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# Discussing History of Mental Illness in a General Genetic Counseling Setting: Patient and Caregiver Interest and Comfort

Alena Faulkner

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DISCUSSING HISTORY OF MENTAL ILLNESS IN A GENERAL GENETIC  
COUNSELING SETTING: PATIENT AND CAREGIVER INTEREST AND COMFORT

by

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Bachelor of Arts  
Sonoma State University, 2015

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Submitted in Partial Fulfillment of the Requirements

For the Degree of Master of Science in

Genetic Counseling

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University of South Carolina

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## DEDICATION

*“My mother was the rose: bright, bold and beautiful. It was her middle name, her favorite flower, her personality and her essence. She was active, she strove to reach her dreams, she stood out. This made her a great mom. A rose is lustrous and lively; my mom was forever optimistic and welcoming. She was full of warmth and fervor, and always smiled and conversed effusively. However, in life, every rose, every plant, every living thing no matter how brilliant and vivacious, must wither and come to an end.” – Adler Faulkner*

To my mom; this project would not have been imagined without the utmost admiration and inspiration I gained from witnessing your journey with Bipolar Disorder, amongst so much else. I wish you were physically here to fully appreciate the impact that your adversities continue to hold on some of my strongest passions and curiosities. Thank you for raising me to have a compassionate heart, and fueling me to be a champion for those living with mental illnesses. I will continue to carry your energy, ambition, and passion with me in my future endeavors, knowing that you will always be cheering me on.

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I have a tremendous amount of gratitude for my family and friends for continually cheering me on throughout each chapter of my life and lifting me up when I find myself in moments of uncertainty. I am so fortunate to be supported, loved, and encouraged by each of you.

Lastly, I am forever grateful for whatever super power brought my classmates and I together and allowed us to go through the past 21 months side by side. Thank you, thank you, thank you a million times over to each of you; collectively you have all taught me more than I ever imagined I would be leaving this experience with. I am so proud of each of you and honored to call you my colleagues.

## ABSTRACT

**Purpose:** This study explored patient, parent, and/or caregiver interest in and comfort with discussing personal and/or family history of mental illness (MI) with a genetics provider during a general genetics visit.

**Methods:** Participants were seen for initial genetic consultation through offices of the Greenwood Genetic Center (GGC) October 8<sup>th</sup>, 2018 through January 31<sup>st</sup>, 2019.

Following the genetics appointment, participants completed a 38-item questionnaire.

**Results:** Thirty participants completed or partially completed the questionnaire. Most participants had a child being evaluated ( $n=26$ , 87%). Overall, 26/29 participants (90%) indicated some degree of comfort with being directly or hypothetically asked about personal and/or family history of MI. Comfort did not seem to be dependent on positive or negative personal and/or family history of MI. For those who recalled a discussion about MI with the genetics provider ( $n = 10$ ), 90% indicated some degree of comfort with having this discussion. For those that did not discuss MI ( $n = 18$ ), 11 (61%) responded that they would be interested in discussing one or more of the provided mental health topics. Participant depression and anxiety severity measured by the PHQ-9 and GAD-7 scales, respectively, were statistically significantly different (increased) in this study's population compared to a standardized sample ( $p = .012$  and  $p = .0003$ ).

**Conclusion:** These results suggest that patients, parents, and/or caregivers are interested in and comfortable with discussing personal and/or family history of MI with a genetics provider during general genetic counseling. Of note, this sample consisting mostly of caretakers of minors with disabilities or suspected genetic conditions, reported

significantly more depressive and anxious symptoms compared to a general population. This result echoes previous research describing that caregivers of children with chronic conditions report more distress, stress, and worry, and poorer health outcomes and psychosocial well-being for themselves and their families when compared to control groups. This study demonstrates the importance of genetics providers addressing history of MI with patients during general genetics visits, not only to address the etiology of these conditions, but to identify individuals with psychiatric symptoms who could benefit from referral to further support services.

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# CHAPTER I

## BACKGROUND

### 1.1 INTRODUCTION

Approximately 4% of the population is affected in some capacity by mental illness (MI) (Thompson and Thompson Genetics in Medicine, 2016). In 2016, nearly 44 million adults in the United States were affected by MI, estimating about 18% of the American population (Center for Behavioral Health and Statistics and Quality, 2017). Schizophrenia, bipolar disorder, major depressive disorder, and schizoaffective disorder are understood to account for the majority of serious MI. It is estimated that the general population lifetime risk to develop schizophrenia is roughly 1%. The lifetime risk to develop other affective psychoses (e.g. manic-depressive psychosis, depression [including mild to severe], or bipolar disorder) is roughly 5%. (Harper, 2010). In addition to being rather common conditions, MI are also one of the most substantial causes of death worldwide and cause a significant amount of emotional and financial burden on affected individuals and their families (Walker et al. 2015). Like other common conditions such as diabetes, cardiovascular disease, and cancer, adoption and twin studies have shown high heritability for MI, implying complex etiology, involving interactions between both genetic and environmental factors (Hippman et al, 2016). Additionally, MI can be a feature of several genetic conditions in addition to physical symptoms.

### 1.2 GENETICS OF MENTAL ILLNESS

A considerable body of genetic research has been developed over the past several decades, utilizing twin and adoption studies, as well as family history information to

further characterize and understand the complex etiology of MI (Harper, 2010). Twin studies for serious MI such as schizophrenia and bipolar disorder have shown high concordance rates in monozygotic twins when compared to dizygotic twins, implying a strong familial and genetic basis (McGuffin et al., 1995). DNA studies have attempted to identify specific genetic loci involved in causation of MI. In 2014, a study completed by the Schizophrenia Working Group of the Psychiatric Genetics Consortium identified 108 genetic risk loci that reached genome-wide significance for schizophrenia, providing an etiologically relevant foundation for future mechanistic and treatment development studies (Schizophrenia Working Group of the Psychiatric Genomics Consortium, 2014). Similar studies have continued to support the understanding that while there are numerous genetic variants that contribute to one's susceptibility, vulnerability, or predisposition to MI, they are not single causes of these conditions, again highlighting the complex interplay between genetics and environmental experiences. Clearly, there is still much more research that can be done to have a more comprehensive understanding of the genetic variations that can contribute to the development of MI, how to interpret the clinical importance of the genetic risk loci that are associated with MI, and how the combined effects or interaction of multiple genetic variations contribute to vulnerability to MI.

Overall, individuals with psychiatric disorders are among the most highly stigmatized groups in society. Over the past few years, evidence has been accumulating that belief about illness causation, particularly regarding psychiatric disorders, strongly impacts quality of life and psychological adjustment in medical illness in general (Meiser et al., 2007). Meiser et al. assessed the potential impact that endorsing a genetic model, specifically in those with bipolar disorder or schizo-affective disorder has on perceived

stigma. There is debate on the possible impact of genetic attribution on the stigma associated with MI. For some, a genetic explanation can decrease stigma as it shifts causal responsibility away from the individual and towards the role of an uncontrollable biological cause, heredity, which in turn may alleviate blame, anger, and guilt, and increase sympathy and help in affected individuals. Genetic attribution may also lessen fear of individuals with a MI for those that are unaffected. However, a genetic explanation may give way to increased stigma by increasing perceptions of differentness and seriousness, creating an 'us and them' mentality (Meiser et al, 2007, Austin and Honer, 2004).

Phelan (2005), assessed the impact that geneticization or “the ascendancy of genetics as a basis for understanding human beings and human behavior” may have on the stigma associated with deviant behaviors and adopted an intermediate opinion between the two theories arguing that these theories might not be mutually exclusive, but operate simultaneously (Phelan, 2005). They also propose that for some, genetic attribution of MI may have little effect on stigma. Austin and Honer (2004) described that when attributing causes other than a genetic explanation to MI, such as stress or circumstance, it can lead to reduced social distance or decreased avoidance. Therefore, it seems that there is importance for individuals with MI and/or their relatives to understand that contributions of both genetics and the environment can cause a MI condition to manifest. Knowing this could be helpful for individuals with personal and/or family history of MI to avoid deterministic views and potentially aid in decreasing stigma (Austin and Honer, 2004).

In 2006, Austin et al. explored perceptions of genetic risk, associated effects on reproductive decisions, and attitudes towards genetic testing amongst unaffected relatives

of individuals with psychosis, concerned that “geneticization could lead to oversimplified ideas about genetic risk, producing significant social consequences”. Among participants, over-estimating risk was associated with reproductive decisions favoring fewer children, and more positive attitudes towards genetic testing. Although the chance to pass MI on to future generations is often overestimated in individuals that have a personal and/or family history of MI, there is *some* increased chance of recurrence for future offspring or relatives when an individual has a personal and/or family history of serious MI (Costain et al., 2012; Harper, 2010; Havinga et al., 2017). Currently, an increased risk of MI can be identified through detailed family history information. The empiric risk of developing a MI has been determined for many of these conditions and can be used as a general guide in clinical settings (Hill and Sahhar, 2006).

### 1.3 GENETIC COUNSELING AND MENTAL ILLNESS

As defined by the National Society of Genetic Counselors’ Task Force, genetic counseling is the process of helping people understand and adapt to the medical, psychological, and reproductive implications of the genetic contributions to disease (Resta et al. 2006). Genetic counselors are specialized healthcare professionals who deliver genetic counseling services by gathering information, assessing risk, educating, providing support and facilitating decision-making, and providing follow-up support and counseling to their patients (Hodgkinson et al., 2001). Genetic counseling has applications in many different areas, but arguably the best established and characterized of these would be: prenatal diagnosis of genetic conditions, pediatric and adult onset disorders exhibiting simple Mendelian inheritance patterns, and genetic syndromes (Austin and Honer, 2004). Although there are no clinically useful genetic tests available for establishing, refining, confirming, or excluding a psychiatric diagnosis, genetic

counseling for psychiatric conditions has shown promise for increasing patient knowledge, adaptation, positive self-identity, empowerment, accurate risk perception, along with improving quality of life for family members and improving overall prognosis for affected individuals (Jenkins and Arribas-Ayllon, 2016; Hippman et al, 2016; Austin and Honer, 2004).

In 2012, the first specialist psychiatric genetic counseling clinic of its kind opened in Canada. This specialty clinic aims to provide psychiatric genetic counseling services to individuals with non-syndromic psychiatric disorders and their families. Using data collected throughout the first year of providing psychiatric genetic counseling services, Inglis et al (2015) described uptake and impact of these services based on patient completion of clinical assessment tools, meant to measure patient empowerment and self-efficacy related to illness management, prior to and after genetic counseling sessions. Participants' primary reasons for attending psychiatric genetic counseling were to understand cause of MI, to understand the ways to protect mental health, and to understand the chances for recurrence of illness among other family members. Inglis and colleagues found that mean scores on the two clinical assessment tools increased significantly after genetic counseling, supporting that these specialized services are highly beneficial both to those with and without a personal history of MI, and adding to the growing body of literature describing the positive effects of genetic counseling for people with MI and their families.

In 2016, Jenkins and Arribas-Ayllon explored the potential value and uptake of psychiatric genetic counseling services in the United Kingdom (UK) by sampling from psychiatric health professionals. They found three relevant themes; first, that there was a demand for psychiatric genetic counseling, second, that there is a professional

responsibility for psychiatric genetic counseling provision, and last, given that psychiatric genetic counseling services are not currently being offered in the UK, there are barriers for referral of genetic counseling services. All participants agreed that a specialized psychiatric genetic counseling service would be needed in the future and most reported that affected individuals and their families would benefit from such a service, but that poor patient demand and predictability were factors hindering such a service from coming to fruition. Jenkins and Arribas-Ayllon's study was the first of its kind to explore healthcare professionals' accounts of psychiatric genetic counseling in the UK. Several other studies outside of the UK have explored patient perspectives on the value and outcomes of psychiatric genetic counseling.

In an additional study, Hippman et al. (2016) conducted the first pilot randomized clinical trial and evaluated the impact of genetic counseling for individuals with serious MI, as compared to receiving an educational intervention or waitlist. Participants completed outcome measures assessing knowledge, risk perception, internalized stigma, and perceived control over illness at baseline and at a one-month follow-up. They found that genetic counseling and the educational booklet improved knowledge and that genetic counseling, but not the educational booklet, improved risk perception accuracy in study participants. Qualitatively, they found that participants felt that genetic counseling was more useful than the educational booklets based on mean scores. They suggest that increasing knowledge and risk perception accuracy may play a role in empowering patients to make informed decisions about managing their mental health, and for some, may play a role in reproductive decision-making too.

#### 1.4 CAREGIVER STRAIN

Individuals with a family history of MI have an increased genetic susceptibility to develop MI and those with a personal history of MI have an increased chance of passing on more genetic susceptibility for MI to an offspring. Similarly, living with a lifelong or chronic genetic condition, especially one that significantly decreases quality of life, or caring for a child with a chronic genetic condition can add additional environmental factors that can contribute to vulnerability of experiencing symptoms of MI in the patient and/or caregiver. Several previous studies have shown examples of individuals in the pediatric and adult general populations that are affected by a genetic condition and in some capacity experience MI. Several studies have explored parental stress and caregiver strain in parents or guardians responsible for the care of a child with chronic genetic conditions such as syndromes associated with intellectual disability, cystic fibrosis, mitochondrial diseases, and Duchenne muscular dystrophy. These studies collectively describe that caregivers of children with chronic conditions report more symptoms of distress, increased levels of stress and worry, and poorer health outcomes and psychosocial well-being overall for both themselves and their families when compared to control groups (Cantwell, J et al. 2015; Besier T, et al. 2011; Senger, B, et al. 2015; Abi Daud, MS et al., 2004).

Cantwell et al. (2015) explored the synergistic relationship between stigma, self-esteem, and social support as predictors of depressive symptoms in parents of children with disabilities (e.g. Autism and Down syndrome). Through measuring perceived stigma, self-esteem, social support, and depressive symptoms in parents of children with disabilities and control parents, they found that parents of children with disabilities reported more depressive symptoms and that stigma, self-esteem, and social support were

associated with depressive symptomology. Their results highlight the necessity for specific support that aid in helping them with social support, self-esteem, and dealing with stigma.

Similarly, Besier et al (2011) assessed the prevalence of symptoms of anxiety and depression and the extent of life satisfaction in parents caring for children with cystic fibrosis. They enrolled 650 caregivers of 564 children with cystic fibrosis and had them complete standardized anxiety and depression scales. They found that more than one-third of parents showed elevated levels of anxious symptoms and that significantly more parents showed elevated levels of depressive symptoms compared to a community sample (28% versus 21%, respectively). Overall, these two elevated levels were associated with lower life satisfaction. They ultimately suggested that annual symptom screening is warranted to identify those at risk and therefore provide referrals and intervention for those in need.

### 1.5 LIVING WITH A GENETIC CONDITON

Other studies have shown that individuals living with certain genetic conditions, such as skeletal dysplasias or neurofibromatosis, experience a decreased quality of life when compared to controls, and can be at higher risk to manifest MI too, sometimes due to the progressive or uncertain nature of having a lifelong condition (Page, P et al., 2006, Apajasalo M et al., 1998, Jennings S et al. 2019). For example, using standardized questionnaires, Jennings et al. (2019) examined the prevalence of depression and anxiety in adults with skeletal dysplasias, and assessed any correlation with pain in individuals living with these conditions. Out of 336 usable survey responses which sampled a large variety of individuals in the skeletal dysplasia population, the study found that 16% of participants had scores consistent with current depression while 17% had scores

consistent with current anxiety, and that many participants (76%) experience pain. Additionally, the data described that 29% of individuals indicated a prior diagnosis of depression and 25% listed a prior anxiety diagnosis, with about 17% of individuals having a prior diagnosis of both. Their data suggested that there are likely a substantial number of individuals living with skeletal dysplasias with undiagnosed or undertreated MI. This prompted them to conclude that future studies should be concerned with investigating barriers to service or treatment of MI in this population. They also encourage education and open conversation to be fostered to reduce stigma surrounding MI in general.

Additional current literature has described individuals with genetic conditions, such as 22q deletion syndrome, in which MI can be a clinical manifestation associated with the genetic change. Approximately 25-30% of individuals with 22q deletion syndrome develop schizophrenia or other psychotic disorders (Morris E, et al. 2013). Individuals with other genetic conditions including, but not limited to, Huntington disease, certain metabolic conditions such as Niemann-Pick disease, Fragile X syndrome premutation carriers, and some sex chromosome aneuploidy conditions can experience MI as a part of these genetic differences too.

## 1.6 RATIONALE FOR CURRENT RESEARCH

Genetic counselors and medical geneticists see patients for a variety of indications. Common indications in a general genetic counseling visit include children with multiple birth defects, developmental delays, intellectual disabilities, autism, growth delays or overgrowth, suspicion of a known genetic condition, or presence of a familial genetic condition. Adults are commonly referred to a general genetics clinic to be evaluated by a genetic counselor and/or geneticist for similar reasons. Typically during a

general genetic counseling or general genetic evaluation visit, genetic counselors will ascertain information pertinent to finding a diagnosis for the child or adult, may recommend appropriate genetic testing, and may formulate a suitable management plan for the individual and their family. Part of this information involves gathering a family history, in which MI within the patient, the family, or the pediatric patient's caregiver can be a topic of discussion, especially if MI is a feature of a suspected or confirmed genetic condition.

As summarized by Inglis et al. (2017) in an article providing guidance to healthcare professionals seeking to provide prenatal psychiatric genetic counseling, the commonality of MI in the general population and the implicit relevance to many patients in the prenatal setting expresses the importance for clinicians to routinely ask their patients about personal and family history of MI to identify individuals who could benefit from psychiatric genetic counseling and further referrals and support. Similarly, MI is an important topic to be addressed in a general genetic counseling session due to the prevalence of caregiver burden present in the caregiver population and the degree that chronic medical conditions can take a toll on the quality of life and the mental health of a patient and/or caregiver, especially those that have a degenerative or progressive nature. Investigating patient comfort with and interest in discussing MI with a genetics professional in a general genetic clinic visit is important and has the potential to produce improved clinical practice implications for genetics providers.

As noted above, previous studies about psychiatric genetic counseling have shown benefits of directly addressing and discussing personal or family history of MI during a genetic counseling session. A recent master's thesis (Nimrichter, 2018) studied patient interest in and comfort with discussing MI in a prenatal genetic counseling

setting. Participants included prenatal patients referred to prenatal genetic counseling for routine prenatal screening or testing. This study found that most participants expressed interest in and comfort with discussing MI with a genetic counselor regardless of the presence of a family history, at 70% and 72%, respectively. Additionally, the absence or presence of depressive symptoms in the participant did not dictate interest level. The study suggested that discussion of family and personal history of MI is often welcome or well received by genetic counseling patients in the prenatal setting. Additional study in other genetic counseling patient populations are warranted to determine if similar themes emerge in other areas of genetic counseling (Nimrichter, 2018).

#### 1.7 PURPOSE AND HYPOTHESIS

To our knowledge, no previous studies have assessed patient interest in and comfort with discussing MI with a genetic counselor during a general genetics clinic visit. This study explored patient, parent, and/or caregiver interest in and comfort with discussing MI with a genetic counselor, medical geneticist, or other genetics provider (e.g. physician's assistant) during a general genetic counseling or genetics evaluation visit. The study hypothesizes that individuals with a personal and/or family history of MI will be more comfortable with discussing personal or family history of MI with a genetics provider than individuals who have no such personal or family history. It has the potential to elucidate what information patients would be interested in discussing as it relates to personal or family history of MI. This study also has the potential to emphasize the importance and positive impact of discussing MI with patients during a general genetics clinic appointment, adding to previous data showing that patients have significant interest in and comfort with discussing MI with a prenatal genetic counselor, with or without the presence of a personal or family history of MI. The study may show

that there are differences in the levels of comfort and needs of addressing MI between a general genetics population versus the previously studied prenatal genetics population. The results of this study can be compared to those in the aforementioned prenatal genetic study to determine if the recommendation that genetics professionals include a discussion of personal and/or family history of MI should be routinely addressed in the general genetics population.

CHAPTER II: MANUSCRIPT  
DISCUSSING HISTORY OF MENTAL ILLNESS IN A GENERAL GENETIC  
COUNSELING SETTING: PATIENT AND CAREGIVER INTEREST AND  
COMFORT<sup>1</sup>

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<sup>1</sup> Faulkner, A., Austin, J., Jones, A., & Vincent, V. To be submitted to *Journal of Genetic Counseling*.

## 2.1 ABSTRACT

**Purpose:** This study explored patient, parent, and/or caregiver interest in and comfort with discussing personal and/or family history of mental illness (MI) with a genetics provider during a general genetics visit.

**Methods:** Participants were seen for initial genetic consultation through offices of the Greenwood Genetic Center (GGC) October 8<sup>th</sup>, 2018 through January 31<sup>st</sup>, 2019.

Following the genetics appointment, participants completed a 38-item questionnaire.

**Results:** Thirty participants completed or partially completed the questionnaire. Most participants had a child being evaluated ( $n=26$ , 87%). Overall, 26/29 participants (90%) indicated some degree of comfort with being directly or hypothetically asked about personal and/or family history of MI. Comfort did not seem to be dependent on positive or negative personal and/or family history of MI. For those who recalled a discussion about MI with the genetics provider ( $n = 10$ ), 90% indicated some degree of comfort with having this discussion. For those that did not discuss MI ( $n = 18$ ), 11 (61%) responded that they would be interested in discussing one or more of the provided mental health topics. Participant depression and anxiety severity measured by the PHQ-9 and GAD-7 scales, respectively, were statistically significantly different (increased) in this study's population compared to a standardized sample ( $p = .012$  and  $p = .0003$ ).

**Conclusion:** These results suggest that patients, parents, and/or caregivers are interested in and comfortable with discussing personal and/or family history of MI with a genetics provider during general genetic counseling. Of note, this sample consisting mostly of caretakers of minors with disabilities or suspected genetic conditions, reported significantly more depressive and anxious symptoms compared to a general population. This result echoes previous research describing that caregivers of children with chronic

conditions report more distress, stress, and worry, and poorer health outcomes and psychosocial well-being for themselves and their families when compared to control groups. This study demonstrates the importance of genetics providers addressing history of MI with patients during general genetics visits, not only to address the etiology of these conditions, but to identify individuals with psychiatric symptoms who could benefit from referral to further support services.

## 2.2 INTRODUCTION

In 2016, nearly 44 million adults or 18% of the population in the United States were affected by mental illness (MI) (Center for Behavioral Health and Statistics and Quality, 2017). Schizophrenia, bipolar disorder, major depressive disorder, and schizoaffective disorder are understood to account for the majority of serious MI. Similar to other common conditions such as diabetes, cardiovascular disease, and cancer, adoption and twin studies have shown high heritability for MI, implying complex etiology involving interactions between both genetic and environmental factors (Hippman et al, 2016). Additionally, MI can be a feature of several genetic conditions in addition to physical symptoms. Although there are no clinically useful genetic tests available for establishing, refining, confirming, or excluding a psychiatric diagnosis, genetic counseling for MI has shown promise for increasing patient knowledge, adaptation, positive self-identity, empowerment, and accurate risk perception along with improving quality of life for family members and improving overall prognosis for affected individuals (Jenkins and Arribas-Ayllon, 2016; Hippman et al, 2016; Austin and Honer, 2004). Previous studies have shown that individuals with personal and/or family history of MI are interested in psychiatric genetic counseling. Using data collected throughout the first year of providing psychiatric genetic counseling services, Inglis et al

(2015) described that this specialty service increased patient empowerment and self-efficacy, supporting that these services are highly beneficial to both those with and without a personal history of MI.

Individuals with a family history of MI have an increased genetic susceptibility to develop MI and those with a personal history of MI have an increased chance of passing on more genetic susceptibility for MI to an offspring. Similarly, living with a lifelong or chronic genetic condition, especially one that significantly decreases quality of life, or caring for a child with a chronic genetic condition can add additional environmental factors that can contribute to vulnerability of experiencing symptoms of MI in the patients and or caregivers. Several previous studies have shown examples of individuals in the pediatric and adult general populations that are affected by a genetic condition and in some capacity, experience MI. For example, studies have shown that individuals living with certain genetic conditions, such as skeletal dysplasias or neurofibromatosis can be at higher risk to manifest MI, sometimes in relation to the progressive or uncertain nature of having a lifelong condition (Page, P et al., 2006, Apajasalo M et al., 1998, Jennings S et al. 2019).

Additionally, several studies have explored parental stress and caregiver strain in parents or guardians responsible for the care of a child with chronic genetic conditions like syndromes associated with intellectual disability, cystic fibrosis, mitochondrial diseases, and Duchenne muscular dystrophy. These studies collectively describe that caregivers of children with chronic conditions report higher psychological symptoms, increased levels of stress and worry, and poorer health outcomes and psychosocial well-being overall for both themselves and their families, when compared to control groups (Cantwell, J et al. 2015; Besier T, et al. 2011; Senger, B, et al. 2015; Abi Daud, MS et al.,

2004). For these reasons, the caregiver, parent, and adult patient population may benefit from exploring their comfort with and interest in discussing MI in a general genetic counseling setting.

Genetic counselors and medical geneticists see patients for a variety of indications. Common indications in a general genetic counseling setting include children with multiple birth defects, developmental delays, intellectual disabilities, autism, growth delays or overgrowth, suspicion of a known genetic condition, or presence of a familial genetic condition. Adults are commonly referred to a general genetics clinic to be evaluated by a genetic counselor and/or geneticist for similar reasons. Typically during a general genetic counseling or general genetic evaluation visit, genetic counselors will ascertain information pertinent to finding a diagnosis for the child or adult, may recommend appropriate genetic testing, and may formulate a suitable management plan for the individual and their family. Part of this information involves gathering a family history, in which MI within the patient, the family, or the pediatric patient's caregiver can be a topic of discussion, especially if MI is a feature of a suspected or confirmed genetic condition.

This study explored patient's and/or their caregiver's interest in and comfort with discussing MI with a genetic counselor, medical geneticist, or other genetics provider (e.g. physician's assistant) during a general genetic counseling or genetics evaluation visit. The study hypothesizes that individuals with a personal and/or family history of MI will be more comfortable with discussing personal or family history of MI with a genetics provider than individuals who have no such personal or family history.

## 2.3 MATERIALS AND METHODS

### 2.3.1 *Participants and Study Design*

The Institutional Review Boards at the University of South Carolina and Self Regional Healthcare approved this study for data collection during the four-month period (October 2018 through January 2019). Study participants consisted of patients, parents, and/or caregivers referred for initial genetic evaluation or consultation through offices of the Greenwood Genetic Center (GGC). Patients were referred for a variety of indications, for example, to confirm a suspected diagnosis, to evaluate symptoms, or to determine if they carry a familial variant. At this appointment, a genetic counselor, medical geneticist, or genetics provider (e.g. physician's assistant) traditionally takes a comprehensive pregnancy, developmental, and family history, performs a physical examination, provides counseling or education regarding genetic conditions and/or testing options, and obtains informed consent for any testing.

Eligibility requirements for this study were as follows:

- participants must be patients or the parents, legal guardians, or primary caregivers of minor patients who were seen for an initial consultation at one of the Greenwood Genetic Center clinics,
- participants must 18 years or older,
- participants must be English-speaking and able to read English, and
- participants must have received general genetic counseling, during which a detailed family history was obtained or reviewed.

Exclusion criteria were as follows:

- participants less than 18 years old,
- participants who do not speak English and are unable to read English,

- participants seen at the Greenwood Genetic Center for a follow-up visit during which a family history was not reviewed,
- participants seen at the Greenwood Genetic Center for a cancer genetic counseling indication, and
- participants who received a devastating diagnosis at the visit (e.g. a condition with a progressive nature, or a condition which shortens life expectancy), to avoid the potential risk of increased emotional stress.

### *2.3.2 Procedure*

Recruitment and data collection for this study was conducted through the Greenwood Genetic Center at three general genetics clinics in Columbia, SC, Greenwood, SC, and Greenville, SC. Participants were recruited through each of these three centers during or after their visit with a genetic counselor, medical geneticist, and/or genetics provider.

Potential participants were given a paper copy of the invitation to participate, questionnaire, raffle drawing entry sheet, and mental health resource page. These items were provided by the genetic counselor, medical geneticist, or genetics provider who performed their genetic counseling or genetic evaluation (materials can be found in Appendix A). Greenwood Genetic Center genetic counselors, medical geneticists, or genetics providers distributed questionnaires to their patients either while the counselor or genetics provider stepped out of the room mid-visit to consult the geneticist, or immediately following the patient's visit. The invitation stated the option to complete the questionnaire online through SurveyMonkey.com or by the provided hard copy. Upon completion of the paper copy, the questionnaire could be returned to the front desk of a Greenwood Genetic Center waiting room in the folder provided or by a self-addressed

envelope in the mail. Completed questionnaires were collected by the principal investigator (PI) and analyzed after the data collection was complete.

Participants were informed that by beginning and submitting the questionnaire, they were agreeing and consenting to be a participant in the study, and that their care at the Greenwood Genetic Center would not be affected by choosing to participate in the study or not. In the invitation letter, they were informed that all responses gathered from the questionnaire were kept anonymous and confidential and that the results of this study may be published or presented at academic meetings; however, participants would not be identified. They were further informed that their participation in this research is voluntary. Participants also were made aware that some questions in the questionnaire may make them feel uncomfortable and that they do not have to answer any of the questions that make them feel uncomfortable.

In order to increase recruitment, individuals could voluntarily participate in a monthly drawing for a gift card upon completion of the survey. The gift cards each valued \$20, and were from popular restaurants, grocery stores, or online retail sites. A winner was chosen once a month between October 2018 and January 2019, for a total of four winners throughout the data collection period. The PI contacted each winner by email and requested a physical address from winners to mail them a gift card. Email and physical addresses were destroyed upon completion of this study.

### *2.3.3 Methodology/Instrumentation*

This study used survey methodology. A purpose-designed questionnaire was adapted from a previous master's thesis with the intent to assess participants' comfort with being asked about or discussing MI in a general genetic counseling setting, and to assess interest in discussing these conditions (Nimrichter, 2018). The purpose-designed

questionnaire consisted of 38 total items. Parts of the questionnaire used skip logic. As part of the 38-item questionnaire, there were eight demographic questions to establish who was referred for the genetics appointment, which clinic the participant was seen at, the reason for the patient's visit, age, gender, ethnicity, and highest degree or level of education completed.

The nine-item Patient Health Questionnaire (PHQ-9), a validated depression severity screening tool, was administered as part of the 38-item questionnaire ( $\alpha = .82$ ). The PHQ-9 asks participants to check the answer that comes closest to how they have felt in the past two weeks. Answers are scored 0-3, with 0 being not at all, to 3 being nearly every day. Scores are tallied for a total score out of 27. A score of zero indicates no depression severity. Scores between 1 to 4 indicate minimal depression severity, scores between 5 to 9 indicate mild depression severity, scores between 10 to 14 indicate moderate depression severity, scores between 15 to 19 indicate moderately severe depression severity, and scores between 20 to 27 indicate severe depression severity (Kroenke et al., 2001).

The seven-item Generalized Anxiety Disorder Scale (GAD-7), a validated generalized anxiety disorder screening tool, was also administered as part of the 38-item questionnaire ( $\alpha = .90$ ). Like the PHQ-9, the GAD-7 asks participants to check the answer that comes the closest to how they have felt in the past two weeks and answers are scored to indicate anxiety severity. A score of zero indicates no anxiety severity. Scores between 1 to 4 indicate minimal anxiety severity, scores between 5 to 9 indicate mild anxiety severity, scores between 10 to 14 indicate moderate anxiety severity, and scores between 15 to 21 indicate severe anxiety severity (Spitzer RL, et al. 2006).

### *2.3.4 Data Analysis*

Data collection continued from October 2018 through the end of January 2019. Quantitative data analysis was performed using SPSS statistical analysis software. Descriptive and univariate statistics were utilized to investigate study hypotheses. A probability value of 0.05 ( $p = 0.05$ ) was used.

## 2.4 RESULTS

A total of 30 participants completed or partially completed the questionnaire either online or by paper copy. Three participants completed the questionnaire online and 27 by paper copy. All 30 participant's questionnaires were considered in reporting the results. The number of participants varied per question.

### *2.4.1 Demographics*

A summary of the participant's demographics can be found in Table 2.1. All participants indicated the nature of their relationship with the individual that was referred for the genetics appointment. Most participants had a child (biological or adopted) seen at the Greenwood Genetic Center ( $n=26, 87\%$ ). One adult patient participant completed a questionnaire and three participants selected "other". Of the participants who indicated "other", one responded that the genetics appointment was for a child who was a blood relative of theirs, but that they were not the child's parent or legal guardian.

All participants indicated the location of the clinic where they were seen. Most participants were seen at the Greenville location ( $n=17, 57\%$ ), seven were seen at the Columbia location, and six were seen at the Greenwood location.

Twenty-six out of the 30 participants responded to the reason for their or the child's visit. Responses varied from family history of a genetic condition, to determining if their child's symptoms were indicative of a genetic condition, to further genetic

evaluation after a formal diagnosis of autism. All responses were common referral indications for evaluation at a general or pediatric genetics clinic. Four participants did not complete this question.

Table 2.1 Demographic Characteristics of Participants

<b>Participant Characteristics</b>	<b>n</b>	<b>%</b>
<b>Individual Referred for Genetics Appointment</b>	30	
Self	1	3
Child (biological or adopted)	26	87
Other	3	10
<b>Clinic Location</b>	30	
Greenville, SC	17	57
Columbia, SC	7	23
Greenwood, SC	6	20
<b>Personal Identity</b>	30	
Cisgender female	28	93
Cisgender male	2	7
<b>Race</b>	30	
Black or African American	7	23
Hispanic or Latino	1	3
White	22	73
<b>Highest Degree or Level of Education Completed</b>	30	
Less than High School	1	3
High School Graduate or GED	8	27
Some college, no degree	14	47
Associate's Degree	1	3
Bachelor's Degree	3	10
Master's Degree	3	10

All participants provided their age. The age of the participants ranged from 20 to 74 years, with the mean age being 36.90 years (SD = 12.11). All participants indicated their biological sex and gender identity. The majority were cisgender female (93%). Two participants were cisgender male. All participants indicated their ethnicities with the majority describing themselves as white (n = 22, 73%). Seven participants described themselves as African-American, and one described himself or herself as Hispanic or Latino. All participants indicated their highest degree or level of education completed. The most common response was some college, no degree (n = 14, 47%), followed by high school graduate or equivalent (e.g. GED) (n = 8, 27%). One participant indicated they had less than a high school education, one participant had an associate's degree, three participants had a bachelor's degree, and three participants had a master's degree. No participants had a degree beyond a master's education.

#### *2.4.2 Directly Asked About Mental Illness and Associated Comfort*

All participants designated whether the genetics provider asked them directly about a personal and/or family history of MI, as illustrated in Figure 2.1. Twenty out of 30 (67%) indicated that the genetics provider directly asked about a personal and/or family history of MI. Of these participants, the majority (n = 18, 90%) indicated being comfortable with being asked. One participant indicated they felt neutrally about being asked. Only one participant indicated that they were very uncomfortable being directly asked about a personal and/or family history of MI by the genetics provider.

Ten participants indicated they were not directly asked about personal and/or family history of MI (n = 8) or were unsure if they were directly asked (n = 2). Of these combined ten participants, nine indicated hypothetical comfort with being directly asked about personal and/or family history of MI by the genetics provider and one participant

did not complete this question. None of the participants in this group indicated they would have had any level of discomfort had they been directly asked about personal and/family history of MI. Hypothetical comfort levels within this group are shown in Figure 2.1.

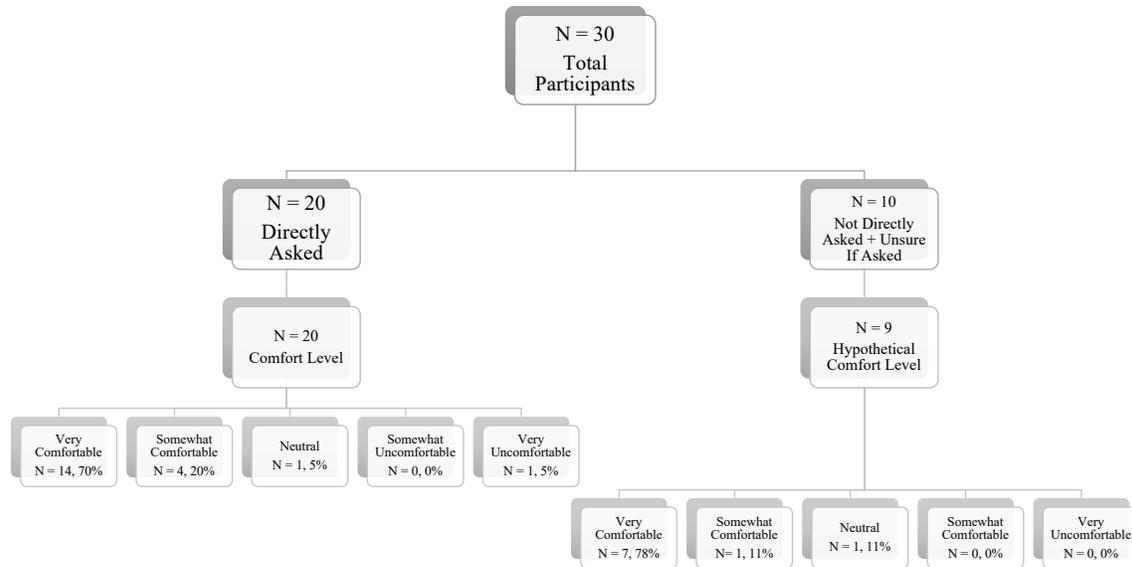


Figure 2.1 Directly Asked about Personal and/or Family History of Mental Illness and Associated Comfort

Overall, 26/29 participants (90%) indicated some degree of comfort with being directly or hypothetically asked about personal and/or family history of MI, with the majority (n = 21/29, 72%) indicating very comfortable. Only one participant indicated that they were uncomfortable with being asked. Overall participant comfort level associated with a genetics provider directly or hypothetically asking about a personal and/or family history of MI is summarized in Figure 2.2 (n = 29).

### 2.4.3 Discussion of Mental Illness during General Genetic Counseling

#### 2.4.3.1 Participants Who Had a Discussion

Out of the 30 total participants, 20 were directly asked about personal and/or family history of MI. Out of those 20 participants who indicated the genetics provider directly asked about history of MI, ten participants (50%) had a follow-up discussion

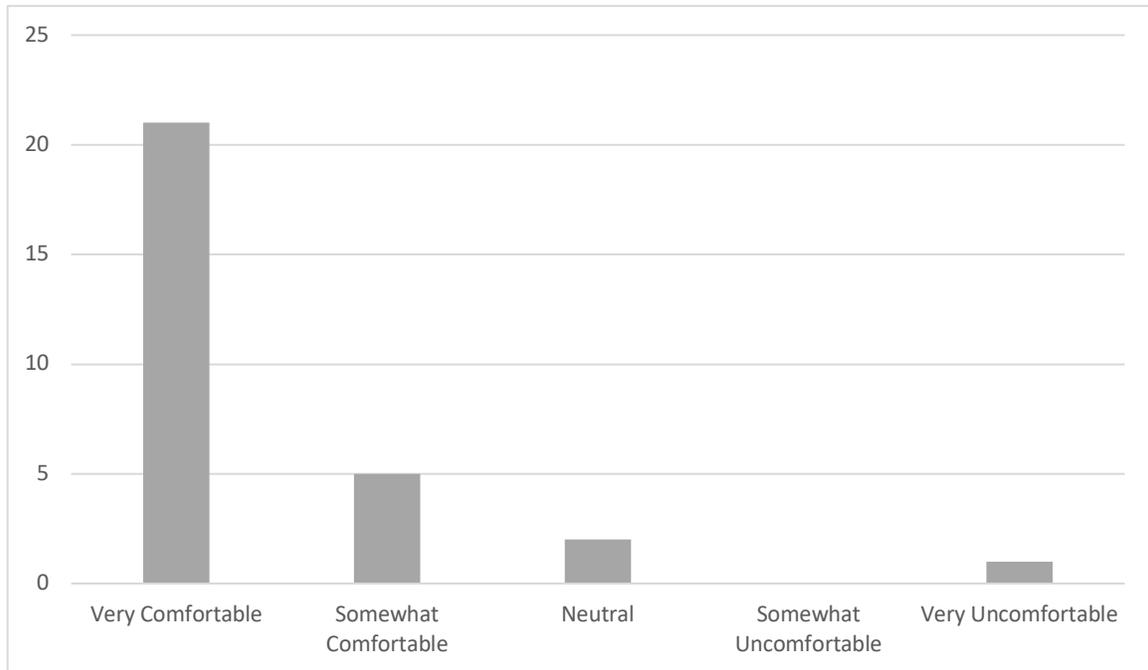


Figure 2.2 Overall Participant Comfort Level Associated with a Genetics Provider Directly or Hypothetically Asking about a Personal and/or Family History of Mental Illness (n = 29)

about mental health topics with the genetics provider. The most common mental health topic discussed was genetic testing (n = 7/10), followed by factors that can cause MI (n = 5/10), chance for family members to develop MI (n = 4/10), resources (e.g. information about mental health professionals) (n = 2/10), and “other” (n = 2/10). One participant who checked “other” indicated that they also discussed their postpartum depression in addition to discussing the MI in the family. A summary of the mental health topics that were discussed are illustrated in Figure 2.3 (n = 10).

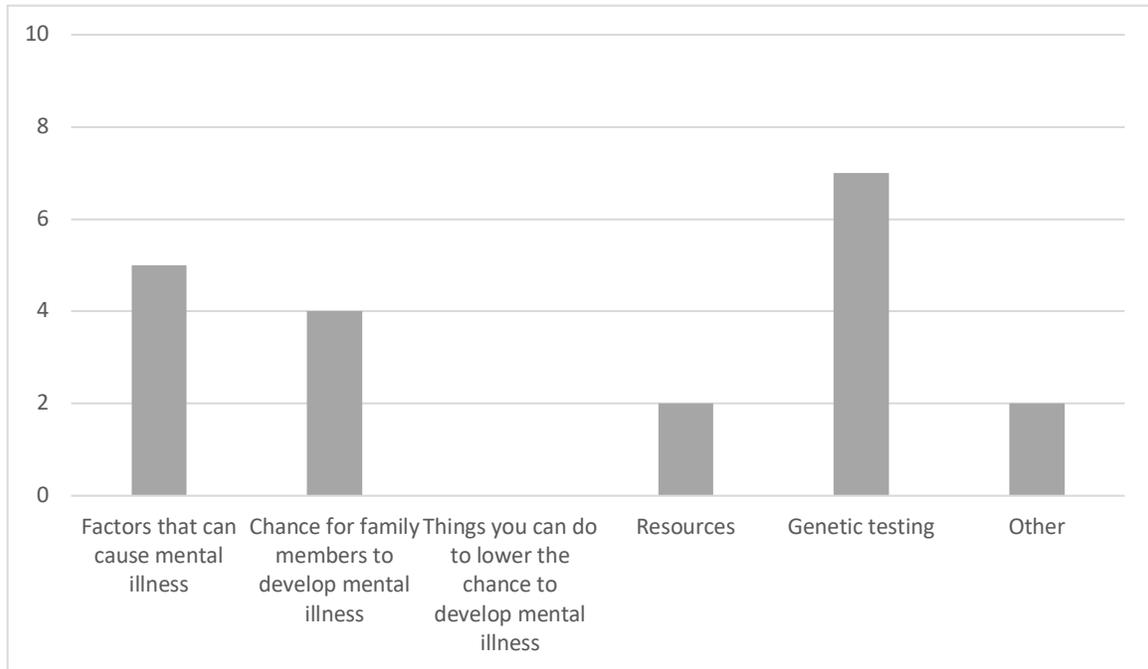


Figure 2.3 Mental Health Topics Discussed (n = 10)

Comfort level associated with discussing MI with a genetics provider was assessed. Of the ten respondents that had a discussion during their genetics appointment, the majority (n = 8, 80%) were very comfortable with having a discussion, and overall, nine (90%) indicated some degree of comfort. One participant indicated that they were very uncomfortable with having this discussion. This participant indicated that the genetics provider discussed genetic testing associated with MI with them and that there were no other topics that they would have liked to discuss. Comfort level associated with discussing MI with a genetics provider is illustrated in Figure 2.4 (n = 10).

The ten participants that discussed MI during the genetic counseling session answered a survey question about other topics they were interested in discussing that the genetics provider did not talk about. Eight (80%) indicated there were no other topics that they would have liked covered, two indicated interest in discussing mental health topics that the genetics provider did not talk about, and one participant that indicated further

interest in discussing factors that can cause MI. One participant indicated interest in discussing all mental health topics listed, although they had also indicated that they had discussed all topics with the genetics provider in their discussion about MI during the genetic counseling session.

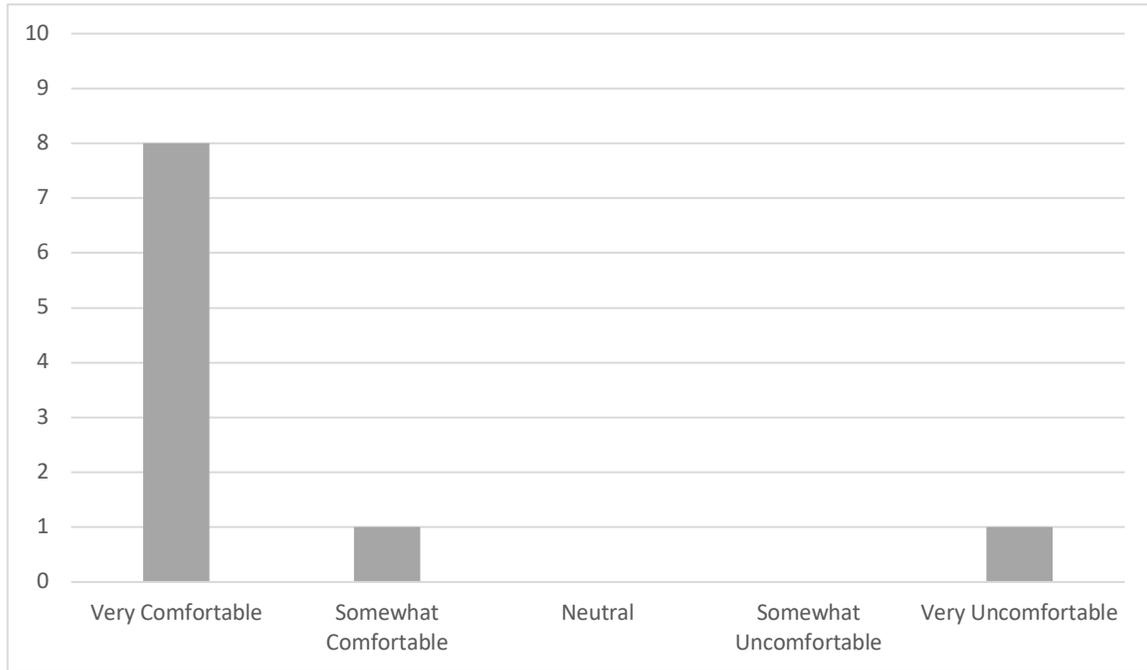


Figure 2.4 Participant Comfort Level Associated with Discussion about Mental Illness with a Genetics Provider (n = 10)

#### 2.4.3.2 Participants Who Did Not Have a Discussion

Twenty participants did not discuss MI during the genetics appointment. These participants were either not directly asked about history of MI in the first place, were unsure if they were asked, or there was no follow-up discussion after they were asked if they had a personal and/or family history of MI. Eighteen of the 20 participants (90%) indicated their interest in discussing topics related to MI and two did not respond to this question, illustrated in Table 2.2 (n = 18). In this group of 18 participants that did not have a discussion about mental health topics and indicated interest, 11 (61%) responded that they would be interested in discussing one or more of the following topics: factors

Table 2.2 Participant Interest in Discussing Mental Health Topics (n = 18)

<b>Mental Health Topic</b>	<b>n</b>
Factors that can cause MI	6
Chance for family members to develop MI	6
Things you can do to lower the chance to develop MI	1
Resources (e.g. information about mental health professionals)	0
Genetic testing	5
Not applicable (e.g. no family or personal history of MI)	4
None – I would not want to discuss this with the genetics provider	4

that cause MI, the chance for family members to develop MI, genetic testing, and things you can do to lower the chance to develop MI (e.g. exercise, adequate sleep, etc.). Four participants indicated that they would not want to discuss MI with the genetics provider. Another four participants indicated that discussing MI with the genetics provider was not applicable to them due to not having a family or personal history of MI. One of the participants who indicated that discussing MI was not applicable to them also indicated that they would be interested in discussing factors that cause MI and things you can do to lower the chance to develop MI. No participants indicated interest in discussing resources (e.g. information about mental health professionals).

#### *2.4.4 Personal History of Mental Illness*

Participants were asked if they had a mental health diagnosis. Thirteen out of 30 participants (43%) indicated a diagnosis of MI including, anxiety, depression, Postpartum Depression, Bipolar Disorder, Post-Traumatic Stress Disorder (PTSD), and Schizophrenia. Seven participants indicated that they have never had a mental health diagnosis, but suspect that they have a MI. Two-thirds ( $n = 20/30$ , 67%) indicated a positive personal history of MI (diagnosed or suspected).

Figure 2.6 illustrates the breakdown of participant personal history of MI and whether the participants in each group were directly asked about personal and/or family history of MI or not, or if they discussed mental health topics or not. Of the 20 participants with a personal history of MI (diagnosed or suspected), seven were directly asked about personal and/or family history of MI and had a follow up discussion, five were directly asked about personal and/or family history of MI and without a follow up discussion, and eight participants were not directly asked or were unsure if they were they were directly asked and therefore did not have a follow up discussion.

Of the ten participants with no reported personal history of MI or those that were unsure if they have ever had a diagnosis of MI, three were directly asked about personal and/or family history of MI and had a follow up discussion, five were directly asked about personal and/or family history of MI and did not have a follow up discussion, and two participants were not directly asked or were unsure if they were directly asked and therefore did not have a follow up discussion.

##### *2.4.4.1 Personal History of Mental Illness and Comfort*

Overall, participant comfort level did not seem to be dependent on positive or negative personal history of MI, as most participants within each group reported some

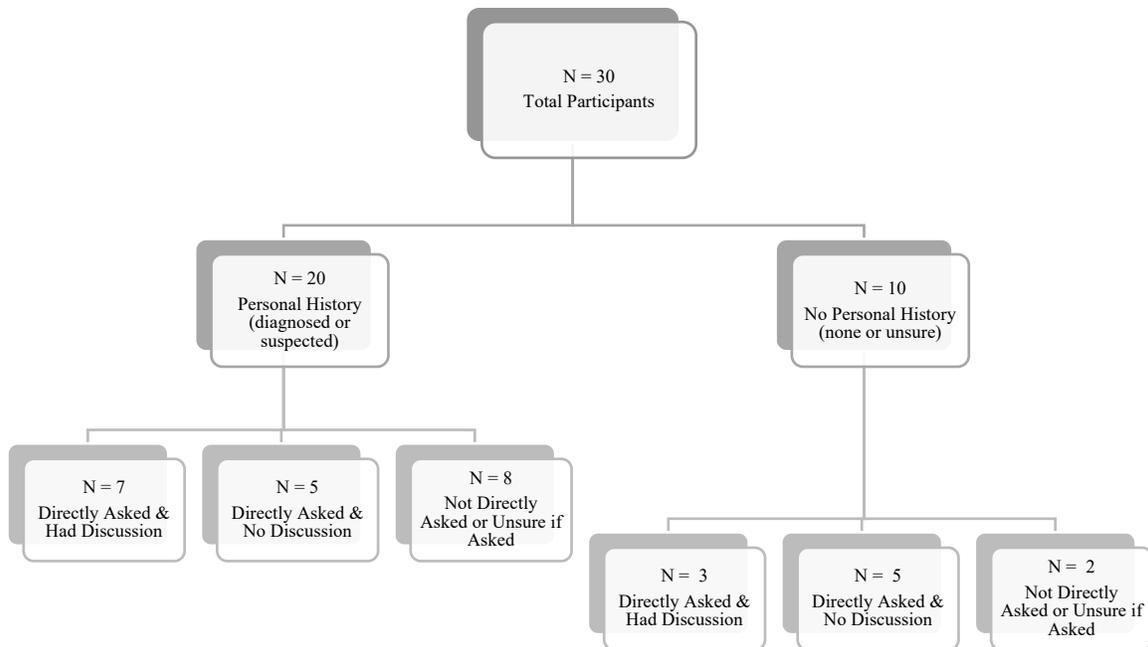


Figure 2.5 Participant Personal History of Mental Illness

degree of comfort with being directly asked and having a follow up discussion about MI (see Figure 2.7). Eighteen of the 20 participants (90%) who indicated a personal history of MI indicated that they were or would be very or somewhat comfortable with being asked about personal and/or family history of MI. Eight of the nine participants (89%) who indicated no personal history of MI and indicated comfort said that they were or would be very or somewhat comfortable with being asked about personal and/or family history of MI. One participant with no personal history did not indicate comfort or hypothetical comfort associated with being asked about a history of MI.

Of the ten participants who had a follow-up discussion regarding mental health topics with a genetics provider, seven participants indicated that they had a positive personal history (diagnosed or suspected) and three had a negative personal history of MI. Of the seven participants with a personal history of MI, none indicated any discomfort with discussing mental health topics. Of the three participants with no

personal history of MI, two indicated being very comfortable and one indicated being very uncomfortable with discussing mental health topics.

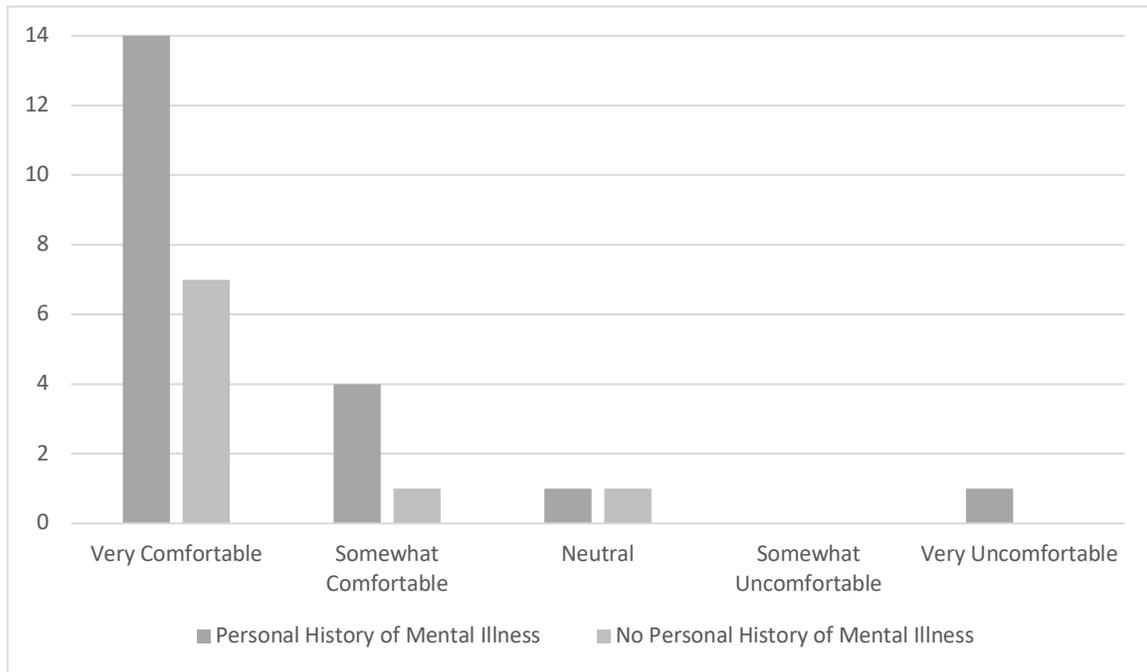


Figure 2.6 Personal History of Mental Illness and Comfort/Hypothetical Comfort with Being Directly Asked About Mental Illness

#### 2.4.5 Family History of Mental Illness

All participants indicated whether or not they had a family history of MI. The majority of participants (n = 19, 63%) indicated that they had a close family member with a diagnosis of MI including, depression, Post-Traumatic Stress Disorder (PTSD), Schizophrenia, Bipolar Disorder, anxiety, Obsessive Compulsive Disorder (OCD), Oppositional Defiant Disorder (ODD), eating disorders, Postpartum Depression, Postpartum Anxiety, and Borderline Personality Disorder (BPD). Five participants indicated that they suspected that a family member might have a MI. Together, twenty-four participants (80%) indicated a positive family history of MI (diagnosed or suspected). Five participants indicated that they did not have a close family member with MI, and one indicated that they were unsure.

Figure 2.8 illustrates the breakdown of participant family history of MI and whether the participants in each group were directly asked about personal and/or family history of MI or not, or if they discussed mental health topics or not. Of the 24 participants with a family history of MI (diagnosed or suspected), one-third recalled being asked and having a discussion about MI with the genetics provider, one-third were asked about personal and/or family history of MI but did not recall having a follow up discussion about MI with the provider, and one-third, did not recall the topic of MI being raised with the genetics provider.

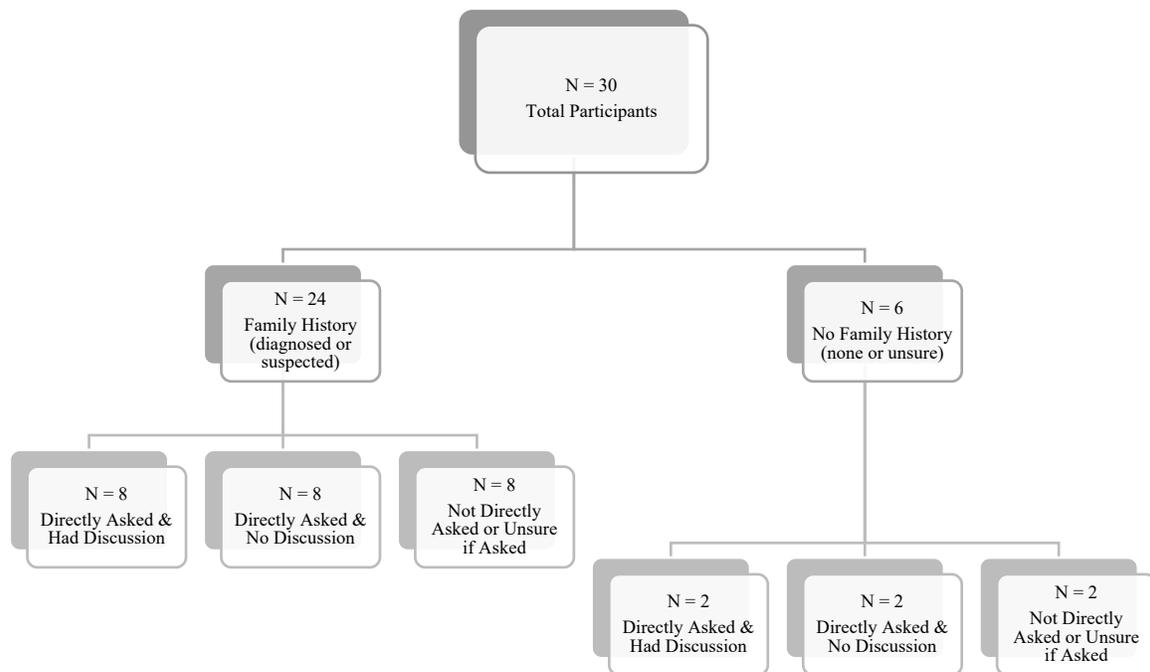


Figure 2.7 Participant Family History of Mental Illness

#### 2.4.5.1 Family History of Mental Illness and Comfort

Overall, participant comfort level did not seem to be dependent on positive or negative family history of MI, as the majority of participants within each group reported being very comfortable and/or somewhat comfortable with being directly asked and having a follow up discussion about MI. Comfort associated with being directly asked or

hypothetically being asked and participant family history of MI is illustrated in Figure 2.9 (n = 29). Twenty-two of the 24 participants (92%) who indicated a family history of MI indicated that they were or would be very or somewhat comfortable with being asked about personal and/or family history of MI. Only one participant with a positive family history of MI indicated discomfort with being directly asked about personal/and or family history of MI and only one participant with no family history of MI indicated they were not comfortable discussing MI with the genetics provider.

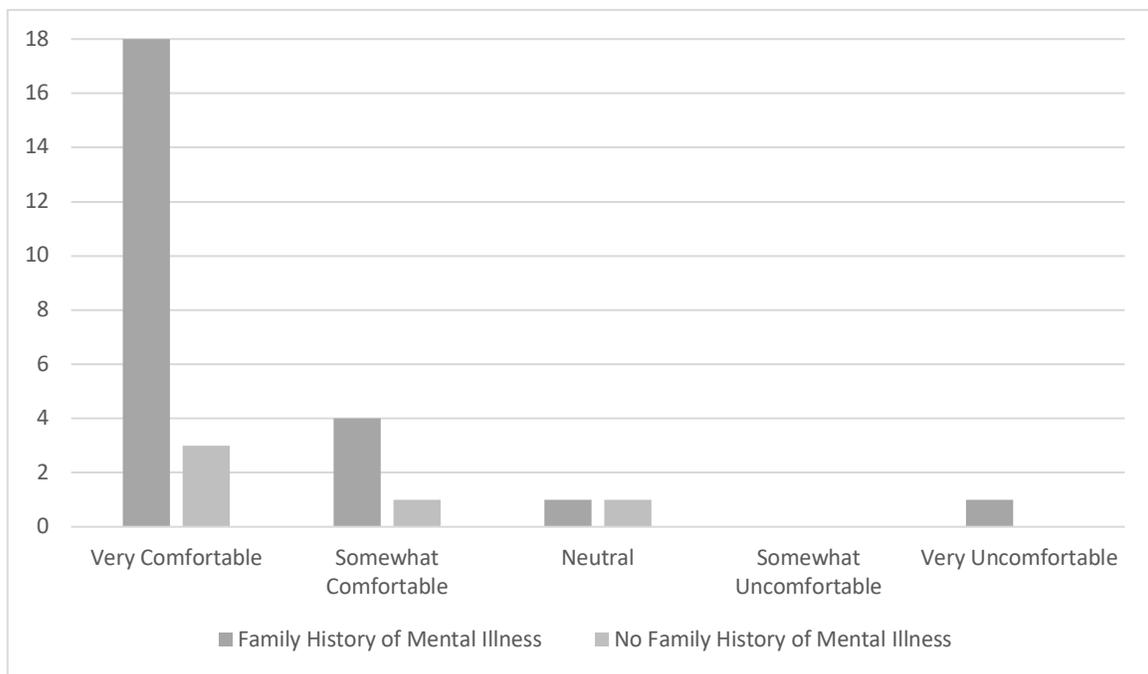


Figure 2.8 Family History of Mental Illness and Comfort/Hypothetical Comfort with Being Directly Asked About Mental Illness (n = 29)

#### 2.4.6 Child Personal and Family History of Mental Illness

All of the 29 participants that had a child being evaluated at the genetics visit indicated whether or not the child had a diagnosis of MI. Nineteen (66%) participants reported that their child did not have a diagnosis of MI, while six participants indicated that their child does not have a diagnosis, but suspect that their child might have a mental health condition. Three participants indicated that their child had a mental health

diagnosis (i.e. ADHD, ADD, OCD, and anxiety). One participant was unsure if her child had a mental health diagnosis. Of the nine participants who indicated their child has a positive personal history of MI (diagnosed or suspected), four participants had a discussion about mental health topics with the genetics provider, while five did not.

#### *2.4.7 Participant Anxiety and Depression Severity (PHQ-9 and GAD-7 scores)*

Twenty-nine participants completed the PHQ-9. Participants were classified into six groups: zero (n = 7, 29%), minimal (n = 11, 38%), mild (n = 7, 24%), moderate (n = 4, 14%), moderately severe (n = 0), and severe level of depression severity (n = 0). The majority of participants scored in the minimal level of depression severity range, with a mean PHQ-9 score of 4.07 (SD = 4.21). The maximum PHQ-9 score was 14, in the moderate level of depression severity range.

The total raw scores obtained on the PHQ-9 in this sample were statistically significantly different ( $p = .012$ ) than those obtained in the standardization sample as reported in Kocalevent et al. (2013) study of standardization of the PHQ-9 in the general population (n = 5018, M = 2.91, SD = 3.52). The participants' mean raw PHQ-9 scores in this study (n = 29, M = 4.07, SD = 4.21) were higher than those reported in the standardization/normative sample.

Twenty-eight participants completely answered the GAD-7. One participant partially completed the GAD-7 and an average score was calculated based on their completed responses, making a total of 29 participants who completed the GAD-7. This participant had an average score of 2.33 and was considered to be in the minimal level of anxiety severity. Participants were classified into five groups: zero (n = 5, 17%), minimal (n = 10, 34%), mild (n = 10, 34%), moderate (n = 1, 3%), and severe level of anxiety severity (n = 3, 10%). The majority of participants scored in the minimal and mild levels

of anxiety severity range. The mean GAD-7 score was 5.25 (SD = 5.05), in the mild level of anxiety severity range. The maximum GAD-7 score was 18, in the severe level of anxiety severity range.

The total raw scores obtained on the GAD-7 in this sample were statistically significantly different ( $p = .0003$ ) than those obtained in the standardization sample as reported in Lowe et al. (2008) study of validation and standardization of the GAD-7 in the general population ( $n = 5030$ ,  $M = 2.95$ ,  $SD = 3.41$ ). The participants' mean raw scores in this study ( $n = 29$ ,  $M = 5.25$ ,  $SD = 5.05$ ) were higher than those reported in the standardization/normative sample.

## 2.5 DISCUSSION

This study used survey methodology to explore patient, caregiver, and/or parent reported interest in and comfort with discussing personal and/or family history of MI with a genetics provider during a general genetic counseling visit. This is the first study known to explore this topic within a general genetic counseling population. Overall, results indicate that the majority of participants expressed comfort with directly being asked about personal and/or family history of MI and with discussing mental health topics with a genetics provider during their genetic counseling visit. The majority of participants who did not discuss mental health topics indicated interest in discussing one or more topics related to MI as well as hypothetical comfort with being explicitly asked about personal and/or family history of MI.

We hypothesized that individuals with a personal and/or family history of MI would be more comfortable with discussing their history of MI with a genetics provider than individuals who have no such personal or family history. Our study did not show a statistically significant difference between these two groups, suggesting that comfort with

discussing MI is not dependent of the presence of personal and/or family history. Overall, most participants with and without a personal or family history of MI expressed some degree of comfort with discussing MI with the genetics provider. Most participants that did not discuss MI with the genetics provider expressed that they would have been interested in discussing one or more mental health topics with the genetics provider.

Only one participant expressed discomfort being asked about family history of MI and one participant expressed discomfort with discussing MI. Interestingly, the one participant who reported being very uncomfortable being asked also indicated that they were very comfortable with having a discussion about MI with the genetics professional. This participant reported both a personal and family history of MI and scored in the moderate depression severity level and mild anxiety severity level around the time of their visit. For the one participant who reported being uncomfortable with discussing MI, they reported no personal or family history of MI and had scores of zero on both of the depression and anxiety severity screening tools. Prior to having a discussion, this participant indicated that they felt very comfortable with being explicitly asked about a personal or family history of MI. This particular participant indicated that the genetics provider discussed genetic testing associated with MI with them and that there were no other topics that they would have liked to discuss. It is possible that there was a discussion about MI in this participant's session because the genetics provider was suspicious of a genetic condition in which MI is a feature in the child that was being evaluated. There is no way to determine if this was the case, but if so, the topic may have been uncomfortable for this particular participant. Of the participants who were not directly asked or were unsure if they were asked about personal and/or family history of

MI, most expressed hypothetical comfort and no participants reported any hypothetical degree of discomfort.

A recent master's thesis (Nimrichter et al., 2018) studied patient interest in and comfort with discussing MI in a prenatal genetic counseling setting. This study found that a majority of participants expressed interest in and comfort with discussing MI with a genetic counselor regardless of the presence of a family history, at 70% and 72%, respectively. Additionally, the absence or presence of depressive symptoms in the participant did not dictate interest level. The study suggested that discussion of family and personal history of MI is often welcome or well received by genetic counseling patients in the prenatal setting. Our study's sample reported higher levels of depressive symptoms than in the prenatal group. However, high levels of comfort with and interest in addressing personal and/or family history of mental illness within the general genetic counseling population remains similar.

In contrast to the prenatal genetic counseling study, the current study reported higher levels of depressive symptoms than in the prenatal group. In addition, the current study also found increased levels of anxiety. This may not be such an unexpected finding in this population when considering that there are a number of studies which describe that caregivers of children with chronic conditions (intellectual disabilities, cystic fibrosis, mitochondrial disease, and Duchenne muscular dystrophy) report higher psychological symptoms, increased levels of stress and worry, and poorer health outcomes and psychosocial well-being overall for both themselves and their families, when compared to control groups (Cantwell, J et al., 2015; Besier, T, et al., 2011; Senger, B, et al., 2015; Abi Daud, MS et al., 2004).

The current study echoes previous research on the degree of psychiatric symptoms in patients affected by chronic conditions and the caregiver population experience when compared to controls. While the majority of participants scored in the zero to mild ranges for depression and anxiety severity, the total raw scores obtained on the PHQ-9 measuring level of depression severity and GAD-7 measuring level of anxiety severity in this sample were higher than those reported in the normative samples for each screening tool (Kocalevent et al. 2013; Lowe et al. 2008). These results underscore the importance of addressing MI within the general genetic counseling patient and caregiver population, based on reported participant depression and anxiety severity in this sample.

A study exploring genetic counselor' perceptions of and attitudes toward schizophrenia found that a large proportion of genetic counselors are reluctant to ask patients about psychiatric illness when taking a family history, and have apprehension feeling that their services might not be helpful in the context of schizophrenia (Monaco, Conway, Valverde, and Austin, 2010). Roughly 33% of genetic counselors in that study felt that patients are uncomfortable being asked. When a family history of schizophrenia did come up during a counseling session, many of the genetic counselors reported providing little details about the etiology of MI. The authors theorized that this could have possibly been due to lack of knowledge, time constraints, or belief that it may not be beneficial to discuss a condition for which there is no genetic testing for establishing, refining, confirming, or excluding a psychiatric diagnosis (Monaco, Conway, Valverde, and Austin, 2010). The responses from the current study suggest that patients, caregivers, and/or parents are or would be comfortable with being directly asked about personal and/or family history of MI, regardless of personal and/or family history, and provides further support for the recommendation that genetics providers should routinely

incorporate asking about MI when taking a family history and should be equipped to have a follow up discussion with the patient or family. A 2017 study by Inglis et al. demonstrates that when clinicians routinely ask patients about a personal/family history of MI, and explore potential associated concerns with them, the conversation can foster the opportunity for patients to share their experiences, reduce feelings of stigma, and ensure that they are receiving the appropriate support (Inglis et al, 2017).

The present study has three major limitations. First, the study was conducted within three general genetic counseling clinics in South Carolina with a limited number of participants. There may be sample biasing in that many of the participants scored high on the depression and anxiety scales and may have been more interested in completing a study about MI. It is possible that patients, parents, and/or caregivers in different general genetic counseling clinics may not report the same level of depression or anxiety as seen in this study, which may affect the level of interest and/or comfort. Second, because it was at the discretion of the genetics professional who met with the participant to invite them to take the survey, there may have been unintentional bias as to who was given the survey. For example, if a potential participant disclosed a personal or family history of MI, the genetics provider may have been more apt to invite them to participate than if no such history was revealed. Third, the genetics providers may have been more likely to directly ask potential participants about a personal and/or family of MI since they were aware that this study was being conducted within each of their clinics. If this were the case, this could have potentially skewed the data.

Future studies might replicate this study in other general genetic counseling clinics and regions of the United States, in an attempt to produce a larger scale study. Additionally, future research could be done in more specialized genetics clinics, such as

in a muscular dystrophy clinic or neurogenetics clinic to see if similar or different themes are present in specific populations.

### CHAPTER III: CONCLUSION

This is the first known study to explore patient, parent, and/or caregiver interest in and comfort with discussing a personal and/or family history of MI with a genetic counselor during a general genetic counseling visit. The majority of participants expressed interest in and comfort with discussing MI with a genetics professional regardless of the presence of a personal and/or family history. Genetics providers who practice in general genetic counseling clinics should routinely ask about MI while taking the family history and be prepared to follow up by discussing mental health topics (i.e. etiology, recurrence risk, resources, etc.) with those who disclose a personal and/or family history, or with those who express depressive and/or anxious symptoms. Replication of this study in other general or specialty genetic counseling clinics is warranted to provide further support to these claims.

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## APPENDIX A: QUESTIONNAIRE MATERIALS



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My name is Alena Faulkner and I am a genetic counseling student in the University of South Carolina's Genetic Counseling Program. You are invited to participate in a study to see what information you would be interested in receiving from a genetic counselor or doctor in a genetic evaluation/counseling visit. Your participation in this research is voluntary. Your care at the Greenwood Genetic Center will not be affected whether you complete this study or not.

If you decide to participate, this brief questionnaire will take 5-10 minutes of your time. Most of the questions are asking about your interest in and comfort level with talking about mental health conditions during review of family medical history with a genetic counselor or doctor. You may feel uncomfortable answering some of the questions. You do not have to answer any of the questions that make you feel uncomfortable. By beginning and submitting this questionnaire, you are agreeing to participate.

Each participant has the option to enter in a drawing by completing the form at the end of the questionnaire, which will include a chance to win a gift card of \$20 to popular stores, online retail sites, or restaurants (i.e. Chick-Fil-A, Wal-Mart, Amazon, Target, etc.). One winner will be randomly drawn in the last week of each month from October 2018 to April 2019. To enter into the drawing, please write down your email address on the small sheet attached to the back of the survey. Return the small slip of paper with your email address to the office's front desk or by mail using one of the two self-addressed envelopes. Please tear this front sheet away and keep it for your records. If you are a winner, look for an email with the heading titled "USC SURVEY WINNER" from this email address: [Alena.Faulkner@uscmed.sc.edu](mailto:Alena.Faulkner@uscmed.sc.edu)

All responses gathered from the questionnaire and your contact information submitted for the drawing for a gift card will be kept anonymous and confidential. The results of this study may be published or presented at academic meetings; however, participants will not be identified.

We will be happy to answer any questions you have about the study. You may contact me at (803) 545-5775 and [Alena.Faulkner@uscmed.sc.edu](mailto:Alena.Faulkner@uscmed.sc.edu) or my faculty advisor, Vicki Vincent at (803) 545-5727 and [Victoria.Vincent@uscmed.sc.edu](mailto:Victoria.Vincent@uscmed.sc.edu) if you have study related questions or problems. If you have any questions about your rights as a research participant, you may contact the Office of Research Compliance at University of South Carolina at (803) 777-7095.

Thank you for your consideration. If you would like to participate, please type the following link [ <https://www.surveymonkey.com/r/WV9JPWZ> ] or scan the QR code at the bottom of this page to complete the questionnaire online, or fill out the provided paper copy. When you are done, please submit the online questionnaire or return the completed paper copy to a Greenwood Genetic Center staff member, or by using one of the two attached self-addressed envelopes. No postage is necessary.

Best,

Alena Faulkner  
(803) 545-5775  
[Alena.Faulkner@uscmed.sc.edu](mailto:Alena.Faulkner@uscmed.sc.edu)



**Section A:**

- 1) Who was the person that was referred for the appointment with the genetic counselor and/or doctor today?
- Me
  - My child (Biological or adopted)
  - A child who is a blood relative to me, and for whom I am a legal guardian (e.g. grandchild, nephew, niece, cousin, etc.)
  - A child who is not a blood relative to me, but for whom I am a legal guardian (e.g. foster child, step child, etc.)
  - Other: \_\_\_\_\_
- 2) Which clinic are you or your child being seen at today:
- Greenwood Genetic Center – Columbia
  - Greenwood Genetic Center – Greenville
  - Greenwood Genetic Center – Greenwood
- 3) What was the reason for your visit today (e.g. To talk about my family history of a known genetic condition such as muscular dystrophy, to determine whether I or my child might have a genetic syndrome, etc.)?
- 

**Section B: General information (of the individual filling out this form)**

- 4) What is your age: \_\_\_\_\_
- 5) What was the biological sex that you were assigned at birth?
- Male
  - Female
- 6) What is your current gender identity?
- Male
  - Female
  - Other (please specify): \_\_\_\_\_
- 7) What ethnicity (or ethnicities) would you describe yourself as: (Choose all that apply)
- American Indian or Alaska Native
  - Asian
  - Black or African American
  - Hispanic or Latino
  - Native Hawaiian or Other Pacific Islander
  - White
  - Other

8) Highest Degree or Level of Education Completed (select one):

- Less than high school
- High school graduate or equivalent (e.g. GED)
- Some college, no degree
- Associate's degree
- Bachelor's degree
- Master's degree
- Other advanced degree beyond a Master's degree

**Section C: During the first genetics visit, a genetic counselor or doctor usually asks questions about family medical history including medical conditions, birth defects, developmental delays, etc...**

9. Did your genetic counselor or doctor ask you directly about a personal or family history of mental illness (schizophrenia, bipolar disorder, anxiety disorders, depression, OCD, eating disorders, etc.)?

- No (**please go to section D, question 15**)
- Unsure (**please go to section D, question 15**)
- Yes

10. If you answered yes to question 9, how did you feel about being asked about a personal or family history of mental illness?

- Very comfortable
- Somewhat comfortable
- Neutral; neither comfortable nor uncomfortable
- Somewhat uncomfortable
- Very uncomfortable

11. If you answered yes to question 9, did you have a discussion about mental illness with the genetic counselor or doctor?

- No (**please go to section D, question 16**)
- Unsure (**please go to section D, question 16**)
- Yes

**If you answered yes to question 11, “did you have a discussion about mental illness with the genetic counselor or doctor”, please answer questions 12 to 14.**

12. What mental health topic(s) did the genetic counselor or doctor discuss? (Select all that apply)

- Factors that can cause mental illness
- Chance for family members to develop mental illness
- Things you can do to lower the chance to develop mental illness (e.g. exercise, adequate sleep, etc.)
- Resources (e.g. information about mental health professionals)
- Genetic Testing
- Other: \_\_\_\_\_

13. Were there topics about mental health conditions you wish that the genetic counselor or doctor would have talked about? (Select all that apply)

- Factors that can cause mental illness
- Chance for family members to develop mental illness
- Things you can do to lower the chance to develop mental illness (e.g. exercise, adequate sleep, etc.)
- Resources (e.g. information about mental health professionals)
- Genetic Testing
- Other: \_\_\_\_\_
- No, there were no other topics I would have liked covered

14. How comfortable were you discussing these mental health topics with your genetic counselor or doctor?

- Very comfortable
- Somewhat comfortable
- Neutral; neither comfortable nor uncomfortable
- Somewhat uncomfortable
- Very uncomfortable

**Please go to Section E, question 17.**

**Section D: If you answered NO to question 9, “Did your genetic counselor or doctor ask you directly about a personal or family history of mental illness”, please answer questions 15 and 16.**

15. How would you have felt if your genetic counselor or doctor asked you directly about a personal or family history of mental illness?

- Very comfortable
- Somewhat comfortable
- Neutral; neither comfortable or uncomfortable
- Somewhat uncomfortable
- Very uncomfortable

16. Please select the topics related to mental illness that you would have liked to have discussed with your genetic counselor or doctor (select all that apply):

- Factors that cause mental illness
- Chance for family members to develop mental illness
- Things you can do to lower the chance to develop mental illness (e.g. exercise, adequate sleep, etc.)
- Resources (e.g. information about mental health professionals)
- Genetic Testing
- Not applicable (e.g. no family or personal history of mental illness)
- None – I would not want to discuss this with the doctor/genetic counselor

**Section E:**

17. Have you ever had a diagnosis of mental illness (e.g. schizophrenia, bipolar disorder, anxiety disorders, depression, OCD, eating disorder, etc)?

- Yes (please write diagnosis here): \_\_\_\_\_
- No, but I suspect I have a mental illness (please write suspected diagnosis here): \_\_\_\_\_
- No
- Unsure

18. Do you have a close family member (i.e. brother, sister, parent, or child) who has been diagnosed with a mental illness (e.g. schizophrenia, bipolar disorder, anxiety disorders, depression, OCD, eating disorder, etc.)?

- Yes (please write diagnosis here): \_\_\_\_\_
- No, but I suspect a family member might have a mental illness (please write suspected diagnosis here): \_\_\_\_\_
- No
- Unsure

**Section F: If the person referred for the appointment today is a child in your care, please answer questions 19-20:**

19. Does the child have a diagnosis of mental illness (e.g., schizophrenia, bipolar disorder, anxiety disorders, depression, OCD, eating disorder, etc.)?
- Yes (please write diagnosis here) \_\_\_\_\_
  - No, but I suspect they might have a mental illness (please write suspected diagnosis here): \_\_\_\_\_
  - No
  - Unsure
20. Does the child have a family history (i.e. brother, sister, or parent) of mental illness (schizophrenia, bipolar disorder, anxiety disorders, depression, OCD, eating disorders, etc.)?
- Yes
  - No

**Section G: Please complete questions 21-30 by checking the answer that comes closest to how you have felt over the last 2 weeks, not just today.**

**Over the last 2 weeks, how often have you been bothered by the following problems?**

21. Little interest or pleasure in doing things
- Not at all
  - Several days
  - More than half the days
  - Nearly every day
22. Feeling down, depressed, or hopeless
- Not at all
  - Several days
  - More than half the days
  - Nearly every day
23. Trouble falling asleep or staying asleep, or sleeping too much
- Not at all
  - Several days
  - More than half the days
  - Nearly every day

24. Feeling tired or having little energy

- Not at all
- Several days
- More than half the days
- Nearly every day

25. Poor appetite or overeating

- Not at all
- Several days
- More than half the days
- Nearly every day

26. Feeling bad about yourself – or that you are a failure or have let yourself or your family down

- Not at all
- Several days
- More than half the days
- Nearly every day

27. Trouble concentrating on things, such as reading the newspaper or watching television

- Not at all
- Several days
- More than half the days
- Nearly every day

28. Moving or speaking so slowly that other people could have noticed? Or, the opposite – being so fidgety or restless that you have been moving around a lot more than usual

- Not at all
- Several days
- More than half the days
- Nearly every day

29. Thoughts that you would be better off dead or hurting yourself in some way

- Not at all
- Several days
- More than half the days
- Nearly every day

30. If you checked off any problems in Section G, how difficult have these problems made it for you to do your work, take care of things at home, or get along with other people?

- Not difficult at all
- Somewhat difficult
- Very difficult
- Extremely difficult

**Section H: Please complete questions 31-38 by checking the answer that comes closest to how you have felt over the last 2 weeks, not just today.**

**Over the last 2 weeks, how often have you been bothered by the following problems?**

31. Feeling nervous, anxious or on edge

- Not at all
- Several days
- More than half the days
- Nearly every day

32. Not being able to stop or control worrying

- Not at all
- Several days
- More than half the days
- Nearly every day

33. Worrying too much about different things

- Not at all
- Several days
- More than half the days
- Nearly every day

34. Trouble relaxing

- Not at all
- Several days
- More than half the days
- Nearly every day

35. Being so restless that it is hard to sit still

- Not at all
- Several days
- More than half the days
- Nearly every day

36. Becoming easily annoyed or irritable

- Not at all
- Several days
- More than half the days
- Nearly every day

37. Feeling afraid as if something awful might happen

- Not at all
- Several days
- More than half the days
- Nearly every day

38. If you checked off any problems in Section H, how difficult have these problems made it for you to do your work, take care of things at home, or get along with other people?

- Not difficult at all
- Somewhat difficult
- Very difficult
- Extremely difficult

**Thank you for your participation! Please return your completed survey to a Greenwood Genetic Center staff member, or mail it using the self-addressed envelope.**



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**If you or someone you know has a mental illness, there are ways to get help. These resources can be used to find help for you, a friend, or family member.**

**Get Immediate Help**

National Suicide Prevention Lifeline (1-800-273-8255)

Veterans, Military, & Families Crisis Line (1-800-273-8255, Press 1)

**Finding Health Care Providers and Treatment in Your Area**

South Carolina Mental Health Centers by County (<http://www.state.sc.us/dmh/cmhc.htm>)

Columbia Area Mental Health Center (803) 898-4800/(803) 898-8888

Lexington County Mental Health Center (803) 996-1500/(803) 395-3545

Orangeburg Area Mental Health Center (803) 536-1463

Beckman Center for Mental Health Services (864) 229-7120

Greenwood Mental Health Clinic (864) 223-8331

Greenville Mental Health Center (864) 241-1040

Palmetto Health Day Treatment Program (Columbia, SC) (803) 296-8765

<https://www.palmettohealth.org/medical-services/behavioral-care>

Carolina Center for Behavioral Health (Greer, SC) (864) 235-2335/800-866-4673

<https://thecarolinacenter.com/>

Inpatient, Partial Hospitalization, and Intensive Outpatient Programs

Greenville Health System (Greenville, SC) <http://ghspsych.org/>

Inpatient, Outpatient, Partial Hospitalization, and Intensive Outpatient Programs

Substance Abuse and Mental Health Services Administration Referral Helpline (1-800-662-4357)

Substance Abuse and Mental Health Services Administration Behavioral Health

Treatment Locator (<https://findtreatment.samhsa.gov/>)

Local Therapists ([www.psychologytoday.com](http://www.psychologytoday.com))

\*Contact your insurance company for a list of providers

### **National Agencies, Advocacy Groups, and Professional Organizations**

Anxiety and Depression Association of America (<https://www.adaa.org>)

Depression and Bipolar Support Alliance ([www.dbsalliance.org](http://www.dbsalliance.org) / (800) 826-3632 toll free)

Mental Health America (<http://www.mentalhealthamerica.net> / (800) 969-6642 toll free)

National Alliance on Mental Illness ([www.nami.org](http://www.nami.org) / (800) 950-6264)

Family Caregiver Alliance (<https://www.caregiver.org/> / (800) 445-8106 toll free)

Postpartum Support International (<http://www.postpartum.net> / (800) 944-4773 toll free helpline)

### **Help for Service Members and Their Families**

Current service members, veterans, and their families may face different mental health issues than the general population. Please visit <https://www.mentalhealth.gov/get-help/veterans/index.html> for a list of resources.

### **More Information about Mental Health Conditions**

National Institute of Mental Health (<https://www.nimh.nih.gov/health/topics/index.shtml>)

National Society of Genetic Counselors: Mental Health and Genetics

(<http://aboutgeneticcounselors.com/Genetic-Conditions/Mental-Health-Conditions>)

Women's Mental Health ([www.womensmentalhealth.org](http://www.womensmentalhealth.org))



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**Enter to win!**

**Winners will be chosen at random for a gift card once a month between October 2018-January 2019. Emails will be kept secure and confidential and will be shredded after each drawing.**

**Email:** \_\_\_\_\_

**Please tear off this page from the questionnaire and return it either by using the self-addressed envelope, or to the box/folder in the Greenwood Genetic Center waiting room. Thank you for your participation!**