serving their patients because of the counselors' symptoms, which can include avoidance behaviors and emotional disturbances (Benoit et al., 2007). The study conducted by Udipi, Veach, Kao, and LeRoy (2008) found that nearly one in four genetic counselors were at high risk for compassion fatigue.

Genetic counselors who practice in predictive testing for HDBDs may be at greater risk for compassion fatigue than genetic counselors who do not see this patient population. This hypothesis comes from the idea that HDBDs are permanent, progressive, and incurable disorders with tragic consequences, often with high inheritance risk and symptoms that alter the very state of an affected individual's mind. The devastating nature of these adult-onset neurodegenerative disorders may predispose genetic counselors who work with HDBD patients to be susceptible to compassion fatigue due to the pressures and responsibilities of their job to provide empathy along with life-changing information (Benoit et al., 2007). Udipi et al. (2008) found that compassion fatigue is more likely to affect counselors who repeatedly expose themselves to distressed patients. Thus, this thesis will attempt to answer the question: do genetic counselors perceive their HDBD patients to be distressed and in need of extra empathy?

### **1.5 Perceptions of Genetic Counselors**

The stress of testing for a HDBD and receiving these life-changing results can threaten patients with increased anxiety, depression, and suicidal thoughts (Goldman et al., 2011). However, more often than not, those who pursue genetic testing for AD, one of the most recognizable examples of a HDBD, are not necessarily subject to severe psychological impacts (Rahman et al., 2012). In fact, genetic testing can help patients feel more in control of their health (Goldman et al., 2011; Rahman et al., 2012). After all, those who pursue testing for dementia often claim that their chief reasons are to address early symptoms, develop health and financial planning for themselves or their children, eliminate uncertainty, and aid researchers (Goldman et al., 2011; Rahman et al., 2012). In another study known as the Risk Evaluation and Education for Alzheimer's Disease

(REVEAL), there was no substantial increase in anxiety, depression, or test-related distress measured in at-risk adults for AD who received presymptomatic testing for their *APOE*  $\epsilon$ 4 carrier status, an allele that increases the risk for AD but does not guarantee the development or absence of AD (Arribas-Ayllon, 2011; Galvin & Sadowsky, 2012; Goldman et al., 2011; Holtzman, Morris, & Goate, 2011; Rahman et al., 2012). Furthermore, by evaluating at-risk HDBD patients for stress levels before genetic testing and referring for psychological or psychiatric assessments when necessary, severe psychological impact can usually be avoided (Rahman et al., 2012).

But what do genetic counselors believe their patients at risk for HDBDs are experiencing? After all, the uptake for genetic testing of HD and AD is lower than expected. Surveys conducted before genetic testing for HD was available concluded that 5 to 7 out of every 10 individuals at risk for HD would want to undergo predictive testing (Keenan et al., 2013). However, now that presymptomatic testing is available, fewer than 1 out of 10 Americans at risk for HD have pursued genetic testing (Shoulson & Young, 2011). This seems to indicate that a definite diagnosis for a HDBD is too unnerving for some individuals to accept (Tibben, 2007). Is this how genetic counselors see their patients?

The low uptake rate may not be the only reason that genetic counselors could have a different view of individuals at risk for HDBDs as opposed to other patient populations. It has been documented that depression can threaten many individuals with terminal dementia, and sometimes depression itself can mimic the symptoms of dementia (Goldman, 2001; Holtzman et al., 2011). This can cause a misdiagnosis for AD, often by the patients themselves, when in fact they are only depressed (Goldman, 2001; Holtzman

et al., 2011). This pursuit to find proof of a disease within oneself can be known as symptom-seeking behavior (SSB) (Wain, Uhlmann, Heidebrink, & Roberts, 2009). For example, Goldman (2001) describes a patient who was distressed because she believed that her forgetfulness was due to AD, which she believed she had inherited from her father who developed AD before the age of 65 years. Neuropsychological tests revealed that her memory failings were more likely due to depression than AD (Goldman, 2001). Wain et al. (2009) further suggests that stress levels are high for patients with a family history of early-onset Alzheimer disease (EOAD) which, by definition, presents with symptoms before 65 years of age.

Perhaps the high level of stress, the seeming predisposition to depression, and the SSB result from the fear of HDBDs, such as HD and AD, because of the severity of symptoms and the lack of treatment or cure. These factors, combined with a relatively high penetrance and inheritance risk, can create family-wide dynamics with unique situations and emotional issues for genetic counselors to handle. How do these factors affect the perceptions made by genetic counselors of their patients living at risk for HDBDs?

## **1.6 Rationale**

HDBDs, such as HD, are present at rates comparable to other inherited disorders for which genetic counselors frequently provide genetic evaluation and counseling; however there are relatively few genetic counselors who see patients at-risk for HDBDs. For example, approximately 30,000 people in the U.S. have HD, and up to 200,000 are living at risk (HDSA, 2011). In contrast, only 1.5% of the genetic counselors who

participated in the 2012 NSGC Professional Status Survey reported neurogenetics as their primary specialty (NSGC, 2012).

The need for more of these professionals to counsel this patient population is apparent. However, the literature available about how these genetic counselors work with their patients and what lessons they have learned is, to some extent, lacking. This thesis project may be able to provide guidance to genetic counselors new to this patient population and provide a framework so that they are at lower risk for compassion fatigue.

### **1.7 Purpose**

This study is intended to benefit both counselors and their patients by providing counselors an opportunity to share their knowledge and understanding with the medical professionals who work with patients at risk for HDBDs. These patients and their families can benefit over the long-term, as the results of this study may help provide a framework to counselors entering this field by recognizing verified methods of counseling.

## Chapter 2: Manuscript

Working with Patients at risk for Hereditary Degenerative Brain Disorders<sup>1</sup>

### 2.1 Abstract

Hereditary degenerative brain disorders (HDBDs) are a unique class of genetic conditions that result in progressive loss of function within the nervous system, many of which present during adulthood. Given this, the diagnosis of a HDBD can be daunting for both the patient and the genetic counselor assisting in medical care. The purpose of this study was to find themes among genetic counselors who see this patient population and help provide a framework to counselors entering this field by recognizing verified methods of HDBD counseling. Sixteen genetic counselors who routinely see patients at risk for HDBDs were interviewed concerning how they prepared for and engaged with their patients, how they handled the complex emotions frequently associated with presymptomatic HDBD counseling, and how they perceived the stress levels and coping mechanisms of their patients. The results of this study showed that genetic counselors who see patients at risk for HDBDs utilize agendas and/or protocols in preparing for and directing sessions. Additionally, these genetic counselors perceive their HDBD patients as capable of coping with their own presymptomatic testing results. There was not enough evidence from this study to determine if genetic counselors who see patients at risk for HDBDs are at any greater risk for compassion fatigue than other genetic counselors. Overall, the genetic counselors who see patients at risk for HDBDs are more

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similar to than different from genetic counselors who do not see this patient population. Given that a significant number of people are living at risk for HDBDs, experiences in case observation and counseling alongside expert HDBD counselors could benefit genetic counselors and the patients they serve.

*Keywords:* Hereditary degenerative brain disorders, Huntington disease, Alzheimer disease, adult-onset neurodegenerative disorders, presymptomatic genetic testing, genetic counseling

## **2.2 Introduction**

Hereditary degenerative brain disorders (HDBDs) are a unique class of genetic conditions that result in progressive loss of function within the nervous system. In many of these disorders, such as Alzheimer disease (AD), amyotrophic lateral sclerosis, or frontotemporal dementia, the affected individuals experience physical and/or mental regression as the brain degenerates during adulthood. Given this, the diagnosis of a HDBD can be daunting for both the patient and the genetic counselor assisting in medical care. Huntington disease (HD) is a HDBD and an ideal model genetic disease for this study as the guidelines for the molecular genetics predictive test in Huntington's disease (IHA & WFN, 1994) provide a widely accepted model for presymptomatic testing for other adult-onset disorders (Goldman et al., 2011; Guimarães et al., 2013)

HD is an autosomal dominant disorder defined by motor deterioration and dementia accompanied by behavioral changes (Ha & Fung, 2012; Shoulson & Young, 2011). Chorea may start in the distal limbs and in the facial muscles (Ha & Fung, 2012). Over time, chorea may decrease, and increased rigidity and dystonia become more pronounced (Ha & Fung, 2012). Throughout the course of the disease, features such as

bradykinesia, ideomotor apraxia, motor impersistence, dysarthria, and dysphasia may be present (Ha & Fung, 2012; Heemskerk & Roos, 2012). Progressive loss of executive functioning, attention, visuospatial and construction skills can occur from dementia (Ha & Fung, 2012). Behavioral changes such as irritability, aggression, psychosis, and sometimes suicidal ideation may be present at any time during the disease (Ha & Fung, 2012). There is no cure or treatment to stop the progression of HD, and death tends to occur between the first and second decade after the onset of HD (Heemskerk & Roos, 2012; Keenan et al., 2013; Nance, 2012; Semaka et al., 2012).

Though a clinical diagnosis for HD is sufficient, genetic testing is available (Ha & Fung, 2012; Nance, 2012). HD results from having a CAG trinucleotide expansion in the *HTT* gene on chromosome 4 (Ha & Fung, 2012; Heemskerk & Roos, 2012; Koutsis et al., 2013; MacDonald et al., 1993; Shoulson & Young, 2011). Most individuals in the general population have fewer than 26 CAG repeats in *HTT* and will not display the disease (Semaka et al., 2012; Shoulson & Young, 2011). Individuals who have between 35 to 39 CAG repeats will display reduced penetrance of symptoms, but individuals with 40 or more CAG repeats have complete penetrance of Huntington disease (Ha & Fung, 2012; Roze et al., 2010; Semaka et al., 2012; Shoulson & Young, 2011). Those with greater than 60 to 80 CAG repeats may experience juvenile HD (Koutsis et al., 2013; Semaka et al., 2012). The age of onset of HD can be quite variable among individuals and is frequently family specific, with the mean age of onset about 40 years (Ha & Fung, 2012; Heemskerk & Roos, 2012; Myers et al., 1982).

Genetic counselors are healthcare professionals trained to help patients understand the role of inheritance in genetic conditions and guide them through decisions

on genetic testing as well as supporting them through the complexities of diagnosis, management, and coping with these medical disorders. As health professionals with training and experience in the personal and emotional situations affecting the livelihood of their patients, genetic counselors acquire a skill set that includes the ability to promote the resilience and endurance of their patients in the face of new diagnoses and extreme hardship. Genetic counselors who practice in predictive testing for HDBDs may be at greater risk for compassion fatigue than genetic counselors who do not see this patient population.

Compassion fatigue is the state of being unable to act on behalf of a client to the best of one's abilities because of a drain on the medical professional's empathy (Benoit et al., 2007). Compassion fatigue can prevent genetic counselors from best serving their patients because of the counselors' symptoms, which can include avoidance behaviors and emotional disturbances (Benoit et al., 2007). The hypothesis that genetic counselors who see at-risk HDBD patients are at a higher risk for compassion fatigue comes from the fact that HDBDs are permanent, progressive, and incurable disorders with tragic consequences, often with high inheritance risk and symptoms that alter the very state of an affected individual's mind. The stress of testing for a HDBD and receiving these life-changing results can threaten patients with increased anxiety, depression, and suicidal thoughts (Goldman et al., 2011). Symptom-seeking behavior (SSB), the anticipation and self-diagnosis of disease onset, can sometimes present itself in at-risk individuals (Wain et al., 2009). The uptake for genetic testing of HDBDs is relatively low; in fact, fewer than 1 out of 10 Americans at risk for HD pursue genetic testing (Shoulson & Young, 2011). This seems to indicate that a definite diagnosis for a HDBD is too unnerving for

some individuals to accept (Tibben, 2007). Furthermore, the symptoms of dementia or psychiatric disturbances may prevent the patients from understanding and directing their medical care appropriately (Goldman et al., 2011). Altogether, the devastating nature of these adult-onset neurodegenerative disorders may predispose genetic counselors who work with HDBD patients (and thus repeatedly expose themselves to distressed patients) to be susceptible to compassion fatigue due to the pressures and responsibilities of their job to provide empathy along with life-changing information (Benoit et al., 2007; Udipi et al., 2008).

There are relatively few genetic counselors who see patients at risk for HDBDs. In fact, only 1.5% of the genetic counselors who participated in the 2012 NSGC Professional Status Survey reported neurogenetics as their primary specialty (NSGC, 2012). The need for more of these professionals to counsel this patient population is apparent. However, the literature available about how these genetic counselors work with their patients and what lessons they have learned is, to some extent, lacking. In this study, genetic counselors who routinely work with this patient population were interviewed and asked to answer how they prepared for and engaged with their patients, how they handled the complex emotions frequently associated with presymptomatic HDBD counseling, and how they perceived the stress levels and coping mechanisms of their patients. This interview study may be able to provide guidance to genetic counselors who are new to presymptomatic HDBD counseling and provide them with a framework so that they are at lower risk for compassion fatigue.

Our hypotheses for this study were as follows: Genetic counselors who serve patients at risk for hereditary degenerative brain disorders (1) utilize agendas/protocols in

preparing and directing sessions, (2) are at a significant risk for compassion fatigue, and (3) perceive their patients as being unable to cope with the information presented during genetic counseling sessions. Significant differences may be seen among genetic counselors who see at-risk HDBD patients as a single provider and those who see at-risk HDBD patients as part of a HDBD interdisciplinary clinical team.

### **2.3 Materials and Methods**

The institutional review board at the University of South Carolina approved this study in September of 2012. Genetic counselors who were members of the National Society of Genetic Counselors (NSGC) Neurogenetics Special Interest Group electronic mailing list were invited by email to participate in this interview study. Other genetic counselors, including personal contacts or counselors participating in the Dominantly Inherited Alzheimer Network study were also sent the same invitation to participate. Later in the study, two e-blast email invitations were sent to the entire NSGC electronic mailing list inviting all counselors with experience counseling patients at risk for HDBDs. Counselors who responded to the primary author arranged dates and times to conduct interviews.

The interviews took place over the phone, Skype, or Adobe Connect, the latter two being different internet-based conferencing media. The medium used was based on the preference of the participant. Audio content of the interviews was recorded for analysis, and no video content was recorded of participants using either Skype or Adobe Connect. Participants were informed of these recording conditions ahead of the interview.

The identity of each genetic counselor has been kept confidential. Numerical codes were assigned to each of the 16 participating genetic counselors. All transcripts

were void of names. Only the primary author had access to identifying information in the original transcripts. The audio and coded lists of the interviewees were stored in separate documents secured in a password-protected laptop. No identifying information was shared during the qualitative analysis in order to protect the privacy of the participants.

The interview questions began with quantitative, closed-ended questions and proceeded to qualitative, open-ended questions (See Appendix). All interviewees were asked the same questions in the same order. Any question that needed to be clarified was done so in a consistent fashion by the primary author. Questions that required additional examples to add context were done so in a consistent fashion by the primary author. Each interviewee was allowed to elaborate on any question as she chose. Additional questions were asked based on the flow of conversation, but may not have been added to the study. Interviews ranged from 22 to 53 minutes in length.

The qualitative analysis software Atlas.ti 7.0 was used to store and organize data as well as transcribe audio to text. Interviews were transcribed in accordance with simple transcription procedure (Thorsten Dresing, 2012) near verbatim, excluding words and phrases not contributing to answering study questions. Due to a recording error, one of audio recordings of the participants was never recovered. The participant was informed and agreed to check and verify her statements transcribed by the primary author during and shortly after the interview as well as add any missing information. This qualitative study used methods based on grounded theory, a widely used methodology for genetic counseling research (Grubs & Piantanida, 2010).

## 2.4 Results

A total of sixteen ( $N = 16$ ) genetic counselors from the United States and Canada participated in this study. Participants were asked to identify their practice specialties, which included prenatal, pediatric, cancer, general genetics, research, and neurogenetics (see Table 1). Three participants chose two primary specialties, and in each of these cases, neurogenetics was one of the specialties. In total, half of the participants ( $n = 8$ ) considered neurogenetics to be, at least in part, their primary specialty. Most participants ( $n = 11$ ) had more than 10 years of experience in genetic counseling. For half of the participants ( $n = 8$ ), HDBD cases made up less than 25% of their total caseload when compared to other specialties, such as pediatric or prenatal cases. However, all participants ( $n = 16$ ) currently see HDBD patients. Most participants ( $n = 12$ ) considered themselves part of a HDBD interdisciplinary clinical team. Only one participant evaluates patients' stress levels with a structured questionnaire, and a minority of participants ( $n = 5$ ) use standard measures for anxiety and/or depression with patients at risk for HDBDs.

Table 1: Demographics of Practice Information of Study Participants	
<i>Study Participants (N = 16)</i>	<i>n (%)</i>
<b>Practice Specialty*</b>	
Prenatal	2 (13%)
Pediatric	3 (19%)
Cancer	1 (6%)
General genetics	3 (19%)
Research	2 (13%)
Neurogenetics	8 (50%)
<b>Length of Practice</b>	
0-5 years	2 (13%)
6-10 years	3 (19%)
11-15 years	3 (19%)
16-20 years	1 (6%)
21 or more years	7 (44%)
<b>Percentage of caseload is for HDBD patients</b>	
≤25%	8 (50%)
26-50%	2 (13%)
51-75%	2 (13%)
≥76%	4 (25%)
<b>Part of a HDBD interdisciplinary clinical team</b>	
Yes	12 (75%)
No	4 (25%)
<b>Currently see HDBD patients</b>	
Yes	16 (100%)
No	0 (0%)
<b>Evaluate HDBD patients' stress levels with structured questionnaire</b>	
Yes	1 (6%)
No	15 (94%)
<b>Use standard measures for anxiety and/or depression with HDBD patients</b>	
Yes	5 (31%)
No	11 (69%)

*\*Percentages do not add up to 100% due to three participants choosing two primary practice specialties*

**2.4.1 What do you to prepare for a session with an individual at risk for a HDBD?** As might be expected, the way the participants prepare for a session with an individual at risk for a HDBD is similar to the preparation of any genetic counselor before a session. Most participants ( $n = 12$ ) described obtaining and reviewing the

medical records and family history. Five participants mentioned they may request additional records on family members to confirm the genetic mutation that their patient may have inherited. Other preparations may come through attending disease-specific meetings, reviewing the medical literature, or readying resources for the patient. Half of the participants claimed to call the patient before the first clinical session to discuss topics such as the patient's expectations of presymptomatic testing, their reasons for wanting testing, their experience with the genetic disorder in question, and/or details about their support network. A few genetic counselors used the pre-visit phone call to anticipate possible needs and concerns a patient could bring to the session and/or anticipate what referrals might be necessary.

Two of the participants, both with more than 10 years genetic counseling experience, made it clear that they needed minimal preparation for presymptomatic HDBD sessions. Participant #8 claimed that because of her many years of experience, she was, "...more involved in getting a good sense for the patient than in doing any type of literature reviews unless something in particular comes up." Participant #16 explained, "...I've been doing it enough times, long enough now, that I don't really sit and prepare what I'm going to say or those kinds of things so much anymore." However, most counselors claimed that HDBD sessions required some degree of preparation.

#### **2.4.2 How do you direct a session with an individual at risk for a HDBD?**

When discussing how a genetic counselor may direct a session, two major categories seemed to emerge: what parts make up a session and how to use an agenda. The parts of the session will appear familiar to all genetic counselors: contracting, gathering information from the patient, educating, and counseling. Many counselors made special

note of gathering information about why their patient wanted presymptomatic testing, what their expectations were of the testing protocol, and what their prior experience with the HDBD in their family was.

All but two participants stated their use of some form of check list, protocol, or agenda to structure their HDBD sessions. How they used this agenda seemed to reflect on their idea of who was directing the session. A majority of participants spoke of modifying agendas to complement the individuality of their patients. For example, Participant #11 stated the following:

Yeah, I mean, I would say that we have an agenda of points that we want to make sure that we hit. But how much time we spend on each of those points and what information is covered in what detail is basically patient led. So it's kind of, it's both.

Several participants wanted the patient's questions and concerns to direct the session. As Participant #13 stated, "I have kind of a list of things that have to be covered, but I start out with letting them direct the session and then I fill in what doesn't come up." Other participants described using detailed agendas that could be modified for patients, but their adherence to these outlines was more consistent as this quote from Participant #3 portrays, "And I do try to let it be somewhat patient controlled or directed, but I also have my own agenda as well so I do just try to find the compromises in that." Overall, the desire of genetic counselors to use the agenda as a means to meet the needs of the patients was consistent among participants.

**2.4.3 How do you handle the patient's needs versus the patient's family's needs?** Participants seemed to address this question from three different approaches that

became the main themes: (1) what counseling strategies are used to resolve family conflicts, (2) how involved was the family member, and (3) whose needs were priority? When this interview question was asked, the primary author always paired it with the following example. The participant was told of a situation in which their patient at risk for HD had a parent who did not want to know his or her own HD carrier status. The participants were then asked what they would do under such circumstances.

The first theme consisted of several counseling strategies. These included asking about the relationship and situation between the conflicting family members, giving extra time for the family to make a decision, and/or making referrals for professional counseling. Over half of the participants ( $n = 9$ ) mentioned that they would encourage their patient to discuss these issues with their parent.

The second theme concerned how involved the parent was in the counseling process. Half of the participants spoke of bringing the family member into the session or arranging for a separate counseling session. Some would talk to both parties independently as individual patients, and others might try to facilitate a contract between the family members. For example, Participant #13 said, “We try to have the two family members make a contract with one another about how they're going to handle information flow within the family.” A few counselors spoke of trying to get the parent tested first instead of their original patient. According to Participant #4, “If possible, have parent get tested instead so children won't have to be tested if test negative.”

Concerning the other aspect of the second theme, if the patient did not involve the parent in a counseling session, then a few of the participants voiced that they would make

sure the patient was aware of how going through with predictive testing could negatively affect that relationship. As Participant #7 stated:

The patient in front of me is always the person that I'm most directly concerned with and would then make sure that that patient was aware of what actions may affect other family members and how: so how testing that person might provide information to the parent without them anticipating those consequences and how that could maybe be dealt with by talking to the father or getting an idea of what communication lines are open between them already.

In reference to the third theme, many of the participants stressed the importance of aiming for compromise. However, if no compromise could be reached as in the quotation above, all but three participants spoke of how the patient's needs take priority. That is, most of these genetic counselors would pursue presymptomatic testing for their patient even if the parent disagreed. Participant #8 further elaborated this concept, "I treat the patient's needs as...they are the determining factor, but we do discuss the family needs and I try to work it so that we respect everyone's needs. But ultimately, the patient's needs and decisions are paramount." Participant #16 hesitated with this line of thinking. When the primary author asked her if she would still allow for testing when no compromise could be found, she responded, "We have. Could I say that I always will? I don't know that I can, but I can tell you that we have done it." Out of the three participants who did not state that they would continue to pursue testing under these circumstances, one of them, who worked in a research setting, stated that she would release the patient from the study, and the other two did not specify how they would resolve the impasse. Participant

#16 had previously explained, "...it's hard. It's not easy. Whatever, you know, there's no right answer for those."

#### **2.4.4 Questions concerning HDBD testing results.**

##### ***2.4.4.1 How do you give results for genetic testing, both positive and negative?<sup>2</sup>***

Several recurring ideas seemed to define how participants would address this question, including when and where to give results, the utility of planning ahead, and how involved phone communication was. All participants routinely give results to patients at a later genetic counseling session. Minor differences existed on how to give the results during the session, such as what patient-friendly language to use or who else would be present at the results disclosure. Whatever the differences were, planning the results session seemed to be important to the participants. Participant #13 noted that planning ahead was important to the patient also:

I recommend that they have a plan in place about what the expectations of people in their families and their support system know about when they're getting their results so that, you know, if there's 10 people on their list who are imminently waiting to hear their results, they have a plan about how that's going to happen because I think people for the most part are not very good about predicting how they feel afterwards.

Perhaps the most surprising comments arose on the use of phone communication. Most counselors were quick to state that they would not call patients with positive or negative results, but that patients were expected to come in for their results during a later session. A few participants ( $n = 4$ ) made concessions that they would give results for rare

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<sup>2</sup> Codes between the first and second part of question four were sometimes used interchangeably given their similarities and proximity to each other during the interview.

exceptions, such as for patients who were separated by significant geographic distance across states and countries. One participant noted that she disapproved of colleagues that would give negative results over the phone.

Two participants favored re-visiting the idea of calling out results to these presymptomatic patients. Participant #5 said it this way:

In our hospital, we do require a post-test face-to-face visit, which I find very frustrating, I'll be honest. We do do that. We say, you know, "It's better to give results in-person." I actually personally disagree...I just think it's too hard to give results in-person because people don't have time to process the information and ask questions versus, even in prenatal, in the prenatal world...the genetic counselor will call out abnormal results over the phone and then schedule a visit for the next day, which I think would make a lot more sense if patients had time to hear the news and let it sink in. Even one day is not long a time to process the information, but enough time to get over the initial shock and then say, "Okay, now what?" You know, it's hard. I've had not a lot, but I've had a handful of patients just walk out of the room. It's just like, "I can't hear anything you have to say. I will call you later." and they leave. And it's SO hard, because I want to be like, "No, just sit here for a minute." But it just doesn't, they just shut down.

***2.4.4.2 How does giving results for a HDBD differ from giving other genetic test results, if at all?*** Many participants seemed to believe that giving results for HDBDs could differ from giving results for other genetic tests due to the very nature of the

disease and because of the particular counseling responsibilities required; however some participants also remarked on the similarities between all genetic test result disclosures. Many participants stated or implied that, by nature, HDBDs are more intense than other genetic diseases. Participant #16 described this theme with her answer, “Yeah, I don't know. I would just say it is more intense. That's what I can tell you.” During the interviews, participants recognized that HDBDs can affect the mind, but there is no cure and there are no effective treatment options. Other participants talked of how giving these results were different in that these at-risk individuals were presymptomatic and this information impacted their whole future. Participants described giving these results as “life-altering” (P#9), “...like you're giving them a death sentence” (P#2), and that the example of HD has, “...a very, very severe phenotype” (P#15). Participant #13 stated that “...there's a lot more anxiety.” Participant #10 spoke of the need for extra support for those receiving these results:

I don't think in any other testing situation do we really require them to bring in somebody else in with them because we know the significant psychological magnitude of the reaction and what it could potentially lead to. That's why we screen very carefully looking for the depression and the suicidal ideation.

Participant #5 summarized a lot of the participants' feelings in her quote:

In this situation, I feel like there's a big difference between telling somebody that they're going to, you know, be at risk for some condition where there is medical treatment, or that it's going to affect their health but not their mind. When you affect your mind, it's scary. I mean, I think

there's a whole level more of the uncertainty and "How do I know?" and "I'm going to worry every time I forget." You know, I mean, we all have those moments: you walk into a room and you're like, "Alright, what was I supposed to do here? I forgot... I was doing something." And people who are at risk for these types of conditions, they find those things so much more stressful. It's that other element of your mind. It's different. I also feel like for a lot of these hereditary degenerative brain diseases there's not a lot of treatment options. There's not much we can do today for a lot of them and that's hard.

Continuing on the theme of the nature of HDBDs and test disclosure, some participants took special notice of how HDBDs are inextricably tied to the family. Many HDBDs are autosomal dominant, and thus numerous patients have seen the disorder in their family before. Because of this, patients seem to know what to expect, as Participant #16 describes, "It's a, you know, a future that usually people have witnessed, you know, with their parents or have 'Googled' and like, you know, are like imagining the worst." Participant #9 gave the example of HD and said, "...because most of these families have seen it already, it just changes their whole outlook, you know?" Leading up to the results disclosure, there can be a "buildup" to the dreaded family disease. Positive results have an impact on the rest of the family as well. When Participant #13 compared this to giving results in pediatric genetic counseling in which the patient is displaying unique symptoms, she said this:

It's a very different dynamic than a family where they know what it is and the whole buildup about the testing is finding out if they have what they're

dreading or not and the ripple effect of that result through their life and their family's life and their whole future. So there's a lot more anxiety.

There's a lot more buildup to getting the results. It's just a really different dynamic than in individuals who they're already dealing with something.

The other theme that emerged was that the counseling responsibilities differ when giving these results. Six participants noted here that these results cannot be given over the phone, and one participant noted that there was more telephone contact with the patient overall. Participants noted the extra disclosure session, the increased follow-up, and the coordination of bringing a support person, all of which can require more planning. One participant noted that the sessions are less about symptoms and more about psychosocial issues, which changes the level of counseling indicated.

That being said, not all participants agreed that giving HDBD results was much different from giving other genetic test results. Many participants recognized that HDBDs are hardly the only genetic disorders to be difficult to accept. Participant #16 exclaimed that giving any bad news, “sucks.” Participant #11 admitted that, although giving HDBD results can be somewhat different, “...the raw emotion that comes out of getting, you know, an upsetting test result is, you know, is pretty universal.” Participant #1 was reluctant to compare giving HDBDs results to all other possible genetic tests, “But there's many genetic conditions that are, you know, that I'm sure people look at as equally devastating in many ways, so it really depends on the condition that we're talking about.” Though most participants would agree that some degree of difference exists, many did not want to single HDBDs out as having the absolute worst possible test disclosures.

**2.4.5 Do you prepare yourself emotionally for a session with an individual at risk for a HDBD?** Out of the 16 participants, 12 claimed that they prepare emotionally for a session on some level. How they prepared seemed to consist either of practical or emotional solutions. Practical solutions were often implemented before a session. Some examples included reviewing the patient's chart and family history, creating a list of talking points, using the pre-visit phone call to anticipate difficult patients, setting aside a "quiet time" (P#13) before the session, and preparing for possible scenarios. Two participants scheduled HDBD cases so that they were not consecutive, that is, they would not have to go directly from one HDBD session to another. Another participant would not look at the results until just before the session so that she would not dwell on the results. Three participants mentioned talking with other staff members to receive emotional support.

Participants often had emotional solutions to help themselves prepare for a session. When asked if they prepared emotionally, several participants had memorable quotations about how to do this. Participant #8 said,

Oh, yes! They can be quite heavy. You know, I really just kind of gear myself up for it. I don't do anything in particular except to realize that what I'm walking into could be a heavy situation and so, "Let's go with it!"

Participant #9 answered, "Yes, I do. I have to every time. And, you know, I've given hundreds of results. But, you know, I have to have my counselor face on." Participant #11 said, "...the geneticist and I kind of mentally prepare and kind of have to steel ourselves to be ready for that session." Participant #16 answered, "Oh, absolutely. Yeah,

put on a little bit more armor. You know, you get more emotionally prepared to sort of be able to handle whatever comes back at you.” Participant #2 answered thoroughly:

So yeah, you do emotionally prepare yourself, mentally also. I mean, you have to think, think about what the patient's going through. Wow, you know, they're coming to potentially find out if they're going to have this life-changing condition and you are going to be the one to tell them that. That's pretty profound. But also there has to be a little bit of separation. You can't wrap all of that up and carry it yourself because that's a huge burden, too. So I don't know that any counselor has that completely perfected. We always want to care deeply but we have to somewhat separate ourselves emotionally. So definitely an ongoing challenge.

Four of the participants agreed that they may prepare emotionally, but it would be conditional. Participant #5 prepared more for positive result disclosures:

I get more nervous when it's positive results, you know, just since I know it's going to be difficult. But for the first visit I don't do a lot...I just try to be the most prepared. I mean, I think that's how I feel more comfortable is just knowing that I am the most prepared I can be.

Other answers included only preparing emotionally at the results disclosure or preparing as much for HBDB sessions as with any other genetic counseling sessions. Still another participant stated that the emotional preparation depended on extent of her relationship with the patient. Though the emotional preparation for these four participants was conditional, it seems that all the participants agreed that they prepared emotionally for HBDB sessions sometimes.

**2.4.6 How do you handle emotions during the session, both that of yours and your patient's?**<sup>3</sup> The most common theme for how participants handled emotions during their sessions was to be supportive of their patients ( $n = 15$ ). Other common themes included letting patients have their emotions ( $n = 14$ ), counselors holding back their own emotions ( $n = 11$ ), counselors showing their own emotions ( $n = 7$ ), and how to help patients long-term ( $n = 7$ ).

Participants most often described how they would support their patients when they became emotional. Many of their techniques look similar to those of other counseling sessions. Participants may remind patients of their reasons for testing, reframe questions to suit the patient, talk through fears, encourage coping mechanisms, clarify misconceptions, and allow time for emotions, questions, and reactions. Some participants described themselves as compassionate, would try to understand where the patient was coming from, would develop rapport, and would try to address the cause of the emotions.

Within this theme of showing support, seven participants expressed how they can give their patients long-term support. Participants may offer resources such as support groups, make referrals for additional counseling, or plan follow-up sessions or phone calls. Participant #11 even mentioned rescheduling appointments for patients, “who are just like shut down and don't want to talk.” Though supportive techniques may differ as well as their timing, Participant #2 describes this as an evolving process for all counselors, “handling patient emotions, I think that's a skill that every counselor has to work on and continually develop.”

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<sup>3</sup> Quotations for this question could be derived from the answers to question 5, especially since the primary author often inferred that these two questions were linked. e.g. “Along the same lines, how do you handle emotions during the session, both that of yours and your patient's?”

The second most common theme was, as Participant #5 said, “let people have their emotions.” Participant #5 goes further to explain:

If they're mad, let them be mad. It's appropriate to be mad...I think I just try to stay calm...I just validate. You know, most of the feelings that people have are completely appropriate and so just let them feel that way. And let them know, you know, of course they should be upset and mad and life's not fair and all those things that people say... [I] try to keep my emotions out of it. I've been affected by things people say and I just tell them, you know, "Wow, that's really awful", and, you know, just be real.

Several participants made it clear that emotions are appropriate and that they try to normalize feelings. Other participants spoke of being attentive to the needs of the patients and to give patients extra space or time if needed.

The third most common theme, which 11 participants voiced, was that counselors should hold back their own personal emotions. The underlying reason seemed to be that the patient should not have to be burdened by the counselor's emotions, as Participant #1 describes:

There are times when I tear up if it's a very sad discussion. And I really don't go beyond that. I do everything in my power because I don't want my patients to have to take care of me. That is not okay with me.

Participant #6 echoed this concept and the need to stay reserved for the patient:

I have not had a situation yet where I myself have broken down with emotions because I feel that is not clearly my job. My job is to be strong

and just accept whatever response they have and be there to be supportive and, you know, sort of a safe zone for them to react.

Several counselors agreed with the need to stay calm for the patient and to not let personal emotions get in the way. Sometimes this required mental preparation such as when Participant #16 reminds herself to keep her emotions, “in perspective that, you know, this isn't about you. It's about them at the moment while you're in the room with the patient.” Sometimes it requires taking a break from the session or to relinquish the session to another health professional who is in a better position to remain neutral. However it is done, many participants find ways to hold back their own emotions from the patient.

However, not every participant believed that all emotions should be held back. Specifically, seven of the participants admitted to tearing up or crying with patients in emotional situations. Participant #9 describes such a transition between holding back personal emotions to crying with patients:

I'm pretty seasoned professional, and you know, I keep my emotions in check but I think it's probably clear to people when they get to me. I was trained never to cry in front of a patient. I think that that's wrong. I think it should be done rarely, but there's a couple of times, particularly some of these families I've now seen and tested three generations of family members. And so I know their story. I know what happened. And so and one more test positive and you might cry with them because it is sad.

Two participants further elaborated that counselors should allow themselves to be genuine to themselves and to their patients. Participant #8 gave another example of not only handling patient emotions, but also her own:

You know, often times the patients cry during my sessions. I think it's inherent in their concern for having inherited this devastating neurologic disorder that's progressive and they've seen many of their family members get it so often they do cry. I hand them a tissue. You know, I am empathic and understand that this is scary. And I try to let them feel their emotions where, you know, instead of deflecting it with humor or something I want them to know that I recognize as well that this is scary. This is important. This could be life-changing and it is important so it's okay if their emotions are involved. And for myself I really try not to show my emotions too much, but you know, if I tear up, I tear up. I don't hide that. I think it shows that I'm human.

When handling emotions during a session, a counselor can help patients not only by being supportive and letting people experience emotions, but also by sharing emotions with patients as Participant #2 summarized:

I will say I have definitely cried with patients. And I don't know that that's bad. The patients that I have gotten emotional with I feel like expressed appreciation and expressed to me, "Wow! It's so nice to feel like you're connecting with us and you understand how hard this is for us." ... I just sort of felt like, what would I do if this was my friend, you know? What would I do? And sometimes that's giving them a hug. Sometimes that's cry

with them. Sometimes that's just sorting it all out with them, putting things in perspective. You know, there's different coping mechanisms that we all have and that we encourage our patients to use. But we're all human and so I think connecting is not always a bad thing. Being emotional is not a bad thing. But sometimes it's just giving them the chance to be emotional. I find that a lot of patients at risk for Huntington's, they just kind of go about their day everyday and shove it off to the side. It's always in the back of their mind, but they're never going to let it surface. Sometimes when they're sitting with you in that office they do, for that hour, let it come to the surface. And that's an emotional thing. And just letting them cry or letting them be angry, letting them be so mad that this is happening in their family is cathartic for them and definitely, I think, healing for the counselors, too.

**2.4.7 What do you do if you cannot keep from “taking the session home” with you? Who do you talk to?<sup>4</sup>** Participants took this opportunity to talk about how difficult situations can arise from their work as well as the activities and thoughtful solutions they use to alleviate the stress of “taking the session home.” Most participants seemed to acknowledge that “taking the session home” does happen at least occasionally. Some are affected more than others. Participant #1 said:

Oh, I do take sessions home with me. I don't leave stuff at the door. I don't think that's possible. I think about things all the time. I dream about things.

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<sup>4</sup> The first part of this question was always asked, but the second part of “who do you talk to?” was only asked if the participant did not mention whether or not they spoke to anyone to deal with the issue of “taking the session home”. One participant answered part of this question in the prior question, “How do you handle emotions...?”

I'm not one of these people who turns it off when I walk out the door. I don't think many of us are.

“Taking the session home” does happen, and there are innumerable ways genetic counselors deal with it. Almost all of the participants ( $n = 15$ ) said that they would talk to their colleagues, and most of them said this during the interview without any prompting (see previous footnote). Half of the participants would talk with their friends or family members, and one participant consulted with a personal counselor if needed. Participants described activities to take their minds off of the session. These could include things such as listening to music, relaxing at home, going to the movies, or getting time alone. The most common activity cited was getting exercise ( $n = 7$ ).

Participants thought of several thoughtful solutions to combat “taking the session home.” Preventative solutions included protecting oneself from emotions before and during the session. After the session, participants may use techniques learned from graduate school to take control of feelings, use the time to think of ways to help the patient, or process the feelings in another way. Participant #5 spoke of a mentor who reminded her, “‘It's hard, but you have to realize it's not... you didn't cause these results to be positive,’ which obviously you know cognitively, but emotionally it's hard to feel that way.” Many participants recognized the potential of letting themselves “take the session home,” but many also recognized as Participant #8 did, “You discuss it and you've got to move on. It's hard.”

#### **2.4.8 How do you believe your patients handle being at risk for a HDBD?<sup>5</sup>**

Participants believed that their patients could live with uncertainty well or poorly, often based on the individuality of the patient, but many also concluded that these individuals are a self-selected population and that we have limited knowledge of patients who are living at risk yet do not come in for genetic counseling. Altogether, 10 participants praised their patients for living with uncertainty well. Participant #9 stated, “I think they handle it pretty well. I give them a lot of credit for strength and courage and bravery.” However, just as many participants ( $n = 10$ ) had examples of how patients could handle their at-risk status poorly, such as with destructive behaviors or by ignoring the possibility of a positive result. Participant #16 summarized it as:

Oh, I think it's really, really hard. I mean everybody that I talk to about it, no one's ever said to me, ‘Oh, it's okay. It's what it is, you know, it's fine. It's easy.’ Everyone tells you it's hard.

Even more of the majority of participants ( $n = 11$ ) spoke of or implied that their patients were subject to a spectrum of emotions and that the way patients handle risk is as individualized as they are. As Participant #1 stated, “I think it's as personalized as they are. I mean, I can see 100 people and there'll be 100 stories.”

An interesting conclusion several counselors made about their HDBD patients was that they are a self-selected subpopulation. For example, Participant #4 presented the idea that “Patients who come in for presymptomatic testing are a self-selected group; they tend to cope well with result (positive or negative) because it is the uncertainty that they have the hardest time coping with.” The patients that present for genetic counseling

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<sup>5</sup> Some answers to this question were shared with the next question regarding coping mechanisms. In such cases, quotes were analyzed interchangeably between the two questions.

believe that their uncertainty about their at-risk status is worse than knowing their genetic answer, positive or negative. In fact, we know relatively little about those who are at risk and are not receiving genetic counseling. Participant #7 put it this way:

I mean, we're only seeing probably the tip of the iceberg of the people who are at risk and considering the testing. And I think a lot of people are getting testing in the community and getting very little counseling with it. And I don't think we have an idea of the consequences because we're not seeing those people.

Participant #13 made a valid point when she said, "I think the ones who aren't coping well or don't really want to know never show up. So I think on the whole, genetic counselors may not really know how people are dealing with their at-risk status." Indeed, the perception of genetic counselors may not adequately reflect how all individuals in this patient population feel about their at-risk status, especially those who refrain from genetic testing.

**2.4.9 What are the different methods you have seen your patients use to cope during genetic counseling and genetic testing?** Perhaps the better question would have been, "What coping methods have you NOT seen your patients use?" Dozens of coping methods with different layers of complexity were mentioned, and a number of participants spoke of how any and every coping method could be seen. As Participant #3 put it, "It's sort of a whole spectrum of different coping mechanisms. Pretty much, if the coping mechanism exists and it's documented in our textbooks, I've probably seen it."

Among the more common coping mechanisms were talking to friends and family, using humor, using spirituality and/or prayer, being angry, seeking all knowledge that

could be found on their HDBD, denial, getting professional counseling, crying, and finding support groups. Though many of the coping mechanisms could be helpful, several could be destructive such as substance abuse or domestic violence.

**2.4.10 Please describe your perception of these patients' stress levels before the counseling session has begun and after the counseling session. Please describe your perception of your patients' stress levels post-session.** Questions 10 and 11 both asked the participants to describe their perceptions of the patients' stress levels before the first session, after the first session, and during follow-up. Some overlap of answers existed with question #9 as well.<sup>6</sup> When the answers to these combined questions were analyzed, three main themes emerged: when are patients stressed, why are patients stressed, and how can stress be reduced?

When are patients stressed? Every participant perceived that patients come into their first genetic counseling session with more stress or are more upset coming into a session than when they leave. Participant #14 provides an example of this sentiment:

I think my perception is that most people come in at a higher stress level at the beginning or prior to the session than after. We tend to try to gauge our session around trying to help alleviate some of that stress to the extent you can... certainly gear it towards what the patient needs to make them feel comfortable, and I'd rather not send a patient out the door knowing that their stress level just went up after a session ... but overall, I'd like to think that stress level is at a more manageable level by the end of the session than prior to the beginning of a counseling session.

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<sup>6</sup>By mistake, Participant #5 was not asked to describe her perception of her patients' stress levels post-session.

From there, the perception of stress level varies. Some spoke of high levels of stress leading up to the session, during the first session, during the neurological exam, or at the blood draw. Others spoke of stress levels peaking while waiting for the results or during the results session. Many participants ( $n = 7$ ) believed that stress levels decreased during follow-up, and some of these participants believed that stress levels continued to decrease over time.

Why are patients stressed? Some participants voiced that it depends on the patients themselves. Some patients are strong, others calm, others angry, others nervous, and some stay stressed throughout all their appointments. There could be a whole slew of variables within the patients' lives that contribute to how they deal with their genetic information, as Participant #1 elaborated:

I am not a believer in one-size-fits-all. I am absolutely not. I think that every family I see, I start with a clean slate because people are always surprising me. People are incredibly unique in their needs, their style, their background, their history, the burden of what they're facing. One size does not fit all, absolutely.

Participants had more explanations for why patients may be stressed. Patients may be stressed because of the complications surrounding the testing protocol. They may not know what to expect. They may previously have had little contact with medical care. They may be worried about finances. One participant even said they may be stressed because they are worried that their stress may disqualify themselves from testing.

The information the results provide can also help explain why patients are stressed. Several participants noted that negative test results generally come with an

immediate drop in stress level. In direct contrast, several participants noted that positive results are inherently stressful. There may be a state of shock after receiving positive results. Positive results could incite patients to envision themselves declining like an affected relative. Positive results can lead to SSB. Positive results can also be so stressful that they call for professional counseling.

How can stress be reduced? Many participants paired their perceptions about stress levels with thoughts about how to help alleviate stress. Participants may help reduce stress levels by eliminating the fear of the unknown by answering questions, by correcting misconceptions, and by familiarizing the patient with the testing protocol. Participants also spoke of ways to reduce stress by helping patients be able to plan their testing protocol or plan what their life could look like after testing. Four participants noted that some patients can have their stress reduced just by knowing that they are not in this alone; that there is a support system that they can plug into and rely upon. Overall, there seemed to be a consensus that genetic counselors, in fact, help reduce stress by fulfilling supportive roles in the medical management of genetic testing.

**2.4.11 Do you follow-up with your patients?** Perhaps the clearest theme from this question is that there is no obvious theme. Most participants engaged in some sort of follow-up with their patients, and this was most often done by phone calls. However, details such as how often to follow-up, whether or not to follow-up with patients with negative results, and when to stop pursuing follow-up were varied across the participants. Even the decision to follow a formal protocol was varied.

If follow-up was not pursued, it seemed to emanate from the participant's belief that the patient did not want to be reminded of their positive test result. When Participant #5 was asked if she followed-up with her patients, she described this example:

Not usually, no. I did for a while, and then it just didn't seem like patients wanted that. So I'd say... "Do you want me to give you a call in a couple weeks and see?" And they'll say, "No, I'll call you if I want to."

Though most participants did pursue follow-up, several of these participants seemed unsure of what level of follow-up was necessary or wanted by the patients.

**2.4.12 How often do patients assign symptoms of a HDBD when their symptoms may also be indicative of another disorder?** Most of the participants, but not all, have recognized SSB during presymptomatic HDBD testing, and several of these participants had ideas of who is at greater risk for SSB. When this interview question was asked, the primary author always paired it with the following example. The participant was told of a situation in which their patient, who has a family history of EOAD, begins to have symptoms of memory loss and thus diagnoses herself with EOAD. However, the memory loss appears to be more indicative of depression. The participants are then asked to describe how often this type of SSB occurs in their practices.

Out of the 16 participants, 13 believed that this type of SSB does occur. Estimates of how often range from "almost always" (P#14) to "rare" (P#8) and everything in between. Two of these 13 participants believed that SSB happens roughly less than half the time, and another three of these participants are unsure of how often SSB occurs. The other eight of these participants believed it happens with a greater degree of frequency. When asked how often she sees SSB for HDBD patients, Participant #2 said:

I would say a lot. I mean, I think that's just human nature and that makes sense. If you have this condition that plagues your family, of course you're going to think every time you forget your keys or, you know, forget where you put your keys that, "Oh, this could be it!" Or the Huntington's patients, every time they might trip and fall, they think, "Was that really just...did I just trip or was that just pure clumsiness and this is related to the Huntington's." So yeah, a lot! I think that's completely normal.

Participants also had answers as to who is susceptible to SSB. Some examples of to whom SSB occurs include those who struggle with alcoholism, those who are asymptomatic, those who are symptomatic, and those with a family history of mental illness, anxiety, stress, and/or dysfunction. Two participants believe that SSB motivates people to come in for testing. Two participants stated that parents can have SSB for their children with other symptoms. Participant #9 stated that patients with positive results are even more susceptible to SSB:

But then after we disclose people, they have their HD when they test positive, they have their HD radar on and so they think everything is Huntington's disease! You know, and it could be the flu and they think it's somehow Huntington's disease.

However patients display it, these 13 participants believe that SSB is present during HDBD sessions.

Three participants disagreed that SSB occurs in this kind of presymptomatic testing scenario. Participant #15 did not recognize any such cases in her practice.

Participants #5 and #10 found that the opposite was true, that patients are in denial about symptoms. Participant #5 described it this way:

I've actually found the opposite to be true. I find that when people come in and I'm thinking, "Oh my goodness, you're symptomatic! Like you're not pre-symptomatic, you're symptomatic." I think that some people will say, "No, no! I'm not! Like I think I'm just a stressful person," or, "You know, I've got a lot going on so I think that my depression is due to, like, all of these other things. I don't think that I'm symptomatic for HD." You know, I think that people try to hold onto the hope and to not like, deny that what they're experiencing is symptoms, sometimes, to protect themselves.

**2.4.13 What has been the most rewarding aspect of your work with patients at risk for HDBDs?** Participants found that helping their patients and connecting with families and communities to be the most rewarding aspects of their work with this patient population among other aspects. For 14 of the participants, the most rewarding aspect of working with patients at risk for HDBDs is the opportunity to help patients and/or their families. Participant #16 explained it this way:

I mean just in general, it's just helping people through, you know, a difficult time... And so to contribute, like, in a way where you're feeling like you're doing the best that you can do to help people get the information that they want and to help guide them through that process is very rewarding.

Participants also stated that they enjoyed being a resource to patients, identifying the family disease, helping people cope with the information, hearing gratitude

and positive feedback from patients, helping with family planning, empowering and supporting patients, and providing opportunities in research. Three participants specifically mentioned that giving good news, which is negative results, to patients is very rewarding. Participant #8 summarized this and the main theme when she answered:

Gosh, being able to tell someone that they're not going to have this horrible disease that has devastated their family is wonderful. It's an absolutely fantastic feeling and I get lots of hugs for it (laughter). But I think either way it's rewarding because I get to help these people at a crossroads in their lives and I find that rewarding.

The second most common theme was the reward of connecting with patients, their families, and their communities ( $n = 5$ ). Participant #13 captured these sentiments with her answer:

I think the most rewarding thing has been really being able to know whole families and being help them in a sense... I really believe that people cope better when they have good information and good support. And so I think being in a position to provide that because of my dual role, both in the program as well as through the support group and through a larger HD community, has been really rewarding.

Participant #1 also expounded on this point:

And so I think the most rewarding moments I have are when a family connects well with me. They are grateful for my presence and I know that in some way I have given them, whether it's support or validation or just

enough of my, you know, ability to help them cope with the situation at hand, to tolerate what's going on, those moments that I realize that perhaps I've been able to do that for them, that's rewarding.

Other rewarding aspects mentioned by participants included the opportunity to use counseling skills extensively and working as a team with other medical professionals. In all of these ways, the participants found something to cherish in their work with HDBD patients.

**2.4.14 What has been the most challenging aspect of your work with patients at risk for HDBDs?** In addition to several other themes about the difficulties associated with HDBD counseling, the most common theme, which half of the participants voiced, was that giving bad news was the most challenging part of their job. Sometimes, giving bad news was difficult because there is little or no treatment to offer. Participant #9 voiced this sentiment when she said:

The most challenging aspect is that I don't have anything in my bag of tricks that really is going to make them feel better. You know, I don't have any effective treatment or cure, and so I still feel somewhat powerless, you know, in that aspect of the relationship.

Other times, it was difficult because the news was for presymptomatic individuals. Participant #6 was struck by both the lack of treatment and the presymptomatic nature of the results:

The most challenging, I think, is always... is having to tell people that they are a carrier, that they do have the genetic change. It's difficult information to tell people. It's hard to be the bearer of that news, you know, bad news.

And one of the reasons it is so challenging is, at this point, you know, let alone the fact that they now know they do have the gene and will most likely develop the disease as long as they live old enough, we don't have any preventative options. There's nothing that I can tell them that they can do to reduce their risk in any way.

The reason giving bad news was challenging was sometimes less about the information itself and more about how to give the information. Participant #5 again noted that results should be given over the phone because positive results cause patients to “shut down” and further counseling ceases to benefit patients that day. Participant #14 specifically noted it is difficult to give bad news and the appropriate counseling to patients who have had testing ordered through physicians who do not follow presymptomatic testing protocol:

And then the negative impacts that that can have on patients has been very challenging because ultimately then, what I have seen many individuals that do come back to us for counseling and it's very difficult. You can't undo the damage that was done.

Some other challenging aspects of working with patients at risk for HDBDs included the emotional burden on a counselor ( $n = 3$ ), the nature of the patient population ( $n = 3$ ), having conflicts with patients ( $n = 2$ ), losing contact with the patients ( $n = 2$ ), and other non-counseling aspects of the job ( $n = 4$ ). The emotional burden to the counselor could include watching a patient decline in health and independence, witnessing juvenile onset of the disease, or trying not to take the harder cases “home with you” (P#8). The HDBD patient population itself could create challenges. Participant #11 commented that

some of these patients could be “unpredictable,” “intimidating,” and have, “a lot of emotional issues.” Participant #16 thought some patients were, “really paranoid,” and Participant #15 thought some patients were not, “necessarily as grateful.” Challenges such as defending the need for a testing protocol to patients or being subjected to verbal abuse were mentioned. Two participants noted how patients would be lost to follow-up. Four participants commented that non-counseling aspects, such as administrative work, organizing a clinic with patients who are losing executive functioning, or having few resources available for patients are among the most challenging aspects of working with this patient population. These were some of the more common challenging aspects of working with at-risk HDBD patients with giving bad news being the most prevalent.

**2.4.15 What advice would you give to a genetic counselor beginning to work in this field?** Though it had been assumed that advice among the participants would be similar, every participant had an original idea. Nonetheless, themes emerged. Participants had advice about counseling ( $n = 9$ ), about getting experience ( $n = 6$ ), about preparing for sessions ( $n = 3$ ), and about working with a team ( $n = 3$ ) as well as thoughts about the field of HDBD counseling itself ( $n = 4$ ).

The most common advice was about how to counsel this patient population. Four participants emphasized the importance of listening to patients. Participant #9 said to, “really listen to the stories of the families... you will learn more about HD from these family stories than you will from any book or any symposium or any article.” More than knowledge, listening will enable counselors to engage with your patients, as Participant #1 implores counselors to:

...not think so much about what you're going to say, but think much more about what is being said. Listen; LISTEN to your families because they're going to tell you what they need. Listen; and they're going to tell you their story. So be patient, be mindful, and be present, and listen carefully.

Other counseling advice included treating every patient as an individual, being flexible in counseling methods, never assuming anything about patients, and the importance of finding the balance between being empathic and not “taking the session home.”

The second most common piece of advice regarded getting experience. Counselors beginning to work with this patient population should get prior experience in job shadowing, in clinical exposure, in supervision, in participating with the affected community, and in getting life experience with affected individuals. This theme tied in with another theme: working with a team. Recommendations included getting a mentor, accepting help from other medical professionals, and creating coalitions of medical professionals with whom to share cases.

Some advice regarded preparing for sessions. This included creating a protocol to standardize practice with all patients and reading medical literature to prepare for any issue that could come up during the session. One participant specifically advised discussing possible insurance and discrimination issues surrounding presymptomatic testing with patients during a pre-visit phone call.

Other advice described the field of HDBD counseling itself. Participant #3 had this to say: “Keep in mind it's going to be hard, that it's challenging but it's

promising, that with HDBDs, there's a lot of research going on.” Participant #15 had more sobering words for counselors considering this field:

I think they should think about it carefully and try to attend a couple clinics and sit in and watch before they buy into it and sign themselves up to get involved because I've seen senior genetic counselors who do cancer genetic counseling, excellent genetic counselors, unable to manage. So it's not for everyone, you know. There's a niche for everybody, clearly, but I don't think this area's for everyone.

Participant #7, however, encouraged more counselors to get involved:

Don't be afraid of it... it's really rewarding aspect of genetic counseling and you can be an expert in this area like no one else can. You're really some of the best professions prepared to do this kind of work. So get out there and do it!

The collective advice of all the participants offers invaluable resources and tips to assist counselors that indeed wish to counsel patients at risk for HDBDs.

## **2.5 Discussion**

As had been previously stated, there are relatively few genetic counselors who counsel patients at risk for HDBDs. Out of the over 2,850 NSGC members, approximately 1.5% specialize in neurogenetics (Finucane, 2012; NSGC, 2012). In the end, only 16 genetic counselors replied to our request for interviews, eight of whom identified their primary specialty as neurogenetics, thus approximately 19% of the estimated neurogenetics counselors participated in this study. Though these interviews were rich in content, there were too few participants to explore possible statistical

significance by quantitative analysis. For this reason, comparison of themes to demographics such as practice specialty, length of practice, participation in a HDBD interdisciplinary clinical team, and use of standard measures was excluded from this discussion.

We hypothesized that genetic counselors who serve this population would report using specific agendas in working with patients at risk for HDBDs. As had been expected, the participants utilized agendas and/or protocols in preparing for and directing sessions. There was no apparent difference in many of the ways participants prepared for HDBD sessions when compared to case preparation for any other indication for genetic counseling such as described by Uhlmann (2009). However, one difference may be that half of the participants call their at-risk HDBD patients ahead of time, a potentially time-consuming preparation that may not be seen as often in other practice specialties based on the experience of the primary author. All but two participants stated their use of some form of check list, protocol, or agenda to structure their HDBD sessions, and it seemed as though the agendas were used as a means to meet the needs of the patients.

The specific agendas were unique for each participant rather than being exact copies of the guidelines for the molecular genetics predictive test in Huntington's disease (IHA & WFN, 1994). Guimarães et al. (2013) suggested that counselees receiving presymptomatic testing for late-onset neurodegenerative disorders desire more flexibility in protocol to match their expectations and psychological needs. A majority of participants already claimed to do this by allowing their patients to lead or co-lead the session and help establish the agenda for the session.

Though all participants routinely give results to patients at a result disclosure session, the protocol exceptions for calling out results and/or the amount of follow-up pursued varied widely. Also, there was no consensus on protocol for how to deal with the patients' desire to be tested and the conflicting desires not to know among at-risk family members. A majority of participants ( $n = 13$ ) stated that, in the end, they would pursue presymptomatic testing for their original patient if no compromise could be made. Despite these differences, it was a common theme to utilize agenda and protocols to assist in preparing for and directing sessions with patients at-risk for HDBDs.

There is not enough evidence from this study to determine if genetic counselors who see patients at-risk for HDBDs are at a significant risk for compassion fatigue. However, certain themes indicate that the risk for compassion fatigue is present for these genetic counselors and that they have found ways to reduce the risk, such as by talking to friends and colleagues, engaging in activities outside of work, and mentally and emotionally preparing for and enduring through sessions. The main theme that indicates an increased risk for compassion fatigue is the repeated exposure to distressing clinical events as described by Udipi et al. (2008).

Seven intrapersonal and situational characteristics were identified by Udipi et al. (2008) to be predictors of compassion fatigue, including: burnout, self-criticism and giving up, increased number of distressing events, increased patient load, using religion to cope, having no children, and seeking support to cope. The variable of seeking support was minimized given the fact that participants of this interview study were asked who they talked to when they "took sessions home with them." Out of the remaining six

variables, the increased number of distressing events seemed most relevant to the participants of this interview study.

Participants of this study were affected by the distressing events that were among the most prevalent in the Udipi et al. (2008) study, including giving bad news (i.e. positive genetic test results), feeling helpless to assist patients, feeling invested emotionally in the patients' well-being, and feeling burdened by the knowledge of positive test results. Giving bad news, the most common distressing event from the Udipi et al. (2008) study, was listed as the most challenging aspect of working with patients at risk for HDBDs by half of the participants. Some of the reasons cited for why giving bad news was so difficult included having no treatment to offer, no hope to give, and the feeling of taking away the patients' future with positive results. When participants answered how giving HDBD results differed from giving other genetic test results, similar themes emerged, such as lack of treatment and the intensity of results disclosure, that appear to correlate with the distressing event of feeling helpless to assist patients.

In contrast, connecting with patients, their families, and their communities was the second most common rewarding aspect of working with patients at risk for HDBDs. Feeling invested emotionally in the patients' well-being may become a distressing event for these genetic counselors if they overextend empathic reserves into their patients. However, counselees of late-onset neurodegenerative disorders have expressed the need for empathic engagement from their genetic counselors (Guimarães et al., 2013), and many participants echoed the importance of connecting with the patient. Lastly, feeling burdened by the knowledge of positive test results was not a common theme in and of

itself, though one participant admitted that she would not look at test results until just before the disclosure session so as to avoid dwelling on the knowledge of the test results.

The increased risk for compassion fatigue based on triggers such as delivering bad news and consequently feeling responsible for patient suffering are supported by a previous study by Benoit et al. (2007). However, the increased risk for compassion fatigue described by Udipi et al. (2008) is caused by a combination of the seven predictor variables, not just exposure to distressing events. More so, there is no reason to think that these distressing events are isolated to HDBD counseling. Therefore, this current interview study does not have enough data to verify an actual increased risk for compassion fatigue for genetic counselors who see patients at risk for HDBDs. Though we were not able to measure compassion fatigue using a structured questionnaire, participants were able to recognize the potential for a problem and they have found ways to cope with the stress of counseling.

Do genetic counselors perceive their patients as being unable to cope with the information presented during a genetic counseling session, namely presymptomatic testing results? This study suggests the opposite, that this patient population is capable of coping with their own genetic information. When asked to describe their perception of how patients handled their at-risk status, the most common theme among the participants was that the patients' ability to handle risk is as individualized as they are. An equal number of participants thought that patients handle their risk status well or poorly. Interestingly, several participants noted that the patients that come to clinic are a self-selected population that cope well with knowledge but not well with uncertainty. Neither was there a common theme of the coping methods used by this patient population.

Rather, the examples were a spectrum of different, yet common coping methods used by the patients; such as talking to friends and family, using humor, using spirituality and/or prayer, being angry, seeking all knowledge that could be found on their HDBD, and denial.

Though many participants stated that HDBD test results were intense in nature and that the stress surrounding presymptomatic testing was high, this in and of itself did not imply that patients would be unable to cope with the results. In fact, many participants gave examples of how they could help reduce the stress levels of patients during the sessions and during follow-up. It is unlikely that so many participants would try to reduce their patients' stress levels if they perceived their patients as being unable to cope anyway. Additionally at many testing centers, the patients who are perceived to be able to cope would be allowed to interact with the genetic counselors rather than being referred to therapy and deferred from testing.

Interestingly, all participants believed that patients leave their initial genetic counseling session less stressed than when they came in. This seems to imply that the at-risk HDBD patient population is capable of coping; indeed, patients can cope right before the genetic counselors' eyes. This line of thinking seems to agree with the REVEAL study which demonstrated that even positive results for AD can be "well tolerated" by patients who have received genetic counseling (Arribas-Ayllon, 2011). Another explanation could be that genetic counselors want to believe that they are helping patients. Therefore, perceived reduction of stress could be a desired perception by genetic counselors. Whatever the explanation, there was less of a consensus on the ability to cope

during follow-up, but many participants believed that stress levels decreased over time thus implying the ability to cope eventually.

One could argue that SSB, by definition, is the inability to cope with uncertainty. Thirteen of the participants reported SSB in this patient population, but there is no clear answer on how often this occurs. Therefore, it is unclear whether this patient population is less able to cope because of their SSB.

Though some of the patient population was sometimes labeled as unpredictable, highly emotional, and/or more paranoid, these attributes do not confer the inability to cope with information. Even so, these thoughts were expressed by a minority of participants. Considering all of the interviews, it does not appear that genetic counselors by default assume their at-risk HDBD patients are unable to cope with the information presented during genetic counseling sessions.

The limitations of this study include the total number of interviews and the potential for the omission of themes. As discussed earlier, there were not enough participants for quantitative analysis to hold statistical significance. Larger study groups could help answer some of the questions raised by this study, particularly the effects of practice specialty, length of practice, participation in a HDBD interdisciplinary clinical team, and use of standard measures on counseling methods and interpretations. Given the semi-structured nature of the interviews, genetic counselors may have given implications by omission. Without asking each counselor specifically about each theme discussed, it is unclear how many may identify with or disagree with any given theme.

## 2.6 Conclusions

We hypothesized that genetic counselors who serve patients at risk for hereditary degenerative brain disorders (1) utilize agendas/protocols in preparing and directing sessions, (2) are at a significant risk for compassion fatigue, and (3) perceive their patients as being unable to cope with the information presented during genetic counseling sessions. Our study demonstrated that genetic counselors who see patients at risk for HDBDs utilize agendas and/or protocols in preparing for and directing sessions, though there were differences among participants in how these were used. Contrary to our hypothesis, genetic counselors perceive their HDBD patients as capable of coping with their own presymptomatic testing results. The most common theme of the participants' risk perception was that the patients' ability to handle risk is as individualized as they are. Both the ability to cope and the coping methods used were on a wide, variable spectrum.

Lastly, there was not enough evidence from this study to determine if genetic counselors who see patients at risk for HDBDs are at any greater risk for compassion fatigue than other genetic counselors. However, certain themes such as exposure to distressing events and feeling invested emotionally in the patients' well-being indicate that the risk for compassion fatigue is present for these genetic counselors. Nonetheless, these participants have found ways to reduce the risk, such as by talking to friends and colleagues, engaging in activities outside of work, and both mentally and emotionally preparing for and enduring through sessions. Additionally, the participants listed positive feelings experienced by counseling these patients that balance the negative feelings surrounding the job.

Overall, the genetic counselors who see patients at risk for HDBDs are more similar to than different from genetic counselors who do not see this patient population. Genetic counselors who may wish to serve this patient population will have most of the necessary skills to succeed in providing the best care for presymptomatic testing and follow-up. Several participants in this study gave advice to incoming genetic counselors to seek experience with the HDBD community and medical expertise before counseling this patient population. The addition of HDBD case observation and HDBD patient counseling to the curriculum of any board-certified Genetic Counseling program has the potential to greatly assist new counselors in gaining this type of experience. Given that a significant number of people are living at risk for adult-onset neurodegenerative disorders, these learning experiences could benefit genetic counselors and the patients they serve.

### **Chapter 3: Conclusions**

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## **Appendix**

### **Interview Questions**

The following list of questions was used as an outline for the interview questions. Where appropriate, the interviewees were asked to expand upon their answers.

#### **QUANTITATIVE QUESTIONS**

1. What is your practice specialty?
  - a. Prenatal
  - b. Pediatric
  - c. Cancer
  - d. General genetics
  - e. Assisted reproduction
  - f. Research
  - g. Laboratory
  - h. Neurogenetics
  - i. Other
2. How long have you been a practicing genetic counselor?
  - a. 0-5 years
  - b. 6-10 years
  - c. 11-15 years
  - d. 15-20 years
  - e. 20 or more year

3. What percentage of your total caseload is spent with patients at-risk for a hereditary degenerative brain disorders (HDBD) versus other patients (e.g. pediatric, cancer, etc.)?
  - a. Less than 25% with HDBD patients
  - b. 26%-50% with HDBD patients
  - c. 51%-75% with HDBD patients
  - d. More than 75% with HDBD patients
4. Are you part of a hereditary degenerative brain disorders interdisciplinary clinical team?
  - a. Yes
  - b. No
5. Do you routinely see patients at-risk for a hereditary degenerative brain disorder or with a hereditary brain disorder for continuing evaluation and/or treatment?
  - a. Yes
  - b. No
6. Do you evaluate patients' stress levels with a structured questionnaire?
  - a. Yes
  - b. No
7. Do you or your clinic use standard measures for anxiety and/or depression with patients at-risk for a hereditary degenerative brain disorder?
  - a. Yes
  - b. No

## QUALITATIVE QUESTIONS

1. What do you do to prepare for a session with an individual at-risk for a HDBD?
2. How do you direct a session with an individual at-risk for a HDBD?
3. How do you handle the patient's needs versus the patient's family's needs?
4. How do you give results for genetic testing, positive and negative? How does giving results for a HDBD differ from giving other genetic tests results, if at all?
5. Do you prepare yourself emotionally for a session with an individual at-risk for a HDBD?
6. How do you handle emotions during the session, both that of yours and your patient's?
7. What do you do if you cannot keep from "taking the session home" with you?  
Who do you talk to?
8. How do you believe your patients handle being at-risk for a HDBD?
9. What are the different methods you have seen your patients use to cope during genetic counseling and genetic testing?
10. Please describe your perception of these patients' stress levels before the counseling session has begun and after the counseling session.
11. Do you follow-up with your patients? If so, how? Please describe your perception of your patients' stress levels post-session.
12. How often do patients assign symptoms of HDBD when their symptoms may also be indicative of another disorder?
13. What has been the most rewarding aspect of your work with patients at-risk for HDBD?

14. What has been the most challenging aspect of your work with patients at-risk for HDBD?
15. What advice would you give to a genetic counselor beginning to work in this field?