Genetic Disorders with Psychiatric Manifestations: A Survey of Genetic Counselors

Master's Thesis

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Abstract

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A thesis presented to the Genetic Counseling Program

Graduate School of Arts and Sciences
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Waltham, Massachusetts

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In recent years it has become recognized that many genetic disorders, such as VCFS, may include psychiatric manifestations as part of the phenotype. Individuals diagnosed with one of these disorders have an increased risk to develop psychiatric illness in childhood or as adults. Previous research by Sachs (2008) demonstrated that parents of children with VCFS preferred to learn about the risk of psychiatric illness from a medical provider at the time that the diagnosis is made. However, in practice many of the parents learned about the increased risk for psychiatric conditions through the internet. In the current study, we wished to explore the opinions and practices of genetic counselors regarding the disclosure of risk for psychiatric illness that accompanies VCFS, Klinefelter syndrome, Prader-Willi and Williams syndrome. We recruited
prenatal and pediatric genetic counselors through the NSGC listserv to complete an anonymous, online survey that included multiple choice and Likert-scale questions. A total of 42 prenatal and 38 pediatric counselors completed the survey, for a total of 80 responses. Overall, the prenatal counselors were more likely than the pediatric counselors to discuss the risk to develop psychiatric illness with families. However, the prenatal counselors reported that they often only mention the increased risk to families while the pediatric counselors reported they were more likely to discuss details, such as treatment options, diagnostic criteria and natural history. Our participants reported that if there were better treatment and/or preventative options to offer families they would feel more comfortable discussing the risk with families. Interestingly, the respondents reported that the ability to offer predictive genetic testing for mental illness would not have a major impact on their likelihood to discuss the increased risk. Our findings suggest that future efforts should focus on increasing counselors’ knowledge of and comfort regarding mental illness.
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Introduction

Mental illness is a serious disease that affects a large percentage of the population. In the United States roughly 26.2% of adults have been diagnosed with some form of a mental illness (Kessler et al., 2005). Studies have found that about 15% of children have been identified as suffering from some type of psychological disorder (In-Albon et al., 2010). The prevalence of mental illness has been increasing and this could be due to a true increase in the disorder, increased awareness of mental illness, or changes in the diagnostic criteria (Barbaresi et al., 2009). Regardless of the reason or reasons behind the increase in the observed incidence of mental illness, it is increasingly affecting more and more people.

Mental illness is an umbrella term that defines a group of individuals who are suffering or are in some way incapacitated by an emotional, cognitive, or personality disorder. The Diagnostic and Statistical Manual of Mental Disorders 4th ed. (2000) defines mental illness as

“a clinically significant behavioral or psychological syndrome or pattern that occurs in an individual and that is associated with present distress (e.g., a painful symptom) or disability (i.e., impairment in one or more important areas of functioning) or with a significantly increased risk of suffering death, pain, disability, or an important loss of freedom.”
This covers a wide spectrum of conditions including depression, attention deficit disorder (ADD), generalized anxiety disorder, personality disorders and schizophrenia. The age of onset can be anywhere from childhood to late adulthood. Mental illness typically affects a person’s ability to function or produces some form of a disability which is an impairment or limitation in participation or activities (WHO, 2001).

Presently, much research is aimed at finding the causative genes for many different types of mental illness. The hope is that by determining many of the genes and alleles that predispose people to developing a mental illness, testing could be pursued that could determine who is at risk for developing the disorder, predict the severity or course of the illness, and predict the best treatment options (Hebebrand et al., 2010). Prior research has shown that if testing were available that could predict whether a person would be affected with a mental illness or how they would respond to specific treatment many people would support the opportunity to undergo testing. Trippitelli et al. (1998) asked patients and their spouses whether or not they would take advantage of presymptomatic or prenatal testing for bipolar disorder if they became available. The majority said they would, indicating that the benefits of knowing the information outweighed the risks. The respondents ranked the ability to obtain preventative treatment as a primary benefit. To date the progress of research to identify psychiatric risk genes has been slow, due in part to the fact that mental illness in most instances is likely due to complex inheritance rather than Mendelian inheritance. The research has so far been contradictory due to the polygenic nature of mental illness and the potential utility of testing may never reach its full potential (Price, 2005).
Currently, the treatment of mental illness is based on the specific diagnosis that is made. Possible therapeutic interventions include pharmaceuticals, cognitive therapy, and alternative treatments. Pharmaceutical intervention is aimed at correcting the chemical change in the brain that is the cause of the illness and the symptoms the patient is experiencing (Escalas et al., 2010). Cognitive therapy is any form of therapy that employs talking in attempt to shape behavior or to diminish abnormal or unhealthy thoughts and feelings (Marriott & Kellett, 2009). Treatments are typically considered effective if they are able to elicit a change in the person’s abnormal thoughts, behavior, or feelings (Biering, 2010). Treatment for mental illness is often designed to ameliorate the symptoms of the condition.

A subset of mental illness is due to an underlying genetic condition in which the mental disorder is a part of a larger cluster of symptoms. It is imperative to recognize those patients whose mental disorder is the result of a genetic condition because the presence of a genetic condition could affect the treatment options available, increase the chance for other medical conditions, or influence the recurrence risk and the long term prognosis (Smoller et al., 2008). The correct diagnosis of an underlying genetic disorder is imperative in the correct medical management of the condition. Not only is the management of the condition dependent on the diagnosis, but the risk to other family members is impacted if a Mendelian inherited condition is diagnosed. There are many genetic conditions that have a psychiatric manifestation as a feature of the condition. Some examples are Velocardiofacial syndrome (VCFS), Klinefelter syndrome, Prader-Willi syndrome, and Williams syndrome. Though each of the four diagnoses
includes mental illness as a feature of the disorder, the specific types of mental illness
varies with each genetic condition.

Velocardiofacial syndrome is caused by a 1.5-3 Mb deletion of chromosome 22
that occurs in 1 out of every 4,000 births (Liao et al., 2004). Children with VCFS typically
have conotruncal heart defects, velopharyngeal insufficiency, thymic and parathyroid
defects, cleft palate, characteristic facial features and learning disabilities. The clinical
features of VCFS show variable expressivity and penetrance among different patients
(McDermid & Morrow, 2002; Aggarwal & Morrow, 2008). There is an increased
incidence of ADHD, oppositional defiance disorder, phobias, obsessive compulsive
disorders, generalized anxiety disorder and autism in people with VCFS (Gothelf, Schaer,
&Eliez, 2008). Adults with VCFS have been found to be at an increased risk to develop
bipolar disorder, schizophrenia and also schizoaffective disorder (Murphy, 2002).

Klinefelter syndrome is due to an extra X chromosome found in males, resulting
in a karyotype of XXY rather than XY. Klinefelter syndrome is believed to occur in about
1 in every 500 live born males, but only about 35% of males with Klinefelter are ever
diagnosed with the condition. Many go undiagnosed because they do not have any
medical problems. The physical characteristics found in people with Klinefelter include
above average height, gynecomastia, inadequate virilization, hypogonadism, and
infertility (Verri et al., 2010). Males with Klinefelter are believed to have normal to low
normal IQ scores, but express variable difficulties in language and reading (Boada et al.,
2009). Adolescents and adults with Klinefelter are at an increased risk to experience
anxiety and depression (Visootsak & Graham, 2006). In one study, almost one quarter (24%) of men with Klinefelter syndrome had experienced a depressive episode (Bruining et al., 2009).

Prader-Willi syndrome occurs in about 1 out of 22,000-25,000 live births and is caused by loss of the paternally inherited genes in 15q11-13 (Whittington & Holland, 2010). The cardinal features of Prader-Willi syndrome include low birth weight, hypotonia and feeding difficulties in infancy followed by hyperphagia and obesity in childhood. Other features include short stature, small hands and feet, characteristic facial features, intellectual disability and a distinctive behavioral profile (Buiting, 2010). The behavioral profile includes obsessiveness, hoarding of food, temper tantrums, mood swings and skin picking (Whittington & Holland, 2010). Almost all children with Prader-Willi syndrome will develop a psychotic disorder (Biliya & Bulla, 2010).

Williams syndrome is a neurodevelopmental disorder that is caused by a micro-deletion of 7q11.23. Children with Williams tend to have behavioral differences including being overly friendly, less reserved towards strangers, and more approaching and affectionate (Klein-Tasman et al., 2009). Individuals with Williams syndrome have a distinct facial appearance that is typified by a short nose with a broad nasal tip, puffiness around the eyes, wide mouth, full cheeks and lips and a small chin. People with Williams syndrome also have intellectual disability, cardiac anomalies, and growth failure (NINDS, 2008; Pober, 2010). The most prevalent psychiatric disorder diagnosed in children with Williams syndrome is attention deficit hyperactivity disorder (ADHD).
Nearly 65% of children with Williams will develop ADHD (Lo-Castro et al., 2010). Other mental illnesses that are more prevalent in people with Williams syndrome are anxiety disorders, obsessive-compulsive disorder, and autism (Stinton et al., 2010).

Previous research completed by Sachs (2008) looked at parents' perceptions of the risk of psychiatric manifestations associated with their child's diagnosis of VCFS. The results of the study showed that most parents knew that there was an increased risk for psychiatric conditions in people with VCFS. Parents most often reported learning about the increased chance for a psychiatric condition from the internet, but most said they would have preferred learning the information from a medical professional. Many parents also stated that they would have liked to have received the information at the time of diagnosis which was in contrast to the reported experience of most of the parents. Though most parents did not seem overly concerned about the possibility of their child developing a psychiatric disorder, when asked they did state they would be most concerned if their child developed schizophrenia or bipolar disorder. Many parents also reported that if given the opportunity they would use predictive genetic testing to determine if their child would develop an adult-onset psychiatric disorder. Nearly half (45.9%) of the parents stated that their child with VCFS had already been diagnosed with a mental illness, and 27.1% had more than one psychiatric diagnosis. Parents also reported that they found the internet to be the most useful source of educational information when learning about their child's psychiatric diagnosis. Although they turned to medical providers as a resource with a similar frequency as that of the
internet, parents reported medical providers as a useful source of educational information and emotional support with less frequency.

The future of genetic counseling is likely to include educating an increasing number of patients about the psychiatric manifestations of genetic conditions and the genetics surrounding psychiatric illness. Though genetic counselors are trained to teach the basics of genetics and support their patients through the counseling session, most have little education surrounding mental illness. Genetic counselors do not have adequate knowledge surrounding diagnosis, prognosis and treatment options for mental illness (Finn, 2006). Another difficulty regarding counseling for psychiatric conditions is the ambiguity surrounding mental illness. Symptoms of psychiatric illnesses can overlap, are subjective, and there is no laboratory test to confirm their existence (Anonymous, 2006). Though genetic counselors may not be trained in all aspects of mental illness, they have the opportunity to make a difference in the lives of those affected and their family members. By improving the prognosis of the affected individual genetic counselors could improve the quality of life for the whole family (Austin & Honer, 2004). Genetic counselors also have the chance to improve patients' lives by offering correct information regarding the course, prognosis, and treatment options and dispelling inaccurate beliefs (Finn & Smoller, 2006). Though genetic counselors are uniquely trained to discuss issues concerning heritable conditions and most say they feel comfortable discussing psychiatric conditions many say they rarely discuss information regarding mental illness. Monaco et al. (2009) found that 71% of genetic counselors reported they experience no discomfort when asking patients about
a family history of mental illness, but 75% reported that they rarely or never discuss early signs of psychiatric illness with their patients. Genetic counselors are trained to discuss heritable conditions and can offer families and those affected with mental illness a lot of information and comfort, though many do not take the opportunity when it is presented to them.

The goal of this study was to investigate genetic counselors’ opinions and practices when presenting information regarding psychiatric conditions that are possible symptoms of childhood genetic illnesses. Specifically, I aimed to find out if genetic counselors counsel parents about the increased risk to their child to develop a psychiatric condition after a genetic diagnosis and how much information genetic counselors discuss regarding the natural history, risk, and treatment of the psychiatric manifestation. I also aimed to investigate how comfortable counselors are discussing the possible psychiatric conditions with parents, and what could help to increase their comfort level when counseling about the risk for mental illnesses associated with a genetic diagnosis.
Methods

Subject Characteristics and Recruitment Procedures

This project received human subjects approval from the Brandeis University IRB. Genetic counselors that specialize in either prenatal or pediatric genetic counseling were eligible to participate in the survey. Subjects had to be over 18 years old and be fluent in English. The participants had to be currently working in a clinical setting and must have been counseling for at least one year. We recruited participants through the National Society of Genetic Counselors listserv. The Principle Investigator posted the recruitment notice containing a link to the research survey (see Appendix A) on the NSGC listserv. The Principle Investigator also posted a reminder recruitment notice on the NSGC listserv approximately two weeks after the original notice was posted. Potential participants who followed the link to the survey first saw an introduction to the survey (see Appendix B) describing the purpose of the study.

Data Collection

The survey was an anonymous online survey on www.qualtrics.com. There were two versions of the survey (see Appendix C), one for prenatal genetic counselors and one for pediatric genetic counselors. Each survey contained 56 potential
questions that were designed to collect data regarding counselors’ opinions and practices surrounding counseling about the increased risk to develop psychiatric manifestations associated with specific genetic diagnoses. The majority of the questions were multiple choice questions. There were 16 Likert-type questions to assess counselors’ comfort level with the specific psychiatric illnesses and also their opinions regarding testing for those mental illnesses. There were multiple questions that allowed for respondents elaborate or explain the answers they provided to previous questions.

The survey consisted of five sections. The first section was designed to collect demographic information on the respondents. Depending on how the potential participants responded to some of the questions they may have been funneled out of the survey, directed to the prenatal version, or directed to the pediatric version of the survey. The remaining four sections were designed to collect information on counselors’ opinions and practices regarding counseling for the psychiatric manifestations that are possible symptoms of four different genetic conditions.

The questions were also designed to elicit counselors’ comfort level with mental illness and counseling for possible psychiatric manifestations. Participants were also asked what, if anything, could help to increase their comfort level when counseling for psychiatric conditions. The questions in each of the remaining four sections (sections two through five) were identical except for the genetic condition in question. The conditions were chosen based on the severity of the associated mental illness and the age of onset of psychiatric manifestations. The second section of the survey asked
respondents about VCFS and looked at counselors’ opinions and clinical practices when the associated psychiatric illness is severe and has an adult onset. The third section asked respondents about Klinefelter syndrome and compares counselors’ opinions and clinical practices when the associated mental illness is less severe and also has an adult onset. The fourth section asked respondents about Prader-Willi syndrome and adresses counselors' opinions and practices when the possible presenting psychiatric condition is severe and has a childhood onset. The final section asked respondents about Williams syndrome and presents a less severe psychiatric illness with a childhood onset to assess counselors' opinions and clinical practices.

Data Analysis

The data was quantitatively analyzed using the data analysis program SPSS 18.0. The information was analyzed to calculate correlations and descriptive statistics. The groups were compared based on only the demographic information they provided.
Results

There were 117 surveys attempted. Of the 117 surveys that were started 80 participants fit all of the survey inclusion criteria. As shown in table 1 the majority of the participants were female (95%) and between the ages of 25 and 40 (81.3%). The greatest number of participants were located in region 4 (31.3%) and region 2 (22.5%). There were an equal number of participants who stated that they practiced in regions 1, 5, and 6 (11.3%). Almost half of all of the respondents had recently graduated (between 2006-2009) from a genetic counseling program (47.5%). Almost three quarters of counselors reported that they had graduated after the year 2000 (73.8%). Over 70% of respondents admitted to having a personal experience, family member, or close friend diagnosed with a mental illness. Just over half (52.5%) of the respondents said that they worked as a prenatal genetic counselor while pediatric genetic counselors made up close to half (47.5%) of the survey population. There were no statistically significant differences between the prenatal and pediatric counselor populations.
## Table 1.

<table>
<thead>
<tr>
<th>Age Range in Years</th>
<th>Frequency</th>
<th>Percent</th>
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<tbody>
<tr>
<td>25-29</td>
<td>31</td>
<td>38.8</td>
</tr>
<tr>
<td>30-34</td>
<td>21</td>
<td>26.3</td>
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<td>35-39</td>
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<td>16.3</td>
</tr>
<tr>
<td>40-44</td>
<td>6</td>
<td>7.5</td>
</tr>
<tr>
<td>45-49</td>
<td>3</td>
<td>3.8</td>
</tr>
<tr>
<td>50-54</td>
<td>4</td>
<td>5</td>
</tr>
<tr>
<td>55-59</td>
<td>1</td>
<td>1.3</td>
</tr>
<tr>
<td>60-64</td>
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<td>1.3</td>
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<tr>
<td><strong>Total</strong></td>
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<td><strong>100</strong></td>
</tr>
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</table>

<table>
<thead>
<tr>
<th>Gender</th>
<th>Frequency</th>
<th>Percent</th>
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<tbody>
<tr>
<td>Male</td>
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<td>5</td>
</tr>
<tr>
<td>Female</td>
<td>76</td>
<td>95</td>
</tr>
<tr>
<td><strong>Total</strong></td>
<td><strong>80</strong></td>
<td><strong>100</strong></td>
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<thead>
<tr>
<th>Region of Practice</th>
<th>Frequency</th>
<th>Percent</th>
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</thead>
<tbody>
<tr>
<td>1 (CT, MA, ME, NH, RI, VT, CN Maritime Provinces)</td>
<td>9</td>
<td>11.3</td>
</tr>
<tr>
<td>2 (DC, DE, MD, NJ, NY, PA, VA, WV, PR, VI, Quebec)</td>
<td>18</td>
<td>22.5</td>
</tr>
<tr>
<td>3 (AL, FL, GA, KY, LA, MS, NC, SC, TN)</td>
<td>10</td>
<td>12.5</td>
</tr>
<tr>
<td>4 (AR, IA, IL, IN, KS, MI, MN, MO, ND, NE, OH, OK, SD, WI, Ontario)</td>
<td>25</td>
<td>31.3</td>
</tr>
<tr>
<td>5 (AZ, CO, MT, NM, TX, UT, WY, Alberta, Manitoba, Sask.)</td>
<td>9</td>
<td>11.3</td>
</tr>
<tr>
<td>6 (AK, CA, HI, ID, NV, OR, WA, British Columbia)</td>
<td>9</td>
<td>11.3</td>
</tr>
<tr>
<td><strong>Total</strong></td>
<td><strong>80</strong></td>
<td><strong>100</strong></td>
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<tr>
<th>Year Graduated from a Genetic Counseling Program</th>
<th>Frequency</th>
<th>Percent</th>
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<tr>
<td>1975-79</td>
<td>1</td>
<td>1.3</td>
</tr>
<tr>
<td>1980-84</td>
<td>3</td>
<td>3.8</td>
</tr>
<tr>
<td>1985-89</td>
<td>1</td>
<td>1.3</td>
</tr>
<tr>
<td>1990-94</td>
<td>4</td>
<td>5</td>
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<tr>
<td>1995-99</td>
<td>12</td>
<td>15</td>
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<tr>
<td>2000-05</td>
<td>21</td>
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</tr>
<tr>
<td>2006-09</td>
<td>38</td>
<td>47.5</td>
</tr>
<tr>
<td><strong>Total</strong></td>
<td><strong>80</strong></td>
<td><strong>100</strong></td>
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</table>

<table>
<thead>
<tr>
<th>Primary Specialty Area</th>
<th>Frequency</th>
<th>Percent</th>
</tr>
</thead>
<tbody>
<tr>
<td>Prenatal genetics</td>
<td>42</td>
<td>52.5</td>
</tr>
<tr>
<td>Pediatric genetics</td>
<td>38</td>
<td>47.5</td>
</tr>
<tr>
<td><strong>Total</strong></td>
<td><strong>80</strong></td>
<td><strong>100</strong></td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Personal Experience with a Psychiatric Illness</th>
<th>Frequency</th>
<th>Percent</th>
</tr>
</thead>
<tbody>
<tr>
<td>Yes</td>
<td>58</td>
<td>72.5</td>
</tr>
<tr>
<td>No</td>
<td>22</td>
<td>27.5</td>
</tr>
<tr>
<td><strong>Total</strong></td>
<td><strong>80</strong></td>
<td><strong>100</strong></td>
</tr>
</tbody>
</table>
As shown in figure 1, 76.2% of prenatal genetic counselors have counseled at least one family with a fetal diagnosis of Velocardiofacial syndrome (VCFS). All (100%) of the pediatric counselors responded that they had counseled a family regarding VCFS. Almost half of the prenatal counselors stated (40.5%) that they had counseled between 1-4 families with a diagnosis of VCFS. Nearly half (47.4%) of the pediatric counselors stated that they had counseled greater than 13 families regarding this diagnosis.

**Figure 1**

![Number of Families Counseled About VCFS](image)

As shown in figure 2, of the prenatal counselors who said they had counseled at least one family about VCFS, 62.5% said that they discussed the possibility for the fetus to later develop schizophrenia. An additional 28.1% said that they sometimes discussed the risk for a fetus to develop schizophrenia with a diagnosis of VCFS. Almost half (47.4%) of the pediatric counselors stated that they discuss the risk for a child with a diagnosis of VCFS to later in life develop schizophrenia. Half (50%) of the pediatric counselors stated that they sometimes counseled families regarding the risk to develop schizophrenia.
Nearly all of the counselors stated that at some point they discuss the risk of schizophrenia when counseling a family about VCFS.

**Figure 2**

As shown in figure 3, approximately 79% percent of prenatal genetic counselors reported that they had counseled at least one family regarding a fetal diagnosis of Klinefelter syndrome. More than half of these counselors (52.4%) had counseled between 1 and 4 families about a diagnosis of Klinefelter. Almost all (94.7%) of the pediatric counselors stated that they had counseled at least one family regarding a diagnosis of Klinefelter syndrome. Of the pediatric counselors who had seen a family regarding this diagnosis, nearly half (47.4%) of the counselors had seen between one and four families regarding Klinefelter syndrome.
As shown in figure 3, of the nearly 80% of prenatal counselors who had counseled at least one family regarding Klinefelter syndrome, 30.3% of those counselors indicated that they discuss the risk of depression with a diagnosis of Klinefelter. An additional 27.3% of the prenatal counselors stated that they sometimes discussed the risk of depression with a diagnosis of Klinefelter syndrome. Of the 36 pediatric counselors who responded that they had counseled for Klinefelter syndrome, 16.7% said that they discuss the increased risk to develop depression. An additional two-thirds of the counselors (63.9%) said that they sometimes discussed the increased risk to develop depression with families of children with Klinefelter.
As shown in figure 5, only about a third of prenatal counselors (35.7%) had ever counseled for a fetal diagnosis of Prader-Willi syndrome. Of those who had, 26.2% had counseled between 1-4 families about Prader-Willi. All but six (83.3%) of the pediatric counselors answered that they had counseled at least one family regarding Prader-Willi syndrome. More than half (55.6%) of the pediatric counselors had met with either 1-4 or 5-8 families with this diagnosis.
As shown in figure 6, of the prenatal counselors who had counseled a family regarding a prenatal diagnosis of Prader-Willi, one third (33.3%) discuss the risk of psychosis. Twenty percent (20%) reported that they *sometimes* discuss the risk of psychosis when counseling about Prader-Willi. Only 7 (23.3%) of the pediatric counselors responded that they discuss the risk of psychosis when they counsel families regarding a diagnosis of Prader-Willi. One third (33.3%) of the pediatric counselors reported that they *sometimes* discussed the risk of psychosis with this diagnosis.
As shown in figure 7, 23.8% of the prenatal counselors stated that they had counseled a family regarding a fetal diagnosis of Williams syndrome. The majority of the counselors who had seen a family for Williams syndrome had counseled between 1-4 families about the condition. Thirty-one (86.1%) of the pediatric counselors stated that they had counseled at least one family regarding a diagnosis of Williams syndrome. Of the counselors who had seen a family about this diagnosis, most (41.7%) had met with between one and four families.
As shown in figure 8, of the prenatal counselors who had counseled for Williams, 30% responded that they discuss the risk of ADD with a diagnosis of Williams. The same amount (30%) of prenatal counselors stated that they sometimes discuss the risk of ADD. Of the pediatric counselors who had met with a family regarding a diagnosis of Williams syndrome, twenty (64.5%) of the counselors said that they do discuss the risk of developing ADD. Of the remaining pediatric counselors who had counseled a family regarding Williams syndrome, all of them stated that they sometimes discuss with families the increased risk to develop ADD with a diagnosis of Williams syndrome. None of the pediatric counselors who have met with a family regarding a diagnosis of Williams syndrome said that they do not discuss the increased risk of ADD with a diagnosis of Williams.
As shown in figure 9, both the prenatal and pediatric genetic counselors had an opportunity to rate how severe they believed each of the four mental illnesses (schizophrenia, depression, psychosis, and ADD) to be on a scale of 1-7. The prenatal counselors rated psychosis to be the most severe with a mean score of 5.75 (SD= 0.672). The pediatric counselors also rated psychosis as the most severe condition with a mean score of 6.34 (SD=0.614). Both the prenatal and pediatric counselors rated ADD to be the least severe condition with a mean score of 4.10 (SD= 1.287) and 3.43 (SD=1.040), respectively.
As shown in figure 10, when asked which topics about each mental illness the counselors discussed during a session, the counselors were allowed to choose more than one option. The topics regarding VCFS and schizophrenia prenatal counselors discuss in order of frequency were

- mention the increased possibility [of schizophrenia] without discussing specific numbers (n=23)
- discuss the percentage of patients with VCFS who will later develop schizophrenia (n= 6)
- talk about the natural history of schizophrenia (n=4)

The topics regarding Klinefelter syndrome and depression prenatal counselors discuss in order of frequency were

- mention the increased possibility [of depression] without discussing specific numbers (n=16)
• discuss possible treatments and prevention (n=9)
• discuss the percentage of patients with Klinefelter who will later develop depression (n=3)
• discuss the natural history of depression (n=3)

Prenatal counselors indicated that they discuss the same information with parents when counseling about Prader-Willi syndrome and psychosis. The topics they discuss were

• mention the increased possibility [of psychosis] without discussing specific numbers (n= 5)
• discuss the percentage of patients with Prader-Willi who will later develop psychosis (n=3)
• talk about the natural history of psychosis (n=3)

The topics prenatal counselors discuss in order of frequency when counseling about Williams syndrome and ADD are

• mention the increased possibility [of ADD] without discussing specific numbers (n= 4)
• discuss the percentage of patients with Williams who will later develop ADD (n=2)
• discuss possible treatments and prevention (n=1)
• discuss diagnostic criteria/symptoms (n=1)
As shown in Figure 11, the frequency with which pediatric counselors discuss specific topics about schizophrenia during a session about VCFS were similar to the prenatal frequency. They

- mention the increased possibility [of schizophrenia] without discussing specific numbers (n=29)
- discuss the percentage of patients with VCFS who will later develop schizophrenia (n=18)
- discuss diagnostic criteria/symptoms (n=11)
The topics pediatric counselors discuss regarding depression with parents of children with Klinefelter were

- mention the increased possibility [of depression] without discussing specific numbers (n=29)
- discuss possible treatments or prevention (n=8)
- discuss diagnostic criteria/symptoms (n=6)

The topics pediatric counselors felt discuss regarding psychosis when counseling a family about a diagnosis of Prader-Willi were

- mention the increased possibility [of psychosis] without discussing specific numbers (n=17)
- discuss diagnostic criteria/symptoms (n=6)
- discuss possible treatments or prevention (n=4)

The topics that pediatric counselors were more likely to discuss regarding ADD and a diagnosis of Williams syndrome were

- mention the increased possibility [of ADD] without discussing specific numbers (n=30)
- discuss possible treatments or prevention (n=16)
- discuss diagnostic criteria/symptoms (n=10)

Both the prenatal and pediatric counselors were most likely to mention the increased possibility of a mental illness with families without going into specifics about the condition like the natural history of the condition, the percentage of patients diagnosed or the possible treatment or preventative options available. The pediatric counselors
were more likely than the prenatal counselors to discuss more topics regarding the possible mental illness with families.

**Figure 11**

![Figure 11](image_url)

As shown in figure 12 and figure 13, the vast majority of the prenatal and pediatric counselors felt that the most appropriate time to discuss with parents the possible psychiatric manifestations that are features of their child's genetic diagnosis is at the time they are diagnosed with the genetic condition.
Figure 12

Prenatal Counselors Beliefs Regarding the Appropriate Time to Discuss Psychiatric Manifestations

- VCFS
- Klinefelter
- Prader-Willi
- Williams

Times to Discuss

Frequency (n)
Figure 13 shows that the counselors rated their likeliness to offer families predictive testing for a mental illness if such testing were available on a Likert-scale between 1 and 7. Of the four mental illnesses the prenatal counselors indicated they would be most likely to offer predictive testing for schizophrenia with families of fetuses with VCFS. Prenatal counselors rated their likeliness to offer this to families of a fetus with VCFS with a mean score of 5.72 (SD= 1.326). The prenatal counselors were least likely to offer predictive testing for depression to families of a fetus diagnosed with Klinefelter syndrome. The mean score was 4.97 (SD= 1.571). The pediatric counselors felt that they
would be most likely to offer predictive testing for depression to families of children with a diagnosis of Klinefelter. The mean score was 5.11 (SD= 1.568). Pediatric counselors rated predictive testing for schizophrenia in children with VCFS the test they would be least likely to offer with a mean score of 3.87 (SD= 1.018).

Figure 14

As shown in figure 15, the counselors rated how comfortable they felt with their knowledge of specific mental illnesses and how comfortable they felt discussing specific mental illnesses with families on a Likert-scale between one and seven. Prenatal counselors felt most comfortable with their personal knowledge of depression with a mean score of 5.12 (SD= 1.293). The prenatal counselors rated their comfort with their personal knowledge of psychosis as the lowest with a mean score of 3.40 (SD= 1.454). Pediatric counselors felt most comfortable with their personal knowledge of ADD with a
mean score of 5.32 (SD = 1.249). The pediatric counselors were least comfortable with their personal knowledge of psychosis with a mean score of 3.57 (SD = 1.654).

**Figure 15**

![Bar chart showing counselors comfort level with their knowledge of mental illness](chart)

Figure 16 shows of the four mental illnesses, prenatal counselors were most comfortable discussing ADD with families that had a fetal diagnosis of Williams syndrome. The mean score was 5.00 (SD = 1.00). The prenatal counselors were least comfortable discussing psychosis with families that had a fetal diagnosis of Prader-Willi. The mean score was 3.60 (SD = 1.765). The pediatric counselors were most comfortable discussing ADD with families that had a child diagnosed with Williams syndrome with a mean score of 5.55 (SD = 1.091). Pediatric counselors rated their comfort with discussing psychosis with families that had a child diagnosed with Prader-Willi lowest with a mean score of 3.83 (SD = 1.683).
As shown in figure 17, for all of the four conditions the prenatal counselors indicated that if they had more knowledge surrounding the specific mental illnesses, that they would feel more comfortable discussing them. Having the ability to offer parents better treatment or prevention options would also increase their comfort discussing mental illness. Very few of the prenatal counselors, regardless of the condition, felt that there was nothing that could increase their comfort discussing mental illness with families. In figure 18, for all of the four conditions the pediatric counselors also indicated that if they had more knowledge about the specific mental illnesses they would feel more comfortable discussing them with families. Again, for all of the four conditions the pediatric counselors indicated that having better treatments or preventative options to offer families would help increase their comfort in these discussions. Like the prenatal counselors, the pediatric counselors also very rarely chose “nothing” when asked what would increase their comfort to discuss mental illness with families.
Figure 17

Ways to Increase Prenatal Counselors Comfort Discussing Mental Illness

Options to Increase Comfort Level

Figure 18

Ways to Increase Pediatric Counselors Comfort Discussing Mental Illness

Options to Increase Comfort Level
As shown in figure 19, for the prenatal counselors who responded no or sometimes when asked if they discussed the risk for psychiatric manifestations selected that the “fetus may never develop [the mental illness]” with the highest frequency when asked why they do not discuss the increased risk for mental illness with families. The prenatal counselors felt that the second most common reason why they do not discuss the possible psychiatric manifestations is that there are “more important possible symptoms to discuss”. Figure 20 shows the most common reasons the pediatric counselors chose as to why they do not discuss the risk for psychiatric manifestations with families. The pediatric counselors ranked the “children may never develop [the mental illness]” as the most common reason they do not discuss the risk for schizophrenia with families of children with VCFS and depression with families of children with Klinefelter. For Prader-Willi and Williams syndrome the most common reason pediatric counselors do not discuss the risk for psychosis and ADD, respectively, is because there are “more important possible symptoms to discuss”. For all four conditions the pediatric counselors were least likely to select it is “another provider's responsibility to discuss” when asked the reason they do not discuss the possible psychiatric manifestations with families.
Figure 19

Reasons Prenatal Counselors Do Not Discuss Psychiatric Manifestations

Figure 20

Reasons Pediatric Counselors Do Not Discuss Psychiatric Manifestations
As seen in figure 21, of the prenatal counselors who responded no or sometimes when asked if they discuss the risk for psychiatric manifestations the prenatal counselors selected (n=6) “geneticist” with the highest frequency when asked who should be responsible for discussing the risk of schizophrenia with a diagnosis of VCFS. When discussing the risk of depression with a diagnosis of Klinefelter syndrome the prenatal counselors selected (n=9) “genetic counselors” with the highest frequency as the provider responsible to discuss this risk with families. The prenatal counselors selected with equal frequency (n=5) “geneticists” and “genetic counselors” as the providers responsible to discuss the risk of psychosis with a diagnosis of Prader-Willi syndrome. The prenatal counselors felt that (n=3) “genetic counselors” should be the provider responsible to discuss the risk of ADD with a diagnosis of Williams syndrome.

Figure 21
Figure 22 shows the pediatric counselors who selected no or sometimes when asked if they discuss the risk of psychiatric manifestations who they selected as the appropriate provider to discuss the risk for mental illness. The pediatric counselors responded (n=12) “geneticist” as the most appropriate provider to discuss the risk of schizophrenia with a diagnosis of VCFS. “Geneticist” was also the provider who the pediatric counselors selected (n=13) as the provider who should be responsible to discuss the risk to develop depression with a diagnosis of Klinefelter. The pediatric counselors responded (n=9) “geneticist” as the most appropriate provider to discuss the possible risk of psychosis with a diagnosis of Prader-Willi. The pediatric counselors selected with equal frequency (n=4) both “geneticist” and “genetic counselor” as the most appropriate providers to discuss the risk to develop ADD with a diagnosis of Williams syndrome.

Figure 22

Pediatric Counselors Beliefs About the Provider Responsible to Discuss Psychiatric Manifestations

![Bar chart showing frequencies of provider selection for different conditions]
For most conditions a counselor’s comfort with discussing the possible psychiatric manifestations correlated with their comfort with their knowledge of the mental illness. Prenatal counselors’ feelings about their comfort discussing the risk of depression correlated with their comfort of their knowledge of depression $r= 0.469$, $\text{sig}= 0.006$. The prenatal counselors’ feelings regarding their comfort discussing psychosis correlated with their comfort regarding their knowledge of psychosis $r= 0.596$, $\text{sig}= 0.039$. The pediatric counselors’ comfort discussing schizophrenia correlates with their comfort of their knowledge regarding schizophrenia $r= 0.568$, $\text{sig}= 0.000$. Pediatric counselors’ comfort discussing depression with families correlates with their comfort with their knowledge of depression $r= 0.499$, $\text{sig}= 0.002$. The pediatric counselors’ comfort discussing the risk of psychosis correlated with the comfort they had regarding their knowledge of psychosis $r= 0.667$, $\text{sig}= 0.000$. Finally, pediatric counselors’ comfort discussing ADD with parents correlated with the counselors’ comfort with their knowledge of ADD $r= 0.649$, $\text{sig}= 0.000$. The prenatal counselors’ personal experience with a mental illness correlated with their likelihood to discuss the risk of depression with families of fetuses with Klinefelter syndrome $r= -0.350$, $\text{sig}= 0.046$. There were no statistically significant correlations between the prenatal counselors’ personal experience with a mental illness and their likelihood to discuss schizophrenia, psychosis or ADD. There were also no statistically significant correlations between pediatric counselors’ personal experience with a mental illness and their likelihood to discuss the risk to develop schizophrenia, depression, psychosis, or ADD.
Discussion

Demographics

The population of genetic counselors in this survey was on average younger than the average genetic counselor. In this survey the vast majority (65.1%) of the counselors were between the age of 25 and 34. Based on the 2010 Professional Status Survey completed by the NSGC the majority (56%) of genetic counselors are between the ages of 25 and 34. It is possible that the younger population of counselors in this study would explain the lack of experience counseling families regarding a diagnosis of VCFS, Klinefeter, Prader-Willi, and Williams syndrome.

Experience and Practice Counseling for Psychiatric Manifestations

The genetic counselors that we surveyed had more experience counseling for VCFS and Klinefelter syndrome than for Williams and Prader-Willi syndrome. This would make sense given that VCFS and Klinefelter are much more common conditions then Prader-Willi and Williams syndrome. The prevalence of VCFS is about 1 in 4,000 live births while Klinefelter is believed to occur in one out of 500 live born males (Liao at al. 2004; Verri et al. 2010). Whereas, Prader-Willi only occurs in 1 out of every 22,000 to 25,000 births and Williams has a prevalence of 1 of every 10,000 live births (Wittington & Holland, 2010; Pober, 2010). Given a vastly greater prevalence of VCFS and Klinefelter
compared to Prader-Willi and Williams it is not surprising they were the conditions more often seen by our sample.

Overall, the prenatal counselors were more likely to discuss the risk for developing a psychiatric manifestation with families. Of the four genetic conditions and their associated psychiatric manifestations, prenatal counselors were most likely to discuss the increased risk of schizophrenia with a diagnosis of VCFS. The link between VCFS and schizophrenia is well established and also well publicized. The wide-spread understanding of the link may explain why prenatal counselors were more likely to discuss the increased risk with parents of affected fetuses. The pediatric genetic counselors were also very likely to discuss the increased incidence of schizophrenia in adults with VCFS. Interestingly, all of the pediatric counselors said that they discuss the risk of ADD either all or some of the time with parents of children with Williams syndrome. ADD is a childhood onset condition. It is possible that the pediatric counselors discuss the risk because they felt comfortable with a less severe condition like ADD that is familiar to many parents. It is also possible that the genetic counselors were more likely to discuss the increased risk to develop ADD with families because parents most likely perceive it as less severe and are not as devastated when hearing of the possibility to later develop ADD. Genetic counselors’ likelihood to discuss ADD with parents of children with Williams may also be due to their own familiarity with the condition and that the children are or may soon be at the age when symptoms would begin to show.
Counselors’ Beliefs Regarding Severity

Both prenatal and pediatric counselors rated psychosis as the most severe of the four mental conditions. Most people would probably consider psychosis to be severe because the symptoms, including delusions and hallucinations, are atypical. Not only are the symptoms of psychosis unpredictable, but due to the lower prevalence it is likely that most people do not have a personal experience with the condition. Due to the rarity and severity of the condition it is not unexpected that psychosis is considered to be the most severe by the counselors.

The prenatal and pediatric counselors all felt that ADD and depression were less severe than psychosis and schizophrenia. Not only are the symptoms of ADD and depression often considered milder, but due to the commonness of the conditions most people may be more comfortable with them. One of the study participants described depression as follows

“To me, depression is not that big of a deal, that is probably very paternalistic of me, but, it’s treatable, it’s very common.”

Since depression and ADD are relatively common in the general population it is likely that most people have had a personal experience with the conditions and it is possible they may find it less severe due to their understanding and experience with the conditions.

Discussion of Psychiatric Manifestations
Prenatal and pediatric counselors reported with all four conditions that they most likely only mention the increased risk of the psychiatric manifestation, but do not go into much depth discussing the symptoms of the condition or the actual risk. Genetic counselors may feel that when a diagnosis is made that there is only so much time to discuss all facets of a condition and therefore only have time to mention the increased risk for a psychiatric manifestation. It may also be due to the comfort level of the counselors. Counselors may not feel confident enough with their understanding of the mental illness to feel comfortable discussing the ins and outs of the psychiatric manifestation and therefore only mention the increased risk. There were a large number of prenatal counselors who stated that they also discuss treatment and preventative options for depression with parents whose fetus has a diagnosis of Klinefelter syndrome. Depression is comparatively well understood when weighed against the other mental illnesses, and successful treatment exists for depression. It is possible that the prenatal counselors are more apt to discuss the risk and treatment options for depression with families of fetuses with Klinefelter so the families have the information and possibly feel less concerned regarding their child's future if it includes depression. Given that treatment and preventative options exist it is not surprising that counselors are more comfortable discussing depression with families of fetuses with Klinefelter syndrome.

Pediatric counselors on the other hand were willing to discuss more aspects of depression and ADD then the prenatal counselors. The pediatric counselors reported they are more likely to discuss the percentage of patients who will develop ADD, discuss
treatment and preventative options for depression and ADD, and discuss the diagnostic criteria and symptoms of depression and ADD then they were for schizophrenia and psychosis. It is possible that pediatric counselors are more likely to discuss these risks because their patients may have been diagnosed with depression or ADD or possibly showing signs or symptoms of the conditions. Both depression and ADD are milder conditions that are prevalent in the general population. Most people are likely to be aware of ADD and depression either through a personal experience, media coverage, or other avenues that counselors and families comfort level with both conditions is most likely much greater. Due to that greater comfort level it is not surprising that counselors discuss more aspects of both depression and ADD with families. One of the counselors stated that in regards to depression they discuss

"I explain that it is a condition that may develop, and that it will need to be considered as the child is growing, as will many other aspects of this condition. I also explain that it is not treated any differently on the basis of the diagnosis."

Interestingly, a large number of pediatric counselors responded that they discuss the percentage of patients with VCFS who will develop schizophrenia. The link between VCFS and schizophrenia is well understood and a large body of research exists surrounding the prevalence of schizophrenia in adults with VCFS. Since the connection has been well researched pediatric counselors' most likely feel confident in the published percentages and therefore feel more confident discussing the percentage of patients with VCFS who will develop schizophrenia.

The vast majority of prenatal and pediatric counselors felt that the most appropriate time to discuss the risk of psychiatric manifestations is at the time of
diagnosis for the genetic condition. It is interesting that the majority of both prenatal and pediatric counselors felt that the risk for a mental illness should be discussed at the time of diagnosis because that seems to be at odds with the reported experience of parents. Sachs (2008) found that most parents learned of the increased risk for multiple psychiatric conditions that have an increased incidence in people with VCFS including schizophrenia via the internet. Most of the parents stated that they did not initially learn about the risks from a medical provider, but that they learned of the risk on their own. Almost half (46.4%) of the parents did state that they would have liked to learn the psychiatric risk information at the time their child was diagnosed with VCFS. It is possible that the parents in Sachs’ study who reported learning about the increased risk for psychiatric illness via the internet were among those that reported never having met with a genetic counselor (approximately 44% of the families in Sachs’ study reported they had never met with a genetic counselor). It is also possible that counseling practices have changed since 2008, and that more counselors are now discussing psychiatric risks. A third possibility is that counselor responses to the current survey were based on what respondents felt was appropriate to report, and not based on their actual practices. Regardless, based on the current findings and those of Sachs it appears that patient reports of their experiences are not entirely consistent with genetic counselor reports of their practices.

The prenatal counselors reported that they would be more likely to discuss genetic testing for a mental illness with parents of fetuses diagnosed with VCFS or Prader-Willi. Both schizophrenia and psychosis are severe mental illnesses that place a
tremendous burden on family members. Based on the severity of the conditions, prenatal counselors may be more willing to discuss testing as another piece of information to be gathered in order to decide how to continue with an affected pregnancy. If testing showed that a fetus would later develop schizophrenia or psychosis, families may choose to not continue the pregnancy. Prenatal counselors may be more willing to discuss testing for a mental illness as another tool to aide parents who are deciding on whether to continue with a pregnancy with a diagnosis of VCFS or Prader-Willi.

Reasons Counselors Don't Discuss the risk for mental illness

Of the counselors who do not discuss the risk to develop a psychiatric manifestation the prenatal counselors reported that the most important reasons for not discussing the increased risk with parents is because the mental illness may never develop and that there are more important symptoms to discuss. Given the large amount of information that prenatal counselors need to discuss and the shorter amount of time they have to discuss it, it is not surprising that they felt that there are more important symptoms to discuss (such as physical manifestations, possible treatment options, recurrence risk, and pregnancy outcomes including termination or adoption) or that they would rather discuss symptoms that are more likely to develop. One of the prenatal counselors described discussing depression prenatally as follows:

“Because depression is so prevalent in the general population, it would feel akin to saying ‘your fetus may one day have high blood pressure’. I could say that to practically any patient, and see no need to alarm them further.”
The pediatric counselors who do not discuss the risk for psychiatric manifestations reported that the most important reason why they do not discuss the increased risk for psychiatric manifestations is because there are more important symptoms to discuss. Often children are diagnosed with a genetic condition after they begin to show symptoms of an illness. It is not surprising that pediatric counselors who do not discuss the increased risk for a mental illness report their reason is because of the importance of other symptoms that the child is most likely experiencing at that moment.

**Provider Responsibility**

Both the prenatal and pediatric counselors felt that it is the geneticist and genetic counselor’s responsibility to discuss the increased risk of psychiatric manifestations with specific diagnoses. Geneticists and genetic counselors’ training focuses on genetic disorders and the symptoms that accompany those conditions, so both would be the most knowledgeable regarding the risk. Interestingly, with Klinefelter and Williams syndrome around one quarter of the prenatal counselors and a minority of pediatric counselors felt that it should be the primary care physician’s (PCP) responsibility to discuss the increased risk for psychiatric manifestations. It is possible that those counselors felt that since both depression and ADD are so common in the general population that PCPs address the issue of both more often than a geneticist or genetic counselor would and therefore they would be the most appropriate provider to address the increased risk.
Comfort with Psychiatric Manifestations

Both the prenatal and pediatric counselors felt most comfortable with their knowledge surrounding depression and ADD. The prenatal and pediatric counselors also reported being most comfortable discussing ADD with families. This was not surprising based on how common the conditions are and also how often they are discussed or covered in the media. It is likely that most people have had personal experiences with these conditions which would also increase the counselors’ comfort with their knowledge of the conditions.

When asked what would help increase counselors’ comfort level when discussing the psychiatric manifestations of genetic diagnoses both the prenatal and pediatric counselors most often reported that if they had more knowledge surrounding the mental illnesses and had more to offer parents like better treatment or preventative options they would feel more comfortable discussing the increased risk with parents. Mental illnesses are complex, ambiguous, variable, and are often unfamiliar to genetic counselors leading to feelings of uncertainty (Peay et al., 2007). Having more knowledge about something is a standard way to feel more comfortable about a topic so it is not surprising that most of the counselors responded this way. Previous research completed by Travassos (2008) found that counselors who participated in educational initiatives had a greater level of knowledge regarding psychiatric genetic counseling and were more comfortable providing the information to patients. Increasing genetic counselors knowledge surrounding mental illness either through training during graduate
programs, immersion in populations with mental illness or through continuing education programs will have a positive impact on their comfort with mental illness.

If counselors had more options to offer families regarding treatment or preventative options then families may not feel so overwhelmed, scared, or troubled by the increased risk and therefore counselors would feel more comfortable discussing the increased risk. Prenatal counselors also felt that if they had the ability to offer predictive testing they would feel more comfortable discussing the risks for mental illness with families. Having the option of predictive testing would offer families another tool to gain important information about their fetus before deciding how to proceed with the pregnancy. For prenatal counselors being able to give parents a potentially crucial piece of information about their fetus not surprisingly would make prenatal counselors feel more comfortable discussing the risk.

Not unexpectedly, the more comfortable the prenatal and pediatric counselors were regarding their knowledge of the mental illness the more likely they were to discuss the increased risk with families. This positive correlation was noted across almost all of the conditions regardless of specialty. The only time a personal experience with a mental illness was found to have a statistically significant impact on the likelihood a counselor discussed the risk for a psychiatric manifestation was when prenatal counselors discussed the risk for depression with Klinefelter syndrome. Given the greater comfort and experience with depression it is not unexpected that the counselors...
had had a personal experience with depression and it lent them to be more comfortable discussing it with families.

A major limitation of this study was the small sample size. Though there are 80 total responses, once the groups were separated based on the specialty there were only about 40 respondents in each group. Though the numbers are large enough to statistically analyze, the small sample may not be representative of the opinions and practices of the larger population of genetic counselors. Another limitation is that we only analyzed the opinions and practices of counselors who had experience working with the specific condition. There were many counselors who were unable to answer a lot of the questions because they did not have a personal experience discussing one or more of the conditions. Given the participants were younger, more recently graduated, and most likely less experienced then the average genetic counselor could explain their lack of experience with the genetic conditions and because they are right out of school and could have covered the recent literature discussing the link between genetics and mental illness may explain their comfort discussing some of the mental illnesses. It may be interesting in future studies to find out how counselors who have not counseled a family regarding a diagnosis would feel about some of these issues.
Conclusion

Mental illness is a disease that is far reaching and extremely common. This study provides an insight into the opinions and practices of prenatal and pediatric genetic counselors when it comes to the psychiatric manifestations that are features of a genetic disorder. Overall, most counselors feel more comfortable with and were more likely to discuss conditions such as depression and ADD that may be perceived as milder than conditions such as schizophrenia and psychosis. Though the counselors stated they felt comfortable discussing the increased risk with families, the topics the counselors were most likely to discuss were often limited to just mentioning the increased risk and not going into detail regarding the symptoms, treatment or preventative options, etc.

Previous research has shown that parents of children with VCFS prefer to learn about the risk of schizophrenia from a medical provider and at the time of diagnosis. These parents felt that if they had the information earlier they would be better equipped to recognize the onset of symptoms sooner and begin treatment earlier. Our respondents agreed that the appropriate time to discuss the increased risk was at the time of diagnosis. It is possible that if parents had the information regarding the increased risk for psychiatric manifestations they would be more comfortable with the possible risk and more comfortable living and coping with a mental illness.
In order to increase counselors’ comfort level with mental illness and therefore improve the counseling they can offer families there needs to be an increase in knowledge and training surrounding psychiatric conditions. This needs to begin during the graduate training with increased exposure to mental illness through lectures, placements, speakers, etc. It is possible that by exposing students to the information earlier they will gain a greater understanding of the conditions and therefore feel more comfortable approaching and discussing the increased risk for psychiatric manifestations with families.

Future research should focus on the lack of training surrounding mental illness in graduate programs and how this can be addressed and corrected. Research should also investigate other providers, i.e. primary care physicians, psychiatrists and psychologists, understanding of the connection between genetic disorders and psychiatric manifestations. Given many of the counselors felt that these other providers should be responsible for discussing the increased risk, it would be helpful to explore whether healthcare providers in general are aware of the risk, are comfortable discussing the risk, and feel able to manage the psychiatric symptoms before we expect or rely on them to discuss the increased risk.
References


Appendix A: Recruitment Notice

Subject Line of Email to NSGC Listserv: Invitation to Participate in a Study about Psychiatric Manifestations of Genetic Disease

Text of Recruitment Notice:

My name is Meghan Jackson, and I am a second year student in the Brandeis University Genetic Counseling Program. For my master’s thesis, I am conducting a research study to learn about genetic counselors’ opinions and practices when presenting information regarding psychiatric manifestations that are possible symptoms of childhood onset genetic illnesses. If you are a pediatric or prenatal genetic counselor with at least one year of clinical experience, you are invited to participate in this study, which consists of an anonymous, online survey that will take approximately 20 minutes to complete. Participants will have the opportunity to enter a raffle for one of three $25 gift certificates. If you are interested in participating, please click on the following link to access the survey: http://survey.qualtrics.com/SE/?SID=SV_efltWbM2hHTTsUY.

If you have questions, please contact me at: mjackson@brandeis.edu.
Appendix B: Introduction to Survey

Dear Genetic Counselor,

Thank you for your interest in participating in my master’s thesis research study. The purpose of the study is to investigate genetic counselors’ opinions and practices when presenting information regarding psychiatric manifestations that are possible symptoms of childhood onset genetic illnesses. I am also interested in learning more about methods that could help to increase counselors’ comfort level when counseling about the risk for mental illnesses associated with a genetic diagnosis. The study is an anonymous online survey that will take approximately 20 minutes to complete. If you are a prenatal or pediatric counselor with at least one year of clinical experience you are eligible to participate. Participation is voluntary. The Principle Investigator for the project is Brandeis University Genetic Counseling Program faculty member Beth Rosen-Sheidley, MS, CGC. If you have any questions about the project please contact me, Meghan Jackson, the Student Researcher, at mjackson@brandeis.edu. If you have questions about your rights as a research subject please contact the Brandeis Institutional Review Board at irb@brandeis.edu or 781-736-8133.
Appendix C: Survey

Q1 What is your age range in years?

- 20-24 (1)
- 25-29 (2)
- 30-34 (3)
- 35-39 (4)
- 40-44 (5)
- 45-49 (6)
- 50-54 (7)
- 55-59 (8)
- 60-64 (9)
- 65-69 (10)
- 70-74 (11)
- 75+ (12)

Q2 What is your gender?

- Male (1)
- Female (2)
- Unknown (3)

Q3 What region do you practice in?

- 1 (CT, MA, ME, NH, RI, VT, CN Maritime Provinces) (1)
- 2 (DC, DE, MD, NJ, NY, PA, VA, WV, PR, VI, Quebec) (2)
- 3 (AL, FL, GA, KY, LA, MS, NC, SC, TN) (3)
- 4 (AR, IA, IL, IN, KS, MI, MN, MO, ND, NE, OH, OK, SD, WI, Ontario) (4)
- 5 (AZ, CO, MT, NM, TX, UT, WY, Alberta, Manitoba, Sask.) (5)
- 6 (AK, CA, HI, ID, NV, OR, WA, British Columbia) (6)
Q4 What year did you graduate from a Genetic Counseling Program?

- 1970-74 (1)
- 1975-79 (2)
- 1980-84 (3)
- 1985-89 (4)
- 1990-94 (5)
- 1995-99 (6)
- 2000-05 (7)
- 2006-2009 (8)

Q5 Have you, a family member, or close friend ever been diagnosed with a psychiatric illness such as depression, schizophrenia, ADD/ADHD or psychosis?

- Yes (1)
- No (2)

Q6 Do you currently see patients clinically (either full time or part time)?

- Yes (1)
- No (2)

If No Is Selected, Then Skip To End of Survey
If Yes Is Selected, Then Skip To Have you worked with patients clinically...

Q7 Have you worked with patients clinically for at least 1 year?

- Yes (1)
- No (2)

If No Is Selected, Then Skip To End of Survey
If Yes Is Selected, Then Skip To What is your primary specialty area?
Q8 What is your primary specialty area?

- Prenatal genetics (1)
- Pediatric genetics (2)
- Cancer genetics (3)
- Psychiatric genetics (4)
- Other adult genetics (5)
- Other (6)

If Cancer genetics is selected, then skip to end of survey. If Psychiatric genetics is selected, then skip to end of survey. If Other adult genetics is selected, then skip to end of survey. If Other is selected, then skip to end of survey.

Q76 About how many families of patients with fetuses with Velo-cardio-facial syndrome (VCFS) have you counseled?

- 0 (1)
- 1-4 (2)
- 5-8 (3)
- 9-12 (4)
- 13+ (5)

If 0 is selected, then skip to about how many families of children w...

Q77 In your opinion, how severe a condition is schizophrenia?

- Not very severe (1)
- (2)
- (3)
- (4)
- (5)
- (6)
- Very severe (7)
Q78 Individuals with VCFS have a higher risk of developing schizophrenia. When a fetus is diagnosed with VCFS, do you discuss with the parents the possibility for the fetus to later develop schizophrenia?

- Yes (1)
- No (2)
- Sometimes (3)

If No Is Selected, Then Skip To When do you feel is the most appropri...

Q79 Which of the following, regarding schizophrenia, do you discuss with parents of fetuses with VCFS? (select all that apply)

- Mention the increased possibility without discussing specific numbers (1)
- Discuss the percentage of patients with VCFS who will later develop schizophrenia (2)
- Talk about the natural history of schizophrenia (3)
- Discuss possible treatments or prevention (4)
- Discuss diagnostic criteria/symptoms (5)
- Other (6)

Answer If Which of the following, regarding schizophrenia, do you d... Other Is Selected

Q80 Please specify

Q81 When do you feel is the most appropriate time to discuss with parents of a fetus with VCFS the increased risk for their fetus to later develop schizophrenia?

- At the time of diagnosis of VCFS (1)
- At a later point in pregnancy (2)
- At birth (3)
- After all of the physical manifestations of VCFS are being well managed (4)
- Later in childhood (5)
- In adolescence (6)
- At the time of diagnosis for schizophrenia, if the fetus is ever diagnosed with schizophrenia (7)
- There is no appropriate time (8)
- Other (9)
Q82 Please specify

Q83 If testing were available for patients with VCFS that could predict if the fetus would later develop schizophrenia, how likely would you be to discuss it with parents?

- Very Unlikely (1)
- (2)
- (3)
- (4)
- (5)
- (6)
- Very Likely (7)

Q84 How comfortable do you feel discussing with parents of fetuses with VCFS the increased risk for their fetus to develop schizophrenia?

- Not at all comfortable (1)
- (2)
- (3)
- (4)
- (5)
- (6)
- Very comfortable (7)

Q85 How comfortable are you with your knowledge about schizophrenia?

- Not at all comfortable (1)
- (2)
- (3)
- (4)
- (5)
- (6)
- Very comfortable (7)
Q86 What would/could help to increase your comfort to discuss the increased chance to develop schizophrenia with a diagnosis of VCFS? (select all that apply)

- More knowledge surrounding schizophrenia and the increased risk (1)
- More training about mental illness (2)
- More to offer parents like better treatment/prevention options (3)
- Ability to offer predictive genetic testing (4)
- Nothing (5)
- Other (6)

Answer If What would/could help to increase your comfort to discuss... Other Is Selected

Q87 Please explain

Answer If Individuals with VCFS have a higher risk of developing sc... No Is Selected Or Individuals with VCFS have a higher risk of developing sc... Sometimes Is Selected

Q88 Why do you not discuss with parents of a fetus with VCFS the increased chance for their fetus to later on develop schizophrenia? (select all that apply)

- Fetus may never develop schizophrenia (1)
- More important possible symptoms to discuss (2)
- Lack of comfort discussing schizophrenia (3)
- Another provider’s responsibility to discuss (4)
- Other (5)

Answer If Why do you not discuss with parents of a&nbsp;fetus with ... Other Is Selected

Q89 Please explain
Q90 In your opinion, whose responsibility is it to discuss with parents of a fetus with VCFS the increased chance for the fetus to later develop schizophrenia?

- Primary Care Physician (1)
- Geneticist (2)
- Genetic Counselor (3)
- Psychiatrist/Psychologist (4)
- Other (5)

Q91 Please specify

Q92 If testing were available for patients with VCFS that could predict if a fetus would later develop schizophrenia how likely would it change if you would discuss with their parents the increased chance to develop schizophrenia?

- Very Unlikely (1)
- (2)
- (3)
- (4)
- (5)
- (6)
- Very Likely (7)
Q93 About how many families of patients with fetuses with Klinefelter syndrome have you counseled?

- 0 (1)
- 1-4 (2)
- 5-8 (3)
- 9-12 (4)
- 13+ (5)

If 0 Is Selected, Then Skip To About how many families of children w...

Q94 In your opinion, how severe a condition is depression?

- Not very severe (1)
- (2)
- (3)
- (4)
- (5)
- (6)
- Very severe (7)

Q95 Individuals with Klinefelter syndrome have a higher risk of developing depression. When a fetus is diagnosed with Klinefelter, do you discuss with the parents the possibility for the fetus to later develop depression?

- Yes (1)
- No (2)
- Sometimes (3)

If No Is Selected, Then Skip To When do you feel is the most appropri...
Q96 Which of the following, regarding depression, do you discuss with parents? (select all that apply)

- Mention the increased possibility without discussing specific numbers (1)
- Discuss the percentage of patients with Klinefelter syndrome who will later develop depression (2)
- Talk about the natural history of depression (3)
- Discuss possible treatments or prevention (4)
- Discuss diagnostic criteria/symptoms (5)
- Other (6)

Answer If Which of the following, regarding depression, do you disc... Other Is Selected

Q97 Please explain

Q98 When do you feel is the most appropriate time to discuss with parents of a fetus with Klinefelter syndrome the increased chance for their fetus to later develop depression?

- At the time of diagnosis of Klinefelter (1)
- At a later point in pregnancy (2)
- At birth (3)
- After all of the physical manifestations of Klinefelter are being well managed (4)
- Later in childhood (5)
- In adolescence (6)
- At the time of diagnosis for depression, if the fetus is ever diagnosed with depression (7)
- There is no appropriate time (8)
- Other (9)

Answer If When do you feel is the most appropriate time to discuss ... Other Is Selected

Q99 Please explain
Q100 If testing were available for patients with Klinefelter that could predict if a fetus would later develop depression, how likely would you be to discuss it with parents?

- Very Unlikely (1)
- (2)
- (3)
- (4)
- (5)
- (6)
- Very Likely (7)

Q101 How comfortable do you feel discussing with parents of fetuses with Klinefelter the increased risk for their fetus to develop depression?

- Not at all comfortable (1)
- (2)
- (3)
- (4)
- (5)
- (6)
- Very comfortable (7)

Q102 How comfortable are you with your knowledge about depression?

- Not at all comfortable (1)
- (2)
- (3)
- (4)
- (5)
- (6)
- Very comfortable (7)
Q103 What would/could help to increase your comfort to discuss the increased chance to developing depression with a diagnosis of Klinefelter syndrome? (select all that apply)

- More knowledge surrounding depression and the increased risk (1)
- More training about mental illness (2)
- More to offer parents like better treatment/prevention options (3)
- Ability to offer predictive testing (4)
- Nothing (5)
- Other (6)

**Answer If** What would/could help to increase your comfort to discuss... Other Is Selected

Q104 Please explain

**Answer If** Individuals with Klinefelter syndrome have a higher risk... No Is Selected Or Individuals with Klinefelter syndrome have a higher risk... Sometimes Is Selected

Q105 Why do you not discuss with parents of fetuses with Klinefelter syndrome the increased chance for the fetus to later on develop depression? (select all that apply)

- Fetus may never develop depression (1)
- More important possible symptoms to discuss (2)
- Lack of comfort discussing depression (3)
- Another providers responsibility to discuss (4)
- Other (5)

**Answer If** Why do you not discuss with parents of children with Klin... Other Is Selected

Q106 Please specify
Q107 Whose responsibility is it to discuss with parents of a fetus with Klinefelter syndrome the increased chance for the fetus to later develop depression?

- Primary Care Physician (1)
- Geneticist (2)
- Genetic Counselor (3)
- Psychiatrist/Psychologist (4)
- Other (5)

Q108 Please specify

Q109 If testing were available for patients with Klinefelter that could predict if a fetus would later develop depression how likely would it change if you would discuss with their parents the increased chance to develop depression?

- Very Unlikely (1)
- (2)
- (3)
- (4)
- (5)
- (6)
- Very Likely (7)
Q110 About how many families of patients with fetuses with Prader-Willi syndrome have you counseled?

- 0 (1)
- 1-4 (2)
- 5-8 (3)
- 9-12 (4)
- 13+ (5)

If 0 Is Selected, Then Skip To About how many families of children w...

Q111 In your opinion, how severe a condition is psychosis?

- Not very severe (1)
- (2)
- (3)
- (4)
- (5)
- (6)
- Very severe (7)

Q112 Individuals with Prader-Willi have a higher risk of developing psychosis. Psychosis is characterized by an inability to think and communicate effectively, understand reality, and behave appropriately. Symptoms of psychosis include hallucinations and delusions. When a fetus is diagnosed with Prader-Willi, do you discuss with the parents the possibility for the fetus to later develop psychosis?

- Yes (1)
- No (2)
- Sometimes (3)

If No Is Selected, Then Skip To When do you feel is the most appropri...
Q113 Which of the following, regarding psychosis, do you discuss with parents of fetuses with Prader-Willi? (select all that apply)

- Mention the increased possibility without discussing specific numbers (1)
- Discuss the percentage of patients with Prader-Willi who will later develop psychosis (2)
- Talk about the natural history of psychosis (3)
- Discuss possible treatments or prevention (4)
- Discuss diagnostic criteria/symptoms (5)
- Other (6)

**Answer If Which of the following, regarding psychosis, do you discuss... Other Is Selected**

Q114 Please specify

Q115 When do you feel is the most appropriate time to discuss with parents of a fetus with Prader-Willi the increased chance for their fetus to later develop psychosis?

- At the time of diagnosis of Prader-Willi (1)
- At a later point in pregnancy (2)
- At birth (3)
- After all of the physical manifestations of Prader-Willi are being well managed (4)
- Later in childhood (5)
- In adolescence (6)
- At the time of diagnosis for psychosis, if the child is ever diagnosed with psychosis (7)
- There is no appropriate time (8)
- Other (9)

**Answer If When do you feel is the most appropriate time to discuss... Other Is Selected**

Q116 Please specify
Q117 If testing were available for patients with Prader-Willi that could predict if a fetus would later develop psychosis how likely would you be to discuss it with parents?

- Very Unlikely (1)
- (2)
- (3)
- (4)
- (5)
- (6)
- Very Likely (7)

Q118 How comfortable do you feel discussing with parents of fetuses with Prader-Willi the increased risk for their fetus to develop psychosis?

- Not at all comfortable (1)
- (2)
- (3)
- (4)
- (5)
- (6)
- Very comfortable (7)

Q119 How comfortable are you with your knowledge about psychosis?

- Not at all comfortable (1)
- (2)
- (3)
- (4)
- (5)
- (6)
- Very comfortable (7)
Q120 What would/could help to increase your comfort to discuss the increased chance to develop psychosis with a diagnosis of Prader-Willi? (select all that apply)

- More knowledge surrounding psychosis and the increased risk (1)
- More training about mental illness (2)
- More to offer parents like better treatment/prevention options (3)
- Ability to offer predictive genetic testing (4)
- Nothing (5)
- Other (6)

**Answer If What would/could help to increase your comfort to discuss... Other Is Selected**

Q142 Please explain

**Answer If Individuals with Prader-Willi have a higher risk of develop... No Is Selected Or Individuals with Prader-Willi have a higher risk of develop... Sometimes Is Selected**

Q121 Why do you not discuss with parents of a fetus with Prader-Willi the increased chance for the fetus to later on develop psychosis? (select all that apply)

- Fetus may never develop psychosis (1)
- More important possible symptoms to discuss (2)
- Lack of comfort discussing psychosis (3)
- Another providers responsibility to discuss (4)
- Other (5)

**Answer If Why do you not discuss with parents of a fetus with ... Other Is Selected**

Q143 Please explain
Q122 Whose responsibility is it to discuss with parents of a fetus with Prader-Willi the increased risk for the fetus to later develop psychosis?

- Primary Care Physician (1)
- Geneticist (2)
- Genetic Counselor (3)
- Psychiatrist/Psychologist (4)
- Other (5)

Q144 Please specify

Q123 If testing were available for patients with Prader-Willi that could predict if a fetus would later develop psychosis how likely would it change if you would discuss with their parents the increased chance to develop psychosis?

- Very Unlikely (1)
- (2)
- (3)
- (4)
- (5)
- (6)
- Very Likely (7)
Q124 About how many families of patients with fetuses with Williams syndrome have you counseled?

- 0 (1)
- 1-4 (2)
- 5-8 (3)
- 9-12 (4)
- 13+ (5)

If 0 Is Selected, Then Skip To End of Survey

Q125 In your opinion, how severe a condition is ADD?

- Not very severe (1)
- (2)
- (3)
- (4)
- (5)
- (6)
- Very severe (7)

Q126 Individuals with Williams have a higher risk of developing attention deficit disorder (ADD). ADD is characterized by an inability to pay attention, impulsivity, and hyperactivity. When a fetus is diagnosed with Williams, do you discuss with the parents the possibility for the fetus to later develop ADD?

- Yes (1)
- No (2)
- Sometimes (3)

If No Is Selected, Then Skip To When do you feel is the most appropri...
Q127 Which of the following, regarding ADD, do you discuss with parents of fetuses with Williams? (select all that apply)

- Mention the increased possibility without discussing specific numbers (1)
- Discuss the percentage of patients with Williams who will later develop ADD (2)
- Talk about the natural history of ADD (3)
- Discuss possible treatments or prevention (4)
- Discuss diagnostic criteria/symptoms (5)
- Other (6)

Answer If Which of the following, regarding ADD, do you discuss with parents of fetuses... Other Is Selected

Q128 Please explain

Q129 When do you feel is the most appropriate time to discuss with parents of a fetus with Williams the increased risk for their fetus to later develop ADD?

- At the time of diagnosis of Williams (1)
- At a later point in pregnancy (2)
- At birth (3)
- After all of the physical manifestations of Williams are being well managed (4)
- Later in childhood (5)
- In adolescence (6)
- At the time of diagnosis for ADD, if the fetus is ever diagnosed with ADD (7)
- There is no appropriate time (8)
- Other (9)

Answer If When do you feel is the most appropriate time to discuss ... Other Is Selected

Q130 Please explain
Q131 If testing were available for patients with Williams that could predict if a fetus would later develop ADD how likely would you be to discuss it with parents?

- Very Unlikely (1)
- (2)
- (3)
- (4)
- (5)
- (6)
- Very Likely (7)

Q132 How comfortable do you feel discussing with parents of fetuses with Williams syndrome the increased risk for their fetus to develop ADD?

- Not at all comfortable (1)
- (2)
- (3)
- (4)
- (5)
- (6)
- Very comfortable (7)

Q133 How comfortable are you with your knowledge about ADD?

- Not at all comfortable (1)
- (2)
- (3)
- (4)
- (5)
- (6)
- Very comfortable (7)
Q134 What would/could help to increase your comfort to discuss the increased chance to develop ADD with a diagnosis of Williams syndrome? (select all that apply)

- More knowledge surrounding ADD and the increased risk (1)
- More training about mental illness (2)
- More to offer parents like better treatment/prevention options (3)
- Ability to offer predictive genetic testing (4)
- Nothing (5)
- Other (6)

**Answer If What would/could help to increase your comfort to discuss... Other Is Selected**

Q135 Please explain

**Answer If Individuals with Williams have a higher risk of developing... No Is Selected Or Individuals with Williams have a higher risk of developing... Sometimes Is Selected**

Q136 Why do you not discuss with parents of a fetus with Williams syndrome the increased chance for the fetus to later on develop ADD? (select all that apply)

- Fetus may never develop ADD (1)
- More important possible symptoms to discuss (2)
- Lack of comfort discussing ADD (3)
- Another providers responsibility to discuss (4)
- Other (5)

**Answer If Why do you not discuss with parents of a child with Willi... Other Is Selected**

Q137 Please explain
Q138 Whose responsibility is it to discuss with parents of a fetus with Williams syndrome the increased chance for the fetus to later develop ADD?

- Primary Care Physician (1)
- Geneticist (2)
- Genetic Counselor (3)
- Psychiatrist/Psychologist (4)
- Other (5)

Q139 Please specify

Q140 If testing were available for patients with Williams that could predict if a fetus would later develop ADD how likely would it change if you would discuss with their parents the increased chance to develop ADD?

- Very Unlikely (1)
- Very Unlikely (2)
- Very Unlikely (3)
- Very Unlikely (4)
- Very Unlikely (5)
- Very Unlikely (6)
- Very Likely (7)
Q9 About how many families of children with Velo-cardio-facial syndrome (VCFS) have you counseled?

- 0 (1)
- 1-4 (2)
- 5-8 (3)
- 9-12 (4)
- 13+ (5)

If 0 Is Selected, Then Skip To About how many families of children w...

Q10 In your opinion, how severe a condition is schizophrenia?

- Not very severe (1)
- (2)
- (3)
- (4)
- (5)
- (6)
- Very Severe (7)

Q11 Individuals with VCFS have a higher risk of developing schizophrenia. When a child is diagnosed with VCFS, do you discuss with the child’s parents the possibility for the child to later develop schizophrenia?

- Yes (1)
- No (2)
- Sometimes (3)

If No Is Selected, Then Skip To When do you feel is the most appropri...
Q12 Which of the following, regarding schizophrenia, do you discuss with parents of children with VCFS? (select all that apply)

- Mention the increased possibility without discussing specific numbers (1)
- Discuss the percentage of patients with VCFS who will later develop schizophrenia (2)
- Talk about the natural history of schizophrenia (3)
- Discuss possible treatments or prevention (4)
- Discuss diagnostic criteria/symptoms (5)
- Other (6)

**Answer If Which of the following, regarding schizophrenia, do you d... Other Is Selected**

Q13 Please specify

Q14 When do you feel is the most appropriate time to discuss with parents of a child with VCFS the increased risk for their child to later develop schizophrenia?

- At the time of diagnosis of VCFS (1)
- After all of the physical manifestations of VCFS are being well managed (2)
- Later in childhood (3)
- In adolescence (4)
- At the time of diagnosis for schizophrenia, if the child is ever diagnosed with schizophrenia (5)
- There is no appropriate time (6)
- Other (7)

**Answer If When do you feel is the most appropriate time to discuss ... Other Is Selected**

Q15 Please specify
Q16 If testing were available for patients with VCFS that could predict if the child would later develop schizophrenia, how likely would you be to discuss it with parents?

- Very Unlikely (1)
- (2)
- (3)
- (4)
- Very Likely (5)

Q17 How comfortable do you feel discussing with parents of children with VCFS the increased risk for their child to develop schizophrenia?

- Not at all comfortable (1)
- (2)
- (3)
- (4)
- (5)
- (6)
- Very Comfortable (7)

Q18 How comfortable are you with your knowledge about schizophrenia?

- Not at all comfortable (1)
- (2)
- (3)
- (4)
- (5)
- (6)
- Very Comfortable (7)
Q19 What would/could help to increase your comfort to discuss the increased chance to develop schizophrenia with a diagnosis of VCFS? (select all that apply)

- More knowledge surrounding schizophrenia and the increased risk (1)
- More training about mental illness (2)
- More to offer parents like better treatment/prevention options (3)
- Ability to offer predictive genetic testing (4)
- Nothing (5)
- Other (6)

Answer If What would/could help to increase your comfort to discuss... Other Is Selected

Q20 Please explain

Answer If Individuals with VCFS have a higher risk of developing sc... No Is Selected Or Individuals with VCFS have a higher risk of developing sc... Sometimes Is Selected

Q21 Why do you not discuss with parents of a child with VCFS the increased chance for their child to later on develop schizophrenia? (select all that apply)

- Child may never develop schizophrenia (1)
- More important possible symptoms to discuss (2)
- Lack of comfort discussing schizophrenia (3)
- Another provider’s responsibility to discuss (4)
- Other (5)

Answer If Why do you not discuss with parents of a child with VCFS ... Other Is Selected

Q22 Please explain
Q23 In your opinion, whose responsibility is it to discuss with parents of a child with VCFS the increased chance for the child to later develop schizophrenia?

- Primary Care Physician (1)
- Geneticist (2)
- Genetic Counselor (3)
- Psychiatrist/Psychologist (4)
- Other (5)

Q24 Please specify

Q25 If testing were available for patients with VCFS that could predict if a child would later develop schizophrenia how likely would it change if you would discuss with their parents the increased chance to develop schizophrenia?

- Very Unlikely (1)
- (2)
- (3)
- (4)
- (5)
- (6)
- Very Likely (7)

Q26 About how many families of children with Klinefelter syndrome have you counseled?

- 0 (1)
- 1-4 (2)
- 5-8 (3)
- 9-12 (4)
- 13+ (5)

If 0 Is Selected, Then Skip To About how many families of children w...
Q27 In your opinion, how severe a condition is depression?

- Not very severe (1)
- (2)
- (3)
- (4)
- (5)
- (6)
- Very Severe (7)

Q28 Individuals with Klinefelter syndrome have a higher risk of developing depression. When a child is diagnosed with Klinefelter, do you discuss with the child’s parents the possibility for the child to later develop depression?

- Yes (1)
- No (2)
- Sometimes (3)

If No Is Selected, Then Skip To When do you feel is the most appropri...

Q29 Which of the following, regarding depression, do you discuss with parents? (select all that apply)

- Mention the increased possibility without discussing specific numbers (1)
- Discuss the percentage of patients with Klinefelter syndrome who will later develop depression (2)
- Talk about the natural history of depression (3)
- Discuss possible treatments or prevention (4)
- Discuss diagnostic criteria/symptoms (5)
- Other (6)

Answer If Which of the following, regarding depression, do you disc... Other Is Selected

Q30 Please explain
Q31 When do you feel is the most appropriate time to discuss with parents of a child with Klinefelter syndrome the increased chance for their child to later develop depression?

- At the time of diagnosis of Klinefelter (1)
- After all of the physical manifestations of Klinefelter are being well managed (2)
- Later in childhood (3)
- In adolescence (4)
- At the time of diagnosis for depression, if the child is ever diagnosed with depression (5)
- There is no appropriate time (6)
- Other (7)

**Answer If When do you feel is the most appropriate time to discuss ... Other Is Selected**

Q32 Please explain

Q33 If testing were available for patients with Klinefelter that could predict if a child would later develop depression, how likely would you be to discuss it with parents?

- Very Unlikely (1)
- (2)
- (3)
- (4)
- (5)
- (6)
- Very Likely (7)

Q34 How comfortable do you feel discussing with parents of children with Klinefelter the increased risk for their child to develop depression?

- Not at all comfortable (1)
- (2)
- (3)
- (4)
- (5)
- (6)
- Very comfortable (7)
Q35 How comfortable are you with your knowledge about depression?

- Not at all comfortable (1)
- (2)
- (3)
- (4)
- (5)
- (6)
- Very comfortable (7)

Q36 What would/could help to increase your comfort to discuss the increased chance to developing depression with a diagnosis of Klinefelter syndrome? (select all that apply)

- More knowledge surrounding depression and the increased risk (1)
- More training about mental illness (2)
- More to offer parents like better treatment/prevention options (3)
- Ability to offer predictive testing (4)
- Nothing (5)
- Other (6)

Answer If What would/could help to increase your comfort to discuss... Other Is Selected

Q37 Please explain

Answer If Individuals with Klinefelter syndrome have a higher risk ... No Is Selected Or Individuals with Klinefelter syndrome have a higher risk ... Sometimes Is Selected

Q38 Why do you not discuss with parents of children with Klinefelter syndrome the increased chance for the child to later on develop depression? (select all that apply)

- Child may never develop depression (1)
- More important possible symptoms to discuss (2)
- Lack of comfort discussing depression (3)
- Another providers responsibility to discuss (4)
- Other (5)
Answer If Why do you not discuss with parents of children with Klin... Other Is Selected

Q39 Please specify

Answer If Individuals with Klinefelter syndrome have a higher risk ... No Is Selected Or Individuals with Klinefelter syndrome have a higher risk ... Sometimes Is Selected

Q40 Whose responsibility is it to discuss with parents of a child with Klinefelter syndrome the increased chance for the child to later develop depression?

- Primary Care Physician (1)
- Geneticist (2)
- Genetic Counselor (3)
- Psychiatrist/Psychologist (4)
- Other (5)

Answer If Whose responsibility is it to discuss with parents of a c... Other Is Selected

Q41 Please specify

Answer If Individuals with Klinefelter syndrome have a higher risk ... No Is Selected Or Individuals with Klinefelter syndrome have a higher risk ... Sometimes Is Selected

Q42 If testing were available for patients with Klinefelter that could predict if a child would later develop depression how likely would it change if you would discuss with their parents the increased chance to develop depression?

- Very Unlikely (1)
- (2)
- (3)
- (4)
- (5)
- (6)
- Very Likely (7)
Q43 About how many families of children with Prader-Willi syndrome have you counseled?

- 0 (1)
- 1-4 (2)
- 5-8 (3)
- 9-12 (4)
- 13+ (5)

If 0 Is Selected, Then Skip To About how many families of children w...

Q44 In your opinion, how severe a condition is psychosis?

- Not very severe (1)
- (2)
- (3)
- (4)
- (5)
- (6)
- Very severe (7)

Q45 Individuals with Prader-Willi have a higher risk of developing psychosis. Psychosis is characterized by an inability to think and communicate effectively, understand reality, and behave appropriately. Symptoms of psychosis include hallucinations and delusions. When a child is diagnosed with Prader-Willi, do you discuss with the child’s parents the possibility for the child to later develop psychosis?

- Yes (1)
- No (2)
- Sometimes (3)

If No Is Selected, Then Skip To When do you feel is the most appropri...
Q46 Which of the following, regarding psychosis, do you discuss with parents of children with Prader-Willi? (select all that apply)

- Mention the increased possibility without discussing specific numbers (1)
- Discuss the percentage of patients with Prader-Willi who will later develop psychosis (2)
- Talk about the natural history of psychosis (3)
- Discuss possible treatments or prevention (4)
- Discuss diagnostic criteria/symptoms (5)
- Other (6)

Answer If Which of the following, regarding psychosis, do you discu... Other Is Selected

Q47 Please specify

Q48 When do you feel is the most appropriate time to discuss with parents of a child with Prader-Willi the increased chance for their child to later develop psychosis?

- At the time of diagnosis of Prader-Willi (1)
- After all of the physical manifestations of Prader-Willi are being well managed (2)
- Later in childhood (3)
- In adolescence (4)
- At the time of diagnosis for psychosis, if the child is ever diagnosed with psychosis (5)
- There is no appropriate time (6)
- Other (7)

Answer If When do you feel is the most appropriate time to discuss ... Other Is Selected

Q49 Please specify
Q50 If testing were available for patients with Prader-Willi that could predict if a child would later develop psychosis how likely would you be to discuss it with parents?

- Very Unlikely (1)
- (2)
- (3)
- (4)
- (5)
- (6)
- Very Likely (7)

Q51 How comfortable do you feel discussing with parents of children with Prader-Willi the increased risk for their child to develop psychosis?

- Not at all comfortable (1)
- (2)
- (3)
- (4)
- (5)
- (6)
- Very comfortable (7)

Q52 How comfortable are you with your knowledge about psychosis?

- Not at all comfortable (1)
- (2)
- (3)
- (4)
- (5)
- (6)
- Very comfortable (7)
Q53 What would/could help to increase your comfort to discuss the increased chance to develop psychosis with a diagnosis of Prader-Willi? (select all that apply)

- More knowledge surrounding psychosis and the increased risk (1)
- More training about mental illness (2)
- More to offer parents like better treatment/prevention options (3)
- Ability to offer predictive genetic testing (4)
- Nothing (5)
- Other (6)

Answer If What would/could help to increase your comfort to discuss... Other Is Selected
Q139 Please explain

Answer If Individuals with Prader-Willi have a higher risk of developing psychosis...
Q55 Why do you not discuss with parents of a child with Prader-Willi the increased chance for the child to later on develop psychosis? (select all that apply)

- Child may never develop psychosis (1)
- More important possible symptoms to discuss (2)
- Lack of comfort discussing psychosis (3)
- Another providers responsibility to discuss (4)
- Other (5)

Answer If Why do you not discuss with parents of a child with Prade... Other Is Selected
Q140 Please explain
Q57 Whose responsibility is it to discuss with parents of a child with Prader-Willi the increased risk for the child to later develop psychosis?

- Primary Care Physician (1)
- Geneticist (2)
- Genetic Counselor (3)
- Psychiatrist/Psychologist (4)
- Other (5)

Q141 Please specify

Q59 If testing were available for patients with Prader-Willi that could predict if a child would later develop psychosis how likely would it change if you would discuss with their parents the increased chance to develop psychosis?

- Very Unlikely (1)
- (2)
- (3)
- (4)
- (5)
- (6)
- Very Likely (7)

Q60 About how many families of children with Williams syndrome have you counseled?

- 0 (1)
- 1-4 (2)
- 5-8 (3)
- 9-12 (4)
- 13+ (5)

If 0 Is Selected, Then Skip To End of Survey
Q61 In your opinion, how severe a condition is ADD?

- Not very severe (1)
- (2)
- (3)
- (4)
- (5)
- (6)
- Very severe (7)

Q62 Individuals with Williams have a higher risk of developing attention deficit disorder (ADD). ADD is characterized by an inability to pay attention, impulsivity, and hyperactivity. When a child is diagnosed with Williams, do you discuss with the child’s parents the possibility for the child to later develop ADD?

- Yes (1)
- No (2)
- Sometimes (3)

If No Is Selected, Then Skip To When do you feel is the most appropri...

Q63 Which of the following, regarding ADD, do you discuss with parents of children with Williams? (select all that apply)

- Mention the increased possibility without discussing specific numbers (1)
- Discuss the percentage of patients with Williams who will later develop ADD (2)
- Talk about the natural history of ADD (3)
- Discuss possible treatments or prevention (4)
- Discuss diagnostic criteria/symptoms (5)
- Other (6)

Answer If Which of the following, regarding ADD, do you discuss wit... Other Is Selected

Q64 Please explain
Q65 When do you feel is the most appropriate time to discuss with parents of a child with Williams the increased risk for their child to later develop ADD?

- At the time of diagnosis of Williams (1)
- After all of the physical manifestations of Williams are being well managed (2)
- Later in childhood (3)
- In adolescence (4)
- At the time of diagnosis for ADD, if the child is ever diagnosed with ADD (5)
- There is no appropriate time (6)
- Other (7)

**Answer If When do you feel is the most appropriate time to discuss ... Other Is Selected**

Q66 Please explain

Q67 If testing were available for patients with Williams that could predict if a child would later develop ADD how likely would you be to discuss it with parents?

- Very Unlikely (1)
- (2)
- (3)
- (4)
- (5)
- (6)
- Very Likely (7)

Q68 How comfortable do you feel discussing with parents of children with Williams syndrome the increased risk for their child to develop ADD?

- Not at all comfortable (1)
- (2)
- (3)
- (4)
- (5)
- (6)
- Very comfortable (7)
Q69 How comfortable are you with your knowledge about ADD?

- Not at all comfortable (1)
- (2)
- (3)
- (4)
- (5)
- (6)
- Very Comfortable (7)

Q70 What would/could help to increase your comfort to discuss the increased chance to develop ADD with a diagnosis of Williams syndrome? (select all that apply)

- More knowledge surrounding ADD and the increased risk (1)
- More training about mental illness (2)
- More to offer parents like better treatment/prevention options (3)
- Ability to offer predictive genetic testing (4)
- Nothing (5)
- Other (6)

**Answer If What would/could help to increase your comfort to discuss... Other Is Selected**

Q71 Please explain

**Answer If Individuals with Williams have a higher risk of developing... No Is Selected Or Individuals with Williams have a higher risk of developing... Sometimes Is Selected**

Q72 Why do you not discuss with parents of a child with Williams syndrome the increased chance for the child to later on develop ADD? (select all that apply)

- Child may never develop ADD (1)
- More important possible symptoms to discuss (2)
- Lack of comfort discussing ADD (3)
- Another providers responsibility to discuss (4)
- Other (5)
Q73 Please explain

Q74 Whose responsibility is it to discuss with parents of a child with Williams syndrome the increased chance for the child to later develop ADD?

- Primary Care Physician (1)
- Geneticist (2)
- Genetic Counselor (3)
- Psychiatrist/Psychologist (4)
- Other (5)

Q75 Please specify

Q76 If testing were available for patients with Williams that could predict if a child would later develop ADD how likely would it change if you would discuss with their parents the increased chance to develop ADD?

- Very Unlikely (1)
- (2)
- (3)
- (4)
- (5)
- (6)
- Very Likely (7)