EVALUATION OF GENETIC COUNSELING SERVICES OFFERED IN A HUNTINGTON DISEASE CLINIC

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ABSTRACT

Studies of outcomes associated with the provision of genetic counseling services began to emerge in the 1990's. The goals of this outcome-based research are often focused on examining the effectiveness of the services offered and determining appropriate means for improvement of services. The outcomes measured vary depending on the specific goals of the study and beliefs concerning the nature of counseling. Research utilizing outcome measurements choose the most appropriate measurement based on the desired goal of the research. This study employed a client satisfaction measurement, specifically a Satisfaction with Genetic Counseling Scale (SGCS), a self-reporting measure provided to clients in the form of a survey who were seen at the Huntington Disease Specialty Clinic in Pittsburgh, Pennsylvania. The hypothesis for this study was that patients in the HD Clinic are generally satisfied with the genetic counseling services they have received. Descriptive statistics were used to measure the general satisfaction and test the hypothesis. Responses to open-ended questions were categorized into themes to gather more insight into patient perceptions of genetic counseling services. Thirty-three individuals completed the survey, and analysis of the results revealed that patients are generally satisfied with the genetic counseling. Results were broken down into subscales measuring general satisfaction of inherent characteristics of a genetic counselor and attributes specific to the counseling. Patients were overall generally satisfied with each subscale. This research is of

particular significance to the field of public health because assessing the provision of genetic counseling services ensures that clients' needs are being met as well identifies potential avenues for improvement of services. Research of this nature is becoming increasingly more important because genetic counselors, with their expertise in molecular diagnosis, are expected to play an essential role in providing personalized and preventative health care in the growing field of genome-guided medicine. This research may inform outcome research in other settings, providing additional insight to the genetic counseling field.

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PREFACE

The completion of this master's thesis would not have been possible without the combined efforts of multiple individuals. First, my sincerest gratitude extends to Robin Grubs, MS, PhD, LCGC, director of the genetic counseling program. She served multiple roles in this process including serving as one my committee members, thesis advisor, and mentor throughout my time at the University of Pittsburgh. Her dedication to my professional growth and understanding within the scope of genetic counseling, and extending further, will never be forgotten. Dr. Grubs' dedication to the education of students in the genetic counseling program is a commendable attribute that I am so fortunate to have been a part of. Her continuous mentorship and accessibility over the past two years has aided in the achievement of the completion of graduate school, my most current professional goal.

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1.0 INTRODUCTION

Huntington disease (HD) is an inherited, progressive neurodegenerative condition characterized by motor disturbances, behavioral changes, and cognitive decline. The disease results from an expanded CAG repeat in the *huntingtin* (HTT) gene located on chromosome 4. In the general population, individuals carry on average 17-20 CAG repeats in this gene. Individuals with >40 repeats will develop HD with nearly 100% certainty. The individuals who inherit the disease progress from completely asymptomatic to presenting with symptoms that worsen in severity over time. Early signs of the disease generally present by the age of 40, with death typically occurring 15-20 years after the age of diagnosis. The prognosis is poor, and there still remains no cure for the disease; however, treatment is available pharmacologically and nonpharmacologically for management of some of the symptoms (Dayalu & Albin, 2015).

Families can face a number of challenges after receiving a diagnosis of HD. At-risk individuals are often burdened with the complexity of living in the shadow of risk for developing HD, some families are placed in the position of being caregivers for those affected with HD, which can create psychological stress, and family dynamics can transform significantly leading to unexpected consequences such as divorce. Health care and support provided to families with HD has the potential to alleviate some of these stressors, which is the intent of the Huntington Disease Clinic of UPMC in Pittsburgh, Pennsylvania. The clinic is a dedicated resource for families with HD in Western Pennsylvania and the geographical surrounding areas.

Genetic counseling has been an integral service provided in the UPMC HD Clinic and in the broader HD community for greater than twenty years. Genetic counselors have collaborated with the Western Pennsylvania Chapter of the Huntington Disease Society of America (HDSA) to provide services in the Clinic. Predictive testing, one of the services offered in the clinic, is available to at-risk individuals. Recognizing the potential psychological impact of learning one's risk status, a predictive testing protocol was developed by the HDSA. Genetic counseling is an important component of this protocol (Meyers et al., 2003).

Although HD predictive testing has been available for greater than twenty years, minimal research has been conducted to evaluate the effectiveness of genetic counseling services within a HD clinic setting. This type of outcome research is important to ensure that patients and their families affected with HD are receiving the genetic counseling services necessary in order to enhance their coping and minimize the stressors and impacts that a diagnosis may have on a family.

1.1 HYPOTHESIS AND SPECIFIC AIMS

1.1.1 Hypothesis

Patients in the Huntington Disease Clinic of UPMC in Pittsburgh, Pennsylvania are generally satisfied with the genetic counseling services that they have received.

1.1.2 Specific Aims

Aim 1: To evaluate the effectiveness of genetic counseling services by distributing a client satisfaction survey to patients or their family members/caregivers receiving these services from the Huntington Disease Clinic of UPMC in Pittsburgh, Pennsylvania.

Aim 2: To statistically analyze the results from the client satisfaction survey to evaluate the effectiveness of the genetic counseling services offered through the Huntington Disease Clinic of UPMC in Pittsburgh, Pennsylvania.

1.2 BACKGROUND AND SIGNIFICANCE

1.2.1 Huntington Disease

Huntington disease (HD) is an inherited, progressive neurodegenerative condition characterized by motor disturbances, behavioral changes, and cognitive decline. It is estimated to affect approximately 30,000 individuals in the United States and Canada. Approximately 150,000 individuals are at risk for inheriting the disease. HD most frequently affects individuals of European decent, which the prevalence is considered to be about 10 to 15 per 100,000 (Dayalu & Albin, 2015).

A clinical triad is often used to characterize the features associated with Huntington disease. Progressive motor disorder is one part of the triad. Perhaps the most recognizable feature of HD is the chorea movements, which remain only a small part of the motor dysfunction but can

be the most debilitating. Chorea often begins as fidgety movements most visible in the distal extremities. As the disease progresses, the chorea often becomes more pronounced involving larger, proximal muscles. Individuals may experience other motor difficulties including saccadic eye movements, dystonia, bradykinesia, and ataxia of speech, limbs or gait. Progressive motor function can lead to even more serious complications as the disease progresses. Individuals often have significant weight loss and aspiration as a result of dysphagia. Additionally, the progressive motor failure and chorea can be accident-provoking leading to serious injury (Dayalu & Albin, 2015).

The second part of the clinical triad is the cognitive disorder. The cognitive decline culminates in dementia, which serves as a significant disturbance to an individual's daily living. It typically presents early in an individual's life, affecting the capability to complete normal activities. Multitasking, focus, short-term memory, and learning new skills are also difficulties individuals may experience and may affect social as well as occupational functioning (Dayalu & Albin, 2015).

The third part of the clinical triad in HD is the psychiatric changes that may include behavioral problems, anxiety disorders, delusional behavior, or hallucinations. As with all of the features associated with Huntington disease, the degree and symptoms of the psychiatric disturbances varies between individuals. Some individuals experience significant depression, obsessive-compulsive behaviors, irritability, and outbursts not consistent with their typical personality type. Individuals receiving a diagnosis of HD should be monitored for suicide, as the rate is increased for individuals receiving a new diagnosis (Dayalu & Albin, 2015).

1.2.1.1 Molecular Genetics

Huntington disease is as an autosomal dominant disorder. The majority of cases are caused by the inheritance of the expanded allele from an affected parent. However, approximately 10% of cases result from a new expansion in an individual. The disease results from an expanded CAG repeat in the *huntingtin* (HTT) gene located on chromosome 4. The length of the CAG repeat is associated with the age of onset and presence of symptoms. In the general population, individuals carry on average 17-20 CAG repeats in this gene. Typically, individuals with 6-26 CAG repeats are considered in the normal range and will not develop the disease. Those with 27-35 repeats are considered in the intermediate range. Generally these individuals do not develop HD; however, there have been a few reported cases in the literature. An allele with a repeat size in the intermediate range is considered unstable and has the chance to expand in future generations to a disease causing repeat number. Studies have shown that this happens more frequently when transmitted through a male parent. Moreover, a repeat number in the range of 36-39 is considered the 'gray zone'. Individuals with a repeat number in this range may or may not develop the disease as a result of incomplete penetrance. However, CAG repeats >40 indicate that the individual will develop HD in their lifetime with nearly 100% certainty.

Highly expanded repeat numbers are associated with an early age of onset of HD symptoms. Individuals with a highly expanded repeat number, >60, generally develop symptoms at an early age compared to others with a lower number of repeats. The reason for this is unknown at this time. Individuals with <45 CAG repeats have a more variable age of onset. Smaller repeat lengths such as <45 are more common than highly expanded repeat numbers. Furthermore, age of onset may be influenced by other genetic and environmental factors, to which the extent is currently unknown (Dayalu & Albin, 2015).

1.2.1.2 Treatment and Management

At present, there is no cure for HD. Pharmacologic treatments and nonpharmacologic supportive measures are used for symptom management.

Pharmacologic treatments are mainly directed towards psychiatric symptoms due to the adequate response to treatment. Additionally, there are pharmacologic treatments for chorea movements. These treatments seem to reduce chorea but do not completely eliminate it from the individual's symptomatology.

Nonpharmacologic management may include comprehensive care by a wide range of specialists including but not limited to: primary care physicians, neurologists, psychiatrists, geneticists, physical and occupational therapists, speech pathologists, nutritionists, social workers, and genetic counselors. Nondrug interventions are just as important as pharmacologic treatments in the care of an individual with HD. Physical and occupational therapy is vital for management of gait disturbances and modifications to the home that are important for the safety of the individual as the disease progresses. Speech therapy may be necessary for an individual for management of dysarthria and dysphagia. A nutritionist often manages dietary concerns, particularly regarding extreme weight loss. Social workers and counselors participate in managing the complex needs and psychosocial concerns an individual with HD or their family has before and after receiving a diagnosis (Dayalu & Albin, 2015).

The management for an individual with HD is complex and requires the attention of multiple health care professionals. Care and treatment should be tailored to the specific individual. Given that there is no cure for the disease and pharmacotherapy is solely focused on managing symptoms, research is currently centered on identifying a disease-modifying therapy (Dayalu & Albin, 2015).

1.2.2 The Role of Genetic Counseling in Huntington Disease

Predictive genetic testing is currently being offered for a variety of different conditions including single gene disorders, such as Huntington disease, and other conditions such as hereditary cancer predispositions. HD paved the way in predictive genetic testing, having been the first predictive testing program offered to individuals who are at risk. Predictive genetic testing has the potential to engender emotional, cognitive, and behavioral sequelae in the individual undergoing testing. It is recommended that HD predictive testing be offered within a predictive testing protocol due to the complexities that are potentially involved in the process (Broadstock et al., 2000). This predictive testing protocol includes elaborate pre- and post- test evaluations, which includes genetic counseling, to ensure that the individual desiring the predictive testing is making an informed choice, is competent enough to receive their results, and to reduce the risk of adverse psychological consequences as a result of the testing (i.e. severe depression, suicidality) (Grubs et al., 2014).

The Huntington Disease Society of America (HDSA) published a revised protocol in 2003 for predictive genetic testing in HD. The HDSA recommends these guidelines but states that they may need adapted by a health care professional according to the clinic of care or the individual seeking the testing. The predictive testing protocol recommended by the HDSA can be seen below in Table 1:

Table 1. HDSA Recommended Predictive Testing Protocol

Predictive Testing Protocol
1. Initial telephone contact
2. Neurological evaluation
3. Genetic counseling
4. Documentation of informed consent
5. Psychological assessment
6. Review of the potential impact of the test
7. Disclosure of results in person
8. Follow-up after testing

Genetic counselors have a number of responsibilities in the predictive genetic testing process. The HDSA describes the roles of genetic counseling in the predictive testing process as follows:

> "This includes a review of the family history, confirmation of the family diagnosis, and explanation of the applicant's risk status. Genetic principles that relate to HD as the gene test are reviewed, including the risks, benefits, and limitations of the test (such as the possibility of results in the intermediate range, or the inability to predict the age of onset based on repeat number alone). Alternatives to genetic testing are discussed. Often the genetic counselor will explore the applicant's experience with HD and perceptions of the disease, and discuss the potential burden of the test results on the individual and the family. Pre-test tasks are discussed and scheduled or performed if necessary (such as identifying a local counselor, confirming the family diagnosis by testing an affected person, evaluating neurological or psychiatric symptoms, or obtaining insurance)."

The above quotation from the HDSA delineates the importance of genetic counseling in the HD predictive genetic testing protocol. Outcome research on the effectiveness of genetic counseling in a HD setting is important to ensure that the counseling is fulfilling its role in the predictive genetic testing process. Due to the major involvement of genetic counseling in the predictive testing protocol, an outcome study of client satisfaction involving the elucidation of patients' perceptions in the setting of the Huntington Disease Clinic of UPMC would identify areas of proficiencies and areas in need of improvement. In addition, the results of this study have the potential to contribute information not only for genetic counseling in its involvement with HD predictive genetic testing but also with predictive testing in other specialties of genetic counseling. Discovering inherent characteristics of a genetic counselor and components of a

genetic counseling session in which patients are most satisfied with is applicable knowledge for any specialty of genetic counseling. In an era where consumer-oriented practice is becoming more commonplace, outcome measure research utilizing client satisfaction in genetic counseling will become increasingly more important.

1.2.3 Huntington Disease Clinic of UPMC: Pittsburgh, Pennsylvania

The Huntington Disease Clinic of UPMC in Pittsburgh, Pennsylvania offers predictive and confirmatory/diagnostic genetic testing and counseling to individuals affected with HD, or those at-risk, and their families. Additionally, the clinic provides comprehensive care for individuals with HD in Pittsburgh and surrounding regions. When appropriate, patients are offered the opportunity to participate in HD-related research. Leaders in the field of HD, Robert Y. Moore, MD, Professor of Neurology, and Elizabeth Gettig, MS, LCGC founded the clinic. Presently, the clinic is still providing services under the direction of Valerie R. Suski, DO, Assistant Professor of Neurology.

The clinic offers support and resources for individuals and their families affected by HD. Contributing to the care of these individuals in this center are physicians, genetic counselors, social workers, and research coordinators. Comprehensive care is provided to ensure that individuals receive the tailored care necessary in the management of their disease. Among the services offered are the following: neurological evaluation, genetic counseling, pharmacologic treatment for movement disorders and psychiatric symptoms, cognitive and psychiatric evaluation, physical, occupational, and speech therapy, caregiver services, social services, and nutritional counseling. Additionally, this center is a leader in Huntington disease research and offers individuals the opportunity to participate in clinical trials and research studies.

1.3 OUTCOME MEASURES IN GENETIC COUNSELING

Outcome measures in genetic counseling are designed with the goal of evaluating counseling effectiveness in meeting client needs as well as discovering areas for improvement in the delivery of counseling services. The evaluation of genetic counseling largely depends on the belief about the nature and the goals of the counseling (Pilnick and Dingwall, 2001). Pilnick and Dingwall (2001) noted that the outcome measures used most frequently in studies measuring counseling effectiveness are centered on reproductive decision-making, anxiety reduction, and client satisfaction. However, other studies have focused on client information recall as a means for studying counseling effectiveness. This is largely due in part to the fact that one aspect of a genetic counselor's role is educational in nature. Given the transmission of factual information in genetic counseling interactions, measuring client information recall is one logical approach to measuring counseling effectiveness. Additionally, genetic counseling has obvious psychological consequences for the client following the delivery of particularly difficult information. Outcome measures such as an individual's general wellbeing and perceived personal control (PPC) may be most appropriate in settings where clients' psychological consequences are of interest to the researcher (Davey et al., 2005). Ultimately, the appropriate outcome measure to be used in a study is largely dependent on the client population, the specific intent of the study, and the beliefs about the nature and goals of the counseling.

Historically, outcome research in genetic counseling has rarely been performed in the setting of the Huntington disease population. In this study, we chose to use the outcome measure of client satisfaction to evaluate the effectiveness of the genetic counseling services offered in the Huntington Disease Clinic of UPMC in Pittsburgh, Pennsylvania. For the purposes of this thesis, various outcome measures are briefly discussed to explicate why we felt client satisfaction

was the most appropriate measurement to be used in satisfying the aims of this study. For each of the outcome measures discussed, the literature extends beyond the scope of this paper. References to appropriate literature are provided should further research on the specific outcome measure be desired.

1.3.1.1 Knowledge Measures

Knowledge outcome measures have been used historically as a means of evaluating the effectiveness of genetic counseling services. One aim of genetic counseling is to aid in patient informed decision making by helping the patient to "comprehend the medical facts, including the diagnosis, the probable course of the disorder and the available management." (Michie et al., 1997). In some respects, knowledge measures as part of outcome research are used to measure the extent to which patients receiving genetic counseling are informed after receiving those services. Several knowledge scales have been developed and validated for target populations as a means of using this measure in outcome research. However, researchers in the field are generally moving away from this measure in evaluating genetic counseling services. One reason is that relevant knowledge is perceived differently among individuals, and groups, particularly when from different cultures. Therefore, it has been argued that transferring knowledge measures between research studies is fairly difficult (Kasparian et al., 2007).

A comprehensive literature review on knowledge measures is beyond the scope of this thesis. However, the following references provide additional information regarding this measure: Lippman-Hand et al., 1979, Michie et al., 1997, Pilnick, A., & Dingwall, R., 2001, Rowley et al., 1984, Seidenfield, MJ. & Antley, RM., 1981, and Sorenson et al., 1981, Clarke et al. 1996, Drake et al. 1999; Hunter et al. 2005; Pieterse et al. 2007, Armeli et al. 2005; Decruyenaere et al. 1992; Gordon et al. 2003; Grody et al. 1997; Leonard et al. 1995; Meiser et al. 2005; Sujansky et

al. 1990, Ondrusek et al. 1999, Miller et al. 2005a, Warner et al. 2003, Erblich et al. 2005, Marteau et al. 1988, Lesser & Rabinowitz 2001, Goel et al. 1996, Rostant et al. 2003.

1.3.1.2 Decision-related Measures

Client decision-making is often a central component of genetic counseling interactions. In general, two different approaches are used in research to evaluate decision-making and a client's satisfaction with his/her decision. One approach that does not appear to have been used to date in settings of genetic counseling is assessing patient satisfaction with the decision-making process. Reasons for this are unclear. However, scales have been developed for the purpose of this measurement. Additionally, several scales have been developed to measure a second approach used in research regarding decision-making and client satisfaction. These involve assessing outcomes to a client's decision as well as investigating client's evaluations of their decisions (Kasparian et al., 2007). See the following references for a more thorough discussion of decision-related outcome measures: Miller & Starr, 1967, Sainfort & Booske 2000, Pierce 1993, Holmes-Rovner et al., 1996, Green et al., 2004, Nagle et al., 2006, O'Connor 1997, O'Connor 1995, O'Connor et al. 1998, O'Connor et al. 2006, Drake et al. 1999, Hunter et al. 2005, Kaiser et al. 2002, Matloff et al. 2006, Peterson et al. 2006, Schwartz et al. 2001, Tiller et al. 2006, Wakefield et al. 2007, Mancini et al. 2006, Brehaut et al. 2003, Stalmeier et al. 2005, Michie et al. 2002, Jaques et al. 2005, and Rowe et al. 2006.

1.3.1.3 Psychological Adjustment Measures

Self-reporting measures have been developed in outcome research for the purpose of identifying individuals that may be psychologically impacted by genetic risk assessment. Conveying genetic information in a genetic counseling session has the potential to significantly

impact the psychological wellbeing of a client. For example, one may think of counseling a client regarding familial cancer risk to be taxing on the client's psychological state. These measures have also been utilized in the setting of genetic counseling to identify clients who may benefit from additional interventions or more thorough psychological evaluation. A number of scales have been developed that are either tailored to a specific hereditary condition or provide a general indication of psychological distress (Kasparian et al., 2007).

An individual affected with Huntington disease may have an altered psychological state as a consequence of the disease. The validity of a study may be questioned when an outcome measure studying the effectiveness of genetic counseling utilizes the psychological state of a client. The psychological state of the individual would potentially serve as a confounding variable in the study, unless this variable is controlled for in an appropriate manner. The following references provide a more thorough discussion of different scales that have been developed: Zigmond and Snaith 1983, Bjelland et al. 2002, Keller et al. 2002, Nordin et al. 2002, Lobb et al. 2002a, Lodder et al. 2001, Reichelt et al. 2004, Chapman and Bilton 2004, Spielberger 1983, Hunter et al. 2005, Tercyak et al. 2001b, van Zuuren 1993, Meiser et al. 2005, Nisselle et al. 2004, Brain et al. 2002, Cull et al. 1999, Huiart et al. 2002, Julian-Reynier et al. 1999, Watson et al. 1999, Pieterse et al. 2007, Grosfeld et al. 2000, Gordon et al. 2003, Grody et al. 1997, Decruyenaere et al. 2003, Marteau and Bekker 1992, Radloff 1977, Myers and Weissman 1980, McDowell and Newell 1996, Verdier-Taillefer et al. 2001, Weissman et al. 1977, Vadaparampil et al. 2005, Lerman et al. 1997b, McBride et al. 2002, Horowitz et al. 1979, Cella et al. 1990, Zilberg et al. 1982, Schwartz et al. 2003b, Cella et al. 2002, Skirton 2001, Roy and Andrews 1999, Read et al. 2005.

1.3.1.4 Perceived Personal Control (PPC)

Perceived Personal Control (PPC) is an outcome measure studying whether a client feels he/she possesses the appropriate knowledge, support system, and resources necessary to respond to a stressor in their life in a way that decreases its negative effect. Specifically, it is an outcome measure that examines an individual's response to an event that is occurring in their life. In a genetic counseling context, outcome research utilizes cognitive and decisional components of PPC that serve as predictors of satisfaction with genetic counseling services (Davey et al., 2005).

An individual affected with Huntington disease may have an altered psychological state and cognitive disorder as a consequence of the disease. The validity of a study may be questioned when it uses an outcome measure studying the effectiveness of genetic counseling on the psychological wellbeing of a client. Furthermore, considering a cognitive disorder is one part of the clinical triad of Huntington disease, PPC would not be an appropriate outcome measure in this study. PPC as an outcome measure incorporates a cognitive component referring to an individual's ability to process information in a way to alleviate some stress from a stressful situation. As individuals with HD progress into the later stages of disease, decision-making and cognitive abilities diminish. Hence, PPC as an outcome measure was deemed not appropriate for the HD population. However, the following references will provide more details regarding Perceived Personal Control as an outcome measure: Shiloh et al. 2002, Thompson et al. 1993, Taylor 1983, Berkenstadt et al. 1999, Averill 1973, Carver 1997, Carver et al. 1989, Helder et al. 2002, Biesecker et al. 2000, Tercyak et al. 2004, Scheier and Carver 1985, Scheier et al. 1994, Smith et al. 1989, Miller 1987, 1995, 1996, Miller and Mangan 1983, Steptoe and O'Sullivan 1986, Lerman et al. 1993, Miller et al. 1988, Phipps and Zinn 1986; van Zuuren 1993, Tercyak et al. 2001b, Christensen et al. 1997, Tercyak et al. 2001c.

1.3.1.5 Perceived Risk and Risk Accuracy Measures

There are different methodological approaches that have been developed for assessing a client's perceived risk after receiving genetic counseling. Frequently, perceived lifetime risk for developing a hereditary condition is evaluated. This is often done using a visual analog scale. Additionally, some outcome research is also done to evaluate or assess accuracy of perceived risk, especially in circumstances when accurate objective risk estimates for a condition are known. A comprehensive review of this literature is beyond the scope of this thesis. The following references provide detailed information on this outcome measure: Kasparian et al., 2007, Killgore 1999, McCormack et al. 1988, Miller and Ferris 1993, Nyenhuis et al. 1997, Wewers and Lowe 1990, Codori et al. 2001, Epstein et al. 1997, McCaul et al. 1996, Schwartz et al. 2003a, Marteau et al. 1991, Tercyak et al. 2001b, Lerman et al. 1997b, Watson et al. 1999, McBride et al. 2002, Miller et al. 2005b, Erblich et al. 2000, Kent et al. 2000, Lloyd et al. 1996, Meiser et al. 2000, Rees et al. 2004, Zakowski et al. 1997, Julian-Reynier et al. 1999.

1.3.1.6 Family Functioning and Family Communication Measures

Genetic counseling outcome measures that assess a family's communication pattern and dynamics pre- and post- counseling is a challenging process and are mostly done in the setting of hereditary cancer predispositions. Kasparian et al., 2007 states that "…the aim of past research has been to: (1) characterize the patterns of family functioning for those families presenting for genetic counseling (Koehly et al. 2003); (2) describe the extent to which family members communicate with one another about genetic counseling and/or genetic test results (Hughes et al. 2002; McGivern et al. 2004); (3) describe the nature of, and motivations for, family communication about genetic risk (Hughes et al. 2002; McGivern et al. 2004); and (4) identify the various "roles" played by different members within a family (e.g. caretaker, informer;

DudokdeWit et al. 1997; Foster et al. 2004; Koehly et al. 2003).” Given the difficulty in the utilization of this outcome measure as well as its lack of use in populations other than hereditary cancer populations, this outcome measure was not chosen for this study. More detailed information about this outcome measure may be found in the following references: Koehly et al. 2003, McInerney-Leo et al. 2005, Peterson et al. 2003, Green et al. 1997, Julian-Reynier et al. 2000, 1996, Lerman et al. 1998, Wilson et al. 2004, Blandy et al. 2003, Foster et al. 2004, Gaff et al. 2005, Hallowell et al. 2005, Hamilton et al. 2005, Kenen et al. 2004, Bowen et al. 2004, Claes et al. 2003, Hughes et al. 1999, Tercyak et al. 2001a, 2002, Koehly et al. 2003, Hughes et al. 2002, McGivern et al. 2004, DudokdeWit et al. 1997, Mesters et al. 1997, van Oostrom et al. 2007a, b, 2003, Cappelli et al. 2005, Skinner et al. 1995, Olson et al. 1985, Hudson 1992, Holahan and Moos 1986, Beavers et al. 1985.

1.3.1.7 Client Satisfaction

Shiloh *et al.* (1990) developed a client satisfaction outcome measure to evaluate the effectiveness of genetic counseling services. In their study, the aim was to design a questionnaire and to elicit patient satisfaction by administering the questionnaire to 76 clients in a genetic counseling center as well as to 56 parents in a pediatric outpatient clinic, which served as the control group. The researchers wanted to determine the quality and outcome of genetic counseling by studying clients’ general level of satisfaction. Additionally, they were interested in determining the composition and determinants of satisfaction in a genetic counseling setting. They constructed a measure of satisfaction using three components of satisfaction: instrumental, affective, and procedural. Instrument refers to a client’s perspective on the healthcare professional possessing the required skills necessary in the delivery of services. The affective component relates to the healthcare professional showing affect in the delivery of services,

specifically exhibiting interest and care. Lastly, the procedural component refers to the administrative aspect of the delivery of the services.

Client satisfaction has not been widely used in genetic counseling outcome research. Shiloh *et al.* (1990) suggest that this measure is not commonly used due to a "lack of an acceptable measure of this factor". In other words, they argue that a standardized measure of satisfaction in a genetic counseling setting has not been developed. Shiloh *et. al.* (1990) state that their study developed a measure of satisfaction that is standardized and acceptable for use in client satisfaction outcome research.

Presently, debate still exists over the use of client satisfaction as a measure for evaluating effectiveness in genetic counseling. Several factors complicate its use as an outcome measure. Genetic counseling is believed to be less satisfying than other medical services from a client perspective. Often, clients are not receiving their desired outcome of the services; i.e. receiving results of genetic testing that may not explain an adverse health outcome. Additionally, genetic counselors often provide information to a family that is perceived negatively by clients. These two factors may confound or negatively impact the judgment of clients and affect their evaluation of the genetic counseling services. The evaluation may not adequately reflect their view of the service itself. Rather, their negative interpretation of the information delivered to them resulted in an undesirable evaluation of the services.

Despite these concerns for utilizing patient satisfaction in outcome research, in this study we adapted the Shiloh *et al.* (1990) comprehensive measure of satisfaction. This was deemed appropriate for the HD population for multiple reasons. Measuring client satisfaction allows for adequate evaluation of genetic counseling services and is not dependent on psychological or cognitive abilities of the client. Rather, the study utilized two of the three components of

satisfaction including instrumental and affective. Additionally, considering the central role genetic counseling has in the predictive testing process, client perceptions of genetic counseling services and their satisfaction with these services are important in ensuring the genetic counselor is providing satisfactory services and meeting clients' expectations. Outcome research on genetic counseling in the HD population has not been completed to date and a client satisfaction outcome study would contribute knowledge on the effectiveness of counseling in a predictive testing setting. The results of the study have the potential to inform outcome research conducted in other predictive testing settings. In addition, the ease of obtaining satisfaction data is ideal in a patient population whose mental capacity could be too limited to participate in outcome research using measures that rely on cognitive processes such as knowledge measures. Individuals affected with HD or their family members or caregivers can easily assess the components of satisfaction, therefore making satisfaction an appropriate outcome measure for this study. Outcome measures such as decision-related measures, perceived risk and risk accuracy measures, and family functioning and family communication measures may be appropriate in a HD setting for other research studies. However, the goal of this research was to determine effectiveness of counseling services by utilizing and determining patient perceptions. A satisfaction outcome measure was considered the most appropriate measure in order to achieve the aims of this research.

2.0 MATERIALS AND METHODS

This thesis project was reviewed by the University of Pittsburgh's Institutional Review Board and was determined to meet criteria for exemption (Appendix A).

2.1 PARTICIPANTS

This research study sought to elicit perceptions of satisfaction with genetic counseling from three groups of individuals: individuals affected with Huntington disease, family members (individuals at-risk based on their family history of HD), or caregivers for individuals with HD. Several groups of participants were recruited for this study. Individuals who attend the monthly Huntington Disease (HD) Clinic of UPMC in Pittsburgh, Pennsylvania and received genetic counseling services were invited to participate in the study. Individuals with Huntington disease who were members of the Pittsburgh Institute for Neurodegenerative Diseases (PIND) Movement Disorders Research Registry and had received genetic counseling in the past were also invited to participate. Additionally, the Huntington Disease Society of America Inc. Western Pennsylvania Chapter holds a monthly support group in the city of Pittsburgh. Individuals who had previously received genetic counseling services through the HD Clinic and currently attend the support group meetings were also invited to participate.

2.2 INSTRUMENTATION

2.2.1 Survey Design

The final draft of the survey instrument, which can be seen in Appendix C, was constructed and adapted from the comprehensive measure of satisfaction instrument seen in Appendix D developed by Shiloh *et al.* (1990). Primary author, Dr. Shoshana Shiloh, gave her written permission for the use of the Satisfaction with Genetic Counseling instrument in this study. Shiloh *et al.* (1990) instrument was adapted to include all of the relevant questions pertaining to the genetic counseling services offered in the Huntington Disease Clinic of UPMC in Pittsburgh, Pennsylvania.

The survey was evaluated several times by Robin E. Grubs, MS, PhD, LCGC, a leader in the field of outcome research in genetic counseling. The primary author of this thesis project, Alicia Martinez, BS, worked in collaboration with Robin Grubs, MS, PhD, LCGC to add, remove, and restructure questions in the survey making it appropriate for participants from the HD population. Genetic epidemiologist, John R. Shaffer, PhD, recommended and assisted in constructing the Likert scale, which is the psychometric scale measuring levels of agreement/disagreement used for the responses to the questions in the survey.

The first part of the survey consisted of an introduction, served the purpose of describing the study to the participants and its purpose of evaluating patient satisfaction with genetic counseling services. It provided the participants with the names of the two genetic counselors who have provided these services in the HD Clinic over the years, Betsy Gettig, LCGC and Christa Lorenchick, CGC. The introduction also provided the participants with basic instructions on filling out the survey and stressed that responses would be anonymous. The HD population in

Pittsburgh, PA is relatively small; therefore, there was the potential for participants to feel uncomfortable answering questions in the survey if they felt their answers would not be anonymous. It was described in the introduction that there were no foreseeable risks associated with the study, nor were there any direct benefits to the participants. The introduction outlined that participation in the study was voluntary and individuals were given the opportunity to opt out at any time.

The main body of the survey instrument was comprised of 26 questions, demographic questions, and two open-ended questions. Demographic questions asked age, gender, reason for current/last visit to the clinic, and who is completing the survey. Additional questions were targeted toward analyzing participants' feelings toward the genetic counseling services they had received in clinic. Participants were given the opportunity to choose "N/A" if a question was not applicable to their experience with these services. They were asked to provide commentary regarding the satisfaction with the genetic counseling services they received as well as their satisfaction with the genetic counselor who delivered the services to determine any trends in data.

Participants who completed the paper version of the survey were given the option of omitting answering certain questions as well as choosing more than one answer for a given question. The online version of the survey through Qualtrics Survey System prohibited participants from moving on to different parts of the survey without completing every question.

2.2.2 Recruitment E-mail

E-mails were drafted and sent out to invite individuals to participate in an electronic version of the survey if they did not attend the Huntington Disease Clinic of UPMC on a monthly basis. A copy of the initial e-mail and reminder e-mail can be seen in Appendix B.1 and B.2. The social workers for the HD Clinic, Peggy Humbert and Tammy Makoul, and Larry Ivanco, MSW, BSC sent out the e-mails to participants in order to ensure anonymity.

2.3 PROCEDURES

2.3.1 Survey Distribution

Surveys were distributed to willing participants by two different means. Individuals affected with Huntington disease and their families who attend the monthly Huntington Disease Clinic of UPMC and received genetic counseling services were given paper versions of the survey. This version of the survey was distributed to the participants by the healthcare professionals who contribute to the care of the patient and their families in the HD Clinic. Valerie Suski, DO, the neurologist for the clinic, and the social workers, Tammy Makoul and Peggy Humbert, distributed the survey to participants in the clinic.

For those individuals and their families who do not attend the HD clinic on a regular basis, the survey was made available electronically through the University of Pittsburgh's Qualtrics Survey System. An e-mail was sent by Larry Ivanco, MSW, BSC and the social workers to individuals in the Pittsburgh Institute for Neurodegenerative Diseases (PIND)

Movement Disorders Research Registry and the Huntington Disease Society of America Inc. Western Pennsylvania Chapter monthly support group respectively with an anonymous link to the electronic survey. Some individuals at the monthly support group received a paper copy of the survey to be filled out during the support group, which was distributed by the social workers. The primary author of this project did not handle the distribution of the survey to maintain anonymity.

The individuals with the role of distributing the survey to participants had to strategically determine whom to offer to complete the survey. The inclusion/exclusion process had to be completed because some current patients of the clinic have not received genetic counseling services and thus would not be appropriate candidates for this study.

2.3.2 Results Collection

Responses to the survey were collected electronically and through the gathering of the completed paper versions of the survey. Several surveys were distributed to participants by hand during clinic and responses were received by mail. In this circumstance, participants returned the survey with an unmarked envelope without a return address to the social workers in order to ensure anonymity. The primary author of this project did not handle collecting the paper surveys directly from the participants in order to maintain anonymity.

2.4 STATISTICAL ANALYSIS

Quantitative statistical analysis of data obtained from this study was completed under the guidance of a genetic epidemiologist at the University of Pittsburgh. Microsoft Excel® was the tool used to compile the raw data exported from Qualtrics Survey System and the paper versions of the survey. Much of the descriptive statistics was performed through Microsoft Excel® analysis tools; however some values were computed by hand. The primary author attempted to analyze the responses to the open-ended questions by categorizing the responses into themes. The themes were utilized in order to gain additional patient perspectives about satisfaction with genetic counseling services that were otherwise not captured through the 26 multiple-choice questions of the survey.

The Likert scale was scored 1-5 for analysis of 'positive' worded questions of the survey: "Strongly agree" = 5, "Agree" = 4, "Neither agree nor disagree" = 3, "Disagree" = 2, and "Strongly disagree" = 1. For 'negative' worded questions, a reverse scoring method was used for analysis: "Strongly disagree" = 5, "Disagree" = 4, "Neither agree nor disagree" = 3, "Agree" = 2, "Strongly agree" = 1. Participants were given the opportunity to answer questions with "N/A" if the question did not apply to the genetic counseling services they received at the Huntington Disease Clinic. In the circumstance where participants chose "N/A" for a question, the response was not included as part of the analysis. When a participant did not answer a certain question, that question was also not included as part of the analysis.

Of note, one participant partially completed an online version of the survey through the Qualtrics Survey System. This participant's responses were not included in the data analysis.

2.4.1 Likert Scales and Data Analysis

A commonly debated topic is proper statistical analysis of Likert scale data. Specifically, disagreement exists in the literature upon whether Likert items should be considered ordinal data or interval data. Ordinal data is a statistical data type consisting of numerical scores that exist on an arbitrary numerical scale with the purpose of ranking data points. Interval data is a statistical data type that generally consists of integer data where ordering and distance measuring between data points is possible (Norman, 2010).

The desired analysis for this project was to measure general satisfaction of genetic counseling in the Huntington Disease Clinic by using normal statistical procedures, including computing the normal mean and normal confidence intervals around the mean. Some will argue that Likert scales consisting of asymmetric categories, representing ordinal-type data, inhibit the use of normal statistics due to potential bias toward a specific outcome or categories that are not equidistant. However, Likert scales presenting with symmetry of categories about a midpoint are argued to behave like interval-type data where normal statistical procedures can be utilized (Norman, 2010).

In this study, a Likert scale was constructed to include symmetric categories about a midpoint. The investigators in this study assumed the Likert scale utilized approximated interval data and used normal statistical procedures for the data analysis.

2.4.2 Genetic Counseling Satisfaction Survey Questions

Table 2 lists the 26 questions that were that were asked in the genetic counseling satisfaction survey that utilized the Likert scale for which participants could choose their response. For sake

of ease, it can be referenced when viewing tables and figures to view the full question that corresponds to the question number.

Table 2. Genetic Counseling Satisfaction Survey Questions

Question #	Question
Q1	The genetic counselor showed interest in your personal problems beyond what is medically required. For example, did she provide you with additional resources, etc?
Q2	I considered going to another genetic counselor regarding Huntington disease (HD).
Q3	The genetic counselor explained HD to you clearly.
Q4	The genetic counselor met your expectations of her/him.
Q5	You believe that the genetic counselor cares for you as a person.
Q6	You feel comfortable calling the genetic counselor to ask another question.
Q7	The genetic counselor reassured you.
Q8	The genetic counselor listened to what you had to say.
Q9	The genetic counselor was considerate of your emotional state during the meeting.
Q10	You were satisfied with the way in which information was transmitted to you.
Q11	The genetic counselor showed enough dedication in treating you, your family member, or the individual you are caring for with HD.
Q12	The genetic counselor understood what was bothering you (if anything).
Q13	I think I could have received more considerate care from another genetic counselor.
Q14	The genetic counselor made you feel you are "in good hands".
Q15	The genetic counselor made you feel that she/he knows how to handle HD.
Q16	The genetic counselor gave you enough of her/his time.
Q17	The genetic counselor was sensitive and tactful during your conversation.
Q18	The genetic counselor gave you the necessary treatment.
Q19	The genetic counselor is an expert in HD.
Q20	The counseling that you received helped you cope better with your situation.
Q21	The genetic counselor lessened your worries.
Q22	You are satisfied with the information you received in counseling.
Q23	You were comfortable to talk about yourself (or family member) during the counseling session.
Q24	I think I could have gotten better treatment from another genetic counselor.
Q25	You believe that the counseling was given in the appropriate setting.
Q26	Overall you are satisfied with the counseling you received.

3.0 RESULTS

3.1 DEMOGRAPHICS

Approximately 50 individuals were invited to participate in the study, and of these individuals, 33 agreed to participate and completed the survey. Thus, the response rate was approximately 66%. The majority of the participants completed the paper version of the survey (87.9%; n=29). Four individuals (12.1%) completed the electronic version of the survey through the University of Pittsburgh's Qualtrics Survey System. Of the 33 participants, 22 were female (67%) and 11 were male (33%). The mean age of participants who completed the survey was 47 years with the range being from 23 to 74 years.

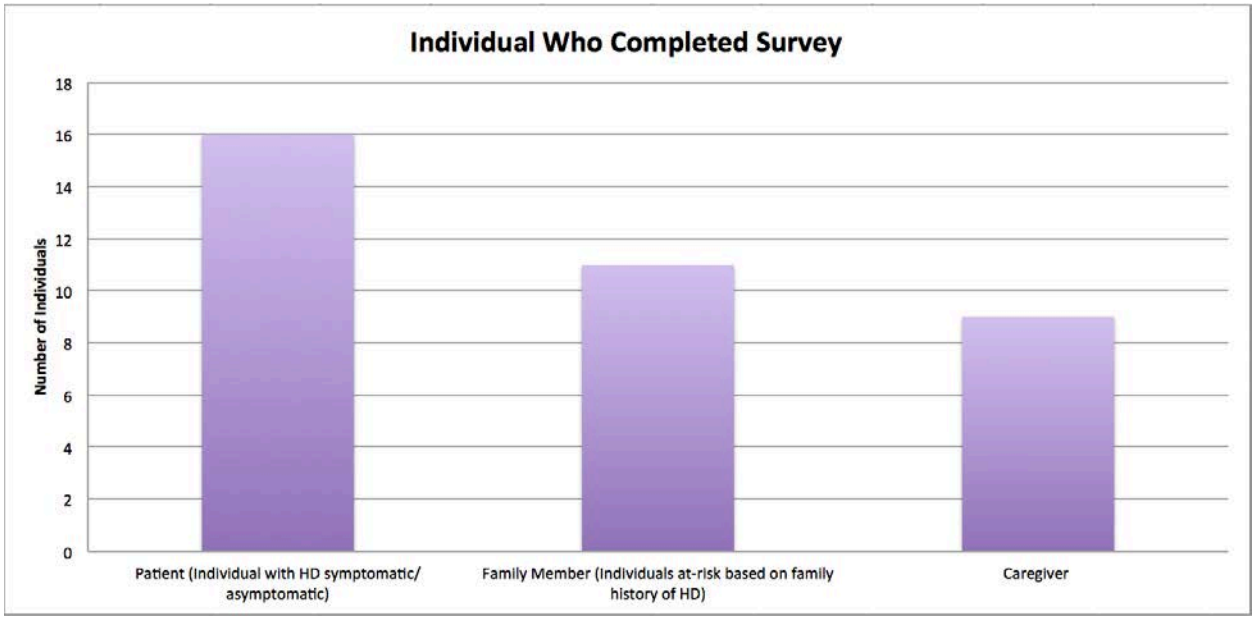

Figure 1. Individual Who Completed Survey

Sixteen participants were patients of the Huntington Disease Clinic of UPMC who were either symptomatic or asymptomatic, as can be seen in Figure 1. Eleven were family members (individuals at-risk based on a family history of HD) and nine were caregivers.

3.2 SATISFACTION DATA

To complete the second aim of the study, descriptive statistics and analysis of open-ended responses was conducted.

3.2.1 Descriptive Statistics

Table 3. Responses to Questions by Counts and Percentages

Tabulated Responses by Question in Counts and Percentages									
Question	Strongly agree	Agree	Neither agree nor disagree	Disagree	Strongly disagree	N/A	Missing	Means	95% Confidence Intervals
1	18 (55%)	11 (33%)	1 (3%)	0	2 (6%)	1 (3%)	0	4.34	±0.35
2	1 (3%)	1 (3%)	2 (6%)	11 (33%)	17 (52%)	1 (3%)	0	4.31	±0.33
3	21 (64%)	12 (36%)	0	0	0	0	0	4.64	±0.17
4	25 (76%)	8 (24%)	0	0	0	0	0	4.76	±0.15
5	23 (70%)	10 (30%)	0	0	0	0	0	4.7	±0.16
6	19 (58%)	12 (36%)	2 (6%)	0	0	0	0	4.52	±0.21
7	19 (58%)	14 (42%)	0	0	0	0	0	4.58	±0.17
8	24 (73%)	9 (27%)	0	0	0	0	0	4.73	±0.15
9	22 (67%)	11 (33%)	0	0	0	0	0	4.67	±0.16
10	24 (73%)	9 (27%)	0	0	0	0	0	4.73	±0.15
11	23 (70%)	10 (30%)	0	0	0	0	0	4.7	±0.16
12	20 (61%)	11 (33%)	0	0	0	2 (6%)	0	4.63	±0.17
13	0	0	0	11 (34%)	20 (61%)	2 (6%)	0	4.58	±0.23
14	24 (73%)	9 (27%)	0	0	0	0	0	4.73	±0.15
15	21 (64%)	12 (36%)	0	0	0	0	0	4.64	±0.17
16	21 (64%)	12 (36%)	0	0	0	0	0	4.64	±0.17
17	25 (76%)	8 (24%)	0	0	0	0	0	4.76	±0.15
18	17 (52%)	12 (36%)	0	0	0	3 (9%)	1 (3%)	4.59	±0.17
19	17 (52%)	15 (45%)	1 (3%)	0	0	0	0	4.48	±0.19
20	20 (61%)	12 (36%)	1 (3%)	0	0	0	0	4.58	±0.19
21	14 (42%)	12 (36%)	6 (18%)	1 (3%)	0	0	0	4.18	±0.29
22	18 (55%)	14 (42%)	0	0	0	1 (3%)	0	4.56	±0.17
23	20 (61%)	13 (39%)	0	0	0	0	0	4.61	±0.17
24	1 (3%)	1 (3%)	0	8 (24%)	21 (64%)	2 (6%)	0	4.52	±0.32
25	18 (55%)	14 (42%)	1 (3%)	0	0	0	0	4.52	±0.19
26	23 (70%)	10 (30%)	0	0	0	0	0	4.7	±0.16

In order to assess general satisfaction, participants were asked to rate how they felt about each question related to their genetic counseling services ranging from strongly agree to strongly disagree. Table 3 displays the responses to questions 1-26 in counts and percentages. Two (6%,

n=33) or fewer participants responded negatively to each question. Table 3 does not reflect the reverse scoring method used for the 'negative' worded questions, 13 and 24. Figure 2 is an alternative way of presenting the satisfaction data. It is a graphical representation of the mean response for questions 1-26 with 95% confidence intervals. Each response was scored 1-5, with 'Strongly agree' corresponding to a score of 5 and 'Strongly disagree' corresponding with a score of 1 for 'positive' worded questions. For the 'negative' worded questions, a reverse scoring method was utilized and also depicted in Figure 2. In this case, 'Strongly disagree' corresponded to a score of 5 and 'Strongly agree' corresponded to a score of 1. The mean response for each question is depicted below. The mean response for all of the questions combined was 4.59 on the scale of 1-5.

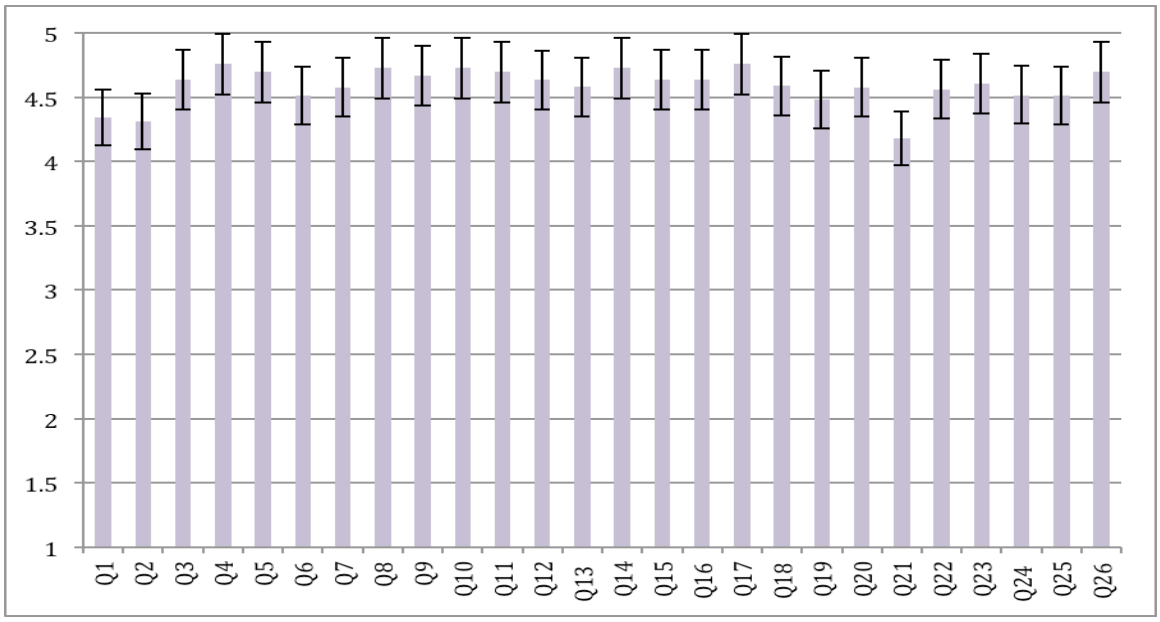

Figure 2. Mean Responses and Confidence Intervals for Q1-Q26 (95%)

3.2.1.1 Subscales

In order to further assess satisfaction, the results were stratified into subscales that appraised two specific components of a genetic counseling session in the HD Clinic. The first subscale includes

all of the questions in the survey that were centered around assessing inherent characteristics of the genetic counselor, i.e. the counselor's affect, genetic expertise on Huntington disease, and whether the genetic counselor provided a setting that was comfortable for the patient to discuss themselves or their family members. The second subscale focuses on assessing the responses to the questions in the survey that were centered on the patient's perceptions during/after the counseling services, i.e. whether the patient considered going to another genetic counselor for services, whether the counselor met the patient's expectations, and their overall satisfaction with the counseling. Table 4 below shows the stratification of questions in the survey into subscales. To view the entire question corresponding to the question number, please reference Table 2 above.

Table 4. Subscales

	Subscale 1-Inherent characteristics of the genetic counselor	Subscale 2-Patient perceptions during/after counseling
Question #	1, 3, 5, 7, 8, 9, 11, 12, 14, 15, 17, 19, 21, 22, 23	2, 4, 6, 10, 13, 16, 17, 18, 20, 24, 25, 26

In order to assess general satisfaction of the two subscales, mean and error bars were computed for each subscale. The mean for each subscale was calculated by averaging the mean responses from Figure 2 for each question in a subscale. The mean response for each subscale was greater than 4.5. Figure 3 below shows the mean and 95% confidence intervals for each subscale.

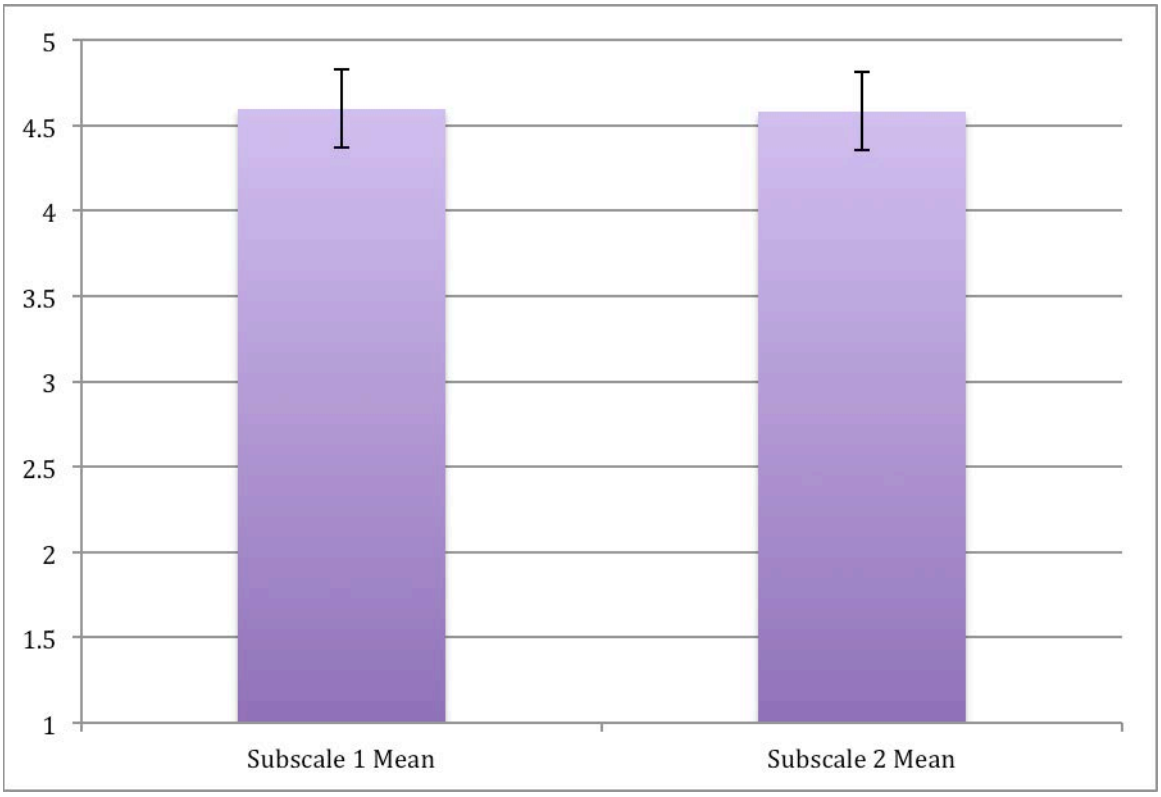

Figure 3. Subscale Mean Response and Confidence Intervals (95%)

3.2.2 Open-Ended Responses

Participants were asked to comment on their general satisfaction with specific components of the genetic counseling services they had received in two open-ended questions. The responses to these questions were categorized into themes. The first open-ended question focused on specific components of the counseling that the participant was most satisfied with. Tables 5, Table 6, and Table 7 contain participants' responses to the first question. The three themes that emerged through the analysis include attributes of the genetic counselor, general feelings of satisfaction, and the setting of the counseling. The comments elicited from the first open-ended question support the hypothesis that patients in the HD Clinic are generally satisfied with the genetic counseling services they have received. The first question read as follows: "Please comment on

how satisfied you were with the counseling services you received and what you were most satisfied with."

Table 5. Open-Ended Responses Question 1, Theme 1

Attributes of the Genetic Counselor
It is reassuring to have a counselor available whose interest is to help you with every problem you may encounter. My most valuable information I have received about HD has come from the counselor
The counselor spoke on a difficult subject on our level-very caring
Concern of well being and mental state understanding of what patients are dealing with
Very satisfied, the demeanor of the counselor was caring, interested and empathetic
The counseling services prompted important information and care to myself, children, and spouse. This is a reason we changed doctor groups, for the overall attention to all phases of this disease.
The patience with which they have.
She was very caring, compassionate, and understanding
I am very satisfied with the genetic counseling services the counselor helped me with. She ultimately gave me hope for my future.

Table 6. Open-Ended Responses Question 1, Theme 2

General Feelings of Satisfaction
During my visit in which I received my results, I honestly couldn't have done it w/o genetic counselor
I would never go anywhere else
The counselor was exceptional
Very satisfied I liked having a support team present during counseling services.
I'm very satisfied
I and my family were very pleased with the services we received.
The patience with which they have.
The counseling greatly helped our family. In many ways.

Table 7. Open-Ended Response Question 1, Theme 3

Setting of the Counseling
Setting was very cramped in small office. But wouldn't make a difference in outcome. Comfort factor? Intimidating a bit!

The second open-ended question focused on asking participants to comment on the genetic counselor and their general satisfaction with her. The question was constructed in order to have participants comment on specific attributes about the genetic counselor that they found to be most effective and attributes with which they were most satisfied. The second question read as follows: "Please comment on your satisfaction with the genetic counselor who provided you with the genetic counseling services, i.e. what did you like most about the genetic counselor?" Table 8 and Table 9 contain all of the participants' responses to the second open-ended question. The two themes that emerged through the analysis include inherent characteristics of the genetic counselor and general feelings of satisfaction. Participants' responses to the second open-ended question support the hypothesis of this research study.

Table 8. Open-Ended Response Question 2, Theme 1

Inherent Characteristics of the Genetic Counselor
She was very patient and listened to [patient's] needs
Caring
Caring/funny
Very personable and knowledgeable willing to discuss details and willing to help in any way
Peaceful manner who talked and listened with both me and the Huntington patient.
Friendly
The counselor was knowledgeable and personable. Providing a comfortable setting to deal with this disease.
She is organized.
She was very kind, compassionate, caring, and understanding with each of us.
Caring, understanding, informative
Friendly, personal, sympathetic easily understood

Table 9. Open-Ended Response Question 2, Theme 2

General Feelings of Satisfaction
The genetic counseling was helpful in understanding of my HD needs. They got me through the appointment.
Their effort in helping people is amazing I am truly satisfied
We were along the way before coming to UPMC-everyone very helpful and caring
The counselor was phenomenal. She went out of her way to maintain my confidentiality. She was great about trying to keep me upbeat without being hopelessly optimistic. She will be sincerely missed.
The counselor explained to me how I need to live life now and go after all of my dreams. She told me not to give up and always have faith in my decision to never let HD be a reason to give up and always have hope and faith in a cure!

4.0 DISCUSSION

The results from the client satisfaction survey were utilized in evaluating the effectiveness of genetic counseling services in the UPMC HD Clinic to accomplish the first aim of this research study. The results were statistically analyzed to achieve the second aim of the study to determine support for the study's hypothesis. The results of the study suggest that the patients in the UPMC HD Clinic are generally satisfied with the genetic counseling services they have received. Services in the HD clinic have not been evaluated prior to this study, and this research has the potential to inform the provision of services in the clinic. Additionally, this study has the potential to inform genetic counseling outcome studies in other settings.

4.1 DEMOGRAPHICS

Approximately 50 individuals were invited to participate in this study through the two means of survey distribution, i.e. paper version and online format. Thirty-three participants agreed to participate in the study and subsequently completed the survey. The majority of the participants were female (67%, n=33) and the average age of participants who completed the survey was 47.

The majority of participants completed the paper version of the survey, and most were current patients of the UPMC HD Clinic. A few participants chose more than one answer when answering the question of who was filling out the survey. It is possible that a participant is

someone who is at-risk for developing HD and is also a caregiver. Additionally, two participants omitted answering this question. For this reason, the number of individuals in Figure 1 does not reflect a total of 33 responses. A total of 36 participants were included in the analysis of the demographics.

4.2 DESCRIPTIVE STATISTICS

The data support our hypothesis that patients are generally satisfied with the genetic counseling services in the UPMC Huntington Disease Clinic. General satisfaction was measured according to a Likert scale scored from 1-5. Five on the scale represents the highest level of satisfaction with the genetic counseling services, and one represents the lowest level of satisfaction. The mean response for each question asked in the survey was greater than 4.18. The mean response for all of the questions combined was 4.59 on the scale of 1-5. This corresponds to a high level of patient satisfaction with the counseling services in the clinic.

The data were further stratified into subscales with the intent of evaluating the multiple choice questions that were focused on satisfaction with inherent characteristics of the genetic counselor and those that were centered on patient's perceptions during/after the counseling services. The mean response for all of the questions in the first subscale was 4.60. The mean response for all of the questions in the second subscale was 4.58. These findings suggest that the participants in the study were generally highly satisfied with the genetic counselor providing the counseling services as well as the counseling itself.

Limited research exists on client satisfaction of genetic counseling services in the Huntington disease population. Furthermore, limited studies exist on client satisfaction in any

setting offering genetic counseling services. However, our data compared similarly with others in the literature measuring global client satisfaction with genetic counseling services in other settings. Shiloh *et al.* (1990) determined that the mean level of satisfaction in their study was 3.13 on a scale of 1-4. Davey *et al.* (2005) evaluated genetic counseling services provided by Genetic Services of Western Australia (GSWA) by utilizing several different outcome measures, one being client satisfaction. These researchers measured client satisfaction using the instrument developed by Shiloh *et al.* (1990). Overall, satisfaction of genetic counseling services was measured to be 3.7 on a 4-point scale, corresponding to relatively high global satisfaction with genetic counseling services.

Davey *et al.* (2005) reported in their study that participants felt there were two components of their genetic counseling services that were essential in their satisfaction with these services. The first component, the instrumental component of satisfaction, describes the genetic counselor as a competent source of knowledge. The second component, the affect component of satisfaction, addresses the manner in which patients felt the counselor related to them as a person. The data in our study support this same finding. Approximately 97% of participants either strongly agreed or agreed that the genetic counselor was an expert in HD. The majority of participants also responded either strongly agreed or agreed to questions in our survey that pertained to the affect component of satisfaction. These findings would also support an argument that the instrumental and affect components to satisfaction are important components to patients' general satisfaction with genetic counseling services.

While this study demonstrated that overall clients are satisfied with genetic counseling services in the HD Clinic, it also identified an area for improvement or need for further exploration. Approximately 18% (6, n=33) of participants responded that they neither agreed nor

disagreed to question 21. Question 21 addressed whether the genetic counselor lessened the client's worries. This finding could suggest several possibilities. One possibility is that the counselor is not sufficiently addressing the clients' worries in the clinic. Another possibility is that the client responded with 'neither agree nor disagree' because the client did not have any worries at the time they were receiving genetic counseling and answered the survey question in a neutral manner for this reason. In addition, a client's worries/concerns may be difficult to mitigate regarding HD because some worries are unable to be assuaged, even by the most experienced genetic counselor. Regardless, this finding suggests an area for possible improvement in genetic counseling services, keeping in mind the challenge of generalizing this finding due to the small patient population (see Study Limitations below). Acknowledging this finding, genetic counselors may wish to pay extra attention to ensure that they are addressing client's concerns/worries when delivering genetic counseling services.

4.3 OPEN-ENDED QUESTION RESPONSES

The responses to the open-ended questions of the survey were categorized into themes in order to identify additional patient perspectives that may not have been captured in the 26 multiple choice questions of the survey. The majority of the responses contributed to the assessment of high general satisfaction with genetic counseling services in the UPMC HD Clinic. The themes that emerged from the analysis of the open-ended responses revealed that attributes and inherent characteristics of a genetic counselor play a significant role in client satisfaction with services. Additionally, one response commented on the setting of the delivery of services being less comfortable than what the participant had expected, but it did not affect the outcome of the

services. This response provides insight into another possible area of improvement. The researchers assessed this response and concluded that the participant who commented negatively on the setting where the genetic counseling took place would favor a room with greater space. Improving the setting where the genetic counseling takes place in the UPMC Huntington Disease Clinic has the potential to increase the overall satisfaction of services.

4.4 STUDY LIMITATIONS

This was an exploratory study that aimed at examining participant perceptions of genetic counseling services by measuring client satisfaction with these services. Some potential limitations to the study-design are noted here. Participants in the study were given the opportunity to complete the survey and were also able to opt out of the study at any time. Self-selection bias has to be acknowledged for this reason. Additionally, the sample size was relatively small in this study and not chosen in an unbiased way. Due to these reasons, generalizations about satisfaction with genetic counseling services in this setting as well as other settings cannot be made. This is because this small sample size is likely not representative of all participants who receive genetic counseling either in a predictive testing setting, such as HD, or other settings where genetic counseling is offered. Additionally, participants in the study were chosen by the neurologist and social workers in the clinic in order to recruit those that have received genetic counseling services. Considering selection for this study was not random, bias could be introduced in this way. Trends observed in this study would be better supported in a study with a larger sample size and whose selection for recruitment is random. Rather, these

findings could be used to examine particular aspects of genetic counseling in which patients are satisfied with and could be used inform further research on the topic.

Another aspect of the study design that made interpretation difficult was the design of the instrument itself. The structure and wording of the questions posed difficulties for several participants completing the survey. The researchers rescored two of the participants' responses due to inconsistencies with their responses to the multiple-choice questions and open-ended questions. For example, these two participants expressed high satisfaction with the genetic counseling services in the open-ended questions, but their responses to all of the multiple choice questions reflected low levels of satisfaction. In other words, these participants' responses were either a 1 or 2 on our scale of 1-5. The researchers attributed these discrepancies to the participants not completely reading the multiple-choice answers before answering the question. Therefore, the researchers found it appropriate to rescore them. Moreover, there were some outliers in the data, i.e. responses to questions that reflected low satisfaction in a patient's survey that otherwise had responses reflecting a high level of satisfaction in all other questions. The researchers did not rescore these questions, but it is possible these findings could be a result of the participant not fully reading the multiple-choice question and/or the responses.

Another aspect of the study that limited the interpretation is the inconsistency between the two means of survey distribution. Mixed-mode surveys (i.e. utilizing more than one mode of data collection) are relatively common in research practices. Debate in the literature exists over potential advantages and disadvantages to this approach. Several advantages are that this approach may increase response rate as well as reduce survey "non-response error". (Brogger et al, 2002). Some disadvantages to this approach may exist in relation to the "mode effects", which refer to the differences in the way respondents answer questions in the survey depending on what

mode of data collection that is being used. Other disadvantages could be survey errors that are influenced by the mode of data collection. Non-response and sampling errors are an example of this. In our study there were some survey errors and inconsistencies between the modes of data collection. Participants completing the online survey were forced to answer questions before proceeding to the next question or part of the survey. This holds true for the open-ended questions in the electronic version. Individuals who were filling out the paper version of the survey were not forced to answer a question before proceeding on. Hence, individuals filling out the paper-version were able to skip answering a question. Furthermore, individuals filling out the electronic version of the survey were not permitted to choose multiple answers for demographic questions. Several participants who completed the paper version responded with several answers to the demographic questions. These inconsistencies could result in data analysis that is not completely accurate. However, in concordance with the conclusions made in a study by Meckel et al. 2005 the researchers felt that the advantages of this mixed-mode survey justified and took precedence over the disadvantages. Future studies should pay particular attention to ensure that all modes for survey distribution are consistent and congruent.

A second major limitation to this study is that the intent focused primarily on assessing general satisfaction with genetic counseling services and not satisfaction with the genetic counseling process itself. The majority of outcome research on genetic counseling services has the intent of providing suggestions for the ways in which the services could be improved. While this study elicited some suggestions for improving services, i.e. the setting that the counseling is delivered or addressing client's worries/concerns, a study focused on investigating the genetic counseling process itself could yield greater suggestions for improvements of this process (Davey *et al.*, 2005). Lastly, a study that is comprised of participants who better represent patient

populations that present for genetic counseling would better inform potential proficiencies and improvements in genetic counseling services.

4.5 FUTURE RESEARCH RECOMMENDATIONS

In the future, it would possibly be beneficial to embark on a more complete quantitative analysis of patient satisfaction potentially utilizing a random sample and more advanced statistical procedures for data analysis. This might be helpful in identifying specific aspects of counseling that are most satisfying for patients or most consistently achieve the desired outcomes. Additionally, it may be useful to incorporate a qualitative approach in an outcome study. With its ability to explore complex social processes and elicit participants' experiences in rich detail (Beeson, 1997), qualitative research would allow for a more robust understanding on patients' perspectives of their genetic counseling experience. Moreover, qualitative research has the potential to identify issues that were not anticipated by researchers (Beeson, 1997) and therefore, may uncover aspects of genetic counseling outcomes that generate novel ideas. Future studies could also employ a different measure to obtain patient perspectives on other outcomes of counseling in a predictive testing setting, such as HD.

A study focused on investigating the genetic counseling process itself could yield suggestions for improvements of genetic counseling services. According to Biesecker and Peters, 2001: "Process studies investigate the content, behavior and relationships that make up genetic counseling sessions. They strive to answer questions about what happens in practice and how it works." In contrast, outcome studies "…assess measurable changes in client outcomes as a result of genetic counseling." (Biesecker and Peters, 2001). Future studies may employ combining

process studies and outcome studies using quantitative approaches to analyze potential impacts of genetic counseling process on client outcomes (Meiser et al. 2008). Results from this study utilizing this approach could yield potential improvements of genetic counseling services because the content of a genetic counseling session may affect patient perceptions of the counseling as well as more broadly affect the outcome of the counseling services (Butow and Lobb 2004, Biesecker and Peters 2001, Clarke et al. 1996, Meiser et al. 2008).

5.0 CONCLUSION

This study had the aim of eliciting patient perspectives of satisfaction with genetic counseling services in the UPMC Huntington Disease Clinic in Pittsburgh, Pennsylvania. Minimal outcome research has been conducted in the HD setting, and this study provided an opportunity to gather information on patient perspectives of genetic counseling services in a predictive testing setting such as HD. Outcome research in genetic counseling in any setting is important to evaluate the services to ensure that they are being provided in a satisfactory manner and may even identify avenues for improvement of these services. The researchers felt that the most appropriate outcome measure for this research study was client satisfaction.

Research on client satisfaction with genetic counseling service is needed as the profession grows and becomes more integrated into health care. Genetic counselors with their expertise in molecular diagnostics are expected to play an increasingly important role in medicine, as genomics continues to become incorporated into health care. Ascertaining patient perceptions of genetic counseling services can help ensure that counseling is meeting the needs of patients and can identify areas for continued improvement.

While changes within the genetic counseling profession were part of the rationale for choosing client satisfaction for this study, the study population was an additional consideration. Huntington disease is a debilitating disorder, with psychiatric disturbances and diminished cognitive abilities being common features. Outcome measures requiring client information recall

after receiving genetic counseling or measures evaluating psychological impact of genetic counseling may be challenging in the HD population due to the characteristic features. However, assessing client satisfaction utilizing the instrumental and affect component of satisfaction was felt to be appropriate for this population in this study. The ease of obtaining satisfaction data is ideal in a population where diminishing mental capacity is of concern. Responding to questions in a survey that are targeted to the instrumental and affect components of satisfaction does not typically require high cognitive abilities. In essence, it requires the patient to reflect back to the genetic counseling services they have received and evaluate their feelings about these services. For these reasons, client satisfaction was felt to be an effective way of gaining patient perceptions of genetic counseling services in the UPMC HD Clinic.

As anticipated, the researchers discovered that patients are generally satisfied with the genetic counseling services in the HD Clinic. On a scale of 1-5, the mean satisfaction of the services was determined to be 4.59. These results compared similarly with other outcome studies utilizing client satisfaction. The stratification of the results of this study indicated that patients are generally satisfied with inherent characteristics of the counselor herself, as well as the counseling. This research also uncovered some potential areas for improvement. One participant commented that the setting of their genetic counseling was not comfortable. This may have decreased the participant's overall satisfaction with the services. The researchers recommend that genetic counselors pay particular attention to the setting where they are delivering services to ensure that the setting is comfortable for the patient. Additionally, responses to the survey elicited concerns that the genetic counselor was not sufficiently addressing the worries of the client. This raises opportunities for potential improvement of services. Genetic counselors may wish to pay particular attention during their counseling sessions to address worries/concerns that

the patient expresses. A way to achieve this could be to directly ask the patient to express any worries/concerns they have during contracting, an important component of the genetic counseling process. While there are limitations and challenges of generalizing the findings of this study due to the small study population chosen in a biased manner, these findings have local relevance of elucidating potential improvements of genetic counseling services in the HD clinic, which is important to ensure that the counseling in the clinic is meeting patient expectations. The findings from this study can also inform further research on this topic.

The information gathered from this research is of particular significance to the field of public health because genetic counseling is expected to play an important role in providing personalized and preventative health care. As O'Daniel notes (2010), "With expertise in genetic science, risk assessment and communication, and a patient-centered practice approach, genetic counselors are poised to play a critical role in facilitating the incorporation of genomic health risks into the burgeoning field of genome-guided preventative medicine." As the profession continues to grow and genome-based testing (Mills and Haga, 2013) expands, it is increasingly important to assess the provision of genetic counseling services to ensure that it is meeting the needs of patients. This study explored patient perspectives in one clinic setting and determined that the participants were satisfied with their genetic counseling experience. This research may inform outcome research in other settings.

APPENDIX A: IRB EXEMPTION LETTER

University of Pittsburgh
Institutional Review Board

3500 Fifth Avenue
Pittsburgh, PA 15213
(412) 383-1480
(412) 383-1508 (fax)
http://www.irb.pitt.edu

Memorandum

To: Alicia Martinez
From: IRB Office
Date: 10/20/2014
IRB#: PRO14090556
Subject: The Evaluation of Patient Satisfaction with Genetic Counseling Services Offered in a Huntington Disease Clinic

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The above-referenced project has been reviewed by the Institutional Review Board. Based on the information provided, this project meets all the necessary criteria for an exemption, and is hereby designated as "exempt" under section 45 CFR 46.101(b)(2).

Please note the following information:

- Investigators should consult with the IRB whenever questions arise about whether planned changes to an exempt study might alter the exempt status. Use the "**Send Comments to IRB Staff**" link displayed on study workspace to request a review to ensure it continues to meet the exempt category.
- It is important to close your study when finished by using the "**Study Completed**" link displayed on the study workspace.
- Exempt studies will be archived after 3 years unless you choose to extend the study. If your study is archived, you can continue conducting research activities as the IRB has made the determination that your project met one of the required exempt categories. The only caveat is that no changes can be made to the application. If a change is needed, you will need to submit a NEW Exempt application.

Please be advised that your research study may be audited periodically by the University of Pittsburgh Research Conduct and Compliance Office.

APPENDIX B: RECRUITMENT E-MAILS

B.1 INITIAL E-MAIL

Subject: **Request for Participation: Genetic Counseling Research Study in Huntington disease**

Greetings,

The Huntington Disease Clinic in Pittsburgh, Pennsylvania has been providing genetic services to patients and at-risk individuals for over 10 years. The purpose of this research study is to evaluate patient satisfaction of these genetic services, particularly the genetic counseling services. An example of these services may include those delivered by Betsy Gettig, and more recently Christa Lorenchick, at the Pittsburgh clinic. We will be surveying individuals who are followed at the University of Pittsburgh Medical Center Huntington Disease Clinic, or their family members or caregivers, and ask them to complete a brief (approximately 10 minute) questionnaire. If you are willing to participate, our questionnaire will ask about your feelings about the genetic counseling services that have been provided to you and some additional background information (age, gender, who is filling out this questionnaire, and reason for current/last visit). Please specify who is filling out this questionnaire as the person whose opinions will be reflected in the answers to the questions.

There are no foreseeable risks associated with this project, nor are there any direct benefits to you. This is an entirely anonymous questionnaire so your responses will not be identifiable in any way. All responses are confidential, and results will be kept under lock and key. Your participation is voluntary. This study is being conducted by Alicia Martinez, Genetic Counseling Master's Degree Student, who can be reached at aem99@pitt.edu, if you have any questions.

Please answer each question based on how you feel. You may check N/A if the question is not applicable to the services that you have received.

Please access this questionnaire through the following anonymous survey link:

https://pitt.co1.qualtrics.com/SE/?SID=SV_3rfbzzTbileWnyZ

Thank you for your time and participation in this research study.

B.2 REMINDER E-MAIL

Subject: **Request for Participation: Genetic Counseling Research Study in Huntington disease**

Greetings,

Please disregard this e-mail if you have already participated in the study by filling out the online questionnaire. This is just a friendly reminder for the opportunity to participate in a research study.

The Huntington Disease Clinic in Pittsburgh, Pennsylvania has been providing genetic services to patients and at-risk individuals for over 10 years. The purpose of this research study is to evaluate patient satisfaction of these genetic services, particularly the genetic counseling services. An example of these services may include those delivered by Betsy Gettig, and more recently Christa Lorenchick, at the Pittsburgh clinic. We will be surveying individuals who are followed at the University of Pittsburgh Medical Center Huntington Disease Clinic, or their family members or caregivers, and ask them to complete a brief (approximately 10 minute) questionnaire. If you are willing to participate, our questionnaire will ask about your feelings about the genetic counseling services that have been provided to you and some additional background information (age, gender, who is filling out this questionnaire, and reason for current/last visit). Please specify who is filling out this questionnaire as the person whose opinions will be reflected in the answers to the questions.

There are no foreseeable risks associated with this project, nor are there any direct benefits to you. This is an entirely anonymous questionnaire so your responses will not be identifiable in any way. All responses are confidential, and results will be kept under lock and key. Your participation is voluntary. This study is being conducted by Alicia Martinez, Genetic Counseling Master's Degree Student, who can be reached at aem99@pitt.edu, if you have any questions.

Please answer each question based on how you feel. You may check N/A if the question is not applicable to the services that you have received.

Please access this questionnaire through the following anonymous survey link:

https://pitt.co1.qualtrics.com/SE/?SID=SV_3rfbzzTbileWnyZ

Thank you for your time and participation in this research study.

APPENDIX C: QUESTIONNAIRE FINAL DRAFT

University of Pittsburgh

Department of Human Genetics

[redacted]

The Huntington Disease Clinic in Pittsburgh, Pennsylvania has been providing genetic services to patients and at-risk individuals for over 10 years. The purpose of this research study is to evaluate patient satisfaction of these genetic services, particularly the genetic counseling services. An example of these services may include those delivered by Betsy Gettig, and more recently Christa Lorenchick, at the Pittsburgh clinic. We will be surveying individuals who are followed at the University of Pittsburgh Medical Center Huntington Disease Clinic, or their family members or caregivers, and ask them to complete a brief (approximately 10 minute) questionnaire. If you are willing to participate, our questionnaire will ask about your feelings about the genetic counseling services that have been provided to you and some additional background information (age, gender, who is filling out this questionnaire, and reason for current/last visit). Please specify who is filling out this questionnaire as the person whose opinions will be reflected in the answers to the questions.

There are no foreseeable risks associated with this project, nor are there any direct benefits to you. This is an entirely anonymous questionnaire so your responses will not be identifiable in any way. All responses are confidential, and results will be kept under lock and key. Your participation is voluntary. This study is being conducted by Alicia Martincz, Genetic Counseling Master's Degree Student, who can be reached at aem99@pitt.edu, if you have any questions.

Please answer each question based on how you feel. You may check N/A if the question is not applicable to the services that you have received.

Age:_____

Gender:_____

Please check who is filling out this questionnaire:

☐ Patient (Individual with HD symptomatic/asymptomatic) ☐ Family Member (Individuals at-risk based on family history of HD) ☐ Caregiver

Please check the reason for your current/last visit:

☐ Presymptomatic testing ☐ Follow-up ☐ Other (specify)_____

1. The genetic counselor showed interest in your personal problems beyond what is medically required. For example, did she provide you with additional resources, etc?

☐ Strongly disagree ☐ Disagree ☐ Neither agree nor disagree
☐ Agree ☐ Strongly agree ☐ NA

2. I considered going to another genetic counselor regarding Huntington disease (HD).

☐ Strongly disagree ☐ Disagree ☐ Neither agree nor disagree
☐ Agree ☐ Strongly agree ☐ NA

3. The genetic counselor explained HD to you clearly.

☐ Strongly disagree ☐ Disagree ☐ Neither agree nor disagree
☐ Agree ☐ Strongly agree ☐ NA

4. The genetic counselor met your expectations of her/him.

☐ Strongly disagree ☐ Disagree ☐ Neither agree nor disagree
☐ Agree ☐ Strongly agree ☐ NA

5. You believe that the genetic counselor cares for you as a person.

☐ Strongly disagree ☐ Disagree ☐ Neither agree nor disagree
☐ Agree ☐ Strongly agree ☐ NA

6. You feel comfortable calling the genetic counselor to ask another question.

☐ Strongly disagree ☐ Disagree ☐ Neither agree nor disagree
☐ Agree ☐ Strongly agree ☐ NA

7. The genetic counselor reassured you.

☐ Strongly disagree ☐ Disagree ☐ Neither agree nor disagree
☐ Agree ☐ Strongly agree ☐ NA

8. The genetic counselor listened to what you had to say.

☐ Strongly disagree ☐ Disagree ☐ Neither agree nor disagree
☐ Agree ☐ Strongly agree ☐ NA

9. The genetic counselor was considerate of your emotional state during the meeting.

☐ Strongly disagree ☐ Disagree ☐ Neither agree nor disagree
☐ Agree ☐ Strongly agree ☐ NA

10. You were satisfied with the way in which information was transmitted to you.

☐ Strongly disagree ☐ Disagree ☐ Neither agree nor disagree
☐ Agree ☐ Strongly agree ☐ NA

11. The genetic counselor showed enough dedication in treating you, your family member, or the individual you are caring for with HD.

☐ Strongly disagree ☐ Disagree ☐ Neither agree nor disagree
☐ Agree ☐ Strongly agree ☐ NA

12. The genetic counselor understood what was bothering you (if anything).

☐ Strongly disagree ☐ Disagree ☐ Neither agree nor disagree
☐ Agree ☐ Strongly agree ☐ NA

13. I think I could have received more considerate care from another genetic counselor.

☐ Strongly disagree ☐ Disagree ☐ Neither agree nor disagree
☐ Agree ☐ Strongly agree ☐ NA

14. The genetic counselor made you feel you are "in good hands".

☐ Strongly disagree ☐ Disagree ☐ Neither agree nor disagree
☐ Agree ☐ Strongly agree ☐ NA

15. The genetic counselor made you feel that she/he knows how to handle HD.

☐ Strongly disagree ☐ Disagree ☐ Neither agree nor disagree
☐ Agree ☐ Strongly agree ☐ NA

16. The genetic counselor gave you enough of her/his time.

☐ Strongly disagree ☐ Disagree ☐ Neither agree nor disagree
☐ Agree ☐ Strongly agree ☐ NA

17. The genetic counselor was sensitive and tactful during your conversation.

☐ Strongly disagree ☐ Disagree ☐ Neither agree nor disagree
☐ Agree ☐ Strongly agree ☐ NA

18. The genetic counselor gave you the necessary treatment.

☐ Strongly disagree ☐ Disagree ☐ Neither agree nor disagree
☐ Agree ☐ Strongly agree ☐ NA

19. The genetic counselor is an expert in HD.

☐ Strongly disagree ☐ Disagree ☐ Neither agree nor disagree
☐ Agree ☐ Strongly agree ☐ NA

20. The counseling that you received helped you cope better with your situation.

☐ Strongly disagree ☐ Disagree ☐ Neither agree nor disagree
☐ Agree ☐ Strongly agree ☐ NA

21. The genetic counselor lessened your worries.

☐ Strongly disagree ☐ Disagree ☐ Neither agree nor disagree
☐ Agree ☐ Strongly agree ☐ NA

22. You are satisfied with the information you received in counseling.

☐ Strongly disagree ☐ Disagree ☐ Neither agree nor disagree
☐ Agree ☐ Strongly agree ☐ NA

23. You were comfortable to talk about yourself (or family member) during the counseling session.

☐ Strongly disagree ☐ Disagree ☐ Neither agree nor disagree
☐ Agree ☐ Strongly agree ☐ NA

24. I think I could have gotten better treatment from another genetic counselor.

☐ Strongly disagree ☐ Disagree ☐ Neither agree nor disagree
☐ Agree ☐ Strongly agree ☐ NA

25. You believe that the counseling was given in the appropriate setting.

☐ Strongly disagree ☐ Disagree ☐ Neither agree nor disagree
☐ Agree ☐ Strongly agree ☐ NA

26. Overall you are satisfied with the counseling you received.

☐ Strongly disagree ☐ Disagree ☐ Neither agree nor disagree
☐ Agree ☐ Strongly agree ☐ NA

Please comment on how satisfied you were with the genetic counseling services you received and what you were most satisfied with.

Please comment on your satisfaction with the genetic counselor who provided you with the genetic counseling services, i.e. what did you like most about the genetic counselor?

Questionnaire adopted from:

Shiloh S, Avdor O, Goodman R. Satisfaction with genetic counseling: dimensions and measurement.*Am J Med Genet*1990;37:522–9.

APPENDIX D: **Satisfaction with Genetic Counseling Instrument** Shiloh et al. (1990)

TABLE 1. Means and Standard Deviations of Satisfaction Subscores and Items (1 = low, 4 = high) in Genetic Counseling (n = 76) and Control (n = 56) Groups

Items	Genetic counseling group (mean ± SD)	Control group (mean ± SD)	t
Instrumental subscore	3.31 ± 0.44	3.43 ± 0.40	1.62
Affective subscore	3.33 ± 0.44	3.25 ± 0.44	1.02
Procedural subscore	3.04 ± 0.50	2.80 ± 0.62	2.37*
1. Did the doctor show interest in your personal problems beyond what is medically required?	2.86 ± 1.08	2.59 ± 1.34	1.27
2. Did you consider turning to another doctor regarding your problem?	2.68 ± 0.57	2.55 ± 0.68	1.19
3. Did the doctor explain your condition to you clearly?	3.27 ± 0.70	3.36 ± 0.75	0.70
4. Did the doctor meet your expectations of him?	3.02 ± 0.83	3.21 ± 0.76	1.34
5. Do you think the doctor cares for you as a person?	3.55 ± 0.64	3.50 ± 0.76	0.38
6. How comfortable would you feel to call the doctor to ask another question?	3.17 ± 0.67	3.07 ± 0.71	0.80
7. Did the doctor reassure you?	3.05 ± 0.84	3.36 ± 0.70	2.18*
8. Did the doctor listen to what you had to say?	3.69 ± 0.59	3.64 ± 0.55	0.45
9. Was the doctor considerate of your emotional state during the meeting?	3.16 ± 0.69	2.81 ± 0.97	2.27**
10. How satisfied are you with the way in which information was transmitted to you?	2.96 ± 0.84	3.21 ± 0.76	1.79
11. Did the doctor show enough dedication in treating your problem?	3.30 ± 0.84	3.64 ± 0.58	2.71**
12. How would you rate the level of service that you received?	3.42 ± 0.67	3.42 ± 0.66	0.04
13. Did the doctor understand what was really bothering you?	3.66 ± 0.55	3.50 ± 0.60	1.61
14. Do you think that you could get more considerate care from a private doctor?	3.30 ± 0.99	2.91 ± 1.07	2.16*
15. Did the doctor make you feel you are "in good hands"?	3.73 ± 0.55	3.70 ± 0.49	0.18
16. Did the doctor make you feel that he knows how to handle problems like yours?	3.34 ± 0.84	3.64 ± 0.55	2.53**
17. Did the doctor give you enough of his time?	3.53 ± 0.76	3.57 ± 0.71	0.35
18. Was the doctor sensitive and tactful during your conversation?	3.67 ± 0.30	3.66 ± 0.64	0.10
19. Did the doctor give you the necessary treatment?	3.07 ± 1.07	3.43 ± 0.95	1.98**
20. Do you think the doctor is an expert in the field in which you need help?	3.84 ± 0.44	3.69 ± 0.60	1.53
21. Can the counseling that you received help you cope better with your problem?	3.11 ± 0.79	3.48 ± 0.61	3.00**
22. Did the doctor lessen your worries?	2.94 ± 0.88	3.19 ± 0.83	1.57
23. How did you rate the length of time you waited since you first contacted the clinic and until your visit?	2.93 ± 0.83	2.66 ± 0.98	1.70
24. How satisfied are you with the information you got in counseling?	2.94 ± 0.71	3.27 ± 0.62	2.83**
25. How comfortable did you feel to talk about yourself during the counseling session?	3.17 ± 0.66	3.20 ± 0.52	0.26
26. How satisfied were you with the administrative procedures required for your visit (documents, lines, etc.)?	3.08 ± 0.66	3.00 ± 0.79	0.62
27. How do you rate the length of time you waited since your arrival at the clinic and until you entered the doctor's office?	2.79 ± 0.73	2.22 ± 1.03	3.54**
28. If an acquaintance of yours needed a similar help, would you recommend this clinic to him/her?	3.61 ± 0.54	3.33 ± 0.67	2.60**
29. Do you think you could get better treatment from a private doctor?	3.17 ± 0.97	2.87 ± 1.03	1.68
30. How satisfied are you with the treatment you got from the medical staff, besides for the doctor—the nurse, the secretary, etc.?	3.36 ± 0.58	3.26 ± 0.62	0.94
31. Do you think that the counseling was given in the appropriate setting for the sort of problem you had?	3.39 ± 0.75	3.47 ± 0.80	0.57
32. In summary, how would you rate your satisfaction with the counseling?	3.13 ± 0.70	3.40 ± 0.53	2.46**

*P < .05.
**P < .01.

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