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Evaluation of the Perceived Benefit of a Psychiatric Resource for Parents of Children With 22Q11.2 Deletion Syndrome

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EVALUATION OF THE PERCEIVED BENEFIT OF A PSYCHIATRIC RESOURCE FOR PARENTS OF CHILDREN WITH 22Q11.2 DELETION SYNDROME

by

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ABSTRACT

The objective of this study was to assess the perceived utility of an educational resource booklet addressing the psychiatric manifestations and mental health considerations within 22q11.2 deletion syndrome (22q11.2DS).

Seventy-three parents of children with 22q11.2DS completed online surveys before and after reading the booklet. The surveys assessed personality factors and feelings of empowerment, worry, self-stigma, and ability to tolerate uncertainty.

Participants reported that the booklet was easy to understand, provided better understanding of 22q11.2DS and mental illness, answered questions about mental illness associated with the condition, improved knowledge of strategies for protecting the mental health of children with the condition, raised confidence levels in recognizing early warning signs of mental illness, and would be helpful for other families with children affected by the condition. Participants' feelings of empowerment increased by the end of the process while feelings of worry decreased. It was reported that the information contained in the booklet would be beneficial to receive at the time of the diagnosis.

The participants had overall high satisfaction with the booklet. Their knowledge and understanding of mental health within 22q11.2DS reportedly increased after viewing the booklet. The study results suggest a potential benefit in distributing this educational resource to other parents of children with 22q11.2DS.

TABLE OF CONTENTS

LIST OF TABLES

LIST OF FIGURES

CHAPTER 1

LITERATURE REVIEW

1.1 22q11.2 deletion syndrome background

With an estimated prevalence of 1 in 2,000 to 1 in 4,000 live births, 22q11.2 deletion syndrome (22q11.2DS) is the most common microdeletion syndrome (Bassett et al., 2011; Fung et al., 2015). The condition shows variable expression among individuals with a heterogeneous manifestation of symptoms and features. These include congenital heart defects (CHD), immunodeficiency, hypoparathyroidism, cleft palate, psychiatric illness, developmental delay, intellectual disability, learning disability, characteristic facial features, behavioral differences, and hypernasal speech (Bassett et al., 2011; McDonald-McGinn et al., 2015). The syndrome has been termed the second leading cause of major congenital heart disease and developmental delay. It accounts for an estimated 10-15% of individuals with tetralogy of Fallot and 2.4% of people with developmental disabilities (Bassett et al., 2011).

Most cases of 22q11.2DS (90-95%) are newly occurring, or *de novo* (McDonald-McGinn et al., 2015). In the remainder of cases, the condition is inherited from a parent in an autosomal dominant fashion with a 50% chance of the parent passing on the condition to their child. As the medical management of the condition improves, the percentage of inherited cases may increase in the future (McDonald-McGinn et al., 2015). The cause of 22q11.2DS is most

commonly (85%) a large, 3 Mb deletion on the long arm of chromosome 22 which is a region containing approximately 45 functional genes (Bassett et al., 2011). Smaller, atypical, or ‘nested’ deletions within this region account for other cases. The deletions leading to the features observed in 22q11.2DS can range between 0.7 Mb to 3 Mb in size. They occur due to homologous sequences in low copy repeat sequences (LCR) throughout this region (Babcock et al., 2003; Bailey et al., 2002). Due to the similarity of the LCRs, errors are enabled where the LCRs align incorrectly with each other during nonallelic homologous recombination in meiosis. This causes aberrant crossover events that lead to the loss of genetic material (Babcock et al., 2003; Bailey et al., 2002).

Genetic differences causing 22q11.2DS can be identified typically through fluorescent in situ hybridization (FISH), multiplex ligation-dependent probe amplification (MLPA), or array comparative genomic hybridization (aCGH). When a child has 22q11.2DS, parents may be offered genetic testing to confirm if the deletion is inherited or *de novo* due to the variable expressivity of the condition (Bassett et al., 2011).

1.2 22q11.2DS manifestations

Common manifestations of 22q11.2DS include developmental delay, congenital anomalies, and medical and psychiatric disorders with varying ages of onset. Patients typically have at least one prominent phenotypic feature of the condition (Fung et al., 2015). The most prevalent features reported in approximately 75% include congenital heart defects, immunodeficiency, and palatal abnormalities. Hypocalcemia and gastrointestinal issues (including

feeding and swallowing difficulties) are reported in approximately 50%, and genitourinary anomalies (such as renal agenesis) are described in approximately 30% (McDonald-McGinn et al., 2015). Characteristic facial features (bulbous nose, micrognathia, asymmetric crying facies, nasal dimple, and hooded eyelids), hypernasal speech, language delays, learning disabilities, short stature, hearing loss, and seizures can also be present in affected individuals (McDonald-McGinn et al., 2015).

Cardiac defects are considered the leading cause for mortality of children with 22q11.2DS, accounting for around 87% of deaths in this stage of life (McDonald-McGinn et al., 2015). Identification of these heart abnormalities may lead to an individual's diagnosis and can sometimes be detected in the prenatal or neonatal period. In addition to cardiac defects, immune system anomalies are also prevalent within the syndrome, most commonly presenting as recurrent infections (35-40%), autoimmune diseases, and low and impaired T-cell function (Bassett et al., 2011). Palatal abnormalities often manifest in a mild manner (65%), presenting as bifid uvula, velopharyngeal dysfunction, or occult submucosal cleft palate. Around 11% of pediatric patients with 22q11.2DS have overt cleft palate (McDonald-McGinn et al., 2015).

Other features described in 22q11.2DS include hypoparathyroidism, gastrointestinal anomalies, and genitourinary anomalies. Hypoparathyroidism within the condition is thought to lead to hypocalcemia, occurring in over 50 to 60% of cases. Hypocalcemia may result in symptoms of stridor, tetany, fatigue, feeding difficulty, and seizures (McDonald-McGinn et al., 2015). Gastrointestinal

anomalies affect around 30% of individuals with 22q11.2DS. Difficulties in swallowing and feeding, if severe, can necessitate placement of a feeding tube (McDonald-McGinn et al., 2015). Additional anomalies may be present in the genitourinary system which can present as cryptorchidism, absent uterus, dysplastic or cystic kidneys, inguinal hernia, hydronephrosis, or hypospadias. Around one-third of patients will have one genitourinary feature (McDonald-McGinn et al., 2015).

Developmental delay is another feature of 22q11.2DS. In an estimated 70% of cases involving affected children, there is a delay in language onset resulting in either a deficit in words used by the age of 24 months or a nonverbal state (McDonald-McGinn et al., 2015). There is also a difference in intelligence quotient (IQ) scores between the general population and patients with the condition. The average IQ of an individual with 22q11.2DS is around 70, whereas the average IQ for the general population is 100 (McDonald-McGinn et al., 2015). Mild intellectual disabilities occur in around 30-40% of cases (Fung et al., 2015). Severe intellectual disabilities appear to be uncommon among affected individuals, but children experiencing prolonged hypocalcemia, cardiac arrest, neonatal seizures, or primary brain malformations tend to have poorer cognitive outcomes (McDonald-McGinn et al., 2015). Mathematics and language comprehension appears to be the most prevalent areas where learning difficulties are present for preschool to elementary school aged patients (McDonald-McGinn et al., 2015).

Another prominent manifestation of 22q11.2DS is psychiatric illness. Psychiatric illnesses are considered the most common later-onset conditions in 22q11.2DS (Fung et al., 2015). Their prominence and impact make them the most highly reported point of concern among patients and their families. These manifestations are the most likely among the adolescent and adult population to require medical attention and affect the daily life of the individual (Fung et al., 2015). The most prevalent psychiatric conditions experienced by patients with 22q11.2DS are schizophrenia (25% of adults), autism (20% of children), anxiety (24-56% of adults), mood disorders (18-41% of adults), and attention-deficit/hyperactivity disorder (ADHD; 22-33% of adolescence) (Biswas & Furniss, 2016; McDonald-McGinn et al., 1999). Obsessive-compulsive disorder (OCD) can also be observed in some cases (Biswas & Furniss, 2016). With a nearly 20-fold increased chance for schizophrenia (SCZ), this makes the 22q11.2 deletion the most prominent molecular genetic risk factor for this psychiatric illness (Fung et al., 2015; McDonald-McGinn et al., 2015).

1.3 22q11.2DS management

Due to the variable expressivity of 22q11.2DS causing a wide spectrum of symptoms and manifestations, management must be tailored to address patient-specific needs. This customized management allows for the most effective treatment for the patient's features, severity, age or developmental stage, and need for treatment (Bassett et al., 2011). Common management typically involves specialties and services including pediatrics, surgery, general medicine,

interventional therapies, psychiatry, psychology, and genetics services such as consultations with a geneticist and genetic counselor (McDonald-McGinn et al., 2015).

The cardiac manifestations are typically treated as they would be in an individual without 22q11.2DS, and surgical intervention is pursued when required for repair of a congenital heart defect. Special considerations might include increased attention to preventing hypocalcemia, bronchospasm and airway bleeding, and immunological depression around the time of the surgery (McDonald-McGinn et al., 2015). Many adult patients who previously underwent intracardiac repair during childhood require repeat cardiac interventions and follow-up (Fung et al., 2015).

Immune system dysfunction in 22q11.2DS patients requires testing to determine the exact cause of the abnormality. Depending on the severity, a thymus transplant may be required; however, it is not typical for the transplant stage to be reached (McDonald-McGinn et al., 2015). If a low T-cell count is discovered, isolation is recommended for the patient's safety, and live viral vaccines are not administered. Allergies and recurrent infections are corrected through sinus rinses, ear tubes, and allergy treatment (McDonald-McGinn et al., 2015).

Additional areas for treatment and management include those for palatal anomalies, gastrointestinal anomalies, and genitourinary complications. Treatment for palatal anomalies for patients with 22q11.2DS is usually identical to that of a patient without the condition. Surgical repair is typically pursued when

the patient is about 4 to 6 years of age. This repair allows for an improved quality of life by enabling normal speech production and effective communication (McDonald-McGinn et al., 2015). For gastrointestinal anomalies, a feeding tube may be necessary for more extreme feeding and swallowing difficulties. For genitourinary complications, consultation with a nephrologist, urologist, or gynecologist may be appropriate (McDonald-McGinn et al., 2015).

Another point of management includes treating thyroid dysfunction and hypocalcemia. Calcium levels in patients should be monitored in infancy to avoid seizures. In addition, surveillance for calcium levels should be completed when the patient is experiencing biological stress, such as puberty, pregnancy, delivery, or before an operation (McDonald-McGinn et al., 2015). Thyroid function should also be assessed annually in adult patients. Appropriate thyroid medication or calcium and vitamin D supplements may be recommended if necessary (McDonald-McGinn et al., 2015).

Cognitive differences in 22q11.2DS may also entail specific management recommendations depending on areas of concern and severity. Accommodations such as assistance with managing money, completing forms, and making complex life and work decisions may be necessary (Fung et al., 2015). Deficits in verbal learning can be addressed through visual reminders, which have been found to reduce the frustrations of both caregivers and patients (Fung et al., 2015). Throughout the lifespan of a patient with 22q11.2DS, sign language, educational supports, early intervention, and vocational counseling serve as management strategies for the cognitive differences (Bassett et al., 2011).

When individuals experience psychiatric illness as a part of 22q11.2DS, it is most effectively treated when detected and managed early (Gothelf et al., 2007). Therefore, timely detection of psychiatric illness is essential in providing effective management. In a study by Weisman et al. (2017), research was conducted to examine subthreshold psychosis in patients (ranging from 6-55 years old with an average age of 17.1) with 22q11.2DS. It was found that the highest rate of subthreshold psychosis occurred in the age range of 13-25 years old. Additionally, individuals with these symptoms were found to have higher rates of anxiety disorders and ADHD and were found to have lower IQ and global functioning scores (Weisman et al., 2017). In another study, Gothelf et al. (2007) researched risk factors regarding psychotic disorders in patients with 22q11.2DS. This study involved affected and unaffected children, both around 12 years old. An internal longitudinal comparison was completed of the two groups when they reached 16-17 years old. At the follow-up, 32.1% of affected individuals had developed psychotic disorders compared to 4.3% of the unaffected individuals (Gothelf et al., 2007). The main predictors for this development were anxiety and OCD in childhood. Similarly, low verbal IQ in childhood was suggested to be a risk factor for psychotic disorders among individuals with 22q11.2DS (Gothelf et al., 2007). This data supports early identification and intervention for children with these subthreshold signs of psychosis which may help improve the prognosis and outcome of patients.

Additionally, it is recommended that changes in behavior, functioning, thinking, physical state, and emotion be monitored routinely for individuals with

22q11.2DS who have a mental health condition. A knowledgeable clinician should perform this monitoring due to the possibility of communication difficulties with patients who have moderate to severe intellectual disability (Fung et al., 2015). Treatment is similar to those with psychiatric illness without 22q11.2DS (Bassett et al., 2011). Changes such as avoidance of substance use, eating a healthy diet, and implementing mental and physical exercise serve as recommendations that might help reduce the chance for psychiatric illness (Fung et al., 2015).

Another form of management for patients with 22q11.2DS is genetic counseling. Recommendations for individuals with the condition include receiving multiple appointments with genetic counselors throughout their lives to obtain information about their condition (Fung et al., 2015). This information would include discussions about the inheritance pattern and recurrence risk, etiology, variability, medical and psychiatric manifestations, and interventions (Bassett et al., 2011).

1.4 Previous studies on psychiatric illness in 22q11.2DS and parent education

Although patients with 22q11.2DS have a higher chance of experiencing psychiatric conditions, the affected adolescent population is no more likely to receive mental health care than the general, age-matched population (Young et al., 2011). It has been reported that early intervention may reduce the risk of developing a psychotic disorder (Gothelf et al., 2007). Studies have been completed looking at subthreshold indicators for psychosis in patients with

22q11.2DS. Anxiety disorders, low IQ scores, ADHD, and OCD have been associated with later onset psychosis in these patients (Gothelf et al., 2007; Weisman et al., 2017). An information deficit exists among parents of children with 22q11.2DS regarding the increased chance for mental illness. Parents typically receive information about the nonpsychiatric manifestations of the condition from healthcare providers, but they do not always receive information regarding the psychiatric manifestations. Parents often report obtaining information about this aspect of the condition through nonmedical sources such as the internet (61.5%) (Hercher & Bruenner, 2008).

Interestingly, psychiatric presentations are reported to invoke greater anxiety levels among parents than medical presentations (Hercher & Bruenner, 2008). A study found that the possibility of psychiatric illness was not shared at the time of diagnosis for most cases (only 38.5% had received this information during the diagnosis), and it was not frequently brought up later by healthcare providers such as pediatricians (only 2.6% received this information from the pediatrician) (Hercher & Bruenner, 2008). This remained true for medical geneticists, with only 38.1% of parents reporting that they received information about their child's chance of developing a psychiatric illness from a medical geneticist. This is concerning considering this manifestation of the condition is deemed to have a high perceived burden among caregivers and patients themselves (Karas et al., 2014). The reason behind this deficit in knowledge surrounding the psychiatric manifestations of the condition was explored in a later study. Through a questionnaire and parent interviews, it was found that

genetic counselors felt that the risk for psychiatric illness was important to report; however, they were significantly less likely to discuss this risk than they were to discuss risks for other possible features of the condition (41% versus 83%, respectively) when the initial counseling session took place when the patient was an infant (Martin et al., 2012). When asked about the appropriate time for disclosing the psychiatric risk, the genetic counselors answered incongruently with a wide spectrum across infancy, childhood, and adolescence (Martin et al., 2012). This increases the chance that the information may miss being reported since there is no standardized delivery time. The small sample of parents in the study (n = 4) expressed that they preferred to know about the psychiatric risks before the onset of their child's symptoms (Martin et al., 2012). In a more extensive study with parents (n = 37) of children with 22q11.2DS, it was determined that the parents wanted to learn more about preventing or coping with their child's mental health complications (Alugo et al., 2017). The parents also gave suggestions regarding information that should be included in an educational resource. They proposed including education regarding early warning signs of psychiatric illness, talking with their child about the condition, and managing their child's mental illness (Alugo et al., 2017). This need could be met with psychiatric genetic counseling, but there is currently limited availability of this specialized service (Inglis et al., 2015).

There are various examples of the use of educational booklets to supplement topics discussed in genetic counseling sessions. Written educational materials have been produced for genetic conditions such as cystic fibrosis,

Down syndrome, and hereditary breast cancer (Bryant et al., 2001; Clayton et al., 1995; Mancini et al., 2006; Vadaparampil et al., 2011). In a study by Lewis et al. (2012), an informational booklet was created and evaluated for parents of children who have not yet received a diagnosis for conditions with suspected genetic etiologies. A booklet to support the psychosocial needs of the parents was made through the combination of parent interviews, professional focus groups, and a review of the literature. The booklet was then evaluated through a questionnaire that assessed the relevance, usefulness, and scope of the information. It was found that the parents had a high patient satisfaction score with the information offered in the booklet (Lewis et al., 2012). Another study by Hunt et al. (2020), used a written informational resource to increase parent knowledge surrounding the inheritance and causation of congenital heart disease. The brochure was piloted with a small group of parents of children with congenital heart disease. It was then further evaluated with a larger sample size. During the study, the parents answered questionnaires before and after viewing the brochure. It was found that the resource increased the parents' knowledge regarding the causation of congenital heart disease, and many parents reported a reduced feeling of guilt after viewing the resource (Hunt et al., 2020).

1.5 Research trajectory

This study aims to evaluate the use of an educational resource that describes the psychiatric manifestations of 22q11.2DS to bridge the knowledge deficit expressed by parents of children with the condition. The educational material is in the form of a booklet designed by a University of British Columbia

(UBC) genetic counseling candidate in collaboration with the UBC Department of Psychiatry's psychiatric genetic counseling team (Chieffo, 2018). An existing written mental health resource inspired the booklet's content with additional direction provided by the input of parents of children with 22q11.2DS. The booklet contains a description of 22q11.2DS and information about mental illnesses. It provides an overview of mental illness, the causes, protective factors, early detectable signs, mental health resources, and mental illness as seen in 22qDS. The current study assessed the utility of the resource, and it evaluated the perceived changes in the parents' feelings of empowerment, worry, and self-stigma surrounding the content. Parents were asked to complete surveys relevant to their experience before and after viewing the resource. Based on the results of this study, it may suggest a benefit in distributing this booklet to other parents of children with 22q11.2DS. This will allow parents to access a professionally developed resource rather than finding information through unregulated sources such as the internet. The resource discusses multiple previously identified parent concerns by addressing the condition's etiology and highlighting signs of emerging mental illness. It also provides information about mental health resources for the families.

CHAPTER 2

EVALUATION OF THE PERCEIVED BENEFIT OF A PSYCHIATRIC RESOURCE FOR PARENTS OF CHILDREN WITH 22Q11.2 DELETION SYNDROME[1]

[1]Blankenship, K., Prijoles, E., Batallones, R., Slomp, C., Morris, E., Hill-Chapman, C., and Austin, J. To be submitted to *Patient Education and Counseling*

2.1 Abstract

2.1.1 Objectives

The objective of this study was to assess the perceived utility of an educational resource booklet addressing the psychiatric manifestations and mental health considerations within 22q11.2 deletion syndrome (22q11.2DS).

2.1.2 Methods

Seventy-three parents of children with 22q11.2DS completed online surveys before and after reading the booklet. The surveys assessed personality factors and feelings of empowerment, worry, self-stigma, and ability to tolerate uncertainty.

2.1.3 Results

Participants reported that the booklet was easy to understand, provided better understanding of 22q11.2DS and mental illness, answered questions about mental illness associated with the condition, improved knowledge of strategies for protecting the mental health of children with the condition, led to raised confidence levels in recognizing early warning signs of mental illness, and would be helpful for other families with children affected by the condition. Participants' feelings of empowerment increased by the end of the process while feelings of worry decreased. It was reported that the information contained in the booklet would be beneficial to receive at the timing of the diagnosis.

2.1.4 Conclusions

The participants had overall high satisfaction with the booklet. Their knowledge and understanding of mental health within 22q11.2DS reportedly increased after viewing the booklet.

2.1.5 Practice Implications

The study results suggest a potential benefit in distributing this educational resource to other parents of children with 22q11.2DS.

2.2 Introduction

With an estimated prevalence of 1 in 2,000 to 1 in 4,000 live births, 22q11.2 deletion syndrome (22q11.2DS) is the most common microdeletion syndrome (Bassett et al., 2011; Fung et al., 2015). The condition shows variable expression among individuals with a heterogeneous manifestation of symptoms and features. These include congenital heart defects (CHD), immunodeficiency, and palatal abnormalities in approximately 75% of individuals. Hypocalcemia and gastrointestinal issues (including feeding and swallowing difficulties) are reported in approximately 50%, and genitourinary anomalies such as renal agenesis are described in approximately 30% (McDonald-McGinn et al., 2015). Characteristic facial features (bulbous nose, micrognathia, asymmetric crying facies, nasal dimple, and hooded eyelids), hypoparathyroidism, psychiatric illness, developmental delay, intellectual disability, learning disability, behavioral differences, seizures, hearing loss, and hypernasal speech can also be present in affected individuals (Bassett et al., 2011; McDonald-McGinn et al., 2015). The syndrome has been termed the second leading cause of major congenital heart

disease and developmental delay. It accounts for an estimated 10-15% of individuals with tetralogy of Fallot and 2.4% of people with developmental disabilities (Bassett et al., 2011).

Most cases of 22q11.2DS (90-95%) are newly occurring, or *de novo* (McDonald-McGinn et al., 2015). In the remainder of cases, the condition is inherited from a parent in an autosomal dominant fashion with a 50% chance of the parent passing on the condition to their child. As the medical management of the condition improves, the percentage of inherited cases may increase in the future (McDonald-McGinn et al., 2015). The cause of 22q11.2DS is most commonly (85%) a large, 3 Mb deletion on the long arm of chromosome 22, which is a region containing approximately 45 functional genes (Bassett et al., 2011). Smaller, atypical, or 'nested' deletions within this region account for other cases. Genetic differences causing 22q11.2DS can be identified typically through genetic testing. When a child has 22q11.2DS, parents may be offered genetic testing to confirm if the deletion is inherited or *de novo* due to the variable expressivity of the condition (Bassett et al., 2011).

A prominent manifestation of 22q11.2DS is psychiatric illness. Psychiatric illnesses are considered the most common later-onset conditions in 22q11.2DS (Fung et al., 2015). Their prominence and impact make them the most highly reported point of concern among patients and their families. These manifestations are the most likely among the adolescent and adult population to require medical attention and affect the daily life of the individual (Fung et al., 2015). The most prevalent psychiatric conditions experienced by patients with

22q11.2DS are schizophrenia (25% of adults), autism (20% of children), anxiety (24-56% of adults), mood disorders (18-41% of adults), and attention-deficit/hyperactivity disorder (ADHD; 22-33% of adolescence) (Biswas & Furniss, 2016; McDonald-McGinn et al., 1999). Obsessive-compulsive disorder (OCD) can also be observed in some cases (Biswas & Furniss, 2016). With a nearly 20-fold increased chance for schizophrenia (SCZ), this makes the 22q11.2 deletion the most prominent molecular genetic risk factor for this psychiatric illness (Fung et al., 2015; McDonald-McGinn et al., 2015).

When individuals experience psychiatric illness as a part of 22q11.2DS, it is most effectively treated when detected and managed early (Gothelf et al., 2007). Therefore, timely detection of psychiatric illness is essential in providing effective management. In a study by Weisman et al. (2017), research was conducted to examine subthreshold psychosis in patients (ranging from 6-55 years old with an average age of 17.1) with 22q11.2DS. It was found that the highest rate of subthreshold psychosis occurred in the age range of 13-25 years old. Additionally, individuals with these symptoms were found to have higher rates of anxiety disorders and ADHD and were found to have lower IQ and global functioning scores (Weisman et al., 2017). In another study, Gothelf et al. (2007) researched risk factors regarding psychotic disorders in patients with 22q11.2DS. This study involved affected and unaffected children, both around 12 years old. An internal longitudinal comparison was completed of the two groups when they reached 16-17 years old. At the follow-up, 32.1% of affected individuals had developed psychotic disorders compared to 4.3% of the unaffected individuals

(Gothelf et al., 2007). The main predictors for this development were anxiety and OCD in childhood. Similarly, low verbal IQ in childhood was suggested to be a risk factor for psychotic disorders among individuals with 22q11.2DS (Gothelf et al., 2007). This data supports early identification and intervention for children with these subthreshold signs of psychosis which may help improve the prognosis and outcome of patients.

Additionally, it is recommended that changes in behavior, functioning, thinking, physical state, and emotion be monitored routinely for individuals with 22q11.2DS who have a mental health condition. Treatment is similar to those with psychiatric illness without 22q11.2DS (Bassett et al., 2011). Changes such as avoidance of substance use, eating a healthy diet, and implementing mental and physical exercise serve as recommendations that might help reduce the chance for psychiatric illness (Fung et al., 2015).

Another form of management for patients with 22q11.2DS is genetic counseling. Recommendations for individuals with the condition include receiving multiple appointments with genetic counselors throughout their lives to obtain information about their condition (Fung et al., 2015). This information would include discussions about the inheritance pattern and recurrence risk, etiology, variability, medical and psychiatric manifestations, and interventions (Bassett et al., 2011).

Although patients with 22q11.2DS have a higher chance of experiencing psychiatric conditions, the affected adolescent population is no more likely to receive mental health care than the general, age-matched population (Young et

al., 2011). This indicates a possible information deficit among parents of children with 22q11.2DS regarding the increased chance for mental illness. Parents typically receive information about the nonpsychiatric manifestations of the condition from healthcare providers, but they do not always receive information about the psychiatric manifestations. Parents often report obtaining information about this portion of the condition through nonmedical sources such as the internet (61.5%) (Hercher & Bruenner, 2008).

Interestingly, psychiatric presentations of 22q11.2DS are reported to invoke greater anxiety levels among parents than medical presentations (Hercher & Bruenner, 2008). However, a study found that the possibility of psychiatric illness was not shared at the time of diagnosis for most cases (only 38.5% had received this information during the diagnosis), and it was not frequently brought up by later healthcare providers such as pediatricians (only 2.6% received this information from the pediatrician) (Hercher & Bruenner, 2008). This remained true for medical geneticists, with only 38.1% of parents reporting that they received information about their child's chance of developing a psychiatric illness from a medical geneticist. This is concerning considering this manifestation of the condition is deemed to have a high perceived burden among caregivers and patients themselves (Karas et al., 2014). The reason behind the deficit in knowledge surrounding the psychiatric manifestations of the condition was explored in a later study. Through a questionnaire and parent interviews, it was found that genetic counselors felt that the risk for psychiatric illness was important to report; however, they were significantly less likely to discuss this risk

than they were to discuss risks for other possible features of the condition (41% versus 83%, respectively) when the initial counseling session took place when the patient was an infant (Martin et al., 2012). When asked about the appropriate time for disclosing the psychiatric risk, the genetic counselors answered incongruently with a wide spectrum across infancy, childhood, and adolescence (Martin et al., 2012). This increases the chance that the information may miss being reported since there is no standardized delivery time. The small sample of parents in the study (n = 4) expressed that they preferred to know about the psychiatric risks before the onset of their child's symptoms (Martin et al., 2012). In a more extensive study with parents of children with 22q11.2DS (n = 37), it was determined that the parents wanted to learn more about preventing or coping with their child's mental health complications (Alugo et al., 2017). The parents also gave suggestions regarding information that should be included in an educational resource. They proposed including education regarding early warning signs of psychiatric illness, talking with their child about the condition, and managing their child's mental illness (Alugo et al., 2017). This need could be met with psychiatric genetic counseling, but there is currently limited availability of this specialized service (Inglis et al., 2015).

Educational booklets have been used in various medical settings to address knowledge deficits surrounding certain conditions. There are numerous examples of the use of educational booklets to supplement topics discussed in genetic counseling sessions. Written educational materials have been produced for genetic conditions such as cystic fibrosis, Down syndrome, and hereditary

breast cancer (Bryant et al., 2001; Clayton et al., 1995; Mancini et al., 2006; Vadaparampil et al., 2011). In a study by Lewis et al. (2012), an informational booklet was created and evaluated for parents of children who have not yet received a diagnosis for conditions with suspected genetic etiologies. A booklet to support the psychosocial needs of the parents was made through the combination of parent interviews, professional focus groups, and a review of the literature. The booklet was then evaluated through a questionnaire that assessed the relevance, usefulness, and scope of the information. It was found that the parents had a high patient satisfaction score with the information offered in the booklet (Lewis et al., 2012). Another study by Hunt et al. (2020), used a written informational resource to increase parent knowledge surrounding the inheritance and causation of congenital heart disease. The brochure was piloted with a small group of parents of children with congenital heart disease. It was then further evaluated with a larger sample size. During the study, the parents answered pre- and post-questionnaires before and after viewing the brochure. It was found that the resource increased the parents' knowledge regarding the causation of congenital heart disease, and many parents reported a reduced feeling of guilt after viewing the resource (Hunt et al., 2020).

This study aimed to evaluate the parent perception of an educational resource addressing the psychiatric manifestations of 22q11.2DS. If successful, this resource could aid in bridging the knowledge deficit surrounding parent knowledge of psychiatric conditions involved in 22q11.2DS. The educational material is in the form of a booklet designed by a University of British Columbia

(UBC) genetic counseling candidate in collaboration with the UBC Department of Psychiatry's psychiatric genetic counseling team (Chieffo, 2018). An existing written mental health resource inspired the booklet's content with additional direction provided by the input of parents of children with 22q11.2DS. The booklet contains a description of 22q11.2DS and information about mental illnesses. It provides an overview of mental illness, the causes, protective factors, early detectable signs, mental health resources, and mental illness as seen in 22q11.2DS.

2.3 Materials and Methods

2.3.1 Participants and Recruitment

Parents of children diagnosed with 22q11.2 deletion syndrome were recruited to participate in the study. Due to limitations in translation services, the recruited parents were English-speaking. Recruitment occurred through two methods, including the online distribution of a flyer by social media posts and a patient letter sent directly to the parents of children affected with the condition. Multiple 22q11.2 deletion syndrome support groups were contacted to aid in distributing the online flyer. Communications were sent successfully to the 22q Family Foundation, 22q and You Center (associated with the Children's Hospital of Philadelphia), and multiple individually run state support groups in the states of South Carolina, California, Maryland, New Hampshire, New Jersey, Ohio, Pennsylvania, and Texas. In addition, patient letters were sent to parents of children seen through the Greenwood Genetic Center in South Carolina.

2.3.2 General Procedure and Ethics

The study was conducted through online questionnaires which were completed before, immediately after, and one month after reading an educational booklet containing information about the psychiatric manifestations of 22q11.2 deletion syndrome. These questionnaires were referred to as the Pre-Resource Viewing Questionnaire (T1), Immediate Post-Viewing Survey, and One Month Post-Viewing Questionnaire (T2), respectively. The Pre-Resource Viewing Questionnaire contained a combination of validated scales including the Mini-International Personality Item Pool (Mini-IPIP), Intolerance of Uncertainty Short Version (IUS Short Version), Problem-Focused Style of Coping (PF-SOC), Genetic Counseling Outcome Scale (GCOS), Self-Stigma in Relatives of People with Mental Illness Scale (SSRMI; participants only completed this section of the questionnaire if they reported that their child had a mental illness), and Worry scale (adapted from the modified cancer worry scale) (Carleton et al., 2007; Donnellan et al., 2006; Douma, 2010; Heppner et al., 1995; McAllister et al., 2011). The Immediate Post-Viewing Questionnaire included a question asking if the participants completed the booklet, a question asking how long it took them to complete the booklet, seven 5-point Likert scale questions (with 1 = “strongly agree” and 5 = “strongly disagree”) assessing the participants’ perceptions of the resource, and six short answer questions asking the participants for their thoughts about the resource. The One Month Post-Viewing Questionnaire included the repeated measures of the IUS Short Version, GCOS, SSRMI (if the

participant reported having a child with a mental illness), and Worry scale (Carleton et al., 2007; Donnellan et al., 2006; Douma, 2010; Heppner et al., 1995; McAllister et al., 2011).

Interested individuals were invited to click a link to the questionnaire, which directed them to the study consent form on REDCap (Research Electronic Data Capture), a secure, online application designed to collect data for research purposes (Harris et al., 2009; Harris et al., 2019). All participants included in the study indicated consent to participate. Next, participants were asked to enter their email address into a REDCap field to allow individual follow-up questionnaire links and a study honorarium to be sent to them. This study was done following the review and approval of the University of South Carolina's Institutional Review Board (Pro00112654).

2.3.3 Resource Booklet for 22q11.2DS Psychiatric Manifestations

After completing the Pre-Resource Viewing Questionnaire, the participants were asked to view an educational booklet titled, "22q11.2 Deletion Syndrome: What does it mean for mental health?" containing 22 pages of patient-friendly information relevant to mental health conditions seen in 22q11.2DS. This educational booklet was previously designed during a research study by a University of British Columbia (UBC) genetic counseling candidate in collaboration with the psychiatric genetic counseling group at the UBC Department of Psychiatry (Chieffo, 2018). The content of the booklet was directed by the input of parents of children with 22q11.2DS who had had psychiatric genetic counseling. They were initially given a generic mental illness

booklet and then interviewed to identify what they would like to be included in a 22q11.2DS psychiatric symptom-specific resource. Using the themes from these interviews, the booklet was created and reassessed by ten parents of children with the condition. Feedback was obtained, and the resource was further modified. The booklet contains a description of 22q11.2DS, mental illness, the causes of mental illness, protective factors in mental illness, mental illness as seen in 22q11.2DS, and early signs of mental illness. The booklet also provides a section including mental health resources.

2.3.4 Mini-International Personality Item Pool (Mini-IPIP)

The Mini-IPIP was used in this study to measure personality dimensions (Donnellan et al., 2006). This scale contains 20 questions in a 5-point Likert scale fashion (with 1 = “very inaccurate” and 5 = “very accurate”) that measure the “Big Five” personality traits which are labeled as extraversion, agreeableness, conscientiousness, neuroticism, and openness to experience. Each personality trait serves as a subscale and is assigned four questions. Questions 6, 7, 8, 9, 10, 15,16, 17, 18, 19, and 20 were reverse scored as outlined in the directions for using the scale. The scoring is completed by calculating the sum of responses within each of the three subscales (Donnellan et al., 2006). The results of this study demonstrated reliability for each of the factors of extraversion ($\alpha = 0.896$), agreeableness ($\alpha = 0.794$), conscientiousness ($\alpha = 0.892$), neuroticism ($\alpha = 0.731$), and openness to experience ($\alpha = 0.706$).

2.3.5 Intolerance of Uncertainty Short Version (IUS Short Version)

The IUS Short Version was used to measure participants' reactions to ambiguity, uncertainty, and the future. This scale includes 12 questions in a 5-point Likert scale fashion (with 1 = "not characteristic of me" and 5 = "entirely characteristic of me") and is scored as a total sum of all responses (Carleton et al., 2007). The results of this study indicated adequate reliability of the scale (α = 0.795).

2.3.6 Problem-Focused Style of Coping (PF-SOC)

The PF-SOC was used to measure coping style, and it included 18 questions in a 5-point Likert scale fashion (with 1 = "almost never" and 5 = "almost all of the time") (Heppner et al., 1995). This scale separates coping styles into reflective, suppressive, and reactive categories. Each of the three coping styles serves as a subscale with assigned questions. Of the 18 questions, seven pertain to reflective coping, six to suppressive coping, and five to reactive coping. The results of this study demonstrated reliability for each of the factors of reflective coping (α = 0.791), suppressive coping (α = 0.921), and reactive coping (α = 0.876).

2.3.7 Genetic Counseling Outcome Scale (GCOS)

The GCOS was used to measure the empowerment of the participants (McAllister et al., 2011). This scale contains 24 questions in a 7-point Likert scale fashion (with 1 = "strongly agreed" and 7 = "strongly disagree"), and it is scored as a total sum of all responses. The results of this study indicated adequate reliability of the scale (α = 0.863).

2.3.8 Self-Stigma in Relatives of People with Mental Illness Scale (SSRMI)

Participants completed the SSRMI if they reported that their child had a mental illness. This scale measures the self-stigma of first-degree relatives of individuals with serious mental illness (Morris et al., 2018). The scale contains ten questions in a 5-point Likert scale fashion (with 1 = “strongly disagree” and 5 = “strongly agree”) and is scored as a total sum of all responses. The results of this study indicated adequate reliability of the scale (α = 0.748).

2.3.9 Worry Scale

The Worry scale was used to measure the participants’ level of worry concerning their child’s chances to have a mental illness, and it was adapted from the modified cancer worry scale (Douma, 2010). It consisted of 8 questions. The first question, “How often have you thought about your child’s chance of getting a mental illness (again)?” was scored on a 4-point scale (with 1 = “never” and 4 = “almost always”). The second question, “Have these thoughts affected your mood?” was scored on a 4-point scale (with 1 = “no, not at all” and 4 = “yes, a lot”). The third question, “Have these thoughts interfered with your ability to do daily activities?” was scored on the same 4-point scale as the second question. The fourth question, “How concerned are you about the possibility of your child getting a mental illness (again) one day?” was scored on a 4-point scale (with 1 = “not concerned” and 4 = “very concerned”). The fifth question, “How often do you worry about your child developing a mental illness (again)?” was scored on a 4-point scale (with 1 = “never” and 4 = “almost always”). The sixth question, “How much of a problem is this worry?” was scored on a 4-point scale (with 1 = “not at

all a problem" and 4 = "very much a problem"). The seventh question, "How concerned are you about the possibility that your child will ever need treatment for a mental illness (again)?" was scored on the same 4-point scale as the fifth question. The scores of each question were combined to create a total worry score. The results of this study indicated adequate reliability of the scale (α = 0.903).

2.3.10 Data Analysis

All quantitative statistical analyses were completed using the Statistical Package for the Social Sciences (SPSS). Descriptive statistics were calculated for the participant demographic information and the Immediate Post-Viewing Questionnaire. Reliability analyses were run for each scale used in the research study including the Mini-IPIP, IUS Short Version, GCOS, SSRMI, and Worry scale. Then a paired sample t-test was run to compare the T1 responses to the T2 responses. Following this, Pearson's correlation coefficients were determined for both the Mini-IPIP and PF-SOC versus the results of the IUS Short version, GCOS, Worry scale, and SSRMI for both T1 and T2.

The qualitative data collected through the short answer responses on the Immediate Post-Viewing Questionnaire were analyzed for themes. Themes were determined for each question based on repetitive answers provided by participants, and the themes for each of the questions were synthesized to create three compiled themes.

2.4 Results

2.4.1 Demographic Information

There was a total of 73 participants included in the study. Demographic information of the participants can be seen in Table 2.1, participant diagnosis information can be seen in Table 2.2, and information about the participants' children can be seen in Table 2.3. Of the 73 participants, 17 said their child received a diagnosis prenatally (23.3%), while 56 did not receive a diagnosis prenatally (76.7%). Of the 73, 32 said their child received a diagnosis of 22q11.2DS before the age of 1 (43.8%), 22 said their child did not receive a diagnosis before the age of 1 (30.1%), and 19 participants did not answer this question (26.0%). The average age of diagnosis for the children was 6.11 years old (SD = 7.138). The most frequently reported age of diagnosis was three years old.

2.4.2 Pre-Viewing versus One Month Post-Viewing (T1 vs T2)

Of the 73 participants, 47 completed the GCOS at T1 and T2, 46 completed the Worry scale at T1 and T2, 17 completed the SSRMI at T1 and T2, and 9 completed the IUS Short Version at T1 and T2. A two-sided paired sample t-test was used to compare T1 and T2. The GCOS measure increased from T1 ($M = 127.640$, $SD = 20.231$) to T2 ($M = 135.043$, $SD = 19.868$), with a difference in the means of 7.403, 95% CI [-11.401, -3.408], $t(47) = -3.729$, $p < 0.05$, $d = -0.544$ (Figure 2.1). The Worry scale measure decreased from T1 ($M = 18.239$, $SD = 5.626$) to T2 ($M = 17.304$, $SD = 5.028$), with a difference in the means of -0.935, 95% CI [-0.00096, 1.871], $t(46) = 2.012$, $p < 0.05$, $d = 0.297$ (Figure 2.2).

The SSRMI measure had nearly no detected change between T1 (M = 23.471, SD = 5.433) and T2 (M = 23.294, SD = 8.161), with a difference in the means of -0.177, 95% CI [-2.790, 3.143], t(17) = 0.126, p = 0.90, d = 0.031 (Figure 2.3). The IUS Short Version measure seemed to decrease from T1 (M = 27.333, SD = 7.583) to T2 (M = 24.889, SD = 7.441), with a difference in the means of -2.444, 95% CI [-0.137, 5.026], t(9) = 2.184, $p < 0.05$, d = 0.728 (Figure 2.4).

2.4.3 Correlations

After this, Pearson's correlations were run to look at the relationships between the Mini-IPIP and PF-SOC versus T1 (IUS Short Version, GCOS, Worry scale, and SSRMI) and the Mini-IPIP and PF-SOC versus T2 (IUS Short Version, GCOS, Worry scale, and SSRMI). All 73 participants were included. There were no statistically significant correlations between Mini-IPIP and PF-SOC versus T1 or the Mini-IPIP and PF-SOC versus T2.

2.4.4 Immediate Post-Viewing Survey

Of the 73 participants, 51 completed the Immediate Post-Viewing Survey. The majority of participants reported that the booklet was easy to understand (Figure 2.5), provided them with a better understanding of 22q11.2DS and mental illness (Figure 2.6), answered questions they had regarding mental illness associated with 22q11.2DS (Figure 2.7), would be helpful for other families who have a child with 22q11.2DS (Figure 2.8), helped them understand strategies they could use to protect their child's mental health (Figure 2.9), and helped them to feel more confident in their ability to recognize early warning signs of mental illness (Figure 2.10). Responses from participants were not uniform when they

were asked if the resource led them to feel less worried about the risks for psychiatric disorders in 22q11.2DS (Figure 2.11).

The short answer questions on the Immediate Post-Viewing Questionnaire were answered by 25 of the participants. Themes were extracted from each question and then compiled into three major themes, as seen in Table 2.4.

2.5 Discussion and Conclusion

2.5.1 Discussion

The goal of this study was to evaluate the parent perception of a resource booklet focusing on the psychiatric manifestations of 22q11.2DS. The sample population was parents of children with 22q11.2DS. This research aimed to assess the utility of the booklet; to improve awareness and understanding about the psychiatric manifestations and strategies to protect and improve mental health; and to evaluate the perceived changes in the parents' feelings of empowerment, worry, self-stigma, and perceived ability to deal with the uncertainty surrounding the mental health of their children. Personality factors were also examined to determine if they influenced the participants' responses. It was hypothesized that the parents would find the resource to be helpful. It was also hypothesized that different personality characteristics might alter the way participants interact with the resource.

The study results found that the parents experienced increased empowerment and decreased worry after reading the resource. The literature has previously observed the amelioration of feelings within parents after viewing medical-oriented educational resources pertaining to their children. For example,

in a study completed by Hunt et al. (2020), it was found that viewing an educational booklet about congenital heart disease aided in changing the demeanor of the parents with an affected child. For these parents, parental guilt was diminished after viewing the resource. The increase in empowerment and decrease in worry for parents of children with 22q11.2DS could potentially increase their confidence in their ability to provide care for their child's needs. This could be beneficial regarding their status as the caregiver for their child if this empowerment leads to a subsequential decrease in perceived caregiver burden. A study by Karas et al. (2014) demonstrated that psychiatric manifestations of 22q11.2DS are deemed to have a high perceived burden among caregivers. If this knowledge helps parents feel confident in their abilities to help their children manage this portion of the condition, this may alleviate this perceived burden.

The majority of participants in the current study reported that the booklet was easy to understand, provided them with a better understanding of 22q11.2DS and mental illness, answered questions they had regarding mental illness associated with 22q11.2DS, would be helpful for other families who have a child with 22q11.2DS, helped them understand strategies they could use to protect their child's mental health, and helped them feel more confident in their ability to recognize early warning signs of mental illness. This data appeared to address the concerns raised by parents in a previous study where they expressed the need for an educational resource that would include education on early warning signs of psychiatric illness and how to manage their child's mental

illness (Alugo et al., 2017). However, responses from participants were not uniform when they were asked if the resource led them to feel less worried about the risks for psychiatric disorders in 22q11.2DS. This was interesting since the participants' worry decreased from T1 to T2. It could be that the increased time to process the information in the booklet from the Immediate Post-Viewing Questionnaire to the One Month Post-Viewing Questionnaire could have aided in decreasing the worry among the parents. Family members may also still feel some degree of worry and uncertainty that may be difficult to quantify until specifically faced with psychiatric illness in their child.

Themes from the Immediate Post-Viewing Questionnaire indicated that the booklet was easy to understand, helpful, and included useful illustrations; the booklet could be improved by providing more detailed information and more mental health resources; and the parents would have wanted to know information about the psychiatric manifestations of 22q11.2DS at the diagnosis of their child, but this may be considered overwhelming for other parents. This supported the hypothesis that parents would find the educational resource helpful. It also highlighted areas of improvement for the resource. The timing of the delivery of the information of the psychiatric conditions within 22q11.2DS remained consistent with the previous literature where parents reported that they would want to receive this information before the onset of the child's psychological symptoms (Martin et al., 2012). Interestingly, the parents involved in this study expressed their preference to hear about the psychiatric manifestations at the time of diagnosis. Still, they expressed concern that this may overwhelm other

parents. This may be due to the parents in this study likely being inherently information-seeking since they volunteered to contribute to a research study. They could be recognizing that other parents may not want as much information.

Correlations between different personality factors and responses to the T1 and T2 questionnaires were not statistically significant. This may be due to the small sample size, or it could suggest that personality factors may not influence the participants' responses.

2.5.2 Conclusion

The timing and delivery of information about the psychiatric manifestations of 22q11.2DS are not standardized among healthcare professionals. This leads to parents of children with the condition being unaware of the potential for their child to have a mental illness and less prepared to recognize when their child needs psychiatric care. This information is best addressed through a consultation with a psychiatric genetic counselor, but the resources for this service are limited. In order to address the need for providing this information, an educational resource booklet was provided to parents of children with 22q11.2DS for evaluation. It was determined that the booklet increased empowerment among the parents surrounding the topic and it decreased worry. The parents reported that the booklet was easy to understand, provided them with a better understanding of 22q11.2DS and mental illness, answered questions they had about mental illness associated with the condition, would be helpful for other families who have a child with the condition, helped them understand strategies they could use to protect their child's mental health, and helped them feel more

confident in their ability to recognize early warning signs of mental illness. The parents suggested that the booklet be improved with more detailed information and additional mental health resources. This study also revealed that the parents would have preferred to hear about the psychiatric manifestations of the condition at the time of diagnosis of their child; however, the sampled parents emphasized that other parents may feel overwhelmed with this same timing.

2.5.3 Practice Implications

Based on the results of this study, there could be a potential benefit in distributing this booklet to other parents of children with 22q11.2DS. Providing this resource to parents of children with 22q11.2DS would allow access to medically sourced information rather than unregulated information sources such as the internet. In addition, the resource addresses multiple previously identified parent concerns by highlighting signs of emerging mental illness and mental health management. This booklet would not replace psychiatric genetic counseling services, but it could aid in addressing the current limited availability of this specialized service (Inglis et al., 2015).

2.5.4 Limitations and Future Research

Limitations of the study include the participant population and the small sample size. The participants consisted mainly of white females and individuals of higher education levels (the most frequently reported terminal degree was a bachelor’s degree followed by a master’s degree). It would be interesting to pursue further research to determine if the perspectives represented in the findings of this study would change with populations that include more males and

individuals with different ethnic and educational backgrounds. The study population also likely consisted of participants who are more prone to seek information since they volunteered to be involved in a research study. The viewpoints of other parents who prefer to know less information may not have been represented as prominently. The statistical analysis of this study was limited due to the small sample size. Additional data could be collected in the future for further analysis.

Future studies could be completed to refine the educational resource following the suggestions submitted by the parents. There were requests for more detailed information and support resources to be included in the booklet. Further examination could also be completed to determine any moderators that may affect the perception of the resource's utility. Personality factors were not found to be moderating variables in this study, but additional avenues may be explored. Another area of further research could address the timing of the provision of the resource to families. The participants reported that they were unsure if other parents would want this information during the diagnosis. A study could be completed with two cohorts where one is given the booklet at the time of the diagnosis and the other is given the booklet at a later stage.

Table 2.1 Participant Demographics

Participant characteristic	Total (n)	Percent (%)
Age		
20-24	2	2.7
25-30	0	0
31-35	11	15.1
36-40	27	37.0
41-45	8	11.0
46-50	9	12.3

51-55	8	11.0
56-60	3	4.1
61-65	4	5.5
66-70	1	1.4
Sex		
Male	7	9.6
Female	66	90.4
Ethnicity		
White/Caucasian/European	63	86.3
Black or African American	1	1.4
Hispanic or Latino	3	4.1
Asian	3	4.1
Other	3	4.1
Location		
Canada	1	1.4
United States	66	90.4
Europe	4	5.5
Asia	1	1.4
Australia/South Pacific	1	1.4
Education		
Some high school, no diploma	3	4.1
High school graduate, diploma, or equivalent (e.g., GED)	5	6.8
Some college credit, no degree	7	9.6
Trade/technical vocational training	8	11.0
Bachelor's degree	27	37.0
Master's degree	18	24.7
Doctorate	5	6.8

Table 2.2 Participant Diagnoses Information

Participant's diagnosis status	Total (n)	Percent (%)
Diagnosed with 22q11.2DS		
Yes	3	4.1%
No	31	42.5%
Unspecified	39	53.4%
Diagnosed with a mental illness		
Yes	20	27.4
No	53	72.6

Table 2.3 Demographics and Diagnoses Information of the Children

Information about child	Total (N)	Percent (%)
Age		
0-5	22	30.1
6-10	14	19.2
11-15	10	13.7
16-20	10	13.7
21-25	7	9.6
26-30	1	1.4
31-35	2	2.7
36-40	3	4.1
Unspecified	4	5.5
Age when diagnosed with 22q11.2DS		
0-5	15	20.5
6-10	6	8.2
11-15	0	0
16-20	0	0
21-25	0	0
26-30	0	0
31-35	0	0
36-40	1	1.4
Unspecified	51	69.9
Diagnosed with a mental illness		
Yes	29	39.7
No	44	60.3
Mental illness diagnosis*		
Anxiety	26	35.6
Depression	12	16.4
Schizophrenia	2	2.7
Obsessive-Compulsive Disorder (OCD)	11	15.1
Bipolar Disorder	4	5.5
Other psychiatric diagnoses	8	11.0
Treatment received for psychiatric diagnosis		
Yes	26	35.6
No	3	4.1
Unspecified	44	60.3
Mental health assessment		
Yes	8	11.0
No	36	49.3
Unspecified	29	39.7

*This category is a question for which participants could select more than one answer. Due to this, the percentages do not reach a sum of 100%.

Table 2.4 Short Response Themes for Immediate Post-Viewing Survey

Question	Themes	Compiled Themes
What did you like about the booklet?	1) The booklet was easy to understand. 2) The illustration of the jar model was appreciated.	1) The booklet was easy to understand, helpful, and included illustrations that were useful. 2) The booklet could be improved by providing more detailed information and more mental health resources. 3) The parents expressed that they would have wanted to know this information when their child was first diagnosed with 22q11.2DS, but they recognized that this may be too overwhelming for other parents.
What did you not like about the booklet?	1) The participants would have liked to see missing pieces of information included. 2) The booklet should provide links to mental health resources.	
Please comment on when you think this resource should be given to parents of children with 22q11.2DS.	1) This resource should be given to parents of children with 22q11.2DS at diagnosis. 2) For families of children with 22q11.2DS, this resource should be provided before the teenage years.	
Was there anything that you felt was missing from the booklet?	1) The resource should focus more on providing strategies for protecting against mental illness in childhood.	
If you have any additional thoughts or comments about the booklet, please write them here.	1) More details could have been included in the booklet. 2) The resource could possibly overwhelm parents with younger and newly diagnosed children. 3) The booklet was appreciated and thought to be helpful.	

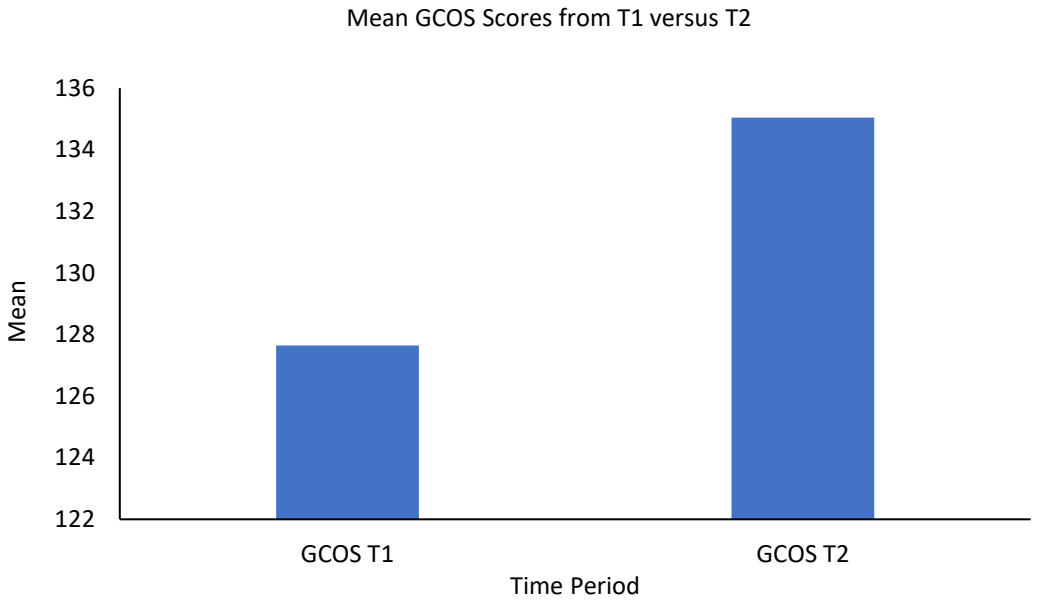

Figure 2.1 Comparison of the mean GCOS scores from T1 versus T2.

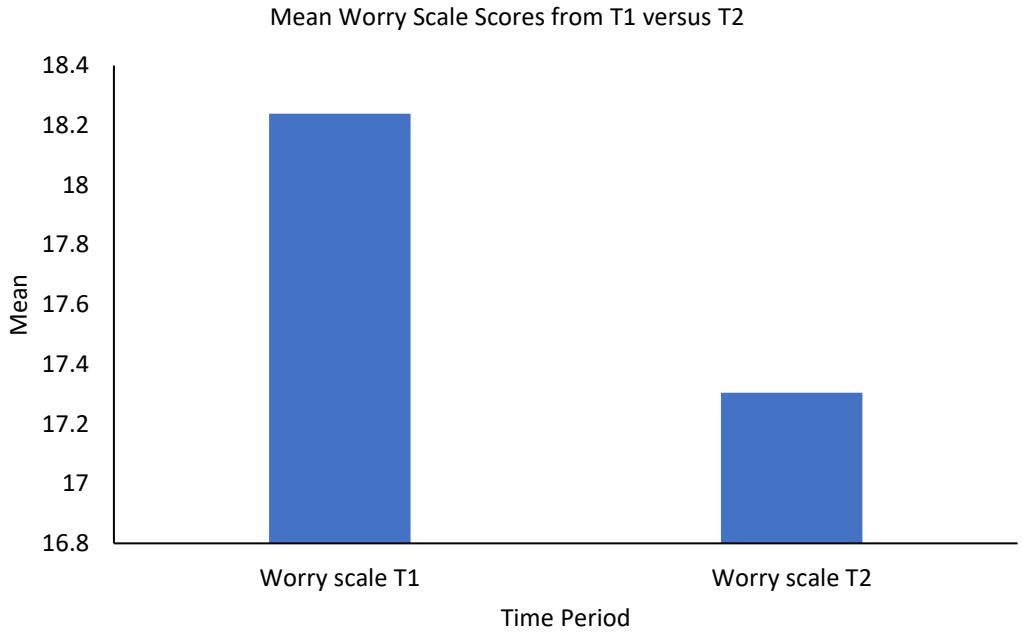

Figure 2.2 Comparison of the mean Worry scale scores from T1 versus T2.

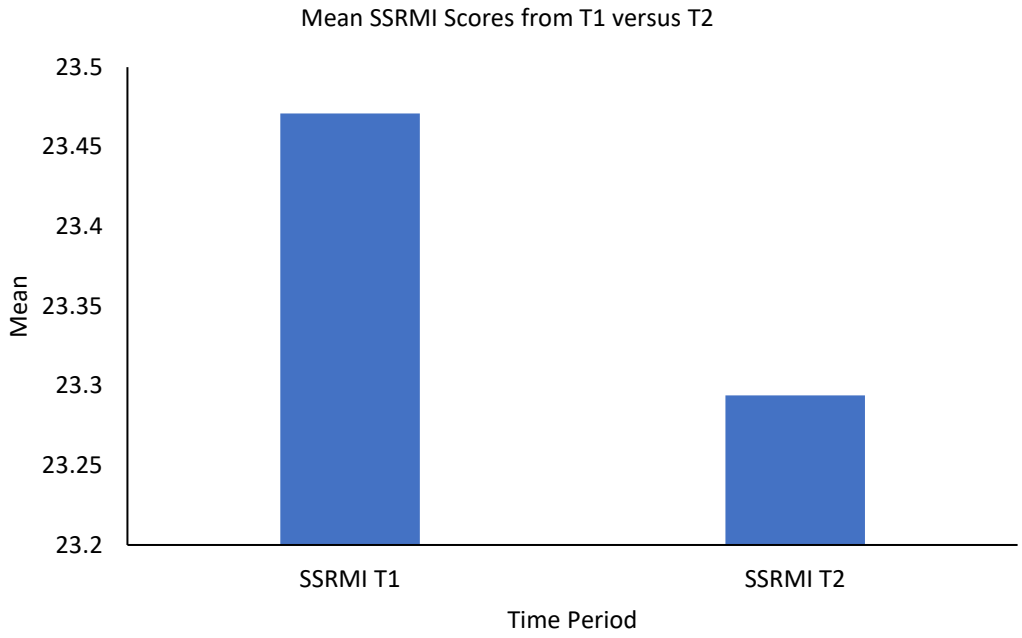

Figure 2.3 Comparison of the mean SSRMI scores from T1 versus T2

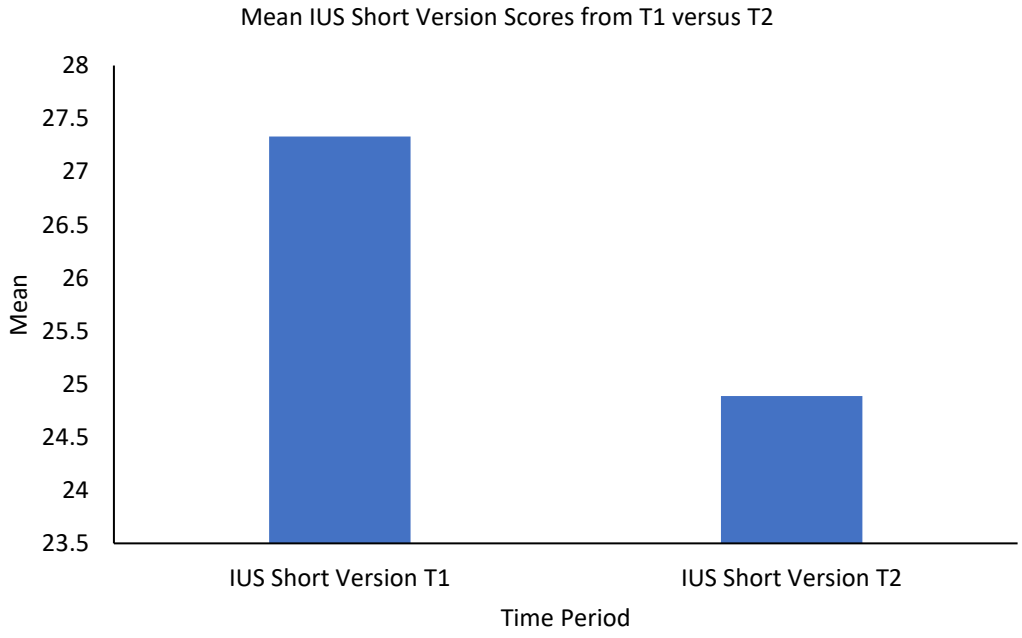

Figure 2.4 Comparison of the mean IUS Short Version scores from T1 versus T2

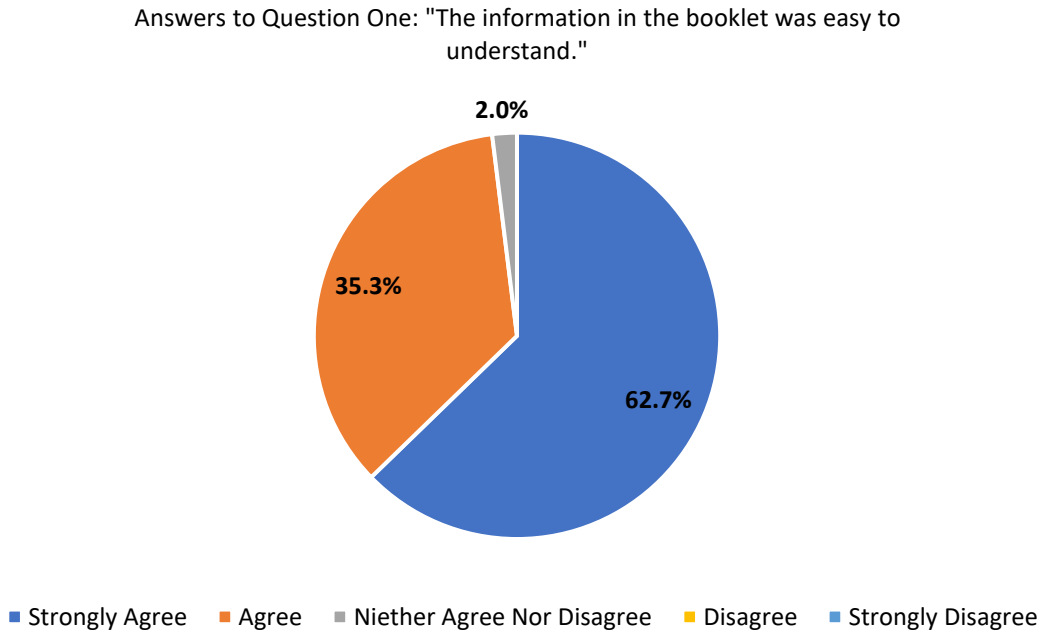

Figure 2.5 Percentage of participants stating strongly agree, agree, disagree, neither agree nor disagree, disagree, or strongly disagree in response to Immediate Post-Viewing Questionnaire question one, “The information in the booklet was easy to understand.”

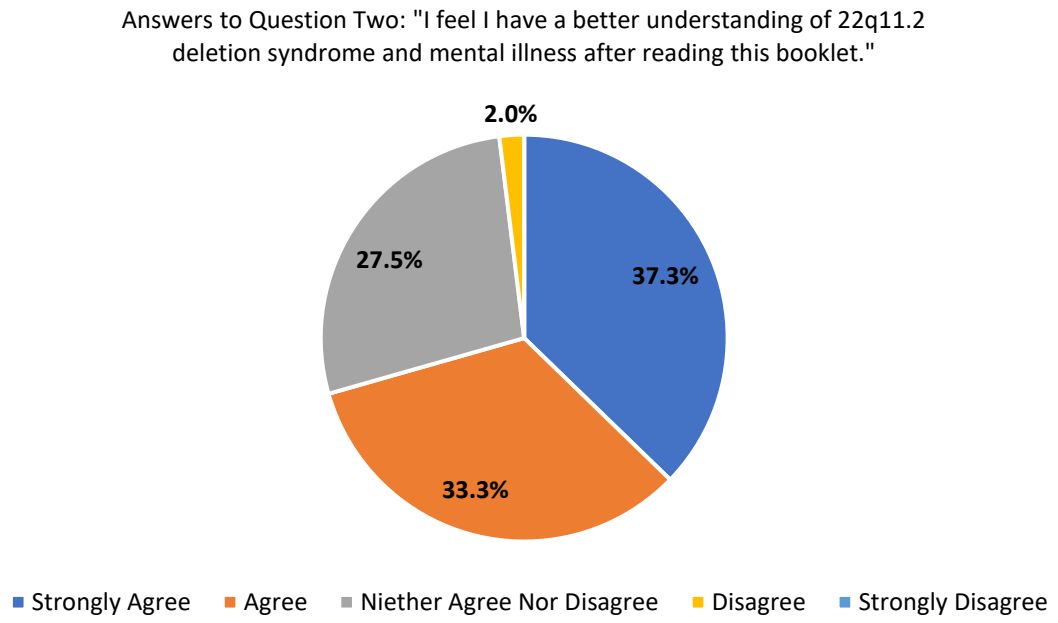

Figure 2.6 Percentage of participants stating strongly agree, agree, disagree, neither agree nor disagree, disagree, or strongly disagree in response to Immediate Post-Viewing Questionnaire question two, “I feel I have a better understanding of 22q11.2 deletion syndrome and mental illness after reading this booklet.”

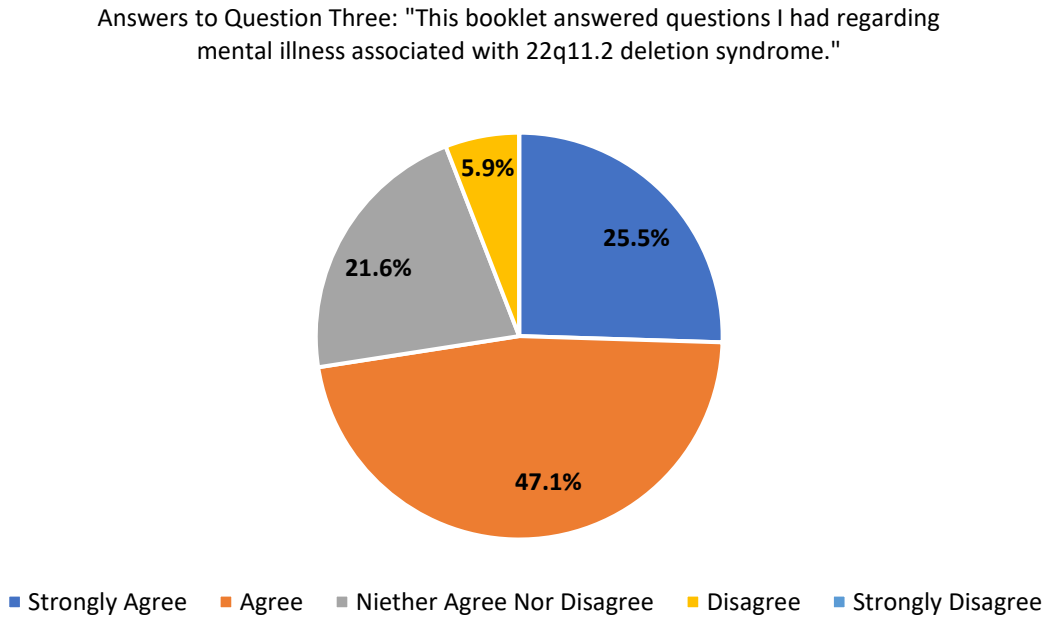

Figure 2.7 Percentage of participants stating strongly agree, agree, disagree, neither agree nor disagree, disagree, or strongly disagree in response to Immediate Post-Viewing Questionnaire question three, “This booklet answered questions I had regarding mental illness associated with 22q11.2 deletion syndrome.”

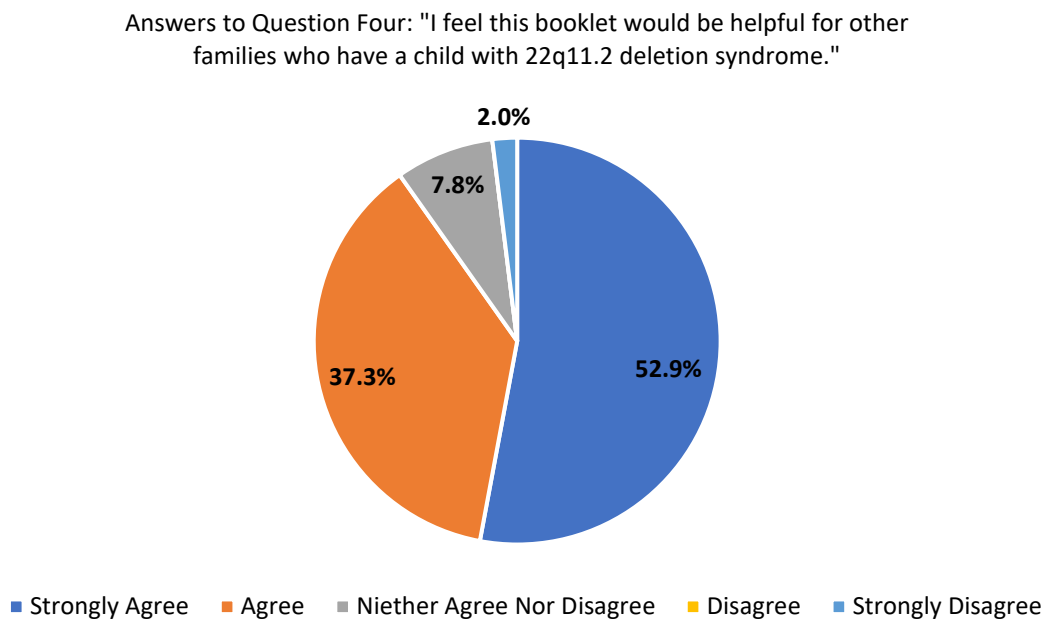

Figure 2.8 Percentage of participants stating strongly agree, agree, disagree, neither agree nor disagree, disagree, or strongly disagree in response to Immediate Post-Viewing Questionnaire question four, “I feel this booklet would be helpful for other families who have a child with 22q11.2 deletion syndrome.”

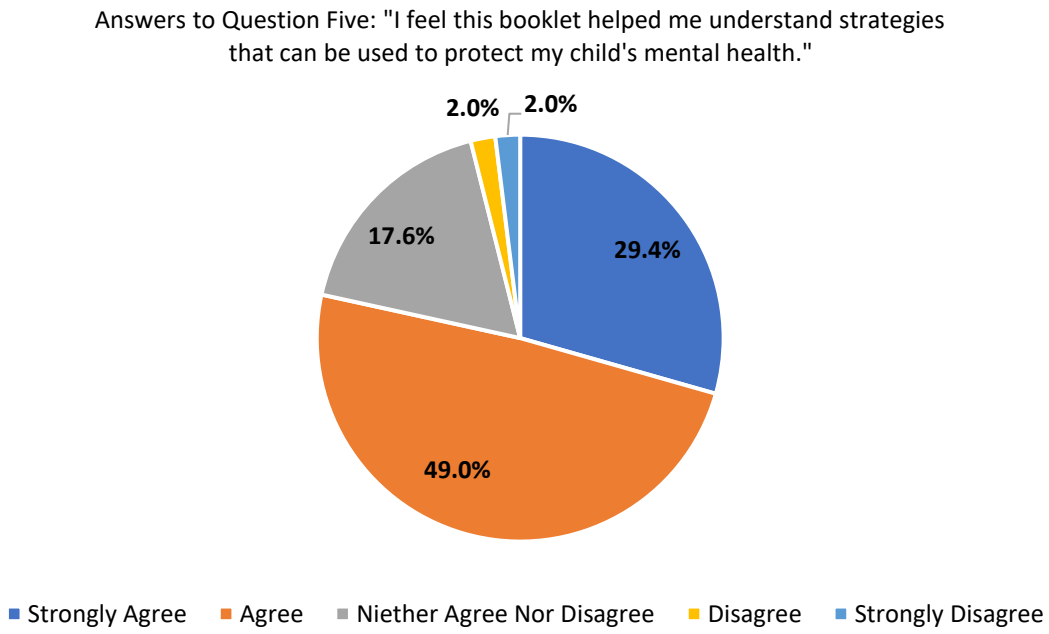

Figure 2.9 Percentage of participants stating strongly agree, agree, disagree, neither agree nor disagree, disagree, or strongly disagree in response to Immediate Post-Viewing Questionnaire question five, “I feel this booklet helped me understand strategies that can be used to protect my child’s mental health.”

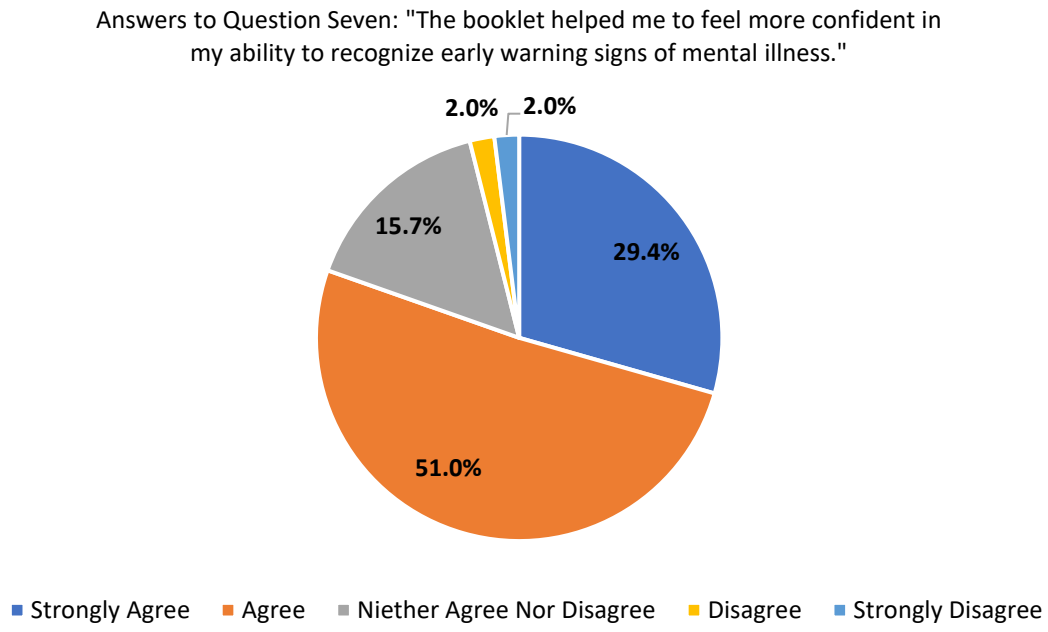

Figure 2.10 Percentage of participants stating strongly agree, agree, disagree, neither agree nor disagree, disagree, or strongly disagree in response to Immediate Post-Viewing Questionnaire question seven, “The booklet helped me to feel more confident in my ability to recognize early warning signs of mental illness.”

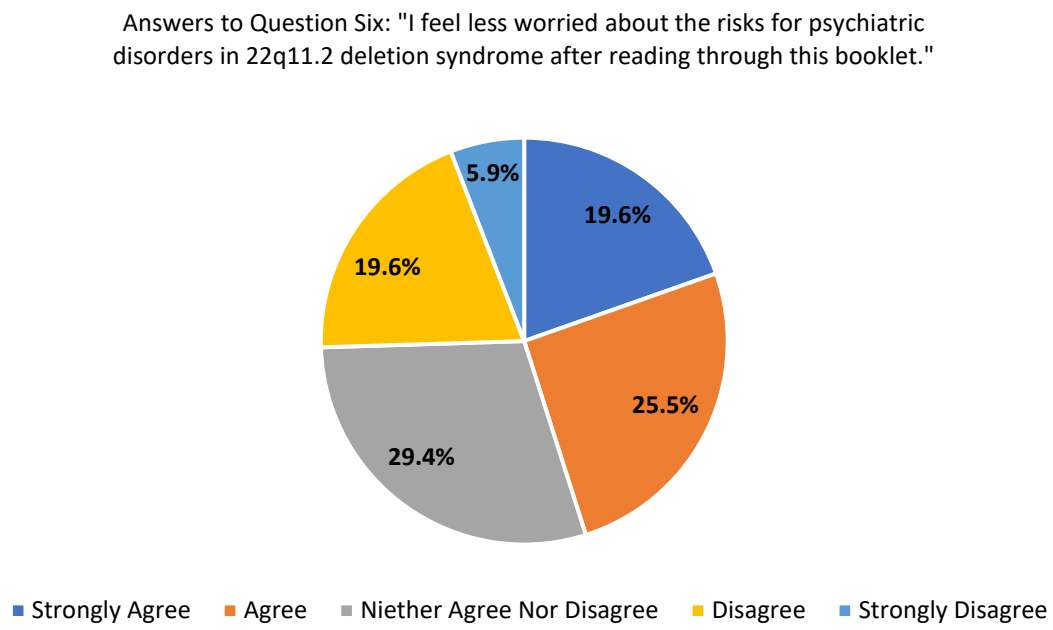

Figure 2.11 Percentage of participants stating strongly agree, agree, disagree, neither agree nor disagree, disagree, or strongly disagree in response to Immediate Post-Viewing Questionnaire question six, “I feel less worried about the risks for psychiatric disorders in 22q11.2 deletion syndrome after reading through this booklet.”

CHAPTER 3

CONCLUSIONS

The timing and delivery of information about the psychiatric manifestations of 22q11.2DS are not standardized among healthcare professionals. This leads to parents of children with the condition being unaware of the potential for their child to have a mental illness and less prepared to recognize when their child needs psychiatric care. This information is best addressed through a consultation with a psychiatric genetic counselor, but the resources for this service are limited. In order to address the need for providing this information, an educational resource booklet was created. This booklet outlines what mental illness is, its causes, mental illness as seen in 22q11.2DS, protective factors, mental health resources, and early signs of mental illness. The booklet was presented to parents of children with the condition to determine whether it would be suitable for other parents. The perceptions and feedback of the parents were collected through a series of questionnaires. It was determined that the booklet increased empowerment among the parents surrounding the topic, and it decreased worry. It was also found that the parents thought that the booklet was easy to understand, provided them with a better understanding of 22q11.2DS and mental illness, answered questions they had about mental illness associated with the condition, would be helpful for other families who have a child with the condition, helped them understand strategies they could use to protect their child's mental

health, and helped them feel more confident in their ability to recognize early warning signs of mental illness. The parents suggested that the booklet be improved with more detailed information and additional mental health resources. This study also revealed that the parents would have preferred to hear about the psychiatric manifestations of the condition at the time of diagnosis of their child; however, the sampled parents emphasized that other parents may feel overwhelmed with this same timing. Based on the results of this study, it may suggest a benefit in distributing this booklet to other parents of children with 22q11.2DS. Providing this resource would allow parents to access this information through a trusted medical source rather than unregulated information sources such as the internet.

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APPENDIX A

STUDY CONSENT FORM

The participants were required to view and electronically consent before they were included in the study. The consent form is included below.

Evaluation of an online resource for parents regarding the psychiatric manifestations of 22q11.2 deletion syndrome

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Introduction:

You are being invited to take part in this research study because you are a parent of someone with 22q11.2 deletion syndrome (22qDS). Participation in this study is voluntary. It is your decision to choose whether you would like to participate in this study or not. Before you decide, it is important for you to understand what the research involves. This consent form will tell you what this study is about, what your involvement would be if you choose to participate, and the possible benefits, risks, and discomforts that may occur. Take the time to read through this form carefully and keep a copy of the form for your records.

What is the purpose of this study?

Families living with 22qDS often have questions or concerns about the psychiatric (mental health) symptoms of 22qDS. Researchers are interested in finding ways to provide more education and support for these issues. We previously made an educational booklet for families about 22qDS and mental health. This resource was made using feedback from a small group of parents that had received psychiatric genetic counseling. We want to understand if a booklet about mental illness and 22qDS would be helpful for families of children with the condition by helping parents worry less, feel more prepared, or alter their view of psychiatric illnesses. We want to know if this resource could help other parents feel more familiar with psychiatric illnesses sometimes seen with 22qDS. We also want to know if this

resource could help other parents better understand strategies to protect and improve mental health.

What will my participation involve?

You are being asked to provide feedback about the educational resource by completing online surveys. If you agree to participate, you will be directed to these surveys through a secure online database called REDCap. There will be several questionnaires to complete before viewing the resource (this set of questions will be called the "pre-viewing" survey). You will then be able to view the online resource. This will be followed by some additional questions about your thoughts about the resource (this set of questions will be called the "post-viewing" survey). We expect this to take approximately 45 minutes - 1.5 hours depending on how much time you spend reviewing the resource.

One month later, you will be sent another link to complete the final round of questionnaires (this final round of questions will be called the "1-month follow-up" survey). We expect this part to take approximately 10-25 minutes.

The questionnaires will include questions to help us better understand the background of you and your child such as age, ethnicity, and 22qDS diagnosis status as well as other questions such as, "This booklet answered questions I had regarding mental illness associated with 22q11.2DS" and "The booklet helped me to feel more confident in my ability to recognize early warning signs of mental illness."

10/19/2021 8:15pm

Confidentiality:

Your confidentiality will be respected. REDCap (Research Electronic Data Capture) is a secure software that is used to display the online surveys. Study data will be accessible through this software only to research staff. They will have personal passwords, and they will have a two-step login verification process to follow. This provides a security since the data is stored on secure University servers. Minimal contact information (email address) will be kept in REDCap. This contact information will be used to send you an individual follow-up survey link. It will also be used to provide you with a gift card if the study is completed. Only study team members will have access to your email address. Your email address will be marked as confidential information. This means that it will not be allowed to be downloaded from REDCap with any of the study data. Identifying information will be removed from your survey responses. All survey responses from participants will be combined for analysis. Your survey responses will not be linked back to your personal information, and they will only be accessible to members of the study team.

After 5 years, data from the surveys will be destroyed. This is the policy for the university.

For participants in Canada: Emails sent using some accounts such as Gmail, Hotmail, or others, are sometimes stored or directed outside of Canada (for example, to the United States). Emails may contain personal information about you, such as your first or last name contained in your email address. The Freedom of Information and Protection of Privacy Act requires that we get your consent before we continue. We will only send your personal information to the email address you have provided to us. All of the information you provide to us will be kept completely confidential. Providing your email address means that you voluntarily agree and give your consent for the study team to email your personal information (such as your email address) to you.

Compensation:

You will receive a $10.00 gift card upon your completion of the study. After completing the final 1-month follow-up survey, you will be asked whether you would like to receive an electronic gift card for Amazon.ca or Amazon.com.

Potential risks:

As with all research, you may be faced with small risks if you participate in this study. It is possible that thinking about your child's diagnosis and/or mental health when filling out a survey or reviewing the educational booklet may upset you. Options for where to access support are included in the resource. If your participation in the study has upset you or if the provided resources are insufficient, please contact the principal investigator Kayla Blankenship (kayla.blankenship@uscmed.sc.edu) or the study supervisor Crystal ¬¬¬¬¬¬¬Hill-Chapman (chillchapman@fmarion.com) and we will try to connect you with resources as needed.

Though unlikely, there is a chance that confidentiality could be compromised if the University servers are hacked from the outside.

Potential benefits:

There are no direct benefits to you from taking part in the study, although some people may find it helpful to review the information provided in the educational resource. Some people may view this study as a way to help benefit other 22q11.2DS families in the future. You will also receive an electronic copy of the finished resource when the study is complete.

Contact:

If you have any questions or would like more information, please contact Kayla Blankenship (kayla.blankenship@uscmed.sc.edu).

If you have any concerns or complaints about your rights as a research participant and/or your experiences while participating in this study, contact the Research Participant Complaint Line in the University of British Columbia Office of Research Ethics by email at RSIL@ors.ubc.ca or by phone at 604-822-8598 (Toll Free: 1-877-822-8598). Please reference the study number (H17-01148) when contacting the Complaint Line so the staff can better assist you.

Consent:

Your participation is entirely voluntary. It is up to you to decide whether or not to take part in this study. If you do your participation in the study will not have any negative consequences to the medical care or other services you or your child are currently receiving or will receive in the future.

Entering your email and continuing with the questionnaires indicates that you have read through and understood this consent form.

After reading through the consent form do you consent to participating in the study? ◯ Yes ◯ No

---

Please enter your email address: _____

We recommend that you save a copy of the consent form for your records. By clicking on the link a PDF version of the consent form will open on your computer, allowing you to save it if you wish.

[Attachment: "22q consent form.docx"]

Do you give permission for the research team to contact you regarding future research opportunities? ◯ Yes ◯ No

Figure A.1 Study consent form.

APPENDIX B

STUDY QUESTIONNAIRES

The following questionnaires were administered to the participants.

Demographics	
What sex were you assigned at birth, meaning on your original birth certificate?	◯ Male ◯ Female
Which best describes your current gender identity?	◯ Male ◯ Female ◯ Indigenous or other cultural gender minority identity (e.g. two-spirit) ◯ Something else (e.g. gender fluid, non-binary)
What gender do you currently live as in your day-to-day life?	◯ Male ◯ Female ◯ Sometimes male, sometimes female ◯ Something other than male or female
Enter your age:	_____ (Please enter a number only)
What is your ethnic origin (or race)?	◯ White/Caucasian/European ◯ Black or African American ◯ Hispanic or Latino ◯ Indigenous or Aboriginal ◯ Asian ◯ Other/Mixed
Specify:	_____
Where do you currently live?	◯ Canada ◯ The United States of America ◯ Europe ◯ South America ◯ Africa ◯ Asia ◯ Australia / South Pacific ◯ Other
What is the highest degree or level of school that you have completed? If currently enrolled, select the highest degree you have already received.	◯ Some high school, no diploma ◯ High school graduate, diploma or the equivalent (for example, GED) ◯ Some college credit, no degree ◯ Trade/technical/vocational training ◯ Bachelor's degree ◯ Master's degree ◯ Doctorate degree ◯ Other (please specify below)
Specify:	_____

Have you been diagnosed with 22q11.2 deletion syndrome?	◯ Yes ◯ No
Have you been diagnosed with a mental illness?	◯ Yes ◯ No
Do you feel you've experienced symptoms of a mental illness that have not been diagnosed?	◯ Yes ◯ No

Please answer the following questions regarding your child with 22q11.2 deletion syndrome

Is your child with 22q11.2 deletion younger than 1 year of age?	◯ Yes ◯ No
Please enter the current age of your child with 22q11.2 deletion syndome	_____ (years (enter number only))
Was your child diagnosed with 22q11.2 deletion during the pregnancy (prenatally)?	◯ Yes ◯ No
Was your child diagnosed with 22q11.2 deletion syndrome before the age of 1?	◯ Yes ◯ No
At what age was your child diagnosed with 22q11.2 deletion syndrome?	_____ (years (enter number only))
Please indicate how severely you feel your child is affected by their 22q11.2 deletion syndrome by using the sliding scale.	1 (Very Mild) 5 (Average) 10 (Very Severe) *(Place a mark on the scale above)*
Has your child ever been diagnosed with a psychiatric disorder / mental illness?	◯ Yes ◯ No
Please check all that apply	☐ Anxiety ☐ Depression ☐ Schizophrenia ☐ Obsessive Compulsive Disorder (OCD) ☐ Bipolar Disorder ☐ Other psychiatric diagnosis
Please specify the other psychiatric diagnosis:	_____
Has your child received any treatments (e.g. counselling, therapies, medications) for their [child_psych_dx] ?	◯ Yes ◯ No
Has your child ever had a mental health assessment?	◯ Yes ◯ No

	Very Inaccurate	Moderately Inaccurate	Neither accurate nor inaccurate	Moderately Accurate	Very Accurate
I am the life of the party.	◯	◯	◯	◯	◯
I sympathize with others' feelings.	◯	◯	◯	◯	◯
I get chores done right away.	◯	◯	◯	◯	◯
I have frequent mood swings.	◯	◯	◯	◯	◯

I have a vivid imagination.	○	○	○	○	○
I don't talk a lot.	○	○	○	○	○
I am not interested in other people's problems.	○	○	○	○	○
I often forget to put things back in their proper place.	○	○	○	○	○
I am relaxed most of the time.	○	○	○	○	○
I am not interested in abstract ideas.	○	○	○	○	○
I talk to a lot of different people at parties.	○	○	○	○	○
I feel others' emotions.	○	○	○	○	○
I like order.	○	○	○	○	○
I get upset easily.	○	○	○	○	○
I have difficulty understanding abstract ideas.	○	○	○	○	○
I keep in the background.	○	○	○	○	○
I am not really interested in others.	○	○	○	○	○
I make a mess of things.	○	○ Text	○	○	○
I seldom feel blue.	○	○	○	○	○
I do not have a good Imagination.	○	○	○	○	○

	Not at all characteristic of me	A little characteristic of me	Somewhat characteristic of me	Very characteristic of me	Entirely characteristic of me
Unforeseen events upset me greatly.	○	○	○	○	○
It frustrates me not having all the information I need.	○	○	○	○	○
Uncertainty keeps me from living a full life.	○	○	○	○	○
One should always look ahead so as to avoid surprises.	○	○	○	○	○
A small unforeseen event can spoil everything, even with the best of planning.	○	○	○	○	○
When it's time to act, uncertainty paralyzes me.	○	○	○	○	○
When I am uncertain I can't function very well.	○	○	○	○	○
I always want to know what the future has in store for me.	○	○	○	○	○
I can't stand being taken by surprise.	○	○	○	○	○
The smallest doubt can stop me from acting.	○	○	○	○	○
I should be able to organize everything in advance.	○	○	○	○	○
I must get away from all uncertain situations.	○	○	○	○	○

	Almost never	Occasionally	About half of the time	Often	Almost all of the time
I am not really sure what I think or believe about my problems.	○	○	○	○	○
I don't sustain my actions long enough to really solve my problems.	○	○	○	○	○
I think about ways that I solved similar problems in the past.	○	○	○	○	○
I identify the causes of my emotions, which helps me identify and solve my problems.	○	○	○	○	○
I feel so frustrated that I just give up doing any work on my problems at all.	○	○	○	○	○
I consider the short-term and long-term consequences of each possible solution to my problems.	○	○	○	○	○
I get preoccupied thinking about my problems and overemphasize some parts of them.	○	○	○	○	○
I continue to feel uneasy about my problems, which tells me I need to do some more work.	○	○	○	○	○
My old feelings get in the way of solving current problems.	○	○	○	○	○
I spend my time doing unrelated chores and activities instead of acting on my problems.	○	○	○	○	○
I think ahead, which enables me to anticipate and prepare for problems before they arise.	○	○	○	○	○
I think my problems through in a systematic way.	○	○	○	○	○
I misread another person's motives and feelings without checking with the other person to see if my conclusions are correct.	○	○	○	○	○
I get in touch with my feelings to identify and work on problems.	○	○	○	○	○
I act too quickly, which makes my problems worse.	○	○	○	○	○
I have a difficult time concentrating on my problems (i.e., my mind wanders).	○	○	○	○	○
I have alternate plans for solving my problems in case my first attempt does not work.	○	○	○	○	○
I avoid even thinking about my problems.	○	○	○	○	○

Using the scale below, circle a number next to each statement to indicate how much you agree with the statement.
Please answer all the questions. For questions that are not applicable to you (i.e. Questions 1, 14, 23),
please choose option 4 (neither agree nor disagree).

When it refers to "the condition" this is referring to the "psychiatric manifestations of 22q11.2 deletion syndrome"

	Strongly disagree	Disagree	Slightly disagree	Neither disagree nor agree	Slightly agree	Agree	Strongly agree
1. I am clear in my own mind why I am attending the clinical genetics service.	○	○	○	○	○	○	○
2. I can explain what the condition means to people in my family who may need to know.	○	○	○	○	○	○	○
3. I understand the impact of the condition on my child(ren)/any child I may have.	○	○	○	○	○	○	○

	Strongly disagree	Disagree	Slightly disagree	Neither disagree nor agree	Slightly agree	Agree	Strongly agree
4. When I think about the condition in my family, I get upset.	○	○	○	○	○	○	○
5. I don't know where to go to get medical help I/my family needs.	○	○	○	○	○	○	○

	Strongly disagree	Disagree	Slightly disagree	Neither disagree nor agree	Slightly agree	Agree	Strongly agree
6. I can see that good things have come from having this condition in my family.	○	○	○	○	○	○	○
7. I can control how this condition affects my family.	○	○	○	○	○	○	○
8. I feel positive about the future.	○	○	○	○	○	○	○
9. I am able to cope with having this condition in my family.	○	○	○	○	○	○	○
10. I don't know what could be gained from each of the options available to me.	○	○	○	○	○	○	○
11. Having this condition in my family makes me feel anxious.	○	○	○	○	○	○	○
12. I don't know if this condition could affect my other relatives (brothers, sisters, aunts, uncles, cousins).	○	○	○	○	○	○	○

13. In relation to the condition in my family, nothing I decide will change the future for my children/any children I might have.	○	○	○	○	○	○	○
	Strongly disagree	Disagree	Slightly disagree	Neither disagree nor agree	Slightly agree	Agree	Strongly agree
14. I understand the reasons why my doctor referred me to the clinical genetics service.	○	○	○	○	○	○	○
15. I know how to get the non-medical help I/my family needs (e.g. educational, financial, social support).	○	○	○	○	○	○	○
16. I can explain what the condition means to people outside my family who may need to know (e.g. teachers, social workers).	○	○	○	○	○	○	○
	Strongly disagree	Disagree	Slightly disagree	Neither disagree nor agree	Slightly agree	Agree	Strongly agree
17. I don't know what I can do to change how this condition affects me/my children.	○	○	○	○	○	○	○
18. I don't know who else in my family might be at risk for this condition.	○	○	○	○	○	○	○
	Strongly disagree	Disagree	Slightly disagree	Neither disagree nor agree	Slightly agree	Agree	Strongly agree
19. I am hopeful that my children can look forward to a rewarding family life.	○	○	○	○	○	○	○
20. I am able to make plans for the future.	○	○	○	○	○	○	○
	Strongly disagree	Disagree	Slightly disagree	Neither disagree nor agree	Slightly agree	Agree	Strongly agree
21. I feel guilty because I (might have) passed this condition on to my children.	○	○	○	○	○	○	○
22. I am powerless to do anything about this condition in my family.	○	○	○	○	○	○	○
	Strongly disagree	Disagree	Slightly disagree	Neither disagree nor agree	Slightly agree	Agree	Strongly agree
23. I understand what concerns brought me to the clinical genetics service.	○	○	○	○	○	○	○

24. I can make decisions about the condition that may change my child(ren)'s future/the future of any child(ren) I may have.	○	○	○	○	○	○	○

The following questions ask how you currently feel about your family member's (i.e your child with 22q11.2) mental illness. Although we use the term 'mental illness' please think of this in whichever way you feel most comfortable. If you have had a diagnosis of mental illness yourself (or another relative with a mental illness), as you answer the questions, please focus on how you feel about mental illness in your child with 22q11.2 DS, rather than your own diagnosis or that of another relative. Please mark whether you strongly disagree, disagree, feel neutral, agree, or strongly agree.

	Strongly Disagree	Disagree	Neutral	Agree	Strongly Agree
1. I feel embarrassed that I have a family member with a mental illness	○	○	○	○	○
2. Having a family member with a mental illness has made me more concerned about my own mental health	○	○	○	○	○
3. People don't want to talk to me about my family member's mental illness	○	○	○	○	○
4. I am concerned about being labeled as someone who has a family member with a mental illness	○	○	○	○	○
5. People blame me for my family member's mental illness	○	○	○	○	○
6. I blame myself for my family member's mental illness	○	○	○	○	○
7. I feel discriminated against because I have a family member with a mental illness	○	○	○	○	○
8. I feel isolated because I have a family member with a mental illness	○	○	○	○	○
9. I minimize the severity of my family member's mental illness when describing it to people	○	○	○	○	○
10. My self-esteem has been damaged because of my family member's mental illness	○	○	○	○	○

When answering these questions, please consider how you've felt over the PAST MONTH about your child with 22q11.2 deletion syndrome.

How often have you thought about your child's chance of getting a mental illness (again)?

○ 1 never
○ 2 sometimes
○ 3 frequently
○ 4 almost always

Have these thoughts affected your mood?	◯ 1 no, not at all ◯ 2 no, not much ◯ 3 yes, a little ◯ 4 yes, a lot
Have these thoughts interfered with your ability to do daily activities?	◯ 1 no, not at all ◯ 2 no, not much ◯ 3 yes, a little ◯ 4 yes, a lot
How concerned are you about the possibility of your child getting a mental illness (again) one day?	◯ 1 not concerned ◯ 2 only a little concerned ◯ 3 somewhat concerned ◯ 4 very concerned
How often do you worry about your child developing a mental illness (again)?	◯ 1 never ◯ 2 sometimes ◯ 3 frequently ◯ 4 almost always
How much of a problem is this worry?	◯ 1 not at all a problem ◯ 2 only a little bit of a problem ◯ 3 somewhat of a problem ◯ 4 very much a problem
How concerned are you about the possibility that your child will ever need treatment for a mental illness (again)?	◯ 1 not concerned ◯ 2 only a little concerned ◯ 3 somewhat concerned ◯ 4 very concerned
How often do you worry about the chance of other family members developing a mental illness (again)?	◯ 1 never ◯ 2 sometimes ◯ 3 frequently ◯ 4 almost always

Figure B.1 Pre-Viewing Questionnaire (T1).

Below are some questions asking about your thoughts and opinions about the resource.

As you go through the next questions about the resource, it may be helpful for you to refer to the booklet. Here is a downloadable pdf of the resource for your own reference.

[Attachment: "22qDS 2020_finale.pdf"]

I have completed reading the booklet.	◯ Yes ◯ No
How many minutes did it take you to read the booklet?	_____
The information in the booklet was easy to understand.	◯ Strongly agree ◯ Agree ◯ Neither agree nor disagree ◯ Disagree ◯ Strongly disagree
I feel I have a better understanding of 22q11.2 deletion syndrome and mental illness after reading this booklet.	◯ Strongly agree ◯ Agree ◯ Neither agree nor disagree ◯ Disagree ◯ Strongly disagree

This booklet answered questions I had regarding mental illness associated with 22q11.2 deletion syndrome.	◯ Strongly agree ◯ Agree ◯ Neither agree nor disagree ◯ Disagree ◯ Strongly disagree
I feel this booklet would be helpful for other families who have a child with 22q11.2 deletion syndrome.	◯ Strongly agree ◯ Agree ◯ Neither agree nor disagree ◯ Disagree ◯ Strongly disagree
I feel this booklet helped me understand strategies that can be used to protect my child's mental health.	◯ Strongly agree ◯ Agree ◯ Neither agree nor disagree ◯ Disagree ◯ Strongly disagree
I feel less worried about the risks for psychiatric disorders in 22q11.2 deletion syndrome after reading through this booklet.	◯ Strongly agree ◯ Agree ◯ Neither agree nor disagree ◯ Disagree ◯ Strongly disagree
The booklet helped me to feel more confident in my ability to recognize early warning signs of mental illness.	◯ Strongly agree ◯ Agree ◯ Neither agree nor disagree ◯ Disagree ◯ Strongly disagree
What did you like about the booklet?	_____
What did you not like about the booklet?	_____
Please comment on when you think this resource should be given to parents of children with 22q11.2 deletion syndrome:	_____
Was there anything that you felt was missing from the booklet?	_____
Was there anything that you felt was not necessary in the booklet?	_____
If you have any additional thoughts or comments about the booklet, please write them here.	_____

Figure B.2 Immediate Post-Viewing Questionnaire.

In the last month, have you scheduled or had any new mental health assessments for your child?	○ Yes ○ No
In the last month, have you scheduled or had any new mental health treatments or therapies for your child?	○ Yes ○ No

	Very Inaccurate	Moderately Inaccurate	Neither accurate nor inaccurate	Moderately Accurate	Very Accurate
I am the life of the party.	○	○	○	○	○
I sympathize with others' feelings.	○	○	○	○	○
I get chores done right away.	○	○	○	○	○
I have frequent mood swings.	○	○	○	○	○
I have a vivid imagination.	○	○	○	○	○
I don't talk a lot.	○	○	○	○	○
I am not interested in other people's problems.	○	○	○	○	○
I often forget to put things back in their proper place.	○	○	○	○	○
I am relaxed most of the time.	○	○	○	○	○
I am not interested in abstract ideas.	○	○	○	○	○
I talk to a lot of different people at parties.	○	○	○	○	○
I feel others' emotions.	○	○	○	○	○
I like order.	○	○	○	○	○
I get upset easily.	○	○	○	○	○
I have difficulty understanding abstract ideas.	○	○	○	○	○
I keep in the background.	○	○	○	○	○
I am not really interested in others.	○	○	○	○	○
I make a mess of things.	○	○ Text	○	○	○
I seldom feel blue.	○	○	○	○	○
I do not have a good Imagination.	○	○	○	○	○

	Not at all characteristic of me	A little characteristic of me	Somewhat characteristic of me	Very characteristic of me	Entirely characteristic of me
Unforeseen events upset me greatly.	○	○	○	○	○
It frustrates me not having all the information I need.	○	○	○	○	○
Uncertainty keeps me from living a full life.	○	○	○	○	○
One should always look ahead so as to avoid surprises.	○	○	○	○	○
A small unforeseen event can spoil everything, even with the best of planning.	○	○	○	○	○
When it's time to act, uncertainty paralyzes me.	○	○	○	○	○

When I am uncertain I can't function very well.	○	○	○	○	○
I always want to know what the future has in store for me.	○	○	○	○	○
I can't stand being taken by surprise.	○	○	○	○	○
The smallest doubt can stop me from acting.	○	○	○	○	○
I should be able to organize everything in advance.	○	○	○	○	○
I must get away from all uncertain situations.	○	○	○	○	○

	Almost never	Occasionally	About half of the time	Often	Almost all of the time
I am not really sure what I think or believe about my problems.	○	○	○	○	○
I don't sustain my actions long enough to really solve my problems.	○	○	○	○	○
I think about ways that I solved similar problems in the past.	○	○	○	○	○
I identify the causes of my emotions, which helps me identify and solve my problems.	○	○	○	○	○
I feel so frustrated that I just give up doing any work on my problems at all.	○	○	○	○	○
I consider the short-term and long-term consequences of each possible solution to my problems.	○	○	○	○	○
I get preoccupied thinking about my problems and overemphasize some parts of them.	○	○	○	○	○
I continue to feel uneasy about my problems, which tells me I need to do some more work.	○	○	○	○	○
My old feelings get in the way of solving current problems.	○	○	○	○	○
I spend my time doing unrelated chores and activities instead of acting on my problems.	○	○	○	○	○
I think ahead, which enables me to anticipate and prepare for problems before they arise.	○	○	○	○	○
I think my problems through in a systematic way.	○	○	○	○	○
I misread another person's motives and feelings without checking with the other person to see if my conclusions are correct.	○	○	○	○	○

I get in touch with my feelings to identify and work on problems.	○	○	○	○	○
I act too quickly, which makes my problems worse.	○	○	○	○	○
I have a difficult time concentrating on my problems (i.e., my mind wanders).	○	○	○	○	○
I have alternate plans for solving my problems in case my first attempt does not work.	○	○	○	○	○
I avoid even thinking about my problems.	○	○	○	○	○

Using the scale below, circle a number next to each statement to indicate how much you agree with the statement.
Please answer all the questions. For questions that are not applicable to you (i.e. Questions 1, 14, 23),
please choose option 4 (neither agree nor disagree).

When it refers to "the condition" this is referring to the "psychiatric manifestations of 22q11.2 deletion syndrome"

	Strongly disagree	Disagree	Slightly disagree	Neither disagree nor agree	Slightly agree	Agree	Strongly agree
1. I am clear in my own mind why I am attending the clinical genetics service.	○	○	○	○	○	○	○
2. I can explain what the condition means to people in my family who may need to know.	○	○	○	○	○	○	○
3. I understand the impact of the condition on my child(ren)/any child I may have.	○	○	○	○	○	○	○

	Strongly disagree	Disagree	Slightly disagree	Neither disagree nor agree	Slightly agree	Agree	Strongly agree
4. When I think about the condition in my family, I get upset.	○	○	○	○	○	○	○
5. I don't know where to go to get medical help I/my family needs.	○	○	○	○	○	○	○

	Strongly disagree	Disagree	Slightly disagree	Neither disagree nor agree	Slightly agree	Agree	Strongly agree
6. I can see that good things have come from having this condition in my family.	○	○	○	○	○	○	○
7. I can control how this condition affects my family.	○	○	○	○	○	○	○
8. I feel positive about the future.	○	○	○	○	○	○	○
9. I am able to cope with having this condition in my family.	○	○	○	○	○	○	○

10. I don't know what could be gained from each of the options available to me.	○	○	○	○	○	○	○
11. Having this condition in my family makes me feel anxious.	○	○	○	○	○	○	○
12. I don't know if this condition could affect my other relatives (brothers, sisters, aunts, uncles, cousins).	○	○	○	○	○	○	○
13. In relation to the condition in my family, nothing I decide will change the future for my children/any children I might have.	○	○	○	○	○	○	○

	Strongly disagree	Disagree	Slightly disagree	Neither disagree nor agree	Slightly agree	Agree	Strongly agree
14. I understand the reasons why my doctor referred me to the clinical genetics service.	○	○	○	○	○	○	○
15. I know how to get the non-medical help I/my family needs (e.g. educational, financial, social support).	○	○	○	○	○	○	○
16. I can explain what the condition means to people outside my family who may need to know (e.g. teachers, social workers).	○	○	○	○	○	○	○

	Strongly disagree	Disagree	Slightly disagree	Neither disagree nor agree	Slightly agree	Agree	Strongly agree
17. I don't know what I can do to change how this condition affects me/my children.	○	○	○	○	○	○	○
18. I don't know who else in my family might be at risk for this condition.	○	○	○	○	○	○	○

	Strongly disagree	Disagree	Slightly disagree	Neither disagree nor agree	Slightly agree	Agree	Strongly agree
19. I am hopeful that my children can look forward to a rewarding family life.	○	○	○	○	○	○	○
20. I am able to make plans for the future.	○	○	○	○	○	○	○

	Strongly disagree	Disagree	Slightly disagree	Neither disagree nor agree	Slightly agree	Agree	Strongly agree
21. I feel guilty because I (might have) passed this condition on to my children.	○	○	○	○	○	○	○
22. I am powerless to do anything about this condition in my family.	○	○	○	○	○	○	○

	Strongly disagree	Disagree	Slightly disagree	Neither disagree nor agree	Slightly agree	Agree	Strongly agree
23. I understand what concerns brought me to the clinical genetics service.	○	○	○	○	○	○	○
24. I can make decisions about the condition that may change my child(ren)'s future/the future of any child(ren) I may have.	○	○	○	○	○	○	○

The following questions ask how you currently feel about your family member's (i.e your child with 22q11.2) mental illness. Although we use the term 'mental illness' please think of this in whichever way you feel most comfortable. If you have had a diagnosis of mental illness yourself (or another relative with a mental illness), as you answer the questions, please focus on how you feel about mental illness in your child with 22q11.2 DS, rather than your own diagnosis or that of another relative. Please mark whether you strongly disagree, disagree, feel neutral, agree, or strongly agree.

	Strongly Disagree	Disagree	Neutral	Agree	Strongly Agree
1. I feel embarrassed that I have a family member with a mental illness	○	○	○	○	○
2. Having a family member with a mental illness has made me more concerned about my own mental health	○	○	○	○	○
3. People don't want to talk to me about my family member's mental illness	○	○	○	○	○
4. I am concerned about being labeled as someone who has a family member with a mental illness	○	○	○	○	○
5. People blame me for my family member's mental illness	○	○	○	○	○
6. I blame myself for my family member's mental illness	○	○	○	○	○
7. I feel discriminated against because I have a family member with a mental illness	○	○	○	○	○
8. I feel isolated because I have a family member with a mental illness	○	○	○	○	○
9. I minimize the severity of my family member's mental illness when describing it to people	○	○	○	○	○
10. My self-esteem has been damaged because of my family member's mental illness	○	○	○	○	○

When answering these questions, please consider how you've felt over the PAST MONTH about your child with 22q11.2 deletion syndrome.	
How often have you thought about your child's chance of getting a mental illness (again)?	◯ 1 never ◯ 2 sometimes ◯ 3 frequently ◯ 4 almost always
Have these thoughts affected your mood?	◯ 1 no, not at all ◯ 2 no, not much ◯ 3 yes, a little ◯ 4 yes, a lot
Have these thoughts interfered with your ability to do daily activities?	◯ 1 no, not at all ◯ 2 no, not much ◯ 3 yes, a little ◯ 4 yes, a lot
How concerned are you about the possibility of your child getting a mental illness (again) one day?	◯ 1 not concerned ◯ 2 only a little concerned ◯ 3 somewhat concerned ◯ 4 very concerned
How often do you worry about your child developing a mental illness (again)?	◯ 1 never ◯ 2 sometimes ◯ 3 frequently ◯ 4 almost always
How much of a problem is this worry?	◯ 1 not at all a problem ◯ 2 only a little bit of a problem ◯ 3 somewhat of a problem ◯ 4 very much a problem
How concerned are you about the possibility that your child will ever need treatment for a mental illness (again)?	◯ 1 not concerned ◯ 2 only a little concerned ◯ 3 somewhat concerned ◯ 4 very concerned
How often do you worry about the chance of other family members developing a mental illness (again)?	◯ 1 never ◯ 2 sometimes ◯ 3 frequently ◯ 4 almost always

Figure B.3 One Month Post-Viewing Questionnaire.

APPENDIX C

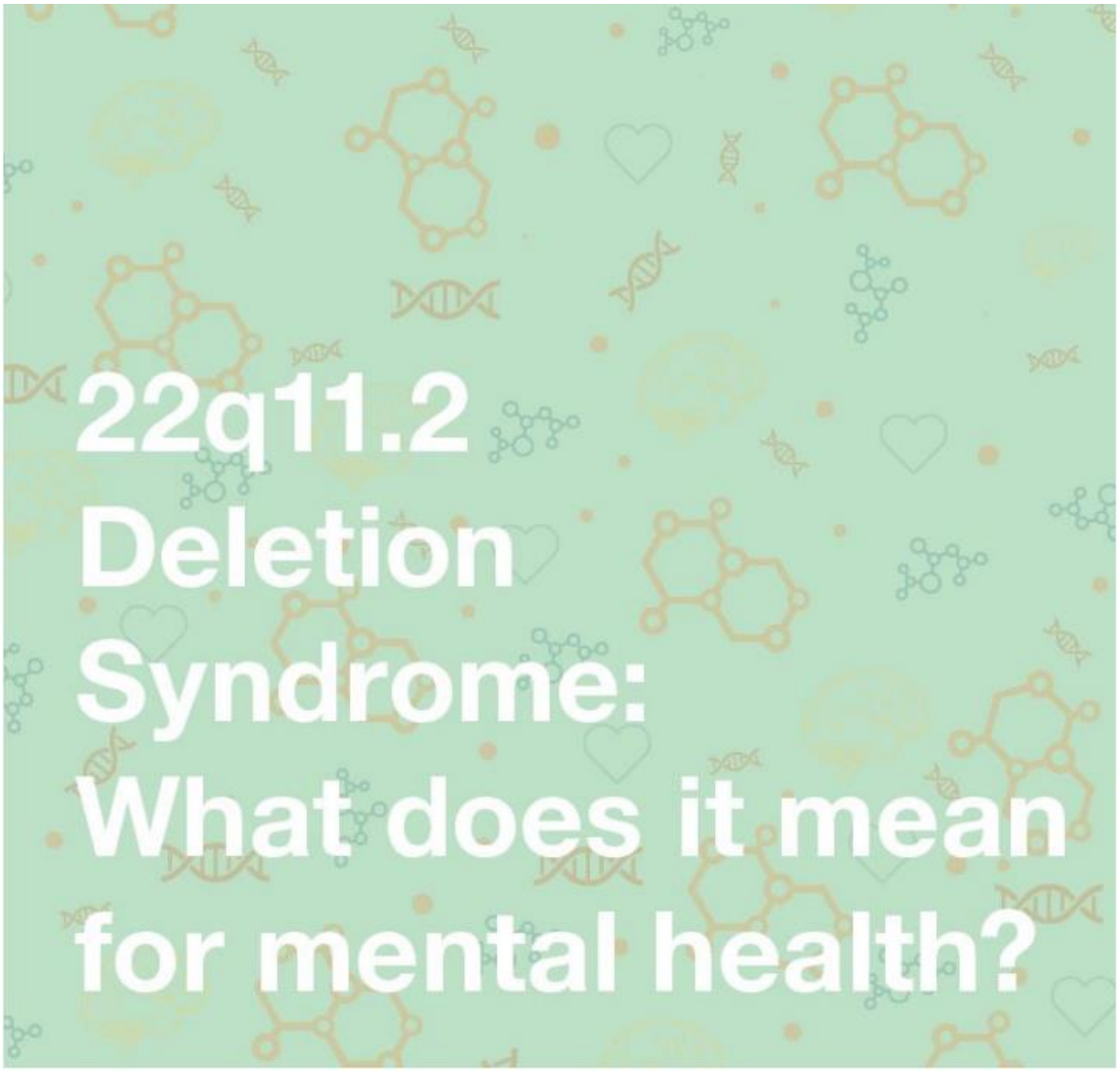
EDUCATIONAL RESOURCE BOOKLET

The resource booklet used in the study is pictured below.

Thank you for completing those questions.

This next section contains the online resource booklet about 22q11.2 deletion syndrome and mental health. Please take your time to review the booklet. You can go back and forth between the pages. You can also click "save and return later" if you need more time to read it.

When you are done reading the resource, you will also have the option of downloading a PDF of the resource booklet for yourself to keep.



Who is this booklet for?

This booklet was developed as a resource about 22q11.2 deletion syndrome (22qDS). It contains information about how 22qDS can impact mental health and provides practical strategies and recommendations to protect and improve mental health.

This is not a comprehensive guide to treating mental illness and you should contact your local mental health providers for further guidance specific to your situation.

This booklet was developed to be a reliable source of information for families. It was developed by genetic counselors and families who live with 22qDS working together.

This booklet is intended to be a supplement to genetic counseling. You can find a genetic counselor in your area using the National Society of Genetic Counselors' Find a Genetic Counselor tool at nsgc.org.

What is 22qDS?

22qDS (22q11.2 deletion syndrome) is also called Velocardiofacial syndrome (VCFS) and DiGeorge syndrome (DGS). About 1 in 4000 people have 22qDS. It is a condition caused by a small missing (deleted) piece of chromosome 22. This missing piece of genetic material can contribute to developmental and/or health issues. There is a lot of variation between people with 22qDS in terms of whether they have health issues, and how serious these are.

Some of the more common health issues that an individual with 22qDS may develop are listed below. Not everyone with 22qDS will have all of the health issues listed.

- Heart defects
- Cleft palate (an opening at the roof of the mouth)
- Learning difficulties
- Immune system problems (being more vulnerable to infections)
- Differences in facial appearance

Additional health issues that may occur are:

- Growth delays
- Feeding difficulties
- Kidney problems
- Low levels of calcium
- Hearing loss
- Developmental delays
- Speech delay
- Seizures
- Bone differences
- Eye problems
- Gastrointestinal problems
- Mental illness*

*This booklet will focus on mental illness.

What is mental illness?

Mental illness refers to a wide range of conditions that affect how people think, feel and/or behave. Mental illness can cause distress but there are a variety of treatments available, and people can recover. Even if a person does not experience mental illness, it is important for everyone to take care of their mental health. A selection of mental illnesses are described below:

Anxiety Disorders

Anxiety disorders involve excessive fear or anxiety beyond that of normal nervousness or anxiousness, and may include specific phobias or panic disorders.

Schizophrenia

Schizophrenia involves delusions (beliefs that are not based in reality), hallucinations (e.g. seeing or hearing things that are not there), and trouble with thinking and concentration.

See page 15 for more information about schizophrenia.

Mood Disorders

"Mood disorders" is a term that covers both depression (low mood) and bipolar disorders (periods of low mood and periods of "high" or irritable mood).

Schizoaffective Disorder

Schizoaffective disorder involves a combination of symptoms of schizophrenia and symptoms of mood disorders.

Obsessive Compulsive Disorder (OCD)

OCD involves symptoms that include uncontrollable, reoccurring thoughts (obsessions) and behaviors (compulsions) that people engage in to try to cope with that obsession.

What causes mental illness?

The Mental Illness Jar

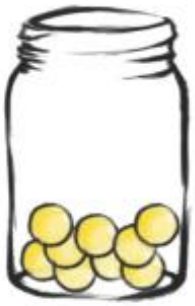
Most of the time mental illness does not have one single cause. Mental illness is typically caused by a combination of genetic and environmental factors ("nature" and "nurture").

The following pictures use a jar to represent how **genetic (inherited) factors** and **environmental factors** work together to cause mental illness. Everyone has a mental illness jar, which can be filled with both genetic and environmental factors.

If the mental illness jar becomes full, a person experiences an episode of mental illness. If the jar is not full, a person is not experiencing an episode of mental illness. The closer the jar becomes to being full, the more vulnerable to mental illness the person is.

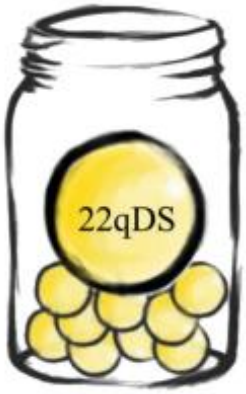
Genetic Factors

Genetic factors are represented in the jar as yellow balls. Research has shown that likely everyone has some genetic factors for mental illness. There are different types of genetic factors for mental illness. The most common type of genetic factor that people can have is represented by a small yellow ball. Some people have a small amount of them and some have a large amount, but most people have a medium amount. Each one of this type of genetic factor makes only a very small difference to whether a person develops mental illness.



22qDS - The Big Yellow Ball

Missing or extra pieces of chromosomes - like the one that causes 22qDS - are a different type of genetic factor for mental illness. These are less common in the general population, but they can make a bigger difference to whether or not a person develops mental illness.



We use a large yellow ball in the mental illness jar to represent 22qDS because studies have shown that this genetic factor makes a bit of a bigger difference to whether or not someone develops mental illness.

People with 22qDS likely have other smaller yellow balls too (just like everyone else does) in their mental illness jar.

Environmental Factors

Environmental factors can be thought of as "life experiences". We don't know what all of the environmental factors for mental illness are, but some of the ones we do know about are listed below.

We are all different in how sensitive we are to environmental factors, like stress. Stress can include both positive and negative experiences. People who are more sensitive may be more likely to develop mental illness after a stressful experience. The differences in how sensitive people are might be due to having different amounts of genetic factors in one's mental illness jar.

Environmental factors that make a person more likely to have either **schizophrenia** or **schizoaffective disorder** or **bipolar disorder**:

- Being born in winter months
- Experiencing stressful life events
- Taking certain illegal drugs (like cannabis or methamphetamine)

Environmental factors that make a person more likely to have **schizophrenia**:

- Being born after a difficult pregnancy or delivery
- Being brought up in a large city
- Being an immigrant
- Having a childhood head injury

Most of these experiences make it only slightly more likely that a person will develop mental illness. For example, for the average person in the general population, the chance to develop schizophrenia is about 1% (1 out of 100 people). But, someone whose birth was difficult is twice as likely to have a mental illness, like schizophrenia, than someone whose birth was not difficult. This means that a person whose birth was difficult has a chance to develop schizophrenia of 2% rather than 1%.

Environmental Factors

22qDS

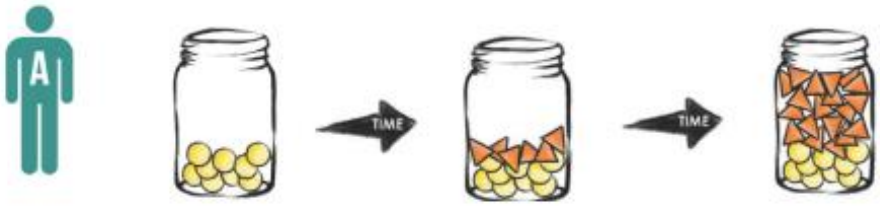
Below are some environmental factors that a person with 22qDS may be more likely to experience than a person without 22qDS.

- Frequent medical appointments or visits to the hospital
- Difficulties making friends
- Feeling different than their peers or siblings, or experiencing bullying
- Finding school stressful ordifficult

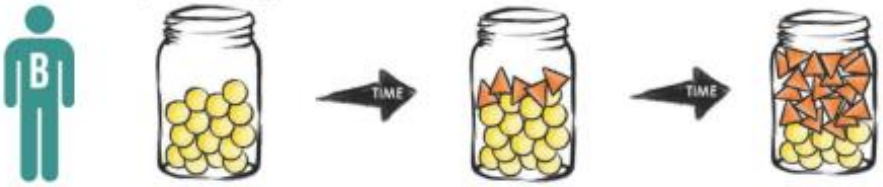
Putting It All Together

Mental illness can be caused by different combinations of genetic and environmental factors. It is not anyone's fault when someone develops a mental illness.

Person A has a small amount of genetic factors, and has experienced a large amount of environmental factors that have filled up their mental illness jar.



Person B has a larger amount of genetic factors and a smaller amount of environmental factors which has resulted in their mental illness jar being filled.



Person C has 22qDS and has some other, more common, genetic factors as well. They have also experienced environmental factors, which filled up their mental illness jar.

A person who has a large amount of genetic factors – including individuals with 22qDS – may be more likely to develop mental illness than someone who has only a small amount.

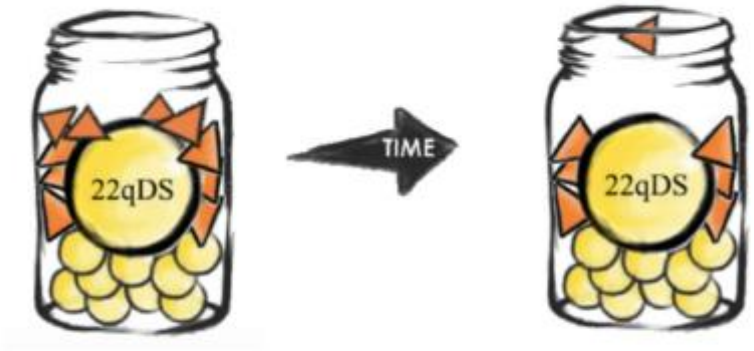
But, a person with 22qDS may not develop mental illness.

Protecting Mental Health in People with 22qDS

People with 22qDS can do things to protect their mental health.

The rest of this booklet will focus specifically on how to take care of mental health. Most of the concepts apply for people without 22qDS as well as those that have 22qDS.

People cannot change their genetic factors; however, some environmental factors can be avoided, changed or removed from the mental illness jar. Someone might remove environmental factors by:

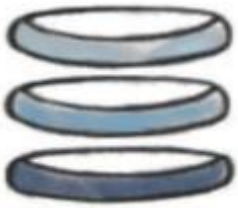


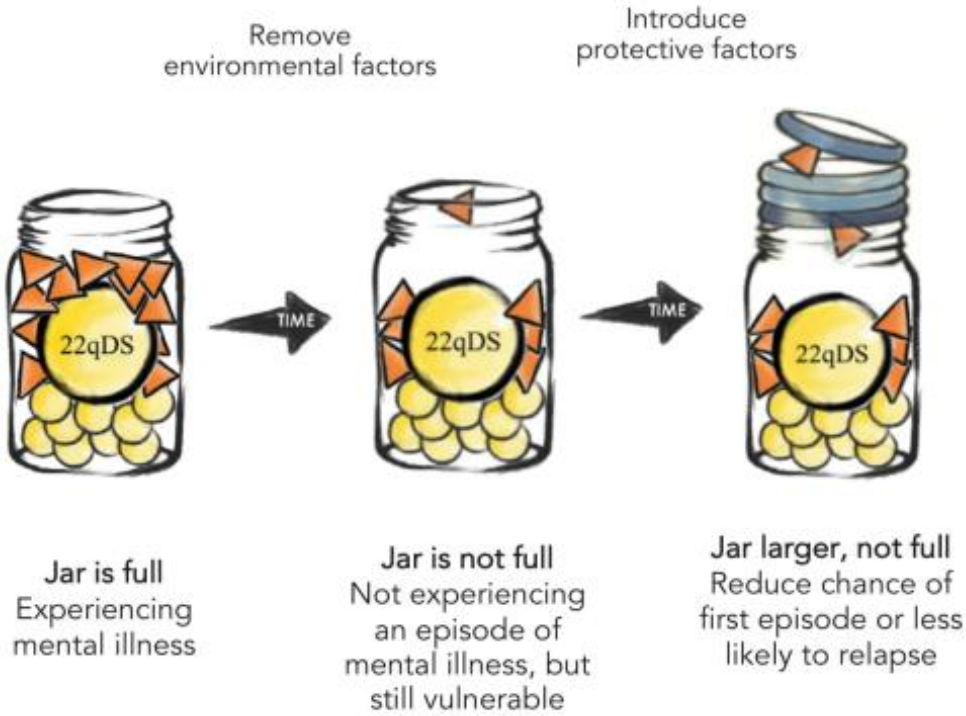
- **Finding ways to reduce stress**
  - See page 12 for some examples of activities to reduce stress

- **Not taking drugs like cannabis and methamphetamine**

Protecting Mental Health in People with 22qDS

People with 22qDS can also protect their mental health by adding protective factors onto their jars. Protective factors can be thought of as rings that stack on top of the jar to make it bigger. This makes it less likely for the jar to fill to the top. Protective factors can make more room if someone's jar is already full and help with recovery.

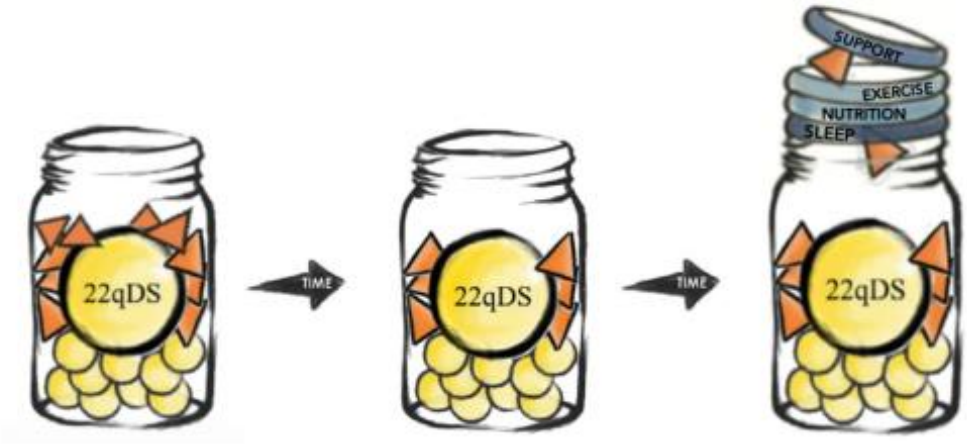




Protecting Mental Health in People with 22qDS

Research has shown that the following things are protective of mental health in everyone:

- **Exercise:** some form of regular exercise or physical activity
- **Nutrition:** regular meals and a healthy balanced diet
- **Sleep:** adequate and consistent sleep
- A good **support system** of people you trust



People with 22qDS are also recommended to have **regular check-ins with a mental health professional** (psychiatrist or psychologist) **starting in early childhood** (i.e. before age 5). These visits can help monitor the individual for any symptoms and can connect parents with appropriate treatments and resources. Early intervention is one of the best ways of improving outcomes for people with mental illness. This professional may be part of a 22qDS clinic or care team, or may be a separate part of your or your child's care.

Protecting Mental Health in People with 22qDS

As well as the protective factors that work for all of us, there are others that different people may find helpful. Some examples are listed below:

- Stretching
- Yoga
- A hot bath
- Meditation
- Spending time with a pet
- Talking to someone
- Taking a break from the stressor
- Doing something that makes you laugh
- Listening to your favorite music
- Looking through old family photos
- Deep breathing
- Reading a book
- Working on a coloring book
- Painting
- Gardening
- Going to the park
- Dancing
- Making a craft
- Playing a game
- Asking for help
- Work through a mental health workbook
- Make a mental health plan

It is important for individuals to find what works best for them and their family. Encouraging healthy habits and having a safe space to discuss feelings can be very helpful for protecting a person's mental health.

What is the chance for a person with 22qDS to develop mental illness?

Mental illness is common; the chance for someone without 22qDS to develop any mental illness is ~20%. The most common mental illnesses are depression and anxiety, and 1% of people develop schizophrenia.

A person with 22qDS is more likely to experience mental illness than a person without 22qDS, but the chance of developing a mental health problem is difficult to estimate. The most common mental illnesses, like for someone without 22qDS, are depression and anxiety.

More studies have been done about schizophrenia and 22qDS:

About 25% (1 in 4) people with 22qDS will develop schizophrenia. This means that most people (75% or 3 out of 4 people) with 22qDS do **not** develop schizophrenia.

*See page 15 for information about schizophrenia.

Out of 100 people with 22qDS, approximately 25 will develop schizophrenia in their lifetime and 75 will not develop schizophrenia.

22qDS and Mental Illness Facts

When mental illness happens, the symptoms most often start during the **late teens to early 20's**. This is similar between people with and without 22qDS.

Early diagnosis and treatment for mental illness leads to the best outcomes for people over the long term. This is why regular mental health monitoring is important for children with 22qDS.

*See page 16-17 for how to recognize emerging symptoms of mental illness and pages 11-12 for protective and wellness factors.

Researchers are trying to determine whether there are treatments for mental illness that are specific to 22qDS. Currently, mental illness treatment options are similar between people who do and do not have 22qDS.

Individualized treatment plans for mental illness can be developed between mental health professionals and the family. Possible treatment options include:

- Medication
- Different types of talk therapy (psychotherapy), including Cognitive Behavioral Therapy (CBT)
- Psychoeducation
- An in-patient stay in the hospital

Treatment often uses a combination of therapy and medication.

Schizophrenia

Schizophrenia can cause a change in the way a person thinks, feels, and behaves. During an active episode, a person with schizophrenia may be unable to distinguish between what is real and what is not real. Types of symptoms that may occur in schizophrenia are listed below.

Positive symptoms are changes in thoughts and behavior that are "added" to a person's experiences. These symptoms are often referred to **psychosis** and represent a loss of touch with reality. Some examples are:

- Hallucinations (hearing, seeing, feeling, tasting or smelling something that is not there)
- Delusions (false or mistaken beliefs that are firmly held)
- Paranoia

Negative symptoms are thoughts or behaviors that exist before a person gets sick, but are not present once a person is ill.

- Less ability to plan, reduced speech or difficulty expressing emotion

Cognitive symptoms

- Problems with attention, concentration or memory
- Confused or disordered ("disorganized") thinking and speech

Symptoms of schizophrenia typically begin between the ages of 16 and 30. People with schizophrenia may not be aware that they are developing symptoms of mental illness, so family or friends may be the first to recognize changes and seek help.

Recognizing Emerging Symptoms of Mental Illness

Early intervention (treatment) has been shown to improve long-term outcomes for people with mental illness. Recognizing emerging symptoms of mental illness can help with getting early treatment. Examples of emerging symptoms are listed below:

Anxiety:

- Constantly worrying or having negative thoughts
- Sleep or appetite changes
- Complaining of physical ailments (e.g. stomach aches and headaches)
- Feelings of panic (e.g. heart racing or difficulty breathing)
- Problems thinking or troubles concentrating
- Drop in functioning (for example in grades or school performance)
- Avoiding specific situations (e.g. school or work)
- Irritability

Mood Disorders:

- Lack of interest or enthusiasm for things that used to be interesting or enjoyable
- Mood changes/extreme shifts between highs and lows
- Feeling disconnected
- Sleep or appetite changes
- Irritability
- Drop in functioning (for example in grades or school performance)
- Difficulty Concentrating
- Low energy
- Withdrawal/spending more time alone than usual

Recognizing Emerging Symptoms of Mental Illness

Psychosis:
In addition to the symptoms listed on the previous page, emerging symptoms of psychosis (schizophrenia) may also include:

- Increased sensitivity to smell, sound, touch, etc.
- Illogical thinking
- Difficulty telling what is real from what is not
- Nervousness or suspiciousness beyond what is typical for them
- Unusual behavior
- Loss of personal hygiene or self-care

A person who experiences some of these symptoms does not necessarily have an emerging mental illness. But if you are concerned about how frequent or severe these symptoms are, or about a sudden change in behavior, it can be helpful to check with a mental health professional, like a psychiatrist or psychologist.

Things To Remember

1. Mental illness is caused by BOTH genes and environment. Usually, inherited (genetic) factors and environmental (lifestyle) factors contribute together to cause mental illness.

2. 22qDS is one genetic factor that contributes to someone's susceptibility to mental illness, but does not by itself cause mental illness.

3. A person who has a larger amount of genetic factors (e.g. a person with 22qDS) may be more likely to develop mental illness than someone who has a smaller amount. But, not everyone with 22qDS will develop a mental illness.

4. People with 22qDS who have had an episode of mental illness can recover. There are treatments for mental illness.

5. Environmental factors can sometimes be removed from the mental illness jar, but we cannot control or prevent all environmental factors.

6. There are things to protect mental health for the future. Some suggestions are listed on pages 11 and 12. Protective factors can reduce the chance of experiencing a first episode of mental illness or make it less likely that another episode occurs.

7. If you are worried that someone might have symptoms of mental illness, get help quickly. Options listed on pages 19 might help.

8. Regular mental health check-ins should be a routine part of care for people with 22qDS.

Mental Health Resources

Emergency mental health resources:

- For anything urgent or life-threatening call 911 or your local emergency number immediately.

- Find a Crisis Line in your area. For help finding a Crisis Line visit yourlifecounts.org.

Routine, ongoing mental health resources:

- See a psychologist or psychiatrist regularly, starting in early childhood if possible.

- Seek help from your primary care provider, 22qDS team, or other health care provider.

Taking care of your own mental health:

- Reach out to close friends or loved ones.

- Contact a minister, spiritual leader or someone in your faith community (if applicable).

- Check out some of the resources on the next page which might be helpful for you.

Mental Health Resources

Organizations that you might find helpful include:

- Canadian Mental Health Association (CMHA)
- Anxiety Canada
- National Alliance on Mental Illness (NAMI)
- Anxiety & Depression Association of America (ADAA)
- The International 22q11.2 Foundation
- Rare Disease Foundation

Meditation or mindfulness websites/apps such as:

- Headspace
- Calm
- Stop, Breath & Think

Kid-friendly books and resources include:

- Mighty Moe: An Anxiety Workbook for Children
- Taming Worry Dragons
- What To Do When You Worry Too Much

Thank You

This resource was created by Stephanie Chieffo, Emily Morris, and Caitlin Slomp, with input from parents of individuals with 22qDS, and designed by Vanessa Macdonald.

The original jar model images are courtesy of Jehannine Austin, Catriona Hippman, and Claudia Li, creators of the booklet called "Mental Illness: Underlying Causes and Approaches to Recovery".

This resource would not have been possible without the generous contributions of the 22qDS families and the funding provided by: **The Rare Disease Foundation** and **The National Society of Genetic Counselors: Education Special Interest Group (SIG).**

Figure C.1 Educational Resource Booklet for 22q11.2DS psychiatric symptoms.

APPENDIX D

PEARSON'S CORRELATION DATA

The raw data for the Pearson's correlations are included below.

Table D.1 Mini-IPIP versus All T1 Variables

		Extraversion	Agreeableness	Conscientiousness	Neuroticism	Openness to Experience
Extraversion	Pearson Correlation	1	.570	.246	-.133	-.381
	Sig. (2-tailed)		.086	.493	.714	.278
	N	10	10	10	10	10
Agreeableness	Pearson Correlation	.570	1	.336	.068	-.140
	Sig. (2-tailed)	.086		.343	.851	.700
	N	10	10	10	10	10
Conscientiousness	Pearson Correlation	.246	.336	1	-.475	-.141
	Sig. (2-tailed)	.493	.343		.166	.697
	N	10	10	10	10	10
Neuroticism	Pearson Correlation	-.133	.068	-.475	1	.681*
	Sig. (2-tailed)	.714	.851	.166		.030
	N	10	10	10	10	10

Openness to Experience	Pearson Correlation	-.381	-.140	-.141	.681*	1
	Sig. (2-tailed)	.278	.700	.697	.030	
	N	10	10	10	10	10
IUS TOTAL 1	Pearson Correlation	.277	.145	.014	.562	.219
	Sig. (2-tailed)	.439	.689	.970	.091	.543
	N	10	10	10	10	10
PF-SOC: Reflective	Pearson Correlation	.129	-.068	.653*	-.651*	-.279
	Sig. (2-tailed)	.723	.852	.041	.041	.434
	N	10	10	10	10	10
PF-SOC: Suppressive	Pearson Correlation	-.132	.079	-.586	.677*	.290
	Sig. (2-tailed)	.717	.828	.075	.031	.416
	N	10	10	10	10	10
PF-SOC: Reactive	Pearson Correlation	-.336	-.038	-.601	.856**	.494
	Sig. (2-tailed)	.343	.917	.066	.002	.147
	N	10	10	10	10	10
imputed_total_gcos_t1	Pearson Correlation	-.336	.413	.313	-.329	-.289
	Sig. (2-tailed)	.343	.235	.379	.353	.418
	N	10	10	10	10	10
Stigma Total Preview	Pearson Correlation	.189	-.866	-.655	.000	-.569
	Sig. (2-tailed)	.879	.333	.546	1.000	.614
	N	3	3	3	3	3

Worry Total Preview	Pearson Correlation	-.164	-.115	.527	.002	-.001
	Sig. (2-tailed)	.651	.751	.117	.996	.999
	N	10	10	10	10	10

*. Correlation is significant at the 0.05 level (2-tailed).
**. Correlation is significant at the 0.01 level (2-tailed).

Table D.2 T1 IUS, PF-SOC, and GCOS versus All T1 Variables

		IUS TOTAL 1	PF-SOC: Reflective	PF-SOC: Suppressive	PF-SOC: Reactive	imputed_total_gcos_t1
Extraversion	Pearson Correlation	.277	.129	-.132	-.336	-.336
	Sig. (2-tailed)	.439	.723	.717	.343	.343
	N	10	10	10	10	10
Agreeableness	Pearson Correlation	.145	-.068	.079	-.038	.413
	Sig. (2-tailed)	.689	.852	.828	.917	.235
	N	10	10	10	10	10
Conscientiousness	Pearson Correlation	.014	.653*	-.586	-.601	.313
	Sig. (2-tailed)	.970	.041	.075	.066	.379
	N	10	10	10	10	10
Neuroticism	Pearson Correlation	.562	-.651*	.677*	.856**	-.329
	Sig. (2-tailed)	.091	.041	.031	.002	.353
	N	10	10	10	10	10
Openness to	Pearson Correlation	.219	-.279	.290	.494	-.289

Experience	Sig. (2-tailed)	.543	.434	.416	.147	.418
	N	10	10	10	10	10
IUS TOTAL 1	Pearson Correlation	1	-.470	.476	.551	-.412
	Sig. (2-tailed)		.171	.164	.099	.236
	N	10	10	10	10	10
PF-SOC: Reflective	Pearson Correlation	-.470	1	-.650*	-.788**	.161
	Sig. (2-tailed)	.171		.042	.007	.656
	N	10	10	10	10	10
PF-SOC: Suppressive	Pearson Correlation	.476	-.650*	1	.904**	-.096
	Sig. (2-tailed)	.164	.042		<.001	.792
	N	10	10	10	10	10
PF-SOC: Reactive	Pearson Correlation	.551	-.788**	.904**	1	-.123
	Sig. (2-tailed)	.099	.007	<.001		.736
	N	10	10	10	10	10
imputed_total_gcos_t1	Pearson Correlation	-.412	.161	-.096	-.123	1
	Sig. (2-tailed)	.236	.656	.792	.736	
	N	10	10	10	10	73
Stigma Total Preview	Pearson Correlation	-.240	.056	.228	.108	-.417*
	Sig. (2-tailed)	.846	.964	.854	.931	.027
	N	3	3	3	3	28
	Pearson Correlation	.408	.168	-.026	.056	-.231

Worry Total Preview	Sig. (2-tailed)	.242	.642	.943	.877	.053
	N	10	10	10	10	71

*. Correlation is significant at the 0.05 level (2-tailed).
**. Correlation is significant at the 0.01 level (2-tailed).

Table D.3 T1 SSRMI and Worry Scale versus All T1 Variables

		Stigma Total Preview	Worry Total Preview
Extraversion	Pearson Correlation	.189	-.164
	Sig. (2-tailed)	.879	.651
	N	3	10
Agreeableness	Pearson Correlation	-.866	-.115
	Sig. (2-tailed)	.333	.751
	N	3	10
Conscientiousness	Pearson Correlation	-.655	.527
	Sig. (2-tailed)	.546	.117
	N	3	10
Neuroticism	Pearson Correlation	.000	.002
	Sig. (2-tailed)	1.000	.996
	N	3	10
Openness to Experience	Pearson Correlation	-.569	-.001
	Sig. (2-tailed)	.614	.999
	N	3	10
IUS TOTAL 1	Pearson Correlation	-.240	.408
	Sig. (2-tailed)	.846	.242
	N	3	10
	Pearson Correlation	.056	.168

PF-SOC: Reflective	Sig. (2-tailed)	.964	.642
	N	3	10
PF-SOC: Suppressive	Pearson Correlation	.228	-.026
	Sig. (2-tailed)	.854	.943
	N	3	10
PF-SOC: Reactive	Pearson Correlation	.108	.056
	Sig. (2-tailed)	.931	.877
	N	3	10
imputed_total_gcos_t1	Pearson Correlation	-.417*	-.231
	Sig. (2-tailed)	.027	.053
	N	28	71
Stigma Total Preview	Pearson Correlation	1	.157
	Sig. (2-tailed)		.445
	N	28	26
Worry Total Preview	Pearson Correlation	.157	1
	Sig. (2-tailed)	.445	
	N	26	71

*. Correlation is significant at the 0.05 level (2-tailed).
**. Correlation is significant at the 0.01 level (2-tailed).

Table D.4 PF-SOC versus All T2 Variables

		Extraversion	Agreeableness	Conscientiousness	Neuroticism	Openness to Experience
Extraversion	Pearson Correlation	1	.570	.246	-.133	-.381
	Sig. (2-tailed)		.086	.493	.714	.278

	N	10	10	10	10	10
Agreeableness	Pearson Correlation	.570	1	.336	.068	-.140
	Sig. (2-tailed)	.086		.343	.851	.700
	N	10	10	10	10	10
Conscientiousness	Pearson Correlation	.246	.336	1	-.475	-.141
	Sig. (2-tailed)	.493	.343		.166	.697
	N	10	10	10	10	10
Neuroticism	Pearson Correlation	-.133	.068	-.475	1	.681*
	Sig. (2-tailed)	.714	.851	.166		.030
	N	10	10	10	10	10
Openness to Experience	Pearson Correlation	-.381	-.140	-.141	.681*	1
	Sig. (2-tailed)	.278	.700	.697	.030	
	N	10	10	10	10	10
GCOS Time 2	Pearson Correlation	-.123	.072	.083	-.384	.032
	Sig. (2-tailed)	.735	.843	.821	.273	.929
	N	10	10	10	10	10
Worry Total Time 2	Pearson Correlation	.001	-.171	-.016	.354	.144
	Sig. (2-tailed)	.999	.636	.966	.315	.691

	N	10	10	10	10	10
IUS Time 2	Pearson Correlation	.304	.047	.217	.541	.321
	Sig. (2-tailed)	.427	.904	.574	.133	.400
	N	9	9	9	9	9
Stigma Time 2	Pearson Correlation	.982	-.500	.756	-1.000**	-.822
	Sig. (2-tailed)	.121	.667	.454	.000	.386
	N	3	3	3	3	3

*. Correlation is significant at the 0.05 level (2-tailed).
**. Correlation is significant at the 0.01 level (2-tailed).

Table D.5 All T2 Variables versus Mini-IPIP and All T2 Variables

		GCOS Time 2	Worry Total Time 2	IUS Time 2	Stigma Time 2
Extraversion	Pearson Correlation	-.123	.001	.304	.982
	Sig. (2-tailed)	.735	.999	.427	.121
	N	10	10	9	3
Agreeableness	Pearson Correlation	.072	-.171	.047	-.500
	Sig. (2-tailed)	.843	.636	.904	.667
	N	10	10	9	3
Conscientiousness	Pearson Correlation	.083	-.016	.217	.756
	Sig. (2-tailed)	.821	.966	.574	.454
	N	10	10	9	3

Neuroticism	Pearson Correlation	-.384	.354	.541	-1.000**
	Sig. (2-tailed)	.273	.315	.133	.000
	N	10	10	9	3
Openness to Experience	Pearson Correlation	.032	.144	.321	-.822
	Sig. (2-tailed)	.929	.691	.400	.386
	N	10	10	9	3
GCOS Time 2	Pearson Correlation	1	-.308*	-.440	-.488*
	Sig. (2-tailed)		.035	.236	.040
	N	47	47	9	18
Worry Total Time 2	Pearson Correlation	-.308*	1	.512	-.031
	Sig. (2-tailed)	.035		.158	.904
	N	47	47	9	18
IUS Time 2	Pearson Correlation	-.440	.512	1	-1.000**
	Sig. (2-tailed)	.236	.158		.000
	N	9	9	9	3
Stigma Time 2	Pearson Correlation	-.488*	-.031	-1.000**	1
	Sig. (2-tailed)	.040	.904	.000	
	N	18	18	3	18

*. Correlation is significant at the 0.05 level (2-tailed).
**. Correlation is significant at the 0.01 level (2-tailed).

APPENDIX E

STUDY FLYER

A flyer was distributed by social media for recruitment.

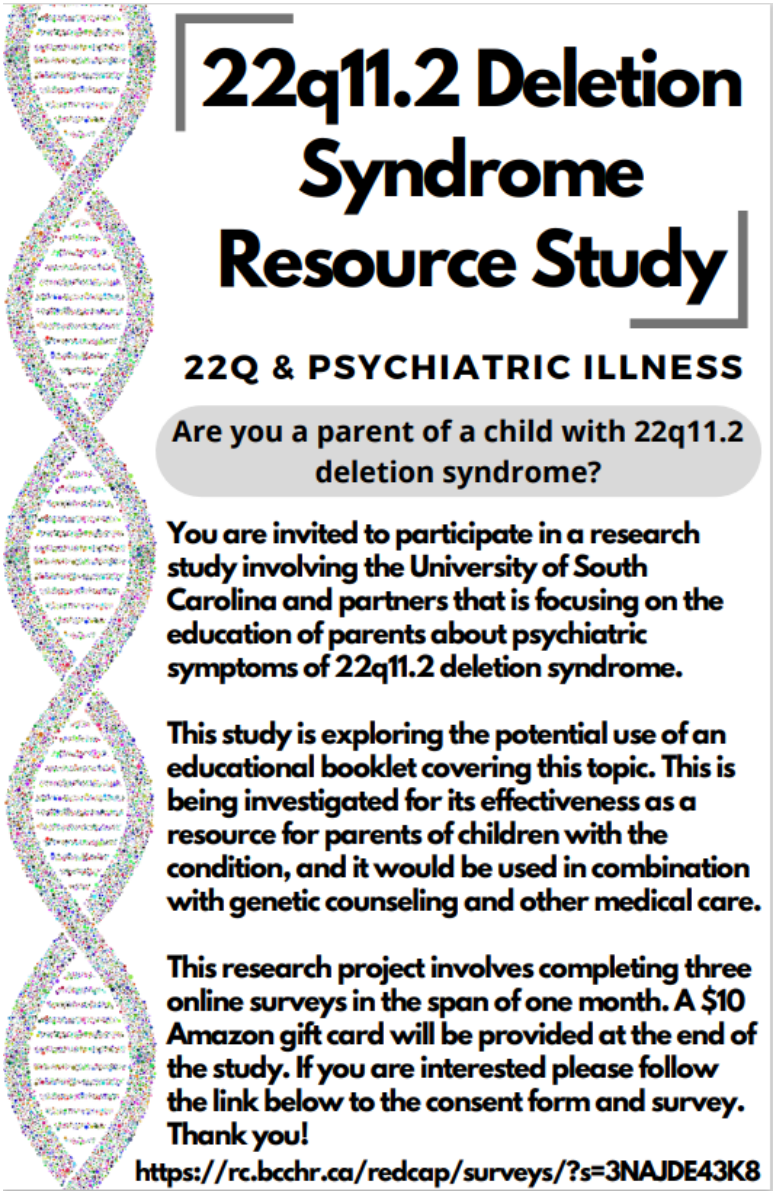


Figure E.1 Study Flyer.

APPENDIX F

PARTICIPANT LETTER

The following letter was sent to potential participants through the Greenwood Genetic Center.

University of South Carolina School of Medicine
Genetic Counseling Program

Dear Potential Participant,

You are invited to participate in a graduate research study focusing on the delivery of information regarding the psychiatric symptoms of 22q11.2 deletion syndrome to parents of children with the condition. My name is Kayla Blankenship, and I am a graduate student in the genetic counseling program at the University of South Carolina School of Medicine. My research investigates the potential benefit of an educational booklet detailing information about the psychiatric symptoms of 22q11.2 deletion syndrome. This booklet would be a resource for parents of children with the condition to use in combination with genetic counseling. The research involves completing three online surveys in the span of one month.

The surveys will include questions to help us better understand the background of you and your child such as age, ethnicity, and 22qDS diagnosis status as well as other questions such as, "This booklet answered questions I had regarding mental illness associated with 22q11.2DS" and "The booklet helped me to feel more confident in my ability to recognize early warning signs of mental illness." If you do not wish to answer a certain question, please skip that question and continue with the rest of the survey.

All responses gathered from the surveys will be kept anonymous and confidential. We do ask for your email for the purpose of sending both the later surveys and to supply a $10.00 amazon gift card after the completion of the study. The results of this study might be published or presented at academic meetings; however, participants will not be identified.

Your participation in this research is voluntary. At any time, you may withdraw from the study by not completing the survey. If you choose to continue with the study, you may follow the link in parentheses to begin the survey process (https://rc.bcchr.ca/redcap/surveys/?s=3NAJDE43K8). There will be a more detailed consent form provided before the first survey.

Thank you for your time and consideration to participate in this survey. Your responses may help inform possible use of this educational resource for parents raising a child with 22q11.2 deletion syndrome. If you have any questions regarding the research, you may contact either myself or my faculty advisor, Crystal Hill-Chapman, MS, CGC, using the contact information below. If you have any questions about your rights as a research participant, you may contact the Office of Research Compliance at the University of South Carolina at (803) 777-7095.

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Figure F.1 Potential Participant Letter.