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Revisiting the Essential Informational Needs of Parents Receiving a Diagnosis of Down Syndrome

Margaret Jean Wilkes

REVISITING THE ESSENTIAL INFORMATIONAL NEEDS OF PARENTS RECEIVING A DIAGNOSIS OF DOWN SYNDROME

by

Margaret Jean Wilkes

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Accepted by:

Richard Ferrante, Director of Thesis

Kathryn Berrier, Reader

Stephanie Meredith, Reader

Campbell Brasington, Reader

Cheryl L. Addy, Vice Provost and Dean of the Graduate School

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ABSTRACT

Down syndrome is a condition characterized by varying degrees of intellectual disability (ID), distinctive facial appearance, and congenital anomalies that results from the presence of a third 21st chromosome. Down syndrome is the most common chromosomal condition, affecting approximately 12.6 per 10,000 live births in the United States, making it imperative that we determine which information is most essential to impart to parents when first presenting the diagnosis. The aim of the present study is to reassess the informational needs of parents during the presentation of a Down syndrome diagnosis. In 2009, data were collected to define the essential information necessary to give a balanced description of Down syndrome. In the present, replicative study, parents and genetic counselors completed online, anonymous surveys used in the 2009 study to reassess which informational items are most essential to present during the initial diagnosis experience. Both groups rated the importance of 100 aspects of Down syndrome. Results identify 30 essential items, of which 19 were highly ranked by all groups. These results were compared to the findings of the 2009 study to identify any changes in perceived importance of different aspects of Down syndrome for both parents and genetic counselors. Comparisons between 2009 and 2019 data reveal a parental emphasis on both the medical and social facets of Down syndrome, whereas genetic counselors generally placed greater weight on the medical facets. Parents also demonstrated a preference for inclusion over specialized programs for their children. Both parents and genetic counselors highlighted the importance of informational

resources and referrals, with parents especially showing the need for a wide array of resource media. Findings of our study reinforce the recommendations of the existing practice guidelines while highlighting both persistent and novel discrepancies between parents and providers.

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LIST OF ABBREVIATIONS

AAP ..... American Academy of Pediatrics

ADA ..... Americans with Disabilities Act

AFP ..... Alpha(α)-Fetoprotein

CVS ..... Chorionic Villus Sampling

DD ..... Developmental Delay

DS ..... Down syndrome

GC(s) ..... Genetic Counselor(s)

ID ..... Intellectual Disability

IDEA ..... Individuals with Disabilities Education Act

MFM ..... Maternal-Fetal Medicine Subspecialist

MS ..... Master of Science degree

NDSC ..... National Down Syndrome Congress

NIPS/T ..... Noninvasive Prenatal Screening/Testing

NSGC ..... National Society of Genetic Counselors

Ob-Gyn ..... Obstetrician-Gynecologist

CHAPTER 1: BACKGROUND

Down syndrome, referred to as Down's syndrome in the United Kingdom, is a condition characterized by varying degrees of intellectual disability (ID), distinctive facial appearance, and congenital anomalies. The condition results from a gain in genetic material due to the presence of three copies of chromosome 21 (Sheets, 2009). The association of a supernumerary chromosome 21 with the features of Down syndrome was first discovered in 1959 (LeJeune et al., 1959). Down syndrome is the most common chromosomal condition, affecting approximately 12.6 per 10,000 live births in the United States (de Graaf et al., 2015). Though one of the most frequent causes of intellectual disability, the degree of cognitive disability in most individuals with Down syndrome is in the mild (IQ of 50-70) to moderate (IQ of 35-50) range (Duffner, 2011). Common features seen in Down syndrome include a flat facial profile, upward-slanting palpebral fissures, hypotonia, hyperflexibility, and a single palmar crease, among others. Specific medical complications can also arise in association with the condition, including hearing loss, vision problems, increased susceptibility to infection, and early-onset Alzheimer's disease (Antonarakis et al., 2020).

In approximately 90-95% of cases, the extra 21st chromosome that causes Down syndrome is derived from a nondisjunction event during cell division. The majority of these nondisjunction events occur maternally in the ovum, however approximately 5% of these cases are due to paternal nondisjunction. This form of the condition is also referred to as trisomy 21, named for the presence of three copies of chromosome 21 rather than

the typical two copies. Down syndrome can also be caused by a translocation in around 2-4% of cases (Coppedè, 2016). A critical portion of chromosome 21, or the full chromosome in some cases, becomes attached to another chromosome, effectively resulting in excess genetic material which produces the phenotype. Clinically, there are no distinguishable differences between trisomy 21 and translocation Down syndrome (NDSS, 2019). The final form of the condition, mosaic Down syndrome, makes up approximately 2-4% of all cases. Individuals with this type have some cells with trisomy 21, and other cells with the typical two copies. Mosaicism occurs when there is an error in cell division after the unification of the ovum and sperm cells, whereas in most cases of trisomy 21, a nondisjunction event occurs in the ovum prior to fertilization. A wide range of phenotypic severity can be seen in individuals with mosaic Down syndrome depending on the number of affected cells present in the body.

Although Down syndrome occurs across all demographics, different factors influence the chance a pregnancy will be affected with Down syndrome. Since approximately 95% of cases of Down syndrome can be attributed to a nondisjunction event, with 95% of these nondisjunction events occurring in the maternal oocyte, maternal age is a factor influencing the development of aneuploidies. Advanced maternal age, defined by the American Congress of Obstetricians and Gynecologists (ACOG) as being of age 35 or older at the time of delivery, is a well-established risk factor for chromosome abnormalities including trisomy 21 (Ryu, 2013). Females are born with the total number of oocytes they will have in their lifetime, and therefore as the oocytes age and develop into ova, there is a higher chance for meiotic errors to occur (Antonarakis et al., 2020).

Certain environmental factors are known to play a role in triggering the nondisjunction events that lead to trisomy 21. Known environmental factors include tobacco use, folic acid supplementation, and oral contraceptive use, among others. It has also been proposed that transgenerational factors, including BPA exposure, may have atypical effects on both sperm and oocyte development that could lead to aneuploidy. Maternal social factors, including exposure to solvents through work, have also been linked to aneuploidy (Antonarakis et al., 2020).

A wide range of variability in physical features and medical comorbidities can be seen in Down syndrome. Approximately 50% of individuals who have Down syndrome are born with one or more birth differences. Congenital heart defects (CHDs), most commonly atrioventricular septal defects, occur in about half of people with the condition and can require surgical intervention. Individuals who have Down syndrome are typically of short stature and present with hypotonia. Common craniofacial features seen with Down syndrome include a flat facial profile and nasal bridge, upslanting palpebral fissures, a small mouth, and small, low-set ears. Individuals with Down syndrome can have a higher susceptibility to certain health conditions, including obstructive sleep apnea, hypothyroidism, autoimmune disease, epilepsy, leukemia, and early-onset Alzheimer's disease. Solid tumors are less common in individuals with Down syndrome than the general population, lowering the risk of certain cancers (Antonarakis et al., 2020).

Outcomes for individuals living with Down syndrome have improved dramatically in the last several decades. Improved outcomes can be largely attributed to better understanding of the condition, advancements in medical care, increased amounts

of supports and services, and shifts in the sociocultural landscape toward inclusivity. In 1970, parents initially receiving a diagnosis of Down syndrome could have been told that over 50% of babies with the condition die within the first year, the child's life expectancy would be around 20 years of age, and that their child would have no right to a public education or employment (Saul & Meredith, 2016). Contemporary statistics show that over 93% of infants with Down syndrome survive past the first year, the life expectancy is around 60 years of age, and children have rights to early intervention services, a public education in the least restrictive environment among typically developing peers under the Individuals with Disabilities Education Act (IDEA), and a right to employment and equal access under the Americans with Disabilities Act (ADA) (Kucik et al., 2012). There are also increased opportunities available to individuals with Down syndrome as well as the disability community as a whole. For instance, there are now over 200 college programs available for students who have intellectual disabilities (Saul & Meredith, 2016; Think College, 2019).

1.1 INITIAL INFORMATION ABOUT PRENATAL TESTING FOR DOWN SYNDROME

As the global population continues to grow, the prevalence of Down syndrome is also increasing. According to the Centers for Disease Control and Prevention (CDC), the number of babies with Down syndrome born between 1979 and 2003 increased by 30%, thought to be due to postponed motherhood (de Graaf, Buckley, & Skotko, 2015). As more pregnancies are affected with Down syndrome, it is imperative that we determine which information is most essential according to the parents and families impacted.

While many cases of Down syndrome are diagnosed postnatally, patients can pursue both screening tests and diagnostic tests prenatally. Before the 1980s, risk of aneuploidy was based solely on advanced maternal age, which alone accurately predicted only 25-30% of fetal aneuploidy. Amniocentesis, a diagnostic test available after 15 weeks gestation, was offered to women of advanced maternal age to formerly diagnose fetal aneuploidy including Down syndrome beginning in the 1970s. In 1984, Merkatz et al. discovered a link between low maternal serum α-fetoprotein (MSAFP) in the second trimester and fetal aneuploidy, particularly trisomy 18. Around this time, measuring MSAFP levels during pregnancy was proposed and implemented as a prenatal screening option for Down syndrome in the second trimester. Other analytes were found to have increased or decreased levels in pregnancies affected with fetal aneuploidy, including free β-human chorionic gonadotropin (β-hCG), inhibin A (DIA), and unconjugated estriol (uE_3). The 'triple screen,' including AFP, β-hCG, and uE_3 used in conjunction with maternal age, debuted in 1988 and was available during the second trimester. This screen increased the detection rate for Down syndrome from ~30% via maternal age to ~60% with a false positive rate of 5% (Russo & Blakemore, 2014). Inhibin A was incorporated into the screen, establishing the 'quadruple screen,' bringing the detection rate of the screen plus maternal age to 75%.

Since the goal of prenatal screening for Down syndrome was to detect an increased chance as early as possible, efforts were made to develop first trimester screening options. This is largely due to the availability of chorionic villus sampling (CVS), a diagnostic test typically performed from 10-13 weeks gestation, beginning in the 1980s. In 1992, an initial study demonstrated the utility of using free β-hCG to screen for both

trisomy 21 and trisomy 18 in the first trimester of pregnancy (Spencer et al., 1992). Further investigation of using free β-hCG as a screening analyte from 9-13 weeks gestation found that it was significantly elevated in pregnancies affected with trisomy 21, which confirmed the utility of the analyte for first trimester screening (Macri et al., 1993). Alongside free β-hCG, pregnancy-associated plasma protein A (PAPP-A) was found to be low in pregnancies affected with Down syndrome from 8-12 weeks. Wald et al. (1992) concluded that PAPP-A too was a measurable screening analyte for trisomy 21 in the first trimester of pregnancy, and the two markers have since been analyzed together in traditional first-trimester maternal serum screening.

Nicolaides et al. (1992) were among the first to hypothesize an association between nuchal edema and chromosomal conditions, including Down syndrome. To test this hypothesis, 827 pregnancies were screened to assess the amount of fluid behind the necks of the fetuses from 10-14 weeks gestation. The incidence of chromosomal conditions when the thickness of translucency between the cervical spine and the skin was 3-8mm was 35%, whereas the rate of chromosomal conditions was 1% when the thickness measurements were smaller (Nicolaides et al., 1992; Russo & Blakemore, 2014). The study concluded that there is an increased chance of chromosomal abnormalities with an increased translucency measurement and introduced the term 'nuchal translucency' into prenatal screening practices. The nuchal translucency, or NT, was used in combination with biochemical markers in the first trimester starting in the mid-1990s. This combined first trimester screening increased the detection rate for Down syndrome specifically to 85%, with a 5% false positive rate. The addition of maternal age as a factor alongside the biochemical markers and NT measurement brought

the detection for Down syndrome to 87% in the first trimester, and the detection for Trisomy 18 to 76% (Orlandi et al., 1997; Russo & Blakemore, 2014).

Noninvasive Prenatal Screening (NIPS), also known as Noninvasive Prenatal Testing (NIPT), was first introduced into clinical practice in 2011. This screening technique involves isolating cell-free DNA from the placenta in the maternal serum to determine if there is an increased chance for aneuploidy. The screen is most frequently used to detect if there is an increased chance for common chromosomal aneuploidies, such as Down syndrome, Trisomy 18, and Trisomy 13, as well as common sex chromosome aneuploidies, including Turner syndrome (Monosomy X) and Klinefelter syndrome (47,XXY) (Allyse et al., 2015). Though not a diagnostic test, NIPS boasts both a sensitivity and specificity of >99% for Down syndrome, with the false positive rate being as low as 1-3% (Allyse et al., 2015). The screen has high detection rates for different aneuploidies in comparison to other tests, albeit not as high as for Down syndrome; other detection rates include 97-99% for Trisomy 18, 87-99% for Trisomy 13, and 92-95% for Turner syndrome. Aside from the benefits of having a high sensitivity, high specificity, and low false positive rate, this screen can be performed with high accuracy without the risk of miscarriage associated with diagnostic testing procedures (Kellogg et al., 2014).

The advent of newer, more accurate prenatal screening tests for Down syndrome and other aneuploidies was met with mixed feelings from the Down syndrome community. Both parents and providers have recognized positive aspects of NIPS, including the accuracy, safety, and ability to test earlier in the pregnancy (van Schendel et al., 2017). Though not a diagnostic test, NIPS has a high detection rate for Down

syndrome and other aneuploidies which may reduce the number of invasive procedures done to confirm the presence of the condition, which is viewed as an advantage by parents (van Schendel et al., 2017). The early screening enabled by NIPS may also lead to increased prenatal diagnoses of Down syndrome, whereas historically the majority of cases were diagnosed postnatally (Skotko, 2009a). This changes the traditional diagnostic landscape, and gives women more time earlier in the pregnancy to consider various reproductive options including continuing the pregnancy, terminating the pregnancy, or adoption.

The same factors that were viewed by parents to be advantages of NIPS were also viewed as disadvantages in different lights. When NIPS was first clinically implemented, there were questions concerning the effects the screen would have on reproductive decision-making. Given that NIPS can be drawn at no risk of miscarriage to the pregnancy, it was postulated that Down syndrome could be detected more often prenatally, and potentially lead to a disproportionate number of terminations of pregnancies affected with Down syndrome (Kellogg et al., 2015; Skotko, 2009c). If less babies are born with Down syndrome, the supports and services available to individuals with the condition could decrease (How et al., 2019). Parents also fear that the normalization of such a prenatal screen would increase discrimination and stigmatization against individuals who have Down syndrome.

When Kellogg et al. (2015) surveyed 73 mothers of children who have Down syndrome regarding NIPS, participants highlighted both positive and negative aspects of the screening. Approximately 50% of those surveyed stated they would opt for NIPS in a subsequent pregnancy, and 67% felt as though the screening test should be available to

all pregnant women to promote autonomy and aid in preparation before birth. Despite these positive sentiments, the majority of these participants (88%) also stated they believed terminations of pregnancies affected with Down syndrome would increase due to this test, and that subsequently, access to services would decrease (64%) (Kellogg et al., 2015). Both parents and medical professionals recognize the potential benefits and harmful effects of NIPS, and its implementation is changing how and when individuals are being diagnosed with Down syndrome.

1.2 THE GENETIC COUNSELING PROFESSION

The first cohort of genetic counselors graduated with master's degrees from Sarah Lawrence College in Bronxville, New York in 1971 (NSGC, 2020). In 1979, the National Society of Genetic Counselors (NSGC) was established to support, promote, and advance the profession of genetic counseling (Sheets et al., 2009). In the early 2000s, since genetic counseling was expanding beyond traditional roles and into other realms of medicine, such as public health, research, other subspecialties, and the laboratory, a task force was assembled to redefine the role of a genetic counselor (Resta et al., 2006). This NSGC task force defined genetic counseling as:

> …"the process of helping people understand and adapt to the medical, psychological and familial implications 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 (Resta et al., 2006, Sheets et al., 2009)."

The American Society of Human Genetics (ASHG) proposed another widely-referenced definition of genetic counseling during the 1975 Ad Hoc Committee on Genetic Counseling, which states that genetic counseling is:

… "a communication process which deals with the human problems associated with the occurrence, or the risk of occurrence, of a genetic disorder in a family. This process involves an attempt by one or more appropriately trained persons to help the individual or family to

1. Comprehend the medical facts, including the diagnosis, prognosis, and management,
2. Appreciate the way heredity contributes to the disorder, including recurrence risk to relatives,
3. Understand the alternatives for dealing with recurrence risk,
4. Choose the course of action which seems to them appropriate in view of their risk, their family goals, and their ethical and religious standards, and to act in accordance with that decision, and
5. To make the best possible adjustment to the disorder in an affected family member and/or to the risk of recurrence of that disorder (ASHG, 1975; Sheets et al., 2009).

The Genetic Alliance (2009) further defines the role of a genetic counselor as "a central resource of information about genetic disorders for other healthcare professionals, patients, and the general public."

The NSGC established a Code of Ethics in 1992 to affirm the ethical responsibilities of genetic counselors. This Code of Ethics was recently revised and adopted by the organization in April 2017 following a majority vote of the full membership. The values of the Code of Ethics stem from the principles of autonomy, beneficence, nonmaleficence, and justice (NSGC, 2017). Autonomy is a crucial component of informed consent and decision-making in the genetic counseling process (Sheets et al., 2009). Historically, autonomy in the context of genetic counseling was viewed as non-interference in patient decision-making, or 'nondirectiveness' (Jamal et al., 2019; Stern, 2009). White (1997) proposed that patient autonomy in genetic counseling had been viewed as a 'negative right:' noninterference in decision-making, and calls for patient autonomy to be viewed as a 'positive right:' a maximally enhanced decision-making process. This idea promoted thoughtful dialogue between counselor and patient, making the deliberative process a central aspect of the genetic counseling session (White, 1997). These early calls to shift away from the 'nondirectiveness' paradigm of genetic counseling have been echoed in recent literature. Jamal et al. (2019) call for genetic counselors to emphasize a commitment to the ethical principles of beneficence and non-maleficence, and recognize that patient autonomy can be respected and promoted in different ways depending on the situation.

The profession of genetic counseling has experienced immense growth and change. Genetic counselors must keep up with the rapid development of new genomic technologies and disseminate complicated information to patients with empathy. Medical providers with this unique skillset are in high demand, and genetic counselors have expanded from their traditional roles into other subspecialties, industry, and research.

Biesecker (2019) argues that despite the ever-evolving nature of the field, the core tenets of genetic counseling remain unchanged. The therapeutic relationship between counselor and patient is central to the profession (Biesecker, 2019). The counselor's role is to elicit the patient's psychosocial and educational needs, provide relevant information in an accessible and empathetic way, and to facilitate informed decision-making. To fully encompass and define the unique role of the genetic counselor, the reciprocal engagement model (REM) was developed to highlight the tenets of practice, which include: 1. Genetic information is key, 2. The relationship is integral, 3. Patient autonomy must be supported, 4. Patients are resilient, and 5. Patient emotions make a difference (Veach et al., 2007). These tenets of practice are still relevant today, and guide the profession of genetic counseling (Biesecker, 2019).

1.3 RECOMMENDATIONS FOR INFORMING THE PARENTS

Recommendations to providers about the disclosure of a genetic diagnosis include providing accurate, up-to-date information in a balanced manner (Hippman et al., 2012; Saul & Meredith, 2016; Sheets et al., 2011). Providers are also advised to share information not only about the medical side of Down syndrome, but also about life outcomes and social supports (Saul & Meredith, 2016). The news should be communicated to parents initially by a medical professional who is knowledgeable about Down syndrome (Skotko et al., 2009b). Parents generally prefer being told about the Down syndrome diagnosis as soon as it is confirmed or even suspected, and they prefer that the news be disclosed to them in-person, in the presence of partners or other support, and in a private place (Skotko et al., 2009b). Parents prefer to be given written information about the condition along with diagnostic information (Saul & Meredith,

2016; Skotko et al., 2009b). It is also recommended that providers give parents information about community resources and services, as well as the contact information of other families raising a child with Down syndrome (Saul & Meredith, 2016; Skotko et al., 2009a; Skotko et al., 2009b). The social side of the condition, including positive aspects and personal stories depicting the potential for individuals with Down syndrome in modern society, should also be shared with new parents (Skotko et al., 2009a).

1.4 HISTORY OF PARENTAL DISSATISFACTION

Down syndrome can be diagnosed both prenatally and postnatally. Historically, the majority of cases of Down syndrome were diagnosed postnatally, however the advent of new prenatal screening options has made detection in utero more and more common. Regardless of diagnosis timeline, there is a well-established history of parental dissatisfaction in receiving a diagnosis of Down syndrome for their child. An early report determined that nearly two-thirds of parents were dissatisfied with how the diagnosis was presented, claiming physician descriptions were brief, inadequate, and confusing (Quine & Pahl, 1987; Sheets, 2009). In a study conducted by Skotko (2005) surveying mothers of children with Down syndrome about their postnatal diagnosis experience in both the United States and Spain, the majority of both groups reported feelings of fear and anxiety upon receiving the diagnosis, especially after physicians mentioned little to no positive aspects of Down syndrome. Mothers also noted that they were aware of the "silence" following the birth of their child, and that the hesitancy expressed by physicians to disclose the diagnosis only deepened feelings of fear and anxiety. At times, the physicians used words insinuating that the birth of a child with Down syndrome was solely a negative event, or even used derogatory or offensive

language during disclosure. This cohort felt as though they were not given up-to-date materials or resources, and that referrals to other parents in similar situations were sorely lacking (Skotko, 2005). This study found that it was rare that the disclosure of a Down syndrome diagnosis was viewed as a positive experience by the patients.

1.5 THE NSGC DOWN SYNDROME PRACTICE GUIDELINES

The present study is replicating research conducted in 2009 that aimed to identify the essential informational items that should be imparted upon parents during the initial diagnosis of Down syndrome. Data from that study were utilized to create the "Practice Guidelines for Communicating a Prenatal or Postnatal Diagnosis of Down Syndrome: Recommendations of the National Society of Genetic Counselors" (Sheets et al., 2011). The goal of the practice guidelines is to provide genetic counselors and other medical professionals information on how to disclose a Down syndrome diagnosis using balanced, up-to-date information while conveying support and respect (Sheets et al., 2011). The guidelines walk through the etiology of Down syndrome, including a clinical description and associated management, the prevalence, the genetics of the condition, recurrence risk, and diagnostic testing options. Recommendations for delivering a diagnosis include informing parents of the diagnosis as soon as possible, in person and in a private setting, and by a knowledgeable medical provider. Neutral, person-centric language should be used in providing a description in an empathetic manner that validates the parents' feelings. Balanced information should be given, including the positive aspects and challenges associated with Down syndrome. Informational resources are key to provide during the initial discussion of the diagnosis, including information on local and national support groups, printed material or fact sheets, books,

and the contacts of families raising a child with Down syndrome. Essential informational content to cover with families include both medical and social facets of Down syndrome.

1.6 RATIONALE

Down syndrome remains the most common chromosomal disorder, and the frequency of the condition continues to increase. According to the Centers for Disease Control and Prevention (CDC), the number of babies born between 1979 and 2003 increased by 30%. The proportion of live births has been on the rise since the 1980s, largely due to postponed motherhood (de Graaf, Buckley, & Skotko, 2015). As more pregnancies are affected with Down syndrome, it is imperative that we revisit which information is most essential according to the parents and families impacted. Historically, parental dissatisfaction with their Down syndrome diagnosis experience is well-established (Cunningham, Morgan, & McGucken, 1984). Sheets et al. (2009) explored how to present a diagnosis in a balanced manner in an effort to increase parental satisfaction in the future. The present study could broaden our understanding of the information most relevant to those receiving a Down syndrome diagnosis at this time.

1.7 PURPOSE

The aim of this study is to reassess the informational needs of parents during the presentation of a Down syndrome diagnosis. In 2009, data were collected to define the essential information necessary to give a balanced description of Down syndrome (Sheets, 2009). The manner in which a diagnosis is presented has a lasting impact on families, and the informational needs of parents have likely changed in the years since the previous study was conducted. As previous data were utilized in the preparation of the National Society of Genetic Counselors (NSGC) Down Syndrome Practice Guidelines

(Sheets et al. 2011a), any shift in informational preferences over the last several years may suggest a need for an update or revision.

CHAPTER 2: REVISITING THE ESSENTIAL INFORMATIONAL NEEDS OF PARENTS RECEIVING A DIAGNOSIS OF DOWN SYNDROME

2.1 ABSTRACT

Down syndrome is a condition characterized by varying degrees of intellectual disability (ID), distinctive facial appearance, and congenital anomalies that results from the presence of a third 21st chromosome. Down syndrome is the most common chromosomal condition, affecting approximately 12.6 per 10,000 live births in the United States, making it imperative that we determine which information is most essential to impart to parents when first presenting the diagnosis. The aim of the present study is to reassess the informational needs of parents during the presentation of a Down syndrome diagnosis. In 2009, data were collected to define the essential information necessary to give a balanced description of Down syndrome. In the present, replicative study, parents and genetic counselors completed online, anonymous surveys used in the 2009 study to reassess which informational items are most essential to present during the initial diagnosis experience. Both groups rated the importance of 100 aspects of Down syndrome. Results identify 30 essential items, of which 19 were highly ranked by all groups. These results were compared to the findings of the 2009 study to identify any changes in perceived importance of different aspects of Down syndrome for both parents and genetic counselors. Comparisons between 2009 and 2019 data reveal a parental emphasis on both the medical and social facets of Down syndrome, whereas genetic counselors generally placed greater weight on the medical facets. Parents also

demonstrated a preference for inclusion over specialized programs for their children. Both parents and genetic counselors highlighted the importance of informational resources and referrals, with parents especially showing the need for a wide array of resource media. Findings of our study reinforce the recommendations of the existing practice guidelines while highlighting both persistent and novel discrepancies between parents and providers.

2.2 INTRODUCTION

Down syndrome is a genetic condition characterized by varying degrees of intellectual disability (ID), distinctive facial appearance, and congenital anomalies. The presence of a third copy of chromosome 21 results in the phenotype. Down syndrome is the most common chromosomal condition, affected approximately 12.6 per 10,000 live births in the United States, and is the most common genetic cause of intellectual disability (de Graaf et al., 2015; Mégarbané et al., 2013). The degree of cognitive impairment is typically mild to moderate (Duffner, 2011). Individuals who have Down syndrome can exhibit a wide range of features, with the most common being a flat facial profile, upward-slanting palpebral fissures, hypotonia, hyperflexibility, and a single palmar crease. Approximately 50% of individuals are born with one or more anomalies, with congenital heart defects occurring in half of the Down syndrome population. Specific medical complications can also arise in association with the condition, including hearing loss, vision problems, increased susceptibility to infection, and early-onset Alzheimer's disease (Antonarakis et al., 2020). There are three forms of Down syndrome: trisomy 21, mosaic trisomy 21, and translocation Down syndrome. Trisomy 21 makes up 90-95% of cases, and results from a random nondisjunction event that

typically occurs in the ovum (Coppedè, 2016). Advanced maternal age is a well-established risk factor for Down syndrome, as it is known to increase the chance of nondisjunction events. Environmental factors, including tobacco use, folic acid supplementation, oral contraceptive use, and BPA exposure, have also been thought to trigger nondisjunction events leading to trisomy 21.

Down syndrome may be diagnosed prenatally or postnatally (Antonarakis et al., 2020). Chorionic villus sampling (CVS), a test available from 10-13 weeks gestation, can diagnose Down syndrome prenatally. Amniocentesis is another diagnostic test available after 15 weeks gestation. Screening tests were developed in an effort to detect an increased chance for Down syndrome without performing an invasive procedure. Since the discovery of the association between maternal analyte levels and the occurrence of Down syndrome, multiple screenings have been developed, including the quadruple screen, first-trimester screen, nuchal translucency, and noninvasive prenatal screening (NIPS) (Russo & Blakemore, 2014).

The implementation of prenatal screening tests for Down syndrome has been met with mixed feelings from the disability community. Parents and providers alike have recognized positive aspects of NIPS, including the accuracy, safety, and ability to test earlier in the pregnancy (van Schendel et al., 2017). Though not a diagnostic test, NIPS has a high detection rate for Down syndrome and other aneuploidies which may reduce the number of invasive procedures done to confirm the presence of the condition, which is viewed as an advantage by parents (van Schendel et al., 2017). The early screening enabled by NIPS may also lead to increased prenatal diagnoses of Down syndrome, whereas historically the majority of cases were diagnosed postnatally (Skotko, 2009).

This changes the traditional diagnostic landscape, and gives women more time earlier in the pregnancy to consider various reproductive options including continuing the pregnancy, terminating the pregnancy, or adoption. The same factors that were viewed by parents to be advantages of NIPS were also viewed as disadvantages in different lights. For instance, parents worry that the noninvasive nature of the test could lead to increased uptake of screening for Down syndrome, and therefore could lead to more terminations on account of the condition. If less babies are born with Down syndrome, the supports and services available to individuals with the condition could decrease (How et al., 2019). Parents also fear that the normalization of such a prenatal screen would increase discrimination and stigmatization against individuals who have Down syndrome. Both parents and medical professionals recognize the potential benefits and harmful effects of NIPS, and its implementation is changing how and when individuals are being diagnosed with Down syndrome.

Regardless of if Down syndrome is found prenatally or postnatally, the majority of parents have reported dissatisfaction with their diagnosis experience. Recommendations to providers about the disclosure of a genetic diagnosis include providing accurate, up-to-date information in a balanced manner (Hippman et al., 2012; Saul & Meredith, 2016; Sheets et al., 2011). Providers are also advised to provide information not only about the medical side of Down syndrome, but also about life outcomes and social supports (Saul & Meredith, 2016). Parents frequently describe opposite experiences, reporting that medical provider descriptions were brief, inadequate, and confusing, often with the use of derogatory or offensive language (Quine & Pahl, 1987; Skotko, 2005; Sheets et al., 2009). Parents also felt as though they were not given

up-to-date materials or resources, and that referrals to other parents in similar situations were sorely lacking (Skotko, 2005).

Genetic counselors may be uniquely equipped to deliver a prenatal or postnatal diagnosis of Down syndrome to families. The American Society of Human Genetics (ASHG) proposed a widely-referenced definition of genetic counseling in 1975 which states that genetic counseling is "…a communication process which deals with the human problems associated with the occurrence, or the risk of occurrence of a genetic disorder in a family," which involves comprehending the medical facts, appreciating the inheritance of the disorder, understanding recurrence risk, choosing an appropriate course of action in accordance with patients' perception of risk, family goals, and ethical and religious standards, and to help patients make the best possible adjustment to the disorder (ASHG, 1975; Sheets et al., 2009). The therapeutic relationship between counselor and patient is central to the profession (Biesecker, 2019). The counselor's role is to elicit the patient's psychosocial and educational needs, provide relevant information in an accessible and empathetic way, and to facilitate informed decision-making. The core tenets of genetic counseling practice call for counselors to provide families balanced, accurate information about the medical and social sides of Down syndrome in an empathetic and non-judgmental manner.

The present study is replicating research conducted in 2009 that aimed to identify the essential informational items that should be imparted upon parents during the initial diagnosis of Down syndrome. Data from that study were utilized to create the "Practice Guidelines for Communicating a Prenatal or Postnatal Diagnosis of Down Syndrome: Recommendations of the National Society of Genetic Counselors" (Sheets et al., 2011).

The goal of the practice guidelines is to provide genetic counselors and other medical professionals information on how to disclose a Down syndrome diagnosis using balanced, up-to-date information while conveying support and respect (Sheets et al., 2011).

The aim of this study is to reassess the informational needs of parents during the presentation of a Down syndrome diagnosis. In 2009, data were collected to define the essential information necessary to give a balanced description of Down syndrome (Sheets, 2009). The manner in which a diagnosis is presented has a lasting impact on families, and the informational needs of parents have likely changed in the years since the previous study was conducted. As previous data were utilized in the preparation of the National Society of Genetic Counselors (NSGC) Down Syndrome Practice Guidelines (Sheets et al. 2011a), any shift in informational preferences over the last several years may suggest a need for an update or revision.

2.3 METHODS

2.3.1 DESIGN AND PARTICIPANTS

The institutional review board at the University of South Carolina approved this study in June of 2019. The present study replicated a prior study completed by Sheets et al. in 2009, and all current methodology is adapted from this prior study. Genetic counselors were identified through the National Society of Genetic Counselors (NSGC) membership listserv. Parents of children with Down syndrome were identified through local, national, and online organizations affiliated with Down syndrome, as well as in person at national conferences. Anonymous, online surveys for both genetic counselors and parents of children with Down syndrome were used in this study. The NSGC and

parent organizations received an invitational letter (Appendices A & B) containing the link to the online surveys inviting genetic counselors and parents to participate, and they distributed this information to their members (Sheets et al., 2009).

Upon completion of the online survey, genetic counselors were encouraged to share the online survey link with other genetic counselors and the link for the parent survey with parents of children with Down syndrome. Likewise, parents were encouraged to share the online survey link with other parents of children with Down syndrome upon completion of the survey (Sheets et al, 2009).

2.3.2 INSTRUMENTATION

The surveys used are adapted from those used by Sheets et al. (2009) to initially collect the data used to define essential information to give during a Down syndrome diagnosis. Minor changes in wording were made, as well as the addition and removal of some items. The frequency of features seen in Down syndrome were also updated. Items were amended or excluded due to outdated terminology or lack of relevance in current practice. The survey designed for genetic counselors consisted of four main sections. Two sections presented a wide range of features of Down syndrome to determine what information is considered essential for parents receiving a prenatal or postnatal diagnosis in the eyes of genetic counselors. The features fall into different categories, which include: (1) genetics of the condition, (2) diagnostic criteria and physical features, (3) associated medical complications, (4) intellectual disability and developmental delay of affected individuals, (5) long-term prognostications, (6) impact on the family, and (7) informational resources and referrals. This list of items was prepared by reviewing related texts (Sheets et al., 2009).

One section directed genetic counselors to rate the importance of the information for a prenatal diagnosis of Down syndrome, whereas another section directed genetic counselors to rate the importance of the items for a postnatal diagnosis. They were asked to rate each informational item as essential (3), important but not essential (2), not too important (1) or unsure (0). Another section was comprised of free response questions. The final section assessed general demographic information, including current specialty area, current work area, year of graduation, and level of contact with individuals with Down syndrome both in and out of their work setting.

The survey designed for the parents of children with Down syndrome was similar to the genetic counselor survey and also consisted of four sections. One section gathered information about their child's Down syndrome diagnosis, including the year of diagnosis, if it was prenatal or postnatal, which medical professional delivered the diagnosis, prenatal screening and/or testing received, whether they received genetic counseling, how informed they were when given the diagnosis, and their overall satisfaction with how they were given the diagnosis.

Another section asked parents to rate informational items about Down syndrome based on how essential it was for an initial discussion with new or prospective parents receiving a diagnosis for their newborn or unborn child. This list of features was identical to those previously described in the first and second sections of the genetic counselor survey, with the exception of minor changes to the wording to simplify medical terminology without losing meaning. Parents were provided the same Likert scale rating format described above. A third section was composed of free response questions. The fourth section recorded basic demographic information (Sheets et al., 2009).

2.3.3 DATA ANALYSIS

For quantitative data analysis, responses were separated into four groups: prenatal genetic counselors, postnatal genetic counselors, prenatal parents, and postnatal parents. Genetic counselor responses were separated into a prenatal group and a postnatal group based on ratings generated from the separate prenatal and postnatal information sections. Parents were also separated into prenatal and postnatal groups based on the timing of diagnosis. Data analysis was performed using SPSS Version 24 and Microsoft Office Excel Version 16, Copyright ©2020 Microsoft Corporation. General genetic counselor and parent groups from the 2009 study were compared to general genetic counselor and parent groups from the 2019 study.

Average ratings were calculated for each informational item using the Likert scale format (essential = 3; important but not essential = 2; not too important = 1; unsure =0) then assembled in rank order to determine the highest rated items. Independent T-Tests were performed to identify significant differences between prenatal parent and postnatal parent responses, and also for multiple comparisons between parent and genetic counselor groups. The qualitative data generated from free response questions were used to add further dimension to the investigation in defining balanced information about Down syndrome. The qualitative analysis and emergent themes were beyond the scope this paper and will be subsequently reported elsewhere (Sheets et al., 2009).

2.4 RESULTS

2.4.1 GENETIC COUNSELOR PARTICIPANTS

Of the 136 genetic counselors who opened the survey, 72 completed a significant portion. Since participants had the option of skipping items, not all participants

completed the same number of items in their respective surveys. The majority of genetic counselor participants were female, Caucasian, and between the ages of 25 and 34 years of age (see Table 1). Most have been working 5 years or less, with the majority working in academic or university-based practices (41.94%) or hospital settings (40.32%). A larger proportion of genetic counselor participants are currently working as prenatal genetic counselors (46.77%).

The majority of genetic counselors reported having some contact with individuals who have Down syndrome in their graduate training (79.03%), whereas only some reported having plenty of contact (12.90%). In current practice, most reported having no contact with individuals with Down syndrome (45.16%) or some contact (41.94%). Outside of a professional context, most genetic counselors had social contact only with individuals who have Down syndrome (50.00%) or no other contact (37.10%). When asked about familiarity with the NSGC Down Syndrome Practice Guidelines, the majority of genetic counselors reported that they had some exposure (51.61%) or plenty of exposure (40.32%). Most genetic counselor participants did not have children (66.13%). Religious activity among genetic counselors varied, with the majority having no religion (47.54%) or reporting being very active religiously (19.67%).

2.4.2 PARENT PARTICIPANTS

Of the 641 parents who opened the survey, 529 completed a significant portion of the questions. Like the genetic counselor survey, parent participants also had the option of skipping items, and therefore not all participants completed the same number of items in their respective surveys. The majority of the parents were female (96.90%) and Caucasian (89.38%) (see Table 2). Most parents were educated, with the majority

holding a Bachelor's degree (38.01%) or a Master's degree (26.37%). The majority of parent participants were married (86.64%) and had one child (20.21%), two children (34.59%), or three children (30.14%). The majority of parents were members of a Down syndrome advocacy group (65.64%), with even more reporting that they were members of 1-5 online support groups, social media groups, and/or online forums (72.11%). Most parent participants reported being either somewhat active (31.96%) or very religiously active (39.18%), and had a household income of $100,000 or more (51.63%).

2.4.3 INFORMATION REGARDING THE DIAGNOSIS

The majority of parents received their child's diagnosis within the last decade (72.11%) with 53.12% occurring postnatally, and 46.88% occurring prenatally (see Table 3). In most cases, parents were informed of the diagnosis of Down syndrome by an obstetrician-gynecologist (28.68%) or a pediatrician (23.02%). The diagnosis was first delivered by a genetic counselor 10.94% of the time. Of those diagnoses delivered by a genetic counselor, 10.00% were delivered by a prenatal genetic counselor, whereas 0.94% were delivered by a pediatric genetic counselor. Of this parent cohort, 33.52% had received pediatric genetic counseling, 23.86% had received prenatal genetic counseling, 9.28% had received both pediatric and prenatal genetic counseling, and 33.33% had none.

For most parents, their child with Down syndrome had been their first (28.60%) or second (25.95%) pregnancy, and their first (36.74%) or second (31.06%) child. During the pregnancy, the majority of parents had multiple prenatal screening and/or diagnostic tests (47.05%), whereas 24.76% of parents elected to have no prenatal screening or diagnostic tests. Regarding their experience of receiving their child's

diagnosis, 54.65% of parents felt they were insufficiently informed about Down syndrome when the diagnosis was given, and 49.81% were somewhat or extremely dissatisfied with the experience overall.

2.4.4 ESSENTIAL INFORMATION IN 2019

The data generated by the Likert scale rating format are considered ordinal, so the distribution or spacing between rating values may not be equal. Therefore, informational items were placed in rank order by average rating to determine the most essential items. Table 4 presents the informational items in rank order by average rating for all four groups of respondents. The higher the average rating, the more essential the item is for the initial discussion of Down syndrome. Nineteen informational items were present among the 30 highest ranked items for each group, which further demonstrate agreement on essential information for the initial discussion of Down syndrome.

Although some informational items were similarly rated as essential and appeared high in the rank order lists for each group, there were differences between the ratings by each group. Table 5 presents analyses of differences between groups for each informational item, and includes respective average ratings. Average ratings were not used for statistical analysis, but are provided in the table as a visual reference.

Prenatal and postnatal genetic counselor ratings were statistically significantly different for nine of the total 100 informational items. Prenatal genetic counselor ratings were significantly different from prenatal parents for 54 items, and postnatal parents for 48 items. Postnatal genetic counselor ratings were significantly different from prenatal parents for 57 of the 100 items, and postnatal parents for 53 items. Prenatal and postnatal parent groups rated 13 of the 100 informational items differently. There were 22

informational items for which there were no statistical differences between any of the groups in 2019 (Table 6).

To assess differences over time, the 2009 parent cohort was compared to the 2019 parent cohort, and the 2009 genetic counselor cohort was compared to the 2019 genetic counselor cohort (Table 8). The 2009 parents were significantly different from the 2019 parents for 51 of the total 100 informational items. The 2009 genetic counselors were significantly different from the 2019 cohort for 45 of the 100 informational items.

Table 2.1. Genetic counselor demographic information

Demographics	Responses	Total *n*	(%)
AGE GROUP (N=62)	20-24	5	8.06%
	25-29	31	50.00%
	30-34	9	14.52%
	35-39	4	6.45%
	40-44	5	8.06%
	45-49	3	4.84%
	50-54	1	1.61%
	55-59	4	6.45%
GENDER (N=62)	Female	58	93.55%
	Male	4	6.45%
RACE / ETHNICITY (N=62)	Black/African American/African	1	1.61%
	White	57	91.94%
	American Indian or Alaskan Native	0	-
	Spanish/Hispanic/Latino	0	-
	Asian Indian	0	-
	Chinese	1	1.61%
	Japanese	0	-
	Other Asian	1	1.61%
	Native Hawaiian or Pacific Islander	0	-
	Other	2	3.23%
YEARS EMPLOYED (N=61)	< 1 year	10	16.39%
	1 - 5 years	30	49.18%
	6 - 10 years	6	9.84%
	11 - 15 years	5	8.20%
	16 - 20 years	4	6.56%
	21 - 25 years	3	4.92%
	26 - 30 years	0	-
	> 30 years	3	4.92%
EMPLOYMENT SETTING (N=62)	Academic/University-based	26	41.94%
	Hospital-based	25	40.32%
	Multiple specialty group	1	1.61%
	Private practice	4	6.45%
	Industry	4	6.45%
	Other	2	3.23%

SPECIALTY AREA (N=62)	Prenatal	29	46.77%
	General Pediatrics	8	12.90%
	Pediatric specialty	3	4.84%
	Cancer	3	4.84%
	Adult	2	3.23%
	Infertility	0	-
	Multiple specialties	6	9.68%
	Laboratory	3	4.84%
	Research	3	4.84%
	Other	5	8.06%
RELIGIOUS ACTIVITY (N=61)	Occasionally active	10	16.39%
	Non-practicing	10	16.39%
	Very active	12	19.67%
	No religion	29	47.54%
NUMBER OF CHILDREN (N=62)	0 children	41	66.13%
	1 child	6	9.68%
	2 children	8	12.90%
	3 children	7	11.29%
	4 children	0	-
EXPOSURE TO DS IN TRAINING (N=62)	None	5	8.06%
	Some	49	79.03%
	Plenty	8	12.90%
EXPOSURE TO DS IN PRACTICE (N=62)	None	28	45.16%
	Some	26	41.94%
	Plenty	8	12.90%
EXPOSURE TO DS OUTSIDE PROFESSIONAL CONTEXT (N=62)	No other contact	23	37.10%
	Social only	31	50.00%
	Family only	2	3.23%
	Both social & family	6	9.68%

EXPOSURE TO NSGC DS PRACTICE GUIDELINES (N=62)	None	5	8.06%
	Some	32	51.61%
	Plenty	25	40.32%

Table 2.2. Parent demographic information

Demographics	Responses	Total *n*	(%)
PARENT GENDER (N=290)	Female	281	96.90%
	Male	9	3.10%
RACE / ETHNICITY (N=292)	Black/African American/African	3	1.03%
	White	261	89.38%
	American Indian or Alaskan Native	0	-
	Spanish/Hispanic/Latino	20	6.85%
	Asian Indian	2	0.68%
	Chinese	2	0.68%
	Japanese	1	0.34%
	Other Asian	1	0.34%
	Native Hawaiian or Pacific Islander	0	-
	Other	2	0.68%
RELIGIOUS ACTIVITY (N=291)	No Religion	41	14.09%
	Non-practicing	43	14.78%
	Occasionally active	93	31.96%
	Very active	114	39.18%
HIGHEST EDUCATION LEVEL (N=292)	No Education	0	-
	Middle school	1	0.34%
	Some high school	3	1.03%
	High school graduate	19	6.51%
	Some college	24	8.22%
	Associate's degree	26	8.90%
	Bachelor's degree	111	38.01%
	Master's degree	77	26.37%
	Professional degree	21	7.19%
	Doctorate degree	8	2.74%
	Other	2	0.68%
MARITAL STATUS (N=292)	Never married	9	3.08%
	Civil union	3	1.03%

	Married	253	86.64%
	Separated	8	2.74%
	Divorced	17	5.82%
	Widowed	2	0.68%
TOTAL NUMBER OF CHILDREN (N=292)	1 child	59	20.21%
	2 children	101	34.59%
	3 children	88	30.14%
	4 children	25	8.56%
	5 children	12	4.11%
	6 children	2	0.68%
	7 children	1	0.34%
	8 children	3	1.03%
	9 children	0	-
	10 children	0	-
	> 10 children	1	0.34%
ADVOCACY GROUP MEMBERSHIP (N=521)	Yes	342	65.64%
	No	179	34.36%
ONLINE SUPPORT GROUP, SOCIAL MEDIA GROUP, ONLINE FORUM (N=527)	Yes: 1-5	380	72.11%
	Yes: 6-10	51	9.68%
	Yes: over 10	28	5.31%
	Not involved	68	12.90%
HOUSEHOLD INCOME (N=277)	< $24,999	14	5.05%
	$25,000 - $49,999	25	9.03%
	$50,000 - $74,999	46	16.61%
	$75,000 - $99,999	49	17.69%
	$100,000 - $124,999	56	20.22%
	$125,000 - $149,999	29	10.47%

	> $150,000	58	20.94%

Table 2.3. Information regarding the diagnosis

Information	Categories	Total *n*	(%)
DIAGNOSIS YEAR (N=527)	1970-1979	1	0.19%
	1980-1989	17	3.23%
	1990-1999	46	8.73%
	2000-2009	83	15.75%
	2010-2019	380	72.11%
TIMING OF DIAGNOSIS (N=529)	Postnatal	281	53.12%
	Prenatal	248	46.88%
INFORMER (N=530)	OB/GYN	152	28.68%
	MFM	55	10.38%
	Pediatrician	122	23.02%
	Pediatric Geneticist	11	2.08%
	Prenatal Genetic Counselor	53	10.00%
	Pediatric Genetic Counselor	5	0.94%
	Nurse	23	4.34%
	Other	109	20.57%
NUMBER PREGNANCY (N=528)	1st	151	28.60%
	2nd	137	25.95%
	3rd	100	18.94%
	4th	62	11.74%
	5th	30	5.68%
	6th	20	3.79%
	7th	7	1.33%
	8th	5	0.95%
	9th	1	0.19%
	10th	3	0.57%
	> 10th	10	1.89%
	Unsure	2	0.38%
NUMBER CHILD (N=528)	1st	194	36.74%
	2nd	164	31.06%
	3rd	98	18.56%
	4th	42	7.95%

	5th	13	2.46%
	6th	7	1.33%
	7th	2	0.38%
	8th	4	0.76%
	9th	0	-
	10th	2	0.38%
	> 10th	2	0.38%
PRENATAL SCREENING / TESTING (N=525)	None	130	24.76%
	Screening Test (Only)	104	19.81%
	Diagnostic Test (Only)	14	2.67%
	Multiple Tests	247	47.05%
	Unsure, but had some form of prenatal screening	30	5.71%
HOW INFORMED AT TIME OF DIAGNOSIS (N=527)	Not very informed	288	54.65%
	Somewhat informed	167	31.69%
	Very well informed	72	13.66%
PARTICIPATION IN GENETIC COUNSELING (N=528)	No GC	176	33.33%
	Prenatal GC	126	23.86%
	Pediatric GC	177	33.52%
	Both Prenatal & Pediatric GC	49	9.28%
SATISFACTION WITH EXPERIENCE (N=524)	Extremely dissatisfied	155	29.58%
	Somewhat dissatisfied	106	20.23%
	Somewhat satisfied	115	21.95%
	Extremely satisfied	73	13.93%
	Unsure	75	14.31%

Table 2.4. Essential informational items for practice in 2019

Caused by extra material from chromosome 21*	Special Education classes
Diagnosis confirmed by chromosome analysis*	Complete high school
Hypotonia*	Supported, competitive employment
One or more congenital abnormalities	Live independently
Heart defect possibly requiring open heart surgery*	**Have friends***
Gastrointestinal defect possibly requiring surgery	Have intimate relationships
Hearing Loss (75%)	Life expectancy (age range of 50s-60s)
Variable range of intellectual disability from mild to moderate*	**More like other children than different***
Developmental delay in achieving milestones*	**Local support groups***
Need for physical therapy*	**National advocacy organizations and websites***
Need for occupational therapy*	Online support groups, social media, or online forums*
Need for speech and language therapy*	Early Intervention centers*
Need for early intervention and case management*	**Printed or written material***
Participate in community sports, activities, and leagues*	Fact sheets/brochures
Inclusion in regular classes	Specialist referral(s)

Note: Items listed were rated *essential* by the majority of respondents in all four groups. *Indicates item among the highest rated (top 30) items for all groups. Items with no statistical difference between any groups appear in bold type.

Table 2.5. Items with no statistically significant differences between groups in 2019

Item	*Average Overall Rating*
Caused by extra genetic material from chromosome 21	2.77
Flat facial profile (90%)	1.87
Upward slanting, almond-shaped eyes (80%)	1.94
Hypotonia (80%)	2.65
Excess skin on back of neck (80%)	1.79
Epicanthal folds	1.72
Single palmar crease	1.68
Short stature	2.05
Gastrointestinal defect possibly requiring surgery (12%)	2.48
Need for physical therapy	2.73
Need for occupational therapy	2.72
Need for speech & language therapy	2.74
Need for Early Intervention & case management	2.87
Participate in community sports, activities, & leagues	2.66
Have friends	2.82
More like other children than different	2.88
Impact on other siblings – less attention & resentful	2.14
Impact on marriage – strains relationship	2.15
Impact on extended family members – limited interaction	1.83
Local support group(s)	2.90
Advocacy organizations & websites	2.77
Printed/Written material	2.73

2.5 DISCUSSION

2.5.1 ESSENTIAL INFORMATION FOR ROUTINE PROVISION

Upon first giving a diagnosis of Down syndrome, it is unrealistic for providers to communicate every detail about the condition to parents, nor are parents able to take in vast volumes of information at that time. Results of this study identified 30 essential informational items for the initial presentation of a Down syndrome diagnosis (see Table 2.4). Of these items, 19 were present among the highest ranked items for all groups, which further characterizes the importance of these items. Of the items deemed essential in the present study, two items, 'Hearing loss (75%)' and 'Online support groups, social media, or online forums' were not categorized as essential in the 2009 study. Certain items that were categorized as essential in 2009 were not rated as essential in the present study, including 'Recurrence risk for future pregnancies,' 'Employed in workshop setting,' 'Live in group home,' 'More time commitment,' 'Books,' and 'Contact with families raising a child with Down syndrome.' As done previously, informational items were placed in rank order to highlight which elements are valued most by prenatal genetic counselors, postnatal genetic counselors, prenatal parents, and postnatal parents (Sheets et al., 2009).

2.5.2 INTERPRETING ESSENTIAL IN 2019

For both the 2009 and 2019 study, items were interpreted as "*essential*" if ≥ 50% of respondents from each of the four groups rated them as a 3, signifying they felt it was essential to share this information with parents at the initial presentation of the diagnosis. Presenting the informational items in rank order highlights that certain items are regarded as more or less important depending on the group. For instance, 'Developmental delay in

achieving milestones' was highly rated among genetic counselor groups, garnering the 3rd highest average rating for prenatal genetic counselors, and as the highest average rating for postnatal genetic counselors. While 'Developmental delay in achieving milestones' still fell within the top 30 ranked items for the parent groups, it earned the ranking of 17th for prenatal parents, and 26th for postnatal parents. This exemplifies that different items classified as 'essential' do not carry equal weight across the different groups.

Differences such as the average rating of 'Developmental delay in achieving milestones' may illustrate the contrasting roles and responsibilities of a provider and a parent. As the item was rated as essential in >50% of cases for both genetic counselor and parent groups, and it appears in the top 30 average rating rankings for each group, its importance is underscored. Genetic counselors appear to find it crucial to discuss the possibility of a child with Down syndrome experiencing developmental delays in achieving milestones at the initial point of diagnosis as a part of their job. While parents agree that this is an important facet of Down syndrome to discuss, overall they place more emphasis on receiving information about resources upon first receiving the diagnosis. This example highlights the difference in provider and parental roles, rather than a disparity in the rating of items.

2.5.3 DIFFERENCES BETWEEN PRENATAL AND POSTNATAL INFORMATION IN 2019

Like the original study, the current study aimed to elucidate any differences in informational disclosure between prenatal and postnatal diagnoses. Prenatal and postnatal genetic counselor ratings were statistically different in 9 of the total 100 items. In 2009, 70 of 100 informational items were statistically different between prenatal and

postnatal genetic counselors. The great reduction in statistically significant differences between prenatal and postnatal counselors suggest that the profession has evolved, and similar emphasis is placed on certain content regardless of specialty.

Though overall there were few statistical differences between prenatal and postnatal genetic counselors, there were some discrepancies between the two groups. This suggests that there is still some information that is considered more relevant in the prenatal or postnatal diagnostic setting. Understandably, prenatal genetic counselors rated 'Diagnosis confirmed by chromosome analysis' significantly higher than postnatal genetic counselors. This is likely due to the time pressure faced by prenatal counselors during an ongoing pregnancy to make sure a patient has access to all reproductive options, and the need to perform screening and diagnostic tests in narrow gestational windows. Though there was a significant difference between the two groups, both gave the item high average ratings, with it being the highest rated item for prenatal genetic counselors, and the 9th highest rated item for postnatal genetic counselors. There was also a significant difference between groups regarding the item 'Recurrence risk for future pregnancies,' with postnatal genetic counselors rating it significantly higher than prenatal counselors. This suggests that prenatal counselors are more focused on supporting the patient through their current pregnancy than they are on discussing possible outcomes of future pregnancies. In the postnatal setting, the child has been born, and more focus can be placed on the discussion of future pregnancy outcomes.

Several differences between prenatal and postnatal genetic counselors are for informational items in the 'Medical Complications Associated with Down Syndrome' category. Postnatal genetic counselors rated 'Thyroid disorders (up to 50%),'

'Obstructive sleep apnea (33%),' 'Vision problems (60%),' 'Hearing loss (75%),' and 'Ear problems/infections (up to 70%)' significantly higher than prenatal genetic counselors. These medical issues, while not experienced by all individuals who have Down syndrome, are listed by the American Academy of Pediatrics (AAP) as the most common medical complications seen with the condition (Bull, 2011). From our study, it is apparent that counselors in the postnatal setting are aware of these AAP health supervision guidelines that direct screenings and clinical care for children with Down syndrome from birth onward. While these medical issues are important to discuss with families postnatally, they are not complications that can be detected prenatally, making these items less relevant for prenatal counselors to emphasize.

Interestingly, prenatal genetic counselors rated 'Inclusion in regular classes' significantly higher than postnatal genetic counselors. This item was ranked as the 13th highest average rating among prenatal counselors, but fell below the top 30 ranked items for postnatal genetic counselors at 32nd. This difference could be attributed to the prenatal counselor's tendency to provide a range of possible outcomes to prospective parents, whereas postnatally counselors can focus on the child's development, individual needs, and the desires of the family.

'Fact sheets/brochures' also was rated significantly higher by prenatal genetic counselors than postnatal genetic counselors. Interestingly, this was one of only two informational items that had no statistically significant differences between any of the four groups in the 2009 study. Well-established parental preference for accurate, balanced information in a written format supports the provision of resources such as fact sheets or brochures when giving a diagnosis (Saul & Meredith, 2016; Sheets et al., 2009).

Prenatal genetic counselors may place slightly higher emphasis on providing this type of resource to help their patients better understand the complexities of prenatal screening and diagnostic testing. Prenatally, also added is reproductive options.

Prenatal and postnatal parents were statistically the same for 87 of the total 100 informational items. Interestingly, prenatal parents had higher average ratings for all thirteen items that were statistically different. The different items fell under several of the study categories. Under the 'Genetics of Down syndrome,' prenatal parents placed higher value on information about reproductive options and adoption than postnatal parents. Reproductive options most commonly are discussed during a current pregnancy, and therefore are more relevant in a prenatal diagnostic setting. Prenatal screening and diagnostic testing for Down syndrome is not done to inform a treatment plan or uncover a cure, but rather to direct pregnancy management decisions, including whether to continue the pregnancy, elect to terminate, or make plans for adoption (Reed & Berrier, 2016). Because of this, it makes sense that prenatal parents rank adoption higher than postnatal parents. However, it is important to note that both reproductive options and adoption were ranked relatively lowly by both groups of parents: Reproductive options was ranked 76 by prenatal parents and 92 by postnatal parents, and adoption was ranked 71 by prenatal parents and 79 by postnatal parents. This study did not include the perspectives of patients who have terminated pregnancies affected with Down syndrome, or of patients who have chosen to adopt out a child who has Down syndrome. Therefore, there could be inherent bias against these options in this parent population.

Other differences between prenatal and postnatal parents include perceived importance of items in the 'Medical Complications Associated with Down syndrome,

'Long-Term Prognosis for Individuals with Down syndrome,' 'The Family of Individuals with Down syndrome,' and 'Informational Resources & Referrals for Individuals with Down syndrome' categories. Generally, prenatal parents rated all discrepant items statistically higher than the postnatal parents. This further demonstrates that prenatal parents want to hear the broad range of features and characteristics that can be seen with Down syndrome, whereas postnatal parents may be more focused on their child's individual needs.

2.5.4 DISCREPANCIES BETWEEN PARENTS AND PROVIDERS

Like the 2009 study, both parents and providers seem to appreciate the importance of similar information to present at the initial diagnosis of Down syndrome. Differing levels of emphasis were placed on certain categories of informational items. Significant differences between parents and providers could signify that there are unmet needs of parents as they go through the diagnosis process. It is important to bring these differences to light to determine if disparities are due to the contrasting roles of a medical professional and parent, or rather to unfulfilled informational needs of parents and families.

Like the previous study, there is still a disparity between parents and providers regarding the importance of reproductive options at the initial point of diagnosis. When considering the rankings of average ratings, prenatal parents and postnatal parents ranked 'Reproductive options' at 76 and 92, respectively. On the other hand, 'Reproductive options' was ranked 27 for prenatal genetic counselors and 36 for postnatal counselors, and the item was considered essential information for both groups. Consistent with the professional standards of genetic counseling, pregnant women should be offered all

reproductive options available to her following a positive screening or diagnostic test in a respectful, non-judgmental way (Sheets et al., 2009). These options include continuing a pregnancy, terminating the pregnancy, and pursuing adoption. This immense divergence in ranking may depict ongoing contentions between genetic counselors and the disability community that were also evident in the 2009 study. A previous study reported that when asked about the collective provision of genetics services, individuals with disabilities expressed worry that these services were contributing to a less tolerant view of disability in society (Roadhouse et al., 2018). Selective termination following a positive prenatal screen or prenatal diagnosis has been viewed as furthering discrimination by members of the disability community. Some have even stated that the mere existence of prenatal testing options is discriminatory (Roadhouse et al., 2018). While genetic counselors are expected to discuss all available reproductive options with patients as a standard of care, they must understand and respect parents' thoughts and perceptions on this sensitive issue.

Parents and genetic counselors placed higher emphasis on contrasting aspects of medical complications that can be seen with Down syndrome. Genetic counselors prioritized the 'One or more congenital abnormalities (59%)' item, with it falling at the 7th highest ranking for prenatal genetic counselors and the 6th highest ranking for postnatal genetic counselors. Notably, this item was not in the top 30 rated items for either of the parent groups, falling at 33 for prenatal parents and 36 for postnatal parents, and there were significant differences between all parent and genetic counselor groups. Though there is a clear disparity between level of emphasis placed by genetic counselors versus parents, this item was still categorized as essential by all four groups.

The item concerning the chance of a child with Down syndrome having a heart defect possibly warranting open heart surgery was rated extremely high by all four groups, with there being only one significant difference in average ratings between postnatal genetic counselors (average 2.95) and postnatal parents (average 2.82). This item fell in the top 30 ratings for all groups, and was considered to be essential. It could be that parents interpret this item to be more relevant or specific than 'One or more congenital abnormalities,' as a heart defect can be one of the more severe and common complications that can be seen in Down syndrome, and can require immediate medical attention at birth if discovered prenatally.

Respiratory problems, orthopedic problems, increased susceptibility to infection, increased susceptibility to autoimmune disease, increased susceptibility to periodontal disease, and tendency for obesity were rated significantly higher in both parent groups than in the genetic counselor groups. These are all more general items that have high frequencies in the general population, but can be seen slightly more often in the Down syndrome population. Since these items are more generalized, it is possible that genetic counselors place less of an emphasis on them. However, these are items that parents may experience more frequently in their child's everyday life. This disparity between genetic counselors and parents highlights the counselors' tendency to focus on the specific, medical side of Down syndrome, and the parents' desire to become informed about the social side of Down syndrome.

Items within the 'Long-Term Prognosis for Individuals with Down syndrome' category highlighted intriguing differences between parent and provider perspectives. Overall, parents seemed to exhibit a preference for inclusion over specialized programs,

whereas genetic counselors placed more value on specialized programs. Inclusion was rated highly by parents, ranking as the 9th highest rated item for prenatal parents and the 13th highest for postnatal parents. In comparison, inclusion in regular classes ranked as the 13th highest rated item for prenatal counselors, yet it was not rated in the top 30 for postnatal counselors with a ranking of 32nd. This difference was statistically different between prenatal and postnatal counselors, and is likely attributed to prenatal counselors wanting to provide information on the full range of possibilities for individuals with Down syndrome, whereas postnatal counselors can focus on the child's individual needs. However, the item 'Special Education classes' was rated significantly higher by both groups of genetic counselors than both parent groups. Special education classes fell in the top 30 rated items for genetic counselors, though it was not present in the top 30 for either parent group. Parents rated 'Complete high school' and 'Attend College or Post-Secondary Education' statistically higher than genetic counselors in three of the four comparisons done between genetic counselor and parent groups for each. In conjunction with these educational trends, parents exhibited a clear preference for 'Live independently' over 'Live in a group home,' with the live independently item ranking at 16th for prenatal parents and 21st for postnatal parents, and live in a group home falling at 47th for prenatal parents and 53rd for postnatal parents. On the other hand, genetic counselors rated the importance of living independently and living in a group home very similarly. Prenatal genetic counselors rated living in a group home at 17th and living independently at 20th, with postnatal counselor rankings being 28th and 29th, respectively. These findings are suggestive of a cultural shift in the disability community; rather than focusing on getting their children involved in specialized, separate programs such as

special education or group homes, parents seem more interested in mainstream inclusion. While genetic counselors appear to value informational items such as inclusion in regular classes and living independently, they are placing equivalent emphasis on specialized programs. It is important that genetic counselors and other medical providers realize the importance of both inclusivity and specialized programs, but they must understand that disability culture may have shifted to favor inclusion in the Down syndrome community.

Notably absent from the informational items deemed essential in Table 2.4 are items from the 'Family of Individuals with Down syndrome' category. This is not for lack of essential ratings from parents; the informational items 'Impact on siblings – more compassionate & caring,' 'Impact on marriage – strengthens relationship,' 'Impact on grandparents – supportive & welcoming,' 'Impact on extended family members – supportive & welcoming,' 'Impact on other relationships – supportive & welcoming,' and 'Time commitment – more' were all categorized as essential by both parent groups. None of these items received a majority of essential ratings by either genetic counselor group, and parents' ratings were statistically higher than genetic counselors in nearly every comparison. Interestingly, there were far fewer statistically significant differences between parents and genetic counselors for the negative side of each of these items. For instance, though parents and genetic counselors were statistically different in every comparison for the item 'Impact on other siblings – more compassionate & caring,' there were no statistical differences between any parent and genetic counselor groups for the item 'Impact on other siblings – less attention & resentful.' This pattern persisted throughout this category. These findings emphasize that parents want to discuss the potential impacts a diagnosis of Down syndrome can have on the family. While it

appears providers place the same value as parents on the potential negative impacts on the family, it seems they are not meeting parental needs when it comes to discussing positive impacts. These positive aspects being categorized as essential by both parent groups highlights the importance of addressing the potential impact on the family during the initial diagnosis conversation.

Generally, the 'Informational Resources & Referrals for Individuals with Down syndrome' category of informational items was regarded highly by both genetic counselors and parents. Notably, 'Early Intervention centers' was the highest-rated item for both prenatal parents and postnatal parents, with average ratings of 2.94 and 2.92, respectively. Both groups of genetic counselors rated the item significantly lower than the parents, with the item receiving a ranking of 15th for prenatal counselors (average rating 2.74) and 16th for postnatal counselors (average rating of 2.74). While 'Early Intervention centers' was still considered an essential item by genetic counselors and parents alike, it is important to highlight that both groups of parents in this study view it as the most essential item. In comparison, genetic counselors placed more value on the medical and developmental aspects of Down syndrome, particularly the items concerning mild to moderate intellectual disability and the likelihood of children experiencing developmental delays in achieving milestones. Interestingly, higher on the lists of both genetic counselor groups than 'Early Intervention centers' was 'Need for Early Intervention & case management,' falling at the 5th ranking for prenatal genetic counselors and at the 7th ranking for postnatal genetic counselors. This subtle difference may show that genetic counselors are quick to acknowledge that children with Down

syndrome may need these services, however they seem less concerned about addressing how to access these specific services.

2.5.5 COMPARISON BETWEEN 2009 AND 2019

Comparison of the essential informational items from the 2009 and 2019 studies reveals both reinforcement of the importance of certain items as well as shifts in the diagnostic and sociocultural landscapes over the last decade. Of the 34 essential items identified in 2009 and the 30 essential items identified in 2019, there were 28 commonalities. Of the items deemed essential in the present study, two items, 'Hearing loss (75%)' and 'Online support groups, social media, or online forums' were not categorized as essential in the 2009 study. The item 'Online support groups, social media, or online forums' was not originally included in 2009, but was added to the current study as the prominence of online support and social media has flourished within the last ten years. Certain items that were categorized as essential in 2009 were not rated as essential in the present study, including 'Recurrence risk for future pregnancies,' 'Employed in workshop setting,' 'Live in group home,' 'More time commitment,' 'Books,' and 'Contact with families raising a child with Down syndrome.' The item 'Employed in workshop setting' was not included in the present study.

The 34 items listed in the essential information table from 2009 were ultimately used in the preparation of the NSGC Down Syndrome Practice Guidelines for Communicating a Prenatal or Postnatal Diagnosis of Down Syndrome. Therefore, the extensive commonalities between the 2009 essential items table and the 2019 essential

items table reinforce the recommendations of the Practice Guidelines. Notably, there were far less significant differences between groups in the 2019 study compared to the 2009 study. There were only two items in 2009, 'Printed or written material' and 'Fact sheets or brochures,' that had no significant differences between groups. In comparison, thirteen items in the 2019 essential informational items table alone had no significant differences between groups. This trend was observed for other items not rated as essential in the 2019 study. The increase in the number of items lacking significant differences embodies a growing consensus between genetic counselors and parents, and prenatal and postnatal viewpoints.

While the essential informational items table demonstrates more of a consensus between groups, the absence of some key items highlight areas of disagreement between genetic counselors and parents. The item 'Contact with families raising a child with Down syndrome' was included in the list of essential informational items in 2009, however it was absent in the 2019 list. The item was highly rated in both parent groups, earning the 2nd highest ranking for prenatal parents (average rating 2.93) and the 5th highest ranking for postnatal parents (average rating 2.86). The vast majority of parents rated the item as 'essential,' including 93.5% of prenatal parents and 88.2% of postnatal parents. In contrast, 41.5% of prenatal genetic counselors and 49.2% of postnatal genetic counselors rated the item as essential, and rankings fell at 37th and 35th, respectively. Given that 'Contact with families raising a child with Down syndrome' is such a highly-rated item among parents, it is important to call attention to this discrepancy. Since the diagnostic landscape of Down syndrome is transitioning from predominantly postnatal to being more evenly distributed between prenatal and postnatal due to the uptake of

prenatal screenings such as NIPS, it is likely that less emphasis is being placed on providing a family contact for parents during the initial diagnosis.

As mentioned previously, a big difference between genetic counselors and parents in 2019 is the level of emphasis placed on discussing the potential impacts a Down syndrome diagnosis can have on family dynamics. Several items from the 'Family of Individuals with Down syndrome' category were rated as essential by both parent groups, yet they were not rated essential by genetic counselors, and therefore were not included in the essential informational items table. When comparing ratings of parents from 2009 to 2019 in this category, many items significantly increased, suggesting that parents are placing even more value on discussing potential family impacts at the time of the initial diagnosis. Increases were seen for both positive and negative impacts, demonstrating that parents value hearing a balanced description. While increases in the ratings of items in this category were seen for genetic counselors, the data from 2019 reveal that they consistently rate them statistically lower. This is more evident for the positive family items, whereas parents and genetic counselors have less differences between them for the negative family items. This disparity emphasizes even more the importance of genetic counselors including 'Contact with families raising a child with Down syndrome' in their repertoire of resources to give parents at the initial diagnosis.

2.6 LIMITATIONS AND FURTHER INVESTIGATION

The conclusions drawn from this study are limited, as responses from parents and genetic counselors reflect voluntary, nonrandom participation. Parent responses were biased, since the majority of parents were recruited through national and local advocacy groups as well as online support platforms. The perspectives of parents who chose

adoption or to terminate a pregnancy affected with Down syndrome were largely excluded. Future studies should explore the perspectives of this portion of the parent population to get a more complete picture.

Recall bias is an inherent part of this study due to the retrospective nature of survey responses. Though the majority of parent respondents in 2019 have received their child's diagnosis in the last decade, parental needs are likely to change with time as their child ages (Sheets et al., 2009). Parental needs may also be inseparable from their child and family's individual needs, and therefore results may not be generalizable to the parent population as a whole. The majority of genetic counselor participants were members of NSGC. Responses may represent the professional standards set forth by the NSGC code of ethics.

Though not likely, minute differences in the language and included items between the 2009 survey and the 2019 survey could have impacted the significance of responses. Some items were not evaluated for differences in ratings from 2009 to 2019, as they were only present in one of the studies. Certain items included in 2009 were excluded in 2019 because of outdated terminology or a lack of relevance in current care practices. New items were added in the present study that were not included in the previous study that have become relevant in the last decade. Finally, slight changes in wording, phrasing, and risk numbers may have influenced how both parents and genetic counselors rated the importance of certain items.

CHAPTER 3: CONCLUSIONS

The current study identified both areas of agreement as well as areas of discrepancy among parents and genetic counselors. The essential informational items to impart on families initially receiving a diagnosis of Down syndrome largely remained the same from 2009 to 2019 with fewer statistically significant differences between groups. The large number of item commonalities with fewer statistical differences emphasizes the importance of these items, and highlights a growing consensus between parents and genetic counselors on what information is truly essential to provide during the initial diagnosis experience. Since the NSGC Down Syndrome Practice Guidelines were heavily influenced by the essential items identified in 2009, the present study reinforces the adherence to the recommendations provided in the guidelines.

Despite a growing consensus between parents and genetic counselors based on the essential informational items that were identified, parental dissatisfaction with the diagnosis experience is still high. This could be because of the items recognized as essential by parent groups that were not considered essential by the genetic counselor groups. Parents rated items pertaining to the social side of raising a child with Down syndrome, such as items related to family impact, possible day-to-day medical complications, and certain resources, consistently higher than genetic counselors. These items would have been included among the essential informational items based solely on parental preferences. Parents also seem to be favoring inclusivity over specialized programs, and prefer to receive information through various mediums. While the

guidelines emphasize the importance of content prioritized by parents and genetic counselors alike, genetic counselors need to be cognizant of information deemed essential by parents in order to increase parental satisfaction with the diagnosis experience.

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APPENDIX A: INVITATIONAL LETTER & ONLINE SURVEY FOR GENETIC COUNSELORS

INVITATION TO PARTICIPATE

Dear Genetic Counselor:

Thank you for your interest in this study. The goal of this Genetic Counseling Master's Thesis Project is to reevaluate the essential informational needs of parents receiving a prenatal or postnatal diagnosis of Down syndrome. Our initial study was conducted by Katie Berrier Sheets in 2009, and the data gathered were used to help establish the National Society of Genetic Counselors Down Syndrome Practice Guidelines in 2011. However, information deemed essential while receiving a diagnosis has likely shifted over the last several years as the social and cultural landscape has changed tremendously.

Major advancements have occurred in the last decade, including the clinical implementation of Noninvasive Prenatal Screening and the establishment of many new postsecondary education programs. These advancements highlight a need to reevaluate how we disclose Down syndrome diagnoses now that more options and opportunities are available to patients.

We aim to assess perspectives of genetic counselors and parents again in this present study. Survey questions include rating informational items presented during both a prenatal and postnatal diagnosis disclosure, free response questions, and demographic information questions. Your involvement is extremely valuable to healthcare professionals involved in distributing information and resources and those involved in the care of individuals with Down syndrome and their families.

Participation involves completing an online survey. The online survey will be available until December 30th, 2019 at midnight, and it will be taken through Qualtrics. The survey will take approximately 20 minutes to complete.

Participation is voluntary. Your consent to participate is given by completing and submitting the online survey. You many choose not to complete the survey and may exit at any time. All responses will be anonymous, and in no way will be identifiable or linked back to you. If you have any questions regarding your rights as a participant in

this project, you may contact the University of South Carolina Office of Research Compliance at 803-777-7095.

If you have questions or difficulty accessing the online survey, please reach out to Margaret Wilkes, the primary investigator, at margaret.wilkes@uscmed.sc.edu. Thank you for your participation.

Sincerely,

Margaret J. Wilkes, BA

Genetic Counseling Program

University of South Carolina

PRENATAL DIAGNOSIS – DEFINING BALANCED INFORMATION ABOUT DOWN SYNDROME

This survey includes two sections for defining balanced information—prenatal and postnatal. You may choose to complete one section or both. This is the PRENATAL section. Please rate each item as you feel is an essential or important component of a balanced presentation for parents receiving a PRENATAL diagnosis of Down syndrome for their unborn child. Numerical data and frequencies (%) are provided where available.

1. Rate each item: Genetics of Down syndrome

   Incidence (1/600 to 1/800 births)
   Increasing incidence with increasing maternal age
   Caused by extra genetic material from chromosome 21
   3 Types: Nondisjunction (95%), Translocation (2-4%) Mosaic (2-4%)
   Diagnosis confirmed by chromosome analysis
   Recurrence risk for future pregnancies
   Reproductive options
   Adoption

2. Rate each item: Diagnostic Criteria & Physical Features of Down syndrome

   Flat facial profile (90%)
   Upward slanting, almond-shaped eyes (80%)
   Hypotonia (80%)
   Hyperflexibility (80%)

Excess skin on back of neck (80%)
Dysplastic ear (60%)
Dysplasia of midphalanx of 5th digit (60%)
Small head
Brushfield spots on the iris of the eye
Epicanthal folds
Short, broad hands with short fingers
5th finger clinodactyly
Single palmar crease
Sandal gap between 1st and 2nd toes
Short stature
Enlarged tongue
Syndactyly of 2nd and 3rd toes

3. Rate each item: Medical Complications Associated with Down syndrome

   Early-onset Alzheimer Disease (<5% under 40; 5-15% by age 40-49; 68-80% by age 65)
   Dry, hyperkeratotic skin (75%)
   One or more congenital abnormalities (59%)
   Heart defect possibly requiring open heart surgery (50%)
   Thyroid disorders (up to 50%)
   Obstructive sleep apnea (50-75%)
   Atlantoaxial and/or atlantooccipital instability (1-2%)
   Gastrointestinal defect possibly requiring surgery (12%)
   Autism Spectrum Disorder (ASD) (1%)
   Epilepsy (1-13%)
   Increased risk of childhood leukemia (1%)
   Congenital or acquired cataracts (15%)
   Vision problems (60%)
   Hearing loss (75%)
   Ear problems/infections (up to 70%)
   Increased susceptibility to periodontal disease (23%)
   Increased susceptibility to infection
   Increased risk of autoimmune disease
   Attention deficit hyperactivity disorder (ADHD)
   Respiratory problems
   Orthopedic problems
   Psychiatric disorders
   Tendency for obesity
   Sterility in males
   Increased risk of testicular cancer in males

4. Rate each item: Intellectual Disability & Developmental Delay for Individuals with Down syndrome

Variable range of intellectual disability from mild to moderate
Mean (average) IQ of 50
Range of IQ from 20 to 70
Developmental delay in achieving milestones
Need for physical therapy
Need for occupational therapy
Need for speech & language therapy
Need for early intervention & case management

5. Rate each item: Long-Term Prognosis for Individuals with Down syndrome

   Participate in community sports, activities, & leagues
   Participate in Special Olympics & therapeutic recreation
   Inclusion in regular classes
   Special Education classes
   Finish high school
   Attend college or post-secondary education
   Supported, competitive employment
   Live independently
   Live in group home
   Have friends
   Have intimate relationships
   Get married
   Life expectancy (age range of 50s-60s)
   More like other children than different

6. Rate each item: The Family of Individuals with Down syndrome

   Impact on other siblings – more compassionate & caring
   Impact on other siblings – less attention & resentful
   Impact on marriage – strengthens relationship
   Impact on marriage – strains relationship
   Impact on grandparents – supportive & welcoming
   Impact on grandparents – limited interaction
   Impact on extended family members – supportive & welcoming
   Impact on extended family members – limited interaction
   Impact on other relationships – supportive & welcoming
   Impact on other relationships – lose social circle
   Financial impact – No difference
   Financial impact – More
   Time Commitment – No Difference
   Time Commitment – More

7. Rate each item: Informational Resources & Referrals for Individuals with Down syndrome

Local support group(s)
National advocacy organizations & websites
Online support groups/social media platforms
Early intervention centers (Birth to 3-Years programs)
Printed/written material
Photographs of children with Down syndrome
Fact sheets/brochures
Books
Contact with families raising a child with Down syndrome
Adoption agencies
Alternative/nonconventional therapies
Specialist referral(s)
Counselor or family therapist referral(s)
Pastoral counseling referral(s)

POSTNATAL DIAGNOSIS – DEFINING BALANCED INFORMATION ABOUT DOWN SYNDROME

This survey includes two sections for defining balanced information—prenatal and postnatal. You may choose to complete one section or both. This is the POSTNATAL section. Please rate each item as you feel is an essential or important component of a balanced presentation for parents receiving a POSTNATAL diagnosis of Down syndrome for their newborn child. Numerical data and frequencies (%) are provided where available.

8. Rate each item: Genetics of Down syndrome

   Incidence (1/600 to 1/800 births)
   Increasing incidence with increasing maternal age
   Caused by extra genetic material from chromosome 21
   3 Types: Nondisjunction (95%), Translocation (2-4%) Mosaic (2-4%)
   Diagnosis confirmed by chromosome analysis
   Recurrence risk for future pregnancies
   Reproductive options
   Adoption

9. Rate each item: Diagnostic Criteria & Physical Features of Down syndrome

   Flat facial profile (90%)
   Upward slanting, almond-shaped eyes (80%)
   Hypotonia (80%)
   Hyperflexibility (80%)
   Excess skin on back of neck (80%)
   Dysplastic ear (60%)
   Dysplasia of midphalanx of 5^{th} digit (60%)

Small head
Brushfield spots on the iris of the eye
Epicanthal folds
Short, broad hands with short fingers
5th finger clinodactyly
Single palmar crease
Sandal gap between 1st and 2nd toes
Short stature
Enlarged tongue
Syndactyly of 2nd and 3rd toes

10. Rate each item: Medical Complications Associated with Down syndrome

    Early-onset Alzheimer Disease (<5% under 40; 5-15% by age 40-49; 68-80% by age 65)
    Dry, hyperkeratotic skin (75%)
    One or more congenital abnormalities (59%)
    Heart defect possibly requiring open heart surgery (50%)
    Thyroid disorders (up to 50%)
    Obstructive sleep apnea (50-75%)
    Atlantoaxial and/or atlantooccipital instability (1-2%)
    Gastrointestinal defect possibly requiring surgery (12%)
    Autism Spectrum Disorder (ASD) (1%)
    Epilepsy (1-13%)
    Increased risk of childhood leukemia (1%)
    Congenital or acquired cataracts (15%)
    Vision problems (60%)
    Hearing loss (75%)
    Ear problems/infections (up to 70%)
    Increased susceptibility to periodontal disease (23%)
    Increased susceptibility to infection
    Increased risk of autoimmune disease
    Attention deficit hyperactivity disorder (ADHD)
    Respiratory problems
    Orthopedic problems
    Psychiatric disorders
    Tendency for obesity
    Sterility in males
    Increased risk of testicular cancer in males

11. Rate each item: Intellectual Disability & Developmental Delay for Individuals with Down syndrome

    Variable range of intellectual disability from mild to moderate
    Mean (average) IQ of 50
    Range of IQ from 20 to 70

Developmental delay in achieving milestones
Need for physical therapy
Need for occupational therapy
Need for speech & language therapy
Need for early intervention & case management

12. Rate each item: Long-Term Prognosis for Individuals with Down syndrome

    Participate in community sports, activities, & leagues
    Participate in Special Olympics & therapeutic recreation
    Inclusion in regular classes
    Special Education classes
    Finish high school
    Attend college or post-secondary education
    Supported, competitive employment
    Live independently
    Live in group home
    Have friends
    Have intimate relationships
    Get married
    Life expectancy (age range of 50s-60s)
    More like other children than different

13. Rate each item: The Family of Individuals with Down syndrome

    Impact on other siblings – more compassionate & caring
    Impact on other siblings – less attention & resentful
    Impact on marriage – strengthens relationship
    Impact on marriage – strains relationship
    Impact on grandparents – supportive & welcoming
    Impact on grandparents – limited interaction
    Impact on extended family members – supportive & welcoming
    Impact on extended family members – limited interaction
    Impact on other relationships – supportive & welcoming
    Impact on other relationships – lose social circle
    Financial impact – no difference
    Financial impact – more
    Time Commitment – no difference
    Time Commitment – more

14. Rate each item: Informational Resources & Referrals for Individuals with Down syndrome

    Local support group(s)
    National advocacy organizations & websites
    Online support groups/social media platforms

Early intervention centers (Birth to 3-Years programs)
Printed/written material
Photographs of children with Down syndrome
Fact sheets/brochures
Books
Contact with families raising a child with Down syndrome
Adoption agencies
Alternative/nonconventional therapies
Specialist referral(s)
Counselor or family therapist referral(s)
Pastoral counseling referral(s)

FREE RESPONSE SECTION

Please share your thoughts and perspective in response to the following questions. There are no restrictions on the content or length of your responses.

15. Please provide a sample description of what you consider to be a balanced presentation of Down syndrome.
16. What positive aspects of Down syndrome should routinely be included as part of a balanced presentation?
17. There are obvious differences between prenatal & postnatal diagnostic settings. Should there be differences in the content of information provided to parents receiving a prenatal diagnosis versus a postnatal diagnosis? If so, how should information differ between settings and why? If not, why not?
18. What legal and ethical obligations, responsibilities, or duties do medical professionals have in terms of informing new or prospective parents about the array of possible medical complications and health risks associated with Down syndrome?
19. Please share any additional comments, suggestions, experiences, etc. as you see helpful for medical professionals.

DEMOGRAPHIC INFORMATION

20. Your current age (in years)
21. Sex
    a. Male
    b. Female

22. Race/Ethnicity
    a. White
    b. Black/African American/African
    c. American Indian or Alaskan Native
    d. Spanish/Hispanic/Latino

e. Asian Indian
f. Chinese
g. Japanese
h. Other Asian
i. Native Hawaiian or Pacific Islander
j. Other ethnicity (please specify)

23. Level of religious activity
    a. Very active
    b. Occasionally active
    c. Non-practicing
    d. No religion

24. Total number of children you have
    a. 0 children
    b. 1 child
    c. 2 children
    d. 3 children
    e. 4 children
    f. 5 children
    g. 6 children
    h. 7 children
    i. 8 children
    j. 9 children
    k. 10 children
    l. > 10 children

25. Year you completed genetic counseling graduate training (YYYY format)
26. Total number of years in practice since graduation
    a. < 1 year
    b. 1 – 5 years
    c. 6 – 10 years
    d. 11 – 15 years
    e. 16 – 20 years
    f. 21 – 25 years
    g. 26 – 30 years
    h. > 30 years

27. Current U.S. state in which you practice
28. Current work setting
    a. Academic/University-based
    b. Hospital-based
    c. Multiple specialty group
    d. Private practice
    e. Industry
    f. Other (please specify)

29. Current specialty area of practice
    a. Prenatal
    b. General Pediatrics
    c. Pediatric specialty
    d. Cancer
    e. Adult
    f. Infertility
    g. Multiple specialties
    h. Laboratory
    i. Research
    j. Other (please specify)

30. Years of practice in current specialty area
    a. < 1 year
    b. 1 – 5 years
    c. 6 – 10 years
    d. 11 – 15 years
    e. 16 – 20 years
    f. 21 – 25 years
    g. 26 – 30 years
    h. > 30 years

31. Previous areas of practice – check all that apply
    a. Prenatal
    b. General Pediatrics
    c. Pediatric specialty
    d. Cancer
    e. Adult
    f. Infertility
    g. Multiple specialties
    h. Laboratory
    i. Research
    j. Other (please specify)

32. Extent of contact with individuals with Down syndrome in graduate training
    a. No contact
    b. Some contact
    c. Plenty of contact

33. Extent of contact with individuals with Down syndrome in current area of practice
    a. No contact
    b. Some contact
    c. Plenty of contact

34. Extent of contact with individuals with Down syndrome outside of a professional context
    a. No other contact
    b. Social contact only
    c. Family contact only
    d. Both social and family contact

35. Amount of exposure to the NSGC Practice Guidelines for Communicating a Prenatal or Postnatal Diagnosis of Down Syndrome

    a. No exposure
    b. Some exposure
    c. Plenty of exposure

THANK YOU

Thank you for completing our survey. Your participation in this study is greatly appreciated and extremely value to all healthcare professionals involved in distributing information and resources, and in the care of individuals with Down syndrome and their families.

We invite you to share this online survey with other Genetic Counselors. Counselors may access the online survey through Qualtrics.

We also invite you to share a second version of this online survey with parents of children with Down syndrome. Parents may access the online survey through Qualtrics.

Thank you again for helping us define balanced information about Down syndrome.

APPENDIX B: INVITATIONAL LETTER & ONLINE SURVEY FOR PARENTS

INVITATION TO PARTICIPATE

Dear Parent:

Thank you for your interest in this study. You are receiving this letter due to your membership/involvement in a Down syndrome advocacy organization, and because you have a child with Down syndrome. The goal of this Genetic Counseling Master's Thesis Project is to reevaluate the informational needs of parents receiving a diagnosis of Down syndrome. Our initial study was conducted by Katie Berrier Sheets in 2009, and these data were used to help establish the National Society of Genetic Counselors Down Syndrome Practice Guidelines in 2011. We expect the essential information given at the time of a diagnosis has changed over the last several years due to advancements in testing, increasing numbers of college programs, and other opportunities.

We aim to assess perspectives of genetic counselors and parents again in this present study. Survey questions include questions about your child's diagnosis, rating informational items presented during diagnosis disclosure, free response questions, and demographic information questions. Your involvement is extremely valuable to healthcare professionals involved in distributing information and resources and those involved in the care of individuals with Down syndrome and their families.

Participation involves an completing an online survey. The online survey will be available until December 30th, 2019 at midnight, and it will be made available through Qualtrics. The survey will take approximately 20 minutes to complete.

Participation is voluntary. Your consent to participate is given by completing and submitting the online survey. You many choose not to complete the survey and may exit at any time. All responses will be anonymous, and in no way will be identifiable or linked back to you. If you have any questions regarding your rights as a participant in this project, you may contact the University of South Carolina Office of Research Compliance at 803-777-7095.

If you have questions or difficulty accessing the online survey, please reach out to Margaret Wilkes, the primary investigator, at margaret.wilkes@uscmed.sc.edu. Thank you for your participation.

Sincerely,

Margaret J. Wilkes, BA

Genetic Counseling Program

University of South Carolina

YOUR CHILD'S DIAGNOSIS

Please provide information about your experience with receiving your child's diagnosis of Down syndrome.

1. What year was your child's diagnosis made? (YYYY format)

2. Was your child's diagnosis of Down syndrome a prenatal diagnosis or postnatal diagnosis?

    a. Prenatal diagnosis (during pregnancy). Enter the gestational age (weeks into the pregnancy) when the diagnosis was made.
    b. Postnatal diagnosis (after birth). Enter your child's age when the diagnosis was made (in days, weeks, months, or years)

3. Who informed you of your child's diagnosis of Down syndrome?

    a. Obstetrician/Gynecologist (OB/GYN)
    b. Maternal Fetal Medicine (MFM) sub-specialist
    c. Pediatrician
    d. Pediatric Geneticist
    e. Prenatal Genetic Counselor
    f. Pediatric Genetic Counselor
    g. Nurse
    h. Other (please specify)

4. What is your child's biological sex?

    a. Male
    b. Female

5. What number pregnancy was this child for you (your wife/partner)?

    a. Unsure
    b. 1st pregnancy
    c. 2nd pregnancy
    d. 3rd pregnancy
    e. 4th pregnancy
    f. 5th pregnancy
    g. 6th pregnancy
    h. 7th pregnancy
    i. 8th pregnancy
    j. 9th pregnancy
    k. 10th pregnancy

l. > 10th pregnancy

6. What number child was this for you?
    a. 1st child
    b. 2nd child
    c. 3rd child
    d. 4th child
    e. 5th child
    f. 6th child
    g. 7th child
    h. 8th child
    i. 9th child
    j. 10th child
    k. > 10th child

7. Did you receive prenatal screening or prenatal diagnostic testing during your pregnancy with your child with Down syndrome? Check all that apply.

    a. None
    b. First trimester blood screen
    c. First trimester Nuchal Translucency (NT) measurement
    d. Noninvasive Prenatal Screening (NIPS/NIPT)
    e. Second trimester multiple marker blood screen (quad screen)
    f. Second trimester blood screen – AFP only
    g. Chorionic Villus Sampling (CVS)
    h. Amniocentesis
    i. Level II/High resolution ultrasound (2nd trimester anatomy scan)
    j. Unsure, but had some form of prenatal screening

8. How well were you informed about Down syndrome when you were given your child's diagnosis?
    a. Very well-informed
    b. Somewhat informed
    c. Not very informed

9. Did you see a Genetic Counselor regarding your child's diagnosis of Down syndrome?

    a. No genetic counseling (if no genetic counseling, provide clarification: e.g., did not know about it/want it, was not available in my region, etc.)
    b. Yes, prenatal (during pregnancy) genetic counseling only
    c. Yes, pediatric (after birth) genetic counseling only
    d. Yes, both prenatal & pediatric genetic counseling

Comments regarding your genetic counseling experience:

10. Rate your overall satisfaction with your experience of receiving your child's diagnosis.

    a. Extremely satisfied
    b. Somewhat satisfied
    c. Unsure
    d. Somewhat dissatisfied
    e. Extremely dissatisfied

11. Are you a member of a Down syndrome advocacy group?

    a. No
    b. Yes. Please enter the organization's name & year the membership was initiated.

12. Are you involved in an online support group, social media group, or online forum concerning your child's diagnosis? If so, which ones?

    a. No
    b. Yes: list

DEFINING BALANCED INFORMATION ABOUT DOWN SYNDROME

Below is an extensive list of features associated with Down syndrome. We would like to know which of these are essential for medical professionals to include in the first discussion with new or expectant parents receiving a diagnosis. Please rate each item as you feel is an essential or important component of a balanced presentation or discussion of Down syndrome. Numerical data and frequencies (%) are provided where available.

13. Rate each item: Genetics of Down syndrome

    Incidence (1/600 to 1/800 births)
    Increasing chance if mother is 35 years old or older
    Caused by extra genetic material from chromosome 21
    3 Types: Nondisjunction (95%), Translocation (2-4%) Mosaic (2-4%)
    Diagnosis confirmed by chromosome analysis
    Recurrence risk for future pregnancies
    Reproductive options
    Adoption

14. Rate each item: Diagnostic Criteria & Physical Features of Down syndrome

    Flattened nose/flat facial profile (90%)
    Upward slanting, almond-shaped eyes (80%)
    Low muscle tone/hypotonia (80%)

Increased flexibility (80%)
Excess skin on back of neck (80%)
Small, simple ears (60%)
Poorly developed 5th finger (60%)
Small head
Brushfield spots on the iris of the eye
Skin folds of upper eyelid covering inner corner of the eye/epicanthal folds
Short, broad hands with short fingers
5th finger curves inward
Single crease on palm of hand
Wide gap between 1st and 2nd toes/sandal gap
Short stature
Enlarged tongue
Fusion/syndactyly of 2nd and 3rd toes

15. Rate each item: Medical Complications Associated with Down syndrome

    Early-onset Alzheimer Disease (<5% under 40; 5-15% by age 40-49; 68-80% by age 65)
    Dry, hardened, flaky skin (75%)
    One or more abnormalities present at birth (50%)
    Heart defect possibly requiring open heart surgery (50%)
    Thyroid disorders (up to 50%)
    Obstructive sleep apnea/pauses in breathing during sleep (50-75%)
    Atlantoaxial and/or atlantooccipital instability (1-2%)
    Blockage in stomach or intestines possibly requiring surgery (12%)
    Autism Spectrum Disorder (ASD) (1%)
    Seizures/epilepsy (1-13%)
    Increased risk of childhood leukemia (1%)
    Cataracts present at birth or developed over time (15%)
    Vision problems (60%)
    Hearing loss (75%)
    Ear problems/infections (up to 70%)
    Increased susceptibility to dental problems (23%)
    Increased susceptibility to infection
    Increased risk of autoimmune disease
    Attention deficit hyperactivity disorder (ADHD)
    Breathing/respiratory problems
    Problems with muscles and/or skeletal system
    Mental health conditions
    Tendency for obesity/weight gain
    Males reduced ability/unable to reproduce
    Increased risk of testicular cancer in males
16. Rate each item: Intellectual Disability & Developmental Delay for Individuals with Down syndrome

Variable range of intellectual disability from mild to moderate
Mean (average) IQ of 50
Range of IQ from 20 to 70
Developmental delay in achieving milestones
Need for physical therapy
Need for occupational therapy
Need for speech & language therapy
Need for early intervention & case management

17. Rate each item: Long-Term Prognosis for Individuals with Down syndrome

Participate in community sports, activities, & leagues
Participate in Special Olympics & therapeutic recreation
Inclusion in regular classes
Special Education classes
Finish high school
Attend college or post-secondary education
Supported, competitive employment
Live independently
Live in group home
Have friends
Have intimate relationships
Get married
Life expectancy (age range of 50s-60s)
More like other children than different

18. Rate each item: The Family of Individuals with Down syndrome

Impact on other siblings – more compassionate & caring
Impact on other siblings – less attention & resentful
Impact on marriage – strengthens relationship
Impact on marriage – strains relationship
Impact on grandparents – supportive & welcoming
Impact on grandparents – limited interaction
Impact on extended family members – supportive & welcoming
Impact on extended family members – limited interaction
Impact on other relationships – supportive & welcoming
Impact on other relationships – lose social circle
Financial impact – no difference
Financial impact – more
Time commitment – no difference
Time commitment – more

19. Rate each item: Informational Resources & Referrals for Individuals with Down syndrome

Local support group(s)

National advocacy organizations & websites
Online support groups/social media platforms
Early intervention centers (Birth to 3-Years programs)
Printed/written material
Photographs of children with Down syndrome
Fact sheets/brochures
Books
Contact with families raising a child with Down syndrome
Adoption agencies
Alternative/nonconventional therapies
Specialist referral(s)
Counselor or family therapist referral(s)
Pastoral counseling referral(s)

FREE RESPONSE SECTION

Please share your thoughts & perspective in response to the following questions. There are no restrictions on the content or length of your responses.

20. There is a need for medical professionals to include more of the positive aspects of Down syndrome when describing the condition to new or expectant parents. What positive aspects of Down syndrome should routinely be included as part of a balanced presentation?

21. Please comment on the presentation of your child's diagnosis, the information you were given about Down syndrome at that time, & your overall satisfaction with your experience.

22. Consider your own personal experience of receiving your child's diagnosis. Do you feel you were given balanced and accurate information about what to expect from raising a child with Down syndrome? If yes, what information was most useful for you? If no, what information should have been provided that was not?

23. Consider your own personal experience of receiving your child's diagnosis. What could have been done differently in order to improve the experience?

24. There are obvious differences between prenatal & postnatal diagnostic settings: In a prenatal setting the Genetic Counselor should provide balanced, adequate information for informed decision-making. Information is general & based on statistics. There is a time constraint on prenatal decision-making. In a postnatal setting, the child is already here. The Genetic Counselor should focus on supporting the family's needs. Information is specific for that child & their unique characteristics. Decision-making involves treatment & management options. Understanding these differences, should there be variation in the content of information provided to parents receiving a prenatal diagnosis for their unborn

child, versus parents receiving a postnatal diagnosis for their newborn? Or should the information be the same in both prenatal & postnatal settings?

25. Please share any additional comments, suggestions, experiences, etc. as you see helpful for medical professionals.

DEMOGRAPHIC INFORMATION

26. Your Current Age (in years)

27. Sex

    a. Male
    b. Female

28. Ethnicity/Race

    a. White
    b. Black/African American/African
    c. American Indian or Alaskan Native
    d. Spanish/Hispanic/Latino
    e. Asian Indian
    f. Chinese
    g. Japanese
    h. Other Asian
    i. Native Hawaiian or Pacific Islander
    j. Other (please specify)

29. Level of Religious Activity

    a. Very active
    b. Occasionally active
    c. Non-practicing
    d. No religion

30. Highest Level of Education

    a. No education
    b. Middle school
    c. Some high school
    d. High school graduate (e.g., Diploma or GED)
    e. Some college
    f. Associate degree (e.g., AA, AS)
    g. Bachelor's degree (e.g., BS, BA)

h. Master's degree (e.g., MA, MS, MBA)
i. Professional degree (e.g., MD, DDS, DVM, LLB, JD)
j. Doctorate degree (e.g., PhD)
k. Other (please specify)

31. Marital Status

a. Married
b. Widowed
c. Divorced
d. Separated
e. Never married
f. Civil union

32. Total Number of Children You Have

a. 1 child
b. 2 children
c. 3 children
d. 4 children
e. 5 children
f. 6 children
g. 7 children
h. 8 children
i. 9 children
j. 10 children
k. > 10 children

33. Current state/place of residency

34. Combined household income

a. ≤ $24,999
b. $25,000 - $49,999
c. $50,000 - $74,999
d. $75,000 - $99,999
e. $100,000 - $124,999
f. $125,000 - $149,999
g. ≥ $150,000

THANK YOU

Thank you for completing our survey. Your participation in this study is greatly appreciated and extremely valuable to all healthcare professionals involved in distributing

information and resources, and in the care of individuals with Down syndrome and their families.

We invite you to share this online survey with other parents of children with Down syndrome. Parents may access the online survey through Qualtrics.

Thank you again for helping us define balanced information about Down syndrome.

APPENDIX C – ITEMS RATED ESSENTIAL BY EACH GROUP

MAJORITY

Informational Item	Group	% Essential (n)	% Important (n)	% Not Important (n)	Total *n*	Average Rating‡
Caused by extra material from chromosome 21	Prenatal GC	81.0% (34)	16.7% (7)	2.4% (1)	42	2.79
	Postnatal GC	85.2% (52)	14.8% (9)	0.0% (0)	61	2.85
	Prenatal Parent	75.7% (165)	19.7% (43)	4.6% (10)	218	2.71
	Postnatal Parent	77.3% (187)	18.6% (45)	4.1% (10)	242	2.73
Diagnosis confirmed by chromosome analysis	Prenatal GC	97.6% (41)	2.4% (1)	0.0% (0)	42	2.98
	Postnatal GC	85.2% (52)	13.1% (8)	1.6% (1)	61	2.84
	Prenatal Parent	72.3% (154)	22.1% (47)	5.6% (12)	213	2.67
	Postnatal Parent	79.8% (194)	16.9% (41)	3.3% (8)	243	2.77
Hypotonia (80%)	Prenatal GC	64.3% (27)	33.3% (14)	2.4% (1)	42	2.62
	Postnatal GC	75.4% (46)	19.7% (12)	4.9% (3)	61	2.70
	Prenatal Parent	68.3% (138)	26.7% (54)	5.0% (10)	202	2.63
	Postnatal Parent	69.7% (154)	23.5% (52)	6.8% (15)	221	2.63
One or more congenital abnormalities (50%)	Prenatal GC	90.5% (38)	2.4% (1)	7.1% (3)	42	2.83
	Postnatal GC	88.5% (54)	9.8% (6)	1.6% (1)	61	2.87
	Prenatal Parent	64.6% (122)	30.7% (58)	4.8% (9)	189	2.60
	Postnatal Parent	59.2% (125)	31.3% (66)	9.5% (20)	211	2.50
Heart defect possibly requiring surgery (40-60%)	Prenatal GC	95.2% (40)	0.0% (0)	4.8% (2)	42	2.90
	Postnatal GC	95.1% (58)	4.9% (3)	0.0% (0)	61	2.95
	Prenatal Parent	89.2% (174)	9.7% (19)	1.0% (2)	195	2.88
	Postnatal Parent	85.4% (182)	10.8% (23)	3.8% (8)	213	2.82
Gastrointestinal defect possibly requiring surgery (12%)	Prenatal GC	52.4% (22)	40.5% (17)	7.1% (3)	42	2.45
	Postnatal GC	56.7% (34)	35.0% (21)	8.3% (5)	60	2.48
	Prenatal Parent	62.2% (120)	28.5% (55)	9.3% (18)	193	2.53
	Postnatal Parent	55.3% (115)	35.1% (73)	9.6% (20)	208	2.46
Hearing loss (75%)	Prenatal GC	51.2% (21)	41.5% (17)	7.3% (3)	41	2.44
	Postnatal GC	76.3% (45)	18.6% (11)	5.1% (3)	59	2.71
	Prenatal Parent	66.3% (128)	27.5% (53)	6.2% (12)	193	2.60
	Postnatal Parent	64.0% (135)	27.5% (58)	8.5% (18)	211	2.55
Variable range of intellectual disability from mild to moderate	Prenatal GC	92.9% (39)	7.1% (3)	0.0% (0)	42	2.93
	Postnatal GC	96.8% (60)	3.2% (2)	0.0% (0)	62	2.97
	Prenatal Parent	70.2% (127)	24.9% (45)	5.0% (9)	181	2.65
	Postnatal Parent	68.7% (138)	24.4% (49)	7.0% (14)	201	2.62
Developmental delay in achieving milestones	Prenatal GC	92.9% (39)	7.1% (3)	0.0% (0)	42	2.93
	Postnatal GC	98.4% (61)	1.6% (1)	0.0% (0)	62	2.98
	Prenatal Parent	77.6% (142)	17.5% (32)	4.9% (9)	183	2.73
	Postnatal Parent	66.7% (136)	28.4% (58)	4.9% (10)	204	2.62
Need for physical therapy	Prenatal GC	66.7% (28)	31.0% (13)	2.4% (1)	42	2.64
	Postnatal GC	83.9% (52)	11.3% (7)	4.8% (3)	62	2.79
	Prenatal Parent	77.6% (142)	19.1% (35)	3.3% (6)	183	2.74
	Postnatal Parent	77.0% (157)	19.1% (39)	3.9% (8)	204	2.73
Need for occupational therapy	Prenatal GC	66.7% (28)	31.0% (13)	2.4% (1)	42	2.64
	Postnatal GC	83.9% (52)	11.3% (7)	4.8% (3)	62	2.79
	Prenatal Parent	77.6% (142)	19.1% (35)	3.3% (6)	183	2.74
	Postnatal Parent	76.1% (156)	20.0% (41)	3.9% (8)	205	2.72
Need for speech and language therapy	Prenatal GC	66.7% (28)	31.0% (13)	2.4% (1)	42	2.64
	Postnatal GC	85.5% (53)	9.7% (6)	4.8% (3)	62	2.81
	Prenatal Parent	79.3% (146)	17.9% (33)	2.7% (5)	184	2.77
	Postnatal Parent	77.6% (159)	19.0% (39)	3.4% (7)	205	2.74
	Prenatal GC	90.5% (38)	9.5% (4)	0.0% (0)	42	2.90
	Postnatal GC	88.7% (55)	9.7% (6)	1.6% (1)	62	2.87

Need for early intervention and case management	Prenatal Parent	87.5% (161)	10.3% (19)	2.2% (4)	184	2.85
	Postnatal Parent	87.3% (179)	11.2% (23)	1.5% (3)	205	2.86
Participate in community sports, activities, and leagues	Prenatal GC	69.0% (29)	28.6% (12)	2.4% (1)	42	2.67
	Postnatal GC	71.7% (43)	21.7% (13)	6.7% (4)	60	2.65
	Prenatal Parent	72.8% (131)	23.9% (43)	3.3% (6)	180	2.69
	Postnatal Parent	67.2% (135)	28.9% (58)	4.0% (8)	201	2.63
Inclusion in regular classes	Prenatal GC	73.8% (31)	26.2% (11)	0.0% (0)	42	2.74
	Postnatal GC	58.3% (35)	33.3% (20)	8.3% (5)	60	2.50
	Prenatal Parent	82.2% (148)	16.1% (29)	1.7% (3)	180	2.81
	Postnatal Parent	77.9% (155)	18.1% (36)	4.0% (8)	199	2.74
Special education classes	Prenatal GC	73.8% (31)	26.2% (11)	0.0% (0)	42	2.74
	Postnatal GC	78.7% (48)	18.0% (11)	3.3% (2)	61	2.75
	Prenatal Parent	54.7% (98)	38.5% (69)	6.7% (12)	179	2.48
	Postnatal Parent	57.1% (112)	33.2% (65)	9.7% (19)	196	2.47
Complete high school	Prenatal GC	59.5% (25)	35.7% (15)	4.8% (2)	42	2.55
	Postnatal GC	55.0% (33)	38.3% (23)	6.7% (4)	60	2.48
	Prenatal Parent	77.1% (138)	19.6% (35)	3.4% (6)	179	2.74
	Postnatal Parent	72.5% (145)	22.5% (45)	5.0% (10)	200	2.68
Supported, competitive employment	Prenatal GC	66.7% (28)	31.0% (13)	2.4% (1)	42	2.64
	Postnatal GC	57.4% (35)	36.1% (22)	6.6% (4)	61	2.51
	Prenatal Parent	76.7% (138)	20.0% (36)	3.3% (6)	180	2.73
	Postnatal Parent	73.4% (146)	21.1% (42)	5.5% (11)	199	2.68
Live independently	Prenatal GC	66.7% (28)	33.3% (14)	0.0% (0)	42	2.67
	Postnatal GC	59.0% (36)	34.4% (21)	6.6% (4)	61	2.52
	Prenatal Parent	76.7% (138)	20.6% (37)	2.8% (5)	180	2.74
	Postnatal Parent	68.5% (135)	28.4% (56)	3.0% (6)	197	2.65
Have friends	Prenatal GC	78.6% (33)	19.0% (8)	2.4% (1)	42	2.76
	Postnatal GC	82.0% (50)	14.8% (9)	3.3% (2)	61	2.79
	Prenatal Parent	87.8% (158)	11.7% (21)	0.6% (1)	180	2.87
	Postnatal Parent	86.5% (173)	11.5% (23)	2.0% (4)	200	2.85
Have intimate relationships	Prenatal GC	55.0% (22)	42.5% (17)	2.5% (1)	40	2.53
	Postnatal GC	51.7% (30)	37.9% (22)	10.3% (6)	58	2.41
	Prenatal Parent	67.2% (121)	27.2% (49)	5.6% (10)	180	2.62
	Postnatal Parent	62.9% (124)	29.9% (59)	7.1% (14)	197	2.56
Life Expectancy (age range of 50s-60s)	Prenatal GC	80.5% (33)	19.5% (8)	0.0% (0)	41	2.80
	Postnatal GC	70.0% (42)	28.3% (17)	1.7% (1)	60	2.68
	Prenatal Parent	58.3% (105)	33.3% (60)	8.3% (15)	180	2.50
	Postnatal Parent	60.3% (120)	30.7% (61)	9.0% (18)	199	2.51
More like other children than different	Prenatal GC	88.1% (37)	7.1% (3)	4.8% (2)	42	2.83
	Postnatal GC	90.0% (54)	10.0% (6)	0.0% (0)	60	2.90
	Prenatal Parent	90.5% (162)	8.4% (15)	1.1% (2)	179	2.89
	Postnatal Parent	90.5% (181)	8.0% (16)	1.5% (3)	200	2.89
Local support group(s)	Prenatal GC	88.1% (37)	11.9% (5)	0.0% (0)	42	2.88
	Postnatal GC	95.1% (58)	4.9% (3)	0.0% (0)	61	2.95
	Prenatal Parent	92.3% (156)	7.1% (12)	0.6% (1)	169	2.92
	Postnatal Parent	87.0% (161)	11.9% (22)	1.1% (2)	185	2.86
National advocacy organizations and websites	Prenatal GC	71.4% (30)	28.6% (12)	0.0% (0)	42	2.71
	Postnatal GC	73.8% (45)	24.6% (15)	1.6% (1)	61	2.72
	Prenatal Parent	82.1% (138)	16.7% (28)	1.2% (2)	168	2.81
	Postnatal Parent	83.8% (155)	15.1% (28)	1.1% (2)	185	2.83
Online support groups/social media platforms	Prenatal GC	66.7% (28)	33.3% (14)	0.0% (0)	42	2.67
	Postnatal GC	60.7% (37)	39.3% (24)	0.0% (0)	61	2.61
	Prenatal Parent	88.6% (148)	10.8% (18)	0.6% (1)	167	2.88
	Postnatal Parent	81.5% (150)	17.4% (32)	1.1% (2)	184	2.80
Early Intervention centers	Prenatal GC	76.2% (32)	21.4% (9)	2.4% (1)	42	2.74
	Postnatal GC	82.0% (50)	9.8% (6)	8.2% (5)	61	2.74
	Prenatal Parent	94.7% (161)	4.1% (7)	1.2% (2)	170	2.94
	Postnatal Parent	93.0% (173)	6.5% (12)	0.5% (1)	186	2.92
Printed/written materials	Prenatal GC	76.2% (32)	23.8% (10)	0.0% (0)	42	2.76
	Postnatal GC	82.0% (50)	14.8% (9)	3.3% (2)	61	2.79
	Prenatal Parent	69.9% (116)	27.7% (46)	2.4% (4)	166	2.67
	Postnatal Parent	71.7% (132)	25.0% (46)	3.3% (6)	184	2.68
Fact sheets or Brochures	Prenatal GC	69.0% (29)	31.0% (13)	0.0% (0)	42	2.69
	Postnatal GC	54.2% (32)	35.6% (21)	10.2% (6)	59	2.44
	Prenatal Parent	68.9% (115)	29.3% (49)	1.8% (3)	167	2.67
	Postnatal Parent	70.3% (128)	24.2% (44)	5.5% (10)	182	2.65

Specialist referral(s)	Prenatal GC	58.5% (24)	29.3% (12)	12.2% (5)	41	2.46
	Postnatal GC	74.6% (44)	15.3% (9)	10.2% (6)	59	2.64
	Prenatal Parent	75.3% (125)	21.7% (36)	3.0% (5)	166	2.72
	Postnatal Parent	79.9% (147)	15.2% (28)	4.9% (9)	184	2.75

APPENDIX D – INFORMATIONAL ITEMS IN RANK ORDER

Rank	*Prenatal GC*		*Postnatal GC*		*Prenatal Parent*		*Postnatal Parent*	
	Item	**Rating**	**Item**	**Rating**	**Item**	**Rating**	**Item**	**Rating**
1	Confirm Diagnosis by Chromosome Analysis	2.98	DD in Achieving Milestones	2.98	Early Intervention Centers	2.94	Early Intervention Centers	2.92
2	Variable Range of ID (Mild-Moderate)	2.93	Variable Range of ID (Mild-Moderate)	2.97	Contact with Families Raising a Child with DS	2.93	More Alike Than Different	2.89
3	DD in Achieving Milestones	2.93	Heart Defect Possibly Requiring Surgery	2.95	Local Support Groups	2.92	Need for Early Intervention and Case Management	2.86
4	Heart Defect Possibly Requiring Surgery	2.90	Local Support Groups	2.95	More Alike Than Different	2.89	Local Support Groups	2.86
5	Need for Early Intervention and Case Management	2.90	More Alike Than Different	2.90	Heart Defect Possibly Requiring Surgery	2.88	Contact with Families Raising a Child with DS	2.86
6	Local Support Groups	2.88	One or More Congenital Abnormalities	2.87	Online Support Groups / Social Media	2.88	Have Friends	2.85
7	One or More Congenital Abnormalities	2.83	Need for Early Intervention and Case Management	2.87	Have Friends	2.87	Advocacy Organizations / Websites	2.83
8	More Alike Than Different	2.83	Caused by Extra Material from Chromosome 21	2.85	Need for Early Intervention and Case Management	2.85	Heart Defect Possibly Requiring Surgery	2.82
9	Life Expectancy	2.80	Confirm Diagnosis by Chromosome Analysis	2.84	Inclusion in Regular Classes	2.81	Online Support Groups / Social Media	2.80
10	Caused by Extra Material from Chromosome 21	2.79	Need for Speech & Language Therapy	2.81	Advocacy Organizations / Websites	2.81	Confirm Diagnosis by Chromosome Analysis	2.77
11	Have Friends	2.76	Need for Physical Therapy	2.79	Impact on Other Siblings – More Compassionate & Caring	2.79	Specialist Referral(s)	2.75
12	Printed / Written Material	2.76	Need for Occupational Therapy	2.79	Need for Speech & Language Therapy	2.77	Need for Speech & Language Therapy	2.74
13	Inclusion in Regular Classes	2.74	Have Friends	2.79	Need for Physical Therapy	2.74	Inclusion in Regular Classes	2.74
14	Special Education Classes	2.74	Printed / Written Material	2.79	Need for Occupational Therapy	2.74	Caused by Extra Material from Chromosome 21	2.73
15	Early Intervention Centers	2.74	Special Education Classes	2.75	Finish High School	2.74	Need for Physical Therapy	2.73
16	Advocacy Organizations / Websites	2.71	Early Intervention Centers	2.74	Live Independently	2.74	Need for Occupational Therapy	2.72

17	Live in Group Home	2.69	Recurrence Risk	2.72	DD in Achieving Milestones	2.73	Finish High School	2.68
18	Fact Sheets / Brochures	2.69	Advocacy Organizations / Websites	2.72	Supported, Competitive Employment	2.73	Supported, Competitive Employment	2.68
19	Participate in Community Sports, Activities, & Leagues	2.67	Hearing Loss	2.71	Specialist Referral(s)	2.72	Impact on Other Siblings – More Compassionate & Caring	2.68
20	Live Independently	2.67	Hypotonia	2.70	Caused by Extra Material from Chromosome 21	2.71	Printed / Written Material	2.68
21	Online Support Groups / Social Media	2.67	Life Expectancy	2.68	Participate in Community Sports, Activities, & Leagues	2.69	Live Independently	2.65
22	Need for Physical Therapy	2.64	Vision Problems	2.67	Books	2.69	Fact Sheets / Brochures	2.65
23	Need for Occupational Therapy	2.64	Thyroid Disorders	2.65	Confirm Diagnosis by Chromosome Analysis	2.67	Hypotonia	2.63
24	Need for Speech & Language Therapy	2.64	Participate in Community Sports, Activities, & Leagues	2.65	Printed / Written Material	2.67	Participate in Community Sports, Activities, & Leagues	2.63
25	Supported, Competitive Employment	2.64	Specialist Referral(s)	2.64	Fact Sheets / Brochures	2.67	Variable Range of ID (Mild-Moderate)	2.62
26	Hypotonia	2.62	Online Support Groups / Social Media	2.61	Obstructive Sleep Apnea	2.66	DD in Achieving Milestones	2.62
27	Reproductive Options	2.57	Ear Problems	2.57	Variable Range of ID (Mild-Moderate)	2.65	Thyroid Disorders	2.60
28	Finish High School	2.55	Live in Group Home	2.56	Impact on Marriage – Strengthens Relationship	2.64	Obstructive Sleep Apnea	2.60
29	Intimate Relationships	2.53	Live Independently	2.52	Hypotonia	2.63	Intimate Relationships	2.56
30	Recurrence Risk	2.48	Obstructive Sleep Apnea	2.51	Attend College or Post-Secondary Education	2.63	Hearing Loss	2.55
31	Specialist Referral(s)	2.46	Supported, Competitive Employment	2.51	Thyroid Disorders	2.62	Books	2.55
32	Gastrointestinal Defect Possibly Requiring Surgery	2.45	Inclusion in Regular Classes	2.50	Intimate Relationships	2.62	Impact on Marriage – Strengthens Relationship	2.54
33	Participate in Special Olympics & Therapeutic Recreation	2.45	Gastrointestinal Defect Possibly Requiring Surgery	2.48	One or More Congenital Abnormalities	2.60	Ear Problems	2.53
34	Hearing Loss	2.44	Finish High School	2.48	Hearing Loss	2.60	Respiratory Problems	2.52
35	Vision Problems	2.39	Contact with Families Raising a Child with DS	2.48	Participate in Special Olympics & Therapeutic Recreation	2.60	Life Expectancy	2.51
36	Attend College or Post-	2.37	Reproductive Options	2.45	Counselor or Family Therapist Referral(s)	2.56	One or More Congenital Abnormalities	2.50

	Secondary Education							
37	Contact with Families Raising a Child with DS	2.37	Fact Sheets / Brochures	2.44	Impact on Grandparents – Supportive & Welcoming	2.55	Participate in Special Olympics & Therapeutic Recreation	2.50
38	Books	2.34	Intimate Relationships	2.41	Ear Problems	2.54	Vision Problems	2.49
39	Adoption	2.33	Impact on Other Siblings – More Compassionate & Caring	2.30	Gastrointestinal Defect Possibly Requiring Surgery	2.53	Attend College or Post-Secondary Education	2.48
40	Thyroid Disorders	2.33	Attend College or Post-Secondary Education	2.29	Respiratory Problems	2.53	Special Education Classes	2.47
41	Get Married	2.32	Adoption	2.28	Photographs of Children with DS	2.53	Gastrointestinal Defect Possibly Requiring Surgery	2.46
42	Impact on Other Siblings – More Compassionate & Caring	2.32	Get Married	2.24	Vision Problems	2.52	Counselor or Family Therapist Referral(s)	2.45
43	Range of IQ	2.29	Participate in Special Olympics & Therapeutic Recreation	2.23	Get Married	2.52	Increased Susceptibility to Infection	2.44
44	Time Commitment – More	2.29	3 Types: Nondisjunction, Translocation, Mosaic	2.21	Impact on Other Relationships – Supportive & Welcoming	2.52	Get Married	2.43
45	Impact on Marriage – Strengthens Relationship	2.26	Books	2.21	Life Expectancy	2.50	Impact on Grandparents – Supportive & Welcoming	2.40
46	Short Stature	2.19	Early-Onset Alzheimer Disease	2.17	Special Education Classes	2.48	Time Commitment – More	2.40
47	Obstructive Sleep Apnea	2.19	Impact on Other Siblings – Less Attention & Resentful	2.16	Live in Group Home	2.47	Impact on Other Relationships – Supportive & Welcoming	2.37
48	Ear Problems	2.19	Time Commitment – More	2.16	Impact on Extended Family Members – Supportive & Welcoming	2.47	Recurrence Risk	2.35
49	Impact on Marriage – Strains Relationship	2.18	Range of IQ	2.11	Increased Susceptibility to Infection	2.46	Increased Flexibility	2.35
50	Impact on Other Siblings – Less Attention & Resentful	2.17	Impact on Marriage – Strains Relationship	2.11	Time Commitment – More	2.45	Increased Risk of Autoimmune Disease	2.35
51	Mean IQ	2.15	More Financial Impact	2.11	3 Types: Nondisjunction, Translocation, Mosaic	2.39	Impact on Extended Family Members – Supportive & Welcoming	2.34
52	Increased Chance with Advanced Maternal Age	2.14	Impact on Marriage – Strengthens Relationship	2.09	Orthopedic Problems	2.39	Orthopedic Problems	2.33
53	3 Types: Nondisjunction,	2.14	Short Stature	2.07	Early-Onset Alzheimer Disease	2.38	Live in Group Home	2.33

	Translocation, Mosaic							
54	More Financial Impact	2.10	Photographs of Children with DS	2.07	Recurrence Risk	2.36	Photographs of Children with DS	2.33
55	Early-Onset Alzheimer Disease	2.07	Increased Risk of Childhood Leukemia	2.05	Increased Risk of Autoimmune Disease	2.35	3 Types: Nondisjunction, Translocation, Mosaic	2.28
56	Counselor or Family Therapist Referral(s)	2.05	Mean IQ	2.05	Pastoral Counseling Referral(s)	2.33	Atlantoaxial and / or Atlantooccipital Instability	2.26
57	Increased Susceptibility to Infection	2.02	Increased Chance with Advanced Maternal Age	2.03	More Financial Impact	2.31	Early-Onset Alzheimer Disease	2.23
58	Photographs of Children with DS	2.02	Counselor or Family Therapist Referral(s)	2.03	Increased Risk of Childhood Leukemia	2.29	Enlarged Tongue	2.22
59	Upward slanting, almond-shaped eyes	1.98	Adoption Agencies	2.00	Increased Flexibility	2.28	Increased Susceptibility to Dental Problems	2.19
60	Increased Risk of Childhood Leukemia	1.98	Upward slanting, almond-shaped eyes	1.95	Enlarged Tongue	2.28	Tendency for Obesity	2.17
61	Respiratory Problems	1.98	Epilepsy	1.95	Alternative / Nonconventional Therapies	2.25	Pastoral Counseling Referral(s)	2.17
62	Adoption Agencies	1.98	Cataracts	1.93	Epilepsy	2.24	Epilepsy	2.16
63	Increased Flexibility	1.95	Increased Flexibility	1.92	Atlantoaxial and / or Atlantooccipital Instability	2.22	More Financial Impact	2.15
64	Increased Risk of Autoimmune Disease	1.90	Enlarged Tongue	1.88	Increased Susceptibility to Dental Problems	2.22	Impact on Marriage – Strains Relationship	2.14
65	Flat Facial Profile	1.88	Sterility in Males	1.88	Financial Impact – No Difference	2.22	Increased Risk of Childhood Leukemia	2.13
66	Epilepsy	1.88	Flat Facial Profile	1.87	Cataracts	2.20	Cataracts	2.11
67	Mental Health Conditions	1.88	Autism	1.86	Tendency for Obesity	2.20	Impact on Other Siblings – Less Attention & Resentful	2.10
68	Impact on Extended Family Members – Limited Interaction	1.87	Tendency for Obesity	1.86	Increased Risk of Testicular Cancer in Males	2.17	Alternative / Nonconventional Therapies	2.10
69	Enlarged Tongue	1.86	Atlantoaxial and / or Atlantooccipital Instability	1.85	Impact on Marriage – Strains Relationship	2.16	Financial Impact – No Difference	2.07
70	Atlantoaxial and / or Atlantooccipital Instability	1.85	Respiratory Problems	1.85	Time Commitment – No Difference	2.14	Mental Health Conditions	2.06
71	Time Commitment – No Difference	1.84	Increased Susceptibility to Infection	1.83	Adoption	2.13	Increased Risk of Testicular Cancer in Males	2.05
72	Orthopedic Problems	1.83	Financial Impact – No Difference	1.82	Mental Health Conditions	2.13	Incidence	2.03
73	Impact on Grandparents – Supportive & Welcoming	1.82	Impact on Other Relationships – Supportive & Welcoming	1.80	Impact on Other Siblings – Less Attention & Resentful	2.13	Time Commitment – No Difference	2.01

74	Impact on Extended Family Members – Limited Interaction	1.82	Increased Risk of Autoimmune Disease	1.78	Adoption Agencies	2.09	Short Stature	1.98
75	Impact on Other Relationships – Supportive & Welcoming	1.82	Impact on Other Relationships – Lose Social Circle	1.78	Autism	2.05	Upward slanting, almond-shaped eyes	1.96
76	Pastoral Counseling Referral(s)	1.82	Time Commitment – No Difference	1.77	Reproductive Options	2.04	Impact on Other Relationships – Lose Social Circle	1.95
77	Excess Skin on Back of Neck	1.81	Mental Health Conditions	1.76	ADHD	2.04	Hyperkeratotic Skin	1.94
78	Cataracts	1.79	Impact on Grandparents – Supportive & Welcoming	1.75	Sterility in Males	2.03	ADHD	1.94
79	Impact on Grandparents – Limited Interaction	1.79	Impact on Extended Family Members – Supportive & Welcoming	1.74	Impact on Other Relationships – Lose Social Circle	2.02	Adoption	1.92
80	Impact on Other Relationships – Lose Social Circle	1.78	Excess Skin on Back of Neck	1.72	Incidence	2.00	Autism	1.92
81	Autism	1.76	Impact on Grandparents – Limited Interaction	1.72	Impact on Grandparents – Limited Interaction	1.99	Flat Facial Profile	1.91
82	Sterility in Males	1.74	Pastoral Counseling Referral(s)	1.72	Increased Chance with Advanced Maternal Age	1.95	Sterility in Males	1.90
83	Financial Impact – No Difference	1.74	Orthopedic Problems	1.71	Short Stature	1.95	Adoption Agencies	1.89
84	ADHD	1.71	Impact on Extended Family Members – Limited Interaction	1.71	Impact on Extended Family Members – Limited Interaction	1.94	Impact on Grandparents – Limited Interaction	1.87
85	Tendency for Obesity	1.71	Incidence	1.71	Range of IQ	1.93	Range of IQ	1.86
86	Small Head	1.69	Epicanthal Folds	1.68	Small Head	1.89	Mean IQ	1.85
87	Increased Susceptibility to Dental Problems	1.67	Hyperkeratotic Skin	1.65	Upward slanting, almond-shaped eyes	1.87	Small Head	1.84
88	Increased Risk of Testicular Cancer in Males	1.66	Increased Susceptibility to Dental Problems	1.65	Mean IQ	1.87	Impact on Extended Family Members – Limited Interaction	1.84
89	Small, Simple / Dysplastic Ears	1.64	Single Palmar Crease	1.62	Small, Simple / Dysplastic Ears	1.86	Increased Chance with Advanced Maternal Age	1.83
90	Epicanthal Folds	1.64	ADHD	1.62	Hyperkeratotic Skin	1.86	Small, Simple / Dysplastic Ears	1.83
91	Incidence	1.62	Short, Broad Hands with Short Fingers	1.61	Excess Skin on Back of Neck	1.82	Excess Skin on Back of Neck	1.81
92	Single Palmar Crease	1.61	Small Head	1.60	Flat Facial Profile	1.81	Reproductive Options	1.80
93	Alternative / Nonconventional Therapies	1.61	Increased Risk of Testicular Cancer in Males	1.60	Epicanthal Folds	1.79	Short, Broad Hands with Short Fingers	1.79

94	Hyperkeratotic Skin	1.57	Sandal Gap Between 1st & 2nd Toes	1.54	Dysplastic Midphalanx of 5th Digit	1.76	Sandal Gap Between 1st & 2nd Toes	1.78
95	Short, Broad Hands with Short Fingers	1.55	Small, Simple / Dysplastic Ears	1.52	Brushfield Spots on the Iris of the Eye	1.74	Single Palmar Crease	1.76
96	Sandal Gap Between 1st & 2nd Toes	1.50	Brushfield Spots on the Iris of the Eye	1.48	Short, Broad Hands with Short Fingers	1.74	Epicanthal Folds	1.75
97	5th Finger Clinodactyly	1.43	Alternative / Nonconventional Therapies	1.48	Single Palmar Crease	1.71	Brushfield Spots on the Iris of the Eye	1.72
98	Dysplastic Midphalanx of 5th Digit	1.38	Syndactyly of 2nd and 3rd Toes	1.47	Syndactyly of 2nd and 3rd Toes	1.71	Dysplastic Midphalanx of 5th Digit	1.71
99	Brushfield Spots on the Iris of the Eye	1.38	5th Finger Clinodactyly	1.45	Sandal Gap Between 1st & 2nd Toes	1.67	Syndactyly of 2nd and 3rd Toes	1.71
100	Syndactyly of 2nd and 3rd Toes	1.38	Dysplastic Midphalanx of 5th Digit	1.43	5th Finger Clinodactyly	1.61	5th Finger Clinodactyly	1.64

APPENDIX E – SIGNIFICANT DIFFERENCES BETWEEN GROUPS IN 2019

Genetics of Down syndrome	**Group**	**Rating**		**Group**	**Rating**	**Sig < 0.05**
Incidence (1/600 to 1/800 births)	Prenatal GC	1.62	vs	Postnatal GC	1.69	0.613
	Prenatal GC	1.62	vs	Prenatal Parent	2.00	**0.002**
	Prenatal GC	1.62	vs	Postnatal Parent	2.03	**0.001**
	Postnatal GC	1.69	vs	Prenatal Parent	2.00	**0.003**
	Postnatal GC	1.69	vs	Postnatal Parent	2.03	**0.001**
	Prenatal Parent	2.00	vs	Postnatal Parent	2.03	0.761
Increasing incidence with advanced maternal age	Prenatal GC	2.14	vs	Postnatal GC	2.03	0.508
	Prenatal GC	2.14	vs	Prenatal Parent	1.95	0.135
	Prenatal GC	2.14	vs	Postnatal Parent	1.83	**0.011**
	Postnatal GC	2.03	vs	Prenatal Parent	1.95	0.459
	Postnatal GC	2.03	vs	Postnatal Parent	1.83	0.058
	Prenatal Parent	1.95	vs	Postnatal Parent	1.83	0.090
Caused by extra genetic material from chromosome 21	Prenatal GC	2.79	vs	Postnatal GC	2.85	0.416
	Prenatal GC	2.79	vs	Prenatal Parent	2.71	0.408
	Prenatal GC	2.79	vs	Postnatal Parent	2.73	0.534
	Postnatal GC	2.85	vs	Prenatal Parent	2.71	0.057
	Postnatal GC	2.85	vs	Postnatal Parent	2.73	0.092
	Prenatal Parent	2.71	vs	Postnatal Parent	2.73	0.685
3 types: Nondisjunction (94%), Translocation (3-4%), Mosaic (2%)	Prenatal GC	2.14	vs	Postnatal GC	2.21	0.610
	Prenatal GC	2.14	vs	Prenatal Parent	2.39	**0.036**
	Prenatal GC	2.14	vs	Postnatal Parent	2.28	0.268
	Postnatal GC	2.21	vs	Prenatal Parent	2.39	0.080
	Postnatal GC	2.21	vs	Postnatal Parent	2.28	0.531
	Prenatal Parent	2.39	vs	Postnatal Parent	2.28	0.096
Diagnosis confirmed by chromosome analysis	Prenatal GC	2.98	vs	Postnatal GC	2.84	0.040
	Prenatal GC	2.98	vs	Prenatal Parent	2.67	**0.001**
	Prenatal GC	2.98	vs	Postnatal Parent	2.77	**0.007**
	Postnatal GC	2.84	vs	Prenatal Parent	2.67	**0.034**
	Postnatal GC	2.84	vs	Postnatal Parent	2.77	0.306
	Prenatal Parent	2.67	vs	Postnatal Parent	2.77	0.051
Recurrence risk for future pregnancies	Prenatal GC	2.48	vs	Postnatal GC	2.72	0.040
	Prenatal GC	2.48	vs	Prenatal Parent	2.36	0.301
	Prenatal GC	2.48	vs	Postnatal Parent	2.35	0.255
	Postnatal GC	2.72	vs	Prenatal Parent	2.36	**< 0.0001**
	Postnatal GC	2.72	vs	Postnatal Parent	2.35	**< 0.0001**
	Prenatal Parent	2.36	vs	Postnatal Parent	2.35	0.901
Reproductive options	Prenatal GC	2.57	vs	Postnatal GC	2.45	0.442
	Prenatal GC	2.57	vs	Prenatal Parent	2.04	**< 0.0001**
	Prenatal GC	2.57	vs	Postnatal Parent	1.80	**< 0.0001**
	Postnatal GC	2.45	vs	Prenatal Parent	2.04	**0.001**
	Postnatal GC	2.45	vs	Postnatal Parent	1.80	**< 0.0001**
	Prenatal Parent	2.04	vs	Postnatal Parent	1.80	**0.003**
Adoption	Prenatal GC	2.33	vs	Postnatal GC	2.28	0.731
	Prenatal GC	2.33	vs	Prenatal Parent	2.13	0.116
	Prenatal GC	2.33	vs	Postnatal Parent	1.92	**0.002**
	Postnatal GC	2.28	vs	Prenatal Parent	2.13	0.196
	Postnatal GC	2.28	vs	Postnatal Parent	1.92	**0.003**
	Prenatal Parent	2.13	vs	Postnatal Parent	1.92	**0.011**
Diagnostic Criteria & Physical Features	**Group**	**Rating**		**Group**	**Rating**	**Sig < 0.05**
Flat facial profile (90%)	Prenatal GC	1.88	vs	Postnatal GC	1.87	0.940
	Prenatal GC	1.88	vs	Prenatal Parent	1.81	0.561

	Prenatal GC	1.88	vs	Postnatal Parent	1.91	0.804
	Postnatal GC	1.87	vs	Prenatal Parent	1.81	0.592
	Postnatal GC	1.87	vs	Postnatal Parent	1.91	0.699
	Prenatal Parent	1.81	vs	Postnatal Parent	1.91	0.157
Upward slanting, almond-shaped eyes (80%)	Prenatal GC	1.98	vs	Postnatal GC	1.95	0.870
	Prenatal GC	1.98	vs	Prenatal Parent	1.87	0.365
	Prenatal GC	1.98	vs	Postnatal Parent	1.96	0.894
	Postnatal GC	1.87	vs	Prenatal Parent	1.87	0.432
	Postnatal GC	1.87	vs	Postnatal Parent	1.96	0.941
	Prenatal Parent	1.87	vs	Postnatal Parent	1.96	0.199
Hypotonia (80%)	Prenatal GC	2.62	vs	Postnatal GC	2.70	0.438
	Prenatal GC	2.62	vs	Prenatal Parent	2.63	0.880
	Prenatal GC	2.62	vs	Postnatal Parent	2.63	0.922
	Postnatal GC	2.70	vs	Prenatal Parent	2.63	0.395
	Postnatal GC	2.70	vs	Postnatal Parent	2.63	0.381
	Prenatal Parent	2.63	vs	Postnatal Parent	2.63	0.935
Hyperflexibility (80%)	Prenatal GC	1.95	vs	Postnatal GC	1.92	0.817
	Prenatal GC	1.95	vs	Prenatal Parent	2.28	**0.008**
	Prenatal GC	1.95	vs	Postnatal Parent	2.35	**0.001**
	Postnatal GC	1.92	vs	Prenatal Parent	2.28	**0.001**
	Postnatal GC	1.92	vs	Postnatal Parent	2.35	**< 0.0001**
	Prenatal Parent	2.28	vs	Postnatal Parent	2.35	0.320
Excess skin on back of neck (80%)	Prenatal GC	1.81	vs	Postnatal GC	1.72	0.544
	Prenatal GC	1.81	vs	Prenatal Parent	1.82	0.949
	Prenatal GC	1.81	vs	Postnatal Parent	1.81	0.970
	Postnatal GC	1.72	vs	Prenatal Parent	1.82	0.388
	Postnatal GC	1.72	vs	Postnatal Parent	1.81	0.367
	Prenatal Parent	1.82	vs	Postnatal Parent	1.81	0.958
Dysplastic ears (60%)	Prenatal GC	1.64	vs	Postnatal GC	1.52	0.389
	Prenatal GC	1.64	vs	Prenatal Parent	1.86	0.092
	Prenatal GC	1.64	vs	Postnatal Parent	1.83	.124
	Postnatal GC	1.52	vs	Prenatal Parent	1.86	**0.002**
	Postnatal GC	1.52	vs	Postnatal Parent	1.83	**0.003**
	Prenatal Parent	1.86	vs	Postnatal Parent	1.83	0.714
Dysplasia of midphalanx of 5th digit (60%)	Prenatal GC	1.38	vs	Postnatal GC	1.43	0.693
	Prenatal GC	1.38	vs	Prenatal Parent	1.76	**0.002**
	Prenatal GC	1.38	vs	Postnatal Parent	1.71	**0.003**
	Postnatal GC	1.52	vs	Prenatal Parent	1.76	**0.001**
	Postnatal GC	1.52	vs	Postnatal Parent	1.71	**0.003**
	Prenatal Parent	1.76	vs	Postnatal Parent	1.71	0.518
Small head	Prenatal GC	1.69	vs	Postnatal GC	1.60	0..527
	Prenatal GC	1.69	vs	Prenatal Parent	1.89	0.111
	Prenatal GC	1.69	vs	Postnatal Parent	1.84	0.233
	Postnatal GC	1.52	vs	Prenatal Parent	1.89	**0.010**
	Postnatal GC	1.52	vs	Postnatal Parent	1.84	**0.033**
	Prenatal Parent	1.89	vs	Postnatal Parent	1.84	0.492
Brushfield spots on the iris of the eye	Prenatal GC	1.38	vs	Postnatal GC	1.48	0.404
	Prenatal GC	1.38	vs	Prenatal Parent	1.74	**0.003**
	Prenatal GC	1.38	vs	Postnatal Parent	1.72	**0.004**
	Postnatal GC	1.48	vs	Prenatal Parent	1.74	**0.012**
	Postnatal GC	1.48	vs	Postnatal Parent	1.72	**0.019**
	Prenatal Parent	1.74	vs	Postnatal Parent	1.72	0.742
Epicanthal folds	Prenatal GC	1.64	vs	Postnatal GC	1.68	0.786
	Prenatal GC	1.64	vs	Prenatal Parent	1.79	0.242
	Prenatal GC	1.64	vs	Postnatal Parent	1.75	0.366
	Postnatal GC	1.68	vs	Prenatal Parent	1.79	0.332
	Postnatal GC	1.68	vs	Postnatal Parent	1.75	0.519
	Prenatal Parent	1.79	vs	Postnatal Parent	1.75	0.581
Short, broad hands with short fingers	Prenatal GC	1.55	vs	Postnatal GC	1.61	0.652
	Prenatal GC	1.55	vs	Prenatal Parent	1.74	0.124
	Prenatal GC	1.55	vs	Postnatal Parent	1.79	**0.040**
	Postnatal GC	1.61	vs	Prenatal Parent	1.74	0.245
	Postnatal GC	1.61	vs	Postnatal Parent	1.79	0.090
	Prenatal Parent	1.74	vs	Postnatal Parent	1.79	0.500
5th finger clinodactyly	Prenatal GC	1.43	vs	Postnatal GC	1.45	0.862
	Prenatal GC	1.43	vs	Prenatal Parent	1.61	0.124

	Prenatal GC	1.43	vs	Postnatal Parent	1.64	0.056
	Postnatal GC	1.45	vs	Prenatal Parent	1.61	0.119
	Postnatal GC	1.45	vs	Postnatal Parent	1.64	**0.049**
	Prenatal Parent	1.61	vs	Postnatal Parent	1.64	0.650
Single palmar crease	Prenatal GC	1.61	vs	Postnatal GC	1.62	0.924
	Prenatal GC	1.61	vs	Prenatal Parent	1.71	0.428
	Prenatal GC	1.61	vs	Postnatal Parent	1.76	0.205
	Postnatal GC	1.62	vs	Prenatal Parent	1.71	0.422
	Postnatal GC	1.62	vs	Postnatal Parent	1.76	0.175
	Prenatal Parent	1.71	vs	Postnatal Parent	1.76	0.439
Sandal gap between 1st & 2nd toes	Prenatal GC	1.50	vs	Postnatal GC	1.54	0.745
	Prenatal GC	1.50	vs	Prenatal Parent	1.67	0.159
	Prenatal GC	1.50	vs	Postnatal Parent	1.78	**0.015**
	Postnatal GC	1.54	vs	Prenatal Parent	1.67	0.220
	Postnatal GC	1.54	vs	Postnatal Parent	1.78	**0.017**
	Prenatal Parent	1.67	vs	Postnatal Parent	1.78	0.095
Short stature	Prenatal GC	2.19	vs	Postnatal GC	2.07	0.405
	Prenatal GC	2.19	vs	Prenatal Parent	1.95	0.054
	Prenatal GC	2.19	vs	Postnatal Parent	1.98	0.095
	Postnatal GC	2.07	vs	Prenatal Parent	1.95	0.294
	Postnatal GC	2.07	vs	Postnatal Parent	1.98	0.431
	Prenatal Parent	1.95	vs	Postnatal Parent	1.98	0.711
Enlarged tongue	Prenatal GC	1.86	vs	Postnatal GC	1.88	0.856
	Prenatal GC	1.86	vs	Prenatal Parent	2.28	**0.001**
	Prenatal GC	1.86	vs	Postnatal Parent	2.22	**0.002**
	Postnatal GC	1.88	vs	Prenatal Parent	2.28	**< 0.0001**
	Postnatal GC	1.88	vs	Postnatal Parent	2.22	**0.001**
	Prenatal Parent	2.28	vs	Postnatal Parent	2.22	0.470
Syndactyly of 2nd and 3rd toes	Prenatal GC	1.38	vs	Postnatal GC	1.47	0.445
	Prenatal GC	1.38	vs	Prenatal Parent	1.71	**0.004**
	Prenatal GC	1.38	vs	Postnatal Parent	1.71	**0.003**
	Postnatal GC	1.47	vs	Prenatal Parent	1.71	**0.014**
	Postnatal GC	1.47	vs	Postnatal Parent	1.71	**0.011**
	Prenatal Parent	1.71	vs	Postnatal Parent	1.71	0.971
Medical Complications Associated with Down syndrome	**Group**	**Rating**		**Group**	**Rating**	**Sig < 0.05**
Early-Onset Alzheimer Disease (11% by age 50; 77% by age 70)	Prenatal GC	2.07	vs	Postnatal GC	2.17	0.462
	Prenatal GC	2.07	vs	Prenatal Parent	2.38	**0.007**
	Prenatal GC	2.07	vs	Postnatal Parent	2.23	0.202
	Postnatal GC	2.17	vs	Prenatal Parent	2.38	**0.031**
	Postnatal GC	2.17	vs	Postnatal Parent	2.23	0.558
	Prenatal Parent	2.38	vs	Postnatal Parent	2.23	**0.030**
Dry, Hyperkeratotic Skin (75%)	Prenatal GC	1.57	vs	Postnatal GC	1.65	0.596
	Prenatal GC	1.57	vs	Prenatal Parent	1.86	**0.018**
	Prenatal GC	1.57	vs	Postnatal Parent	1.94	**0.001**
	Postnatal GC	1.65	vs	Prenatal Parent	1.86	0.054
	Postnatal GC	1.65	vs	Postnatal Parent	1.94	**0.005**
	Prenatal Parent	1.86	vs	Postnatal Parent	1.94	0.223
One or More Congenital Abnormalities (50%)	Prenatal GC	2.83	vs	Postnatal GC	2.87	0.697
	Prenatal GC	2.83	vs	Prenatal Parent	2.60	**0.017**
	Prenatal GC	2.83	vs	Postnatal Parent	2.50	**0.002**
	Postnatal GC	2.87	vs	Prenatal Parent	2.60	**0.001**
	Postnatal GC	2.87	vs	Postnatal Parent	2.50	**< 0.0001**
	Prenatal Parent	2.60	vs	Postnatal Parent	2.50	0.111
Heart Defect Possibly Requiring Open Heart Surgery (40-60%)	Prenatal GC	2.90	vs	Postnatal GC	2.95	0.477
	Prenatal GC	2.90	vs	Prenatal Parent	2.88	0.717
	Prenatal GC	2.90	vs	Postnatal Parent	2.82	0.267
	Postnatal GC	2.95	vs	Prenatal Parent	2.88	0.153
	Postnatal GC	2.95	vs	Postnatal Parent	2.82	**0.034**
	Prenatal Parent	2.88	vs	Postnatal Parent	2.82	0.120
Thyroid Disorders (up to 50%)	Prenatal GC	2.33	vs	Postnatal GC	2.65	**0.009**
	Prenatal GC	2.33	vs	Prenatal Parent	2.62	**0.004**
	Prenatal GC	2.33	vs	Postnatal Parent	2.60	**0.007**
	Postnatal GC	2.65	vs	Prenatal Parent	2.62	0.735
	Postnatal GC	2.65	vs	Postnatal Parent	2.60	0.589
	Prenatal Parent	2.62	vs	Postnatal Parent	2.60	0.766

Obstructive Sleep Apnea (33%)	Prenatal GC	2.19	vs	Postnatal GC	2.51	**0.019**
	Prenatal GC	2.19	vs	Prenatal Parent	2.66	**< 0.0001**
	Prenatal GC	2.19	vs	Postnatal Parent	2.60	**< 0.0001**
	Postnatal GC	2.51	vs	Prenatal Parent	2.66	0.108
	Postnatal GC	2.51	vs	Postnatal Parent	2.60	0.332
	Prenatal Parent	2.66	vs	Postnatal Parent	2.60	0.345
Atlantoaxial and/or Atlantooccipital Instability (10-30%)	Prenatal GC	1.85	vs	Postnatal GC	1.85	0.960
	Prenatal GC	1.85	vs	Prenatal Parent	2.22	**0.006**
	Prenatal GC	1.85	vs	Postnatal Parent	2.26	**0.002**
	Postnatal GC	1.85	vs	Prenatal Parent	2.22	**0.002**
	Postnatal GC	1.85	vs	Postnatal Parent	2.26	**0.001**
	Prenatal Parent	2.22	vs	Postnatal Parent	2.26	0.567
Gastrointestinal Defect Possibly Requiring Surgery (12%)	Prenatal GC	2.45	vs	Postnatal GC	2.48	0.811
	Prenatal GC	2.45	vs	Prenatal Parent	2.53	0.497
	Prenatal GC	2.45	vs	Postnatal Parent	2.46	0.969
	Postnatal GC	2.48	vs	Prenatal Parent	2.53	0.643
	Postnatal GC	2.48	vs	Postnatal Parent	2.46	0.784
	Prenatal Parent	2.53	vs	Postnatal Parent	2.46	0.280
Autism Spectrum Disorder (ASD) (1%)	Prenatal GC	1.76	vs	Postnatal GC	1.86	0.511
	Prenatal GC	1.76	vs	Prenatal Parent	2.05	**0.028**
	Prenatal GC	1.76	vs	Postnatal Parent	1.92	0.205
	Postnatal GC	1.86	vs	Prenatal Parent	2.05	0.105
	Postnatal GC	1.86	vs	Postnatal Parent	1.92	0.605
	Prenatal Parent	2.05	vs	Postnatal Parent	1.92	0.086
Epilepsy (10%)	Prenatal GC	1.88	vs	Postnatal GC	1.95	0.652
	Prenatal GC	1.88	vs	Prenatal Parent	2.24	**0.005**
	Prenatal GC	1.88	vs	Postnatal Parent	2.16	**0.030**
	Postnatal GC	1.95	vs	Prenatal Parent	2.24	**0.010**
	Postnatal GC	1.95	vs	Postnatal Parent	2.16	0.065
	Prenatal Parent	2.24	vs	Postnatal Parent	2.16	0.272
Increased Risk of Childhood Leukemia (2%)	Prenatal GC	1.98	vs	Postnatal GC	2.05	0.613
	Prenatal GC	1.98	vs	Prenatal Parent	2.29	**0.011**
	Prenatal GC	1.98	vs	Postnatal Parent	2.13	0.227
	Postnatal GC	2.05	vs	Prenatal Parent	2.29	**0.027**
	Postnatal GC	2.05	vs	Postnatal Parent	2.13	0.472
	Prenatal Parent	2.29	vs	Postnatal Parent	2.13	**0.031**
Congenital or Acquired Cataracts	Prenatal GC	1.79	vs	Postnatal GC	1.93	0.364
	Prenatal GC	1.79	vs	Prenatal Parent	2.20	**0.001**
	Prenatal GC	1.79	vs	Postnatal Parent	2.11	**0.010**
	Postnatal GC	1.93	vs	Prenatal Parent	2.20	**0.015**
	Postnatal GC	1.93	vs	Postnatal Parent	2.11	0.107
	Prenatal Parent	2.20	vs	Postnatal Parent	2.11	0.222
Vision Problems	Prenatal GC	2.39	vs	Postnatal GC	2.67	**0.024**
	Prenatal GC	2.39	vs	Prenatal Parent	2.52	0.229
	Prenatal GC	2.39	vs	Postnatal Parent	2.49	0.393
	Postnatal GC	2.67	vs	Prenatal Parent	2.52	0.093
	Postnatal GC	2.67	vs	Postnatal Parent	2.49	0.053
	Prenatal Parent	2.52	vs	Postnatal Parent	2.49	0.614
Hearing Loss	Prenatal GC	2.44	vs	Postnatal GC	2.71	**0.025**
	Prenatal GC	2.44	vs	Prenatal Parent	2.60	0.124
	Prenatal GC	2.44	vs	Postnatal Parent	2.55	0.296
	Postnatal GC	2.71	vs	Prenatal Parent	2.60	0.211
	Postnatal GC	2.71	vs	Postnatal Parent	2.55	0.091
	Prenatal Parent	2.60	vs	Postnatal Parent	2.55	0.457
Ear Problems	Prenatal GC	2.19	vs	Postnatal GC	2.57	**0.005**
	Prenatal GC	2.19	vs	Prenatal Parent	2.54	**0.002**
	Prenatal GC	2.19	vs	Postnatal Parent	2.53	**0.003**
	Postnatal GC	2.57	vs	Prenatal Parent	2.54	0.745
	Postnatal GC	2.57	vs	Postnatal Parent	2.53	0.669
	Prenatal Parent	2.54	vs	Postnatal Parent	2.53	0.872
Respiratory Problems	Prenatal GC	1.98	vs	Postnatal GC	1.85	0.355
	Prenatal GC	1.98	vs	Prenatal Parent	2.53	**< 0.0001**
	Prenatal GC	1.98	vs	Postnatal Parent	2.52	**< 0.0001**
	Postnatal GC	1.85	vs	Prenatal Parent	2.53	**< 0.0001**
	Postnatal GC	1.85	vs	Postnatal Parent	2.52	**< 0.0001**
	Prenatal Parent	2.53	vs	Postnatal Parent	2.52	0.828

Increased Susceptibility to Infection	Prenatal GC	2.02	vs	Postnatal GC	1.83	0.181
	Prenatal GC	2.02	vs	Prenatal Parent	2.46	**< 0.0001**
	Prenatal GC	2.02	vs	Postnatal Parent	2.44	**< 0.0001**
	Postnatal GC	1.83	vs	Prenatal Parent	2.46	**< 0.0001**
	Postnatal GC	1.83	vs	Postnatal Parent	2.44	**< 0.0001**
	Prenatal Parent	2.46	vs	Postnatal Parent	2.44	0.776
Increased Susceptibility to Periodontal Disease	Prenatal GC	1.67	vs	Postnatal GC	1.65	0.906
	Prenatal GC	1.67	vs	Prenatal Parent	2.22	**< 0.0001**
	Prenatal GC	1.67	vs	Postnatal Parent	2.19	**< 0.0001**
	Postnatal GC	1.65	vs	Prenatal Parent	2.22	**< 0.0001**
	Postnatal GC	1.65	vs	Postnatal Parent	2.19	**< 0.0001**
	Prenatal Parent	2.22	vs	Postnatal Parent	2.19	0.703
Increased Risk of Autoimmune Disease	Prenatal GC	1.90	vs	Postnatal GC	1.78	< 0.0001
	Prenatal GC	1.90	vs	Prenatal Parent	1.78	0.413
	Prenatal GC	1.90	vs	Postnatal Parent	2.35	**< 0.0001**
	Postnatal GC	1.78	vs	Prenatal Parent	1.78	**< 0.0001**
	Postnatal GC	1.78	vs	Postnatal Parent	2.35	**< 0.0001**
	Prenatal Parent	1.78	vs	Postnatal Parent	2.35	0.981
Orthopedic problems	Prenatal GC	1.83	vs	Postnatal GC	1.71	0.420
	Prenatal GC	1.83	vs	Prenatal Parent	2.39	**< 0.0001**
	Prenatal GC	1.83	vs	Postnatal Parent	2.33	**< 0.0001**
	Postnatal GC	1.71	vs	Prenatal Parent	2.39	**< 0.0001**
	Postnatal GC	1.71	vs	Postnatal Parent	2.33	**< 0.0001**
	Prenatal Parent	2.39	vs	Postnatal Parent	2.33	0.380
Psychiatric Disorders	Prenatal GC	1.88	vs	Postnatal GC	1.76	0.408
	Prenatal GC	1.88	vs	Prenatal Parent	2.13	0.052
	Prenatal GC	1.88	vs	Postnatal Parent	2.06	0.167
	Postnatal GC	1.76	vs	Prenatal Parent	2.13	**0.001**
	Postnatal GC	1.76	vs	Postnatal Parent	2.06	**0.010**
	Prenatal Parent	2.13	vs	Postnatal Parent	2.06	0.372
Tendency for Obesity	Prenatal GC	1.71	vs	Postnatal GC	1.86	0.317
	Prenatal GC	1.71	vs	Prenatal Parent	2.20	**< 0.0001**
	Prenatal GC	1.71	vs	Postnatal Parent	2.17	**< 0.0001**
	Postnatal GC	1.86	vs	Prenatal Parent	2.20	**0.002**
	Postnatal GC	1.86	vs	Postnatal Parent	2.17	**0.007**
	Prenatal Parent	2.20	vs	Postnatal Parent	2.17	0.734
Sterility in Males	Prenatal GC	1.74	vs	Postnatal GC	1.88	0.322
	Prenatal GC	1.74	vs	Prenatal Parent	2.03	**0.025**
	Prenatal GC	1.74	vs	Postnatal Parent	1.90	0.223
	Postnatal GC	1.88	vs	Prenatal Parent	2.03	0.209
	Postnatal GC	1.88	vs	Postnatal Parent	1.90	0.887
	Prenatal Parent	2.03	vs	Postnatal Parent	1.90	0.106
Increased Risk of Testicular Cancer in Males	Prenatal GC	1.66	vs	Postnatal GC	1.60	0.659
	Prenatal GC	1.66	vs	Prenatal Parent	2.17	**< 0.0001**
	Prenatal GC	1.66	vs	Postnatal Parent	2.05	**0.004**
	Postnatal GC	1.60	vs	Prenatal Parent	2.17	**< 0.0001**
	Postnatal GC	1.60	vs	Postnatal Parent	2.05	**< 0.0001**
	Prenatal Parent	2.17	vs	Postnatal Parent	2.05	0.147
Attention Deficit Hyperactivity Disorder (ADHD)	Prenatal GC	1.71	vs	Postnatal GC	1.62	0.480
	Prenatal GC	1.71	vs	Prenatal Parent	2.04	**0.012**
	Prenatal GC	1.71	vs	Postnatal Parent	1.94	0.081
	Postnatal GC	1.62	vs	Prenatal Parent	2.04	**< 0.0001**
	Postnatal GC	1.62	vs	Postnatal Parent	1.94	**0.003**
	Prenatal Parent	2.04	vs	Postnatal Parent	1.94	0.189
Intellectual Disability & Developmental Delay	**Group**	**Rating**		**Group**	**Rating**	**Sig < 0.05**
Variable Range of Intellectual Disability from Mild to Moderate	Prenatal GC	2.93	vs	Postnatal GC	2.97	0.364
	Prenatal GC	2.93	vs	Prenatal Parent	2.65	**0.003**
	Prenatal GC	2.93	vs	Postnatal Parent	2.62	**0.001**
	Postnatal GC	2.97	vs	Prenatal Parent	2.65	**< 0.0001**
	Postnatal GC	2.97	vs	Postnatal Parent	2.62	**< 0.0001**
	Prenatal Parent	2.65	vs	Postnatal Parent	2.62	0.566
Mean (Average) IQ of 50	Prenatal GC	2.15	vs	Postnatal GC	2.05	0.499
	Prenatal GC	2.15	vs	Prenatal Parent	1.87	**0.025**
	Prenatal GC	2.15	vs	Postnatal Parent	1.85	**0.017**

	Postnatal GC	2.05	vs	Prenatal Parent	1.87	0.095
	Postnatal GC	2.05	vs	Postnatal Parent	1.85	0.067
	Prenatal Parent	1.87	vs	Postnatal Parent	1.85	0.824
Range of IQ from 20 to 80	Prenatal GC	2.29	vs	Postnatal GC	2.11	0.232
	Prenatal GC	2.29	vs	Prenatal Parent	1.93	**0.005**
	Prenatal GC	2.29	vs	Postnatal Parent	1.86	**0.001**
	Postnatal GC	2.11	vs	Prenatal Parent	1.93	0.098
	Postnatal GC	2.11	vs	Postnatal Parent	1.86	**0.023**
	Prenatal Parent	1.93	vs	Postnatal Parent	1.86	0.365
Developmental Delay in Achieving Milestones	Prenatal GC	2.93	vs	Postnatal GC	2.98	0.153
	Prenatal GC	2.93	vs	Prenatal Parent	2.73	**0.021**
	Prenatal GC	2.93	vs	Postnatal Parent	2.62	**0.001**
	Postnatal GC	2.98	vs	Prenatal Parent	2.73	**< 0.0001**
	Postnatal GC	2.98	vs	Postnatal Parent	2.62	**< 0.0001**
	Prenatal Parent	2.73	vs	Postnatal Parent	2.62	0.058
Need for Physical Therapy	Prenatal GC	2.64	vs	Postnatal GC	2.79	0.161
	Prenatal GC	2.64	vs	Prenatal Parent	2.74	0.254
	Prenatal GC	2.64	vs	Postnatal Parent	2.73	0.328
	Postnatal GC	2.79	vs	Prenatal Parent	2.74	0.530
	Postnatal GC	2.79	vs	Postnatal Parent	2.73	0.431
	Prenatal Parent	2.74	vs	Postnatal Parent	2.73	0.809
Need for Occupational Therapy	Prenatal GC	2.64	vs	Postnatal GC	2.79	0.161
	Prenatal GC	2.64	vs	Prenatal Parent	2.74	0.254
	Prenatal GC	2.64	vs	Postnatal Parent	2.72	0.379
	Postnatal GC	2.79	vs	Prenatal Parent	2.74	0.530
	Postnatal GC	2.79	vs	Postnatal Parent	2.72	0.371
	Prenatal Parent	2.74	vs	Postnatal Parent	2.72	0.688
Need for Speech & Language Therapy	Prenatal GC	2.64	vs	Postnatal GC	2.81	0.117
	Prenatal GC	2.64	vs	Prenatal Parent	2.77	0.145
	Prenatal GC	2.64	vs	Postnatal Parent	2.74	0.259
	Postnatal GC	2.81	vs	Prenatal Parent	2.77	0.578
	Postnatal GC	2.81	vs	Postnatal Parent	2.74	0.380
	Prenatal Parent	2.77	vs	Postnatal Parent	2.74	0.624
Need for Early Intervention & Case Management	Prenatal GC	2.90	vs	Postnatal GC	2.87	0.631
	Prenatal GC	2.90	vs	Prenatal Parent	2.85	0.445
	Prenatal GC	2.90	vs	Postnatal Parent	2.86	0.468
	Postnatal GC	2.87	vs	Prenatal Parent	2.85	0.766
	Postnatal GC	2.87	vs	Postnatal Parent	2.86	0.825
	Prenatal Parent	2.85	vs	Postnatal Parent	2.86	0.897

Long-term Prognosis	**Group**	**Rating**		**Group**	**Rating**	**Sig < 0.05**
Participate in Community Sports, Activities, & Leagues	Prenatal GC	2.67	vs	Postnatal GC	2.65	0.886
	Prenatal GC	2.67	vs	Prenatal Parent	2.69	0.759
	Prenatal GC	2.67	vs	Postnatal Parent	2.63	0.712
	Postnatal GC	2.65	vs	Prenatal Parent	2.69	0.588
	Postnatal GC	2.65	vs	Postnatal Parent	2.63	0.829
	Prenatal Parent	2.69	vs	Postnatal Parent	2.63	0.264
Participate in Special Olympics & Therapeutic Recreation	Prenatal GC	2.45	vs	Postnatal GC	2.23	0.108
	Prenatal GC	2.45	vs	Prenatal Parent	2.60	0.159
	Prenatal GC	2.45	vs	Postnatal Parent	2.50	0.638
	Postnatal GC	2.23	vs	Prenatal Parent	2.60	**< 0.0001**
	Postnatal GC	2.23	vs	Postnatal Parent	2.50	**0.004**
	Prenatal Parent	2.60	vs	Postnatal Parent	2.50	0.118
Inclusion in Regular Classes	Prenatal GC	2.74	vs	Postnatal GC	2.50	**0.042**
	Prenatal GC	2.74	vs	Prenatal Parent	2.81	0.370
	Prenatal GC	2.74	vs	Postnatal Parent	2.74	0.995
	Postnatal GC	2.50	vs	Prenatal Parent	2.81	**< 0.0001**
	Postnatal GC	2.50	vs	Postnatal Parent	2.74	**0.004**
	Prenatal Parent	2.81	vs	Postnatal Parent	2.74	0.181
Special Education Classes	Prenatal GC	2.74	vs	Postnatal GC	2.75	0.869
	Prenatal GC	2.74	vs	Prenatal Parent	2.48	**0.012**
	Prenatal GC	2.74	vs	Postnatal Parent	2.47	**0.015**
	Postnatal GC	2.75	vs	Prenatal Parent	2.48	**0.002**
	Postnatal GC	2.75	vs	Postnatal Parent	2.47	**0.003**
	Prenatal Parent	2.48	vs	Postnatal Parent	2.47	0.929
Complete High School	Prenatal GC	2.55	vs	Postnatal GC	2.48	0.602

	Prenatal GC	2.55	vs	Prenatal Parent	2.74	**0.037**
	Prenatal GC	2.55	vs	Postnatal Parent	2.68	0.190
	Postnatal GC	2.48	vs	Prenatal Parent	2.74	**0.002**
	Postnatal GC	2.48	vs	Postnatal Parent	2.68	**0.026**
	Prenatal Parent	2.74	vs	Postnatal Parent	2.68	0.263
Attend College or Post-Secondary Education	Prenatal GC	2.37	vs	Postnatal GC	2.29	0.528
	Prenatal GC	2.37	vs	Prenatal Parent	2.63	**0.015**
	Prenatal GC	2.37	vs	Postnatal Parent	2.48	0.298
	Postnatal GC	2.29	vs	Prenatal Parent	2.63	**< 0.0001**
	Postnatal GC	2.29	vs	Postnatal Parent	2.48	**0.043**
	Prenatal Parent	2.63	vs	Postnatal Parent	2.48	**0.028**
Supported, Competitive Employment	Prenatal GC	2.64	vs	Postnatal GC	2.51	0.256
	Prenatal GC	2.64	vs	Prenatal Parent	2.73	0.308
	Prenatal GC	2.64	vs	Postnatal Parent	2.68	0.713
	Postnatal GC	2.51	vs	Prenatal Parent	2.73	**0.006**
	Postnatal GC	2.51	vs	Postnatal Parent	2.68	**0.048**
	Prenatal Parent	2.73	vs	Postnatal Parent	2.68	0.329
Live Independently	Prenatal GC	2.67	vs	Postnatal GC	2.52	0.215
	Prenatal GC	2.67	vs	Prenatal Parent	2.74	0.396
	Prenatal GC	2.67	vs	Postnatal Parent	2.65	0.895
	Postnatal GC	2.52	vs	Prenatal Parent	2.74	**0.007**
	Postnatal GC	2.52	vs	Postnatal Parent	2.65	0.113
	Prenatal Parent	2.74	vs	Postnatal Parent	2.65	0.117
Live in Group Home	Prenatal GC	2.69	vs	Postnatal GC	2.56	0.211
	Prenatal GC	2.69	vs	Prenatal Parent	2.47	**0.032**
	Prenatal GC	2.69	vs	Postnatal Parent	2.33	**0.002**
	Postnatal GC	2.56	vs	Prenatal Parent	2.47	0.327
	Postnatal GC	2.56	vs	Postnatal Parent	2.33	**0.021**
	Prenatal Parent	2.47	vs	Postnatal Parent	2.33	**0.047**
Have Friends	Prenatal GC	2.76	vs	Postnatal GC	2.79	0.798
	Prenatal GC	2.76	vs	Prenatal Parent	2.87	0.091
	Prenatal GC	2.76	vs	Postnatal Parent	2.85	0.253
	Postnatal GC	2.79	vs	Prenatal Parent	2.87	0.141
	Postnatal GC	2.79	vs	Postnatal Parent	2.85	0.359
	Prenatal Parent	2.87	vs	Postnatal Parent	2.85	0.493
Have Intimate Relationships	Prenatal GC	2.53	vs	Postnatal GC	2.41	0.392
	Prenatal GC	2.53	vs	Prenatal Parent	2.62	0.371
	Prenatal GC	2.53	vs	Postnatal Parent	2.56	0.754
	Postnatal GC	2.41	vs	Prenatal Parent	2.62	**0.029**
	Postnatal GC	2.41	vs	Postnatal Parent	2.56	0.130
	Prenatal Parent	2.62	vs	Postnatal Parent	2.56	0.354
Get Married	Prenatal GC	2.32	vs	Postnatal GC	2.24	0.572
	Prenatal GC	2.32	vs	Prenatal Parent	2.52	0.057
	Prenatal GC	2.32	vs	Postnatal Parent	2.43	0.306
	Postnatal GC	2.24	vs	Prenatal Parent	2.52	**0.004**
	Postnatal GC	2.24	vs	Postnatal Parent	2.43	0.056
	Prenatal Parent	2.52	vs	Postnatal Parent	2.43	0.174
Life Expectancy (ranging into 50s-60s)	Prenatal GC	2.80	vs	Postnatal GC	2.68	0.200
	Prenatal GC	2.80	vs	Prenatal Parent	2.50	**0.004**
	Prenatal GC	2.80	vs	Postnatal Parent	2.51	**0.007**
	Postnatal GC	2.68	vs	Prenatal Parent	2.50	**0.047**
	Postnatal GC	2.68	vs	Postnatal Parent	2.51	0.065
	Prenatal Parent	2.50	vs	Postnatal Parent	2.51	0.852
More Like Other Children Than Different	Prenatal GC	2.83	vs	Postnatal GC	2.90	0.436
	Prenatal GC	2.83	vs	Prenatal Parent	2.89	0.348
	Prenatal GC	2.83	vs	Postnatal Parent	2.89	0.386
	Postnatal GC	2.90	vs	Prenatal Parent	2.89	0.902
	Postnatal GC	2.90	vs	Postnatal Parent	2.89	0.845
	Prenatal Parent	2.89	vs	Postnatal Parent	2.89	0.915
Impact on the Family	**Group**	**Rating**		**Group**	**Rating**	**Sig < 0.05**
Impact on Other Siblings - More Compassionate & Caring	Prenatal GC	2.32	vs	Postnatal GC	2.30	0.891
	Prenatal GC	2.32	vs	Prenatal Parent	2.79	**< 0.0001**
	Prenatal GC	2.32	vs	Postnatal Parent	2.68	**0.001**
	Postnatal GC	2.30	vs	Prenatal Parent	2.79	**< 0.0001**
	Postnatal GC	2.30	vs	Postnatal Parent	2.68	**< 0.0001**

	Prenatal Parent	2.79	vs	Postnatal Parent	2.68	0.061
Impact on Other Siblings - Less Attention & Resentful	Prenatal GC	2.17	vs	Postnatal GC	2.16	0.946
	Prenatal GC	2.17	vs	Prenatal Parent	2.13	0.784
	Prenatal GC	2.17	vs	Postnatal Parent	2.10	0.580
	Postnatal GC	2.16	vs	Prenatal Parent	2.13	0.818
	Postnatal GC	2.16	vs	Postnatal Parent	2.10	0.583
	Prenatal Parent	2.13	vs	Postnatal Parent	2.10	0.667
Impact on Marriage - Strengthens Relationship	Prenatal GC	2.26	vs	Postnatal GC	2.09	0.274
	Prenatal GC	2.26	vs	Prenatal Parent	2.64	**< 0.0001**
	Prenatal GC	2.26	vs	Postnatal Parent	2.54	**0.024**
	Postnatal GC	2.09	vs	Prenatal Parent	2.64	**< 0.0001**
	Postnatal GC	2.09	vs	Postnatal Parent	2.54	**< 0.0001**
	Prenatal Parent	2.64	vs	Postnatal Parent	2.54	0.110
Impact on Marriage – Strains Relationship	Prenatal GC	2.18	vs	Postnatal GC	2.11	0.648
	Prenatal GC	2.18	vs	Prenatal Parent	2.16	0.880
	Prenatal GC	2.18	vs	Postnatal Parent	2.14	0.729
	Postnatal GC	2.11	vs	Prenatal Parent	2.16	0.654
	Postnatal GC	2.11	vs	Postnatal Parent	2.14	0.830
	Prenatal Parent	2.16	vs	Postnatal Parent	2.14	0.743
Impact on Grandparents - Supportive & Welcoming	Prenatal GC	1.82	vs	Postnatal GC	1.75	0.665
	Prenatal GC	1.82	vs	Prenatal Parent	2.55	**< 0.0001**
	Prenatal GC	1.82	vs	Postnatal Parent	2.40	**< 0.0001**
	Postnatal GC	1.75	vs	Prenatal Parent	2.55	**< 0.0001**
	Postnatal GC	1.75	vs	Postnatal Parent	2.40	**< 0.0001**
	Prenatal Parent	2.55	vs	Postnatal Parent	2.40	**0.029**
Impact on Grandparents - Limited Interaction	Prenatal GC	1.79	vs	Postnatal GC	1.72	0.626
	Prenatal GC	1.79	vs	Prenatal Parent	1.99	0.144
	Prenatal GC	1.79	vs	Postnatal Parent	1.87	0.570
	Postnatal GC	1.72	vs	Prenatal Parent	1.99	**0.022**
	Postnatal GC	1.72	vs	Postnatal Parent	1.87	0.193
	Prenatal Parent	1.99	vs	Postnatal Parent	1.87	0.150
Impact on Extended Family Members - Supportive & Welcoming	Prenatal GC	1.87	vs	Postnatal GC	1.74	0.363
	Prenatal GC	1.87	vs	Prenatal Parent	2.47	**< 0.0001**
	Prenatal GC	1.87	vs	Postnatal Parent	2.34	**< 0.0001**
	Postnatal GC	1.74	vs	Prenatal Parent	2.47	**< 0.0001**
	Postnatal GC	1.74	vs	Postnatal Parent	2.344	**< 0.0001**
	Prenatal Parent	2.47	vs	Postnatal Parent	2.344	0.110
Impact on Extended Family Members - Limited Interaction	Prenatal GC	1.82	vs	Postnatal GC	1.71	0.485
	Prenatal GC	1.82	vs	Prenatal Parent	1.94	0.350
	Prenatal GC	1.82	vs	Postnatal Parent	1.84	0.850
	Postnatal GC	1.71	vs	Prenatal Parent	1.94	0.050
	Postnatal GC	1.71	vs	Postnatal Parent	1.84	0.251
	Prenatal Parent	1.94	vs	Postnatal Parent	1.84	0.226
Impact on Other Relationships - Supportive & Welcoming	Prenatal GC	1.82	vs	Postnatal GC	1.80	0.913
	Prenatal GC	1.82	vs	Prenatal Parent	2.52	**< 0.0001**
	Prenatal GC	1.82	vs	Postnatal Parent	2.37	**< 0.0001**
	Postnatal GC	1.80	vs	Prenatal Parent	2.52	**< 0.0001**
	Postnatal GC	1.80	vs	Postnatal Parent	2.37	**< 0.0001**
	Prenatal Parent	2.52	vs	Postnatal Parent	2.37	**0.039**
Impact on Other Relationships - Lose Social Circle	Prenatal GC	1.78	vs	Postnatal GC	1.78	0.965
	Prenatal GC	1.78	vs	Prenatal Parent	2.02	0.066
	Prenatal GC	1.78	vs	Postnatal Parent	1.95	0.182
	Postnatal GC	1.78	vs	Prenatal Parent	2.02	**0.049**
	Postnatal GC	1.78	vs	Postnatal Parent	1.95	0.155
	Prenatal Parent	2.02	vs	Postnatal Parent	1.95	0.400
Financial Impact - No Difference	Prenatal GC	1.74	vs	Postnatal GC	1.82	0.648
	Prenatal GC	1.74	vs	Prenatal Parent	2.22	**< 0.0001**
	Prenatal GC	1.74	vs	Postnatal Parent	2.07	**0.016**
	Postnatal GC	1.82	vs	Prenatal Parent	2.22	**0.001**
	Postnatal GC	1.82	vs	Postnatal Parent	2.07	**0.037**
	Prenatal Parent	2.22	vs	Postnatal Parent	2.07	0.070
Financial Impact - More	Prenatal GC	2.10	vs	Postnatal GC	2.11	0.963
	Prenatal GC	2.10	vs	Prenatal Parent	2.31	0.102
	Prenatal GC	2.10	vs	Postnatal Parent	2.15	0.680
	Postnatal GC	2.11	vs	Prenatal Parent	2.31	0.069
	Postnatal GC	2.11	vs	Postnatal Parent	2.15	0.679

	Prenatal Parent	2.31	vs	Postnatal Parent	2.15	**0.045**
Time Commitment - No Difference	Prenatal GC	1.84	vs	Postnatal GC	1.77	0.649
	Prenatal GC	1.84	vs	Prenatal Parent	2.14	**0.032**
	Prenatal GC	1.84	vs	Postnatal Parent	2.01	0.225
	Postnatal GC	1.77	vs	Prenatal Parent	2.14	**0.002**
	Postnatal GC	1.77	vs	Postnatal Parent	2.01	**0.043**
	Prenatal Parent	2.14	vs	Postnatal Parent	2.01	0.121
Time Commitment - More	Prenatal GC	2.29	vs	Postnatal GC	2.16	0.344
	Prenatal GC	2.29	vs	Prenatal Parent	2.45	0.196
	Prenatal GC	2.29	vs	Postnatal Parent	2.40	0.386
	Postnatal GC	2.16	vs	Prenatal Parent	2.45	**0.006**
	Postnatal GC	2.16	vs	Postnatal Parent	2.40	**0.023**
	Prenatal Parent	2.45	vs	Postnatal Parent	2.40	0.501
Informational Resources & Referrals	**Group**	**Rating**		**Group**	**Rating**	**Sig ≤ 0.05**
Local Support Group(s)	Prenatal GC	2.88	vs	Postnatal GC	2.95	0.197
	Prenatal GC	2.88	vs	Prenatal Parent	2.92	0.490
	Prenatal GC	2.88	vs	Postnatal Parent	2.86	0.734
	Postnatal GC	2.95	vs	Prenatal Parent	2.92	0.419
	Postnatal GC	2.95	vs	Postnatal Parent	2.86	0.075
	Prenatal Parent	2.92	vs	Postnatal Parent	2.86	0.114
National Advocacy Organizations & Websites	Prenatal GC	2.71	vs	Postnatal GC	2.95	0.941
	Prenatal GC	2.71	vs	Prenatal Parent	2.81	0.201
	Prenatal GC	2.71	vs	Postnatal Parent	2.83	0.115
	Postnatal GC	2.95	vs	Prenatal Parent	2.81	0.182
	Postnatal GC	2.95	vs	Postnatal Parent	2.83	0.096
	Prenatal Parent	2.81	vs	Postnatal Parent	2.83	0.692
Online Support Groups/Social Media Platforms	Prenatal GC	2.67	vs	Postnatal GC	2.61	0.539
	Prenatal GC	2.67	vs	Prenatal Parent	2.88	**0.001**
	Prenatal GC	2.67	vs	Postnatal Parent	2.80	0.065
	Postnatal GC	2.61	vs	Prenatal Parent	2.88	**< 0.0001**
	Postnatal GC	2.61	vs	Postnatal Parent	2.80	**0.003**
	Prenatal Parent	2.88	vs	Postnatal Parent	2.80	0.068
Early Intervention Centers	Prenatal GC	2.74	vs	Postnatal GC	2.74	0.997
	Prenatal GC	2.74	vs	Prenatal Parent	2.94	**0.001**
	Prenatal GC	2.74	vs	Postnatal Parent	2.92	**0.001**
	Postnatal GC	2.74	vs	Prenatal Parent	2.94	**0.001**
	Postnatal GC	2.74	vs	Postnatal Parent	2.92	**0.001**
	Prenatal Parent	2.94	vs	Postnatal Parent	2.92	0.729
Printed / Written Material	Prenatal GC	2.76	vs	Postnatal GC	2.79	0.789
	Prenatal GC	2.76	vs	Prenatal Parent	2.67	0.316
	Prenatal GC	2.76	vs	Postnatal Parent	2.68	0.382
	Postnatal GC	2.79	vs	Prenatal Parent	2.67	0.144
	Postnatal GC	2.79	vs	Postnatal Parent	2.68	0.186
	Prenatal Parent	2.67	vs	Postnatal Parent	2.68	0.858
Fact Sheets / Brochures	Prenatal GC	2.69	vs	Postnatal GC	2.44	**0.041**
	Prenatal GC	2.69	vs	Prenatal Parent	2.67	0.819
	Prenatal GC	2.69	vs	Postnatal Parent	2.65	0.663
	Postnatal GC	2.44	vs	Prenatal Parent	2.67	**0.007**
	Postnatal GC	2.44	vs	Postnatal Parent	2.65	**0.023**
	Prenatal Parent	2.67	vs	Postnatal Parent	2.65	0.705
Photographs of Children with Down Syndrome	Prenatal GC	2.02	vs	Postnatal GC	2.07	0.794
	Prenatal GC	2.02	vs	Prenatal Parent	2.53	**< 0.0001**
	Prenatal GC	2.02	vs	Postnatal Parent	2.33	**0.016**
	Postnatal GC	2.07	vs	Prenatal Parent	2.53	**< 0.0001**
	Postnatal GC	2.07	vs	Postnatal Parent	2.33	**0.014**
	Prenatal Parent	2.53	vs	Postnatal Parent	2.33	**0.006**
Books	Prenatal GC	2.34	vs	Postnatal GC	2.21	0.351
	Prenatal GC	2.34	vs	Prenatal Parent	2.69	**< 0.0001**
	Prenatal GC	2.34	vs	Postnatal Parent	2.55	**0.049**
	Postnatal GC	2.21	vs	Prenatal Parent	2.69	**< 0.0001**
	Postnatal GC	2.21	vs	Postnatal Parent	2.55	**< 0.0001**
	Prenatal Parent	2.69	vs	Postnatal Parent	2.55	**0.023**
Contact with families raising a child with Down syndrome	Prenatal GC	2.37	vs	Postnatal GC	2.48	0.330
	Prenatal GC	2.37	vs	Prenatal Parent	2.93	**< 0.0001**
	Prenatal GC	2.37	vs	Postnatal Parent	2.86	**< 0.0001**

	Postnatal GC	2.48	vs	Prenatal Parent	2.93	**< 0.0001**
	Postnatal GC	2.48	vs	Postnatal Parent	2.86	**< 0.0001**
	Prenatal Parent	2.93	vs	Postnatal Parent	2.86	0.064
Adoption Agencies	Prenatal GC	1.98	vs	Postnatal GC	2.00	0.849
	Prenatal GC	1.98	vs	Prenatal Parent	2.09	0.404
	Prenatal GC	1.98	vs	Postnatal Parent	1.89	0.491
	Postnatal GC	2.00	vs	Prenatal Parent	2.09	0.471
	Postnatal GC	2.00	vs	Postnatal Parent	1.89	0.325
	Prenatal Parent	2.09	vs	Postnatal Parent	1.89	**0.023**
Alternative / Nonconventional Therapies	Prenatal GC	1.61	vs	Postnatal GC	1.48	0.393
	Prenatal GC	1.61	vs	Prenatal Parent	2.25	**< 0.0001**
	Prenatal GC	1.61	vs	Postnatal Parent	2.10	**0.001**
	Postnatal GC	1.48	vs	Prenatal Parent	2.25	**< 0.0001**
	Postnatal GC	1.48	vs	Postnatal Parent	2.10	**< 0.0001**
	Prenatal Parent	2.25	vs	Postnatal Parent	2.10	0.074
Specialist Referral(s)	Prenatal GC	2.46	vs	Postnatal GC	2.64	0.196
	Prenatal GC	2.46	vs	Prenatal Parent	2.72	**0.008**
	Prenatal GC	2.46	vs	Postnatal Parent	2.75	**0.004**
	Postnatal GC	2.64	vs	Prenatal Parent	2.72	0.350
	Postnatal GC	2.64	vs	Postnatal Parent	2.75	0.215
	Prenatal Parent	2.72	vs	Postnatal Parent	2.75	0.630
Counselor or Family Therapist Referral(s)	Prenatal GC	2.05	vs	Postnatal GC	2.03	0.907
	Prenatal GC	2.05	vs	Prenatal Parent	2.56	**< 0.0001**
	Prenatal GC	2.05	vs	Postnatal Parent	2.45	**0.001**
	Postnatal GC	2.03	vs	Prenatal Parent	2.56	**< 0.0001**
	Postnatal GC	2.03	vs	Postnatal Parent	2.45	**< 0.0001**
	Prenatal Parent	2.56	vs	Postnatal Parent	2.45	0.139
Pastoral Counseling Referral(s)	Prenatal GC	1.82	vs	Postnatal GC	1.72	0.464
	Prenatal GC	1.82	vs	Prenatal Parent	2.33	**< 0.0001**
	Prenatal GC	1.82	vs	Postnatal Parent	2.17	**0.008**
	Postnatal GC	1.72	vs	Prenatal Parent	2.33	**< 0.0001**
	Postnatal GC	1.72	vs	Postnatal Parent	2.17	**< 0.0001**
	Prenatal Parent	2.33	vs	Postnatal Parent	2.17	**0.046**

APPENDIX F – SIGNIFICANT DIFFERENCES BETWEEN 2009 & 2019 GROUPS

Genetics of Down syndrome	**2009 Group**	**Rating**		**2019 Group**	**Rating**	**Sig < 0.05**
Incidence (1/600 to 1/800 births)	2009 GC	1.86	vs	2019 GC	1.65	**0.002**
	2009 Parent	2.14	vs	2019 Parent	2.02	**0.006**
Increasing incidence with advanced maternal age	2009 GC	2.26	vs	2019 GC	2.10	**0.040**
	2009 Parent	1.93	vs	2019 Parent	1.89	0.415
Caused by extra genetic material from chromosome 21	2009 GC	2.87	vs	2019 GC	2.84	0.591
	2009 Parent	2.70	vs	2019 Parent	2.72	0.498
3 types: Nondisjunction (94%), Translocation (3-4%), Mosaic (2%)	2009 GC	2.20	vs	2019 GC	2.22	0.737
	2009 Parent	2.19	vs	2019 Parent	2.33	**0.003**
Diagnosis confirmed by chromosome analysis	2009 GC	2.94	vs	2019 GC	2.87	0.051
	2009 Parent	2.77	vs	2019 Parent	2.72	0.114
Recurrence risk for future pregnancies	2009 GC	2.64	vs	2019 GC	2.66	0.738
	2009 Parent	2.34	vs	2019 Parent	2.35	0.784
Reproductive options	2009 GC	2.76	vs	2019 GC	2.53	**< 0.0001**
	2009 Parent	1.64	vs	2019 Parent	1.91	**< 0.0001**
Adoption	2009 GC	2.54	vs	2019 GC	2.35	**0.009**
	2009 Parent	1.93	vs	2019 Parent	2.02	0.152
Diagnostic Criteria & Physical Features	**Group**	**Rating**		**Group**	**Rating**	**Sig < 0.05**
Flat facial profile (90%)	2009 GC	1.90	vs	2019 GC	1.86	0.595
	2009 Parent	1.86	vs	2019 Parent	1.86	0.909
Upward slanting, almond-shaped eyes (80%)	2009 GC	1.93	vs	2019 GC	1.92	0.833
	2009 Parent	1.90	vs	2019 Parent	1.91	0.698
Hypotonia (80%)	2009 GC	2.46	vs	2019 GC	2.68	**0.001**
	2009 Parent	2.58	vs	2019 Parent	2.63	0.188
Hyperflexibility (80%)	2009 GC	1.78	vs	2019 GC	1.91	0.075
	2009 Parent	2.31	vs	2019 Parent	2.32	0.828
Excess skin on back of neck (80%)	2009 GC	1.64	vs	2019 GC	1.74	0.141
	2009 Parent	1.68	vs	2019 Parent	1.82	**0.004**
Dysplastic ears (60%)	2009 GC	1.38	vs	2019 GC	1.57	**0.003**
	2009 Parent	1.76	vs	2019 Parent	1.84	0.066
Dysplasia of midphalanx of 5th digit (60%)	2009 GC	1.28	vs	2019 GC	1.41	**0.022**
	2009 Parent	1.62	vs	2019 Parent	1.73	**0.016**
Small head	2009 GC	1.66	vs	2019 GC	1.66	0.967
	2009 Parent	1.81	vs	2019 Parent	1.86	0.232
Brushfield spots on the iris of the eye	2009 GC	1.27	vs	2019 GC	1.45	**0.001**
	2009 Parent	1.62	vs	2019 Parent	1.73	**0.016**
Epicanthal folds	2009 GC	1.62	vs	2019 GC	1.67	0.514
	2009 Parent	1.73	vs	2019 Parent	1.77	0.343
Short, broad hands with short fingers	2009 GC	1.40	vs	2019 GC	1.59	**0.005**
	2009 Parent	1.75	vs	2019 Parent	1.76	0.822
5th finger clinodactyly	2009 GC	1.30	vs	2019 GC	1.44	**0.013**
	2009 Parent	1.58	vs	2019 Parent	1.62	0.307
Single palmar crease	2009 GC	1.38	vs	2019 GC	1.59	**0.001**
	2009 Parent	1.68	vs	2019 Parent	1.74	0.225
Sandal gap between 1st & 2nd toes	2009 GC	1.33	vs	2019 GC	1.51	**0.003**
	2009 Parent	1.63	vs	2019 Parent	1.73	**0.042**
Short stature	2009 GC	2.00	vs	2019 GC	2.17	**0.031**
	2009 Parent	1.93	vs	2019 Parent	1.97	0.428
Enlarged tongue	2009 GC	1.76	vs	2019 GC	1.92	**0.045**
	2009 Parent	2.05	vs	2019 Parent	2.25	**< 0.0001**

Syndactyly of 2nd and 3rd toes	2009 GC	1.28	vs	2019 GC	1.43	**0.005**
	2009 Parent	1.58	vs	2019 Parent	1.71	**0.005**
Medical Complications Associated with Down syndrome	**Group**	**Rating**		**Group**	**Rating**	**Sig < 0.05**
Early-Onset Alzheimer Disease	2009 GC	2.04	vs	2019 GC	2.18	0.071
(11% by age 50; 77% by age 70)	2009 Parent	1.91	vs	2019 Parent	2.30	**< 0.0001**
Dry, Hyperkeratotic Skin (75%)	2009 GC	1.42	vs	2019 GC	1.66	**< 0.0001**
	2009 Parent	1.75	vs	2019 Parent	1.90	**0.001**
One or More Congenital	2009 GC	2.84	vs	2019 GC	2.86	0.697
Abnormalities (50%)	2009 Parent	2.48	vs	2019 Parent	2.55	0.116
Heart Defect Possibly Requiring	2009 GC	2.96	vs	2019 GC	2.94	0.450
Open Heart Surgery (40-60%)	2009 Parent	2.82	vs	2019 Parent	2.85	0.411
Thyroid Disorders (up to 50%)	2009 GC	2.31	vs	2019 GC	2.56	**< 0.0001**
	2009 Parent	2.49	vs	2019 Parent	2.61	**0.004**
Obstructive Sleep Apnea (33%)	2009 GC	1.90	vs	2019 GC	2.42	**< 0.0001**
	2009 Parent	2.32	vs	2019 Parent	2.63	**< 0.0001**
Atlantoaxial and/or Atlantooccipital Instability (10-	2009 GC	1.68	vs	2019 GC	1.89	**0.012**
30%)	2009 Parent	2.10	vs	2019 Parent	2.24	**0.007**
Gastrointestinal Defect Possibly	2009 GC	2.45	vs	2019 GC	2.51	0.447
Requiring Surgery (12%)	2009 Parent	2.41	vs	2019 Parent	2.49	0.060
Autism Spectrum Disorder (ASD)	2009 GC	1.94	vs	2019 GC	1.86	0.306
(1%)	2009 Parent	1.89	vs	2019 Parent	1.98	0.067
Epilepsy (10%)	2009 GC	1.83	vs	2019 GC	1.97	0.058
	2009 Parent	1.97	vs	2019 Parent	2.20	**< 0.0001**
Increased Risk of Childhood	2009 GC	1.92	vs	2019 GC	2.08	**0.035**
Leukemia (2%)	2009 Parent	1.95	vs	2019 Parent	2.21	**< 0.0001**
Congenital or Acquired Cataracts	2009 GC	1.71	vs	2019 GC	1.92	**0.004**
	2009 Parent	1.82	vs	2019 Parent	2.15	**< 0.0001**
Vision Problems	2009 GC	2.19	vs	2019 GC	2.62	**< 0.0001**
	2009 Parent	2.27	vs	2019 Parent	2.50	**< 0.0001**
Hearing Loss	2009 GC	2.22	vs	2019 GC	2.65	**< 0.0001**
	2009 Parent	2.29	vs	2019 Parent	2.58	**< 0.0001**
Ear Problems	2009 GC	1.90	vs	2019 GC	2.45	**< 0.0001**
	2009 Parent	2.30	vs	2019 Parent	2.53	**< 0.0001**
Respiratory Problems	2009 GC	1.98	vs	2019 GC	1.96	0.781
	2009 Parent	2.33	vs	2019 Parent	2.52	**< 0.0001**
Increased Susceptibility to	2009 GC	1.98	vs	2019 GC	1.95	0.706
Infection	2009 Parent	2.32	vs	2019 Parent	2.45	**0.006**
Increased Susceptibility to	2009 GC	1.51	vs	2019 GC	1.72	**0.003**
Periodontal Disease	2009 Parent	2.02	vs	2019 Parent	2.21	**< 0.0001**
Increased Risk of Autoimmune	2009 GC	1.62	vs	2019 GC	1.88	**0.001**
Disease	2009 Parent	2.11	vs	2019 Parent	2.35	**< 0.0001**
Orthopedic problems	2009 GC	1.66	vs	2019 GC	1.81	0.051
	2009 Parent	2.13	vs	2019 Parent	2.36	**< 0.0001**
Psychiatric Disorders	2009 GC	1.64	vs	2019 GC	1.87	**0.002**
	2009 Parent	2.01	vs	2019 Parent	2.10	0.104
Tendency for Obesity	2009 GC	1.65	vs	2019 GC	1.86	**0.005**
	2009 Parent	2.09	vs	2019 Parent	2.19	**0.043**
Sterility in Males	2009 GC	1.73	vs	2019 GC	1.87	0.085
	2009 Parent	1.89	vs	2019 Parent	1.96	0.160
Increased Risk of Testicular	2009 GC	1.47	vs	2019 GC	1.69	**0.003**
Cancer in Males	2009 Parent	1.93	vs	2019 Parent	2.11	**< 0.0001**
Intellectual Disability & Developmental Delay	**Group**	**Rating**		**Group**	**Rating**	**Sig < 0.05**
Variable Range of Intellectual	2009 GC	2.98	vs	2019 GC	2.96	0.189
Disability from Mild to Moderate	2009 Parent	2.71	vs	2019 Parent	2.63	**0.049**
Mean (Average) IQ of 50	2009 GC	1.97	vs	2019 GC	2.08	0.140
	2009 Parent	1.86	vs	2019 Parent	1.86	0.989
Range of IQ from 20 to 80	2009 GC	1.98	vs	2019 GC	2.18	**0.011**
	2009 Parent	1.72	vs	2019 Parent	1.90	**0.001**
Developmental Delay in	2009 GC	2.94	vs	2019 GC	2.97	0.275
Achieving Milestones	2009 Parent	2.74	vs	2019 Parent	2.67	**0.029**
Need for Physical Therapy	2009 GC	2.66	vs	2019 GC	2.75	0.123
	2009 Parent	2.76	vs	2019 Parent	2.74	0.493
Need for Occupational Therapy	2009 GC	2.65	vs	2019 GC	2.75	0.095

	2009 Parent	2.75	vs	2019 Parent	2.73	0.489
Need for Speech & Language	2009 GC	2.67	vs	2019 GC	2.75	0.109
Therapy	2009 Parent	2.78	vs	2019 Parent	2.75	0.377
Need for Early Intervention &	2009 GC	2.86	vs	2019 GC	2.90	0.241
Case Management	2009 Parent	2.87	vs	2019 Parent	2.86	0.707
Long-term Prognosis	**Group**	**Rating**		**Group**	**Rating**	**Sig < 0.05**
Participate in Community Sports,	2009 GC	2.52	vs	2019 GC	2.65	0.051
Activities, & Leagues	2009 Parent	2.76	vs	2019 Parent	2.66	**0.004**
Participate in Special Olympics &	2009 GC	2.35	vs	2019 GC	2.33	0.843
Therapeutic Recreation	2009 Parent	2.60	vs	2019 Parent	2.55	0.207
Inclusion in Regular Classes	2009 GC	2.65	vs	2019 GC	2.58	0.271
	2009 Parent	2.77	vs	2019 Parent	2.77	0.923
Special Education Classes	2009 GC	2.73	vs	2019 GC	2.74	0.915
	2009 Parent	2.56	vs	2019 Parent	2.48	**0.048**
Complete High School	2009 GC	2.47	vs	2019 GC	2.51	0.596
	2009 Parent	2.75	vs	2019 Parent	2.70	0.181
Attend College or Post-Secondary	2009 GC	2.02	vs	2019 GC	2.33	**< 0.0001**
Education	2009 Parent	2.58	vs	2019 Parent	2.55	0.519
Supported, Competitive	2009 GC	2.17	vs	2019 GC	2.56	**< 0.0001**
Employment	2009 Parent	2.69	vs	2019 Parent	2.70	0.690
Live Independently	2009 GC	2.42	vs	2019 GC	2.57	**0.048**
	2009 Parent	2.68	vs	2019 Parent	2.69	0.712
Live in Group Home	2009 GC	2.58	vs	2019 GC	2.60	0.744
	2009 Parent	2.40	vs	2019 Parent	2.39	0.904
Have Friends	2009 GC	2.73	vs	2019 GC	2.76	0.576
	2009 Parent	2.89	vs	2019 Parent	2.86	0.262
Have Intimate Relationships	2009 GC	2.22	vs	2019 GC	2.45	**0.006**
	2009 Parent	2.54	vs	2019 Parent	2.59	0.316
Get Married	2009 GC	2.18	vs	2019 GC	2.26	0.856
	2009 Parent	2.45	vs	2019 Parent	2.47	0.570
Life Expectancy (ranging into	2009 GC	2.71	vs	2019 GC	2.76	0.314
50s-60s)	2009 Parent	2.55	vs	2019 Parent	2.51	0.281
More Like Other Children Than	2009 GC	2.68	vs	2019 GC	2.88	**0.002**
Different	2009 Parent	2.95	vs	2019 Parent	2.89	**0.002**
Impact on the Family	**Group**	**Rating**		**Group**	**Rating**	**Sig < 0.05**
Impact on Other Siblings - More	2009 GC	2.15	vs	2019 GC	2.28	0.153
Compassionate & Caring	2009 Parent	2.61	vs	2019 Parent	2.73	**0.007**
Impact on Other Siblings - Less	2009 GC	2.01	vs	2019 GC	2.16	0.116
Attention & Resentful	2009 Parent	1.75	vs	2019 Parent	2.11	**< 0.0001**
Impact on Marriage - Strengthens	2009 GC	1.98	vs	2019 GC	2.16	0.072
Relationship	2009 Parent	2.37	vs	2019 Parent	2.59	**< 0.0001**
Impact on Marriage – Strains	2009 GC	1.92	vs	2019 GC	2.15	**0.029**
Relationship	2009 Parent	1.70	vs	2019 Parent	2.15	**< 0.0001**
Impact on Grandparents -	2009 GC	1.91	vs	2019 GC	1.79	0.821
Supportive & Welcoming	2009 Parent	2.37	vs	2019 Parent	2.47	0.051
Impact on Grandparents - Limited	2009 GC	1.56	vs	2019 GC	1.77	**0.029**
Interaction	2009 Parent	1.58	vs	2019 Parent	1.92	**< 0.0001**
Impact on Extended Family Members - Supportive &	2009 GC	1.64	vs	2019 GC	1.80	0.071
Welcoming	2009 Parent	2.35	vs	2019 Parent	2.40	0.366
Impact on Extended Family	2009 GC	1.55	vs	2019 GC	1.77	**0.014**
Members - Limited Interaction	2009 Parent	1.57	vs	2019 Parent	1.89	**< 0.0001**
Impact on Other Relationships -	2009 GC	1.65	vs	2019 GC	1.82	0.070
Supportive & Welcoming	2009 Parent	2.37	vs	2019 Parent	2.44	0.160
Impact on Other Relationships -	2009 GC	1.57	vs	2019 GC	1.79	**0.017**
Lose Social Circle	2009 Parent	1.58	vs	2019 Parent	1.98	**< 0.0001**
Financial Impact - No Difference	2009 GC	1.62	vs	2019 GC	1.84	**0.038**
	2009 Parent	1.85	vs	2019 Parent	2.14	**< 0.0001**
Financial Impact - More	2009 GC	2.05	vs	2019 GC	2.12	0.464
	2009 Parent	1.94	vs	2019 Parent	2.23	**< 0.0001**
Time Commitment - No	2009 GC	1.63	vs	2019 GC	1.86	**0.031**
Difference	2009 Parent	1.69	vs	2019 Parent	2.07	**< 0.0001**
Time Commitment - More	2009 GC	2.22	vs	2019 GC	2.23	0.884
	2009 Parent	2.30	vs	2019 Parent	2.42	**0.023**

Informational Resources & Referrals	Group	Rating		Group	Rating	Sig ≤ 0.05
Local Support Group(s)	2009 GC	2.89	vs	2019 GC	2.92	0.397
	2009 Parent	2.90	vs	2019 Parent	2.89	0.711
Advocacy Organizations & Websites	2009 GC	2.77	vs	2019 GC	2.74	0.505
	2009 Parent	2.81	vs	2019 Parent	2.82	0.810
Early Intervention Centers	2009 GC	2.70	vs	2019 GC	2.77	0.272
	2009 Parent	2.96	vs	2019 Parent	2.93	**0.024**
Printed / Written Material	2009 GC	2.85	vs	2019 GC	2.81	0.325
	2009 Parent	2.84	vs	2019 Parent	2.68	**< 0.0001**
Fact Sheets / Brochures	2009 GC	2.75	vs	2019 GC	2.60	**0.007**
	2009 Parent	2.78	vs	2019 Parent	2.66	**< 0.0001**
Books	2009 GC	2.52	vs	2019 GC	2.26	**< 0.0001**
	2009 Parent	2.74	vs	2019 Parent	2.62	**0.001**
Contact with families raising a child with Down syndrome	2009 GC	2.61	vs	2019 GC	2.44	**0.007**
	2009 Parent	2.91	vs	2019 Parent	2.89	0.385
Adoption Agencies	2009 GC	1.99	vs	2019 GC	2.02	0.778
	2009 Parent	1.79	vs	2019 Parent	1.98	**0.002**
Alternative / Nonconventional Therapies	2009 GC	1.40	vs	2019 GC	1.57	**0.032**
	2009 Parent	1.91	vs	2019 Parent	2.17	**< 0.0001**
Specialist Referral(s)	2009 GC	2.44	vs	2019 GC	2.58	0.060
	2009 Parent	2.68	vs	2019 Parent	2.74	0.132
Counselor or Family Therapist Referral(s)	2009 GC	2.07	vs	2019 GC	2.04	0.765
	2009 Parent	2.36	vs	2019 Parent	2.50	**0.004**
Pastoral Counseling Referral(s)	2009 GC	1.81	vs	2019 GC	1.80	0.879
	2009 Parent	2.14	vs	2019 Parent	2.25	0.059