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Evaluating the “Family-Centered” Approach of Pediatric Multidisciplinary Down Syndrome Clinics: A Parents’ Perspective

by

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Wake Forest University, 2013

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Dedication

For my parents – for their endless supply of love, support, encouragement, and inspiration.

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Abstract

Down syndrome is the most common genetic cause of intellectual disability. Individuals with Down syndrome usually display mild to moderate intellectual disability, developmental delay, characteristic facial features, and an increased risk for birth defects and various medical problems. Multidisciplinary clinics were established to address the multi-system health concerns for Down syndrome, increase adherence to medical management guidelines, and provide coordinated and comprehensive care for the patient. Research has examined the beneficial effect of a multidisciplinary approach to patient care and medical outcomes; however, no studies have been done which evaluate the psychosocial aspect of the care provided.

We hypothesized that families whose children attend a multidisciplinary clinic will report a higher level of psychosocial support provided by their healthcare team than those families who receive care from independent practitioners. An online survey was distributed through local Down syndrome support groups and national Down syndrome organizations with the intention of identifying trends involving psychosocial support among families of children with Down syndrome.

Responses from 415 parents were used for statistical analysis. Results consistently indicated that significantly higher levels of psychosocial support and care are provided by multidisciplinary Down syndrome clinics. The survey also collected parents' experiences with and opinions of pediatric genetic counseling. This qualitative data was analyzed using grounded theory methods and identified six major themes related

to the specialty. Four themes related to genetic counseling outcomes identified the experience as: (1) specialized, expert information, (2) opportunity for discussion, (3) comprehensive medical care, and (4) generally unhelpful. Two additional themes related to participants' lack of direct experience with genetic counseling were also identified as (5) negative perceptions and experiences and (6) general misunderstanding or lack of knowledge of the profession.

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Chapter 1. Background

1.1 Down Syndrome

Down syndrome is the most common genetic cause of intellectual disability. About one in every 830 children is born with Down syndrome. It is estimated that about 250,000 people have Down syndrome in the United States (Presson et al., 2013). There is a high degree of variability in the phenotype across individuals with Down syndrome. Down syndrome can usually be diagnosed at birth or shortly thereafter. Clinical diagnoses in the newborn period are usually due to a combination of recognizable facial features and hypotonia, decreased muscle tone. Facial features include flat nasal bridge, low set ears, epicanthal folds, and upslanting palpebral fissures. The eyes can have Brushfield spots around the margin of the iris. Signs of hypotonia can be evident through features, such as a protruding tongue, as well as tests that examine muscle tone. Individuals with Down syndrome usually have short stature and have brachycephaly with a flat occiput. The hands are short and broad, with characteristic features such as a single transverse palmar crease and fifth finger clinodactyly. The feet display a "sandal gap," which is a wide space between the first and second toes. Individuals with Down syndrome also show mild to moderate intellectual disability and developmental delay. The IQ range is around 35 to 70 when individuals are old enough to be tested. Developmental delay is usually apparent by the end of the first year of life (Nussbaum, McInnes, & Willard, 2007).

Down syndrome also puts individuals at risk for medical complications throughout life. Congenital heart disease is present in about 50% of babies with Down syndrome. Common heart defects include atrioventricular canal defect, ventricular septal defect, and atrial septal defect. Certain malformations, such as duodenal atresia, are much more common in Down syndrome than in other conditions or in the general population (Torfs & Christianson, 1998). Table 1.1 lists congenital malformations frequently observed in Down syndrome and their relative risk for Down syndrome as described by Torfs & Christianson's study (1998).

Table 1.1 Malformations Frequently Observed in Down Syndrome

Malformation	**Relative risk**
Atrioventricular canal	1,009
Annular pancreas	430
Duodenal atresia	265
Patent ductus arteriosus	152
Small intestine	142
Ventricular septal defect	95
Tricuspid valve defect	84
Hypoplastic aorta	77
Tetralogy of Fallot	77
Atrial septal defect	71
Ectopic anus	67
Cataract	54
Intestinal malrotation	45
Anal atresia/stenosis	34
Tracheo-esophageal fistula	26
Syndactyly	26

Other medical problems become a concern after birth. There is significant risk for hearing loss and recurrent middle ear infections. Eye disease, including cataracts and refractive errors, also affect over half of the Down syndrome population. Obstructive sleep apnea is also a concern for individuals with Down syndrome. Neurological dysfunction, hip dislocation, thyroid disease, celiac disease, transient myeloproliferative disorder and leukemias, and Hirschsprung disease are also medical concerns, but occur less frequently. Table 1.2 shows the most common medical problems associated with Down syndrome and the percentage of individuals who are affected by each (Torfs & Christianson, 1998).

Table 1.2 Medical Problems Common in Down Syndrome

Condition	%
Hearing problems	75
Vision problems	60
Cataracts	15
Refractive errors	50
Obstructive Sleep Apnea	50-75
Otitis media	50-70
Congenital heart disease	40-50
Hypodontia and delayed dental eruption	23
Gastrointestinal atresia	12
Thyroid disease	4-18
Seizures	1-13
Hematological problems	
Anemia	3
Iron deficiency	10
Transient myeloproliferative disorder	10
Leukemia	1
Celiac disease	5
Atlanto-axial instability	1-2
Autism	1
Hirschsprung disease	<1

Due to the need for comprehensive medical management for individuals with Down syndrome, the American Academy of Pediatrics (AAP) published guidelines for the health supervision for children and young adults with Down syndrome (Bull, 2011). The guidelines outline specific physical examinations and laboratory studies that should be completed to manage and monitor health care, but also list important information and anticipatory guidance that should be reviewed with patients and their families at each visit (Bull, 2011). In addition to health care management, individuals with Down syndrome benefit from early intervention programs and other types of therapies. These therapies can include occupational therapy, speech therapy, and physical therapy.

The Down syndrome phenotype – the characteristic facial features, intellectual and developmental disabilities, and medical issues – is caused by an extra copy of genetic material that is located on chromosome 21. The region of the chromosome that contains those genes responsible for the Down syndrome phenotype was studied and identified in 2006. This "Down syndrome critical region" (DSCR), which is located at the 21q22 locus, was found to have the predominant influence over the manifestation of phenotypic features.

There are several different cytogenetic types of Down syndrome. In other words, there are different manners in which there can be a third copy of genetic material from chromosome 21. About 95% of Down syndrome is caused by trisomy 21, an extra copy of the entire 21st chromosome in all cells. This chromosome condition is usually caused by a sporadic, random error that occurs during the process of meiosis. Nondisjunction, the failure of chromosomes to divide equally during meiotic division, results in a gamete with an incorrect number of chromosomes (also known as aneuploidy). In regard to trisomy 21, about 90% of occurrences are due to a maternal meiotic error. The vast majority of these errors happen during meiosis I, with fewer mistakes occurring during meiosis II. Although nondisjunction is a sporadic event, it occurs more frequently as women age. Therefore, there has been an association between an increased incidence of trisomy 21 with advanced maternal age (Gardner, Sutherland, & Shaffer, 2012). This phenomenon has been attributed to that fact that oocytes remain arrested in meiosis from birth until ovulation and therefore this prolonged meiotic time frame leaves more room for error (Nussbaum et al., 2007). Among the small fraction of trisomy 21 caused by paternal meiotic errors, there are an equal number of errors in meiosis I as compared to meiosis II. Because nondisjunction is predominantly a sporadic event, the recurrences are rare. However, a small increased recurrence risk is given to account for the possibility of gonadal mosaicism, parental predisposition to nondisjunction, and chance (Gardner et al., 2012).

About four percent of patients with Down syndrome have 46 chromosomes. One of these chromosomes, however, is a Robertsonian translocation between chromosome 21q and the long arm of another acrocentric chromosome (usually 14 or 22). The translocation chromosome replaces one of the normal acrocentric chromosomes. In effect, patients with a Robertsonian translocation are trisomic for genes on 21q. Unlike trisomy 21, translocation Down syndrome shows no relation to maternal age. Robertsonian translocations, however, can occur sporadically or be inherited from either parent. If the translocation is familial, one of the parents must be a carrier for a balanced translocation. These carrier individuals have only 45 chromosomes – with one copy of each involved chromosomes being replaced by the single translocation chromosome. Consequently, with familial translocation Down syndrome, there is a relatively high recurrence risk especially when the mother is the balanced translocation carrier. For this reason, karyotyping of the parents is essential before determining recurrence risks for the family if a translocation is present (Nussbaum et al., 2007).

About 1-2% of patients with Down syndrome are mosaic. Mosaicism occurs when chromosomes segregate unevenly during mitotic cell division post-zygotically, or after conception. This mosaicism can be the result of a trisomy 21 fetus losing the extra copy of chromosome 21 during cell division (“trisomy rescue”) or a chromosomally typical fetus incurring nondisjunction during mitosis (Gardner et al., 2012).

Although the clinical diagnosis of Down syndrome is usually not difficult, it is crucial that genetic testing be completed in order to confirm the diagnosis and provide a basis for genetic counseling about recurrence risk and etiology of the specific type of Down syndrome (Nussbaum et al., 2007).

1.2 Multidisciplinary Care

Multidisciplinary care involves bringing different specialties together into an open dialogue to discuss patient care and facilitate decision-making. The primary goal of multidisciplinary care is to improve the health care management of the individual patient. This care model is based on the assumption that one multidisciplinary discussion with all involved specialties is more effective, with the joint decision more accurate, than the sum of all individual opinions. Another benefit of multidisciplinary care is the improved communication between different specialties. Cooperation and collaboration is expected to be greater when each discipline understands the roles, possibilities, and limitations of the others. This increased understanding allows a trusting relationship to be developed between the different specialties (Ruhstaller, Roe, Thürlimann, & Nicoll, 2006). Additionally, patients are treated according to the same guidelines and standards of care. A multidisciplinary approach acknowledges the complexities of modern health care and the important role of communication between health care providers in delivering comprehensive care to patients (Kim, Barnato, Angus, Fleisher, & Kahn, 2010).

The field of oncology has been utilizing the multidisciplinary care approach to facilitate the best patient-specific treatment and management plans. These multidisciplinary meetings involve medical oncologists, surgical oncologists, radiation oncologists, pathologists, diagnostic radiologists, plastic surgeons, and genetic counselors. The full patient history is reviewed and discussed. Treatment plans are maintained or modified depending on the evaluation by all specialists involved. In one retrospective study, the outcomes of patients discussed in a multidisciplinary breast cancer center setting were analyzed (Chang et al., 2001). Treatment recommendations made before the team discussion were compared with the consensus recommendation made by the multidisciplinary team. For the 75 patients reviewed, the multidisciplinary team disagreed with the initial treatment recommendations in 43% of the cases (32 patients). Treatment plans were changed for a variety of reasons: breast-conservation was recommended over mastectomy, further work-up was recommended before final decisions were made, treatment recommendations were altered after pathology changed the diagnosis, and the addition of adjunct treatment such as radiation and hormone therapies (Chang et al., 2001).

Not only are major decisions being changed through a multidisciplinary approach, but patient outcomes are also improved. Many studies have looked at differences in patient history and treatment that have led to discrepancies in patient survival. Differences in survival outcome for patients with ovarian cancer in Scotland were explored in the 1990s. This retrospective study reviewed 533 cases and teased out confounding factors such as age, stage, pathology, degree of differentiation, and comorbidities. The study found that patient survival was significantly improved when patients were referred to a multidisciplinary clinic. About 35% of patients seen at a multidisciplinary clinic reported 5-year survival. This was significantly higher than the 19% of patients with 5-year survival who were not seen at a multidisciplinary clinic. By comparing the data, this study reported a statistically significant ($p < 0.001$) relative hazard ratio of 0.60 between the two groups (Junor, Hole, & Gillis, 1994).

Many other studies have been conducted that explore the impact of multidisciplinary care outside of the field of oncology. The geriatric specialty has found that multidisciplinary care significantly increases survival and improves the health outcomes of elderly patients after hospital discharge (Rubenstein et al., 1984; Caplan, Williams, Daly, & Abraham, 2004). Internal medicine has also found that a multidisciplinary approach offers better treatments, follow up, and outcomes in patients with hypertension and decreases mortality rates of Intensive Care Unit patients (Adorian, Silverberg, Tomer, & Wamosher, 1990; Kim et al., 2010). Cardiologists have found that multidisciplinary care improves the outcome and survival for patients who have suffered heart failure (McDonald et al., 2002). Studies extending across a variety of specialties are coming to the same conclusion – multidisciplinary care improves overall patient care and related outcomes.

1.3 Multidisciplinary Down Syndrome Clinics

Down syndrome is a complex health condition to manage. As the medical complications involve multiple body systems, the pediatric care of a child with Down syndrome requires the involvement of many specialties. Peyton Manning Children's Hospital acknowledges that:

> It is clear that individuals with Down syndrome require a multidisciplinary approach, although families often find themselves running from place to place visiting multiple specialists without the specialists being aware of what each other are doing. The number of physician appointments to obtain the required monitoring that individuals with Down syndrome need can be very overwhelming.

In an attempt to facilitate communication between specialists, provide the best possible care for individuals with Down syndrome, and lessen the overwhelming number and nature of specialist appointments for parents, the concept of multidisciplinary Down syndrome specialty clinics was born. Multidisciplinary clinics provide an environment in which patients can meet with multiple specialists in one visit, in order to achieve comprehensive coordination of care.

Multidisciplinary Down syndrome clinics have been introduced across the country to ensure that individuals with Down syndrome can access coordinated and comprehensive health care (Skotko, Davidson, & Weintraub, 2013). There are approximately 60 Down syndrome specialty clinics across 32 states (National Down Syndrome Society, 2012). These specialized clinics make certain that patients receive care from a team of medical professionals and consultants familiar with Down syndrome. Clinics also ensure that their patients remain up-to-date on the screenings and management outlined by the American Academy of Pediatrics (AAP) published guidelines for the health supervision for children and young adults with Down syndrome.

Therefore, Down syndrome specialty clinics are able to offer patients and their families both expertise and support (Skotko et al., 2013). Each clinic, however, is uniquely designed by their medical center and can include their own chosen subset of specialists. Most clinics include health care providers and consultants encompassing the specialties of genetics and development. Other specialties that are often part of the multidisciplinary team include ophthalmology, audiology, otolaryngology, and cardiology. Some clinics also offer a variety of services such as speech, occupational, and physical therapies.

Much like other multidisciplinary care teams, Down syndrome clinics have been interested in exploring the benefits of a multidisciplinary approach. One particular study evaluated the actual medical care provided (Skotko et al., 2013). This study involved a retrospective chart review of 105 new patients with Down syndrome (ages three and over) that were seen within one year of the opening of the clinic. These patients did not previously have access to a Down syndrome clinic and had been followed by independent specialists. Overall, the study found that only 10% of patients were up-to-date on all of the screenings recommended by the AAP and the Down Syndrome Medical Interest Group (DSMIG) guidelines upon registering with the clinic. Only 17% of patients were up-to-date on the AAP guidelines alone. At the patient's clinic visit, each patient received screening tests and information according to both the AAP and DSMIG guidelines. They were also referred to external specialists for concerns specific to the patient such as speech therapy, clinical psychology, and sleep studies. The clinic visits were also able to identify new co-occurring conditions that had previously gone undiagnosed. These new diagnoses included gastrointestinal problems, dermatological conditions, behavioral diagnoses, and expressive language disorders. As a result of multidisciplinary clinic attendance, 83% of patients were brought up-to-date on screenings based on published guidelines, 41% were referred to outside specialists for additional medical care, and 54% were diagnosed with new co-occurring medical problems (Skotko et al., 2013). The results of this study demonstrate the added value and benefits of multidisciplinary Down syndrome clinics to the medical care and medical management of individuals with Down syndrome.

Multidisciplinary Down syndrome clinics also offer added value in other areas in addition to improved medical management. Most of the published goals of the multidisciplinary clinics reflect a holistic approach to health care, stating goals that go above and beyond providing complete and quality medical care. Themes reflecting a "family-centered" approach are apparent in mission statements and goals established for each clinic (Children's Hospital of Wisconsin, 2014). The Down syndrome clinic at the Waisman Center at the University of Wisconsin-Madison, specifically, list their clinic goals as:

> - Educate the family about Down syndrome, associated complications, and the genetic cause.
> - Detect and treat problems related to Down syndrome.
> - Provide health care counseling about potential risks and preventive behaviors.
> - Refer and collaborate with other health professionals to meet [the] child's special needs.
> - Provide information on community resources and services to support [the] child and family.
> - Support and advocate for the patient and family.

Down syndrome specialty clinics are equipped to provide information and updates on community-based support resources. These resources can include conferences, books, and support groups/organizations (Skotko et al., 2013). In addition to referrals to outside support resources, Down syndrome clinics can also provide psychosocial support to patients and their families during clinic appointments. While a "family-centered" or supportive approach is a goal of many multidisciplinary clinics, there have been no formal studies reported to-date that evaluate the efficacy of this approach.

1.4 Psychosocial Support and Genetic Counseling

According to Mosby's Medical Dictionary, the term "psychosocial" means "pertaining to a combination of psychological and social factors" (2012). Therefore, psychosocial support provides guidance and assistance in areas related to social interactions and emotional well-being.

Genetic counselors are healthcare consultants who are part of the genetics team that specialize in providing psychosocial support and genetics education and interpretation to families affected by genetic conditions. In 2006, the National Society of Genetic Counselors (NSGC) published the following updated definition of genetic counseling:

> Genetic counseling is the process of helping people understand and adapt to the medical, psychological, and familial implication of genetic contributions to disease. This process integrates the following:
> - Interpretation of family and medical histories to assess the chance of disease occurrence or recurrence
> - Education about inheritance, testing, management, prevention, resources, and research
> - Counseling to promote informed choices and adaptation to the risk or condition
>
> (National Society of Genetic Counselors' Definition Task Force, 2006).

In order to accomplish the defined objectives for genetic counseling, genetic counselors receive specific training involving psychosocial assessment and the provision of psychosocial support, along with their genetic education.

Many studies have evaluated the outcomes and areas of patient satisfaction regarding genetic counseling across many settings, including cancer and cardiology (Bjorvatn et al., 2007; Christiaans et al., 2009; Clark et al., 2000; DeMarco, Peshkin, Mars, & Tercyak, 2004). The most commonly reported patient perspectives fit five common themes: provision of information/acquisition of knowledge, psychosocial support (immediate and long-term), anticipatory guidance, facilitation of family communication, and aid in decision-making (Bernhardt, Biesecker, & Mastromarino, 2000). These results traverse all settings of genetic counseling where patient satisfaction has been studied. Based on these results, genetic counselors are a provider of psychosocial support for patients and families affected by genetic conditions.

1.5 Genetic Counseling in the Pediatric Setting

Although psychosocial outcomes of genetic counseling in the pediatric setting have yet to be assessed, other benefits resulting from the inclusion of genetic counselors in a pediatric health care team have been evaluated. One study found that adherence to recommended medical management protocols was significantly increased in the pediatric patient population that was seen by a genetic counselor in combination with a geneticist. In a retrospective chart review, the Cincinnati Children's Hospital Medical Center found that all categories of medical management recommendations made during the appointment were more strictly followed by the patient population seen by pediatric genetic counselors in comparison to those who did not (Rutherford, Zhang, Atzinger, Ruschman, & Myers, 2014). Both patient satisfaction and medical adherence studies identify avenues in which the inclusion of genetic counselors into the pediatric health care team can improve patient care and benefit their families.

1.6 Genetic Counseling in Multidisciplinary Down Syndrome Clinics

Based on reported patient satisfaction in the previously mentioned studies, genetic counselors are an important member of the health care team as they provide psychosocial support for patients and families affected by genetic conditions, such as Down syndrome. Therefore, the addition of genetic counselors to the pediatric health care team should contribute to the “family-centered” approach of multidisciplinary clinics. According to the Global Down Syndrome Foundation’s medical care centers database, 29 out of 45 (62%) registered pediatric Down syndrome specialty clinics across the United States report including genetic counselors as part of their multidisciplinary team approach. Nine clinics (19%) do not have their specialties listed on the database website. Nine clinics (19%) report not including genetic counselors in their specialty teams (Global Down Syndrome Foundation). According to this database, the majority of Down syndrome multidisciplinary clinics utilize genetic counseling as part of their practice. However, the function and use of genetic counselors is not specified for each clinic. Some clinics offer genetic counseling as a one-time consultation for new patients, whereas others utilize genetic counselors for other non-clinical roles such as clinic coordination.

Some clinics may also include other professionals in their multidisciplinary team in order to provide psychosocial support for patients and their families. Other health care providers, such as psychologists, psychiatrist, or neuropsychiatrists, may be included in the health care team for psychosocial purposes. Additionally, social workers or patient advocates may play a role in psychosocial management.

Chapter 2. Manuscript

Evaluating the “Family-Centered” Approach of Pediatric Multidisciplinary Down Syndrome Clinics: A Parents’ Perspective[1]

2.1 Abstract

Down syndrome is the most common genetic cause of intellectual disability. Individuals with Down syndrome usually display mild to moderate intellectual disability, developmental delay, characteristic facial features, and an increased risk for birth defects and various medical problems. Multidisciplinary clinics were established to address the multi-system health concerns for Down syndrome, increase adherence to medical management guidelines, and provide coordinated and comprehensive care for the patient. Research has examined the beneficial effect of a multidisciplinary approach to patient care and medical outcomes; however, no studies have been done which evaluate the psychosocial aspect of the care provided. We hypothesized that families whose children attend a multidisciplinary clinic will report a higher level of psychosocial support provided by their healthcare team than those families who receive care from independent practitioners. An online survey was distributed through local Down syndrome support groups and national Down syndrome organizations with the intention of identifying trends involving psychosocial support among families of children with Down syndrome. Responses from 415 parents were used for statistical analysis. Results consistently indicated that significantly higher levels of psychosocial support and care are provided by

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multidisciplinary Down syndrome clinics. The survey also collected parents' experiences with and opinions of pediatric genetic counseling. These qualitative data were analyzed using grounded theory methods and identified six major themes related to the specialty. Four themes related to genetic counseling outcomes identified the experience as: (1) specialized, expert information, (2) opportunity for discussion, (3) comprehensive medical care, and (4) generally unhelpful. Two additional themes related to participant's lack of direct experience with genetic counseling were also identified as (5) negative perceptions and experiences and (6) general misunderstanding or lack of knowledge of the profession.

2.2 Introduction

Down syndrome is the most common genetic cause of intellectual disability, affecting one in every 700 – 800 live born children. Down syndrome is a chromosomal condition that presents when individuals are born with an extra copy of the genetic material on chromosome 21. Down syndrome is associated with mild to moderate intellectual disability, developmental delay, characteristic facial features, and hypotonia. Children with Down syndrome may also be born with birth defects, such as congenital heart disease or gastrointestinal anomalies. Individuals with Down syndrome also have an increased risk of developing a wide variety of medical complications throughout life. These can include, but are not limited to, hearing loss, vision difficulties, obstructive sleep apnea, thyroid disease, celiac disease, and early-onset Alzheimer disease (Korenberg et al., 1994).

The published guidelines for the medical care of individuals with Down syndrome, established most recently by the American Academy of Pediatrics (AAP), have grown in length and complexity in recent years (Bull, 2011). As the medical complications involve multiple body systems, the pediatric care of a child with Down syndrome requires the attention of many specialties. Until the advent of multidisciplinary clinics, the pediatric medical care of children was disjointed with "families… running from place to place visiting multiple specialists without the specialists being aware of what each other are doing" (Peyton Manning Children's Hospital). A multidisciplinary approach acknowledges the complexities of modern health care and the important role of communication between health care providers in delivering comprehensive care to patients (Kim et al., 2010). Multidisciplinary Down syndrome clinics provide an environment in which patients can meet with multiple specialists in one visit, in order to achieve coordination of care. These clinics ensure that patients are receiving the best standard of care by medical professionals and consultants with expertise in Down syndrome. In the United States, there are approximately 60 Down syndrome specialty clinics across 32 states (National Down Syndrome Society, 2012). Each clinic includes their own subset of specialists and can offer a variety of services such as speech, occupational, and physical therapies. Most clinics include health care providers and consultants encompassing the specialties of genetics and development.

Studies have been conducted that evaluate the efficacy and benefit of utilizing a multidisciplinary approach across a wide range of specialties including oncology, internal medicine, and cardiology. The results from these studies concluded that patient outcomes are improved in a multidisciplinary team setting (Chang et al., 2001; Junor et al., 1994; Adorian et al., 1990; Kim et al., 2010; McDonald et al., 2002). Similarly, one particular study evaluated the benefit of attending a Down syndrome specialty clinic in regard to the medical care provided. The study involved a retrospective chart review of 105 new patients with Down syndrome (ages 3 and over) that were seen within one year of the opening of the specialty clinic and did not previously have access to a multidisciplinary Down syndrome clinic. Prior to admission to the clinic, only 17% of patients were up-to-date on the AAP recommended guidelines. By receiving medical care from the multidisciplinary team, the remaining 83% of patients were brought up to date on guideline-recommended screening, 41% of patients were referred to specialists outside the clinic for additional medical care, and 54% were diagnosed with new co-occurring medical problems that had previously gone undiagnosed (Skotko et al., 2013). The overall results of this study clearly demonstrate the improved medical care of the patients, a direct result of the multidisciplinary approach to health care management.

Down syndrome clinics are equipped to provide both expertise and support to patients and their families. Themes reflecting a holistic and “family-centered” approach are apparent in mission statements and goals established for each clinic (Children’s Hospital of Wisconsin, 2014). Not only can clinics provide information and updates on outside resources such as conferences, books, and support groups/organizations, they can also provide psychosocial support to patients and their families during clinic visits (Skotko et al., 2013). Clinics may also elicit the help of health care providers such as psychologists or psychiatrists or social workers and patient advocates in order to provide psychosocial services for patients and their families.

However, the utilization of genetic counselors as part of the multidisciplinary team may provide an avenue of support for patients and their families. Genetic counselors are health care consultants who are a part of the genetics team that receive specialized training in providing psychosocial support along with genetics assessments and education. Their main function as part of the health care team is to help “people understand and adapt to the medical, psychological, and familial implication of genetic contributions to disease” (National Society of Genetic Counselors’ Definition Task Force, 2006). According to the Global Down Syndrome Foundation’s medical care centers database, 29 out of 45 (62%) registered pediatric Down syndrome specialty clinics in the United States include genetic counselors as part of their multidisciplinary team.

Few studies have been conducted to evaluate the benefits of including genetic counselors in a pediatric health care team. One particular study, conducted at Cincinnati Children’s Hospital Medical Center, found that adherence to recommended medical management protocols was significantly increased in the pediatric patient population that was seen by a genetic counselor in combination with a geneticist. In a retrospective chart review, the authors found that all categories of medical management recommendations made during the appointment were more strictly followed by the patient population seen by pediatric genetic counselors in comparison to those who did not (Rutherford et al., 2014). Many studies have evaluated the outcomes and areas of patient satisfaction after genetic counseling across multiple specialties, although none have looked at the pediatric setting specifically (Bjorvatn et al., 2007; Christiaans et al., 2009; Clark et al., 2000; DeMarco et al., 2004). The five most commonly reported areas of patient satisfaction include the provision of information/acquisition of knowledge, psychosocial support (immediate and long-term), anticipatory guidance, facilitation of communication, and aid in decision-making (Benhardt et al., 2000).

To date, current literature is lacking research that examines the effectiveness and potential added value of the psychosocial aspect of the multidisciplinary clinics. The primary objective of this study is to evaluate parents' perspectives concerning the level of satisfaction with care and level of psychosocial support provided by multidisciplinary Down syndrome clinics, in comparison to independent pediatric specialty care. Other objectives of this study include collecting parent commentary on genetic counseling in the pediatric setting in order to better assess and understand the impact of providing psychosocial support to families.

Survey responses were collected from parents of children with Down syndrome with the intention of comparing perspectives on overall satisfaction with care and level of psychosocial support, based on whether or not their child attends a multidisciplinary clinic. The goal is to effectively assess the perspectives and opinions of the parents in regard to the psychosocial aspects of their child's pediatric care. We hypothesize that parents whose children attend multidisciplinary clinics will report a higher level of overall satisfaction with care and higher level of psychosocial support than those who do not.

With the growing number of multidisciplinary clinics across all specialties, it is important to evaluate the effectiveness of the clinics themselves. Although studies have been conducted that evaluate the added benefit in terms of improved health care and management, there are no formal studies evaluating the other important aspects of the clinic, such as support provided to the families. Therefore, this study will not only serve as an evaluation for the clinic system itself, but also attempt to better understand the family-wide benefits of multidisciplinary clinics. This study will provide healthcare professionals, including but not limited to genetic counselors, with insights about how to better serve and support patients and their families. By collecting the parents' feedback, healthcare professionals can better understand the psychosocial benefits of multidisciplinary care and identify areas that need improvement.

2.3 Materials and Methods

This research study collected quantitative and qualitative data from parents of children with Down syndrome. Participants were recruited by an invitation to take the online survey that was distributed through Down syndrome support groups across the nation. Support groups that agreed to help with participant recruitment circulated invitations through mailing lists, newsletters, websites, Facebook pages, and other mediums of communication. Parents (over the age of eighteen) who had at least one child with Down syndrome between 0 – 21 years of age were eligible to participate in this study. Individuals who were under the age of eighteen and/or whose child was older than the specified age range were excluded from this study. Participants who did not meet inclusion criteria, and were therefore disqualified from the study, were denied access to the remainder of the survey (using survey programming settings).

A request for participation was sent out to the Down syndrome support groups with available and active e-mail addresses listed on the Down Syndrome Affiliates in Action (DSAIA) directory in August 2014. E-mails were also sent to organizations for Down syndrome including: the National Down Syndrome Society (NDSS), National Down Syndrome Congress (NDSC), National Association for Down Syndrome (NADS), and LuMind Down Syndrome Research Foundation (formerly known as Down Syndrome Research Foundation and LuMind Foundation). The request explained the purpose of the research study and asked for assistance in distributing an invitational letter (Appendix A) and link for the online survey hosted by SurveyMonkey.com through the support group's main mode of communication. The survey link was distributed between September and October, 2014, and was available for completion through December 1, 2014.

The online survey (Appendix B) consisted of demographic questions and a series of multiple choice, Likert scale, and free response questions designed to assess parents' perspectives on their child's pediatric care. Demographic information was obtained related to age, gender, ethnicity, education level, marital status, and number of children. Demographic information (age and gender) about their child with Down syndrome was also collected. Further questions were asked in order to collect information about the child's health care management.

The majority of the Likert scale questions, specifically those aimed at assessing the psychosocial aspects of care, were adapted from the affective and instrumental sections of the Satisfaction with Genetic Counseling scale (Appendix C) (Shiloh, Avdor, & Goodman, 1990). While wording was slightly altered, in order to make the questions more applicable to the participants, the essence and content of the questions remained the same. The aim of these adapted questions was to adequately capture a broader understanding of the psychosocial elements within pediatric care. As this scale had been previously studied, the reliability and consistency were already evaluated. The Cronbach's alpha reliability measure was calculated to be 0.90 in this original study. The previous reliability coefficients were 0.74 for the affective questions and 0.79 for the instrumental questions (Shiloh et al., 1990). In our survey, Cronbach's alpha was 0.96 for the affective questions and 0.90 for the chosen instrumental questions. Overall, the reliability coefficient for all questions adapted from the Satisfaction with Genetic Counseling scale was 0.97. These reliability coefficients are above the widely accepted 0.70 cutoff, suggesting internal reliability.

The remaining survey questions were constructed to evaluate additional areas of interest involving psychosocial support. Other multiple choice and short answer questions were designed to explore parents' experiences with genetic counseling.

This research study was approved by the Institutional Review Board, Office of Research Compliance, of the University of South Carolina, Columbia, SC in August 2014. A pilot study was created to obtain participant feedback about the quality and clarity of the survey questions and was completed by one participant. Suggested changes were incorporated before collection began.

Quantitative analysis of the online surveys was conducted using SPSS version 22.0 statistical software (SPSS Inc., Chicago IL). Chi-square, independent t-tests, and Mann-Whitney U tests were used to determine statistically significant relationships between each survey group for various questions. Frequencies and percentages were also calculated for each question, with the data set divided by survey group.

Qualitative data collected via the online survey were reviewed and coded by the principal investigator. Overlying themes pertaining to pediatric care, psychosocial support, and genetic counseling were identified and analyzed using standard Grounded Theory methods (Strauss, 1987).

2.4 Results

2.4.1 Participant Demographics

The total number of participants who began the survey was 584 (*N* = 584). Inclusion criteria included both parent and child age. Twenty-five participants were disqualified from the survey because their children did not meet the age criteria (*N* = 559). Inclusion criteria data are displayed in Table 2.1.

Table 2.1 Inclusion Criteria Demographics

		Frequency	Percentage (%)
Age of Parent	*n = 584*		
	18 – 24 years	6	1.03
	25 – 34 years	119	20.38
	35 – 44 years	242	41.44
	45 – 54 years	166	28.42
	55 – 64 years	38	6.51
	64 – 74 years	10	1.71
	75 years or older	3	0.51
Age of Child	*n = 572*		
	0 – 3 years	225	39.34
	4 – 8 years	145	25.35
	9 – 12 years	83	14.51
	13 – 17 years	70	12.24
	18 – 21 years	24	4.20
	22 years or older	25	4.37

In addition, 144 participants only answered demographic questions and did not provide any responses to the questions related to the study. These incomplete entries were not included in the study, as they did not provide adequate information to elicit inclusion (*N* = 415).

2.4.2 Group Differentiation

All of the 415 included participants were divided into two groups based on the type of pediatric care their child receives: pediatric care through a multidisciplinary

Down syndrome (DS) clinic or care by independent pediatric specialists. Comparisons between these two groups were made throughout this study. Table 2.2 shows the breakdown between these two groups.

Table 2.2 Participant Breakdown by Group

	Frequency	Percentage (%)
Multidisciplinary pediatric DS clinic	76	18.3
General pediatric specialist care	339	81.7
	N = 415	

2.4.3 Group Demographics

Demographic information was analyzed separately for the two groups, clinic and general care. Demographic information about both the participating parent and their child was collected in this study. Parent demographic information is displayed in Table 2.3. The majority of parent participants were Caucasian females for both groups. The majority of participants, across both groups, also reported being married and having at least some college education. While there was distribution among age range, almost all participants (over 95%) fell between the 25 and 54 years of age in both groups. Chi square tests were conducted on the demographic information in order to determine if there were significant differences between the two groups' population demographic information. All Chi square results (Appendix D) showed no statistically significant differences between any of the demographic criteria, except for education level (p = *.047*).

Table 2.3 Parent Demographic Information

		Percentage (%)	
		Clinic (*n = 76*)	General Care (*n = 339*)
Gender			
	Male	5.3	5.6
	Female	94.7	94.4
Age			
	18 – 24 years	0.0	1.2
	25 – 34 years	17.1	18
	35 – 44 years	56.6	45.7
	45 – 54 years	23.7	30.4
	55 – 64 years	2.6	4.1
	65 – 74 years	0.0	0.6
	75 years or older	0.0	0.0
Ethnicity			
	American Indian or Alaskan Native	1.3	1.2
	Asian or Pacific Islander	1.3	2.4
	Black or African American	6.6	2.7
	Hispanic or Latino	0.0	7.7
	White/Caucasian	90.8	85.5
	Other	0.0	0.0
	Prefer not to answer	0.0	0.6
Education			
	Some high school, but no diploma	0.0	0.0
	High school diploma (or GED)	1.3	5.9
	Some college, but no degree	11.8	17.4
	2-year college degree	7.9	11.5
	4-year college degree	36.8	38.6
	Graduate-level degree	42.1	26.5
Marital Status			
	Single, never married	3.9	2.7
	Married	92.1	89.4
	Domestic partnership or civil union	2.6	2.7
	Separated	0.0	1.5
	Divorced	0.0	3.8
	Widowed	1.3	0.0

Child demographic information is displayed in Table 2.4. Child demographics were relatively equally distributed in all categories across both groups. Chi square tests were conducted on the demographic information in order to determine if there were significant differences between the study groups. All Chi square results (Appendix D) showed no statistically significant differences between the two groups across the child demographic information that was collected.

Table 2.4 Child Demographic Information

		Percentage (%)	
		Clinic (*n = 76*)	**General Care (*n = 339*)**
Gender			
	Male	52.6	51.9
	Female	47.4	48.1
Age			
	0 – 3 years	50.0	36.6
	4 – 8 years	25.0	29.8
	9 – 12 years	11.8	15.3
	13 – 17 years	10.5	14.2
	18 – 21 years	2.6	4.1
Diagnosis			
	Prenatal	21.1	28.6
	Postnatal	78.9	71.4

2.4.4 Psychosocial Care and Support

Several different types of questions were asked to assess various aspects of the psychosocial component of health care. The primary assessment of the level of psychosocial care was the Likert scale questions adapted from the Satisfaction with Genetic Counseling scale. The questions themselves, along with mean scores for each group, are displayed in Table 2.5. Independent-Samples Mann-Whitney U tests were calculated for each question in order to detect if there were statistically significant differences in participant responses across the two groups. The resulting p-values for each question can also be seen in Table 2.5. The mean scores for all adapted scale questions were higher in the Down syndrome clinic group. The mean score comparisons are also displayed in Figure 2.1. Additionally, the mean scores between the two groups were determined to be statistically significantly different, with p-values less than 0.05, for all questions except for question 11 (*p* = *.595*).

Table 2.5 Psychosocial Assessment using Adapted Scale

		Mean Score		**p value**
		Clinic (*n* = *76*)	**General Care (*n* = *339*)**	**(*p*)**
Q1	The health care providers show interest in our personal problems beyond what is medically required.	5.50	4.93	.007
Q2	The health care providers care for my family as people, not just as patients.	5.78	5.41	.012
Q3	The health care providers are reassuring.	5.99	5.44	.000
Q4	The health care providers listen to what we have to say.	6.09	5.72	.003
Q5	The health care providers are considerate of our emotional state during our visits.	5.95	5.32	.000
Q6	The health care providers show dedication in treating the problem at hand.	6.16	5.91	.015
Q7	The health care providers really understand what is bothering our family.	5.62	5.11	.003
Q8	The health care providers spend enough time with us in appointments.	5.96	5.51	.001
Q9	The health care providers are sensitive and tactful during our conversations.	6.07	5.64	.001
Q10	The health care providers make us feel comfortable at our appointments.	6.09	5.81	.011
Q11	I feel comfortable calling to ask my health care providers more questions.	5.68	5.68	.595
Q12	The help we receive better helps us cope with our problems at home.	5.58	5.01	.003
	Please rate your satisfaction with the level of psychosocial support offered to your family by your child's health care providers.	3.89	3.49	.001

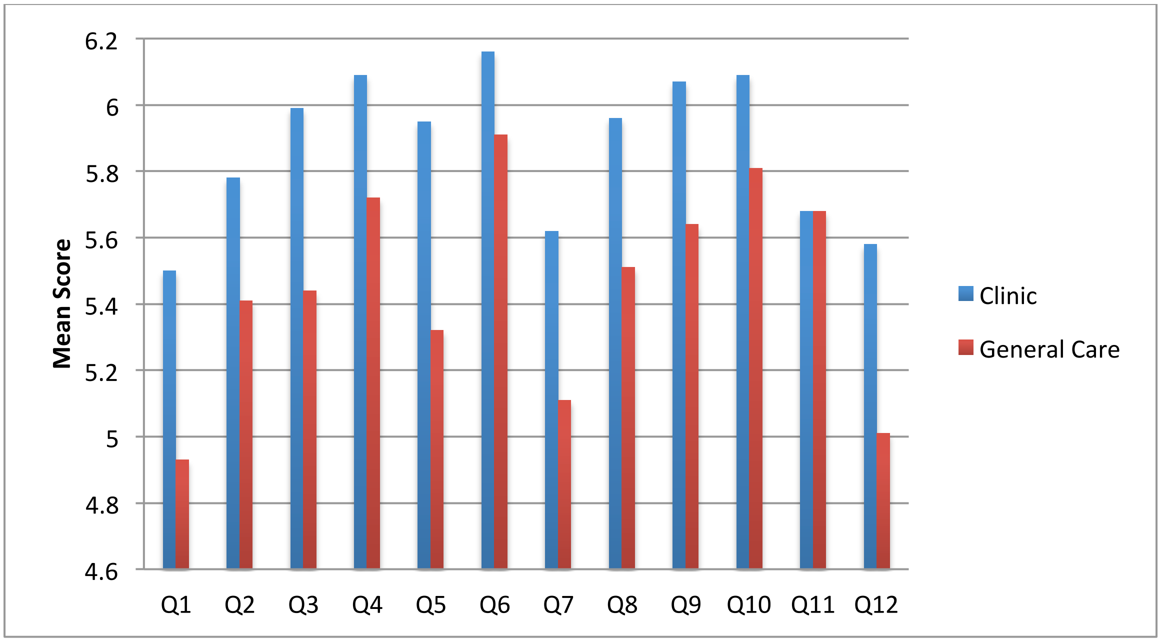

Figure 2.1 Mean Values of Adapted Scale Questions

Additionally, participants were asked to rate their perceived level of psychosocial support provided by their health care providers on a scale from 1 (very dissatisfied) to 5 (very satisfied). The mean scores of both groups and the p-value are included in Table 2.5. The mean score was higher in the Down syndrome clinic group (3.89) as compared to the general care group (3.49). This result can also be seen in Figure 2.2. The Mann-Whitney U test also determined this difference to be significantly different (*p = .001*).

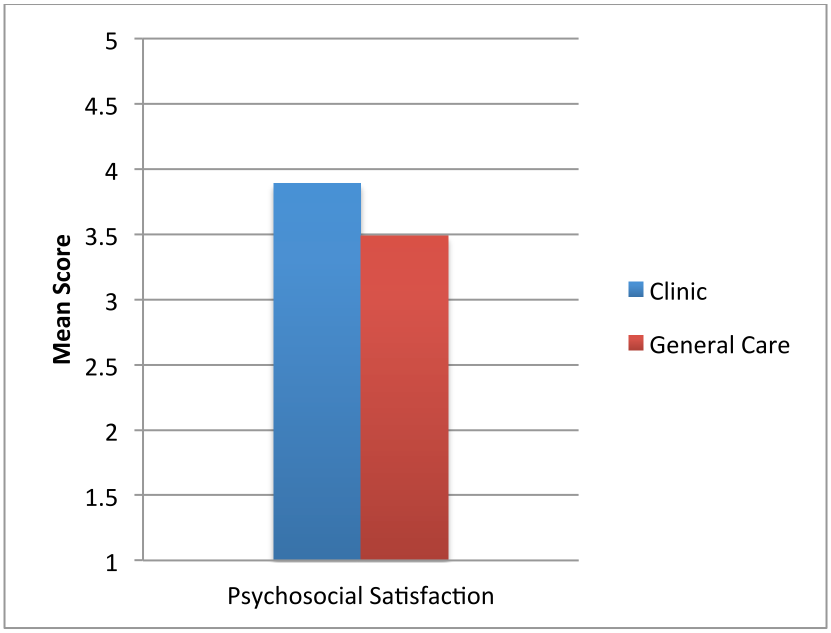

Figure 2.2 Psychosocial Satisfaction Rating

In order to assess other aspects of psychosocial care, participants were asked which resources were offered through their child's health care providers. Across all categories, participants in the Down syndrome clinic group reported being offered more resources than those who receive independent specialist care. The results are displayed in Table 2.6 and Figure 2.3.

Table 2.6 Resources Offered by Health Care Providers

	Percentage (%)	
	Clinic (*n* = *76*)	**General Care (*n* = *339*)**
Books	51.3	23.6
Support groups	50.0	32.7
Conferences	23.7	7.4
Research	32.9	8.6
Civic organizations	11.8	7.7
Play groups	10.5	8.0
Other	10.5	6.8

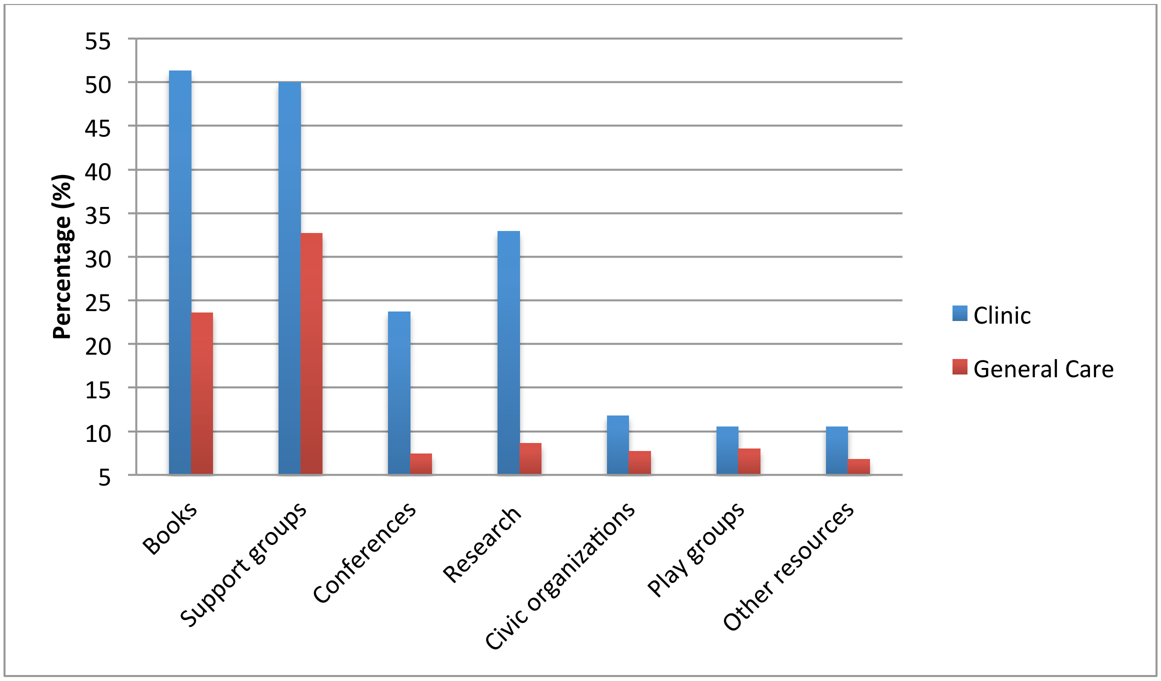

Figure 2.3 Resources Offered by Health Care Providers

The total number of resources offered to each participant was calculated, as well as the average total number of resources for each group. The means were used to determine if there was a statistically significant difference in the number of resources offered to each group, using an Independent Samples t-test. The mean number of resources offered to the Down syndrome clinic group (1.91) was over twice the mean number of the general care group (0.94), and this difference was determined to be statistically significant ($p = .000$).

Participants were asked to rank resources from most supportive to least supportive. Both groups responded very similarly, with friends and family being the most supportive resource in both the Down syndrome clinic (60.5%) and general care (58.7%) groups. Support groups/other parents of children with Down syndrome and health care providers followed in second and third places. Civic and religious groups and written and Internet information were ranked in the bottom three across both groups as well.

Participants were also asked to list their four most supportive health care providers. Results for both groups were similar, with pediatricians being ranked as most supportive by both the Down syndrome clinic (44.7%) and general care (54.9%) groups. The other most common ranked specialists included ophthalmology, otolaryngology, cardiology, genetics, developmental pediatrics, and endocrinology.

2.4.5 Medical Care

Several questions about the child's medical care were also included in the study. Parents were asked to list all the specialists that follow their child regularly. The percentages per group are displayed in Figure 2.4.

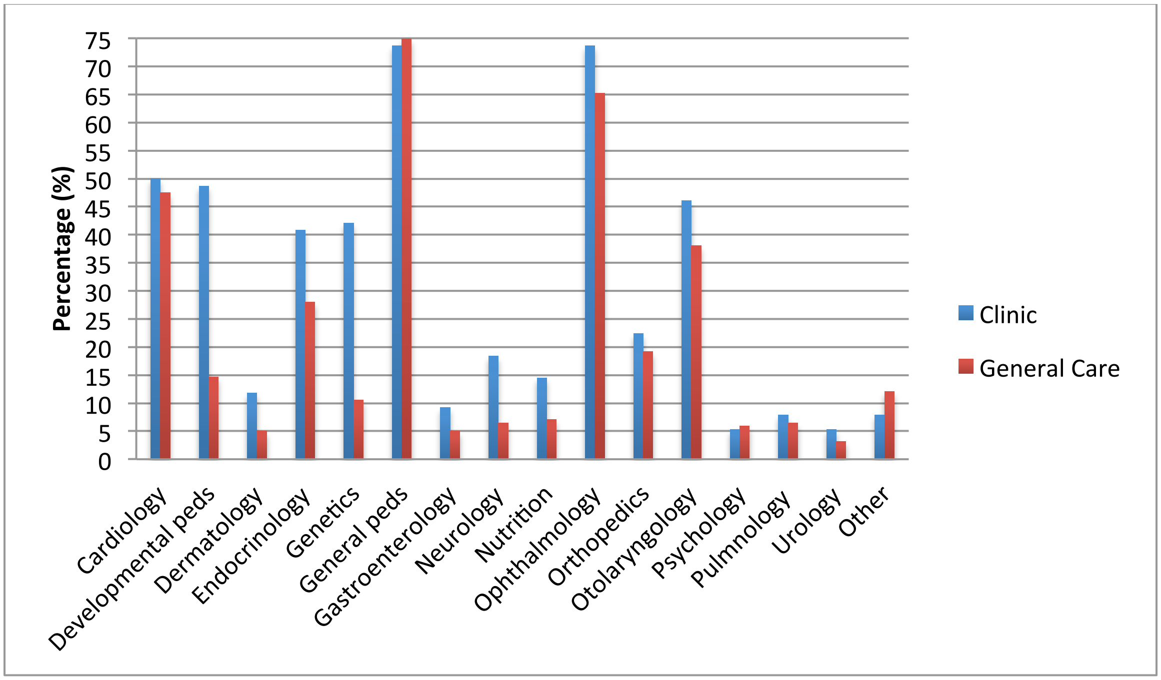

Figure 2.4 Health Care Specialists

Using participant's responses, the total number of specialists seen by each child was calculated and used to determine the mean number of specialists for each group. The means for each group were used to evaluate statistical significance using an Independent Samples t-test. The mean number of specialists seen by patients in the Down syndrome clinic group (4.82) was greater than the general care group (3.53), and this difference was determined to be statistically significant (*p = .000*).

Additionally, participants were asked to rate their overall satisfaction with their child's health care providers on a scale from 1 (very dissatisfied) to 5 (very satisfied). The mean score was higher in the Down syndrome clinic group (4.18) as compared to the general care group (4.04), which can be seen in Figure 2.5. The difference between the two groups trended toward significance (*p = .055*) using an Independent-Samples Mann-Whitney U test.

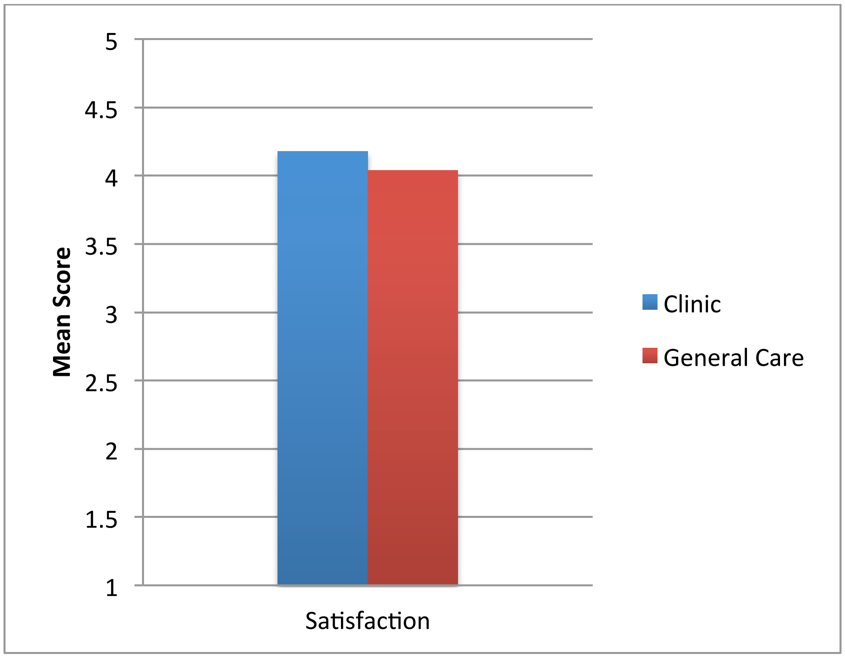

Figure 2.5 Overall Satisfaction Rating

2.4.6 Qualitative Review of Parents' Reflections on Genetic Counseling

Qualitative results were obtained from free response questions in the online survey. One hundred and ninety-two participants responded to the questions related to having experience with a pediatric genetic counselor and 180 participants responded to questions pertinent to those with a lack of personal experience with pediatric genetic counseling. Grounded theory methods were used to reveal major themes in the responses in both categories.

Four major themes were revealed as a response to personal experience with pediatric genetic counseling. Three of these are highlights of the positive aspects, and one was related to negative experiences. In addition, two other themes related to a lack of experience with pediatric genetic counseling were also identified.

Theme 1: Specialized, expert information

A majority of people emphasized the aspect of information sharing at pediatric genetics appointments. A large sub-section highlighted that this information was specialized information, specific to Down syndrome. Participants also mentioned that the information is given by health care providers have "experience and expertise", with lots of knowledge of Down syndrome, and therefore the information is more useful and reliable. Many parents noted that because genetics professionals specialize in Down syndrome, they "feel confident in the advice shared, and what to look for medically and socially." Others highlighted that the information given and received in these appointments is more thorough and in-depth than appointments with other providers. One parent noted that "it was nice to have someone take the time to thoroughly explain things and ensure we understood our [child]'s diagnosis."

Theme 2: Discussion

Themes of talking and discussion were also very prevalent in participant responses. Parents highlighted that there is a larger component of asking and answering questions in genetics appointments. Several described the appointments as "less medical" than other doctor appointments; with others highlighting that these appointments are "more supportive" in nature. Parents noted that the genetics team provides more help and guidance and that they are more caring and understanding than providers in other settings. One parent highlighted that she feels her "son's genetic counselor really cares about [their] entire family unit." Several parents emphasized that appointments are for both the children and their parents – and that discussion is open to family and parental concerns, not just medical ones. Some parents expressed that "[they] wish [they] had more visits to talk… about [their] concerns."

Theme 3: Comprehensive medical care

Many participants noted that genetics appointments involve the "big picture" of their child's medical history. Many emphasized that "genetics visits are all-encompassing health care, where the other health care appointments are for specific concerns." Parents discussed that this "head to toe care" includes not only medical evaluation, but also development, social interactions, school performance, and other important aspects of the child's life. One parent stated that he felt genetics appointment "'connects the dots' so to speak when it comes to [the] child's care." Parents also discussed that these appointments facilitate coordination of health care, with the genetics team ensuring that medical management is optimal and all appropriate and relevant referrals are being made. With their expertise in the field, parents see the value in genetics professionals "keeping [their other doctors] on track as far as Down syndrome-specific care."

Theme 4: "Not helpful"

Other participants did not report positive experiences with pediatric genetic counseling. Some described their experiences as "not helpful". A portion of these respondents voiced that, although information was given, it was information that they had already known/researched themselves. One parent commented that they "knew most of the information. It seemed pointless." Others mentioned that the first genetic counseling session (or first few sessions) was very helpful – in terms of receiving information about Down syndrome – but after they felt knowledgeable on the subject, they felt as if the appointments were not worth their time. Many parents echoed similar opinions in that "past the early years, [appointments were] not very helpful." They no longer saw the point or benefit of the appointments. Many of these respondents reported no longer being followed by genetics, out of personal choice because they "no longer [felt] the need to go." A majority of participants discussed that their pediatric genetic counseling experiences were one-time events. These appointments were reported as mostly return of genetic testing results and NICU consultations. Some respondents also described these limited interactions as being minimally beneficial.

Theme 5: Negative experiences and perceptions

Some participants, without a personal experience of pediatric genetic counseling, reflected on reasons why they chose not to attend genetics appointments. The vast majority of these respondents reflected on either a personal negative experience or relayed a negative experience of someone they knew. Most of these experiences, however, related to prenatal genetic counseling. Some of these recollections included hearing negative information about the diagnosis from prenatal health care professionals and feeling pressured to terminate a pregnancy. One parent recollected that their "prenatal experience with genetic counseling was bad… [they] haven't had great interaction with the profession." A few people reported that friends had told them that genetic counselors provide negative as well as unhelpful information about Down syndrome. Another parent shared that they had "heard too many stories about the misinformation that genetic counselors give parents when Down syndrome is diagnosed. [They] did not feel any need to put [themselves] through that." These respondents also emphasized a need for "honest, unbiased, and up-to-date information about… Down syndrome." Other parents expressed that they wished health care professionals would give balanced information, not simply information focusing on seemingly negative aspects of the diagnosis.

Theme 6: General misunderstanding or lack of knowledge of pediatric genetic counseling

The majority of respondents without personal experience with pediatric genetic counseling reported that genetics appointments had never been discussed with them or offered. Many parents noted that they weren't aware "it was an option or a resource" and that referrals to genetics "were never offered." They also revealed that they did not know the role genetics would play in their child's health care or the purpose of genetics appointments. One parent confessed that they had "no idea what a genetic counselor do besides deliver a definitive diagnosis." Many asked questions about pediatric genetic counseling in their responses due to their lack of awareness of the profession, particularly about its utility. For example, questions such as "what is the purpose?" were frequently recorded.

In addition, many participants' responses indicated confusion between prenatal and pediatric genetic counselors. One particular parent drew attention to this misunderstanding when they said, "I see genetics as a different field from genetic counseling. I see genetic counseling as related to pregnancy and pregnancy planning. I see genetics as a medical specialty working with people with genetic conditions." However, many participants echoed this confusion through their survey responses. Most commonly, participants answered "yes" to the question asking if they had seen a pediatric genetic counselor as part of their child's health care, and then only described experiences with prenatal genetic counselors in their subsequent responses. Several people reported that they did not feel the need to attend pediatric genetic appointments because they weren't having other children and therefore were not interested in the recurrence risk. One parent said that, after their child was born, they "were offered the services of genetic counseling but they didn't understand why it was necessary since [they] didn't plan on having any more children." Generally, it was evident that most participants without personal experience with pediatric genetic counseling were not aware of the role or purpose of genetics in the realm of pediatric health management.

2.5 Discussion

This study explored the psychosocial aspect of pediatric health care related to the medical management of children with Down syndrome. Throughout this study, comparisons were made between two groups: parents whose children attend multidisciplinary Down syndrome clinics and parents whose children receive independent specialist care. These comparisons were made in order to execute the main objective of our study: to evaluate the effectiveness of the psychosocial or “family-centered” care provided at multidisciplinary Down syndrome clinics.

Overall, we found there to be significantly greater psychosocial support provided by the multidisciplinary Down syndrome clinics as reported by parents who responded to the survey. These results were consistent across multiple assessments of psychosocial support including the adapted Likert scale questions, evaluation of resources provided, along with other questions. Parents reported a higher level of psychosocial support across all the affective sub-set questions adapted from the Satisfaction with Genetic Counseling scale, all showing statistically significant differences between the two groups. These questions explored a variety of aspects important to psychosocial care, indirectly assessing satisfaction with psychosocial care. Questions evaluated qualities in health care providers and the health care provided. Topics included the considerate, caring, and understanding qualities of the health care provider and the helpful and thorough nature of the health care provided, along with other measures. The one instrumental question, question 11 (“I feel comfortable calling to ask my health care providers more questions”), that was not found to be significantly different was not a question directly assessing health care providers, but rather was a reflection of the parents themselves. This content difference is a possible explanation for the inconsistency of results as compared to the rest of the Satisfaction with Genetic Counseling scale questions. Additionally, when directly asked to rate their satisfaction with psychosocial support, parents whose children attend Down syndrome clinics reported greater satisfaction (with the numerical score equivalent of “satisfied” response) than those whose children do not (with the numerical score closer to a “neutral” response). This difference between the groups was also statistically significant, further solidifying the similar results obtained from the indirect, Satisfaction with Genetic Counseling scale assessments. Satisfaction with psychosocial care was consistently higher in the Down syndrome clinic group across all measures, both direct and indirect.

Supportive resources were also identified as an important aspect of psychosocial care in our study. Recommending resources, such as written information, support groups, and conferences, allow providers to promote psychosocial wellbeing for patients and their families. It also allows health care providers to foster psychosocial support in areas which they cannot provide support personally or directly. We found that providers within multidisciplinary Down syndrome clinics offer more outside resources more frequently than other independent health care providers. These results were consistent across all categories of resources. Using statistical analysis, these differences in resources offered were significantly different between the two study groups. These additional results intimate that greater psychosocial care is provided to families by Down syndrome clinics than independent specialists. Although no previous research has been done in this specific area, these results are consistent with our predicted hypothesis.

We asked parents to rank supportive outlets in terms of which resources provided the most support for their family. Both groups responded very similarly, reporting that friends and family was the most supportive resource. Parents also conveyed that support groups/other parents of children with Down syndrome and health care professionals provide valuable support. These resources were consistently ranked in the top three for both study groups. Upon specifically inquiring about health care providers, we found that both study groups ranked their pediatrician as most supportive most frequently –

despite the Down syndrome clinic group reporting higher levels of psychosocial support. This finding may be due to the fact that pediatrician appointments occur more frequently than Down syndrome clinic appointments. As a result, parents may have more regular interactions and therefore feel most supported and comforted by their pediatrician. It can be assumed that children who attend Down syndrome clinics in addition to being followed by a pediatrician for healthcare needs unrelated to Down syndrome and regular well visits. It is possible that parents whose children attend multidisciplinary clinics feel more support than the general care study group due to the additional attention and care they are receiving from the clinic visits. However, due to the more frequent interaction with their pediatrician, parents still may view their pediatrician as a more supportive resource for their family.

In the process of assessing psychosocial care, several questions about medical care were included in this study. Our results demonstrated that children who attend multidisciplinary Down syndrome clinics are followed by more specialist doctors than those who receive independent specialist care. The average number of doctors per child demonstrated a statistically significant difference between the two study groups. These results may reflect findings from previous studies focused on the added medical benefit of Down syndrome clinics. A previous study demonstrated that multidisciplinary Down syndrome clinics provide better, more comprehensive medical care to patients. As part of the results, the study found that as a result of multidisciplinary clinic attendance, 83% of patients were brought up-to-date on screenings based on published guidelines, 41% were referred to outside specialists for additional medical care, and 54% were diagnosed with new co-occurring medical problems (Skotko et al., 2013). These additional screenings, referrals, and diagnosed comorbidities would therefore result in more specialist visits. Our results may be a reflection of this previously discovered consequence of attending multidisciplinary clinics for pediatric health care. An alternative explanation may be that patients who attend multidisciplinary clinics are more medically complex and therefore seek out medical care by Down syndrome specialty clinics.

Previous studies and our results indicate both psychosocial and medical advantages to attending pediatric multidisciplinary Down syndrome clinics. Our study directly asked parents to rate their overall satisfaction with their child's health care providers. Although the Down syndrome clinic group reported a higher level of satisfaction, the difference in the two groups was not considered to be statistically significant.

Our comparisons of participant population information supported the integrity of our study's results. Even though our two study groups had unequal numbers, our group populations were shown to be equivalent in terms of demographics of both the parents and the children. Statistical analysis confirmed that the group differentiation did not affect the reliability of the study. The study groups were shown to be comparable, as the demographic information was shown not to be significantly different between the two groups. Results showed that education level of parents varied slightly between the two study groups. However, we do not anticipate this variation to impact other results as almost all participants across both groups reported a minimum of some college education.

As a secondary objective of this study, we also collected parent commentary on their experiences, reflections, and opinions of genetic counseling in the pediatric setting in order to better assess and understand the impact of providing psychosocial support to families. We collected parents' perspectives in the form of free response questions. Multiple themes were discovered during analysis of parent commentary.

When analyzing parents' experiences with pediatric genetics, we discovered four major themes. Three of these themes reflected positive outcomes and areas of satisfaction as a result of genetic counseling. These positive outcomes included specialized and expert information, the opportunity for discussion, and comprehensive medical care. Previous studies have found similar findings, with the common themes: provision of information/acquisition of knowledge, psychosocial support, anticipatory guidance, facilitation of family communication, and aid in decision-making (Bernhardt, Biesecker, & Mastromarino, 2000). Our results mirror these previous studies. The fourth theme was a reflection of a negative outcome of personal experience with genetic counseling. Some parents reported that genetic appointments were "not helpful". Parents who reported this sentiment reflected on either insufficient information or time. Many parents stated that these visits were one-time events in which basic information and/or genetic testing results were given. Many parents also reported that the first few appointments were helpful, but that after the parents knew information about Down syndrome, they no longer saw the point of attending genetics appointments.

In order to evaluate how to better serve our pediatric population, we thought it was also important to understand parents' reasons for not attending genetics appointments. By exploring this topic, we were able to identify two major themes: negative experiences and perceptions and misunderstanding or general lack of knowledge. In terms of negative experiences, many people reported that negative experiences with prenatal genetic counseling influenced their decision to forgo pediatric genetics appointments. Other parents reported that they had been told stories about negative experiences with genetic counseling that influenced their decisions. However, the vast majority of parents without personal experience with genetic counseling reported that this was a result of never being referred to genetics or being offered an appointment with genetics. These parents also indicated a lack of knowledge about the purpose of genetics appointments or what they involve. Other parents displayed a general misunderstanding of pediatric genetics. Most showed confusion between prenatal and pediatric genetics, assuming that both specialties provide the same information and fulfill the same roles in health care provision.

There were, however, limitations to this study – primarily dealing with the participant population. Our population sample consisted of primarily Caucasian, highly educated, and married females across both study groups. As our study population is primarily comprised of this narrow sociodemographic band, the results obtained from this study may not reflect the opinions and experiences of other social, ethnic, or demographic populations. Furthermore, as this study relied on voluntary participation, selection/ascertainment bias may have also influenced results. It is possible that parents who volunteered to participate had stronger or polarized views regarding their child's healthcare as well as other issues explored in this study. Our research study aimed to capture a large number of parent responses in order to explore an extensive number of parent opinions and experiences. This goal was attained through purposive sampling techniques. Although our two study groups' participant populations were homogenous, they were not representative, random samples from the population at large. Therefore, due to these limitations, it is not possible to generalize the study findings or make any definitive statements.

Additionally, in our study we did not capture information about how frequently patients and their families visit their pediatrician and independent specialists or attend multidisciplinary Down syndrome clinic appointments. This information could potentially influence parents' responses on the topics evaluated in this study, and would therefore be critical to collect in future studies on this topic. Future studies could also explore the differences between the different multidisciplinary Down syndrome clinics and evaluate how those factors influence parents' perceived level of psychosocial support. Information collected could be valuable in obtaining feedback from parents and understanding how to best serve patients and their families, in terms of the psychosocial aspects of their healthcare, in the hopes of modifying aspects of the clinics that could provide added value and benefit for attendees.

2.6 Conclusion

This study focused on evaluating the effectiveness of the psychosocial or "family-centered" care provided at pediatric multidisciplinary Down syndrome clinics. We surveyed parents of children with Down syndrome about many facets of psychosocial care as well as additional aspects of their child's medical management. The added psychosocial benefit of attending Down syndrome clinics was assessed by comparing responses between the two study groups: those whose children attend multidisciplinary clinics and those who receive independent specialist care. We hypothesized that due to the "family-centered" approach of multidisciplinary Down syndrome clinics, parents whose children attend these clinics will report a higher level of psychosocial support provided as part of their child's health care. Overall, participants in the Down syndrome clinic group reported significantly greater levels of psychosocial care by their health care providers. These results were consistent across all included measures of psychosocial assessment, including resource referral and recommendations.

Our study demonstrates that attendance at a multidisciplinary Down syndrome clinic might increase and improve the psychosocial care of families. Based on the results of this study and previous research, specialty multidisciplinary Down syndrome clinics are shown to provide added benefits in terms of improved medical management and greater psychosocial support. We believe that referrals to multidisciplinary clinics can improve overall outcomes for patients and their families and should be considered in areas where these clinics are accessible and available.

In addition, our study also found that individuals are not clear on the purpose or scope of practice of genetic counselors outside of prenatal care. Therefore, we also emphasize the importance of education, not only for individuals in the community but also for other health care providers, in the realm of pediatric genetic counseling. Specifically, this education should include the function of genetic counseling as part of the interdisciplinary pediatric health care team and the potential value and benefit for patients and their families. With increased knowledge of the field, health care providers will be well-informed of the purpose of pediatric genetics appointments and therefore be equipped to make appropriate referrals, when available. Likewise, parents will be able to advocate for their own children's medical care and initiate discussion of a referral with their health care providers as well. Education on the topic of genetic counseling seems to be vitally important to rectify misinformation and acquaint those with a lack of experience with the specialty.

Chapter 3. Conclusions

This study focused on evaluating the effectiveness of the psychosocial or "family-centered" care provided at pediatric multidisciplinary Down syndrome clinics. We surveyed parents of children with Down syndrome about many facets of psychosocial care as well as additional aspects of their child's medical management. The added psychosocial benefit of attending Down syndrome clinics was assessed by comparing responses between the two study groups: those whose children attend multidisciplinary clinics and those who receive independent specialist care. We hypothesized that due to the "family-centered" approach of multidisciplinary Down syndrome clinics, parents whose children attend these clinics will report a higher level of psychosocial support provided as part of their child's health care. Overall, participants in the Down syndrome clinic group reported significantly greater levels of psychosocial care by their health care providers. These results were consistent across all included measures of psychosocial assessment, including resource referral and recommendations.

Our study demonstrates that attendance at a multidisciplinary Down syndrome clinic might increase and improve the psychosocial care of families. Based on this study and previous research, specialty multidisciplinary Down syndrome clinics are shown to provide added benefits in terms of improved medical management and greater psychosocial support. We believe that referrals to multidisciplinary clinics can improve overall outcomes for patients and their families and should be considered in areas where these clinics are accessible and available.

In addition, our study also found that individuals are not clear on the purpose or scope of practice of genetic counselors outside of prenatal care. Therefore, we also emphasize the importance of education, not only for individuals in the community but also for other health care providers, in the realm of pediatric genetic counseling. Specifically, this education should include the function of genetic counseling as part of the interdisciplinary pediatric health care team and the potential value and benefit for patients and their families. With increased knowledge of the field, health care providers will be well-informed of the purpose of pediatric genetics appointments and therefore be equipped to make appropriate referrals, when available. Likewise, parents will be able to advocate for their own children's medical care and initiate discussion of a referral with their health care providers as well. Education on the topic of genetic counseling seems to be vitally important to rectify misinformation and acquaint those with a lack of experience with the specialty.

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Appendix A. Invitation to Participate Letter

University of South Carolina School of Medicine
USC Genetic Counseling Program

Dear Potential Participant:

You are invited to take part in a graduate research study focusing on psychosocial support provided to families of children with Down syndrome by pediatric health care professionals. I am a graduate student in the genetic counseling program at the University of South Carolina School of Medicine. My research looks at parent's perspectives on the support given to their family through their child's pediatric health care. The research involves taking a survey that is online. Each survey is meant to be filled out individually by either parent.

The survey contains a series of questions about your child with Down syndrome and the types of medical providers he/she sees on a regular basis. The survey also asks about your opinions and perspectives on the level of psychosocial/emotional support that is offered by your child's pediatric health care providers.

All responses from the surveys will be kept anonymous and confidential. The results of this study might be published or presented at scientific meetings; however, your answers will not be identified in any way. The survey should take about 10-15 minutes to complete.

Your participation in this research is voluntary. By completing the survey, you are consenting that you have read and understand this information. At any time, you may withdraw from the study by exiting out of the survey.

Thank you for your time and consideration for taking part in this study. Your answers may help health care professionals provide the best care for children with genetic conditions and their families. If you have any questions about this research, you may contact my faculty adviser, Richard Ferrante, Ph.D., or me using the information below. If you have any questions about your rights as a research member, you may contact the Office of Research Compliance at the University of South Carolina at (803) 777-7095.

Sincerely,
Devon Haynes, B.A., B.A.
Master of Science Candidate
University of South Carolina School of Medicine

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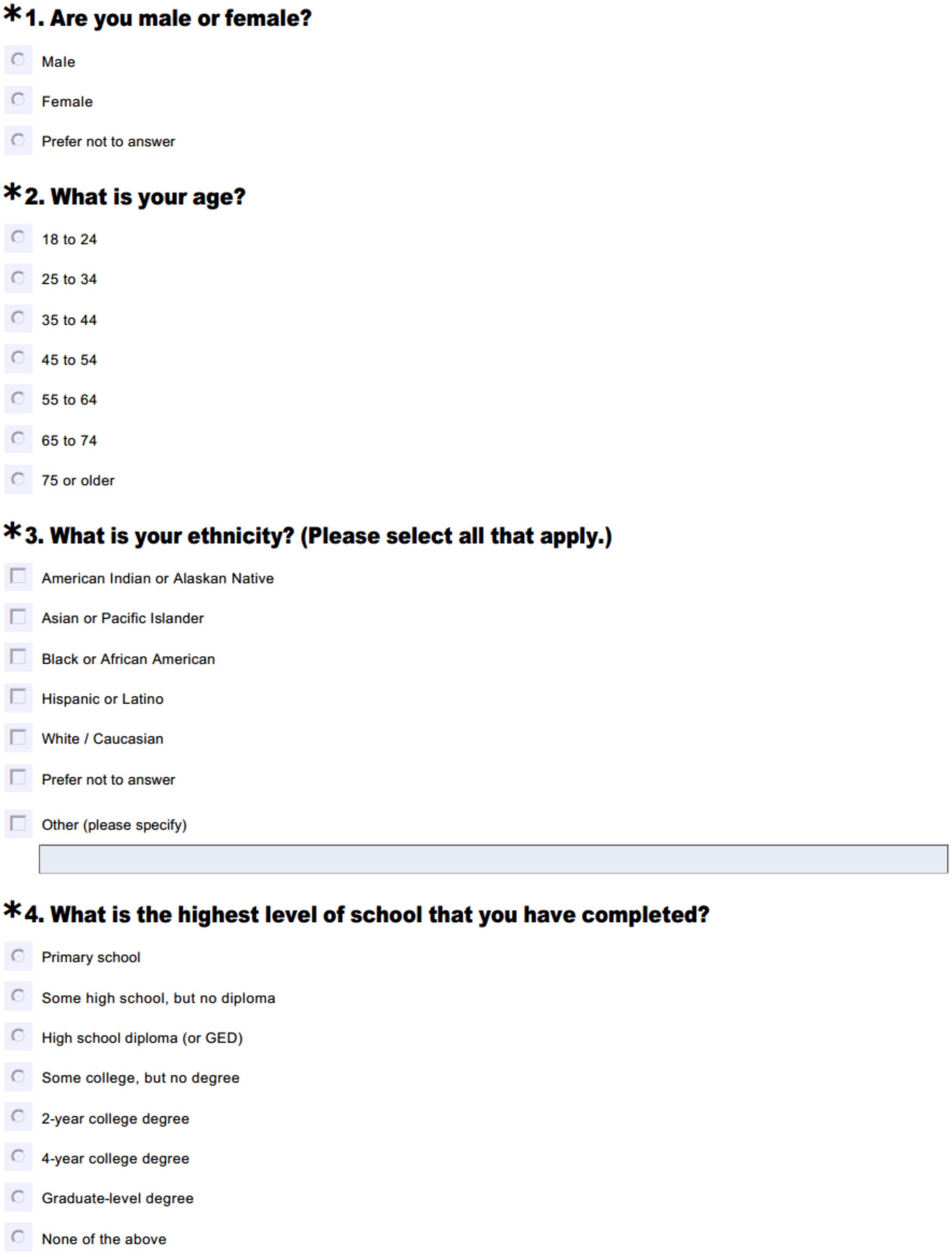
Appendix B. Survey



***1. Are you male or female?**

○ Male

○ Female

○ Prefer not to answer

***2. What is your age?**

○ 18 to 24

○ 25 to 34

○ 35 to 44

○ 45 to 54

○ 55 to 64

○ 65 to 74

○ 75 or older

***3. What is your ethnicity? (Please select all that apply.)**

☐ American Indian or Alaskan Native

☐ Asian or Pacific Islander

☐ Black or African American

☐ Hispanic or Latino

☐ White / Caucasian

☐ Prefer not to answer

☐ Other (please specify)

***4. What is the highest level of school that you have completed?**

○ Primary school

○ Some high school, but no diploma

○ High school diploma (or GED)

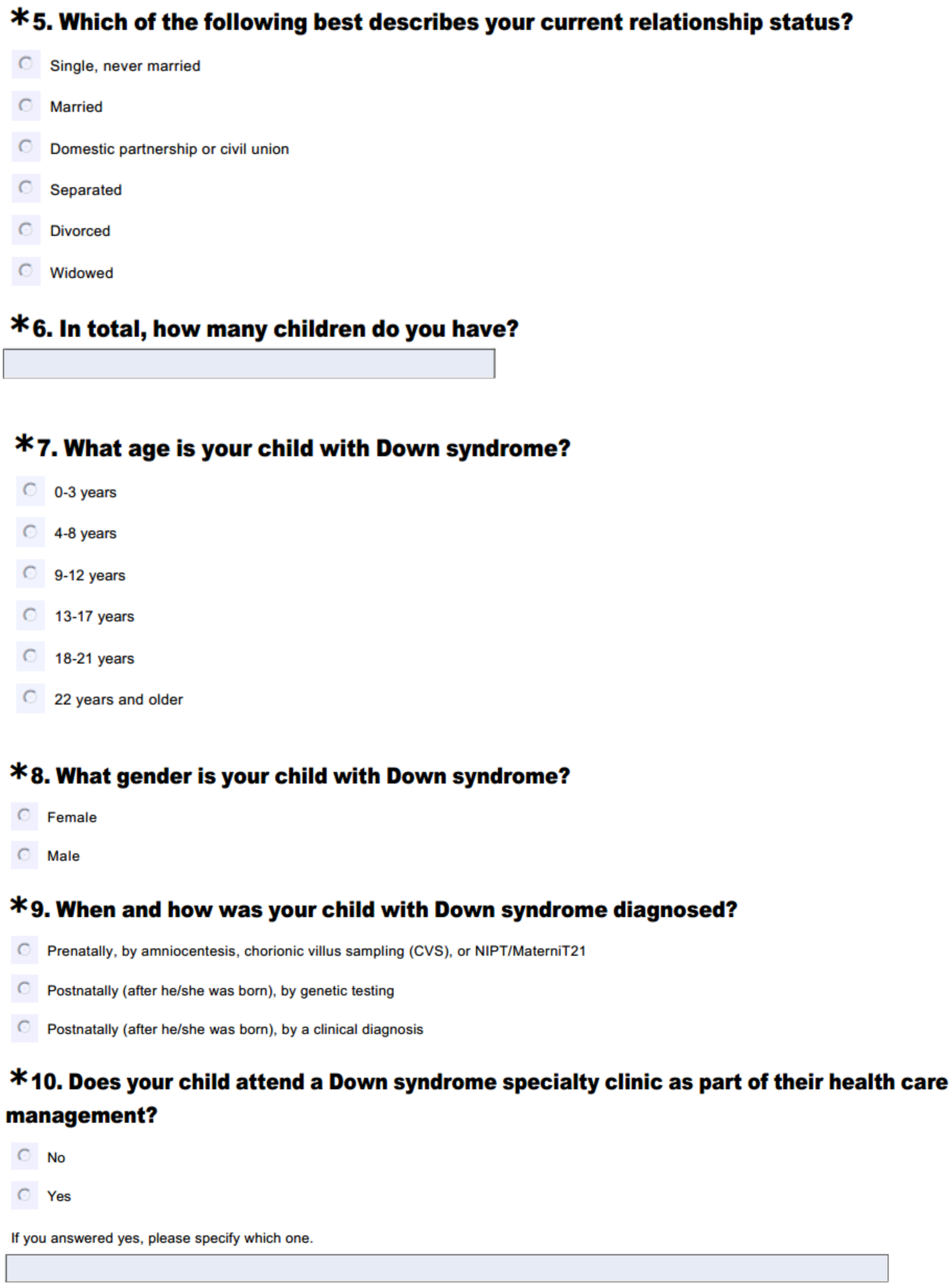
○ Some college, but no degree

○ 2-year college degree

○ 4-year college degree

○ Graduate-level degree

○ None of the above



***5. Which of the following best describes your current relationship status?**

○ Single, never married

○ Married

○ Domestic partnership or civil union

○ Separated

○ Divorced

○ Widowed

***6. In total, how many children do you have?**

***7. What age is your child with Down syndrome?**

○ 0-3 years

○ 4-8 years

○ 9-12 years

○ 13-17 years

○ 18-21 years

○ 22 years and older

***8. What gender is your child with Down syndrome?**

○ Female

○ Male

***9. When and how was your child with Down syndrome diagnosed?**

○ Prenatally, by amniocentesis, chorionic villus sampling (CVS), or NIPT/MaterniT21

○ Postnatally (after he/she was born), by genetic testing

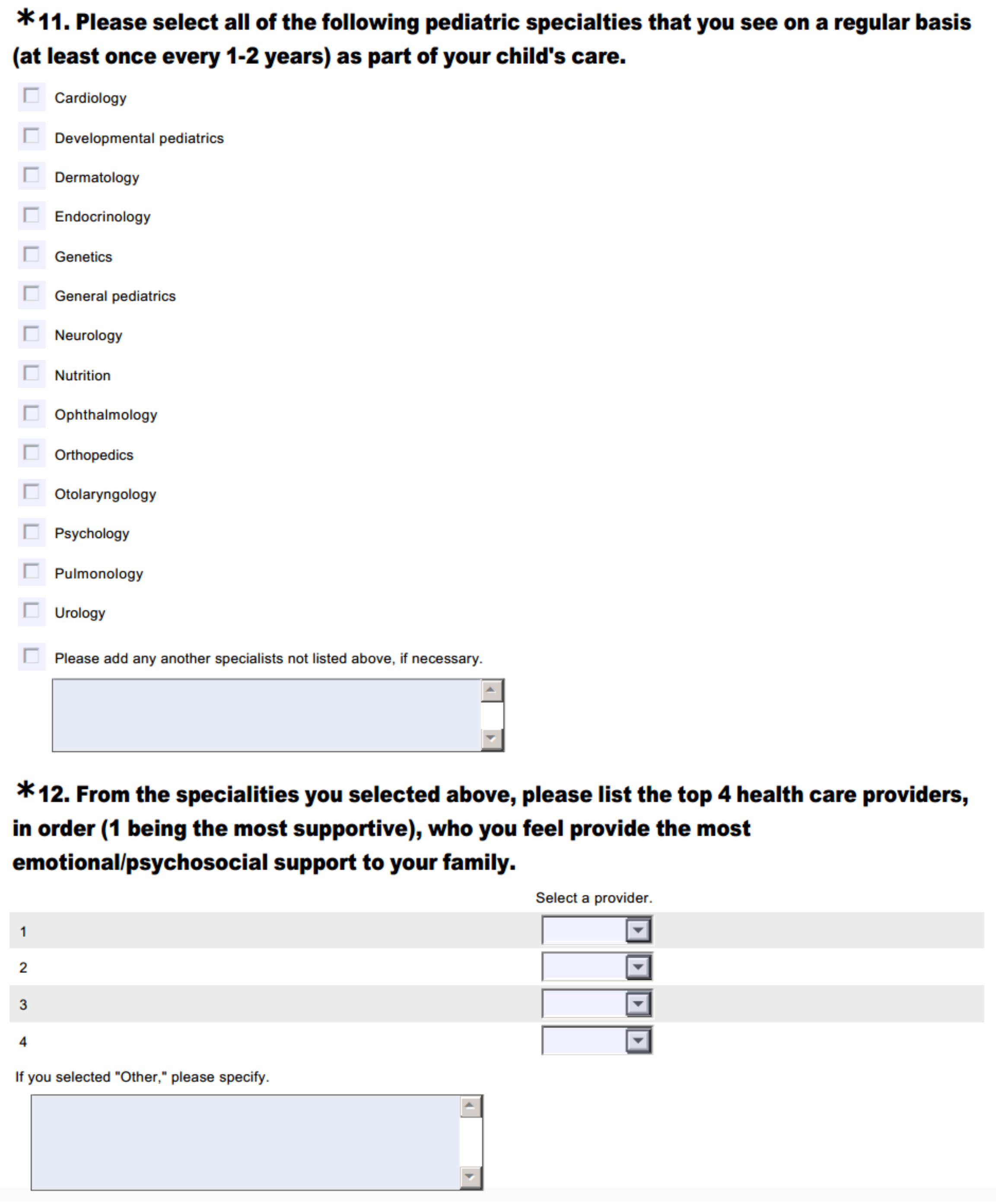
○ Postnatally (after he/she was born), by a clinical diagnosis

***10. Does your child attend a Down syndrome specialty clinic as part of their health care management?**

○ No

○ Yes

If you answered yes, please specify which one.



***11. Please select all of the following pediatric specialties that you see on a regular basis (at least once every 1-2 years) as part of your child's care.**

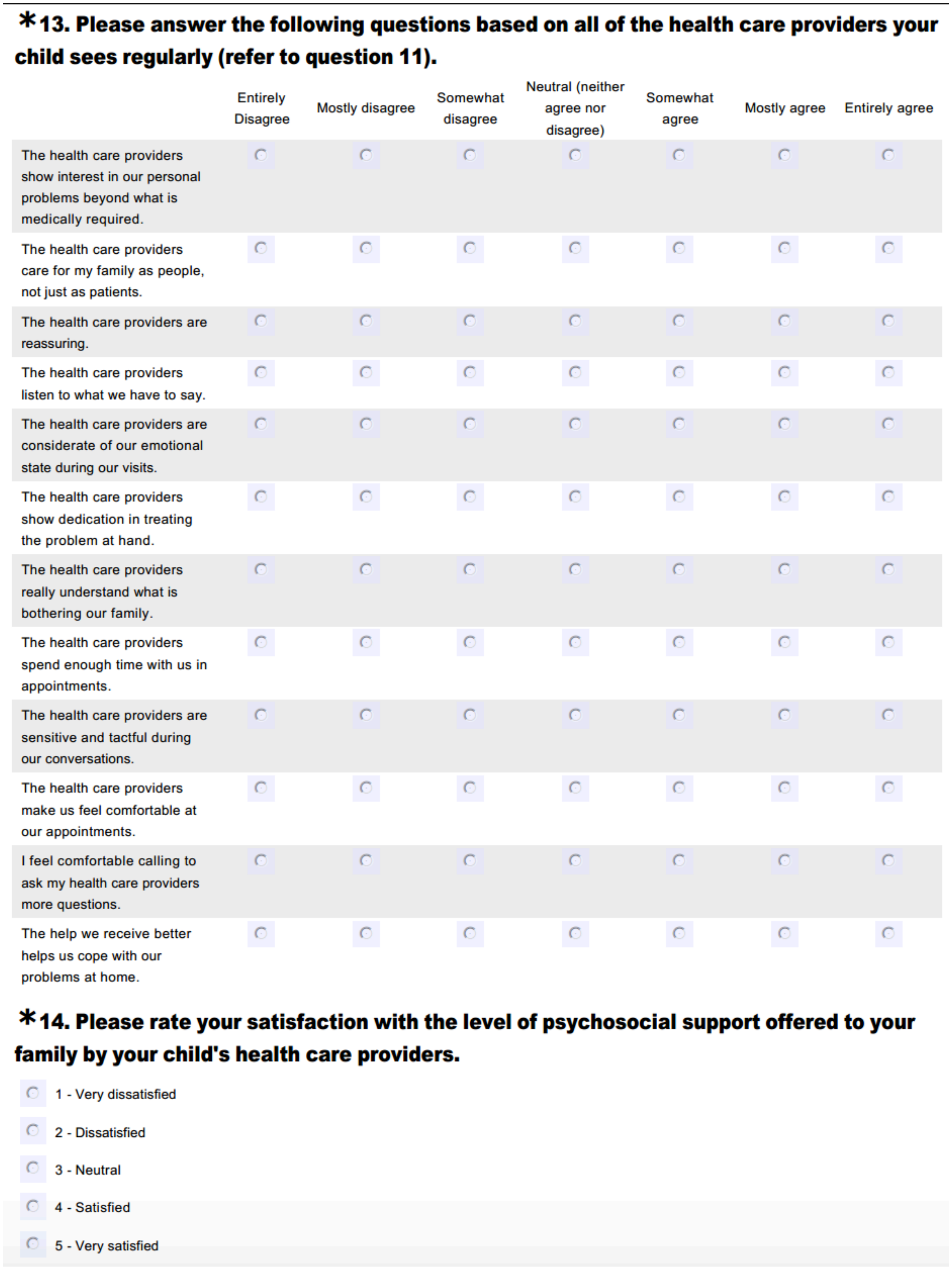
☐ Cardiology
☐ Developmental pediatrics
☐ Dermatology
☐ Endocrinology
☐ Genetics
☐ General pediatrics
☐ Neurology
☐ Nutrition
☐ Ophthalmology
☐ Orthopedics
☐ Otolaryngology
☐ Psychology
☐ Pulmonology
☐ Urology

☐ Please add any another specialists not listed above, if necessary.

***12. From the specialities you selected above, please list the top 4 health care providers, in order (1 being the most supportive), who you feel provide the most emotional/psychosocial support to your family.**

	Select a provider.
1	
2	
3	
4	

If you selected "Other," please specify.

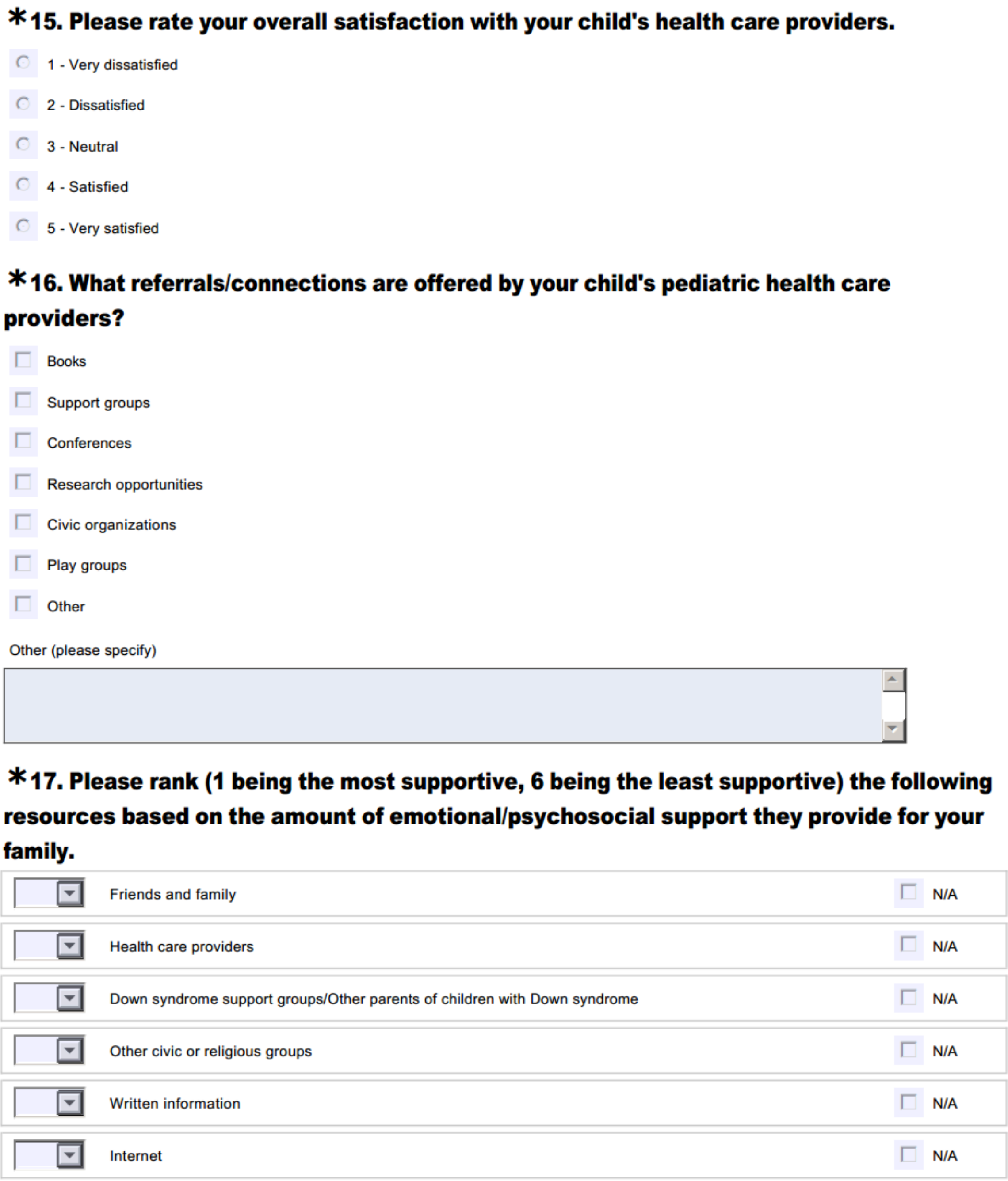


***13. Please answer the following questions based on all of the health care providers your child sees regularly (refer to question 11).**

	Entirely Disagree	Mostly disagree	Somewhat disagree	Neutral (neither agree nor disagree)	Somewhat agree	Mostly agree	Entirely agree
The health care providers show interest in our personal problems beyond what is medically required.	○	○	○	○	○	○	○
The health care providers care for my family as people, not just as patients.	○	○	○	○	○	○	○
The health care providers are reassuring.	○	○	○	○	○	○	○
The health care providers listen to what we have to say.	○	○	○	○	○	○	○
The health care providers are considerate of our emotional state during our visits.	○	○	○	○	○	○	○
The health care providers show dedication in treating the problem at hand.	○	○	○	○	○	○	○
The health care providers really understand what is bothering our family.	○	○	○	○	○	○	○
The health care providers spend enough time with us in appointments.	○	○	○	○	○	○	○
The health care providers are sensitive and tactful during our conversations.	○	○	○	○	○	○	○
The health care providers make us feel comfortable at our appointments.	○	○	○	○	○	○	○
I feel comfortable calling to ask my health care providers more questions.	○	○	○	○	○	○	○
The help we receive better helps us cope with our problems at home.	○	○	○	○	○	○	○

***14. Please rate your satisfaction with the level of psychosocial support offered to your family by your child's health care providers.**

○ 1 - Very dissatisfied
○ 2 - Dissatisfied
○ 3 - Neutral
○ 4 - Satisfied
○ 5 - Very satisfied



***15. Please rate your overall satisfaction with your child's health care providers.**

○ 1 - Very dissatisfied

○ 2 - Dissatisfied

○ 3 - Neutral

○ 4 - Satisfied

○ 5 - Very satisfied

***16. What referrals/connections are offered by your child's pediatric health care providers?**

☐ Books

☐ Support groups

☐ Conferences

☐ Research opportunities

☐ Civic organizations

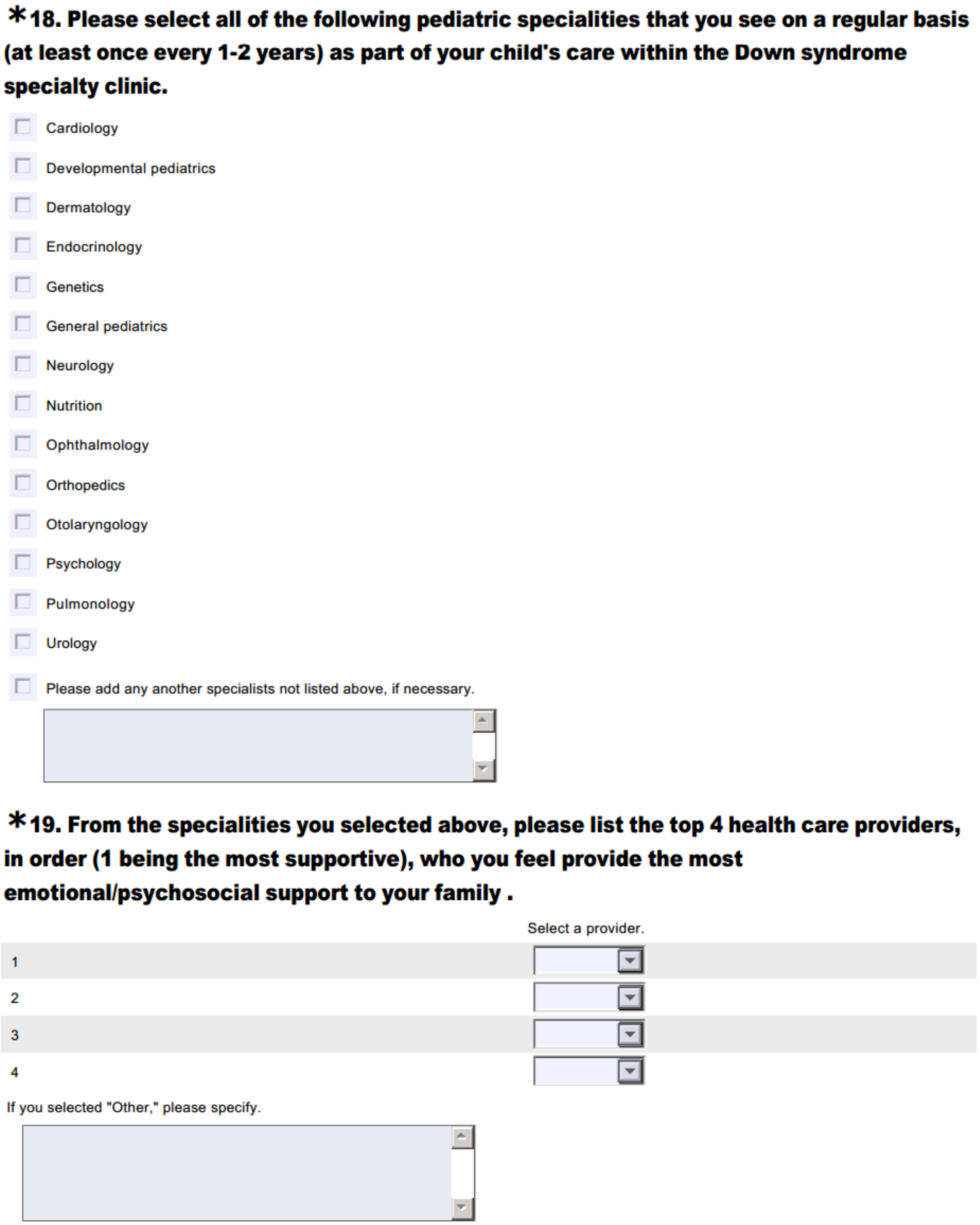
☐ Play groups

☐ Other

Other (please specify)

***17. Please rank (1 being the most supportive, 6 being the least supportive) the following resources based on the amount of emotional/psychosocial support they provide for your family.**

▾	Friends and family	☐ N/A
▾	Health care providers	☐ N/A
▾	Down syndrome support groups/Other parents of children with Down syndrome	☐ N/A
▾	Other civic or religious groups	☐ N/A
▾	Written information	☐ N/A
▾	Internet	☐ N/A



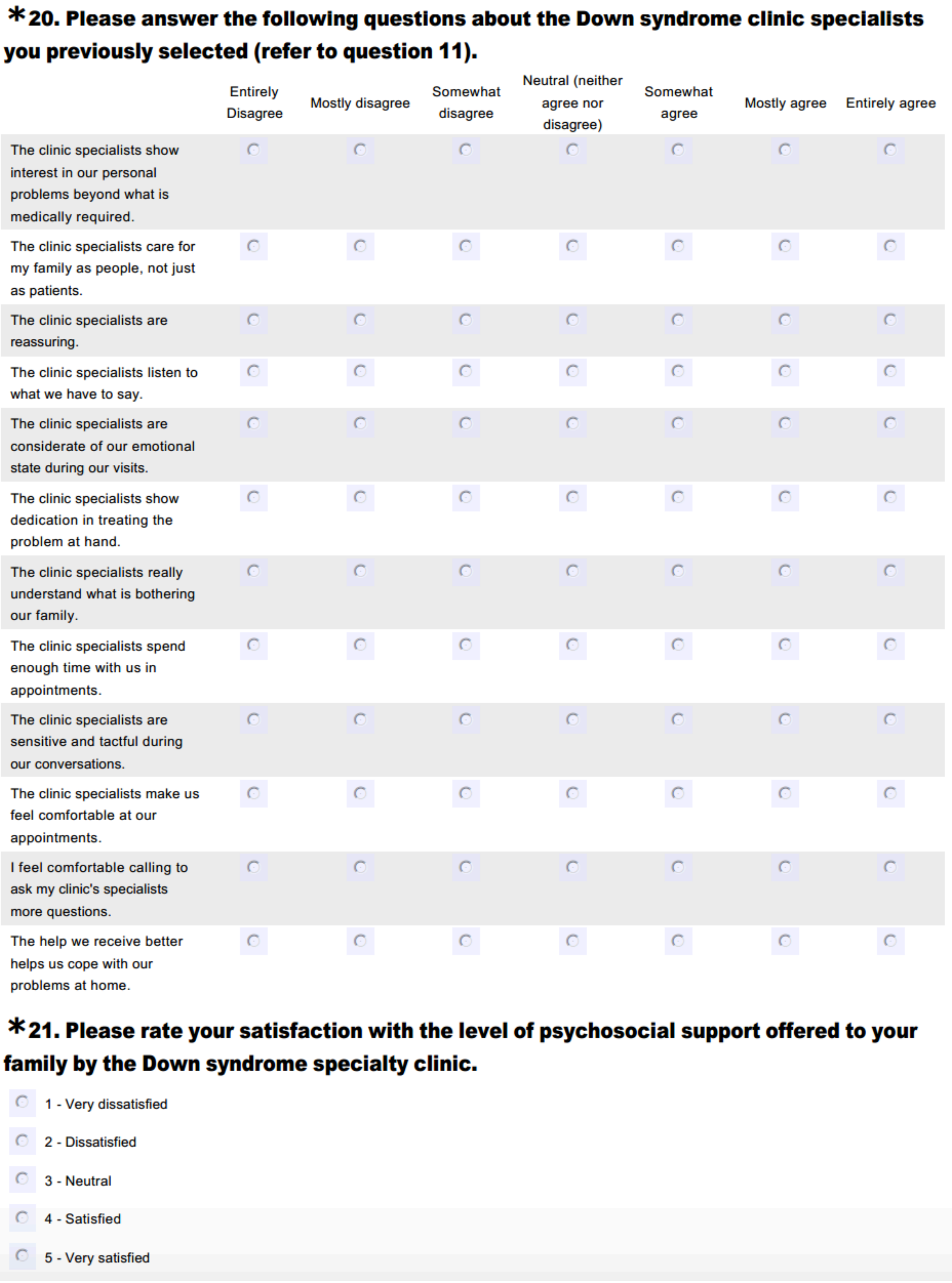
***18. Please select all of the following pediatric specialities that you see on a regular basis (at least once every 1-2 years) as part of your child's care within the Down syndrome specialty clinic.**

☐ Cardiology
☐ Developmental pediatrics
☐ Dermatology
☐ Endocrinology
☐ Genetics
☐ General pediatrics
☐ Neurology
☐ Nutrition
☐ Ophthalmology
☐ Orthopedics
☐ Otolaryngology
☐ Psychology
☐ Pulmonology
☐ Urology
☐ Please add any another specialists not listed above, if necessary.

***19. From the specialities you selected above, please list the top 4 health care providers, in order (1 being the most supportive), who you feel provide the most emotional/psychosocial support to your family .**

	Select a provider.
1	
2	
3	
4	

If you selected "Other," please specify.



***20. Please answer the following questions about the Down syndrome clinic specialists you previously selected (refer to question 11).**

	Entirely Disagree	Mostly disagree	Somewhat disagree	Neutral (neither agree nor disagree)	Somewhat agree	Mostly agree	Entirely agree
The clinic specialists show interest in our personal problems beyond what is medically required.	○	○	○	○	○	○	○
The clinic specialists care for my family as people, not just as patients.	○	○	○	○	○	○	○
The clinic specialists are reassuring.	○	○	○	○	○	○	○
The clinic specialists listen to what we have to say.	○	○	○	○	○	○	○
The clinic specialists are considerate of our emotional state during our visits.	○	○	○	○	○	○	○
The clinic specialists show dedication in treating the problem at hand.	○	○	○	○	○	○	○
The clinic specialists really understand what is bothering our family.	○	○	○	○	○	○	○
The clinic specialists spend enough time with us in appointments.	○	○	○	○	○	○	○
The clinic specialists are sensitive and tactful during our conversations.	○	○	○	○	○	○	○
The clinic specialists make us feel comfortable at our appointments.	○	○	○	○	○	○	○
I feel comfortable calling to ask my clinic's specialists more questions.	○	○	○	○	○	○	○
The help we receive better helps us cope with our problems at home.	○	○	○	○	○	○	○

***21. Please rate your satisfaction with the level of psychosocial support offered to your family by the Down syndrome specialty clinic.**

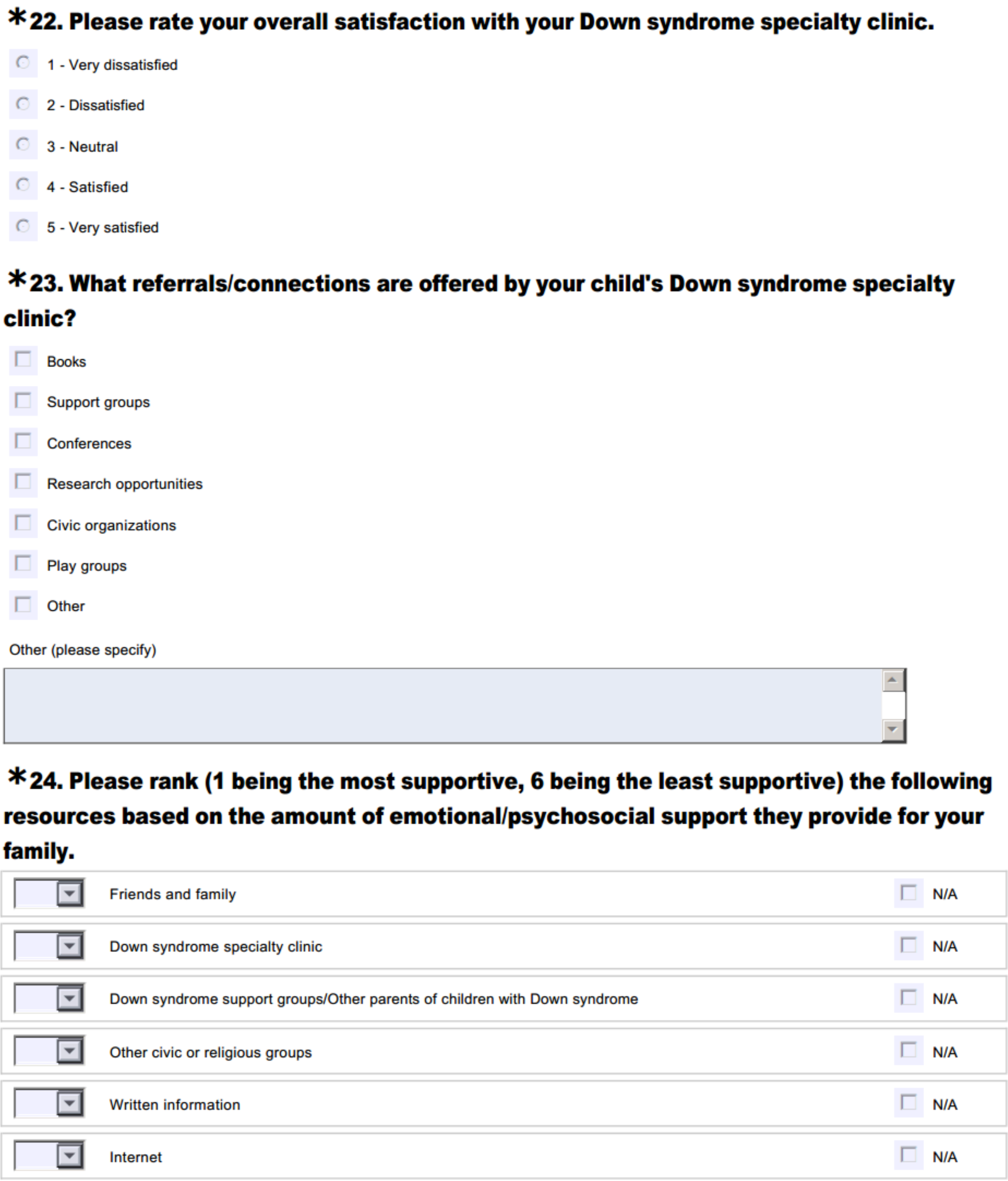
○ 1 - Very dissatisfied

○ 2 - Dissatisfied

○ 3 - Neutral

○ 4 - Satisfied

○ 5 - Very satisfied



***22. Please rate your overall satisfaction with your Down syndrome specialty clinic.**

○ 1 - Very dissatisfied

○ 2 - Dissatisfied

○ 3 - Neutral

○ 4 - Satisfied

○ 5 - Very satisfied

***23. What referrals/connections are offered by your child's Down syndrome specialty clinic?**

☐ Books

☐ Support groups

☐ Conferences

☐ Research opportunities

☐ Civic organizations

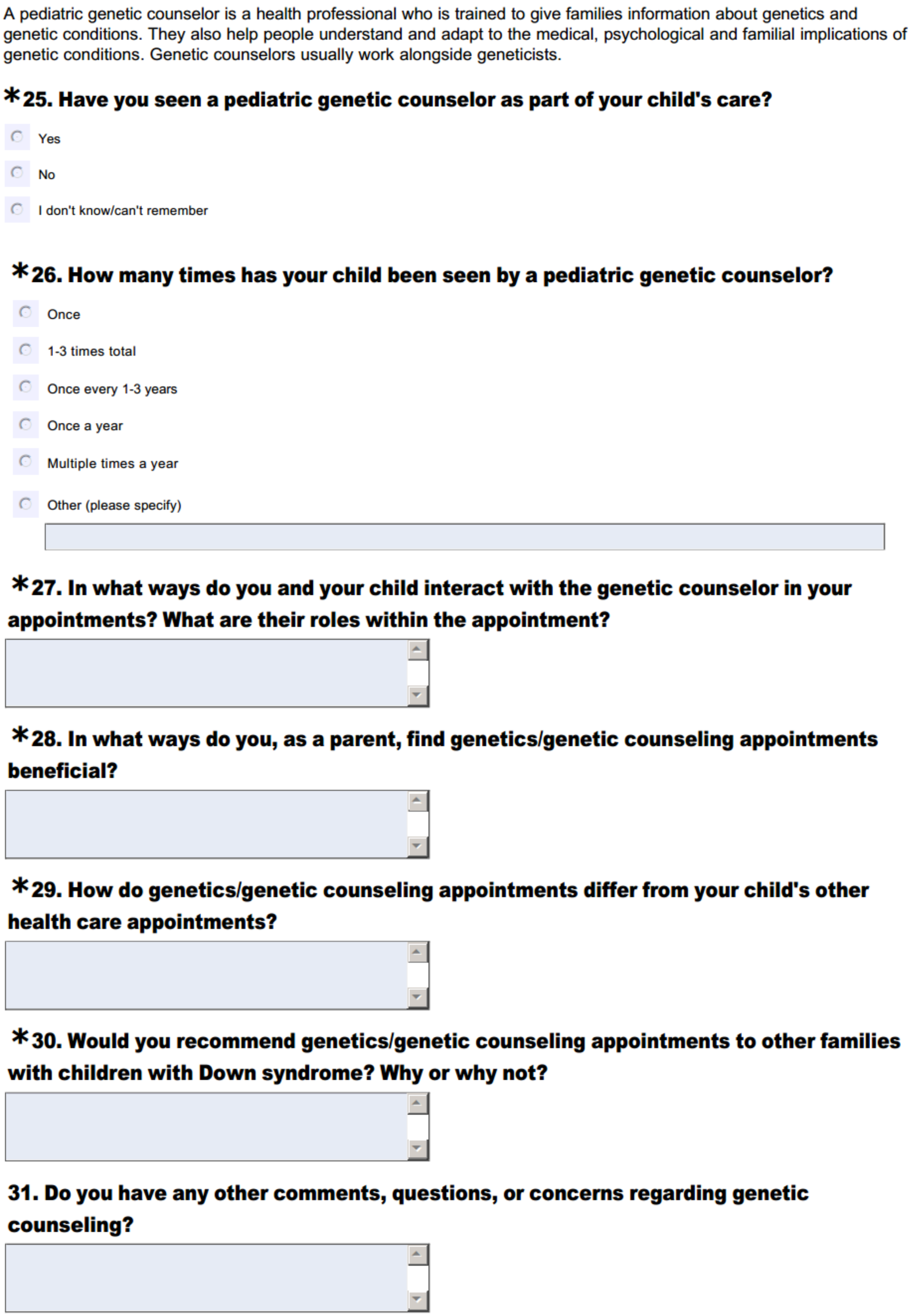
☐ Play groups

☐ Other

Other (please specify)

***24. Please rank (1 being the most supportive, 6 being the least supportive) the following resources based on the amount of emotional/psychosocial support they provide for your family.**

▾	Friends and family	☐ N/A
▾	Down syndrome specialty clinic	☐ N/A
▾	Down syndrome support groups/Other parents of children with Down syndrome	☐ N/A
▾	Other civic or religious groups	☐ N/A
▾	Written information	☐ N/A
▾	Internet	☐ N/A



A pediatric genetic counselor is a health professional who is trained to give families information about genetics and genetic conditions. They also help people understand and adapt to the medical, psychological and familial implications of genetic conditions. Genetic counselors usually work alongside geneticists.

***25. Have you seen a pediatric genetic counselor as part of your child's care?**

○ Yes

○ No

○ I don't know/can't remember

***26. How many times has your child been seen by a pediatric genetic counselor?**

○ Once

○ 1-3 times total

○ Once every 1-3 years

○ Once a year

○ Multiple times a year

○ Other (please specify)

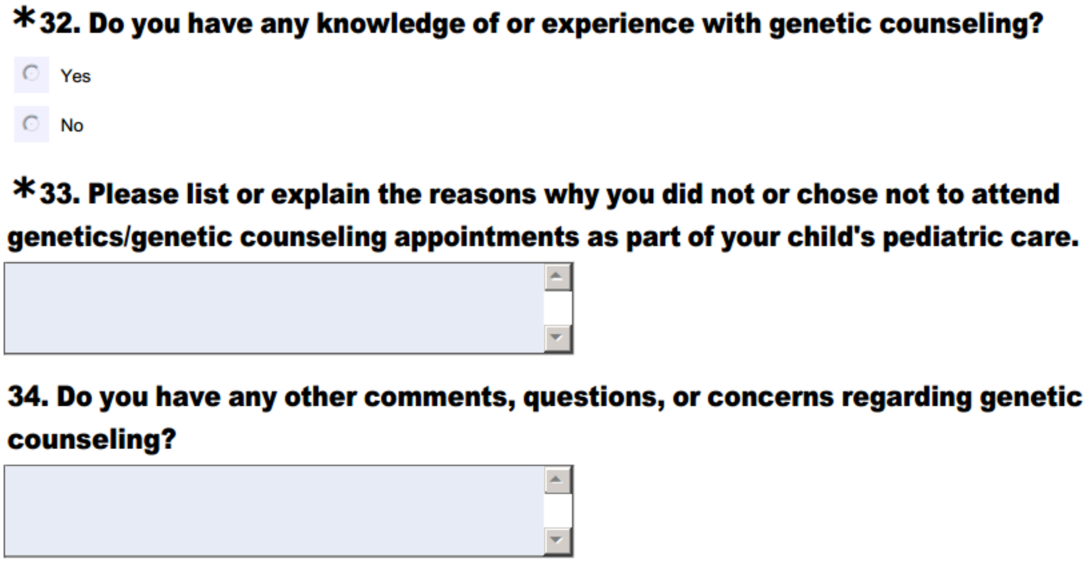
***27. In what ways do you and your child interact with the genetic counselor in your appointments? What are their roles within the appointment?**

***28. In what ways do you, as a parent, find genetics/genetic counseling appointments beneficial?**

***29. How do genetics/genetic counseling appointments differ from your child's other health care appointments?**

***30. Would you recommend genetics/genetic counseling appointments to other families with children with Down syndrome? Why or why not?**

31. Do you have any other comments, questions, or concerns regarding genetic counseling?



***32. Do you have any knowledge of or experience with genetic counseling?**

○ Yes

○ No

***33. Please list or explain the reasons why you did not or chose not to attend genetics/genetic counseling appointments as part of your child's pediatric care.**

34. Do you have any other comments, questions, or concerns regarding genetic counseling?

Appendix C. Satisfaction with Genetic Counseling Scale

Item Number	Item	Sub-scale
1	Did the doctor show interest in your personal problems beyond what is medically required?	Affective
2	Did you consider turning to another doctor regarding your problem?	
3	Did the doctor explain your condition to you clearly?	Instrumental
4	Did the doctor meet your expectations of him?	
5	Do you think the doctor cares for you as a person?	Affective
6	How comfortable would you feel to call the doctor to ask another question?	Affective
7	Did the doctor reassure you?	Instrumental
8	Did the doctor listen to what you had to say?	Instrumental
9	Was the doctor considerate of your emotional state during the meeting?	Affective
10	How satisfied are you with the way in which information was transmitted to you?	Instrumental
11	Did the doctor show enough dedication in treating your problem?	Affective
12	How would you rate the level of service that you received?	
13	Did the doctor understand what was really bothering you?	Affective
14	Do you think that you could get more considerate care from a private doctor?	Affective
15	Did the doctor make you feel you are "in good hands"?	Instrumental
16	Did the doctor make you feel that he knows how to handle problems like yours?	Instrumental
17	Did the doctor give you enough of his time?	Affective
18	Was the doctor sensitive and tactful during your conversation?	Affective
19	Did the doctor give you the necessary treatment?	Instrumental
20	Do you think the doctor is an expert in the field in which you need help?	Instrumental
21	Can the counseling that you received help you cope better with your problem?	Instrumental
22	Did the doctor lessen your worries?	Instrumental
23	How did you rate the length of time you waited since you first contacted the clinic and until your visit?	Procedural
24	How satisfied are you with the information you got in counseling?	

25	How comfortable did you feel to talk about yourself during the counseling session?	Affective
26	How satisfied were you with the administrative procedures required for your visit?	Procedural
27	How do you rate the length of time you waited since your arrival at the clinic and until you entered the doctor’s office?	Procedural
28	If an acquaintance of yours needed similar help, would you recommend this clinic to him/her?	
29	Do you think you could get better treatment from a private doctor?	Instrumental
30	How satisfied are you with the treatment you got from the medical staff, besides the doctor – the nurse, secretary, etc.?	Procedural
31	Do you think that the counseling was given in the appropriate setting for the sort of problem you had?	Instrumental
32	In summary, how would you rate your satisfaction with the counseling?	

Appendix D. Chi Square Results

Table D.1 Parent Demographic χ^2 Calculations

Cross-tabulations	***p***
Group * Gender	.906
Group * Age	.518
Group * Ethnicity	.086
Group * Education	.047
Group * Marital Status	.113

Table D.2 Child Demographic χ^2 Calculations

Cross-tabulations	***p***
Group * Age	.304
Group * Gender	.910
Group * Diagnosis	.403