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# Essential Informational Needs of Parents Receiving a Turner Syndrome Diagnosis: Parent and Genetic Counselor Perspectives

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# ESSENTIAL INFORMATIONAL NEEDS OF PARENTS RECEIVING A TURNER SYNDROME DIAGNOSIS: PARENT AND GENETIC COUNSELOR PERSPECTIVES

by

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ABSTRACT

Turner syndrome affects approximately 1 in 2,500 live female births, and etiology includes the partial or complete loss of the second X-chromosome. Prognosis varies depending on whether a diagnosis is made prenatally or postnatally. Current recommendations state that genetic counselors should be involved in the diagnosis; however, guidelines for what information to include during an initial diagnosis do not exist. The aim of this study was to identify which informational items related to Turner syndrome are considered most essential by parents and genetic counselors.

A survey including 100 informational items related to Turner syndrome was sent to genetic counselors and parents whose children were diagnosed with Turner syndrome. Participants ranked the importance of each informational item for an initial diagnosis. Information that both genetic counselor and parent groups ranked within the top 30 items was deemed "essential" for an initial discussion of a Turner syndrome diagnosis.

Of the top 30-ranked items for each group, 13 items were deemed essential for an overall diagnosis, 21 informational items were deemed essential for a prenatal diagnosis, and 20 informational items were deemed essential for a postnatal diagnosis. There were also statistically significant differences in the item ratings between each survey group, which included prenatal genetic counselors, postnatal genetic counselors, and parents who have received either a prenatal or postnatal diagnosis for their child. Findings of our study may provide a guide for what providers should focus on when presenting an initial Turner syndrome diagnosis to parents in the future.

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# CHAPTER 1: BACKGROUND

## 1.1 TURNER SYNDROME OCCURRENCE

Turner syndrome affects approximately 1 in 2,500 live female births and approximately 1% to 2% of all conceptions (Larizza et al., 2011). Etiology includes the partial or complete loss of the second X-chromosome in phenotypic females (Gravholt et al., 2017; Shankar & Backeljauw, 2018). Previous research has shown that Turner syndrome is the most common chromosomal abnormality in females and that monosomy X is the third most common chromosomal aneuploidy seen in spontaneous abortions during the first trimester (Elsheikh et al., 2002; Jia et al., 2015). Turner syndrome is not significantly correlated with maternal age (Kim et al., 2013). Major differences in prenatal and postnatal features and survivability of Turner syndrome are recognized with a suggested fetal demise rate of 99% from the first trimester to term (Surerus et al., 2003). Individuals who survive to birth have a reduced life expectancy of up to 13 years, but this life expectancy can be increased with appropriate intervention (Elsheikh et al., 2002).

## 1.2 CYTOGENETIC ETIOLOGIES OF TURNER SYNDROME

Typically, Turner syndrome results from partial or complete loss of the second X-chromosome in phenotypic females (Gravholt et al., 2017; Shankar & Backeljauw, 2018). There are several karyotypes found in Turner syndrome live births (Gravholt et al., 2017). A table differentiating the various cytogenetic etiologies of Turner Syndrome and their frequencies can be found below (Table 1.1).

**Table 1.1 Turner syndrome chromosomal frequency and type**

| Diagnosis | Karyotype Result | Frequency |
|---|---|---|
| Monosomy X | 45,X | 40%-50% |
| Monosomy X mosaicism, normal karyotype | 45X/46,XX | 15%-25% |
| Mixed gonadal dysgenesis | 45,X/46,XY | 10%-12% |
| Isochromosome Xq or isodicentric Xp | 46,X i(xq) or 46,X,idic(Xp) | 10% |
| Monosomy X mosaicism with triple X | 45,X/47,XXX or 45,X/46,XX/47,XXX | 3% |
| Unbalanced X-autosome translocation | Variable karyotype | Rare |
| Xp22.3 deletion | 46,XX, del(p22.3) | Unknown |
| Ring X chromosome | 46,X,r(X)/46,XX | Unknown |

Evidence shows that the specific karyotype of an individual patient does not always predict the phenotype. However, compared to patients with a 45,X karyotype, individuals with 45,X/46,XX mosaicism typically have a milder phenotype (Gravholt et al., 2017). A study including 126 women with Turner syndrome who had variable karyotypes found that webbed neck, high arched palate, increased intermammillary distance, low hairline, keloid, epicanthus folds, and all cardiac malformations were more common in women with a 45,X karyotype than women with a 45,X/46,XX karyotype. Additionally, women with 45,X/46,XX mosaicism were diagnosed eight years later on average than women with a 45,X karyotype (ages 18 and 10, respectively) (El-Mansoury et al., 2007). Individuals with 45,X/46,XX mosaicism diagnosed postnatally tend to have a more severe presentation than when identified prenatally. Many individuals who receive a prenatal diagnosis are diagnosed incidentally because karyotype analysis is performed for reasons unrelated to Turner syndrome such as advanced maternal age or abnormal prenatal screening (Gravholt et al., 2017; Gunther et al., 2004). A study comparing phenotype data for 25 patients prenatally diagnosed with 45,X/46,XX mosaicism and 58 patients postnatally diagnosed with 45,X/46,XX mosaicism found that those prenatally diagnosed have milder phenotypes than their postnatally diagnosed

counterparts. Prenatally diagnosed women were more likely to have normal growth and normal secondary sexual development (Tokita & Sybert, 2016).

## 1.3 PRENATAL PRESENTATION, SCREENING, AND DIAGNOSIS OF TURNER SYNDROME

The first signs of Turner syndrome are often found during the prenatal period because of the high incidence of structural anomalies detectable on ultrasound (Redel & Backeljauw, 2018). Turner syndrome is also often initially detected by cell-free fetal DNA screening, also known as non-invasive prenatal testing (NIPT) (Howard-Bath et al., 2018). Since the integration of NIPT in obstetric practice, increasing incidence of early diagnosis of Turner syndrome, before structural anomalies can be detected on an 18-20-week ultrasound, has been noted (Baena et al., 2004).

### *1.3.1 ULTRASOUND FINDINGS*

Screening procedures can indicate a higher risk for Turner syndrome during pregnancy. Ultrasonographic findings can suggest a diagnosis of Turner syndrome in utero. In the first trimester, increased nuchal translucency may suggest Turner syndrome, but the presence of a cystic hygroma increases the likelihood of a Turner syndrome diagnosis significantly (Nicolaides et al., 1992). Other findings including coarctation of the aorta and/or left-sided heart defects, brachycephaly, renal dysplasia (including horseshoe kidney), polyhydramnios, oligohydramnios, and growth restriction indicate a higher likelihood of Turner syndrome (Redel & Backeljauw, 2018).

A study by Baena et al. (2004) found that out of 125 prenatally detected Turner syndrome cases, 84 (67.2%) were detected by ultrasound due to the presence of congenital anomalies. There were only eight cases of Turner syndrome that showed

congenital anomalies at birth that were not detected by ultrasound. Out of the 84 cases detected by ultrasound, 69 had cystic hygroma, 22 had hydrops fetalis, nine had congenital heart defects (most commonly coarctation of the aorta), four had pulmonary defects, four had nuchal thickening, three had renal defects, two had short femurs, two had central nervous system defects, and one had an abdominal wall defect (Baena et al., 2004).

### *1.3.2 FIRST TRIMESTER MATERNAL SERUM SCREENING*

Several other screening methods may lead to a suspicion for Turner syndrome with varying detection rates. Viuff et al. (2015) identified Turner syndrome in 87 out of 100,000 female fetuses out of an expected 209 using first trimester screening, indicating a 42% detection rate. Pregnancies with Turner syndrome showed consistently greater nuchal translucency and lower pregnancy-associated plasma protein A (Viuff et al., 2015).

### *1.3.3 SECOND TRIMESTER MATERNAL SERUM SCREENING*

A study analyzing second trimester maternal serum screening analyte levels in 22 pregnancies affected with Turner syndrome found that maternal serum inhibin A levels were significantly reduced in cases without hydrops, which represented 10 of the 22 cases. Levels of alpha-fetoprotein, unconjugated estriol, and human chorionic gonadotropin hormone were also reduced. In pregnancies with hydrops, inhibin A levels were elevated, with reduced unconjugated estriol, unaffected alpha-fetoprotein, and elevated human chorionic gonadotropin hormone levels (Lambert-Messerlian et al., 1998).

Another study reviewing 130,595 maternal serum samples assayed between January 1989 and December 1997 and their outcomes found that alpha-fetoprotein and unconjugated estriol concentrations were slightly reduced in pregnancies with Turner syndrome, and human chorionic gonadotropin levels were increased, specifically in pregnancies with hydrops. Thus, pregnancies with an increased risk for Down syndrome based on multiple marker screening should be counseled about the possibility of Turner syndrome (Ruiz et al., 1999). Second trimester maternal serum screening detection rates for Turner syndrome were not present in either of these studies.

### *1.3.4 NON-INVASIVE PRENATAL TESTING*

NIPT has been used since 2011 to assess risk for common aneuploidies including Monosomy X (Petersen et al., 2017). A published report indicated that since its introduction into clinics in 2011, NIPT has become the most common screening method for sex chromosome aneuploidies (49%), followed by ultrasound (37%) (Howard-Bath et al., 2018). Given an increased NIPT uptake, there have been more conversations about a possible Turner syndrome diagnosis in pregnancy. However, the number of prenatal sex chromosome aneuploidies diagnosed annually has not significantly increased as a result of this technology becoming widely used (Howard-Bath et al., 2018). Lu et al. (2021) retrospectively analyzed results from 45,773 maternal blood samples from singleton pregnancies that underwent NIPT, and 147 pregnancies screened positive for 45,X. Of the 56 pregnancies that underwent subsequent invasive prenatal diagnosis, seven were true positive, with a 12.50% positive predictive value. The authors hypothesized that the low positive predictive value of NIPT for monosomy X could be due to nonrandom X chromosome inactivation in placental tissue, with the paternal X chromosome tending to

be inactivated in XX female trophoblasts (Lu et al., 2021). Another study reports that the predictive ability of NIPT for sex chromosome aneuploidies could be influenced by age-related X chromosome loss in normal female cells (Xu et al., 2019). Lu et al. (2021) did not observe this phenomenon in the retrospective study, however. NIPT is not considered diagnostic, and it is recommended that a positive result be followed up with prenatal karyotype (Gravholt et al., 2017).

### *1.3.5 CHORIONIC VILLUS SAMPLING AND AMNIOCENTESIS*

Chorionic villus sampling (CVS) or amniocentesis may be performed to obtain karyotype data of the pregnancy and confirm a diagnosis of Turner syndrome (Lu et al., 2021). Typically, CVS is performed between 10- and 13-weeks' gestation (ACOG, 2016). The procedure involves ultrasound-guided aspiration of placental tissue using a percutaneous transabdominal or transvaginal/transcervical approach (Alfirevic et al., 2003). There are two methods in which the placental villi are analyzed. The direct method uses rapidly dividing cells from the cytotrophoblast, and the long-term culture method analyzes fibroblast-like cells cultured from the mesenchymal core of the CVS sample (Goldberg & Wohlferd, 1997). Specimen processing time for CVS typically takes five to seven days. Because results are available earlier in pregnancy than with amniocentesis, CVS results may allow for more pregnancy management options (ACOG, 2016).

Amniocentesis is typically performed between 15- and 20-weeks' gestation and can be performed at a later gestational age (ACOG, 2016). The procedure involves an ultrasound-guided needle puncture through the overlaying skin into the uterus and amniotic cavity which is followed by aspiration of amniotic fluid containing fetal cells

(Alfirevic et al., 2003). Specimen processing time for amniocentesis typically takes seven to 14 days (ACOG, 2016).

Caughey et al. (2006) found that for nonanomalous fetuses with normal karyotype, the pregnancy loss rates for CVS and amniocentesis have decreased over time. For procedures performed at a prenatal diagnostic referral center from 1998 to 2003, no statistically or clinically significant differences in pregnancy loss rates between CVS and amniocentesis were found. In the period from 1998 to 2003, the pregnancy loss rate for CVS was reported to be 1/360 (0.28%), and the pregnancy loss rate for amniocentesis was reported to be 1/370 (0.27%) (Caughey et al., 2006).

A more recent meta-analysis analyzing miscarriage rates for CVS reported a 0.22% procedure-related miscarriage rate before 24 weeks. Another meta-analysis examining miscarriage rates for amniocentesis reported a 0.11% miscarriage rate. Both meta-analyses reported no significant difference in the rate of miscarriage between the procedure and control groups for CVS and amniocentesis (Akolekar et al., 2015). Although there is not a significant difference in termination rates for Turner syndrome between pregnancies diagnosed by CVS vs. amniocentesis, couples in one study were more inclined to terminate when the diagnosis was made during the first trimester (Mezei et al., 2004).

### *1.3.6 PRENATAL KARYOTYPE, CHROMOSOMAL MICROARRAY, AND FLUORESCENCE IN SITU HYBRIDIZATION*

Laboratory tests including karyotype, fluorescence in situ hybridization (FISH), and chromosomal microarray analysis (CMA) can detect Turner syndrome using a CVS or amniocentesis sample. The most common laboratory test to detect Turner syndrome in

a fetus is karyotype analysis. Karyotype analysis must be performed on cultured cells and results typically take seven to 14 days after a sample is obtained. The diagnostic accuracy of karyotype analysis is greater than 99% for Turner syndrome; however, karyotype analysis may not detect mosaicism in the fetus if mosaicism is not present in the cells obtained for genetic testing (ACOG, 2016). Because both CVS and amniocentesis obtain fibroblast cells for analysis, the constitutional karyotype of a fetus diagnosed with Turner syndrome prenatally is uncertain, especially in mosaic individuals. Therefore, chromosome analysis should be repeated after birth in every patient to obtain the most accurate diagnosis (Gravholt et al., 2017).

Turner syndrome can also be prenatally identified by CMA. This technique can be performed directly on uncultured tissue or cultured cells. Results typically take three to seven days to return after a sample is obtained, and because it can be performed on nonviable cells, CMA is preferred over karyotype in the case of a fetal death or stillbirth (ACOG, 2016). In a cohort of 187 patients with Turner syndrome, array-based models of X-chromosome structure were concordant with karyotype in 104/116 (90%) of comparable cases. Although single nucleotide polymorphism (SNP) array did not detect X;autosome translocations in three cases, it did identify two cases of derivative Y chromosomes and 13 large copy number variants not detected through traditional karyotyping (Prakash et al., 2014). If structural anomalies are detected by prenatal ultrasound, CMA is recommended as the primary test to replace karyotype analysis. However, if structural anomalies detected on ultrasound are suggestive of Turner syndrome specifically, karyotype with or without FISH analysis may be offered before CMA (ACOG, 2016).

Preliminary results of a potential Turner syndrome diagnosis can be obtained using FISH analysis, which can probe for specific chromosomes or chromosomal regions. This analysis can be performed on uncultured tissue or cultured cells and results typically become available within two days after a sample is obtained. However, false positive and false negative results from FISH have been reported, and FISH analysis is not considered diagnostic (ACOG, 2016). Therefore, clinical decision-making should be based on either confirmatory chromosome analysis or clinical (ACMG, 2000).

### *1.3.7 PRENATAL DIAGNOSIS AND PREGNANCY TERMINATION*

Pregnancy termination in the case of Turner syndrome is influenced by several factors including the fetal karyotype, the presence or absence of ultrasound anomalies, and parental concerns regarding the postnatal outcomes for their children (Mezei et al., 2004). A French study reporting pregnancy outcomes of 975 prenatally diagnosed Turner syndrome cases noted the overall pregnancy termination rate to be 81% after diagnosis. When first trimester sonographic abnormalities were present, 89% of pregnancies were terminated while 36% of incidentally diagnosed pregnancies were terminated. Pregnancies with a 45,X karyotype were terminated more often (93%) than pregnancies with a mosaic karyotype (36%) (Gruchy et al., 2014). A review analyzing data from several previous studies identified an average pregnancy termination rate of 76%, with a range of 33% to 100% in the case of Turner syndrome (Jeon et al., 2012). Additionally, Turner syndrome pregnancies were found to be terminated more frequently than pregnancies with other sex chromosome aneuploidies including 47,XXY, 47,XXX, and 47,XYY. Although the gestational age at the time of diagnosis played a role, patients' termination decisions were influenced more strongly by the severity of the disorder.

Abnormal sexual development and infertility were perceived to be the most influential factors in patients' decisions to terminate their pregnancies diagnosed with Turner syndrome (Mezei et al., 2004).

## 1.4 POSTNATAL PRESENTATION AND DIAGNOSIS

If a fetus is suspected to have Turner syndrome prenatally, a standard 20-cell karyotype is recommended which can identify at least 10% mosaicism after birth. Additional metaphases can be counted, or FISH can be performed if mosaicism is not demonstrated on karyotype but is highly suspected (Gravholt et al., 2017).

### *1.4.1 NEONATAL PRESENTATION*

The clinical presentation of Turner syndrome varies throughout life. During the neonatal period, clinical manifestations that should prompt consideration for karyotyping include: failure to thrive during the first year (50%), cystic hygroma, hydrops, lymphedema of the extremities (25%), left-sided heart defects [especially coarctation of the aorta (7%-14%) and bicuspid aortic valve (14%-34%)], micrognathia (60%), high-arched palate (35%), epicanthal folds (20%), ptosis (10%), strabismus (15%), external ear differences (15%), low set ears, shield chest (30%), wide-spaced nipples, neck webbing (25%), short neck (87%), low posterior hairline (40%), horseshoe kidney (10%), partially duplicated kidney (5%-10%), absent kidney (2%-3%), multicystic kidney (<1%), ectopic kidney (<1%), Madelung deformity (5%), and nail dysplasia/hypoplasia (10%) (Gravholt et al., 2017; Redel & Backeljauw, 2018).

In general, congenital heart disease occurs in 23%-50% of individuals with Turner syndrome and is the most frequent cause for early mortality. Although coarctation of the aorta and bicuspid aortic valve are the most common congenital heart defects seen with

Turner syndrome, other less frequent anomalies include hypoplastic left heart syndrome, mitral valve anomalies, interrupted inferior vena cava with azygous continuation, cardiac dextroposition, ventricular septal defect, atrioventricular septal defect, pulmonary valve abnormalities, coronary artery anomalies, and patent ductus arteriosus (Gravholt et al., 2017; Mazzanti & Cacciari, 1998; Polkampally et al., 2011; Sybert, 1998; Viuff et al., 2016; Völkl et al., 2005). For certain congenital heart defects, individuals may undergo surgery to repair these abnormalities. A study measuring congenital heart defect surgical outcomes found that for aortic arch repair, those with Turner syndrome have longer operative and postoperative courses. However, individuals with Turner syndrome appear to respond to surgery as well as those without Turner syndrome. Both groups also appear to have low complication rates. Patients with Turner syndrome who have hypoplastic left heart syndrome tend to have poorer post-surgical outcomes than patients without Turner syndrome (Cramer et al., 2014).

### *1.4.2 CHILDHOOD AND ADULTHOOD PRESENTATION*

Throughout life, common features of Turner syndrome include growth failure and reduced adult height (95%-100%), glucose intolerance (15%-50%), type two diabetes (10%), type one diabetes, thyroiditis (15%-30%), hypothyroidism (3.2% annual incidence), hypertension (50%), elevated hepatic enzymes (50%-80%), celiac disease (8%), inflammatory bowel disease (2%-3%), nearsightedness (20%), infection of the middle ear (60%), hearing defects (30%), inverted nipples (5%), increased skin ridge count (30%), multiple pigmented naevi (25%), vitiligo (5%), alopecia (5%), bone age delay (85%), decreased bone mineral content (50%-80%), cubitus valgus (50%), short

fourth metacarpal (35%), genu valgum (35%), congenital hip luxation (20%), scoliosis (10%), and aortic dilation/aneurysm (3%-42%) (Gravholt et al., 2017).

### *1.4.3 NEUROCOGNITIVE AND PSYCHOSOCIAL PRESENTATION*

Although individuals with Turner syndrome typically have normal intelligence, they may have psychosocial or neurocognitive issues. Approximately 10% of females with Turner syndrome, especially those with a small ring X chromosome on karyotype, present with some form of intellectual disability. Many experience challenges in executive functioning including task handling, working memory, and processing speed. Additionally, challenges with visual-spatial perception, mathematics and reading comprehension, facial expression recognition, motor coordination, and motor learning are common (Shankar & Backeljauw, 2018). Approximately 40% have a specific (nonverbal) learning disorder (Gravholt et al., 2017). Individuals with Turner syndrome also commonly have attention deficit and hyperactivity disorder and autism spectrum disorders (Shankar & Backeljauw, 2018). Approximately 40% of individuals exhibit emotional immaturity, and approximately 25% exhibit psychological and behavioral problems including low self-esteem, social isolation, anxiety, and depression (Gravholt et al., 2017; Shankar & Backeljauw, 2018). However, in a study comparing children with Turner syndrome to typically developing children, researchers identified advantages in overall language ability in both receptive and expressive language, phonological skills, word recognition, and emerging lexico-semantic strength in the Turner syndrome cohort (Temple & Shephard, 2012).

### *1.4.4 LONG-TERM LIFE OUTCOMES*

Studies have shown that women and girls with Turner syndrome also have long-term life outcomes which may be different from the general U.S. female population. Gould et al. (2013) published a cross-sectional study comparing 240 women with Turner syndrome (ages 25-67 years old) to the general U.S. female population using normative data. The study found that women with Turner syndrome were more likely to earn a baccalaureate degree or higher than the general U.S. female population (70% vs. 30%). Researchers also found that adults with Turner syndrome were more likely to be employed than the general U.S. female population (80% vs. 70%). Women with Turner syndrome were less likely to marry than the general U.S. female population (50% vs. 78%) (Gould et al., 2013).

## 1.5 HEALTH SURVEILLANCE FOR TURNER SYNDROME

### *1.5.1 HEALTH SURVEILLANCE AT DIAGNOSIS*

Gravholt et al. (2017) made recommendations for screening in Turner syndrome at the time of diagnosis and throughout life. At the time of diagnosis, a weight/BMI, blood pressure, and thyroid function (free T4 and TSH) should be ascertained. Imaging of the thoracic aorta and heart with transthoracic echocardiography (TTE) and CT/cardiac magnetic resonance scan (CMR) should be performed. A renal ultrasound should also be performed as well as a dental and dermatological evaluation. Audiometric evaluation should be done at diagnosis if the patient is nine to 12 months old, and ophthalmological examination should be done at diagnosis if the patient is 12-18 months old. Additionally, clinical investigation for congenital hip dysplasia should be performed in newborns (Gravholt et al., 2017).

### *1.5.2 HEALTH SURVEILLANCE IN CHILDHOOD*

In childhood, weight/BMI and blood pressure should be obtained at every doctor's visit. Thyroid function evaluation and a skin examination should be done annually. From infancy to 16 years, a cardiology exam, TTE, CMR, and electrocardiogram (ECG) should be performed at intervals ranging between every six months to every five years depending on the presence of coarctation of the aorta, bicuspid aortic valve, or hypertension. An audiometric evaluation should be performed every three years. Starting at two years old and thereafter every two years, a celiac screen should be done. A skeletal assessment should be performed at five to six years old and again at 12-14 years old. After 10 years of age, aminotransferase, GGT, alkaline phosphatase, and HbA1c with or without fasting glucose should be measured annually. After nine to 11 years of age, 25-Hydroxyvitamin D should be measured every two to three years. A neuropsychological assessment should be performed at key transitional stages in schooling including preschool, school entry, transition to high school and higher education, or any time difficulties arise (Gravholt et al., 2017).

### *1.5.3 HEALTH SURVEILLANCE IN ADULTHOOD*

In adults with Turner syndrome, weight/BMI and blood pressure should be obtained at every doctor's visit. A cardiology exam, TTE, CMR, and ECG should be performed at intervals ranging from every six months to every five to 10 years depending on the presence of coarctation of the aorta, bicuspid aortic valve, or hypertension. A skin examination should be performed annually. Free T4 and TSH, aminotransferase, GGT, alkaline phosphatase, and HbA1c with or without fasting glucose should be measured annually. If there is at least one cardiovascular risk factor present (hypertension,

overweight, tobacco, diabetes, or physical inactivity), lipids should be measured annually. The levels of 25-Hydroxyvitamin D should be measured every three to five years, and an audiometric evaluation should be performed every five years. Bone mineral density should be ascertained every five years and when discontinuing estrogen, and celiac screening should be performed if the patient has suggestive symptoms. A neuropsychological evaluation should be performed on an as-needed basis (Gravholt et al., 2017).

## 1.6 DELAYED DIAGNOSIS AND ACCESS TO TREATMENT

### *1.6.1 DELAYED DIAGNOSIS OF TURNER SYNDROME*

Often, girls receive a delayed diagnosis of Turner syndrome (Morgan, 2007). Late diagnosis of Turner can lead to missed potential for intervention which can improve health and quality of life (Gravholt et al., 2017). One study reported that from a medical record review of 81 girls with Turner syndrome, a range of age at diagnosis was noted from prenatal to 16.8 years of age, with 49% being diagnosed in childhood or adolescence. Only half of the patients were diagnosed prenatally or in infancy, and the average age for the remaining participants was 8.8 years old.

In this study, unexplained short stature was the most common indication for karyotype in childhood and adolescence with a diagnosis being made seven years on average after patients' short stature were clinically evident on growth curves. Additionally, 26/39 (67%) of patients who were not diagnosed in infancy would have been diagnosed at four years if karyotype analysis had been performed when their height fell below the 5$^{th}$ percentile. The authors explain that early diagnosis is necessary to

maximize growth and development of girls with Turner syndrome and allow for earlier initiation of therapies (Savendahl & Davenport, 2000).

### *1.6.2 IMPACTS OF DELAYED TREATMENT*

Earlier diagnosis of Turner syndrome facilitates treatment and management, especially growth hormone therapy (Gonzalez & Witchel, 2012). If growth hormone treatment is performed early—as early as one year in some females—a normal final height can be achieved (Gravholt et al., 2017). On average, in untreated individuals, height is around 20 cm shorter than the typical population. At birth, the mean length and weight of infants with Turner syndrome typically falls in the low normal range, but deceleration in linear growth velocity can occur as early as 18 months (Gonzalez & Witchel, 2012). As soon as decreased growth velocity presents, growth hormone therapy should be considered (Bondy, 2007). If treatment is initiated before four years of age, height outcomes for girls with Turner syndrome are significantly improved (Linglart et al., 2011). The later that growth hormone treatment is initiated, the less effective it is (Lee & Conway, 2014).

Late diagnosis of Turner syndrome leads to less effective hormone replacement therapy (Lee & Conway, 2014). Gonadal dysgenesis associated with primary ovarian failure is a hallmark feature of Turner syndrome, and most girls require hormone replacement therapy for breast development, uterine growth, and bone health (Gonzalez & Witchel, 2012). If gonadotropins are elevated and LH and FSH concentrations confirm ovarian failure, hormone replacement therapy should begin at 11-12 years of age (Gonzalez & Witchel, 2012; Gravholt et al., 2017). Although higher doses of estrogen administered before 14 years may impact growth and final height potential, low doses of

estrogen do not interfere with growth hormone therapy response (Gravholt et al., 2017). Typically, hormone replacement therapy for women with Turner syndrome ends approximately 40-50 years after initiation. Optimal hormone replacement therapy beginning in adolescence is also important for maintaining bone mineral density, which is typically lower in individuals with Turner syndrome (Gonzalez & Witchel, 2012). Patients who receive hormone replacement therapy at an older age have been shown to have psychological and social/psychosexual activity disadvantages (Christopoulos et al., 2008). In a study examining the psychological well-being in 63 women with Turner syndrome, eight participants who did not receive hormone replacement therapy had a lack of sex hormones and had lower psychological wellbeing than those who had sufficient levels of sex hormones. Six of these participants had a lack of hormone replacement therapy because of a diagnosis later in adult life (Boman et al., 2004).

Later diagnosis can lead to a lost opportunity to preserve oocytes as a possible fertility option. If Turner syndrome is detected early, patients can be offered fertility treatment at a young age (Gravholt et al., 2017). Women with Turner syndrome may possibly conceive a pregnancy through assisted reproductive technologies, and young women with Turner syndrome may have the option of cryopreservation (Karnis, 2012). However, Turner syndrome is considered a contraindication to carrying a pregnancy due to cardiovascular complications, including a risk of dying from aortic dissection or rupture from the increased cardiovascular demands of pregnancy. The estimated maternal mortality rate for women with Turner syndrome who are pregnant is approximately 2%, which is increased above the general population maternal mortality rate of 0.013%. Therefore, while earlier diagnosis may lead to the opportunity for cryopreservation,

women with Turner syndrome are encouraged to achieve pregnancy through gestational surrogacy or to pursue adoption (ASRN, 2012; Karnis, 2012).

Delayed diagnosis of Turner syndrome may lead to later implementation of measures that can address the behavioral traits associated with the condition such as difficulties with mathematical processing, delayed social skills, and feelings of isolation (Lee & Conway, 2014). Delayed diagnosis can also have lasting psychological implications for individuals diagnosed later in life. A research study by Reimann et al. (2018) analyzed the psychosocial profiles of 110 women with Turner syndrome who were 22 and older, 58 who received an early diagnosis (average age 1.5 years) and 52 who received a late diagnosis (average age 16.25 years). Late diagnosis was significantly associated with mild to severe depressive symptoms—12.1% of early diagnosis individuals and 40.4% of late diagnosis individuals had depressive symptoms. Participants who were diagnosed later also reported more severe feelings of self-dislike, worthlessness, personal failure, and suicidal thoughts. Because delayed diagnosis can often lead to inapplicable preventative treatment options, women with later-diagnosed Turner syndrome may feel a loss of control over their well-being (Reimann et al., 2018).

## 1.7 TURNER SYNDROME DIAGNOSTIC GUIDELINES

Multiple guidelines for earlier diagnosis of Turner syndrome have been proposed. Savendahl and Davenport proposed guidelines in 2000 for screening girls for Turner syndrome so that females who have suggestive findings may be diagnosed earlier in life. The guidelines were based on findings that are relatively infrequent in the general population but have an increased frequency in individuals with Turner syndrome. The guidelines propose that Turner syndrome should be considered for any infant, child, or

adolescent girl with unexplained short stature (height <5$^{th}$ percentile), lymphedema, webbed neck, coarctation of the aorta, or delayed puberty. Additionally, the condition should be considered for any girl who has at least two or more other findings consistent with Turner syndrome including nail dysplasia, high arched palate, short fourth metacarpal, or strabismus. Other features including nonverbal learning disability, epicanthal folds, ptosis, cubitus valgus, multiple nevi, renal malformations, bicuspid aortic valve, recurrent otitis media, and need for glasses may also support the diagnosis (Savendahl & Davenport, 2000).

More recent guidelines have been developed that include indications for chromosome analysis to diagnose Turner syndrome. Turner syndrome should be considered in any female with fetal cystic hygroma or hydrops, idiopathic short stature, obstructive left-sided congenital heart defect (bicuspid aortic valve, coarctation of the aorta, aortic stenosis, mitral valve anomalies, and hypoplastic left heart syndrome), unexplained delayed puberty/menarche, a couple with infertility, or characteristic facial features (down-slanted palpebral fissures, epicanthal folds, low-set anomalous pinnae, micrognathia, narrow palate, short broad neck, and webbing). Turner syndrome should also be considered if a female has at least two of the following: renal anomaly (horseshoe, absence, or hypoplasia), Madelung deformity, neuropsychologic problems and/or psychiatric issues, multiple typical or melanocytic nevi, dysplastic or hyperconvex nails, other congenital heart defects (partial anomalous pulmonary venous return, atrial septal defect—secundum type, and ventricular septal defects), or hearing impairment at less than 40 years of age together with short stature (Gravholt et al., 2017).

## 1.8 PARENT PERSPECTIVES AND EXPERIENCES WITH DIAGNOSIS

Parents often find it challenging to cope with their child's Turner syndrome diagnosis. A study where 65 parents were interviewed about their emotional reactions at the time of diagnosis and subsequently at the time of the interview found that most parents felt sadness, shock, anger, and shame at the time of diagnosis. Some parents also experienced guilt. Most parents also found it difficult to cope with their child's infertility, 38% expected fewer opportunities for their daughter to find a job, and 54% anticipated difficulty for their daughter to find a romantic partner (Slijper et al., 1998).

Parent attitudes regarding their diagnostic experiences are variable, and the level of parental satisfaction at the time of diagnosis has been shown to coincide with physician knowledge and support. A previous study interviewing 44 parents (33 mothers and 11 fathers) whose daughters were diagnosed with Turner syndrome found that parents felt it was important to receive a diagnosis because it would make it possible for them to better understand the condition and its effect on their daughters. Of the 44 parents, 23 were dissatisfied with their diagnostic experience and 21 were satisfied. Parental satisfaction when receiving an initial diagnosis was associated with the doctor's ability to provide relevant, understandable, and comprehensive information. Parents appreciated when physicians made referrals to colleagues with more experience with Turner syndrome to provide more information. Parents also felt satisfaction from the ability of the doctor to support their feelings of grief and worries about the future. Dissatisfied parents thought that their feelings were not considered by the physician and left alone with the diagnosis without being offered further consultation with healthcare professionals (Starke & Moller, 2002; Starke et al., 2002).

Researchers also found that these parents' motives for seeking more information about Turner syndrome were related to their experiences at the time of diagnosis and whether they were satisfied or dissatisfied. Dissatisfied parents sought more information about Turner syndrome because doctors had been unable to provide full information about the syndrome. These parents perceived that doctors were incapable of explaining the condition due to a lack of knowledge. Dissatisfied parents also perceived that they were responsible for their daughters' access to appropriate treatment and needed to ensure that health professionals covered all aspects of the condition, adhering to the most recent recommendations. Parents who were satisfied with their diagnostic experience felt that they received 'good information' at the time of diagnosis but were curious and interested in the condition. Satisfied parents also wanted to be able to answer questions about Turner syndrome that were raised by others (Starke & Moller, 2002).

Several parents who participated in the Starke et al. (2002) study reported that the information received during the diagnosis from their physician was problematic. Many recounted that the name of the diagnosis was not given and that its consequences were not explained. They also felt that some information about the diagnosis was incorrect, and some parents reported that they did not receive any information about the condition at the time of diagnosis. Half of the participating parents were informed about possible medical intervention, and over half the group was told that their daughters were presumed to be infertile, which some parents reported as being the worst implications of a Turner syndrome diagnosis (Starke et al., 2002).

When receiving a diagnosis for their child, parents also reported that the doctor should describe various aspects of the condition and emphasize that certain

manifestations do not appear at the same time. Parents also suggested that written information should be given in conjunction with verbal information. Some parents said that parents with a new diagnosis should be able to contact other families who have a child with Turner syndrome, and they should be given information about local and national Turner syndrome foundations (Starke et al., 2002).

With a prenatal Turner syndrome diagnosis, parents' interactions with their providers have been shown to influence pregnancy outcomes. Hall et al. (2003) examined the relationships between the information health professionals presented about sex chromosome anomalies and termination rates in 23 pregnancies. Researchers found that parents who were given a prenatal diagnosis of a sex chromosomal abnormality with a greater amount of negative information were more likely to terminate their pregnancy. Providers included midwives, obstetricians, genetic counselors, and general practitioners. Medical professionals who participated in this study mentioned that they often had little knowledge about the condition they were informing the parents about and found these situations stressful (Hall et al., 2003). Mezei et al. (2004) reported that although obstetricians who delivered the diagnosis of a sex chromosome aneuploidy adhered to the principle of nondirectiveness, 60% of women reported that the counseling for their prenatal diagnosis was directive.

It is recommended that providers include accurate information, both positive and negative, when delivering a diagnosis of sex chromosomal abnormalities. However, the information parents are given when they are first informed about a sex chromosome anomaly is often a matter of chance—providers in the study said that they often delivered diagnoses about conditions that they knew little about themselves. There is also little

consensus on the content of information that parents need about these conditions when receiving an initial diagnosis (Hall et al., 2003).

## 1.9 PHYSICIAN AND GENETIC COUNSELOR GUIDELINES

With increasing availability of prenatal ultrasound and prenatal diagnosis, health professionals including obstetricians, nurses, geneticists, and genetic counselors are often involved in the diagnostic process and discussion of test results (Loscalzo et al., 2006). Current recommendations state that for sex chromosome abnormalities, prenatal genetic counseling by a geneticist or genetic counselor should be provided before and after any prenatal diagnostic procedure (Gravholt et al., 2017). For Turner syndrome, it is essential that prenatal pretest and posttest counseling be provided by genetic counselors, geneticists, and endocrinologists who are highly familiar with the condition. These health professionals should also work with organizations such as Turner syndrome societies that may connect parents with others who have relevant experience (Loscalzo et al., 2006). Physicians and genetic counselors involved in diagnostic counseling need to be fully informed about the prognosis, complications, quality of life, and recent advances in management for Turner syndrome (Saenger et al., 2001). In addition, medical professionals must consider local cultural differences in parental perceptions and try to minimize discrepancies in the information that is delivered between providers (Mezei et al., 2004). Providers should also emphasize that individuals with Turner syndrome can be healthy, happy, and productive members of society (Saenger et al., 2001).

## 1.10 RATIONALE

Turner syndrome is a widely variable condition with vastly different outcomes depending on the cytogenetic etiology, whether features present prenatally or postnatally,

and associated birth defects. With a timely diagnosis by informed healthcare professionals, outcomes for individuals with Turner syndrome have been shown to be improved with a greater quality of life achieved (Gravholt et al., 2017).

Previous studies have shown that interactions with providers when receiving a Turner syndrome diagnosis are often less informative than what parents desire. Many were not presented with a name or information about the condition or given information about medical intervention (Starke et al., 2002). With a prenatal diagnosis, the presentation of information affected whether the couple chose to terminate the pregnancy, although many providers felt uninformed about the diagnosis themselves (Hall et al., 2003). Parents who have received either a prenatal or postnatal diagnosis of Turner syndrome desire to be aware of both positive and negative information about the condition. Greater satisfaction was reported when their providers were knowledgeable of the condition and could present relevant, comprehensive information while also providing emotional support. These findings have suggested that providers should be able to give parents information about Turner syndrome organizations through which they can connect with other families who have a child with Turner syndrome (Hall et al., 2003; Starke et al., 2002).

Balanced information for parents emphasizing both the strengths and challenges of the condition is important when delivering a Turner syndrome diagnosis. Although recommendations for medical interventions following a diagnosis are established, a strong consensus on the content of information that parents need to be presented with and what aspects they feel are most important has not been delineated. Ascertaining the information that parents and providers feel is most essential for an initial diagnosis of

Turner syndrome may guide future recommendations for providers when delivering a diagnosis to parents and ultimately improve overall patient satisfaction. Currently, guidelines for what specifically to include in this presentation do not exist. Identifying areas of overlap and differences between parental and provider responses can provide clearer understanding of the informational disconnect between patients and providers during discussion of a Turner syndrome diagnosis. Identifying areas of overlap or areas for improvement may provide a guide for what providers should focus on when delivering either a prenatal or a postnatal diagnosis to better care for their patients.

**1.11 PURPOSE**

The aim of this study was to identify the information related to Turner syndrome that is considered most essential to an initial prenatal or postnatal diagnosis by parents and providers. The purpose of this study was to also identify discrepancies between the informational preferences of parents and providers. Because a consensus for what to discuss when delivering an initial Turner syndrome diagnosis does not currently exist, the results of this study may provide evidence and insight for developing clinical practice guidelines related to an initial Turner syndrome diagnosis discussion.

# CHAPTER 2

# ESSENTIAL INFORMATIONAL NEEDS OF PARENTS RECEIVING A TURNER SYNDROME DIAGNOSIS: PARENT AND GENETIC COUNSELOR PERSPECTIVES[1]

[1] Wasson, J., Edwards, J., Seal, P., Meredith, S. To be submitted to Journal of Genetic Counseling

## 2.1 ABSTRACT

Turner syndrome affects approximately 1 in 2,500 live female births, and etiology includes the partial or complete loss of the second X-chromosome. Prognosis varies depending on whether a diagnosis is made prenatally or postnatally. Current recommendations state that genetic counselors should be involved in the diagnosis; however, guidelines for what information to include during an initial diagnosis do not exist. The aim of this study was to identify which informational items related to Turner syndrome are considered most essential by parents and genetic counselors.

A survey including 100 informational items related to Turner syndrome was sent to genetic counselors and parents whose children were diagnosed with Turner syndrome. Participants ranked the importance of each informational item for an initial diagnosis. Information that both genetic counselor and parent groups ranked within the top 30 items was deemed "essential" for an initial discussion of a Turner syndrome diagnosis.

Of the top 30-ranked items for each group, 13 items were deemed essential for an overall diagnosis, 21 informational items were deemed essential for a prenatal diagnosis, and 20 informational items were deemed essential for a postnatal diagnosis. There were also statistically significant differences in the item ratings between each survey group, which included prenatal genetic counselors, postnatal genetic counselors, and parents who have received either a prenatal or postnatal diagnosis for their child. Findings of our study may provide a guide for what providers should focus on when presenting an initial Turner syndrome diagnosis to parents in the future.

## 2.2 INTRODUCTION

Turner syndrome is a chromosomal aneuploidy that affects approximately 1 in 2,500 live female births and approximately 1% to 2% of all conceptions (Larizza et al., 2011). Previous research has shown that Turner syndrome is the most common chromosomal abnormality in females and that monosomy X is the third most common chromosomal aneuploidy in spontaneous abortions during the first trimester (Elsheikh et al., 2002; Jia et al., 2015). Turner syndrome is not significantly correlated with maternal age (Kim et al., 2013). Major differences in prenatal and postnatal features and survivability of Turner syndrome are recognized with a suggested fetal demise rate of 99% from the first trimester to term (Surerus et al., 2003). Individuals who survive to birth have a reduced life expectancy of up to 13 years, but this life expectancy can be increased with appropriate intervention (Elsheikh et al., 2002).

Turner syndrome results from partial or complete loss of the second X-chromosome in phenotypic females (Shankar & Backeljauw, 2018). The most common karyotypes found in Turner syndrome live births include: monosomy X [45,X (40%-50%)], Turner syndrome mosaicism with normal karyotype [45,X/46,XX (15%-25%)], mixed gonadal dysgenesis [45,X/46,XY (10%-12%)], and isochromosome Xq or isodicentric Xp [46,X i(xq); 46,X,idic(Xp) (10%)] (Gravholt et al., 2017).

Turner syndrome may be diagnosed prenatally or postnatally. During pregnancy, several screening procedures can indicate a higher risk for Turner syndrome. Ultrasonographic findings including increased nuchal translucency, cystic hygroma, coarctation of the aorta and/or left-sided heart defects, brachycephaly, renal dysplasia (including horseshoe kidney), polyhydramnios, oligohydramnios, and growth restriction

indicate a higher likelihood of Turner syndrome (Nicolaides et al., 1992; Redel & Backeljauw, 2018). Non-invasive prenatal testing (NIPT) may also be used to assess risk for Turner syndrome, with a positive predictive value of 12.5% (Lu et al., 2021). Chorionic villus sampling (CVS) or amniocentesis may be performed to obtain karyotype or chromosomal microarray data and confirm a diagnosis of Turner syndrome (ACOG, 2016; Lu et al., 2021) . Regardless of whether the condition has been diagnosed prenatally, chromosome analysis should be repeated after birth in every patient to obtain the most accurate diagnosis (Gravholt et al., 2017).

After birth, the clinical presentation of Turner syndrome varies throughout life. Common features of Turner syndrome include short stature, gonadal failure, cardiac anomalies, urogenital anomalies, lymphedema, micrognathia, cubitus valgus, low posterior hairline, short neck, high arched palate, short fourth metacarpal, multiple naevi, webbed neck, lymphedema of hands and feet, nail dysplasia, scoliosis, and Madelung deformity (Elsheikh et al., 2002; Redel & Backeljauw, 2018). Although individuals with Turner syndrome typically have normal intelligence, they may have psychosocial or neurocognitive issues (Shankar & Backeljauw, 2018). Additionally, studies have shown that women and girls with Turner syndrome also have higher rates of employment and college education and lower rates of marriage than the general US female population (Gould et al., 2013).

Often, girls receive a delayed diagnosis of Turner syndrome (Morgan, 2007). Late diagnosis of Turner can lead to missed potential for intervention which can improve health and quality of life (Gravholt et al., 2017). Delayed diagnosis of Turner syndrome

can lead to a lost opportunity for treatment and management of health and psychological function (Gravholt et al., 2017; Lee & Conway, 2014).

Parent attitudes regarding their diagnostic experiences are variable, and the level of parental satisfaction at the time of diagnosis has been shown to coincide with physician knowledge and support. Parental satisfaction when receiving an initial diagnosis has been associated with the provider's ability to provide relevant, understandable, and comprehensive information (Starke & Moller, 2002; Starke et al., 2002). It is recommended that providers include accurate information, both positive and negative, when delivering a diagnosis of sex chromosomal abnormalities. However, the information parents are given when they are first informed about a sex chromosome anomaly is often a matter of chance—providers who participated in one study said that they often delivered diagnoses about conditions that they knew little about themselves. There is also little consensus on the content of information that parents need about these conditions when receiving an initial diagnosis (Hall et al., 2003).

Current recommendations state that for sex chromosome abnormalities, genetic counseling by a geneticist or genetic counselor should be provided before and after any diagnostic procedure (Gravholt et al., 2017). Physicians and genetic counselors involved in diagnostic counseling need to be fully informed about the prognosis, complications, quality of life, and recent advances in management for Turner syndrome (Saenger et al., 2001).

Balanced information for parents emphasizing both the strengths and challenges of the condition is important when delivering a Turner syndrome diagnosis. Although recommendations for medical interventions following a diagnosis are established, a

strong consensus on the content of information that parents need to be presented with and what aspects they feel are most important has not been delineated, and guidelines for what specifically to include in this presentation do not exist.

The aim of this study was to identify areas of overlap and differences between parental and provider perspectives on what information is most important to include in a Turner syndrome diagnosis. This information may provide clearer understanding of the informational disconnect between patients and providers during discussion of a Turner syndrome diagnosis. Identifying areas of overlap or areas for improvement may also provide a guide for what providers should focus on when delivering a either a prenatal or a postnatal diagnosis to better care for their patients.

## 2.3 METHODS

### *2.3.1 DESIGN AND PARTICIPANTS*

The institutional review board at the University of South Carolina approved this study in July of 2020. All methods and surveys were adapted from a previous study assessing the informational needs of parents receiving a diagnosis of Down syndrome and a more recent, revised study which re-assessed these needs (Sheets et al., 2011; Wilkes et al., 2020). Informational items, diagnostic information, and free-response questions were adapted to a Turner syndrome diagnosis. For this study, inclusion criteria included:

- parents who had a diagnosis of Turner syndrome for their child or pregnancy,
- genetic counselors with a Master of Science degree in genetic counseling, and
- physicians with a Doctor or Medicine or Doctor of Osteopathic Medicine degree who had delivered a positive screen result or diagnosis of Turner syndrome.

Genetic counselors were accessed through the National Society of Genetic Counselors listserv and received an invitation with a link to a Qualtrics survey through email. Physicians were accessed through their affiliation with multiple professional organizations, through which an invitation with a link to the same survey was shared. Turner syndrome-related advocacy organizations also distributed the survey to genetic counselor and physician affiliates. The link to the genetic counselor/physician survey contained an invitational letter for participants (Appendix A).

Parents whose children had received a diagnosis of Turner syndrome were identified through national and online Turner syndrome organizations and support groups as well as social media groups. An invitation containing a link to a Qualtrics survey was included in emails distributed to parents who were affiliated with several national Turner syndrome organizations, and a link was also posted in multiple Turner syndrome social media groups by administrators. The link to the survey also included an invitational letter for parent participants (Appendix B) (Sheets et al., 2011; Wilkes et al., 2020).

Invitations were also shared with others by individuals who were initially contacted, allowing the survey to reach people who were outside of the original scope. Consent was obtained through participants completing the survey, as completion of the survey was voluntary (Sheets et al., 2011; Wilkes et al., 2020).

### *2.3.2 INSTRUMENTATION*

Parents who received a prenatal diagnosis for their child (prenatal parents) or postnatal diagnosis for their child (postnatal parents) were given a four-section Qualtrics survey. The first section consisted of questions related to their child's diagnosis. The second section included information about various aspects of Turner syndrome and asked

parents to assign a level of importance (*essential* (3), *important but not essential* (2), *not too important* (1), *unsure* (0)) to each feature. The third section included free response questions where parents answered open-ended questions about their experience receiving a diagnosis of Turner syndrome for their child. The fourth section consisted of questions about the demographic information of the parent participants (Sheets et al., 2011; Wilkes et al., 2020).

Genetic counselors and physicians were also given a four-section Qualtrics survey. The first and second sections consisted of informational item ratings about a Turner syndrome diagnosis that were identical to the second section of the parent survey. The first section directed providers to rate the importance of items for a prenatal diagnosis, and the second section directed providers to rate the importance of items for a postnatal diagnosis. Genetic counselors and physicians could either complete the prenatal section (prenatal genetic counselors and prenatal physicians), postnatal section (postnatal genetic counselors and postnatal physicians), or both sections., The third section included a free response section where providers answered open-ended questions about a Turner syndrome diagnosis (Sheets et al., 2011; Wilkes et al., 2020). The fourth section consisted of questions about the demographic information of the provider participants, with questions differentiating depending on whether the participant indicated they were a genetic counselor or physician.

Completing the questionnaire allowed participants to enter into a raffle for a $25.00 Visa gift card. One gift card was available for a raffle in each group (parent, genetic counselor, and physician). The contact information for the raffle was submitted on a questionnaire separate from the questionnaire designed for study content. Thus, no

identifying information was collected in association with survey responses and responses remained anonymous.

### *2.3.3 DATA ANALYSIS*

No physician responses were received. Therefore, four groups were identified based on survey responses: prenatal parents, postnatal parents, prenatal genetic counselors, and postnatal genetic counselors. To analyze quantitative results, data was transferred from Qualtrics to Statistical Package for Social Sciences (SPSS). The average rating for each informational item within the survey was calculated using a Likert scale format (*essential* = 3, *important but not essential* = 2, *not too important* = 1, *unsure* = 0) and assembled into rank order to determine which items were considered most important to least important. Comparative statistical analysis was performed to ascertain the differences between prenatal and postnatal parent groups, prenatal and postnatal genetic counselor groups, prenatal parent and prenatal genetic counselor groups, and postnatal parent and postnatal genetic counselor groups. Qualitative data from free response questions were used to add further dimension to the investigation in defining balanced information about Turner syndrome (Sheets et al., 2011; Wilkes et al., 2020).

## 2.4 RESULTS

### *2.4.1 GENETIC COUNSELOR PARTICIPANTS*

A total of 82 genetic counselors opened the provider survey, and 45 completed a significant portion. Genetic counselors were considered to have completed a significant portion of the survey if they completed at least one of the informational item ranking sections and demographic information section. Some participants completed the section related to a prenatal diagnosis of Turner syndrome, and some completed the section

related to a postnatal diagnosis. Because certain items in the survey were optional, not all participants completed every item. Some genetic counselor participants completed both the prenatal and postnatal sections of the survey. Genetic counselor responses to the free response section of the survey can be found in Appendix C.

The majority of participants were female and white (Table 2.1). Most genetic counselor participants either held no religious beliefs or were non-practicing. The majority of participants had one child. Most participants worked in either an academic/university-based setting or in a hospital-based setting. Several genetic counselors worked in a prenatal or pediatric specialty, and some genetic counselors worked in multiple settings. The majority of genetic counselor participants had practiced for one year, two years, or three years in their current specialty. Most participants had delivered a positive screening result and diagnosis of Turner syndrome while some had delivered a diagnosis only. Most genetic counselor participants had some contact with individuals with Turner syndrome or parents of individuals with Turner syndrome in their training or in their current area of practice. Most have not had contact with individuals with Turner syndrome or parents of individuals with Turner syndrome outside of a professional context.

**Table 2.1 Genetic counselor demographic information**

| Demographic | Responses | Total (n) | Percent |
|---|---|---|---|
| Sex (n=45) | Female | 44 | 97.78 |
| | Male | 1 | 2.22 |
| Ethnicity/Race (n=45) | White | 40 | 88.89 |
| | Black/African American/African | 1 | 2.22 |
| | Spanish/Hispanic/Latino | 1 | 2.22 |
| | Asian Indian | 1 | 2.22 |
| | Chinese | 1 | 2.22 |
| | Other | 1 | 2.22 |

| | | | |
|---|---|---|---|
| Level of Religious Activity (n=45) | Very active | 6 | 13.33 |
| | Occasionally active | 10 | 22.22 |
| | Non-practicing | 10 | 22.22 |
| | No religion | 19 | 42.22 |
| Total Number of Children (n=45) | 1 child | 29 | 64.44 |
| | 2 children | 6 | 13.33 |
| | 3 children | 6 | 13.33 |
| | 4 children | 2 | 4.44 |
| | 5 children | 2 | 4.44 |
| Current State of Residence (n=44) | AL | 2 | 4.55 |
| | AZ | 1 | 2.27 |
| | CA | 1 | 2.27 |
| | CO | 2 | 4.55 |
| | FL | 2 | 4.55 |
| | GA | 1 | 2.27 |
| | IL | 2 | 4.55 |
| | KY | 2 | 4.55 |
| | MA | 2 | 4.55 |
| | MD | 3 | 6.82 |
| | MI | 2 | 4.55 |
| | MS | 2 | 4.55 |
| | NC | 4 | 9.09 |
| | NM | 1 | 2.27 |
| | NY | 2 | 4.55 |
| | OH | 1 | 2.27 |
| | PA | 1 | 2.27 |
| | SC | 1 | 2.27 |
| | TN | 1 | 2.27 |
| | TX | 1 | 2.27 |
| | UT | 1 | 2.27 |
| | VA | 4 | 9.09 |
| | WA | 2 | 4.55 |
| | WI | 3 | 6.82 |
| Current work setting (n=45) | Academic/university based | 15 | 33.33 |
| | Hospital-based | 22 | 48.89 |
| | Private practice | 2 | 4.44 |
| | Industry | 1 | 2.22 |
| | Non-profit research | 1 | 2.22 |
| | Health maintenance organization | 1 | 2.22 |
| | Military facility | 1 | 2.22 |
| | Nonprofit organization | 1 | 2.22 |
| | Physician group | 1 | 2.22 |
| Current specialty area of practice (participants could select more than one answer) | Prenatal | 24 | N/A |
| | General pediatrics | 18 | N/A |
| | Pediatric subspecialty | 9 | N/A |

| | | | |
|---|---|---|---|
| | Cancer | 9 | N/A |
| | Adult genetics | 9 | N/A |
| | Laboratory | 3 | N/A |
| | Research | 1 | N/A |
| Years of practice in current specialty area (n=45) | 1 year | 10 | 22.22 |
| | 2 years | 20 | 44.44 |
| | 3 years | 11 | 24.44 |
| | 4 years | 2 | 4.44 |
| | 5 years | 1 | 2.22 |
| | 8 years | 1 | 2.22 |
| Previous areas of practice (participants could select more than one answer) | Prenatal | 10 | N/A |
| | General pediatrics | 5 | N/A |
| | Pediatric subspecialty | 3 | N/A |
| | Cancer | 3 | N/A |
| | Adult genetics | 2 | N/A |
| | Laboratory | 1 | N/A |
| | Research | 2 | N/A |
| | Lysosomal storage disorders | 1 | N/A |
| | No previous specialty area | 28 | N/A |
| Delivered a positive screening result or diagnosis of Turner syndrome (n=44) | Yes, positive screening result only | 1 | 2.27 |
| | Yes, diagnosis only | 11 | 25.00 |
| | Both | 28 | 63.64 |
| | Neither | 4 | 9.09 |
| Extent of contact with individuals with Turner syndrome or parents of individuals with Turner syndrome in training (n=44) | No contact | 11 | 25.00 |
| | Some contact | 27 | 61.36 |
| | Plenty of contact | 6 | 13.64 |
| Extent of contact with individuals with Turner syndrome or parents of individuals with Turner syndrome in current area of practice (n=44) | No contact | 8 | 18.18 |
| | Some contact | 29 | 65.91 |
| | Plenty of contact | 7 | 15.91 |
| Extent of contact with individuals with Turner syndrome or parents of individuals with Turner syndrome outside of a professional context (n=44) | No other contact | 33 | 75.00 |
| | Social contact only | 11 | 25.00 |

### *2.4.2 PARENT PARTICIPANTS*

A total of 61 parents opened the parent survey, and 44 completed a significant portion. Parents were considered to have completed a significant portion of the survey if they completed the informational item ranking section even if they did not complete the

demographic information section. Parent responses to the free response section of the survey can be found in Appendix D.

The majority of parents were female and white (Table 2.2). Most parents considered their level of religious activity to be either very active or occasionally active. The majority of parents were educated, either holding an associate degree, bachelor's degree, master's degree, or doctorate degree. Most parents were married and had either one child, two children, or three children. The majority of parent participants had a combined household income of less than $75,000. While the majority of parents were not a member of a Turner syndrome advocacy group, most were a member of at least one Turner syndrome online support group, social media group, or online forum.

**Table 2.2 Parent demographic information**

| Demographic | Responses | Total (n) | Percent |
|---|---|---|---|
| Sex (n=32) | Female | 31 | 96.88 |
| | Male | 1 | 3.13 |
| Ethnicity/Race (n=32) | White | 28 | 87.50 |
| | Spanish/Hispanic/Latino | 1 | 3.13 |
| | Asian | 1 | 3.13 |
| | French/Ashkenazi Jewish | 1 | 3.13 |
| | Korean and White | 1 | 3.13 |
| Level of Religious Activity (n=31) | Very active | 9 | 29.03 |
| | Occasionally active | 12 | 38.71 |
| | Non-practicing | 6 | 19.35 |
| | No religion | 4 | 12.90 |
| Highest Level of Education (n=31) | Middle school | 1 | 3.23 |
| | High school graduate | 5 | 16.13 |
| | Some college | 7 | 22.58 |
| | Associate degree | 4 | 12.90 |
| | Bachelor's degree | 7 | 22.58 |
| | Master's degree | 5 | 16.13 |
| | Doctorate degree | 2 | 6.45 |
| Marital Status (n=31) | Married | 24 | 77.42 |
| | Widowed | 1 | 3.23 |
| | Divorced | 1 | 3.23 |
| | Separated | 1 | 3.23 |
| | Never married | 4 | 12.90 |

| | | | |
|---|---|---|---|
| Total Number of Children (n=32) | 1 child | 8 | 25.00 |
| | 2 children | 14 | 43.75 |
| | 3 children | 8 | 25.00 |
| | 4 children | 1 | 3.13 |
| | 8 children | 1 | 3.13 |
| Current State of Residence (n=30) | AL | 1 | 3.33 |
| | CA | 1 | 3.33 |
| | CT | 1 | 3.33 |
| | FL | 4 | 13.33 |
| | KS | 1 | 3.33 |
| | LA | 1 | 3.33 |
| | MA | 1 | 3.33 |
| | MD | 1 | 3.33 |
| | ME | 1 | 3.33 |
| | MI | 1 | 3.33 |
| | MS | 1 | 3.33 |
| | NE | 1 | 3.33 |
| | NH | 1 | 3.33 |
| | NJ | 1 | 3.33 |
| | NY | 1 | 3.33 |
| | OH | 2 | 6.67 |
| | ON | 1 | 3.33 |
| | OR | 1 | 3.33 |
| | SC | 2 | 6.67 |
| | TX | 3 | 10.00 |
| | VA | 1 | 3.33 |
| | WI | 1 | 3.33 |
| | WV | 1 | 3.33 |
| Combined Household Income (n=30) | ≤ $24,999 | 3 | 10.00 |
| | $25,000 - $49,999 | 7 | 23.33 |
| | $50,000 - $74,999 | 6 | 20.00 |
| | $75,000 - $99,999 | 1 | 3.33 |
| | $100,000 - $124,999 | 3 | 10.00 |
| | $125,000 - $149,999 | 4 | 13.33 |
| | ≥ $150,000 | 6 | 20.00 |

### *2.4.3 INFORMATION REGARDING THE DIAGNOSIS*

The majority of parent participants received a diagnosis for their child within the last decade (2010-2020) (Table 2.3). Most parents received a diagnosis postnatally. For parents who received a postnatal diagnosis, the majority received a diagnosis for their child at five years or younger with a maximum age of 16 years. The average age of

postnatal diagnosis was 5.63 years. For parent participants who received a prenatal diagnosis, most pregnancies survived to birth with the majority of prenatal diagnoses being made between 12- and 16-weeks' gestation. This was the first pregnancy for fewer than half of the parent participants.

The majority of diagnoses were either made by an endocrinologist or obstetrician/gynecologist. Genetic counselors delivered fewer than half of the diagnoses; however, most parents did see a genetic counselor at some point during the diagnostic process. The majority of parents who did not see a genetic counselor during their diagnostic process did not see one because they did not know genetic counseling was available.

When asked how well informed they felt about the condition when given an initial diagnosis for their child, some parents felt very well-informed, most felt somewhat informed, and some felt they were not informed. Some parents were extremely satisfied with their experience receiving a diagnosis, some were somewhat satisfied, some were neutral, some were somewhat dissatisfied, and some were extremely dissatisfied.

**Table 2.3 Information regarding the diagnosis**

| Demographic | Responses | Total (n) | Percent |
|---|---|---|---|
| Year of diagnosis (n=48) | 1985 | 1 | 2.08 |
| | 1994 | 1 | 2.08 |
| | 1995 | 1 | 2.08 |
| | 1997 | 1 | 2.08 |
| | 2003 | 1 | 2.08 |
| | 2005 | 2 | 4.17 |
| | 2006 | 1 | 2.08 |
| | 2007 | 1 | 2.08 |
| | 2009 | 3 | 6.25 |
| | 2011 | 5 | 10.42 |
| | 2012 | 2 | 4.17 |
| | 2013 | 3 | 6.25 |
| | 2014 | 2 | 4.17 |

| | | | |
|---|---|---|---|
| | 2015 | 3 | 6.25 |
| | 2016 | 1 | 2.08 |
| | 2017 | 4 | 8.33 |
| | 2018 | 3 | 6.25 |
| | 2019 | 5 | 10.42 |
| | 2020 | 8 | 16.67 |
| Prenatal or postnatal diagnosis (n=58) | Prenatal diagnosis | 19 | 32.76 |
| | Postnatal diagnosis | 39 | 67.24 |
| Gestational age when the diagnosis was made (prenatal diagnosis) (n=15) | 11 weeks | 1 | 6.67 |
| | 12 weeks | 4 | 26.67 |
| | 14 weeks | 2 | 13.33 |
| | 15 weeks | 1 | 6.67 |
| | 16 weeks | 2 | 13.33 |
| | 19 weeks | 1 | 6.67 |
| | 20 weeks | 1 | 6.67 |
| | 21 weeks | 1 | 6.67 |
| | 28 weeks | 1 | 6.67 |
| | 33 weeks | 1 | 6.67 |
| Did baby survive to birth (prenatal diagnosis) (n=16) | Yes | 13 | 81.25 |
| | No | 3 | 18.75 |
| Age when diagnosis was made (postnatal diagnosis) (n=27) | < 1 year | 2 | 7.41 |
| | 1-2 years | 6 | 22.22 |
| | 2-3 years | 2 | 7.41 |
| | 3-4 years | 2 | 7.41 |
| | 4-5 years | 2 | 7.41 |
| | 6-7 years | 2 | 7.41 |
| | 7-8 years | 3 | 11.11 |
| | 8-9 years | 1 | 3.70 |
| | 10-11 years | 3 | 11.11 |
| | 11-12 years | 1 | 3.70 |
| | 14-15 years | 2 | 7.41 |
| | 15-16 years | 1 | 3.70 |
| Who made the Turner syndrome diagnosis (n=47) | Obstetrician/gynecologist | 9 | 19.15 |
| | Maternal fetal medicine sub-specialist | 6 | 12.77 |
| | Pediatrician | 4 | 8.51 |
| | Family physician | 1 | 2.13 |
| | Endocrinologist | 17 | 36.17 |
| | Pediatric geneticist | 2 | 4.26 |
| | Prenatal genetic counselor | 5 | 10.64 |
| | Pediatric genetic counselor | 3 | 6.38 |
| Karyotype of child (n=57) | Monosomy X (45,X) | 22 | 38.60 |
| | Monosomy X mosaicism (45,X/46,XX) | 17 | 29.82 |
| | Other | 11 | 19.30 |

| | | | |
|---|---|---|---|
| | Unknown | 7 | 12.28 |
| What number pregnancy (n=57) | 1st pregnancy | 26 | 45.61 |
| | 2nd pregnancy | 15 | 26.32 |
| | 3rd pregnancy | 10 | 17.54 |
| | 4th pregnancy | 5 | 8.77 |
| | Unsure | 1 | 1.75 |
| What number child (n=56) | 1st child | 28 | 50.00 |
| | 2nd child | 20 | 35.71 |
| | 3rd child | 7 | 12.50 |
| | 4th child | 1 | 1.79 |
| Received prenatal screening or diagnostic testing during pregnancy? (participants could select more than one answer) | None | 14 | N/A |
| | First trimester screening | 18 | N/A |
| | First trimester nuchal translucency measurement | 9 | N/A |
| | Noninvasive prenatal screening | 14 | N/A |
| | Second trimester multiple marker blood screen | 7 | N/A |
| | Second trimester blood screen – AFP only | 1 | N/A |
| | Chorionic villus sampling | 1 | N/A |
| | Amniocentesis | 8 | N/A |
| | Level II ultrasound | 17 | N/A |
| | Unsure, but had some form of prenatal screening | 13 | N/A |
| How well informed about Turner syndrome when given diagnosis (n=57) | Very well-informed | 13 | 22.81 |
| | Somewhat informed | 25 | 43.86 |
| | Not informed | 19 | 33.33 |
| Saw a genetic counselor during diagnostic process (n=57) | No | 21 | 36.84 |
| | Yes (prenatal) | 8 | 14.04 |
| | Yes (postnatal) | 20 | 35.09 |
| | Yes (prenatal and postnatal) | 8 | 14.04 |
| Reason for not seeing a genetic counselor for the diagnosis (if answered no for seeing a genetic counselor) (n=19) | Did not know about genetic counseling | 11 | 57.89 |
| | Genetic counseling was not available in my area | 2 | 10.53 |
| | Did not want to see a genetic counselor | 2 | 10.53 |
| | Was not able to get an appointment due to COVID | 1 | 5.26 |
| | Physician was familiar with Turner syndrome | 1 | 5.26 |
| | Recently received the initial diagnosis and have not scheduled | 1 | 5.26 |

| | | | |
|---|---|---|---|
| | Obstacles related to the genetic counseling appointment | 1 | 5.26 |
| Overall satisfaction with experience of receiving Turner syndrome diagnosis (n=52) | Extremely satisfied | 9 | 17.31 |
| | Somewhat satisfied | 13 | 25.00 |
| | Neutral | 13 | 25.00 |
| | Somewhat dissatisfied | 9 | 17.31 |
| | Extremely dissatisfied | 8 | 15.38 |
| Member of Turner syndrome advocacy group (n=52) | No | 38 | 73.08 |
| | Yes | 14 | 26.92 |
| Advocacy organization name (if answered yes to membership) (n=14) | Turner Syndrome Society of the United States (TSSUS) | 8 | 57.14 |
| | Turner Syndrome Global Alliance | 2 | 14.29 |
| | Turner Syndrome Society of Canada | 1 | 7.14 |
| | Turner Syndrome Foundation | 3 | 21.43 |
| Member of online support group, social media group, or online forum (n=53) | No | 17 | 32.08 |
| | Yes | 36 | 67.92 |
| Online support group, social media, or online forum name (if answered yes to membership) (n=35) | Facebook groups (other) | 10 | 28.57 |
| | Facebook Canadian Turner Syndrome group | 1 | 2.86 |
| | Facebook Turner Syndrome Connections group | 1 | 2.86 |
| | Facebook Turner Syndrome Parent Support group | 1 | 2.86 |
| | Facebook TSSUS Group | 4 | 11.43 |
| | Facebook Oklahoma Turner Syndrome group | 1 | 2.86 |
| | Facebook Turner Syndrome Foundation group | 2 | 5.71 |
| | Facebook Turner Syndrome Mommies and Parents group | 3 | 8.57 |
| | Facebook MAGIC group | 1 | 2.86 |
| | Facebook Turner Syndrome Parents group | 4 | 11.43 |
| | Facebook Florida Turner Syndrome Families group | 1 | 2.86 |
| | Facebook Arizona Turner Syndrome Support group | 1 | 2.86 |
| | Facebook Turner Syndrome Support Chat group | 1 | 2.86 |
| | Online groups (unspecified) | 1 | 2.86 |
| | Yahoo Parents | 1 | 2.86 |
| | Other | 2 | 5.71 |

### *2.4.4 ESSENTIAL INFORMATION FOR DIAGNOSIS*

Informational items were placed in rank order based on average rating to determine which items were deemed most essential for an initial discussion of Turner syndrome. Informational items were presented in rank order by average rating for all four groups of participants: prenatal parents, prenatal genetic counselors, postnatal parents, and postnatal genetic counselors (Appendix E). A higher average rating indicates a more essential item for the initial discussion of Turner syndrome.

Informational items were deemed essential for an overall Turner syndrome diagnosis if they were included in the top 30-ranked items for every group—these groups included prenatal and postnatal parents as well as prenatal and postnatal genetic counselors. Overall, 13 overall essential informational items for a Turner syndrome diagnosis were identified (Table 2.4).

Items were deemed essential for a prenatal diagnosis specifically if they were included in the top 30-ranked items for prenatal genetic counselors and prenatal parents. Twenty-one items were found to be essential for a prenatal diagnosis of Turner syndrome; eight of these items were found to be essential for a prenatal diagnosis only. The eight informational items that were deemed essential for a prenatal diagnosis only in addition to the 13 overall essential items are included in Table 2.4.

Likewise, items were deemed essential for a postnatal diagnosis specifically if they were included in the top 30-ranked items for postnatal genetic counselors and postnatal parents. Twenty items were found to be essential for a postnatal diagnosis of Turner syndrome; seven of these items were found to be essential for a postnatal

diagnosis only. The seven informational items that were deemed essential for a postnatal diagnosis only in addition to the 13 overall essential items are included in Table 2.4.

**Table 2.4 Essential informational items for a Turner syndrome diagnosis**

| Essential Information for an Overall Diagnosis | Additional Prenatal Essential Information | Additional Postnatal Essential Information |
|---|---|---|
| Heart defects and other cardiovascular issues (25%-50%) | Coarctation of the aorta and/or left-sided cardiac defects | Discuss impact of hormones to increase stature or treatments that can allow girls with Turner syndrome to attain a normal adult height (over five feet) |
| Caused by the partial or complete loss of the second X-chromosome in females | Discuss value of ultrasound in detecting prenatal conditions | Women and girls with Turner syndrome typically have normal intelligence |
| Local Turner syndrome support group(s) | Need an echocardiogram (ultrasound of the heart) during pregnancy | Having friends and meaningful relationships |
| Specialist referral(s) | Cystic hygroma (fluid-filled sack on the back of the fetal neck) | More like other children than different |
| May need an evaluation by a pediatric heart specialist | Screening results can be obtained via noninvasive prenatal screening (NIPS/cell-free DNA) | Reduced fertility |
| Discuss available Turner syndrome clinics | Printed/written material | Early intervention services |
| Women with Turner syndrome usually live long lives | Prenatal diagnosis can only be confirmed by chromosome analysis via amniocentesis or CVS | Reproductive capability of a woman with Turner syndrome |
| May have some health conditions but not all | Online support groups/social media platforms | - |
| National advocacy organizations & websites | - | - |

| | | |
|---|---|---|
| Primary ovarian failure (reduced function of ovaries before age 40) (90%) | - | - |
| Chance to reoccur in future pregnancies | - | - |
| Girls with Turner syndrome need hormones to start or complete puberty | - | - |
| Short stature (95%-100%) | - | - |

Although there was overlap in which items were considered essential by participants, there were statistically significant differences between the ratings of informational items by each group ($p<0.05$). The statistically significant differences in the ratings of each informational item for the various participant groups are presented (Appendix F). The average ratings for each item and group are listed in the table as a visual reference.

Prenatal and postnatal genetic counselor ratings were statistically significantly different for 44 of the 100 total informational items ($p<0.05$). Prenatal genetic counselor and prenatal parent ratings were statistically significantly different for 18 of the 100 total informational items ($p<0.05$). Postnatal genetic counselor and postnatal parent ratings were statistically significantly different for 27 of the 100 informational items ($p<0.05$). Prenatal parent and postnatal parent ratings were statistically significantly different for 29 of the 100 informational items ($p<0.05$). There were 18 informational items with statistically significant differences between groups that were $p<0.0001$, and these items are presented in Table 2.5. The complete list of statistically significant differences between each item and group is presented in Appendix F.

**Table 2.5 Items with <0.0001 statistically significant differences between groups**

| Informational Item | Group | Rating | Group | Rating |
|---|---|---|---|---|
| Increased nuchal translucency in first trimester | Prenatal GC | 2.25 | Postnatal GC | 1.29 |
| Pleural effusion | Prenatal GC | 2.02 | Postnatal GC | 1.26 |
| Pericardial effusion | Prenatal GC | 2.06 | Postnatal GC | 1.26 |
| Cystic hygroma | Prenatal GC | 2.46 | Postnatal GC | 1.32 |
| Coarctation of the aorta and/or left-sided cardiac defects | Prenatal GC | 2.71 | Postnatal GC | 2.11 |
| Need an echocardiogram during pregnancy | Prenatal GC | 3.00 | Postnatal GC | 1.34 |
| Polyhydramnios | Prenatal GC | 1.96 | Postnatal GC | 1.26 |
| Oligohydramnios | Prenatal GC | 1.96 | Postnatal GC | 1.26 |
| Growth restriction | Postnatal GC | 2.12 | Postnatal Parent | 2.59 |
| 99% risk for fetal demise when prenatally diagnosed | Prenatal GC | 2.83 | Postnatal GC | 1.38 |
| | Prenatal GC | 2.83 | Prenatal Parent | 2.00 |
| | Postnatal GC | 1.38 | Postnatal Parent | 2.31 |
| Discuss value of ultrasound in detecting prenatal conditions | Prenatal GC | 2.75 | Postnatal GC | 1.37 |
| | Postnatal GC | 1.37 | Postnatal Parent | 2.45 |
| Glucose intolerance (15%-50%) | Prenatal GC | 1.72 | Postnatal GC | 2.36 |
| Frequent ear infections (60%) | Prenatal GC | 1.72 | Postnatal GC | 2.42 |
| Hypothyroidism (15%-30%) | Prenatal GC | 1.88 | Postnatal GC | 2.48 |
| Financial parental impact – more | Postnatal GC | 1.48 | Postnatal Parent | 2.37 |
| Prenatal diagnosis can only be confirmed by chromosome analysis via amniocentesis or CVS | Prenatal GC | 2.82 | Postnatal GC | 1.52 |
| Screening results can be obtained via noninvasive prenatal screening (NIPT/cell-free DNA) | Prenatal GC | 2.74 | Postnatal GC | 1.58 |
| Pregnancy termination resources | Prenatal GC | 2.43 | Postnatal GC | 1.03 |
| | Prenatal GC | 2.43 | Prenatal Parent | 0.83 |

Of the 100 informational items ranked by participants, there were 29 items for which there was no statistically significant differences between the prenatal genetic counselor and postnatal genetic counselor groups, prenatal genetic counselor and prenatal parent groups, postnatal genetic counselor and postnatal parent groups, and prenatal parent and postnatal parent groups. Items with no statistically significant differences between groups are presented below (Table 2.6).

**Table 2.6 Items with no statistically significant differences between groups**

| Item | Average Rating |
|---|---|
| Heart defects and other cardiovascular issues (25%-50%) | 2.81 |
| May have some health conditions but not all | 2.74 |
| Caused by the partial or complete loss of the second X-chromosome in females | 2.74 |
| Women with Turner syndrome usually live long lives | 2.72 |
| Discuss available Turner syndrome clinics | 2.71 |
| Local Turner syndrome support group(s) | 2.70 |
| Primary ovarian failure (90%) | 2.63 |
| Early intervention services | 2.54 |
| Reproductive capability of a woman with Turner syndrome | 2.53 |
| Printed/written material | 2.53 |
| Fact sheets/brochures | 2.43 |
| Possible chromosomal/genetic causes of Turner syndrome: 45,X (40%-50%), 45,X/46,XX (15%-25%), other (25%-45%) | 2.33 |
| Renal anomalies | 2.30 |
| Counselor or family therapist referral(s) | 2.12 |
| Life expectancy (may be reduced up to 13 years) | 1.97 |
| More likely to be employed than the general US female population | 1.96 |
| Photographs of children with Turner syndrome | 1.90 |
| Impact on parental marriage – strengthens relationship | 1.82 |
| Only discuss prenatal issues that are actually confirmed via ultrasound | 1.78 |
| Impact on parental marriage – strains relationship | 1.71 |
| Brachycephaly | 1.68 |
| Impact on other relationships – supportive and welcoming | 1.65 |
| Impact on grandparents – supportive and welcoming | 1.61 |
| Impact on extended family members – supportive and welcoming | 1.52 |
| Impact on other relationships – lose social circle | 1.52 |
| Pastoral counseling referral(s) | 1.45 |
| Financial parental impact – no difference | 1.45 |
| Alternative/nonconventional therapies | 1.44 |
| Impact on grandparents – limited interaction | 1.44 |

## 2.5 DISCUSSION

### *2.5.1 ESSENTIAL INFORMATION FOR A TURNER SYDNROME DIAGNOSIS*

There is a need for balanced information about Turner syndrome when delivering an initial diagnosis to parents for their children. However, consensus on what information to highlight when providing information about sex chromosome abnormalities to parents does not exist (Hall et al., 2003). Historically, parental satisfaction regarding their

diagnostic experience when given a Turner syndrome diagnosis for their child has coincided with provider knowledge and ability to provide relevant, understandable, and comprehensive information (Starke et al., 2002). The present study identified informational items relevant to discussion regarding a prenatal diagnosis, postnatal diagnosis, and overall (prenatal and postnatal) diagnosis of Turner syndrome. Twenty-one informational items were deemed essential for a prenatal diagnosis, 20 items were deemed essential for a postnatal diagnosis, and 13 items were deemed essential for an overall diagnosis. Items were placed in rank order to determine the information most valuable to prenatal parents, postnatal parents, prenatal genetic counselors, and postnatal genetic counselors as done in previous studies (Sheets et al., 2011; Wilkes et al., 2020).

### *2.5.2 EXPERIENCES WITH DIAGNOSIS*

The majority of parents received a postnatal Turner syndrome diagnosis for their child, with approximately 32% receiving a diagnosis prenatally. Most pregnancies that were diagnosed prenatally survived to birth (81.25%). The percentage of pregnancies that survived to birth included in this study is higher than the 1% survival rate quoted on average for a pregnancy diagnosed with Turner syndrome (Surerus et al., 2003). Parents with living children are more likely to become involved and remain involved with social media groups and advocacy organizations related to Turner syndrome, which is where parent participants were recruited from. This may have created a selection bias toward recruiting parents whose children survived to birth to complete the survey. Postnatally, the average age of diagnosis was 5.63 years, which is 3.17 years younger than the average age of diagnosis identified in a previous study performed two decades prior (Savendahl & Davenport, 2000). Multiple guidelines for earlier diagnosis of Turner

syndrome have been proposed since the year 2000 so that females with suggestive findings may be screened for Turner syndrome earlier in life (Gravholt et al., 2017; Savendahl & Davenport, 2000). Creation of these guidelines may have contributed to the younger average age of diagnosis identified in this study, as these guidelines encourage providers to test for the condition when certain symptoms are recognized. Increased uptake of NIPT since 2011 leading to postnatal karyotype testing may also be a contributing factor to the lower average age of postnatal diagnosis seen in this study.

The majority of parents surveyed in this study felt informed about Turner syndrome when given an initial diagnosis for their child (66.67%). However, fewer parents felt satisfied with their overall diagnostic experience (42.31%). The discrepancy between the level of information provided versus overall parental satisfaction may be explained by the fact that many parents felt informed about the condition but felt they could have benefitted from an earlier diagnosis. One parent explained, "I wasn't disappointed with the information as we were relieved to know what was wrong…We could [have] eliminated significant frustrations if we would [have] known sooner!"

Some parents felt that there was too little information provided about Turner syndrome. When asked about the information they were given and satisfaction about their experience, one parent described that "We were given the diagnosis, but nearly no medical information on it…This gave us a lot of unnecessary and harmful worry and stress." Another parent said that "In hindsight, it [is] literally amazing how far we've come with so little info."

Other parents felt that the information provided at the time of diagnosis was overwhelming, with one parent saying, "We were given a book that outlined all the

possibilities with this syndrome. At this point we were not sure whether any of them or all of them were relevant to our daughter!" Another parent explained that the information was "over my head…The doctor used all of these terms I had never heard."

Many parents who were satisfied with their experience explained that their providers were well informed about Turner syndrome and were honest about the unpredictability of the condition. One parent explained,

> "The most useful information I received was the honest truth. They [didn't] really know how each symptom would present during the lifespan. They instead told me what could happen and what to keep an eye on. They reiterated that some things may never happen. The most common symptoms were discussed. That eased the huge anxiety attack I felt thinking all symptoms would arise immediately upon diagnosis."

Another parent mentioned,

> "When I asked about quality of life, [my nurse practitioner] laughed reassuringly and gave me some examples of women living with [Turner syndrome]. It was comforting to hear that my baby had the potential to live a "normal" life. My [nurse practitioner] handled the news with care and provided support while answering my questions as thoroughly as possible."

Responses from these parents coincide with multiple previous studies which state that pretest and posttest counseling must be provided by providers who are highly familiar with the condition. These providers have the ability to convey realistic expectations to parents. The responses also coincide with suggestions found in previous studies that providers must emphasize that individuals with Turner syndrome can be

healthy, happy, and productive members of society (Loscalzo et al., 2006; Saenger et al., 2001).

### *2.5.3 INTERPRETING ESSENTIAL INFORMATION FOR DIAGNOSIS*

Information in this study was interpreted as "essential" for an overall Turner syndrome diagnosis if the informational item was included in the top 30-ranked items for all four groups (prenatal parents, postnatal parents, prenatal genetic counselors, and postnatal genetic counselors). Additionally, information was interpreted as essential for a prenatal diagnosis if the item was included in the top 30-ranked items for prenatal parents and prenatal genetic counselors. For a postnatal diagnosis, information was interpreted as essential if the item was included in the top 30-ranked items for postnatal parents and postnatal genetic counselors. Items were also organized into rank order of importance for each group based on their average ratings.

Overall, 13 items were deemed essential for both a prenatal and postnatal diagnosis of Turner syndrome. Informational items deemed essential were related to health aspects of Turner syndrome (heart problems, primary ovarian failure, short stature, and hormones for puberty), etiology and recurrence risk, referrals and support, lifespan, and overall prognosis. For a prenatal diagnosis, in addition to the overall essential items, information related to pregnancy-related testing and ultrasound findings was also deemed essential for an initial discussion. There was also a greater emphasis on available support resources. For a postnatal diagnosis, in addition to the overall essential items, there was a greater emphasis on long-term life outcomes of the condition and fertility-related information.

### *2.5.4 DIFFERENCES BETWEEN PRENATAL AND POSTNATAL DIAGNOSIS*

Prenatal genetic counselors tended to rate prenatal findings and genetics concepts related to Turner syndrome higher than postnatal counselors. Postnatal genetic counselors rated findings including post-birth health complications, neurocognitive differences, long-term life outcomes, and relationships higher than prenatal counselors. These results underscore the fact that providers tend to highlight information they feel is relevant to the family's situation at the time. For example, postnatal counselors may not feel it is necessary to discuss information that is relevant to the past, such as prenatal findings or risk for fetal demise. A postnatal genetic counselor explained that "there are a lot of things you don't need to discuss at all now that the prenatal time is over (risk of miscarriage, etc.)."

Additionally, in a postnatal environment, counselors described a stronger focus on management and lifestyle factors. Because specific features of the condition tend to be present in the individual at the time of diagnosis postnatally, specific referrals and management recommendations may be made at that time. One genetic counselor stated that "In the postnatal setting, I think it is appropriate to start to talk about longer term management, and of course start to implement therapies/interventions (which does not apply prenatally)."

Genetic counselors also explained that for a postnatal diagnosis, it is important to focus on the specific features seen in the patient when discussing Turner syndrome, with one counselor explaining that "In the postnatal setting, you have a person in front of you to tailor your counseling to." Another counselor said, "I think postnatally, you need to focus on the features right in front of you…Postnatal discussions are less hypothetical

than prenatal." The information about the condition may not be quite as broad in a postnatal discussion because of the ability to provide counseling over several visits throughout the patient's lifetime.

The information during an initial diagnosis postnatally may also differ depending on the patient's age at diagnosis. One counselor explained that in a postnatal setting, the counselor "often has the option to add more information over multiple visits and years." Another counselor stated that "I have had much different conversations with families depending on if the diagnosis occurs around the time of [the] birth/infant period vs. those diagnosed around 13-15 years due to not starting periods."

In contrast with postnatal diagnosis, several counselors explained that prenatally, the main focus of discussion should be related to ultrasound findings as well as diagnostic and pregnancy management options given the high risk for fetal demise. Prenatal counselors also tended to highlight the "big picture" health aspects of the condition and focused less on details related to the family aspects, neurocognitive aspects, or long-term life outcomes. Providers made statements related to this type of discussion which included commentary such as: "Prenatal [genetic counselors] should discuss more about prenatal testing options (screening vs diagnostic), recurrence, and ultrasound findings.", "there needs to be some kind of expectation management", "it is important to discuss the high incidence of pregnancy loss in cases of prenatal diagnosis of Turner syndrome while also presenting a balanced representation of postnatal outcomes", "Memory-making and grief counseling should be anticipated.", and "prenatal should be focused on…helping the family make the decision to do diagnostic testing, continue vs. terminate…what to expect immediately after delivery."

The details about the condition that correspond with a postnatal diagnosis discussion may not be highlighted prenatally because of the variability of Turner syndrome. While ultrasound findings may guide a prognostic discussion, the patient has not yet presented with specific features. Tasks including facilitating decision-making, informing parents about more general aspects of the condition, and providing support for parents tend to take precedence over other elements such as discussing fertility or specific life outcome-related information. It is also important to note that for a prenatal discussion, determining the level of information the family seeks is critical. One counselor explained,

> "[The amount] of information should be tailored to the patient's needs in the prenatal setting as the patient may be making decisions with regard to continuing or not continuing the pregnancy. I tend to…only offer up information that the patient wants at that time since we do not know what features the child will develop."

Another counselor mentioned,

> "The most important [thing] is to provide the family the information they want - if they want to know all the details then that should be shared but if their only question is for life expectancy then you don't need to explain risk for hypertension."

There were also differences between prenatal and postnatal parent responses. Several parents commented that until their child was born, it was difficult for them to conceptualize what Turner syndrome might entail—even if they were given information about the condition by providers. In addition, printed/written material and online support

groups/social media platforms were considered essential for a prenatal Turner syndrome diagnosis but not a postnatal diagnosis. Prenatal parents may appreciate these resources because they may help parents better understand what life is like with Turner syndrome and what to expect after having an initial conversation with a provider. One parent mentioned,

> "Nothing can replace hearing and seeing real life experiences. I wish that providers had more [Turner syndrome]-specific information to share with families immediately so they could use those resources as they wish to do independent research (controlling what and how much they consume) …I think that would help families with locating high quality, accurate information which will ultimately help them to be a better informed patient (and to know what questions to ask once they are feeling less overwhelmed by the initial diagnosis)."

This data suggests that prenatal genetic counselors should identify resources for parents when delivering a diagnosis that demonstrates lived experiences of individuals diagnosed with Turner syndrome. For example, genetic counselors could recommend "Understanding a Turner Syndrome Diagnosis" which is a resource that has been reviewed by the National Society of Genetic Counselors and recommended by the American College of Medical Genetics (Gregg et al., 2016). This resource provides parents with a balanced, accurate depiction of Turner syndrome and includes reliable resources related to the diagnosis (Meredith, 2016).

Postnatal parents placed greater emphasis on non-medical aspects of Turner syndrome than prenatal parents. Items related to normal intelligence, having friends and meaningful relationships, more like other children than different, and early intervention

services were considered essential for a postnatal diagnosis but not for a prenatal diagnosis. Postnatal parents also placed greater emphasis on medical aspects such as fertility and height than prenatal parents. Several postnatal parents made statements such as, “Every Turners girl is unique like every human is unique.”, “The pediatrician simply described her physical attributes and what medical issues she was exhibiting, along with her karyotype test…It was disheartening.”, “I had no idea how much therapy we’d need for our child, how much different our school experience would be.”, “All parents should also learn more about early intervention”, “It would have been helpful to know that not all children will experience all the symptoms.”, “parents also have to know that this disorder isn’t the worst”, “Each child with [Turner syndrome] will display more or fewer of the [Turner syndrome] characteristics.”, and “They lead good lives.”

According to the genetic counselors surveyed in this study, counseling regarding an initial postnatal diagnosis can be more targeted than for a prenatal diagnosis. For a prenatal diagnosis, information needs to be broader to account for variability between individuals. However, for a postnatal diagnosis, the conversation is more tailored to the child’s specific needs and features. Postnatal parents may appreciate more in-depth information about the non-medical aspects of the condition since many of the health-related conditions might not apply to their situation and might not need to be discussed. This allows for a discussion related to the learning, social, and other medical aspects of the condition rather than the more “pressing” issues related to a prenatal diagnosis such as decision-making and the risk for fetal demise.

### *2.5.5 DIFFERENCES IN PARENT AND GENETIC COUNSELOR PERSPECTIVES*

Differences between prenatal parent and genetic counselor responses were identified, with prenatal parents placing more value on items related to informational resources and referrals as well as relationships. Prenatal genetic counselors tended to rate informational items related to post-birth outcomes and genetics of Turner syndrome more highly than prenatal parents. Parents and genetic counselors both highlighted the importance of providing balanced information about Turner syndrome for an initial prenatal diagnosis. Genetic counselors placed significantly greater emphasis on certain non-medical aspects of the condition including intellectual disability, dyscalculia, typically normal intelligence, inclusion in regular classes, living independently, and being more like other children than different. Multiple genetic counselors explained that education about the typical life expectancy and normal intelligence post-birth is part of what they consider to be a balanced presentation. When asked about information related to a balanced presentation, prenatal genetic counselors made statements such as, “girls who are born with Turner syndrome typically do well…most have normal intelligence and can live happy and fulfilling lives.”, “make sure to include that intelligence is likely normal and that there are fertility options if that is wanted one day.”, and “Intelligence is typically normal, but many girls may have a learning disability or different learning methods.”

Counselors also placed significantly greater emphasis on the fertility aspects of the condition than prenatal parents, the chance to reoccur in future pregnancies, and prenatal diagnostic confirmation via chromosome analysis. In contrast, prenatal parents placed significantly greater emphasis on resources like books and contact with other

families than prenatal genetic counselors. Prenatal genetic counselors are tasked with delivering a broad overview of Turner syndrome when delivering a diagnosis and consider non-medical aspects to be part of this presentation. While parents may benefit from hearing this information related to the relatively normal lives individuals with Turner syndrome lead after birth, they may not prioritize this information over resources and support at the time of diagnosis. Many resources cover the same types of information regarding long-term life outcomes that genetic counselors prioritize—parents may be equally interested in this information but could prioritize resources such as books that they can refer to at a later time. These parents may feel a need to process the initial shock of receiving a diagnosis before considering the long-term outcomes.

The strongest discrepancy between prenatal genetic counselors and prenatal parents related to the risk for fetal demise and discussion of available termination resources for affected pregnancies. Out of the 100 informational items, prenatal genetic counselors rated the 99% risk for fetal demise when prenatally diagnosed as the 6$^{th}$ most important item, while prenatal parents rated it as the 48$^{th}$ most important item. Prenatal genetic counselors rated pregnancy termination resources as the 31$^{st}$ most important item, while prenatal parents rated it as the 100$^{th}$ most important item. Most genetic counselors mentioned the importance of discussing the chance for fetal demise and options for ending a pregnancy during a prenatal diagnosis when discussing a balanced presentation. Genetic counselors made statements such as, “Pregnancies with Turner syndrome have a high risk of miscarriage, often due to cystic hygroma or other fluid accumulation.”, “In a prenatal setting, a balanced presentation offers all options for ending or continuing a pregnancy with a focus on the high [likelihood] of fetal demise.”, “most pregnancies

affected with Turner syndrome end in miscarriage", "Most importantly talk about risk of pregnancy loss.", and "Prenatally, I find it hard to discuss too many details of postnatal medical aspects in pregnancy with high risk of demise."

Parents explained that while they understand the necessity of explaining the risk for fetal demise during pregnancy, they felt like it was emphasized too much after they had decided to continue the pregnancy. One parent explained, "for the pregnancy, there [are] statistics and I understand the need to share them with the parents but there are many girls that survive the pregnancy." Another parent stated,

> "The genetic counselor…focused on the statistics telling me of her rare chances of survival. I felt like I knew these things already and was holding onto hope that she would make it and it was kinda like she just wanted to focus on the fact that our daughter was going to die. I was really offended by her."

An additional parent mentioned,

> "We get the situation we're in, if a parent chooses the fight for their child and not terminate, hope and support go a long way. I knew I could lose her but that wasn't what was healthy for me to focus on until that time came if it was going to happen."

Providers have a responsibility to discuss the risk for fetal demise and pregnancy management options to parents during the prenatal period. However, parents may feel that these aspects are overemphasized by multiple providers even if they already understand the risks and have made the decision to continue an affected pregnancy.

Differences between postnatal parent and postnatal genetic counselors were also identified. Overall, postnatal parents rated all of the items related to prenatal physical

features and health complications higher than postnatal genetic counselors. Parents may have applied these items to a theoretical prenatal diagnosis rather than their personal experience with postnatal diagnosis when completing the survey. Parents may have also rated these items higher because they may feel this information is important to review when receiving a postnatal diagnosis, even if it no longer applies to their child.

Given that postnatal parents placed greater emphasis than postnatal genetic counselors on treatment and management options for Turner syndrome, genetic counselors should consider incorporating this information into an initial discussion of a Turner syndrome diagnosis to better meet parents' informational needs. One parent suggested,

> "After questions have been answered, families should be asked what kind of support would be best for them (Are they [struggling] with acceptance? Perhaps they need to be connected with a family therapist. Do they want to connect with families of [Turner syndrome] girls? A community organization serving [Turner syndrome] families may be able to help arrange that. Are they feeling overwhelmed by the future medical appointments/screenings/etc.? They might benefit from joining a local support group for families with children who have special needs and could share advice for navigating the health system) …There can't be a 'one size fits all' approach since the needs will be so varied, but if the physician is well-informed about [Turner syndrome] it will certainly help ease the family's anxiety."

Although this suggestion is geared toward physicians, other providers might ask this question to parents to better understand what types of supports parents hope to benefit from and what needs are still unmet after discussing an initial diagnosis.

Postnatal parents also rated several items related to the family and relationships of individuals with Turner syndrome significantly higher than postnatal genetic counselors, including the impact of the diagnosis on other family members, time commitment of parents, and a greater financial impact. This discrepancy demonstrates a possible need for genetic counselors to include this type of information in a postnatal diagnosis discussion of Turner syndrome. Postnatal parents also rated certain strengths of individuals with Turner syndrome significantly higher than genetic counselors. Parents placed greater emphasis on the fact that women with Turner syndrome are more like to earn a baccalaureate degree or higher than the general US female population and tend to excel at verbal skills. Postnatal genetic counselors placed greater emphasis on certain challenges that individuals with Turner syndrome face, including an increased chance for intellectual disability, dyscalculia, and attention deficit/hyperactivity disorder. However, postnatal genetic counselors also placed a greater emphasis than postnatal parents on the fact that women and girls with Turner syndrome typically have normal intelligence. These results suggest that an initial discussion of a Turner syndrome diagnosis may be enhanced if genetic counselors mention certain non-medical strengths of individuals with the condition in conjunction with certain challenges they may face.

### *2.5.6 LIMITATIONS AND FURTHER INVESTIGATION*

The findings from this study have certain limitations. Parents and providers who participated were recruited from professional organizations, social media groups, and

national advocacy organizations and participated voluntarily, creating bias in responses. Physicians did not participate, and other types of medical providers were not recruited for the study. Future studies could explore the perspectives of these individuals.

The study also included low levels of diversity within each participant group. Within the genetic counselor group, 97.78% of participants were female and 88.89% were white. Within the parent group, 96.88% of participants were female, 87.50% were white, 80.64% attended college, and 77.42% were married. This lack of diversity could have created certain response biases and therefore may not appropriately represent the perspectives of genetic counselors and parents overall. A replicative study that includes participants from more diverse backgrounds would provide a more accurate representation of the general U.S. population.

As this is a retrospective study, recall bias is present for parent participants. Though many parent respondents received their child's diagnosis within the last decade, parental needs may have changed over time. Parental responses may also be tailored to their child's and family's specific needs; therefore, results of this study may not be generalizable to the overall patient population.

Another limitation of this study related to genetic counselor responses—information that is most pertinent for an initial diagnosis may depend on the age of the patient at the time a diagnosis is made. There was not a specific age of diagnosis identified for the provider survey; therefore, responses reflect a more general approach to initial diagnosis. Some information may not be pertinent to an initial diagnosis depending on the patient's age at the time. Future studies analyzing which information is essential for a diagnosis at specific patient ages may ameliorate this discrepancy.

Within the survey, some informational items overlapped with one another. Within the "postnatal physical features and health complications of Turner syndrome" section of each survey, "hypertension (50%)" and "high blood pressure (50%)" were both listed. Within the "long-term life outcomes for individuals with Turner syndrome" section, "special education services" was listed as an informational item—within the "informational resources and referrals for individuals with Turner syndrome and their families" section, "special education supports and services" was also listed as an informational item. These overlapping informational items were not rated statistically significantly differently by any groups, but it is important to note that these overlapping items were included in the survey.

The informational items within the surveys were not exhaustive. There may be other information that is pertinent to an initial Turner syndrome diagnosis that was not included in this study. Future studies could measure the level of importance of other informational items related to Turner syndrome for a more complete assessment of the essential information for diagnosis.

# CHAPTER 3: CONCLUSIONS

The present study identified the information that parents and genetic counselors value most when receiving or delivering an initial Turner syndrome diagnosis either prenatally or postnatally. Thirteen items essential to an overall Turner syndrome diagnosis were identified as well as 21 items essential for prenatal diagnosis and 20 items essential for a postnatal diagnosis. Informational items deemed essential for an overall diagnosis related to specific health aspects of Turner syndrome, etiology and recurrence risk, referrals and support, lifespan, and overall prognosis. Additionally, for a prenatal diagnosis, information about pregnancy-related testing, ultrasound findings, and more support resources was considered essential. For a postnatal diagnosis, information regarding long-term life outcomes and fertility was also deemed essential.

The majority of parents felt informed when they received an initial diagnosis for their child, but fewer than half of parent participants were satisfied with their overall diagnostic experience. Some parents felt they received too little information or an overwhelming amount. Parents who were satisfied felt that providers were well-informed about Turner syndrome and felt these providers were honest with them about expectations.

For a prenatal diagnosis, genetic counselors placed greater emphasis on non-medical aspects of Turner syndrome that may be present after birth, fertility, recurrence risk, and prenatal diagnosis than prenatal parents. A conversation regarding an initial Turner syndrome diagnosis can benefit from an understanding that parents may not

prioritize postnatal non-medical aspects of Turner syndrome at the time of a prenatal diagnosis but instead might appreciate resources with this information that they can later refer to. Parents may desire information about the non-medical aspects of Turner syndrome but might prefer to receive this information after they have had time to process an initial diagnosis. Genetic counselors could instead provide more focus on available supports and resources to better assist parents during these conversations. Prenatal genetic counselors also placed greater emphasis on the risk for fetal demise and termination resources than prenatal parents, who rated these items significantly lower. While parents recognized that providers must present this information during an initial diagnostic conversation, placing less emphasis on this aspect unless the parent is considering termination may be beneficial.

For a postnatal diagnosis, parents placed greater emphasis on prenatal aspects, treatment options, and the family and relationships of individuals with Turner syndrome than postnatal genetic counselors. Genetic counselors should consider incorporating these aspects of Turner syndrome into an initial postnatal diagnosis discussion. Postnatal parents also rated certain strengths of individuals with Turner syndrome higher than postnatal genetic counselors, while postnatal genetic counselors rated certain challenges higher. The informational disconnect between postnatal parents and genetic counselors may be alleviated if genetic counselors mention certain non-medical strengths in conjunction with the challenges that individuals with Turner syndrome face.

Overall, the most important information that should be included when discussing an initial Turner syndrome diagnosis with parents was identified in this study. Discrepancies in informational preferences between parents and genetic counselors were

also identified. Providers can recognize these informational preference discrepancies and incorporate this data into their practice, specifically pinpointing the information that should be included in or excluded from these discussions. This research may also be used to create definitive practice guidelines for communicating a diagnosis of Turner syndrome. Ultimately, with greater provider knowledge about the information deemed most important for an initial Turner syndrome diagnosis, parents can become more informed and satisfied with their experiences in the future.

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## APPENDIX A: INVITATIONAL LETTER & ONLINE SURVEY FOR PROVIDERS

INVITATION TO PARTICIPATE

Dear Provider:

Thank you for your interest in this study. The goal of this Genetic Counseling Master's Thesis Project is to assess the essential informational needs of parents receiving a prenatal or postnatal diagnosis of Turner syndrome. It is critical that parents who receive a diagnosis of Turner syndrome for their child are given balanced information about the condition and are well informed about what to expect. Accurate information is necessary so that parents may access appropriate care.

Participation involves completing an online survey, and it will be taken through Qualtrics. The survey will take approximately 10-15 minutes with the chance to win a $25.00 Visa gift card for completing the survey (odds of winning are estimated to be 1 in 30). 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 Jewel Wasson, the primary investigator, at jewel.wasson@uscmed.sc.edu. Thank you for your participation.

Sincerely,

Jewel L. Wasson, BS

Genetic Counseling Program

University of South Carolina

PRENATAL DIAGNOSIS – DEFINING BALANCED INFORMATION ABOUT TURNER 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 Turner syndrome for their unborn child. Numerical data and frequencies (%) are provided where available.

Rate each item: Prenatal Physical Features and Health Complications of Turner Syndrome (Before Birth)

1. Increased nuchal translucency (excess fluid on the back of the head or neck) in first trimester
2. Pleural effusion ("water in the lungs")
3. Pericardial effusion (heart partially or completely surrounded by fluid)
4. Cystic hygroma (fluid-filled sack on the back of the fetal neck)
5. Coarctation (narrowing) of the aorta and/or left-sided cardiac defects (heart defects)
6. Need an echocardiogram (ultrasound of the heart) during pregnancy
7. May need an evaluation by a pediatric heart specialist
8. Brachycephaly (broad, short skull)
9. Renal anomalies (kidney problems)
10. Polyhydramnios (increase of amniotic fluid volume in pregnancy)
11. Oligohydramnios (decrease of amniotic fluid volume in pregnancy)
12. Growth restriction
13. 99% risk for fetal demise when prenatally diagnosed
14. Only discuss prenatal issues that are actually confirmed via ultrasound
15. Discuss value of ultrasound in detecting prenatal conditions

Rate each item: Postnatal Physical Features and Health Complications of Turner Syndrome (After Birth)

1. Women with Turner syndrome usually live long lives
2. Short stature (95%-100%)
3. Heart defects and other cardiovascular issues (25%-50%)
4. Hearing loss (30%)
5. Vision problems (20%)
6. Hypertension (high blood pressure) (50%)
7. Glucose intolerance (15%-50%)
8. Frequent ear infections (60%)
9. Lymphedema (swelling) in hands and feet (25%)
10. Hypothyroidism (15%-30%)
11. Orthopedic (bone, muscle, joint) problems [scoliosis (10%), decreased bone mineral content (50%-80%), etc.]
12. Kidney issues (10%-15%)

13. High blood pressure (50%)
14. Primary ovarian failure (reduced function of ovaries before age 40) (90%)
15. May have some health conditions but not all
16. Discuss available Turner syndrome clinics
17. Discuss impact of hormones to increase stature or treatments that can allow girls with Turner syndrome to attain a normal adult height (over 5 feet)
18. Girls with Turner syndrome need hormones to start or complete puberty

Rate each item: The Family and Relationships of Individuals with Turner Syndrome

1. Impact on other siblings – more compassionate & caring
2. Impact on other siblings – less attention & resentful
3. Impact on parental marriage – strengthens relationship
4. Impact on parental marriage – strains relationship
5. Impact on grandparents – supportive & welcoming
6. Impact on grandparents – limited interaction
7. Impact on extended family members – supportive & welcoming
8. Impact on extended family members – limited interaction
9. Impact on other relationships – supportive & welcoming
10. Impact on other relationships – lose social circle
11. Financial parental impact – No difference
12. Financial parental impact – More
13. Time commitment of parent – No Difference
14. Time commitment of parent – More

Rate each item: Neurocognitive and Psychosocial Aspects of Turner Syndrome

1. Women and girls with Turner syndrome typically have normal intelligence
2. Intellectual disability (~10%)
3. Poor performance in mathematics (dyscalculia) (50%-75%)
4. Increased risk of attention-deficit/hyperactivity disorder (ADHD) (25%)
5. Lack of working memory which may cause difficulty with multi-tasking, mental calculations, and holding information “in a mind’s eye”
6. Difficulty with problem-solving
7. Inefficiency (e.g., slow and more effortful) when learning through visual means (e.g., pictures and diagrams)
8. Strengths in various aspects of oral and written communication (speaking and reading are commonly strengths for women with Turner syndrome)
9. Clumsiness and delayed motor milestones
10. Difficulty identifying facial emotions
11. Difficulty initiating or maintaining relationships
12. Girls with Turner syndrome may need more support to develop social skills

Rate each item: Long-Term Life Outcomes for Individuals with Turner Syndrome

1. Participating in community activities (clubs, hobbies, sports, volunteer work, etc.)

2. Inclusion in regular classes
3. Special education services
4. Early intervention services
5. Benefit of psychological therapy and support
6. Possible ability to conceive through reproductive assistive technology after a thorough medical examination
7. Alternate ways to become a parent when unable to conceive
8. Finishing high school
9. More likely to earn a baccalaureate degree or higher than the general US female population
10. More likely to be employed than the general US female population
11. Less likely to marry than the general US female population
12. Living independently
13. Having friends and meaningful relationships
14. Having intimate relationships
15. Life expectancy (may be reduced up to 13 years)
16. More like other children than different
17. Excel at verbal skills compared to general US female population
18. Reduced fertility

Rate each item: Genetics of Turner Syndrome

1. Incidence (1/2500 females)
2. No significant increasing incidence with increasing maternal age
3. Caused by the partial or complete loss of the second X-chromosome in females
4. Possible chromosomal/genetic causes of Turner syndrome: 45,X (40%-50%), 45,X/46,XX (15%-25%), Other (25%-45%)
5. Prenatal diagnosis can only be confirmed by chromosome analysis via amniocentesis or CVS
6. Screening results can be obtained via noninvasive prenatal screening (NIPS/cell-free DNA)
7. Chance to reoccur in future pregnancies
8. Reproductive capability of a woman with Turner syndrome
9. Reproductive options for a woman with Turner syndrome (pregnancy, surrogacy, adoption)

Rate each item: Informational Resources & Referrals for Individuals with Turner Syndrome and Their Families

1. Local Turner syndrome support group(s)
2. National advocacy organizations & websites
3. Online support groups/social media platforms
4. Printed/written material
5. Photographs of children with Turner syndrome
6. Fact sheets/brochures
7. Books

8. Contact with families raising a child with Turner syndrome
9. Pregnancy termination resources
10. Alternative/nonconventional therapies
11. Specialist referral(s)
12. Special education supports and services
13. Counselor or family therapist referral(s)
14. Pastoral counseling referral(s)

## POSTNATAL DIAGNOSIS – DEFINING BALANCED INFORMATION ABOUT TURNER 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 Turner syndrome for their child. Numerical data and frequencies (%) are provided where available.

Rate each item: Prenatal Physical Features and Health Complications of Turner Syndrome (Before Birth)

1. Increased nuchal translucency (excess fluid on the back of the head or neck) in first trimester
2. Pleural effusion ("water in the lungs")
3. Pericardial effusion (heart partially or completely surrounded by fluid)
4. Cystic hygroma (fluid-filled sack on the back of the fetal neck)
5. Coarctation (narrowing) of the aorta and/or left-sided cardiac defects (heart defects)
6. Need an echocardiogram (ultrasound of the heart) during pregnancy
7. May need an evaluation by a pediatric heart specialist
8. Brachycephaly (broad, short skull)
9. Renal anomalies (kidney problems)
10. Polyhydramnios (increase of amniotic fluid volume in pregnancy)
11. Oligohydramnios (decrease of amniotic fluid volume in pregnancy)
12. Growth restriction
13. 99% risk for fetal demise when prenatally diagnosed
14. Only discuss prenatal issues that are actually confirmed via ultrasound
15. Discuss value of ultrasound in detecting prenatal conditions

Rate each item: Postnatal Physical Features and Health Complications of Turner Syndrome (After Birth)

1. Women with Turner syndrome usually live long lives
2. Short stature (95%-100%)
3. Heart defects and other cardiovascular issues (25%-50%)
4. Hearing loss (30%)

5. Vision problems (20%)
6. Hypertension (high blood pressure) (50%)
7. Glucose intolerance (15%-50%)
8. Frequent ear infections (60%)
9. Lymphedema (swelling) in hands and feet (25%)
10. Hypothyroidism (15%-30%)
11. Orthopedic (bone, muscle, joint) problems [scoliosis (10%), decreased bone mineral content (50%-80%), etc.]
12. Kidney issues (10%-15%)
13. High blood pressure (50%)
14. Primary ovarian failure (reduced function of ovaries before age 40) (90%)
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7. Inefficiency (e.g., slow and more effortful) when learning through visual means (e.g., pictures and diagrams)

8. Strengths in various aspects of oral and written communication (speaking and reading are commonly strengths for women with Turner syndrome)
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2. Inclusion in regular classes
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6. Possible ability to conceive through reproductive assistive technology after a thorough medical examination
7. Alternate ways to become a parent when unable to conceive
8. Finishing high school
9. More likely to earn a baccalaureate degree or higher than the general US female population
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11. Less likely to marry than the general US female population
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9. Pregnancy termination resources
10. Alternative/nonconventional therapies
11. Specialist referral(s)
12. Special education supports and services
13. Counselor or family therapist referral(s)
14. 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.

- Please provide a sample description of what you consider to be a balanced presentation of Turner syndrome.
- What medical aspects of Turner syndrome should be routinely included in a diagnosis? What non-medical and lifestyle aspects of Turner syndrome should routinely be included in a diagnosis?
- There are obvious differences between prenatal & postnatal diagnostic settings. How should information differ between prenatal and postnatal settings?
- 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 Turner syndrome?
- Please share any additional comments, suggestions, experiences, etc. as you see helpful for medical professionals.

DEMOGRAPHIC INFORMATION

1. Your current age (in years)

2. Sex
    a. Male
    b. Female

3. 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)

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

5. 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

6. Current job title
    a. Genetic counselor
    b. Physician

For genetic counselors only:

1. Year you earned your Master of Science in Genetic Counseling degree (YYYY format)

2. 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

3. Current U.S. state in which you practice (two letter abbreviation format)

4. Current work setting
   a. Academic/University-based
   b. Hospital-based
   c. Multiple specialty group
   d. Private practice
   e. Industry
   f. Other (please specify)

5. Current specialty area of practice – check all that apply
   a. Prenatal
   b. General Pediatrics
   c. Pediatric subspecialty
   d. Cancer
   e. Adult genetics
   f. Infertility
   g. Laboratory
   h. Research
   i. Other (please specify)

6. 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

7. Previous areas of practice – check all that apply
   a. Prenatal
   b. General Pediatrics
   c. Pediatric subspecialty
   d. Cancer
   e. Adult genetics
   f. Infertility
   g. Laboratory
   h. Research
   i. Other (please specify)
   j. No previous specialty area

8. Have you delivered a positive screening result or diagnosis of Turner syndrome?
    a. Yes, positive screening result only
    b. Yes, diagnosis only
    c. Both
    d. Neither

9. Extent of contact with individuals with Turner syndrome or parents of individuals with Turner syndrome in training
    a. No contact
    b. Some contact
    c. Plenty of contact

10. Extent of contact with individuals with Turner syndrome or parents of individuals with Turner syndrome in current area of practice
    a. No contact
    b. Some contact
    c. Plenty of contact

11. Extent of contact with individuals with Turner syndrome or parents of individuals with Turner syndrome outside of a professional context
    a. No other contact
    b. Social contact only
    c. Family contact only
    d. Both social and family contact

For physicians only:

1. Are you a Doctor of Medicine or Doctor of Osteopathic Medicine?

2. Year you earned your Doctor of Medicine or Doctor of Osteopathic Medicine degree (YYYY format)

3. 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

4. Current U.S. state in which you practice (two letter abbreviation format)

5. Current work setting
    a. Academic/University-based

b. Hospital-based
c. Multiple specialty group
d. Private practice
e. Industry
f. Other (please specify)

6. Current specialty area of practice - check all that apply
    a. Obstetrics and Gynecology
    b. Maternal Fetal Medicine
    c. Medical Genetics
    d. Endocrinology
    e. Pediatric
    f. Pediatric Endocrinology
    g. Reproductive Endocrinology
    h. Other (please specify)

7. 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

8. Previous areas of practice – check all that apply
    a. Obstetrics and Gynecology
    b. Maternal Fetal Medicine
    c. Medical Genetics
    d. Endocrinology
    e. Pediatric
    f. Pediatric Endocrinology
    g. Reproductive Endocrinology
    h. Other (please specify)
    i. No previous specialty area

9. Have you delivered a positive screening result or diagnosis of Turner syndrome?
    a. Yes, positive screening result only
    b. Yes, diagnosis only
    c. Both
    d. Neither

10. Extent of contact with individuals with Turner syndrome or parents of individuals with Turner syndrome in training
    a. No contact

b. Some contact
c. Plenty of contact

11. Extent of contact with individuals with Turner syndrome or parents of individuals with Turner syndrome in current area of practice
    a. No contact
    b. Some contact
    c. Plenty of contact

12. Extent of contact with individuals with Turner syndrome or parents of individuals with Turner syndrome outside of a professional context
    a. No other contact
    b. Social contact only
    c. Family contact only
    d. Both social and family contact

*You have reached the end of the survey, but your responses are not yet submitted!*

If you are interested in entering the raffle for taking this survey, please copy the following link into a new browser window to enter your information! Please do so **BEFORE** hitting submit, as the link will not be available to you after you leave this page.

https://uofsc.co1.qualtrics.com/jfe/form/SV_38AHV0CPMcilld3

Do not forget to submit this survey by hitting the arrow below!

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 Turner syndrome and their families.

We invite you to share this online survey with other genetic counselors and/or physicians. They 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 Turner syndrome. Parents may access the online survey through Qualtrics.

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

RAFFLE SURVEY

By entering your information, you are willingly entering a raffle for a $25.00 Visa gift card (odds of winning are estimated to be 1 in 30). You are eligible for this raffle because you completed the research questionnaire, "Essential Informational Needs of Parents Receiving a Turner Syndrome Diagnosis: Parental, Genetic Counselor, and Physician Perspectives". Your information will not be distributed and will not be used to contact you unless you are the winner of the raffle.

If you would like to enter the raffle, please click the next button below. If not, please exit this survey.

Please complete the form below:

- First and last name
- Preferred title (Mr., Ms., Dr., etc.)
- Preferred Email

## APPENDIX B: INVITATIONAL LETTER & ONLINE SURVEY FOR PARENT

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 Turner syndrome advocacy organization, social media group, or support group and because you have received a diagnosis of Turner syndrome for your child or baby. The goal of this Genetic Counseling Master's Thesis Project is to assess the essential informational needs of parents receiving a prenatal or postnatal diagnosis of Turner syndrome. Understanding these perspectives can allow for healthcare professionals to deliver an initial diagnosis of Turner syndrome to parents in a more informative and supportive way.

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 a Turner syndrome diagnosis and their families.

Participation involves completing an online survey, and it will be taken through Qualtrics. The survey will take approximately 10-15 minutes with the chance to win a $25.00 Visa gift card for completing the survey (odds of winning are estimated to be 1 in 30). 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 Jewel Wasson, the primary investigator, at jewel.wasson@uscmed.sc.edu. Thank you for your participation.

Sincerely,

Jewel L. Wasson, BS

Genetic Counseling Program

University of South Carolina

Essential Informational Needs for a Turner Syndrome Diagnosis: Parent Survey

YOUR CHILD'S DIAGNOSIS

Please provide information about your experience with receiving your child's or baby's diagnosis of Turner syndrome.

1. Have you received a diagnosis of Turner syndrome for your child or baby? (yes/no)

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

3. Was your child's diagnosis of Turner 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.
        i. Did your baby survive to birth?

    b. Postnatal diagnosis (after birth). Please enter your child's age when the diagnosis was made (in days, weeks, months, or years)

4. Who informed you of your child's diagnosis of Turner syndrome?

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

5. What is your child's karyotype (chromosomes) result?

    a. Monosomy X (45,X)
    b. Monosomy X mosaicism (45,X/46,XX)
    c. Other
    d. Unknown

6. 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

7. 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

8. Did you receive prenatal screening or prenatal diagnostic testing during your pregnancy with your child with Turner 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

9. How well were you informed about Turner syndrome when you were given your child's diagnosis?

a. Very well-informed
b. Somewhat informed
c. Not very informed

10. Did you see a Genetic Counselor regarding your child's diagnosis of Turner 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:

1. Rate your overall satisfaction with your experience of receiving your child's or baby's diagnosis.

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

2. Are you a member of a Turner syndrome advocacy group?

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

3. 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: Please list which groups or forums you are involved in.

DEFINING BALANCED INFORMATION ABOUT TURNER SYNDROME

Below is an extensive list of features associated with Turner 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 Turner syndrome. Numerical data and frequencies (%) are provided where available.

Rate each item: Prenatal Physical Features and Health Complications of Turner Syndrome (Before Birth)

1. Increased nuchal translucency (excess fluid on the back of the head or neck) in first trimester
2. Pleural effusion ("water in the lungs")

3. Pericardial effusion (heart partially or completely surrounded by fluid)
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15. May have some health conditions but not all
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12. Special education supports and services
13. Counselor or family therapist referral(s)
14. 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.

- There is a need for medical professionals to include both the medical and non-medical aspects of Turner syndrome when describing the condition to current or

expectant parents. What non-medical/life aspects of Turner syndrome should routinely be included as part of a balanced presentation?

- Please comment on the presentation of your child's diagnosis, the information you were given about Turner syndrome at that time, & your overall satisfaction with your experience.
- 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 your pregnancy or from raising a child with Turner syndrome? If yes, what information was most useful for you? If no, what information should have been provided that was not?
- Consider your own personal experience of receiving your child's diagnosis. What could have been done differently in order to improve the experience?
- Please share any additional comments, suggestions, experiences, etc. as you see helpful for medical professionals.

DEMOGRAPHIC INFORMATION

1. Your Current Age (in years)

2. Sex

    a. Male
    b. Female

3. 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)

4. Level of Religious Activity

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

5. 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's 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)

6. Marital Status

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

7. 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

8. Current state/place of residency

9. 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

*You have reached the end of the survey, but your responses are not yet submitted!*

If you are interested in entering the raffle for taking this survey, please copy the following link before submitting this survey. After you have submitted this survey, you may paste the link into a new browser window to enter your information.

https://uofsc.co1.qualtrics.com/jfe/form/SV_9LTIZZUJaS9ExbT

Do not forget to submit this survey by hitting the arrow below!

THANK YOU

Thank you for completing this survey. Your participation in this study is greatly appreciated and extremely valuable to all healthcare professionals involved in distributing information and resources and those involved in the care of individuals with a Turner syndrome diagnosis and their families.

We invite you to share this online survey with other parents who have received a diagnosis of Turner syndrome for their child or baby. Parents may access the online survey through Qualtrics.

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

RAFFLE SURVEY

By entering your information, you are willingly entering a raffle for a $25.00 Visa gift card (odds of winning are estimated to be 1 in 30). You are eligible for this raffle because you completed the research questionnaire, "Essential Informational Needs of Parents Receiving a Turner Syndrome Diagnosis: Parental, Genetic Counselor, and Physician Perspectives". Your information will not be distributed and will not be used to contact you unless you are the winner of the raffle.

If you would like to enter the raffle, please click the next button below. If not, please exit this survey.

Please complete the form below:

- First and last name
- Preferred title (Mr., Ms., Dr., etc.)
- Preferred Email

## APPENDIX C – GENETIC COUNSELOR FREE RESPONSE SECTION

## RESULTS

Please provide a sample description of what you consider to be a balanced presentation of Turner syndrome.

- Depends on the age of the patient when diagnosed. Address how it relates to known symptoms (if applicable), address on-going needs (typically general like endo and homoromes but not details about all endo complications).
- Prenatally that there are increased risk for fetal demise and organ defects, however post-natally if they have a livebirth, it is fairly straightforward managment and typical life expectancy and goals
- Describing characteristic features and management, discuss variability, and discuss strengths of women with Turner syndrome
- Present unbiased information regarding prenatal and postnatal course of Turner syndrome.
- Turner syndrome is a chromosomal condition in which, typically, a person has only one X chromosome rather than two sex chromosomes. Most often, people who have Turner syndrome identify as women or female. Some of the main features of Turner syndrome include short stature, learning disability, absence of puberty and infertility, and sometimes a heart defect. There are other features that may be associated with Turner syndrome. Not every girl diagnosed with Turner syndrome will develop any or all of these features. Some people may not even know that they have Turner syndrome.
- Describing the large variability between girls with TS, that most don't have all the features known to be associated, and that they will be more like their peers/siblings than other girls with TS.
- Turner syndrome is a genetic disorder caused by missing all or part of a second sex chromosome, meaning women with Turner syndrome typically only have one X chromosome. Primary features of Turner syndrome include short stature, heart defects (coarctation of the aorta), and kidney defects. Individuals can also have hypothyroidism, hearing loss, and vision problems. Physical differences can include a broad chest, webbed neck, and low posterior hairline. Most individuals with Turner syndrome have normal intelligence, but there can be some developmental delays and/or specific learning disabilities such as difficulty with spatial reasoning. Girls can have some challenges socially. Girls and women with Turner syndrome typically have early ovarian failure and do not go through puberty on their own. This typically means infertility, but that is not true for all women with Turner syndrome, and there are a variety of ways to have families.

Many of these details and treatment discussions can be further talked about with the appropriate specialists.

- Making sure to touch on all the possible symptoms, but also making sure to discuss that not everyone will have all of them/variable presentation. Additionally, discussing what life looks like for parents, siblings, and for the child with Turner syndrome.
- TS is an issue with the sex chromosomes where girls are missing one x. they are typically shorter than their peers, they have increased risks for learning disabilities and intellectual delays (not necessarily intellectual disability though), and an increased risk for heart and kidney defects. There is also a significantly increased risk for pregnancy loss. (information given in a prenatal setting before any screening and testing)
- Turner syndrome has a wide range of variability and it is not possible to predict exactly what they will experience. I discuss the range of medical and developmental issues so that the family is aware, but make sure to emphasize that each child will follow her own path and we never want to limit opportunities based on the diagnosis.
- Complete or partial absence of the second X chromosome in some or all of an individual's cells, which leads to numerous features which typically include short stature, an increased risk for cardiac defects (particularly left sided lesions), renal anomalies, vision and hearing issues, and ovarian failure which often results in infertility. Intelligence is usually typical in women with Turner syndrome but they may need assistance with particular subjects. Many individuals with Turner syndrome have some unique physical or facial features but these are not as noticeable to the lay individual as say Down syndrome. Every individual with Turner syndrome is unique, just like you and I are unique. Turner syndrome is typically due to a sporadic nondisjunction error in paternal meiosis, however the missing X can be maternal or paternal in origin. There are many cytogenetic variations in Turner syndrome, and some partial X deletions can be below the resolution of karyotype. Unfortunately 99% of conceptions with Turner syndrome are lost as spontaneous miscarriages. There are numerous management and surveillance recommendations, including hormone therapy to assist with growth and puberty. There are numerous ARTs available to assist women with Turner syndrome in having a family. Some women with Turner syndrome can spontaneously conceive pregnancies.
- This is a variable condition. Some women with Turner syndrome have few or no symptoms and never know. MOST will have short stature and fertility issues. Those more severely affected may have some medical concerns such as heart or kidney defects, and an increased risk for some conditions like thyroid problems. They may have trouble with certain types of learning, but usually have normal IQ and do well with support.
- Turner syndrome has a wide variety of features and severity. Most individuals have normal intelligence, however, there can be learning deficits in certain areas. Other areas that can be affected are the heart, stature, and kidneys - it is important for individuals to be screened for these concerns to ensure they get the best care.

- An outline of the different possible medical concerns and what type of intervention may be needed but also outlining that most women with Turner syndrome do not realize their diagnosis. Primary concern is short stature, some learning difficulties and infertility. In prenatal setting however, emphasizing the large likelihood of miscarriage and pregnancy loss.
- Prenatal - women & girls with Turner syndrome may have short stature and issues with puberty and fertility. They are also at risk for certain health complications such as heart problems or kidney problems. Intelligence is typically normal, but many girls may have a learning disability or different learning methods. Pregnancies with Turner syndrome have a high risk of miscarriage, often due to cystic hygroma or other fluid accumulation. If these pregnancies make it to term, most girls with Turner syndrome do quite well.
- In a prenatal setting, a balanced presentation offers all options for ending or continuing a pregnancy with a focus on the high liklelihood of fetal demise. A balanced presentation should represent the actual balance of probabilities of the outcomes. I would focus on pregnancy management and realistic options & outcomes.
- Addressing the symptoms that led to genetics workup, what causes Turner, why additional referrals/treatment may be needed (ie: endocrine for discussion on growth hormone, echo of heart). Then discuss other features as parents voice questions/concerns regarding growth, intelligence, other health concerns. Focus on lack of blame, options for treatment and connecting, emphasize psychosocial processes of grief, changes in expectations. Inform family of resources available, let family dictate what they want or need currently. Check in on each family member present, including patient if old enough to understand.
- Although most pregnancies affected with Turner syndrome end in miscarriage, girls who are born with Turner syndrome typically do well. Many babies with Turner syndrome experience challenges early in life and may have ongoing medical conditions to manage as they get older. However most have normal intelligence and can live happy and fulfilling lives.
- A chromosomal condition that occurs when one of the two copies of the X chromosome is lost sporadically, or by chance. Increased chance of SAB prenatally, however women with Turner syndrome often live long, productive lives. Individuals with Turner syndrome are typically shorter (under 5 feet tall), and may have heart and kidney differences. They often have infertility, however conception through ART may be possible. Other health conditions commonly seen in the general population may be seen more frequently in women with Turner syndrome (HTN, thyroid issues, etc.). Most women with Turner syndrome have normal intelligence, and many finish high school, live independently, hold competitive jobs, and earn advanced degrees. Some women with Turner syndrome may have some learning difficulties in some areas (e.g. problem solving) and excel in other areas (e.g. verbal skills). Developmental delays and intellectual disability can be seen in individuals with Turner syndrome. The initiation of therapies and support early in life can help children with Turner syndrome achieve milestones on their own timelines. Women and children with Turner syndrome are more like other people than they are different.

- Given I'm a prenatal counselor: discussing the prenatal risks (miscarriage, hydrops), risk for birth defects vs healthy at birth, and discussion of what mental abilities/difficulties might be present. Presenting normal IQ and long life expectancy to parents.

What medical aspects of Turner syndrome should be routinely included in a diagnosis? What non-medical and lifestyle aspects of Turner syndrome should routinely be included in a diagnosis?

- Heart defects & evaluation, renal defects & evaluation, infertility issues, hormone replacement options, awareness of learning difficulties and neuropsych eval. Non-medical discussing puberty and approaching differences in children's experiences, expect typical intelligence, independent living
- Cardiac, fertility, short stature are most essential; intelligence and Turner syndrome community are especially important
- Risk for birth defects and pregnancy complications; need for specialist referrals following delivery; overview of medical care to be expected over lifetime
- Heart defects and possibility of kidney abnormalities should be discussed. Learning disabilities should be discussed.
- heart and renal defects, short stature, normal intellect with learning issues, relationship/psych
- The ones that would require intensive interventions particularly the cardiac presentation and risk for gonadoblastoma if mosaic with Y chromosome material. Also important to tailor the information to the presentation of the girl (hearing loss, delayed puberty, short stature...). Should also discuss the normal intelligence and ability to live a full independent life
- Medical: short stature, heart defects, renal defects, hearing loss, ear infections, vision problems, ovarian failure, and infertility. Non-medical/lifestyle: typically normal intelligence, sometimes developmental delay, sometimes learning disability such as difficulty with math and spatial reasoning, sometimes difficulty socially. Positive things: I usually include examples I know of where women with Turner have gone to college or have master's degrees. I also usually talk about several options for having families.
- The main features (short stature, normal intelligence, heart defects) should be discussed. Lifestyle aspects should include life expectancy, fertility, and support groups/psychosocial supportincreased risk of intellectual delays, cardiac defects, kidney defects; sig increased risk of pregnancy loss
- I discuss the possible medical complications routinely even if it hasn't affected the patient yet so that the family can start to anticipate what medical issues may arise in the future and can have a better sense of what to look out for. We discuss early intervention services as well as ways to talk about Turner syndrome with peers and relatives
- cardiac, vision/hearing, recurrent OM, renal anomalies, typically normal intelligence, increased risk for learning disabilities such as issues with mathematics, need for hormone therapy

- Medical: heart defects and infertility. Non-medical: possible learning/developmental delays
- The features that are most prevalent and require referrals and ongoing care - cardiology and endocrinology. Other aspects that should be discussed would be intelligence/milestones/performance in school. Caring for individuals with Turner syndrome should be discussed and an emphasis on the variability should be included - individuals with Turner syndrome do not all have the same features or severity.
- Life expectancy, intellectual function, that this does not define her
- I work in a prenatal clinic so primarily focusing on possible prenatal findings including CDH's and congenital renal anomalies are most important. I like to emphasize that it is not typically associated with ID, however can be associated with learning differences and early intervention is recommended. Also, like to discuss some social concerns problems these women may have and the financial impact. Also, discussing fertility concerns. Most importantly talk about risk of pregnancy loss.
- Depends on timing. Prenatally, I find it hard to discuss too many details of postnatal medical aspects in pregnancy with high risk of demise (due to ultrasound findings). I tend to defer a lot of the specifics to later follow-up if the pregnancy progresses, and focus initial information on overall description & features with specific info on current prognosis and prenatal presentation. I always provide support info (i.e. Turner syndrome society) which I think is essential. Options to connect with other families is also helpful.
- The medically actionable aspects of Turner syndrome should be routinely included in a diagnosis, as well as the pertinent negatives like a typically normal intelligence. In a prenatal setting, I would not feel that lifestyle aspects are an essential piece of information. Helpful, but not essential. Postnatally, I think it is essential to routinely include information about talking to your daughter about her diagnosis so that the family does not keep it a secret.
- Medical: the reason the patient presented to genetics care, parts of care that fall within recommended management (stature, blood pressure, estrogen replacement, neuropsych assessment). Non-medical and lifestyle should be broad (much like peers, may have some learning or social differences), then focus on the family's questions and needs.
- Risk for aortic stenosis or other heart defect, infertility, risk for miscarriage (in prenatal setting). Non-medical aspects should include cognitive challenges and strengths, differences in appearance, how girls with Turner syndrome experience their condition and feel about themselves
- Short stature, possibility for heart defects, possibility for kidney differences, ovarian insufficiency, risk for miscarriage, more alike than different, live independently, inclusion in regular classes, finish high school, hold competitive jobs, go to college, have meaningful relationships, ability to have children (either with ART or via adoption, donor egg, surrogacy, etc.)
- Medical - risks for birth defects and miscarriage, risks for high blood pressure and infertility. Non-medical - thinking about mental abilities and learning styles. Being unable to go through puberty or natural pregnancy.

There are obvious differences between prenatal & postnatal diagnostic settings. How should information differ between prenatal and postnatal settings?

- Postnatal more focused on what child currently has vs. possibilities of miscarriage, new findings
- In the postnatal setting, you have a person in front of you to tailor your counseling to whereas in a prenatal setting, there might be an increased NT, cystic hygroma, or other findings on imaging, but it's important to generally describe all aspects of the condition, including the possibility of miscarriage and option for termination
- This will vary from patient to patient and will depend on the nature of the diagnosis. Information should be tailored to each patient's needs.
- Amount of information should be tailored to the patient's needs in the prenatal setting as the patient may be making decisions with regard to continuing or not continuing the pregnancy. I tend to present less information and support options at the start of a prenatal discussion and only offer up information that the patient wants at that time since we do not know what features the child will develop.
- Prenatal often includes counseling about pregnancy decisions and less about the specific health issues that need to be discussed with a child with TS.
- Prenatal is more pressing issues. A general overview of the condition and discussion about options and faiclitating decision making. Post natal is alot more hands on, specialist related. Follow up appointment allow topics to be addressed as the child meets milestones. Prenatal typically does not have follow up.
- The increased risk for miscarriage should be addressed in a prenatal setting along with the knowledge that multiple ultrasound findings doesn't directly correlate with how many symptoms a girl with TS will develop in the future.
- prenatal is going to be very difficult unless the family actually meets children with Turner syndrome
- I see pediatric patients, and I have discussed the new diagnosis of Turner syndrome many times over the years. I think postnatally, you need to focus on the features right in front of you. Yes, you should discuss things that can come up in the future. However, there are a lot of things you don't need to discuss at all now that the prenatal time is over (risk of miscarriage, etc.). I also think it is important to pay attention to the age at diagnosis. I have had much different conversations with families depending on if the diagnosis occurs around the time of birth/infant period vs those diagnosed around 13-15 years due to not starting periods. Postnatal discussions are less hypothetical than prenatal, in my opinion.
- Prenatal should discuss more about prenatal testing options (screening vs diagnostic), recurrence, and ultrasound findings. Prenatal should also include the major medical aspects. Postnatal counseling should focus on the medical aspects and the future lifestyle aspects.
- prenatal should focus more on information to facilitate decision making
- The big concern prenatally is if the fetus will even make it to delivery. So there needs to be some kind of expectation management, whereas postnatally, the child has clearly survived, so it's more setting the parents and child up for success.
- Prenatal largely depends on what trimester your are talking to your patient and the existing ultrasound anomalies. If you have a large hygroma and cardiac defect the

conversation should focus on more immediate issues, such as the risk of loss, and not long-term information

- In the prenatal setting, it is important to discuss the high incidence of pregnancy loss in cases of prenatal diagnosis of Turner syndrome while also presenting a balanced representation of postnatal outcomes for individuals with Turner syndrome.
- I think prenatal should be focused on big picture and helping the family make the decision to do diagnostic testing, continue vs. terminate etc. And at some point, what to expect immediately after delivery. In the postnatal setting, I think it is appropriate to start to talk about longer term management, and of course start to implement therapies/interventions (which does not apply prenatally).
- Prenatal should include the big factors such as life expectancy and the more common features of cardiac concerns, short stature, and infertility. All the other specifics can come again later in a postnatal setting. The most important this is to provide the family the information they want - if they want to know all the details then that should be shared but if their only question is for life expectancy then you don't need to explain risk for hypertension. Post-natally, you need to give those details more so they understand the medical management requirements moving forward.
- prenatal - u/s findings, pregnancy choices, variability...postnatal - referrals, stages of life, fertility
- There are lots of the prenatal things regarding testing/evaluation that can be skipped for a postnatal diagnosis. Postnatal diagnosis can focus on what are immediiate next steps in care.
- Yes, while Turner syndrome is considered a relatively "mild" disorder in the prenatal setting it is incredibly important to keep in mind that there is a risk of fetal demise. Furthermore, talking about congenital defects is more pressing in the prenatal period and the possibility of different medical disorders vs. what is truly going to be presenting. Talking about the variability is also more important in the prenatal period and especially discussing most pressing concerns such as any ultrasound findings.
- The information provided in a prenatal setting is going to be more tailored to the patient. Have they already decided they are terminating or continuing the pregnancy? Are they undecided and looking for as much information as possible? Ultrasound also plays a role in discussing the prognosis. Was the diagnosis made because of a cystic hygroma on ultrasound? Or a mosaic diagnosis was made after first being picked up on screening, with no ultrasound findings?
- See previous answer. Postnatal should focus less on the prenatal presentations, since these are not highly relevant after delivery. They should focus more on immediate needs in whatever time frame the diagnosis occurs - newborn, childhood, adolescence, etc. Can then expand on etiology, recurrence risk, etc. as appropriate
- Prenatal should focus on the high likelihood of fetal demise and facilitating decision making about continuation vs termination and what to expect after delivery. Postnatal should focus arming the parents with information needed to care for their child now and in the future

- Prenatal needs to focus on concerns and options for the family in short term, and how to manage the rest of pregnancy, with education for postnatal course added over time as delivery approaches. Other than addressing prenatal concerns for 45,X, I don't think postnatal diagnosis disclosure needs to discuss prenatal concerns associated broadly with 45,X. Postnatally often has the option to add more information over multiple visits and years as well.
- Prenatal settings should keep information about potential phenotypes broad and keep in mind the extremely high chance that the pregnancy will not continue to term. Memory-making and grief counseling should be anticipated. In a postnatal session, counseling should be tailored as much as possible to child's specific phenotype and needs. Family planning discussions should also take place.
- High risk of miscarriage is a big consideration in a prenatal diagnostic setting, and a nonissue in postnatal. Reproductive options are also relevant in the prenatal setting, not postnatal. Specialist referrals become more important after the child has been born and can be seen in clinic.
- In prenatal counseling parents may choose to end the pregnancys so this must be considered. In prenatal counseling there remains a high likelihood of prenatal loss so this is also different given it is not always a rosy picture.

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 Turner syndrome?

- I think medical professionals need to be comprehensive and as balanced as possible
- Always stay up to date on all aspects of medical care and information and offer to share this with families.
- I think medical professionals have a responsibility to discuss the most hallmark features of Turner syndrome and stress that not all patients with Turner syndrome will develop every feature.
- There is a moral/ethical obligation to provide the complete medical complications that may be associated but also to keep it balanced with the positive life experiences many women with TS experience
- I'm not completely sure on this one. I think it is important to give a fairly comprehensive and balanced description as well as provide the family with access to reliable resources. However, it would be impossible and not beneficial to the family to exhaust every tiny detail about the condition.
- It is important to inform parents about the main medical complications and health risks so they are fully informed when making decisions about pregnancy management. Additionally, it is important to include the wide variety of medical complications but to make a point that there is variability and not everyone with Turner syndrome will have all medical complications.
- in the event of a confirmed diagnosis, they need to go through all of the information in detail. in the event of a positive screening test they need to make it clear that it is NOT a diagnosis but an increased risk only

- I think it is important to inform of possible health risks, both so that the family can adapt and prepare and also knowing that information is widely available on the internet I think that it helps to build trust to share the array of features they may read about so that they have the opportunity to ask questions rather than finding out through their own reading.
- Balanced information, transparent
- Make sure the parents have the opportunity to talk to someone (like a genetic counselor) who can present a balanced presentation. And we must explain the breadth and variability, and also give the patient the opportunity to research (from reliable sources) on their own.
- To present the information without bias or with an ulterior motive. If a family is unsure about termination do not present only the bad features such as cardiac concerns, but make sure to include that intelligence is likely normal and that there are fertility options if that is wanted one day. Alternatively, don't make the condition out to be fine, families need to know the medical care that will be required.
- I think it is important to provide all of the possible outcomes based on how much information the parents want to know in a non-biased way. A lot of prenatal providers consider it a "mild" disorder whereas others emphasize the adverse outcomes. While this counseling should differ based on the most pressing concerns such as if a cystic hygroma is seen or the likelihood of IUFD is more likely, however talking about the facts and possibilities and allowing parents to make decisions themselves about proceeding with the pregnancy is especially important in the prenatal period.
- the idea of a "balanced" description is always the gold standard - difficult in practice with all the info that must be presented. I think getting the most important and relevant information first is the responsibilities of the providers as appropriate for the specific patient/family.
- The aspects that can affect the health & development of the child are essential
- In today's age of the internet and accessibility, I don't know that there's a legal obligation beyond sharing results and professional recommendations. However, ethically I believe that a postnatal diagnosis is ethically obligated to include information on medical complications that either can be terminal or that have direct management recommendations available, much like the current guidelines for secondary findings on exome and genome sequencing.
- Our responsibility includes showing patients how to access the information they need (from clinics, from websites, scholarly articles, support groups etc.) while also ensuring that patients/their families understand that not every patient will experience every possible phenotype.
- Balanced description is necessary
- It is their duty to be thorough and cover the range of issues that can come up, but also put into perspective the fact that each girl is different and won't have every concern listed.

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

- I did this whole survey on the first page thinking that we had transitioned to the postnatal convo when it had the postnatal header (whoops). All of those answers should be on page 2 instead.
- Turner syndrome may also be caused by the absence of a Y chromosome, not necessarily an X (commenting on one of the bullet points in this survey)
- N/A
- I think sessions discussing the features of TS should be tailored to the presentation while also including information on the spectrum of features. I also try to discuss that this does not necessarily have to define who the patient is, just helps give us guidance as to certain medical conditions to watch for but will still be more similar to the family.
- As stated previously, I think it is important to tweak your discussion based on the age of diagnosis and the features/severity of presentation of your patient.
- My clinic has few geneticists and a very long waitlist. We usually rely on endocrinology to follow the patient and give more detailed information about their management. We don't have enough time or professionals to continue to follow them. So I usually try to talk about why they need to follow endo, inheritance, recurrence risk, and intelligence related information in the result session instead of going super in depth with the other things
- This was a hard survey to fill out because so many items for me are dependent on each specific case.
- I often think about essential information by asking the question "If they never came back to see me again, what information do I absolutely need to make sure they heard"
- I think as set up, this survey doesn't acknowledge that Turner syndrome diagnoses do not routinely have multiple prenatal visits, and some but not all do have multiple postnatal visits. It's hard to fill this out not knowing whether to assume this is information that is only available in a single encounter, or if this is considered a disclosure, initial encounter, etc.

## APPENDIX D – PARENT FREE RESPONSE SECTION RESULTS

There is a need for medical professionals to include both the medical and non-medical aspects of Turner syndrome when describing the condition to current or expectant parents. What non-medical/life aspects of Turner syndrome should routinely be included as part of a balanced presentation?

- High sensory needs, stranger anxiety, harder getting a normal sick diagnosis when TS is in play.
- Every Turners girl is unique like every human is unique. There will be a lot of information given at the time of diagnosis and not all the information may be relevant to your child's situation.
- How this will affect her in life skills, peers, relationships
- The mental strength for parents is most important thing.
- The importance of advocacy for your child's educational rights
- Peer relationship and learning struggles
- Anything that isn't fact based should not be included in a medical presentation. I feel medical doctors could say that TS patients can and do live a long and healthy life but much more than that would be opinion. With each girl being so unique, it is difficult to say for certain what outcomes will happen.
- They lead good lives; social skills and learning concerns
- Social issues that may affect child
- For me the research is so limited with TS and the information that is out there in textbooks is so old that is hard to get an idea of what it is like to have a child with TS. Reading about TS for me was a lot more scary than actually having my daughter here in my arms. She has had some challenges but they have been few and don't define her. She is pretty typical of any other child her age. Also, for the pregnancy, there is statistics and I understand the need to share them with the parents but there are many girls that survive the pregnancy. My daughter had hydrops, cystic hygroma, pericardial and pleural effusions and it was a very scary pregnancy but hers resolved and she is a miracle. There's no way to predict which baby will survive and sadly which baby won't make it but by terminating a pregnancy you would never know if your child could have survived. I've found through the support groups i am in that many doctors push termination and that breaks my heart.
- Cognitive function, neurodiverse conditions, mental health challenges
- In my experience it was a good balance of what to expect
- Emphasis on positives; i.e., normal intelligence, ability to do what other children can, etc.
- Access to the TSSUS website

- I think non-medical is helping your child deal with the diagnosis my daughter was late in life and still will not associate with turner syndrome support systems as a young adult, she has anger issues and I worry about her inability to make relationships. She hates going to doctors because there were so many at the beginning and I think this will affect their ability to monitor her physically like hearing. Also, I think there needs to be regular psychological resources and that the patient should be treated on a wholistic level not just medical.
- The ability to lead a normal life
- It should be stressed that many girls and women with Turner syndrome are happy with fulfilling lives. Connecting with TS groups can be very helpful for parents and children.
- Need for support in developing socially.
- Impact on families, social-emotional difficulties of girls with TS, and executive functioning challenges
- Each child with TS will display more or fewer of the TS characteristics. That is very important to know. I read that women with TS have impaired math ability -- not true of my daughter. But organizational and executive functioning skills are quite common, and helping your daughter to learn them early on is very helpful for her. Watch for difficulties with social interactions, arrange play dates, encourage social skill development.
- The most medically relevant (cardiac, kidney) - parents need to understand. But parents also have to know that this disorder isn't the worst, either.
- We appreciated the OB/GYN telling us that our daughter will most probably be of normal intelligence.
- Treatments and early interventions will provide a child with an advantage to grow and develop along with her peers, which can greatly help foster a positive self-esteem.
- Our biggest impact has been dyscalculia and fine motor skills impact. It would have been nice to have evaluations done at an early age.
- mental health; learning
- Quality of life for TS women (stats related to life achievement/milestones, showcase stories of TS women's success including building families via adoption and/or IVF, academic achievements), share information about any conditions that are associated with TS so parents can informally screen or watch for any indications, definitely share about Early Intervention & community resources the parents can access to provide support for their child's successful growth & development
- Support is needed just because a child is doing well one day take the help from professionals when offered.

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

- Very emotional. Was only met with a genetics counselor once before birth when we go the news and that was it. Most of the time i had to use google to inform myself and other's
- We were given a book that outlined all the possibilities with this syndrome. At this point we were not sure whether any of them or all of them were relevant to our daughter! This was overwhelming until all of her testing was completed and we knew what we had ahead of us. We did not receive any counselling aside from general clinical care.
- With the coarctation the was suspicion of something chromosomal. While in hospital genetics came in and told me but the no follow up. Were still in hospital but I would expect more communication with genetics. Looks like I'm going to call them tomorrow.
- It was a late diagnosis, so we were lacked in many ways
- We were given a brief medical description of Turner's at my daughter's birth when she was diagnosed. The pediatrician simply described her physical attributes and what medical issues she was exhibiting, along with her karyotype test. We didn't receive genetic counseling until a few months later. It was disheartening.
- Diagnosed 5 for low growth. Given basic facts a lot of appointment and information at first.
- I was very overwhelmed with the new diagnosis. Our genetic department did an amazing job of presenting all of the facts regarding TS. I wouldn't change a thing. We were referred to all of the specialists that we needed to see. This is where better information is needed. Most specialists know nothing of Turner Syndrome except what the basic textbook tells them. Information in textbooks needs to change from the outdated "TS girls are doomed to a life of despair" to a better outlook for most TS patients.
- Growth hormones, estrogen,
- Unexpected, received info by phone after 8 weeks waiting on results, was told never have children, call when ready for between hormone. Horrible introduction and I highly recommend it never be considered an appropriate was to receive a diagnosis.
- Very satisfied lots of good info given and resources
- We had 2 MFMs and our first MFM basically told us that she had a 1% chance of survival but was very supportive and told us miracles can happen. We did go to another hospital after referral for shunts for her pleural effusions because our MFM didn't perform that surgery often. The genetic counselor there was very discouraging to me. I was 25 weeks pregnant (diagnosed with hydrops at 11 weeks) and my daughter had been fighting the hydrops for 14 weeks and she still focused on the statistics telling me of her rare chances of survival. I felt like I knew these things already and was holding onto hope that she would make it and it was kinda like she just wanted to focus on the fact that our daughter was going to die. I was really offended by her. The first genetics counselor we saw was precious and gave us info from TSSUS and basically explained the chromosomes of TS 45,x and mosaic Ts.
- In hindsight, it literally amazing how far we've come with so little initial info.

- Our pediatrician didn't suspect it for 7 years. Looking back, it should've been suspected much earlier. Our pediatrician and endocrinologist were accurate and helpful. We were given lots of info and referrals to specialists in a timely manner.
- I was overall very satisfied with how it was explained to us, the only thing not clearly explained is that the tissue that can become cancerous is in the ovaries.
- We were very fortunate to have a healthcare professional who could help us with questions and get her into necessary therapy programs.
- We were given the diagnosis, but nearly no medical information on it. We had to google it, and the wikipedia page made us believe that all children would have all the symptoms. This gave us a lot of unnecessary and harmful worry and stress.
- I felt like I was given very little information and even create a pamphlet for the office to give to other patients.
- Did amnio and in a week they told me my baby was 100% turner syndrome. I am very blessed to have my baby girl
- I never heard of TS before. My doctor was giving me the highlights. While I was crying and trying to process, I kept saying, that doesn't sound so bad.
- Our child was diagnosed prenatally in the third trimester. The MFM doctor provided very little information (stature and intelligence). I was dissatisfied with the experience because of her cold demeanor and her perspective that because most girls with TS have normal intelligence, she could not understand why we would be overwhelmed by the diagnosis. She did well connecting us with testing at birth (echocardiogram, renal ultrasound), but did not provide many resources at the diagnosis beyond basically saying we could Google for information and support groups.
- Minimal info given
- Our daughter was diagnosed at 17 months, after developing hypothyroidism, growth deficiency, swallow diarist, gross and fine motor delays, frequent ear infections. Gastroenterologist refereed is to pediatric geneticist, who performed tests to determine the genetic cause. Was informed of TS diagnosis over the phone and had genetic counseling within 1 week. Highly satisfied with how it went.
- I was given no information except the diagnosis and an explanation that with growth hormone, she could expect to attain an adult height of 5'. I had to find out everything else on my own -- the Turner Syndrome Society of US was absolutely core to my coping with the diagnosis and finding out what I needed to do.
- My least favorite and most favorite person in my prenatal care was the genetics counselors. The first one I had was awful. She was convinced that my daughter didn't have TS and was very dismissive (we had a positive NIPT but refused amnio). The second genetic counselor was amazing - didn't make judgements or predictions. Just stated facts.
- Prenatally we had a NIPT screening and didn't have the diagnosis confirmed until our daughter was 2 weeks old. At that point we got the exact Karyotype and official diagnosis. Seeing a beautiful & overall healthy baby girl when receiving the definite diagnosis made it a lot easier to accept. Prenatally we thought of the worst case scenario. And since we did not want to terminate either way, we chose to hope for the best and not confirm through an amnio.

- After reading through 20 or so negative diagnosis, the physician said she tested positive for TS. He called in residents to view her naked body. He gave a poorly copied booklet with an nonworking website url. It was an extremely painful experience.
- 45x, 46xx, 47xxx. We were not told of how unusual a presentation this was at the time, and only found out about 3 years ago. Since our daughter was normal and healthy except very tiny, we didn't think anything was wrong with her, but as our Pediatric Endocrinologist has told us, we have a very good Pediatrician. An excellent Pediatrician is essential!
- It was over my head, the doctor used all of these terms i had never heard. I had never heard of Turner Syndrome.
- At my 12-week gestational screening, the level of fluid at our daughter's neck was just at the minimum level for concern. Further genetic testing was recommended and bloodwork was done. The testing indicated she likely had Turner syndrome. Initially we had never heard of TS and my main concern was quality of life. The Nurse Practitioner at my OBGYN's office who handled my most recent visit (where excess fluid was noted) called me to share the results. When I asked about quality of life she laughed reassuringly and gave me some examples of women living with TS. It was comforting to hear that my baby had the potential to live a "normal" life. My NP handled the news with care and provided support while answering my questions as thoroughly as possible. I can't imagine this news is easy to relay with any amount of training. I am very satisfied with how it was handled and shared with us.
- It was explained to us with a chromosome mapping without much else. We were told she would need care but it was not elaborated on in much detail. Not verry satisfied.

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 your pregnancy or from raising a child with Turner syndrome? If yes, what information was most useful for you? If no, what information should have been provided that was not?

- No. Alot of information should have been provided that was not. And the information that was provided shouldnt have been.
- I wasn't disappointed with the information as we were relieved to know what was wrong. What to expect was not discussed in great lengths but it would be hard to pinpoint expectations as each girl is so different. We could of eliminated significant frustrations if we would of known sooner!
- Yes. We were told of the issues and their possible solutions.
- No. We weren't given much info and I would have loved to have more. Anything.
- We were not given much information until we saw genetic counseling about 5 months after her birth. I do not feel we were given accurate information or much when we first needed it.
- Fertility stuff should be presented early not waiting until it's too late formiptions
- Yes. The most useful information I received was the honest truth. They didnt really know how each symptom would present during the lifespan. They instead

told me what could happen and what to keep an eye on. They reiterated that some things may never happen. The most common symptoms were discussed. That eased the huge anxiety attack i felt thinking all symptoms would arise immediately upon diagnosis

- It was hard. She was diagnosed when she was 10
- No--health concerns, specialist referrals, support contacts, statistical and factual information on the condition
- Yes considering it was before social platforms & websites had info on them. What I was given most appreciated was an introduction to TS and all aspects social and physical via paper packet. I felt well informed.
- Yes, I feel we were given balanced info and that it's also helpful to talk to other parents and research on your own.
- I was given the very minimum info/ TS and my child would not grow easily. She might struggle with school a bit. I think speaking to others is always helpful so parents should have a way to connect with each other through TSSUS. I had no idea how much therapy we'd need for our child, how much different our school experience would be. How much is need to seek out a TS clinic for the best healthcare.
- Yes, we were given a lot of info about all aspects of TS.
- For me the most useful thing was how knowledgeable and calm all of the doctors are with the diagnosis
- Yes. The availability of various therapy programs.
- Nearly no information was provided to us. It would have been helpful to know that not all children will experience all the symptoms.
- I wish they had done the simple blood test vs a lengthy blood test for other diagnosis
- I was explained so much good information. I am just very thankful my daughters heart is healthy. She goes see's cardiologist in three years just to make sure heart is still healthy.
- The information was mixed. I found a number of doctors who had written papers were very happy to speak to me about their studies. This was enormously helpful and comforting to me. Another thing that was really helpful was meeting a girl with TS. She seemed like a normal teenager, which was very comforting
- No-we received little information. Even having a printout of information from the TSSUS website would have been a helpful start from a reputable resource. It is also helpful to be reminded that as a syndrome, it can manifest in a variety of ways and degrees so not all girls will have the same experience.
- Social development issues
- I was given enough information at the time of diagnosis and our pediatric endocrinologist has kept us abreast of what to expect for the future.
- I received my daughter's diagnosis in May, 1990. I found the Turner Syndrome Society of the US on my own as well and through them most of the info about TS that I needed -- at least as much as was available at the time.
- I often feel like I still know more than the specialists.

- I find the doctor's gave us a worst case scenario. In our case our daughter's karyotype is 45x, 47xxx and we didn't know there are variations between Classic & Mosaic TS. She presents a very mild form. I would have liked to hear that symptoms vary a lot and they could be so mild that they are often unnoticed.
- We went to see a geneticist at a university hospital where she had a complete physical, emotional and family history evaluation. There they explained the extent of her condition. We were told she had a mild form and not to tell anyone. Hmmm? What did that imply?
- Upon diagnosis, there was too much focus on potential medical problems and not enough on learning challenges.
- no; I felt like knowing all around the mental health to the learning to the different lasting effects.
- Yes! However, my daughter has not yet had any issues related to TS which should definitely be taken into account. If she had more medical issues I would likely say that we were not given enough information. Much of that is because I feel like there is a lacking of resources for TS families (likely because it's so rare). We found out our daughter likely had TS prenatally and received confirmation after testing was done when she was born. Based on that, I wish that we had immediately been connected with Early Intervention services. We did not enroll for EI until after my daughter was a year old. The program was amazing - even though my daughter was not showing significant delays, it was really beneficial to have an Interventionist "assigned" to Andy that our family could rely on for questions and help along the way. It was doubly beneficial since we were first time parents and did not know what to expect (even more so related to TS). In SC, children with existing diagnoses can receive Early Intervention services at no cost through BabyNet (via DHHS). If programs like this are available for families in the area in which they are being served, that should definitely be shared and the family should be highly encouraged to connect with that support network.
- No. Resources to reach out for behavioral and social help.

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

- Not bringinf up abortion and the quality of her life on earth would be very low in the first 4 sentences of telling us her diagonis.
- To know sooner about the diagnosis. 4 years to find out and if we would of known sooner, we could of enjoyed the first 4 years instead of struggling as we would of been more equipped.
- Nothing
- I would have liked the providers to be more educated on Turner Syndrome. We were handed a short hand out and told there was a small chance. All through our pregancy "she looked great", even with signs like a horseshoe shaped kidney that is more common in TS. She would have been tested within her first year though due to delayed milestones, growth failure, etc.
- The hospital should have immediately referred us to genetic counseling or at least a social worker to refer us to appropriate clinics.

- Specialist doctors need more information regarding Turner Syndrome. Pediatricians also. So many things were missed
- More doctors need to be aware of turners
- Everything. Face to face opportunity to talk and ask questions, information to take home and read, contact info for support organizations, emotional support
- I wish I would have not received the diagnosis by letter allowing for my mind to wander
- Ultimately, I would say don't focus so much on the negative. Share the facts, if you will, but don't go over it repeatedly unless the parent asks questions. We get the situation we're in, if a parent chooses the fight for their child and not terminate, Hope and support go a long way. I knew I could lose her but that wasn't what was healthy for me to focus on until that time came if it was going to happen.
- Having a professional endocrinologist understand TS, provide info. Much of the info was due to my own searching
- it should've been identified sooner
- I wish that furing the pregnancy that it was more cleatly explained what the amniocentesis could have determined, all I was told is that it would determine the gender as our nipt determined a boy but all of our ultrasounds showed a girl so we ultimately declined doing the amniocentesis
- Nothing. We were relieved to know what we were dealing with and what was available.
- It would have been helpful to know that not all children will experience all the symptoms. Access to the TSSUS website would have been helpful.
- I think she could have been diagnosed earlier in life if pedatrician knew what to look for and had more information about turners
- The genetics department at the hospital was very negative on continuing the pregnancy. Everyone else was very supportive
- A warmer, empathetic approach from the doctor would have helped emotionally with the experience.
- More empathy and reassurance from doctor.
- Getting the results in person rather than over the phone
- If the dr had offered me info on where to get more information and support.
- Softening the blow and telling us she will be a perfectly normal child would've helped calm us a great deal.
- There should have been resources now found on the Turner syndrome Foundation's website to explain the condition, diagnostics, treatment options as well as personal stories of families, women and children living with the disorder.
- I knew there was a problem when I came home to three answering machine messages from three different doctors (not this is the office calling). And the answering machine messages probably dates us!
- We could have been sent to a specialist or a unit dedicated to TS
- Along the way I asked the different providers and networks to be connected with TS families in my community. I was able to speak with one mother by phone but other than that I was not able to make connections. I have shared about my

daughter having TS on my blog and on social media infrequently but have had several parents (even internationally!) reach out to connect. Some have been pregnant moms whose screening had indicated TS. As a woman who has been in that very position, I know how it feels. There's a strong desire to connect with families who are already on the journey. I wanted to hear about their experience and what it is like to raise a daughter with TS. I wanted to hear every detail about how they shared her diagnosis with her, with her caregivers, with her peers, etc. and how they handled the "issues" that come along with it (hormone therapy, injections, ongoing screenings, etc.). Nothing can replace hearing and seeing real life experiences. I wish that providers had more TS-specific information to share with families immediately so they could use those resources as they wish to do independent research (controlling what and how much they consume). Googling on your own is a terrible idea (as we all know!). If parents were given a list of reputable resources (like the TS Foundation & any known support groups), I think that would help families with locating high quality, accurate information which will ultimately help them to be a better informed patient (and to know what questions to ask once they are feeling less overwhelmed by the initial diagnosis).

- Giving us time to process

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

- Listen to the parent. The parent knows best for their child and is with them 24-7 if they feel something is wronf there is a 99% chance there is.
- To be honest I just don't know right now . We just got the diagnosis and still in the hospital from Aug 13. She has be intubated 3 times...to much to type on what's happened till now but we are waiting to have coarc surgery Tuesday. My main fear is the surgery hoping everything goes well but I'm so lost right now with this new diagnosis. I can't help but to be heart broken for her with each and every milestone. All the dr appointmens she will have and not being able to have kids
- None
- People need to be more aware of Turner's
- Be compassionate, my pediatric genetic counselor was very compassionate
- I have found most medical professionals we have had have not been super educated on TS. Most of them know just the same text book info that I can read online. I went to the TS conference and learned so much. It was so nice hearing from docs who had a passion for TS. We haven’t been to a TS clinic though. It would be helpful for me to know if other parents in our area raising girls with TS. That local support for parents and girls could be so helpful esp as the girls grow. If the providers could get consent to share information from their Parents children they see with TS and then connect families I think it would be so helpful for both parties if the families are willing to share info. see with
- Online groups have since become a wealth of info. Drs should at very least provide pertinent info as well as info on TSSUS and local clinics. Not all girls are alike!
- Cardiology care is essential because of increased risk of aortic dissection.

- I wish that our ob and mfm were more familiar with ts as it was never even suggested to us
- The genetics counselor at one of the hospitals offered to give me contact info for a couple of other parents. Talking to them about their
- Having a resource list for parents of TS clinics and professionals with experience working with individuals with TS is helpful as parents try to coordinate care. The clinical care guidelines are an essential resource that all parents should be made aware of at the very least or provided if possible. All parents should also learn more about early intervention of age-appropriate.
- Just after the diagnosis, as I was dressing my daughter, the nurse said to me, "They're really very cute people." It was an awful thing to say and hurt like hell.
- Learn what you need to know, be an advocate for yourself/child and always seek the best possible care/support.
- While I understand that medical professionals rightly focus on medical issues, there needs to be equal attention paid to learning and social challenges these girls will face. It would be nice if the pediatric world would allow a HIPPA waiver so we could try to get the girls to socialize with each other in age brackets. They are each others best sources for answers and information, and meeting in a group environment is very useful for the parents as we share our experiences.
- Having funding provided for people who do not live near the TS specialized hospitals; etc.
- I think the best place for a physician to start is to give the family general (reputable) resources so they can begin researching to the level they feel comfortable with. From there, I think the provider should follow back up with the family to allow them time to ask questions that may have arisen from their research (it's impossible to know what to ask at the time of the diagnosis because it's very likely their first time hearing about TS). After questions have been answered, families should be asked what kind of support would be best for them (Are they strugging with acceptance? Perhaps they need to be connected with a family therapist. Do they want to connect with families of TS girls? A community organization serving TS families may be able to help arrange that. Are they feeling overwhelmed by the future medical appointments/screenings/etc.? They might benefit from joining a local support group for families with children who have special needs and could share advice for navigating the health system). Each family is going to handle a TS diagnosis differently. Additionally, TS can be diagnosed during a range of time periods so I think it's important to assess each situation. There can't be a "one size fits all" approach since the needs will be so varied, but if the physician is well-informed about TS it will certainly help ease the family's anxiety.
- There should be a more immediate way to get help for thoses already dx.

## APPENDIX E – INFORMATIONAL ITEMS IN RANK ORDER

**Table C.1 Rank order of informational item ratings for groups**

| Rank | Prenatal Parent Item | Rating | Prenatal GC Item | Rating | Postnatal Parent Item | Rating | Postnatal GC Item | Rating |
|---|---|---|---|---|---|---|---|---|
| **1** | Coarctation (narrowing) of the aorta and/or left-sided cardiac defects (heart defects) | 2.75 | Need an echocardiogram (ultrasound of the heart) during pregnancy | 3.00 | Discuss impact of hormones to increase stature or treatments that can allow girls with Turner syndrome to attain a normal adult height (over 5 feet) | 2.97 | Women and girls with Turner syndrome typically have normal intelligence | 3.00 |
| **2** | Heart defects and other cardiovascular issues (25%-50%) | 2.69 | Women and girls with Turner syndrome typically have normal intelligence | 2.96 | Specialist referral(s) | 2.94 | Short stature (95%-100%) | 2.94 |
| **3** | Caused by the partial or complete loss of the second X-chromosome in females | 2.69 | Heart defects and other cardiovascular issues (25%-50%) | 2.90 | May have some health conditions but not all | 2.87 | Specialist referral(s) | 2.91 |

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| **4** | Local Turner syndrome support group(s) | 2.69 | May need an evaluation by a pediatric heart specialist | 2.87 | Girls with Turner syndrome need hormones to start or complete puberty | 2.87 | Heart defects and other cardiovascular issues (25%-50%) | 2.88 |
| **5** | Discuss value of ultrasound in detecting prenatal conditions | 2.67 | May have some health conditions but not all | 2.84 | Short stature (95%-100%) | 2.84 | Women with Turner syndrome usually live long lives | 2.85 |
| **6** | Specialist referral(s) | 2.67 | 99% risk for fetal demise when prenatally diagnosed | 2.83 | Discuss available Turner syndrome clinics | 2.84 | National advocacy organizations & websites | 2.85 |
| **7** | May need an evaluation by a pediatric heart specialist | 2.58 | Prenatal diagnosis can only be confirmed by chromosome analysis via amniocentesis or CVS | 2.82 | Women and girls with Turner syndrome typically have normal intelligence | 2.84 | Discuss available Turner syndrome clinics | 2.82 |
| **8** | Need an echocardiogram (ultrasound of the heart) during pregnancy | 2.54 | Caused by the partial or complete loss of the second X-chromosome in females | 2.80 | Women with Turner syndrome usually live long lives | 2.81 | More like other children than different | 2.82 |
| **9** | Discuss available Turner syndrome clinics | 2.54 | Chance to reoccur in future pregnancies | 2.78 | Heart defects and other cardiovascular issues (25%-50%) | 2.77 | May have some health conditions but not all | 2.79 |

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| **10** | Contact with families raising a child with Turner syndrome | 2.54 | Women with Turner syndrome usually live long lives | 2.76 | Primary ovarian failure (reduced function of ovaries before age 40) (90%) | 2.74 | Early intervention services | 2.79 |
| **11** | Fact sheets/brochures | 2.50 | Discuss value of ultrasound in detecting prenatal conditions | 2.75 | Finishing high school | 2.74 | Reduced fertility | 2.79 |
| **12** | Cystic hygroma (fluid-filled sack on the back of the fetal neck) | 2.46 | Primary ovarian failure (reduced function of ovaries before age 40) (90%) | 2.74 | Having friends and meaningful relationships | 2.71 | Local Turner syndrome support group(s) | 2.79 |
| **13** | Women with Turner syndrome usually live long lives | 2.46 | Screening results can be obtained via noninvasive prenatal screening (NIPS/cell-free DNA) | 2.74 | More like other children than different | 2.71 | Caused by the partial or complete loss of the second X-chromosome in females | 2.76 |
| **14** | May have some health conditions but not all | 2.46 | Coarctation (narrowing) of the aorta and/or left-sided cardiac defects (heart defects) | 2.71 | Caused by the partial or complete loss of the second X-chromosome in females | 2.69 | Online support groups/social media platforms | 2.76 |

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| **15** | Screening results can be obtained via noninvasive prenatal screening (NIPS/cell-free DNA) | 2.46 | More like other children than different | 2.70 | Reduced fertility | 2.68 | Discuss impact of hormones to increase stature or treatments that can allow girls with Turner syndrome to attain a normal adult height (over 5 feet) | 2.70 |
| **16** | National advocacy organizations & websites | 2.46 | Short stature (95%-100%) | 2.66 | Local Turner syndrome support group(s) | 2.68 | Girls with Turner syndrome need hormones to start or complete puberty | 2.70 |
| **17** | Special education supports and services | 2.42 | Local Turner syndrome support group(s) | 2.64 | Excel at verbal skills compared to general US female population | 2.67 | Reproductive capability of a woman with Turner syndrome | 2.70 |
| **18** | Primary ovarian failure (reduced function of ovaries before age 40) (90%) | 2.38 | Discuss available Turner syndrome clinics | 2.62 | Chance to reoccur in future pregnancies | 2.67 | Printed/written material | 2.70 |
| **19** | Chance to reoccur in future pregnancies | 2.38 | Early intervention services | 2.60 | Reproductive options for a woman with Turner syndrome (pregnancy, surrogacy, adoption) | 2.67 | Primary ovarian failure (reduced function of ovaries before age 40) (90%) | 2.67 |

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| **20** | Printed/written material | 2.38 | Reduced fertility | 2.60 | Frequent ear infections (60%) | 2.65 | Intellectual disability (~10%) | 2.67 |
| **21** | Increased nuchal translucency (excess fluid on the back of the head or neck) in first trimester | 2.31 | Reproductive capability of a woman with Turner syndrome | 2.60 | Living independently | 2.63 | Chance to reoccur in future pregnancies | 2.67 |
| **22** | Pericardial effusion (heart partially or completely surrounded by fluid) | 2.31 | National advocacy organizations & websites | 2.60 | National advocacy organizations & websites | 2.63 | Vision problems (20%) | 2.64 |
| **23** | Growth restriction | 2.31 | No significant increasing incidence with increasing maternal age | 2.56 | Possible chromosomal /genetic causes of Turner syndrome: 45,X (40%-50%), 45,X/46,XX (15%-25%), Other (25%-45%) | 2.62 | Increased risk of attention-deficit/hyperactivity disorder (ADHD) (25%) | 2.64 |
| **24** | Discuss impact of hormones to increase stature or treatments that can allow girls with Turner syndrome to attain a normal adult height (over 5 feet) | 2.31 | Intellectual disability (~10%) | 2.52 | Early intervention services | 2.61 | Hearing loss (30%) | 2.61 |

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|---|---|---|---|---|---|---|---|---|
| **25** | Girls with Turner syndrome need hormones to start or complete puberty | 2.31 | Printed/written material | 2.52 | Clumsiness and delayed motor milestones | 2.60 | Inclusion in regular classes | 2.61 |
| **26** | Prenatal diagnosis can only be confirmed by chromosome analysis via amniocentesis or CVS | 2.31 | Girls with Turner syndrome need hormones to start or complete puberty | 2.50 | Reproductive capability of a woman with Turner syndrome | 2.60 | Having friends and meaningful relationships | 2.61 |
| **27** | Online support groups/social media platforms | 2.31 | Specialist referral(s) | 2.50 | May need an evaluation by a pediatric heart specialist | 2.59 | Kidney issues (10%-15%) | 2.55 |
| **28** | Counselor or family therapist referral(s) | 2.31 | Cystic hygroma (fluid-filled sack on the back of the fetal neck) | 2.46 | Growth restriction | 2.59 | Poor performance in mathematics (dyscalculia) (50%-75%) | 2.55 |
| **29** | Short stature (95%-100%) | 2.23 | Online support groups/social media platforms | 2.46 | Girls with Turner syndrome may need more support to develop social skills | 2.58 | May need an evaluation by a pediatric heart specialist | 2.54 |
| **30** | Hypertension (high blood pressure) (50%) | 2.23 | Inclusion in regular classes | 2.43 | More likely to earn a baccalaureate degree or higher than the general US female population | 2.58 | Special education services | 2.52 |

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|---|---|---|---|---|---|---|---|---|
| **31** | Frequent ear infections (60%) | 2.23 | Pregnancy termination resources | 2.43 | Special education supports and services | 2.58 | Benefit of psychological therapy and support | 2.52 |
| **32** | Kidney issues (10%-15%) | 2.23 | Renal anomalies (kidney problems) | 2.40 | Coarctation (narrowing) of the aorta and/or left-sided cardiac defects (heart defects) | 2.57 | Reproductive options for a woman with Turner syndrome (pregnancy, surrogacy, adoption) | 2.52 |
| **33** | Finishing high school | 2.23 | Living independently | 2.40 | Orthopedic (bone, muscle, joint) problems [scoliosis (10%), decreased bone mineral content (50%-80%), etc.] | 2.55 | Special education supports and services | 2.52 |
| **34** | Reproductive capability of a woman with Turner syndrome | 2.23 | Discuss impact of hormones to increase stature or treatments that can allow girls with Turner syndrome to attain a normal adult height (over 5 feet) | 2.39 | Strengths in various aspects of oral and written communication (speaking and reading are commonly strengths for women with Turner syndrome) | 2.55 | Hypothyroidism (15%-30%) | 2.48 |
| **35** | Women and girls with Turner syndrome typically have normal intelligence | 2.17 | Having friends and meaningful relationships | 2.38 | Alternate ways to become a parent when unable to conceive | 2.55 | Hypertension (high blood pressure) (50%) | 2.45 |

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|---|---|---|---|---|---|---|---|---|
| **36** | Books | 2.17 | Poor performance in mathematics (dyscalculia) (50%-75%) | 2.36 | Printed/written material | 2.53 | Girls with Turner syndrome may need more support to develop social skills | 2.45 |
| **37** | Renal anomalies (kidney problems) | 2.15 | Fact sheets/brochures | 2.34 | Hearing loss (30%) | 2.52 | Frequent ear infections (60%) | 2.42 |
| **38** | Lymphedema (swelling) in hands and feet (25%) | 2.15 | Girls with Turner syndrome may need more support to develop social skills | 2.28 | Lack of working memory which may cause difficulty with multi-tasking, mental calculations, and holding information "in a mind's eye" | 2.52 | Possible ability to conceive through reproductive assistive technology after a thorough medical examination | 2.42 |
| **39** | High blood pressure (50%) | 2.15 | Possible ability to conceive through reproductive assistive technology after a thorough medical examination | 2.28 | Inclusion in regular classes | 2.52 | Fact sheets/brochures | 2.42 |
| **40** | Early intervention services | 2.15 | Increased risk of attention-deficit/hyperactivity disorder (ADHD) (25%) | 2.26 | High blood pressure (50%) | 2.50 | High blood pressure (50%) | 2.41 |

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| **41** | Benefit of psychological therapy and support | 2.15 | Reproductive options for a woman with Turner syndrome (pregnancy, surrogacy, adoption) | 2.26 | Hypertension (high blood pressure) (50%) | 2.48 | Strengths in various aspects of oral and written communication (speaking and reading are commonly strengths for women with Turner syndrome) | 2.39 |
| **42** | More like other children than different | 2.15 | Increased nuchal translucency (excess fluid on the back of the head or neck) in first trimester | 2.25 | Kidney issues (10%-15%) | 2.48 | Clumsiness and delayed motor milestones | 2.39 |
| **43** | Pleural effusion ("water in the lungs") | 2.08 | Possible chromosomal/genetic causes of Turner syndrome: 45,X (40%-50%), 45,X/46,XX (15%-25%), Other (25%-45%) | 2.24 | Poor performance in mathematics (dyscalculia) (50%-75%) | 2.48 | Participating in community activities (clubs, hobbies, sports, volunteer work, etc.) | 2.39 |
| **44** | Hearing loss (30%) | 2.08 | Hearing loss (30%) | 2.20 | Difficulty with problem-solving | 2.48 | Living independently | 2.39 |

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| **45** | Orthopedic (bone, muscle, joint) problems [scoliosis (10%), decreased bone mineral content (50%-80%), etc.] | 2.08 | Alternate ways to become a parent when unable to conceive | 2.20 | Prenatal diagnosis can only be confirmed by chromosome analysis via amniocentesis or CVS | 2.48 | Possible chromosomal/genetic causes of Turner syndrome: 45,X (40%-50%), 45,X/46,XX (15%-25%), Other (25%-45%) | 2.39 |
| **46** | Possible chromosomal /genetic causes of Turner syndrome: 45,X (40%-50%), 45,X/46,XX (15%-25%), Other (25%-45%) | 2.08 | Vision problems (20%) | 2.14 | Contact with families raising a child with Turner syndrome | 2.48 | Glucose intolerance (15%-50%) | 2.36 |
| **47** | Reproductive options for a woman with Turner syndrome (pregnancy, surrogacy, adoption) | 2.08 | Having intimate relationships | 2.14 | Fact sheets/brochures | 2.47 | Orthopedic (bone, muscle, joint) problems [scoliosis (10%), decreased bone mineral content (50%-80%), etc.] | 2.36 |
| **48** | 99% risk for fetal demise when prenatally diagnosed | 2.00 | Growth restriction | 2.12 | Discuss value of ultrasound in detecting prenatal conditions | 2.45 | Having intimate relationships | 2.36 |

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| **49** | Glucose intolerance (15%-50%) | 2.00 | Special education services | 2.12 | Hypothyroidism (15%-30%) | 2.45 | Alternate ways to become a parent when unable to conceive | 2.33 |
| **50** | Hypothyroidism (15%-30%) | 2.00 | Kidney issues (10%-15%) | 2.08 | Special education services | 2.45 | Contact with families raising a child with Turner syndrome | 2.33 |
| **51** | Reduced fertility | 2.00 | Pericardial effusion (heart partially or completely surrounded by fluid) | 2.06 | Incidence (1/2500 females) | 2.45 | Lymphedema (swelling) in hands and feet (25%) | 2.28 |
| **52** | Incidence (1/2500 females) | 2.00 | Benefit of psychological therapy and support | 2.06 | Renal anomalies (kidney problems) | 2.43 | Renal anomalies (kidney problems) | 2.23 |
| **53** | Oligohydramnios (decrease of amniotic fluid volume in pregnancy) | 1.92 | Finishing high school | 2.06 | Participating in community activities (clubs, hobbies, sports, volunteer work, etc.) | 2.43 | Difficulty with problem-solving | 2.15 |
| **54** | Only discuss prenatal issues that are actually confirmed via ultrasound | 1.92 | Strengths in various aspects of oral and written communication (speaking and reading are commonly strengths for women with Turner syndrome) | 2.04 | No significant increasing incidence with increasing maternal age | 2.43 | Lack of working memory which may cause difficulty with multi-tasking, mental calculations, and holding information "in a mind's eye" | 2.12 |

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| **55** | Impact on other siblings – more compassionate & caring | 1.92 | Pleural effusion ("water in the lungs") | 2.02 | Online support groups/social media platforms | 2.43 | Inefficiency (e.g., slow and more effortful) when learning through visual means (e.g., pictures and diagrams) | 2.12 |
| **56** | Having friends and meaningful relationships | 1.92 | Hypertension (high blood pressure) (50%) | 2.00 | Vision problems (20%) | 2.42 | Finishing high school | 2.12 |
| **57** | Life expectancy (may be reduced up to 13 years) | 1.92 | Special education supports and services | 2.00 | Difficulty identifying facial emotions | 2.39 | Life expectancy (may be reduced up to 13 years) | 2.12 |
| **58** | Photographs of children with Turner syndrome | 1.92 | Polyhydramnios (increase of amniotic fluid volume in pregnancy) | 1.96 | Having intimate relationships | 2.39 | Excel at verbal skills compared to general US female population | 2.12 |
| **59** | Vision problems (20%) | 1.85 | Oligohydramnios (decrease of amniotic fluid volume in pregnancy) | 1.96 | Financial parental impact – More | 2.37 | Coarctation (narrowing) of the aorta and/or left-sided cardiac defects (heart defects) | 2.11 |
| **60** | Impact on parental marriage – strengthens relationship | 1.85 | Contact with families raising a child with Turner syndrome | 1.96 | Inefficiency (e.g., slow and more effortful) when learning through visual means (e.g., pictures and diagrams) | 2.37 | More likely to earn a baccalaureate degree or higher than the general US female population | 2.06 |

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| **61** | Financial parental impact – More | 1.85 | Clumsiness and delayed motor milestones | 1.94 | Glucose intolerance (15%-50%) | 2.35 | No significant increasing incidence with increasing maternal age | 2.06 |
| **62** | Clumsiness and delayed motor milestones | 1.85 | Participating in community activities (clubs, hobbies, sports, volunteer work, etc.) | 1.92 | Intellectual disability (~10%) | 2.35 | Counselor or family therapist referral(s) | 2.00 |
| **63** | Girls with Turner syndrome may need more support to develop social skills | 1.85 | Life expectancy (may be reduced up to 13 years) | 1.92 | Increased risk of attention-deficit/hyperactivity disorder (ADHD) (25%) | 2.35 | Difficulty initiating or maintaining relationships | 1.94 |
| **64** | Inclusion in regular classes | 1.85 | Counselor or family therapist referral(s) | 1.90 | More likely to be employed than the general US female population | 2.35 | More likely to be employed than the general US female population | 1.94 |
| **65** | More likely to earn a baccalaureate degree or higher than the general US female population | 1.85 | Hypothyroidism (15%-30%) | 1.88 | Impact on other siblings – more compassionate & caring | 2.33 | Books | 1.94 |
| **66** | More likely to be employed than the general US female population | 1.85 | High blood pressure (50%) | 1.88 | Benefit of psychological therapy and support | 2.33 | Difficulty identifying facial emotions | 1.88 |

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| **67** | Having intimate relationships | 1.85 | Incidence (1/2500 females) | 1.88 | Lymphedema (swelling) in hands and feet (25%) | 2.32 | Impact on other siblings – more compassion ate & caring | 1.82 |
| **68** | Excel at verbal skills compared to general US female population | 1.85 | Orthopedic (bone, muscle, joint) problems [scoliosis (10%), decreased bone mineral content (50%-80%), etc.] | 1.84 | Possible ability to conceive through reproductive assistive technology after a thorough medical examination | 2.32 | Photographs of children with Turner syndrome | 1.82 |
| **69** | Polyhydramnios (increase of amniotic fluid volume in pregnancy) | 1.77 | Lack of working memory which may cause difficulty with multi-tasking, mental calculations, and holding information "in a mind's eye" | 1.84 | 99% risk for fetal demise when prenatally diagnosed | 2.31 | Impact on parental marriage – strains relationship | 1.79 |
| **70** | Impact on other relationships – supportive & welcoming | 1.77 | Difficulty with problem-solving | 1.84 | Difficulty initiating or maintaining relationships | 2.27 | Incidence (1/2500 females) | 1.79 |
| **71** | Increased risk of attention-deficit/hyper activity disorder (ADHD) (25%) | 1.77 | Lymphedema (swelling) in hands and feet (25%) | 1.78 | Counselor or family therapist referral(s) | 2.26 | Impact on other siblings – less attention & resentful | 1.73 |

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| **72** | Lack of working memory which may cause difficulty with multi-tasking, mental calculations, and holding information "in a mind's eye" | 1.77 | More likely to earn a baccalaureate degree or higher than the general US female population | 1.78 | Impact on other siblings – less attention & resentful | 2.23 | Impact on parental marriage – strengthens relationship | 1.73 |
| **73** | Special education services | 1.77 | Only discuss prenatal issues that are actually confirmed via ultrasound | 1.75 | Screening results can be obtained via noninvasive prenatal screening (NIPS/cell-free DNA) | 2.23 | Pastoral counseling referral(s) | 1.73 |
| **74** | Alternate ways to become a parent when unable to conceive | 1.77 | Impact on other siblings – more compassionate & caring | 1.74 | Books | 2.20 | Screening results can be obtained via noninvasive prenatal screening (NIPS/cell-free DNA) | 1.58 |
| **75** | Living independently | 1.77 | Impact on parental marriage – strains relationship | 1.74 | Photographs of children with Turner syndrome | 2.17 | Growth restriction | 1.57 |
| **76** | Impact on other siblings – less attention & resentful | 1.69 | Glucose intolerance (15%-50%) | 1.72 | Time commitment of parent – More | 2.16 | Impact on grandparents – supportive & welcoming | 1.55 |

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| **77** | Time commitment of parent – More | 1.69 | Frequent ear infections (60%) | 1.72 | Need an echocardiogram (ultrasound of the heart) during pregnancy | 2.07 | Impact on other relationships – supportive & welcoming | 1.55 |
| **78** | Intellectual disability (~10%) | 1.69 | Excel at verbal skills compared to general US female population | 1.72 | Pericardial effusion (heart partially or completely surrounded by fluid) | 2.03 | Impact on other relationships – lose social circle | 1.55 |
| **79** | Strengths in various aspects of oral and written communication (speaking and reading are commonly strengths for women with Turner syndrome) | 1.69 | Books | 1.72 | Impact on parental marriage – strengthens relationship | 2.03 | Time commitment of parent – More | 1.55 |
| **80** | Possible ability to conceive through reproductive assistive technology after a thorough medical examination | 1.69 | Inefficiency (e.g., slow and more effortful) when learning through visual means (e.g., pictures and diagrams) | 1.70 | Time commitment of parent – No Difference | 2.00 | Less likely to marry than the general US female population | 1.52 |

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| **81** | Impact on grandparents – supportive & welcoming | 1.62 | Impact on parental marriage – strengthens relationship | 1.68 | Only discuss prenatal issues that are actually confirmed via ultrasound | 1.97 | Prenatal diagnosis can only be confirmed by chromosome analysis via amniocentesis or CVS | 1.52 |
| **82** | Difficulty with problem-solving | 1.62 | More likely to be employed than the general US female population | 1.68 | Impact on parental marriage – strains relationship | 1.94 | Only discuss prenatal issues that are actually confirmed via ultrasound | 1.49 |
| **83** | Difficulty identifying facial emotions | 1.62 | Photographs of children with Turner syndrome | 1.68 | Impact on grandparents – supportive & welcoming | 1.93 | Financial parental impact – More | 1.48 |
| **84** | Difficulty initiating or maintaining relationships | 1.62 | Impact on other siblings – less attention & resentful | 1.66 | Life expectancy (may be reduced up to 13 years) | 1.93 | Brachycephaly (broad, short skull) | 1.46 |
| **85** | Participating in community activities (clubs, hobbies, sports, volunteer work, etc.) | 1.62 | Financial parental impact – More | 1.56 | Financial parental impact – No difference | 1.85 | Impact on grandparents – limited interaction | 1.45 |
| **86** | No significant increasing incidence with increasing maternal age | 1.62 | Time commitment of parent – More | 1.56 | Less likely to marry than the general US female population | 1.84 | Impact on extended family members – supportive & welcoming | 1.42 |

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| **87** | Poor performance in mathematics (dyscalculia) (50%-75%) | 1.58 | Pastoral counseling referral(s) | 1.50 | Impact on other relationships – supportive & welcoming | 1.83 | Impact on extended family members – limited interaction | 1.42 |
| **88** | Impact on extended family members – supportive & welcoming | 1.54 | Impact on other relationships – supportive & welcoming | 1.46 | Impact on extended family members – supportive & welcoming | 1.81 | Time commitment of parent – No Difference | 1.39 |
| **89** | Inefficiency (e.g., slow and more effortful) when learning through visual means (e.g., pictures and diagrams) | 1.54 | Difficulty identifying facial emotions | 1.44 | Impact on other relationships – lose social circle | 1.73 | 99% risk for fetal demise when prenatally diagnosed | 1.38 |
| **90** | Brachycephaly (broad, short skull) | 1.46 | Difficulty initiating or maintaining relationships | 1.42 | Oligohydramnios (decrease of amniotic fluid volume in pregnancy) | 1.72 | Discuss value of ultrasound in detecting prenatal conditions | 1.37 |
| **91** | Alternative/nonconventional therapies | 1.42 | Impact on other relationships – lose social circle | 1.41 | Impact on grandparents – limited interaction | 1.68 | Financial parental impact – No difference | 1.36 |
| **92** | Impact on parental marriage – strains relationship | 1.38 | Alternative/ nonconventional therapies | 1.39 | Pleural effusion ("water in the lungs") | 1.66 | Need an echocardiogram (ultrasound of the heart) during pregnancy | 1.34 |
| **93** | Impact on other relationships – lose social circle | 1.38 | Brachycephaly (broad, short skull) | 1.35 | Brachycephaly (broad, short skull) | 1.66 | Alternative/ nonconventional therapies | 1.33 |

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| **94** | Time commitment of parent – No Difference | 1.38 | Impact on grandparents – supportive & welcoming | 1.34 | Alternative/nonconventional therapies | 1.63 | Cystic hygroma (fluid-filled sack on the back of the fetal neck) | 1.32 |
| **95** | Less likely to marry than the general US female population | 1.38 | Impact on extended family members – supportive & welcoming | 1.32 | Cystic hygroma (fluid-filled sack on the back of the fetal neck) | 1.62 | Increased nuchal translucency (excess fluid on the back of the head or neck) in first trimester | 1.29 |
| **96** | Impact on grandparents – limited interaction | 1.31 | Impact on grandparents – limited interaction | 1.30 | Impact on extended family members – limited interaction | 1.61 | Pleural effusion ("water in the lungs") | 1.26 |
| **97** | Impact on extended family members – limited interaction | 1.31 | Time commitment of parent – No Difference | 1.30 | Polyhydramnios (increase of amniotic fluid volume in pregnancy) | 1.57 | Pericardial effusion (heart partially or completely surrounded by fluid) | 1.26 |
| **98** | Financial parental impact – No difference | 1.31 | Financial parental impact – No difference | 1.26 | Pastoral counseling referral(s) | 1.48 | Polyhydramnios (increase of amniotic fluid volume in pregnancy) | 1.26 |
| **99** | Pastoral counseling referral(s) | 1.08 | Impact on extended family members – limited interaction | 1.24 | Pregnancy termination resources | 1.47 | Oligohydramnios (decrease of amniotic fluid volume in pregnancy) | 1.26 |

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| **100** | Pregnancy termination resources | 0.83 | Less likely to marry than the general US female population | 1.20 | Increased nuchal translucency (excess fluid on the back of the head or neck) in first trimester | 1.38 | Pregnancy termination resources | 1.03 |

## APPENDIX F – SIGNIFICANT DIFFERENCES BETWEEN GROUPS

**Table C.2 Significant differences between item ratings for groups**

| Prenatal Physical Features and Health Complications of Turner Syndrome | Group | Rating | Group | Rating | Significance |
|---|---|---|---|---|---|
| Increased nuchal translucency in first trimester | Prenatal GC | 2.25 | Postnatal GC | 1.29 | < **.0001** |
| | Prenatal GC | 2.25 | Prenatal Parent | 2.31 | .808 |
| | Postnatal GC | 1.29 | Postnatal Parent | 1.38 | .715 |
| | Prenatal Parent | 2.31 | Postnatal Parent | 1.38 | .050 |
| Pleural effusion | Prenatal GC | 2.02 | Postnatal GC | 1.26 | < **.0001** |
| | Prenatal GC | 2.02 | Prenatal Parent | 2.08 | .836 |
| | Postnatal GC | 1.26 | Postnatal Parent | 1.66 | .122 |
| | Prenatal Parent | 2.08 | Postnatal Parent | 1.66 | .460 |
| Pericardial effusion | Prenatal GC | 2.06 | Postnatal GC | 1.26 | < **.0001** |
| | Prenatal GC | 2.06 | Prenatal Parent | 2.31 | .342 |
| | Postnatal GC | 1.26 | Postnatal Parent | 2.03 | **.002** |
| | Prenatal Parent | 2.31 | Postnatal Parent | 2.03 | .613 |
| Cystic hygroma | Prenatal GC | 2.46 | Postnatal GC | 1.32 | < **.0001** |
| | Prenatal GC | 2.46 | Prenatal Parent | 2.46 | .989 |
| | Postnatal GC | 1.32 | Postnatal Parent | 1.62 | .242 |
| | Prenatal Parent | 2.46 | Postnatal Parent | 1.62 | .064 |
| Coarctation of the aorta and/or left-sided cardiac defects | Prenatal GC | 2.71 | Postnatal GC | 2.11 | < **.0001** |
| | Prenatal GC | 2.71 | Prenatal Parent | 2.75 | .779 |
| | Postnatal GC | 2.11 | Postnatal Parent | 2.57 | .060 |
| | Prenatal Parent | 2.75 | Postnatal Parent | 2.57 | .604 |
| Need an echocardiogram during pregnancy | Prenatal GC | 3.00 | Postnatal GC | 1.34 | < **.0001** |
| | Prenatal GC | 3.00 | Prenatal Parent | 2.54 | **.005** |
| | Postnatal GC | 1.34 | Postnatal Parent | 2.07 | **.004** |
| | Prenatal Parent | 2.54 | Postnatal Parent | 2.07 | .081 |
| May need an evaluation by a pediatric heart specialist | Prenatal GC | 2.87 | Postnatal GC | 2.54 | **.020** |
| | Prenatal GC | 2.87 | Prenatal Parent | 2.58 | .131 |
| | Postnatal GC | 2.54 | Postnatal Parent | 2.59 | .838 |
| | Prenatal Parent | 2.58 | Postnatal Parent | 2.59 | .900 |
| Brachycephaly | Prenatal GC | 1.35 | Postnatal GC | 1.46 | .446 |
| | Prenatal GC | 1.35 | Prenatal Parent | 1.46 | .608 |
| | Postnatal GC | 1.46 | Postnatal Parent | 1.66 | .398 |

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| | Prenatal Parent | 1.46 | Postnatal Parent | 1.66 | .399 |
| Renal anomalies | Prenatal GC | 2.40 | Postnatal GC | 2.23 | .297 |
| | Prenatal GC | 2.40 | Prenatal Parent | 2.15 | .306 |
| | Postnatal GC | 2.23 | Postnatal Parent | 2.43 | .366 |
| | Prenatal Parent | 2.15 | Postnatal Parent | 2.43 | .325 |
| Polyhydramnios | Prenatal GC | 1.96 | Postnatal GC | 1.26 | **< .0001** |
| | Prenatal GC | 1.96 | Prenatal Parent | 1.77 | .477 |
| | Postnatal GC | 1.26 | Postnatal Parent | 1.57 | .157 |
| | Prenatal Parent | 1.77 | Postnatal Parent | 1.57 | .814 |
| Oligohydramnios | Prenatal GC | 1.96 | Postnatal GC | 1.26 | **< .0001** |
| | Prenatal GC | 1.96 | Prenatal Parent | 1.92 | .895 |
| | Postnatal GC | 1.26 | Postnatal Parent | 1.72 | **.040** |
| | Prenatal Parent | 1.92 | Postnatal Parent | 1.72 | .791 |
| Growth restriction | Prenatal GC | 2.12 | Postnatal GC | 1.57 | **.001** |
| | Prenatal GC | 2.12 | Prenatal Parent | 2.31 | .458 |
| | Postnatal GC | 1.57 | Postnatal Parent | 2.59 | **< .0001** |
| | Prenatal Parent | 2.31 | Postnatal Parent | 2.59 | .296 |
| 99% risk for fetal demise when prenatally diagnosed | Prenatal GC | 2.83 | Postnatal GC | 1.38 | **< .0001** |
| | Prenatal GC | 2.83 | Prenatal Parent | 2.00 | **< .0001** |
| | Postnatal GC | 1.38 | Postnatal Parent | 2.31 | **< .0001** |
| | Prenatal Parent | 2.00 | Postnatal Parent | 2.31 | .448 |
| Only discuss prenatal issues that are actually confirmed via ultrasound | Prenatal GC | 1.75 | Postnatal GC | 1.49 | .170 |
| | Prenatal GC | 1.75 | Prenatal Parent | 1.92 | .578 |
| | Postnatal GC | 1.49 | Postnatal Parent | 1.97 | .068 |
| | Prenatal Parent | 1.92 | Postnatal Parent | 1.97 | .763 |
| Discuss value of ultrasound in detecting prenatal conditions | Prenatal GC | 2.75 | Postnatal GC | 1.37 | **< .0001** |
| | Prenatal GC | 2.75 | Prenatal Parent | 2.67 | .567 |
| | Postnatal GC | 1.37 | Postnatal Parent | 2.45 | **< .0001** |
| | Prenatal Parent | 2.67 | Postnatal Parent | 2.45 | .341 |
| **Postnatal Physical Features and Health Complications of Turner Syndrome** | **Group** | **Rating** | **Group** | **Rating** | **Sig < 0.05** |
| Women with Turner syndrome usually live long lives | Prenatal GC | 2.76 | Postnatal GC | 2.85 | .509 |
| | Prenatal GC | 2.76 | Prenatal Parent | 2.46 | .231 |
| | Postnatal GC | 2.85 | Postnatal Parent | 2.81 | .750 |
| | Prenatal Parent | 2.46 | Postnatal Parent | 2.81 | .123 |
| Short stature (95%-100%) | Prenatal GC | 2.66 | Postnatal GC | 2.94 | **.008** |
| | Prenatal GC | 2.66 | Prenatal Parent | 2.23 | **.030** |
| | Postnatal GC | 2.94 | Postnatal Parent | 2.84 | .269 |
| | Prenatal Parent | 2.23 | Postnatal Parent | 2.84 | **.002** |
| Heart defects and other cardiovascular issues (25%-50%) | Prenatal GC | 2.90 | Postnatal GC | 2.88 | .789 |
| | Prenatal GC | 2.90 | Prenatal Parent | 2.69 | .158 |
| | Postnatal GC | 2.88 | Postnatal Parent | 2.77 | .372 |

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| | Prenatal Parent | 2.69 | Postnatal Parent | 2.77 | .665 |
| Hearing loss (30%) | Prenatal GC | 2.20 | Postnatal GC | 2.61 | **.005** |
| | Prenatal GC | 2.20 | Prenatal Parent | 2.08 | .592 |
| | Postnatal GC | 2.61 | Postnatal Parent | 2.52 | .578 |
| | Prenatal Parent | 2.08 | Postnatal Parent | 2.52 | .062 |
| Vision problems (20%) | Prenatal GC | 2.14 | Postnatal GC | 2.64 | **.001** |
| | Prenatal GC | 2.14 | Prenatal Parent | 1.85 | .222 |
| | Postnatal GC | 2.64 | Postnatal Parent | 2.42 | .178 |
| | Prenatal Parent | 1.85 | Postnatal Parent | 2.42 | **.048** |
| Hypertension (50%) | Prenatal GC | 2.00 | Postnatal GC | 2.45 | **.009** |
| | Prenatal GC | 2.00 | Prenatal Parent | 2.23 | .365 |
| | Postnatal GC | 2.45 | Postnatal Parent | 2.48 | .878 |
| | Prenatal Parent | 2.23 | Postnatal Parent | 2.48 | .277 |
| Glucose intolerance (15%-50%) | Prenatal GC | 1.72 | Postnatal GC | 2.36 | < **.0001** |
| | Prenatal GC | 1.72 | Prenatal Parent | 2.00 | .205 |
| | Postnatal GC | 2.36 | Postnatal Parent | 2.35 | .964 |
| | Prenatal Parent | 2.00 | Postnatal Parent | 2.35 | .127 |
| Frequent ear infections (60%) | Prenatal GC | 1.72 | Postnatal GC | 2.42 | < **.0001** |
| | Prenatal GC | 1.72 | Prenatal Parent | 2.23 | **.038** |
| | Postnatal GC | 2.42 | Postnatal Parent | 2.65 | .171 |
| | Prenatal Parent | 2.23 | Postnatal Parent | 2.65 | .066 |
| Lymphedema in hands and feet (25%) | Prenatal GC | 1.78 | Postnatal GC | 2.28 | **.005** |
| | Prenatal GC | 1.78 | Prenatal Parent | 2.15 | .154 |
| | Postnatal GC | 2.28 | Postnatal Parent | 2.32 | .830 |
| | Prenatal Parent | 2.15 | Postnatal Parent | 2.32 | .413 |
| Hypothyroidism (15%-30%) | Prenatal GC | 1.88 | Postnatal GC | 2.48 | < **.0001** |
| | Prenatal GC | 1.88 | Prenatal Parent | 2.00 | .592 |
| | Postnatal GC | 2.48 | Postnatal Parent | 2.45 | .862 |
| | Prenatal Parent | 2.00 | Postnatal Parent | 2.45 | .067 |
| Orthopedic problems [scoliosis (10%), decreased bone mineral content (50%-80%), etc.)] | Prenatal GC | 1.84 | Postnatal GC | 2.36 | **.002** |
| | Prenatal GC | 1.84 | Prenatal Parent | 2.08 | .336 |
| | Postnatal GC | 2.36 | Postnatal Parent | 2.55 | .303 |
| | Prenatal Parent | 2.08 | Postnatal Parent | 2.55 | **.040** |
| Kidney issues (10%-15%) | Prenatal GC | 2.08 | Postnatal GC | 2.55 | **.004** |
| | Prenatal GC | 2.08 | Prenatal Parent | 2.23 | .531 |
| | Postnatal GC | 2.55 | Postnatal Parent | 2.48 | .724 |
| | Prenatal Parent | 2.23 | Postnatal Parent | 2.48 | .243 |
| High blood pressure (50%) | Prenatal GC | 1.88 | Postnatal GC | 2.41 | **.003** |
| | Prenatal GC | 1.88 | Prenatal Parent | 2.15 | .275 |
| | Postnatal GC | 2.41 | Postnatal Parent | 2.50 | .611 |
| | Prenatal Parent | 2.15 | Postnatal Parent | 2.50 | .126 |
| Primary ovarian failure (90%) | Prenatal GC | 2.74 | Postnatal GC | 2.67 | .572 |
| | Prenatal GC | 2.74 | Prenatal Parent | 2.38 | .089 |
| | Postnatal GC | 2.67 | Postnatal Parent | 2.74 | .609 |

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| | Prenatal Parent | 2.38 | Postnatal Parent | 2.74 | .098 |
| May have some health conditions but not all | Prenatal GC | 2.84 | Postnatal GC | 2.79 | .672 |
| | Prenatal GC | 2.84 | Prenatal Parent | 2.46 | .067 |
| | Postnatal GC | 2.79 | Postnatal Parent | 2.87 | .556 |
| | Prenatal Parent | 2.46 | Postnatal Parent | 2.87 | .059 |
| Discuss available Turner syndrome clinics | Prenatal GC | 2.62 | Postnatal GC | 2.82 | .141 |
| | Prenatal GC | 2.62 | Prenatal Parent | 2.54 | .731 |
| | Postnatal GC | 2.82 | Postnatal Parent | 2.84 | .868 |
| | Prenatal Parent | 2.54 | Postnatal Parent | 2.84 | .173 |
| Discuss impact of hormones to increase stature or treatments that can allow girls with Turner syndrome to attain a normal adult height | Prenatal GC | 2.39 | Postnatal GC | 2.70 | .064 |
| | Prenatal GC | 2.39 | Prenatal Parent | 2.31 | .766 |
| | Postnatal GC | 2.70 | Postnatal Parent | 2.97 | **.029** |
| | Prenatal Parent | 2.31 | Postnatal Parent | 2.97 | **.002** |
| Girls with Turner syndrome need hormones to start or complete puberty | Prenatal GC | 2.50 | Postnatal GC | 2.70 | .223 |
| | Prenatal GC | 2.50 | Prenatal Parent | 2.31 | .466 |
| | Postnatal GC | 2.70 | Postnatal Parent | 2.87 | .200 |
| | Prenatal Parent | 2.31 | Postnatal Parent | 2.87 | **.010** |
| **The Family and Relationships of Individuals with Turner Syndrome** | **Group** | **Rating** | **Group** | **Rating** | **Sig < 0.05** |
| Impact on other siblings – more compassionate and caring | Prenatal GC | 1.74 | Postnatal GC | 1.82 | .672 |
| | Prenatal GC | 1.74 | Prenatal Parent | 1.92 | .521 |
| | Postnatal GC | 1.82 | Postnatal Parent | 2.33 | **.024** |
| | Prenatal Parent | 1.92 | Postnatal Parent | 2.33 | .163 |
| Impact on other siblings – less attention and resentful | Prenatal GC | 1.66 | Postnatal GC | 1.73 | .703 |
| | Prenatal GC | 1.66 | Prenatal Parent | 1.69 | .903 |
| | Postnatal GC | 1.73 | Postnatal Parent | 2.23 | **.018** |
| | Prenatal Parent | 1.69 | Postnatal Parent | 2.23 | .087 |
| Impact on parental marriage – strengthens relationship | Prenatal GC | 1.68 | Postnatal GC | 1.73 | .812 |
| | Prenatal GC | 1.68 | Prenatal Parent | 1.85 | .560 |
| | Postnatal GC | 1.73 | Postnatal Parent | 2.03 | .194 |
| | Prenatal Parent | 1.85 | Postnatal Parent | 2.03 | .560 |
| Impact on parental marriage – strains relationship | Prenatal GC | 1.74 | Postnatal GC | 1.79 | .803 |
| | Prenatal GC | 1.74 | Prenatal Parent | 1.38 | .187 |
| | Postnatal GC | 1.79 | Postnatal Parent | 1.94 | .527 |
| | Prenatal Parent | 1.38 | Postnatal Parent | 1.94 | .075 |
| Impact on grandparents – supportive and welcoming | Prenatal GC | 1.34 | Postnatal GC | 1.55 | .244 |
| | Prenatal GC | 1.34 | Prenatal Parent | 1.62 | .243 |
| | Postnatal GC | 1.55 | Postnatal Parent | 1.93 | .082 |
| | Prenatal Parent | 1.62 | Postnatal Parent | 1.93 | .250 |
| Impact on grandparents – | Prenatal GC | 1.30 | Postnatal GC | 1.45 | .367 |

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| limited interaction | Prenatal GC | 1.30 | Prenatal Parent | 1.31 | .973 |
| | Postnatal GC | 1.45 | Postnatal Parent | 1.68 | .320 |
| | Prenatal Parent | 1.31 | Postnatal Parent | 1.68 | .169 |
| Impact on extended family members – supportive and welcoming | Prenatal GC | 1.32 | Postnatal GC | 1.42 | .574 |
| | Prenatal GC | 1.32 | Prenatal Parent | 1.54 | .390 |
| | Postnatal GC | 1.42 | Postnatal Parent | 1.81 | .090 |
| | Prenatal Parent | 1.54 | Postnatal Parent | 1.81 | .327 |
| Impact on extended family members – limited interaction | Prenatal GC | 1.24 | Postnatal GC | 1.42 | .264 |
| | Prenatal GC | 1.24 | Prenatal Parent | 1.31 | .757 |
| | Postnatal GC | 1.42 | Postnatal Parent | 1.61 | .382 |
| | Prenatal Parent | 1.31 | Postnatal Parent | 1.61 | .231 |
| Impact on other relationships – supportive and welcoming | Prenatal GC | 1.46 | Postnatal GC | 1.55 | .630 |
| | Prenatal GC | 1.46 | Prenatal Parent | 1.77 | .233 |
| | Postnatal GC | 1.55 | Postnatal Parent | 1.83 | .195 |
| | Prenatal Parent | 1.77 | Postnatal Parent | 1.83 | .604 |
| Impact on other relationships – lose social circle | Prenatal GC | 1.41 | Postnatal GC | 1.55 | .443 |
| | Prenatal GC | 1.41 | Prenatal Parent | 1.38 | .924 |
| | Postnatal GC | 1.55 | Postnatal Parent | 1.73 | .395 |
| | Prenatal Parent | 1.38 | Postnatal Parent | 1.73 | .111 |
| Financial parental impact – no difference | Prenatal GC | 1.26 | Postnatal GC | 1.36 | .570 |
| | Prenatal GC | 1.26 | Prenatal Parent | 1.31 | .855 |
| | Postnatal GC | 1.36 | Postnatal Parent | 1.85 | .054 |
| | Prenatal Parent | 1.31 | Postnatal Parent | 1.85 | .111 |
| Financial parental impact - more | Prenatal GC | 1.56 | Postnatal GC | 1.48 | .699 |
| | Prenatal GC | 1.56 | Prenatal Parent | 1.85 | .314 |
| | Postnatal GC | 1.48 | Postnatal Parent | 2.37 | **< .0001** |
| | Prenatal Parent | 1.85 | Postnatal Parent | 2.37 | .101 |
| Time commitment of parent – no difference | Prenatal GC | 1.30 | Postnatal GC | 1.39 | .617 |
| | Prenatal GC | 1.30 | Prenatal Parent | 1.38 | .754 |
| | Postnatal GC | 1.39 | Postnatal Parent | 2.00 | **.012** |
| | Prenatal Parent | 1.38 | Postnatal Parent | 2.00 | .060 |
| Time commitment of parent - more | Prenatal GC | 1.56 | Postnatal GC | 1.55 | .941 |
| | Prenatal GC | 1.56 | Prenatal Parent | 1.69 | .637 |
| | Postnatal GC | 1.55 | Postnatal Parent | 2.16 | **.011** |
| | Prenatal Parent | 1.69 | Postnatal Parent | 2.16 | .153 |
| **Neurocognitive and Psychosocial Aspects of Turner Syndrome** | **Group** | **Rating** | **Group** | **Rating** | **Sig < 0.05** |
| Women and girls with Turner syndrome typically have normal intelligence | Prenatal GC | 2.96 | Postnatal GC | 3.00 | .250 |
| | Prenatal GC | 2.96 | Prenatal Parent | 2.17 | **.001** |
| | Postnatal GC | 3.00 | Postnatal Parent | 2.84 | **.046** |
| | Prenatal Parent | 2.17 | Postnatal Parent | 2.84 | **.020** |
| Intellectual disability (10%) | Prenatal GC | 2.52 | Postnatal GC | 2.67 | .268 |
| | Prenatal GC | 2.52 | Prenatal Parent | 1.69 | **.002** |

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| | Postnatal GC | 2.67 | Postnatal Parent | 2.35 | **.011** |
| | Prenatal Parent | 1.69 | Postnatal Parent | 2.35 | **.011** |
| Poor performance in mathematics (dyscalculia) (50%-75%) | Prenatal GC | 2.36 | Postnatal GC | 2.55 | .204 |
| | Prenatal GC | 2.36 | Prenatal Parent | 1.58 | **.006** |
| | Postnatal GC | 2.55 | Postnatal Parent | 2.48 | **.046** |
| | Prenatal Parent | 1.58 | Postnatal Parent | 2.48 | **.006** |
| Increased risk of attention-deficit/hyperactivity disorder (25%) | Prenatal GC | 2.26 | Postnatal GC | 2.64 | **.007** |
| | Prenatal GC | 2.26 | Prenatal Parent | 1.77 | .062 |
| | Postnatal GC | 2.64 | Postnatal Parent | 2.35 | **.034** |
| | Prenatal Parent | 1.77 | Postnatal Parent | 2.35 | .057 |
| Lack of working memory which may cause difficulty with multi-tasking, mental calculations, and holding information "in a mind's eye" | Prenatal GC | 1.84 | Postnatal GC | 2.12 | **.037** |
| | Prenatal GC | 1.84 | Prenatal Parent | 1.77 | .783 |
| | Postnatal GC | 2.12 | Postnatal Parent | 2.52 | .740 |
| | Prenatal Parent | 1.77 | Postnatal Parent | 2.52 | **.021** |
| Difficulty with problem-solving | Prenatal GC | 1.84 | Postnatal GC | 2.15 | **.032** |
| | Prenatal GC | 1.84 | Prenatal Parent | 1.62 | .428 |
| | Postnatal GC | 2.15 | Postnatal Parent | 2.48 | .126 |
| | Prenatal Parent | 1.62 | Postnatal Parent | 2.48 | **.013** |
| Inefficiency when learning through visual means | Prenatal GC | 1.70 | Postnatal GC | 2.12 | **.002** |
| | Prenatal GC | 1.70 | Prenatal Parent | 1.54 | .560 |
| | Postnatal GC | 2.12 | Postnatal Parent | 2.37 | **.035** |
| | Prenatal Parent | 1.54 | Postnatal Parent | 2.37 | **.021** |
| Strengths in various aspects of oral and written communication | Prenatal GC | 2.04 | Postnatal GC | 2.39 | .050 |
| | Prenatal GC | 2.04 | Prenatal Parent | 1.69 | .301 |
| | Postnatal GC | 2.39 | Postnatal Parent | 2.55 | .086 |
| | Prenatal Parent | 1.69 | Postnatal Parent | 2.55 | **.016** |
| Clumsiness and delayed motor milestones | Prenatal GC | 1.94 | Postnatal GC | 2.39 | **.003** |
| | Prenatal GC | 1.94 | Prenatal Parent | 1.85 | .725 |
| | Postnatal GC | 2.39 | Postnatal Parent | 2.60 | .213 |
| | Prenatal Parent | 1.85 | Postnatal Parent | 2.60 | **.008** |
| Difficulty identifying facial emotions | Prenatal GC | 1.44 | Postnatal GC | 1.88 | **.015** |
| | Prenatal GC | 1.44 | Prenatal Parent | 1.62 | .576 |
| | Postnatal GC | 1.88 | Postnatal Parent | 2.39 | .418 |
| | Prenatal Parent | 1.62 | Postnatal Parent | 2.39 | **.023** |
| Difficulty initiating or maintaining relationships | Prenatal GC | 1.42 | Postnatal GC | 1.94 | **.007** |
| | Prenatal GC | 1.42 | Prenatal Parent | 1.62 | .574 |
| | Postnatal GC | 1.94 | Postnatal Parent | 2.27 | .189 |
| | Prenatal Parent | 1.62 | Postnatal Parent | 2.27 | .058 |
| Girls with Turner syndrome may need more support to develop social | Prenatal GC | 2.28 | Postnatal GC | 2.45 | .345 |
| | Prenatal GC | 2.28 | Prenatal Parent | 1.85 | .210 |
| | Postnatal GC | 2.45 | Postnatal Parent | 2.58 | **.015** |

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| skills | Prenatal Parent | 1.85 | Postnatal Parent | 2.58 | **.036** |
| **Long-Term Life Outcomes for Individuals with Turner Syndrome** | **Group** | **Rating** | **Group** | **Rating** | **Sig < 0.05** |
| Participating in community activities (clubs, hobbies, sports, volunteer work, etc.) | Prenatal GC | 1.92 | Postnatal GC | 2.39 | **.010** |
| | Prenatal GC | 1.92 | Prenatal Parent | 1.62 | .288 |
| | Postnatal GC | 2.39 | Postnatal Parent | 2.43 | .828 |
| | Prenatal Parent | 1.62 | Postnatal Parent | 2.43 | **.007** |
| Inclusion in regular classes | Prenatal GC | 2.43 | Postnatal GC | 2.61 | .255 |
| | Prenatal GC | 2.43 | Prenatal Parent | 1.85 | **.033** |
| | Postnatal GC | 2.61 | Postnatal Parent | 2.52 | .592 |
| | Prenatal Parent | 1.85 | Postnatal Parent | 2.52 | **.032** |
| Special education services | Prenatal GC | 2.12 | Postnatal GC | 2.52 | **.011** |
| | Prenatal GC | 2.12 | Prenatal Parent | 1.77 | .188 |
| | Postnatal GC | 2.52 | Postnatal Parent | 2.45 | .732 |
| | Prenatal Parent | 1.77 | Postnatal Parent | 2.45 | **.022** |
| Early intervention services | Prenatal GC | 2.60 | Postnatal GC | 2.79 | .154 |
| | Prenatal GC | 2.60 | Prenatal Parent | 2.15 | .088 |
| | Postnatal GC | 2.79 | Postnatal Parent | 2.61 | .292 |
| | Prenatal Parent | 2.15 | Postnatal Parent | 2.61 | .124 |
| Benefit of psychological therapy and support | Prenatal GC | 2.06 | Postnatal GC | 2.52 | **.006** |
| | Prenatal GC | 2.06 | Prenatal Parent | 2.15 | .726 |
| | Postnatal GC | 2.52 | Postnatal Parent | 2.33 | .358 |
| | Prenatal Parent | 2.15 | Postnatal Parent | 2.33 | .455 |
| Possible ability to conceive through reproductive assistive technology after a thorough medical examination | Prenatal GC | 2.28 | Postnatal GC | 2.42 | .386 |
| | Prenatal GC | 2.28 | Prenatal Parent | 1.69 | **.043** |
| | Postnatal GC | 2.42 | Postnatal Parent | 2.32 | .506 |
| | Prenatal Parent | 1.69 | Postnatal Parent | 2.32 | **.028** |
| Alternate ways to become a parent when unable to conceive | Prenatal GC | 2.20 | Postnatal GC | 2.33 | .429 |
| | Prenatal GC | 2.20 | Prenatal Parent | 1.77 | .117 |
| | Postnatal GC | 2.33 | Postnatal Parent | 2.55 | .145 |
| | Prenatal Parent | 1.77 | Postnatal Parent | 2.55 | **.003** |
| Finishing high school | Prenatal GC | 2.06 | Postnatal GC | 2.12 | .757 |
| | Prenatal GC | 2.06 | Prenatal Parent | 2.23 | .551 |
| | Postnatal GC | 2.12 | Postnatal Parent | 2.74 | **.003** |
| | Prenatal Parent | 2.23 | Postnatal Parent | 2.74 | **.046** |
| More likely to earn a baccalaureate degree or higher than the general US female population | Prenatal GC | 1.78 | Postnatal GC | 2.06 | .177 |
| | Prenatal GC | 1.78 | Prenatal Parent | 1.85 | .823 |
| | Postnatal GC | 2.06 | Postnatal Parent | 2.58 | **.013** |
| | Prenatal Parent | 1.85 | Postnatal Parent | 2.58 | **.010** |
| More likely to be | Prenatal GC | 1.68 | Postnatal GC | 1.94 | .200 |

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| employed than the general US female population | Prenatal GC | 1.68 | Prenatal Parent | 1.85 | .567 |
| | Postnatal GC | 1.94 | Postnatal Parent | 2.35 | .056 |
| | Prenatal Parent | 1.85 | Postnatal Parent | 2.35 | .094 |
| Less likely to marry than the general US female population | Prenatal GC | 1.20 | Postnatal GC | 1.52 | **.044** |
| | Prenatal GC | 1.20 | Prenatal Parent | 1.38 | .394 |
| | Postnatal GC | 1.52 | Postnatal Parent | 1.84 | .105 |
| | Prenatal Parent | 1.38 | Postnatal Parent | 1.84 | .084 |
| Living independently | Prenatal GC | 2.40 | Postnatal GC | 2.39 | .973 |
| | Prenatal GC | 2.40 | Prenatal Parent | 1.77 | **.023** |
| | Postnatal GC | 2.39 | Postnatal Parent | 2.63 | .201 |
| | Prenatal Parent | 1.77 | Postnatal Parent | 2.63 | **.001** |
| Having friends and meaningful relationships | Prenatal GC | 2.38 | Postnatal GC | 2.61 | .174 |
| | Prenatal GC | 2.38 | Prenatal Parent | 1.92 | .106 |
| | Postnatal GC | 2.61 | Postnatal Parent | 2.71 | .510 |
| | Prenatal Parent | 1.92 | Postnatal Parent | 2.71 | **.003** |
| Having intimate relationships | Prenatal GC | 2.14 | Postnatal GC | 2.36 | .235 |
| | Prenatal GC | 2.14 | Prenatal Parent | 1.85 | .328 |
| | Postnatal GC | 2.36 | Postnatal Parent | 2.39 | .891 |
| | Prenatal Parent | 1.85 | Postnatal Parent | 2.39 | **.033** |
| Life expectancy (may be reduced up to 13 years) | Prenatal GC | 1.92 | Postnatal GC | 2.12 | .243 |
| | Prenatal GC | 1.92 | Prenatal Parent | 1.92 | .991 |
| | Postnatal GC | 2.12 | Postnatal Parent | 1.93 | .345 |
| | Prenatal Parent | 1.92 | Postnatal Parent | 1.93 | .743 |
| More like other children than different | Prenatal GC | 2.70 | Postnatal GC | 2.82 | .400 |
| | Prenatal GC | 2.70 | Prenatal Parent | 2.15 | **.039** |
| | Postnatal GC | 2.82 | Postnatal Parent | 2.71 | .386 |
| | Prenatal Parent | 2.15 | Postnatal Parent | 2.71 | .052 |
| Excel at verbal skills compared to the general US female population | Prenatal GC | 1.72 | Postnatal GC | 2.12 | **.028** |
| | Prenatal GC | 1.72 | Prenatal Parent | 1.85 | .649 |
| | Postnatal GC | 2.12 | Postnatal Parent | 2.67 | **.004** |
| | Prenatal Parent | 1.85 | Postnatal Parent | 2.67 | **.006** |
| Reduced fertility | Prenatal GC | 2.60 | Postnatal GC | 2.79 | .183 |
| | Prenatal GC | 2.60 | Prenatal Parent | 2.00 | **.020** |
| | Postnatal GC | 2.79 | Postnatal Parent | 2.68 | .361 |
| | Prenatal Parent | 2.00 | Postnatal Parent | 2.68 | **.011** |
| **Genetics of Turner Syndrome** | **Group** | **Rating** | **Group** | **Rating** | **Sig < 0.05** |
| Incidence (1/2500 females) | Prenatal GC | 1.88 | Postnatal GC | 1.79 | .605 |
| | Prenatal GC | 1.88 | Prenatal Parent | 2.00 | .656 |
| | Postnatal GC | 1.79 | Postnatal Parent | 2.45 | **.001** |
| | Prenatal Parent | 2.00 | Postnatal Parent | 2.45 | .066 |
| No significant increasing incidence with increasing maternal age | Prenatal GC | 2.56 | Postnatal GC | 2.06 | **.003** |
| | Prenatal GC | 2.56 | Prenatal Parent | 1.62 | **.001** |
| | Postnatal GC | 2.06 | Postnatal Parent | 2.43 | **.045** |

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| | Prenatal Parent | 1.62 | Postnatal Parent | 2.43 | **.009** |
| Caused by the partial or complete loss of the second X-chromosome in females | Prenatal GC | 2.80 | Postnatal GC | 2.76 | .718 |
| | Prenatal GC | 2.80 | Prenatal Parent | 2.69 | .555 |
| | Postnatal GC | 2.76 | Postnatal Parent | 2.69 | .630 |
| | Prenatal Parent | 2.69 | Postnatal Parent | 2.69 | .788 |
| Possible chromosomal/genetic causes of Turner syndrome: 45,X (40%-50%), 45,X/46,XX (15%-25%), other (25%-45%) | Prenatal GC | 2.24 | Postnatal GC | 2.39 | .360 |
| | Prenatal GC | 2.24 | Prenatal Parent | 2.08 | .530 |
| | Postnatal GC | 2.39 | Postnatal Parent | 2.62 | .187 |
| | Prenatal Parent | 2.08 | Postnatal Parent | 2.62 | .052 |
| Prenatal diagnosis can only be confirmed by chromosome analysis via amniocentesis or CVS | Prenatal GC | 2.82 | Postnatal GC | 1.52 | **< 0.0001** |
| | Prenatal GC | 2.82 | Prenatal Parent | 2.31 | **.011** |
| | Postnatal GC | 2.31 | Postnatal Parent | 2.48 | **.001** |
| | Prenatal Parent | 1.52 | Postnatal Parent | 2.48 | .596 |
| Screening results can be obtained via noninvasive prenatal screening (NIPT/cell-free DNA) | Prenatal GC | 2.74 | Postnatal GC | 1.58 | **< 0.0001** |
| | Prenatal GC | 2.74 | Prenatal Parent | 2.46 | .183 |
| | Postnatal GC | 1.58 | Postnatal Parent | 2.23 | **.002** |
| | Prenatal Parent | 2.46 | Postnatal Parent | 2.23 | .391 |
| Chance to reoccur in future pregnancies | Prenatal GC | 2.78 | Postnatal GC | 2.67 | .311 |
| | Prenatal GC | 2.78 | Prenatal Parent | 2.38 | **.029** |
| | Postnatal GC | 2.67 | Postnatal Parent | 2.67 | 1.000 |
| | Prenatal Parent | 2.38 | Postnatal Parent | 2.67 | .325 |
| Reproductive capability of a woman with Turner syndrome | Prenatal GC | 2.60 | Postnatal GC | 2.70 | .438 |
| | Prenatal GC | 2.60 | Prenatal Parent | 2.23 | .087 |
| | Postnatal GC | 2.70 | Postnatal Parent | 2.60 | .458 |
| | Prenatal Parent | 2.23 | Postnatal Parent | 2.60 | .150 |
| Reproductive options for a woman with Turner syndrome (pregnancy, surrogacy, adoption) | Prenatal GC | 2.26 | Postnatal GC | 2.52 | .117 |
| | Prenatal GC | 2.26 | Prenatal Parent | 2.08 | .273 |
| | Postnatal GC | 2.52 | Postnatal Parent | 2.67 | .331 |
| | Prenatal Parent | 2.08 | Postnatal Parent | 2.67 | **.025** |
| **Informational Resources and Referrals for Individuals with Turner Syndrome and Their Families** | **Group** | **Rating** | **Group** | **Rating** | **Sig < 0.05** |
| Local Turner syndrome support group(s) | Prenatal GC | 2.64 | Postnatal GC | 2.79 | .178 |
| | Prenatal GC | 2.64 | Prenatal Parent | 2.69 | .782 |
| | Postnatal GC | 2.79 | Postnatal Parent | 2.68 | .325 |
| | Prenatal Parent | 2.69 | Postnatal Parent | 2.68 | .959 |
| National advocacy organizations and websites | Prenatal GC | 2.60 | Postnatal GC | 2.85 | **.029** |
| | Prenatal GC | 2.60 | Prenatal Parent | 2.46 | .537 |
| | Postnatal GC | 2.85 | Postnatal Parent | 2.63 | .051 |
| | Prenatal Parent | 2.46 | Postnatal Parent | 2.63 | .396 |

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| Online support groups/social media platforms | Prenatal GC | 2.46 | Postnatal GC | 2.76 | **.033** |
| | Prenatal GC | 2.46 | Prenatal Parent | 2.31 | .533 |
| | Postnatal GC | 2.76 | Postnatal Parent | 2.43 | **.013** |
| | Prenatal Parent | 2.31 | Postnatal Parent | 2.43 | .470 |
| Printed/written material | Prenatal GC | 2.52 | Postnatal GC | 2.70 | .194 |
| | Prenatal GC | 2.52 | Prenatal Parent | 2.38 | .571 |
| | Postnatal GC | 2.70 | Postnatal Parent | 2.53 | .289 |
| | Prenatal Parent | 2.38 | Postnatal Parent | 2.53 | .489 |
| Photographs of children with Turner syndrome | Prenatal GC | 1.68 | Postnatal GC | 1.82 | .425 |
| | Prenatal GC | 1.68 | Prenatal Parent | 1.92 | .331 |
| | Postnatal GC | 1.82 | Postnatal Parent | 2.17 | .081 |
| | Prenatal Parent | 1.92 | Postnatal Parent | 2.17 | .348 |
| Fact sheets/brochures | Prenatal GC | 2.34 | Postnatal GC | 2.42 | .534 |
| | Prenatal GC | 2.34 | Prenatal Parent | 2.50 | .454 |
| | Postnatal GC | 2.42 | Postnatal Parent | 2.47 | .803 |
| | Prenatal Parent | 2.50 | Postnatal Parent | 2.47 | .965 |
| Books | Prenatal GC | 1.72 | Postnatal GC | 1.94 | .112 |
| | Prenatal GC | 1.72 | Prenatal Parent | 2.17 | **.024** |
| | Postnatal GC | 1.94 | Postnatal Parent | 2.20 | .150 |
| | Prenatal Parent | 2.17 | Postnatal Parent | 2.20 | .681 |
| Contact with Families raising a child with Turner syndrome | Prenatal GC | 1.96 | Postnatal GC | 2.33 | **.011** |
| | Prenatal GC | 1.96 | Prenatal Parent | 2.54 | **.012** |
| | Postnatal GC | 2.33 | Postnatal Parent | 2.48 | .348 |
| | Prenatal Parent | 2.54 | Postnatal Parent | 2.48 | .950 |
| Pregnancy termination resources | Prenatal GC | 2.43 | Postnatal GC | 1.03 | < **0.0001** |
| | Prenatal GC | 2.43 | Prenatal Parent | 0.83 | < **0.0001** |
| | Postnatal GC | 1.03 | Postnatal Parent | 1.47 | **.041** |
| | Prenatal Parent | 0.83 | Postnatal Parent | 1.47 | .060 |
| Alternative/nonconventional therapies | Prenatal GC | 1.39 | Postnatal GC | 1.33 | .736 |
| | Prenatal GC | 1.39 | Prenatal Parent | 1.42 | .912 |
| | Postnatal GC | 1.33 | Postnatal Parent | 1.63 | .186 |
| | Prenatal Parent | 1.42 | Postnatal Parent | 1.63 | .337 |
| Specialist referral(s) | Prenatal GC | 2.50 | Postnatal GC | 2.91 | **.002** |
| | Prenatal GC | 2.50 | Prenatal Parent | 2.67 | .475 |
| | Postnatal GC | 2.91 | Postnatal Parent | 2.94 | .700 |
| | Prenatal Parent | 2.67 | Postnatal Parent | 2.94 | .103 |
| Special education supports and services | Prenatal GC | 2.00 | Postnatal GC | 2.52 | **.001** |
| | Prenatal GC | 2.00 | Prenatal Parent | 2.42 | .067 |
| | Postnatal GC | 2.52 | Postnatal Parent | 2.58 | .716 |
| | Prenatal Parent | 2.42 | Postnatal Parent | 2.58 | .430 |
| Counselor or family therapist referral(s) | Prenatal GC | 1.90 | Postnatal GC | 2.00 | .554 |
| | Prenatal GC | 1.90 | Prenatal Parent | 2.31 | .119 |
| | Postnatal GC | 2.00 | Postnatal Parent | 2.26 | .192 |
| | Prenatal Parent | 2.31 | Postnatal Parent | 2.26 | .979 |

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| Pastoral counseling referral(s) | Prenatal GC | 1.50 | Postnatal GC | 1.73 | .214 |
| | Prenatal GC | 1.50 | Prenatal Parent | 1.08 | .124 |
| | Postnatal GC | 1.73 | Postnatal Parent | 1.48 | .296 |
| | Prenatal Parent | 1.08 | Postnatal Parent | 1.48 | .394 |