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Turner Syndrome: Addressing How Misconceptions Overshadow Opportunities for a “Normal” Life

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Abstract

Turner syndrome (TS) is a genetic condition found only in females. Responsible for an estimated 10% of all miscarriages and stillbirths, an estimated 99% of pregnancies involving monosomy TS (the most predominant subtype) are terminated, either voluntarily or as a result of medical complications. Despite poor prognosis for live birth, children born with TS have significant potential to develop into normal, happy individuals provided they are raised in a supportive environment. Unfortunately, TS issues are relatively underrepresented in counseling literature. In addition to addressing the medical aspects and prognosis of TS, this article examines the individual, couple, and family considerations necessary for providing competent and ethical practice.

Keywords: Turner syndrome, genetic, family counseling, miscarriage, self-esteem

Turner syndrome (TS), also referred to as Turners syndrome, is a complex medical condition found only in females, categorized by the absence of one sex chromosome in some or all of an individual’s cells. Genetically caused, TS occurs with the total or partial loss of the X sex chromosome before or shortly after conception (Rieser & Davenport, n.d.). Although the specific cause of this phenomenon is unknown, researchers noted that the loss of the X chromosome occurs spontaneously and has not been tied to biological factors (Morgan, 2007). Because of this spontaneous occurrence, there is no preliminary way to predict a couple’s potential of having a child with TS prior to conception. In fact, earliest diagnosis of TS frequently occurs through amniocentesis, which is often only conducted in older women, and ultrasounds (Boston Children’s Hospital, 2013). Given variability regarding the onset of signs and symptoms of TS, however, some individuals are not diagnosed until middle childhood.

According to Bondy (2007), the presence of a sex chromosome deficiency is one of the few constants within this syndrome. In fact, Rieser and Davenport (n.d.) noted “every girl with TS is unique and no generalization will apply to every girl, no matter how accurate it is for the group” (p. 2). Specifically, variations in symptoms may be physical, cognitive, or psychological in nature, including short stature, learning disabilities, social apprehensions, depression, and issues with puberty. Although most women with TS are able to live happy, productive lives despite these symptoms, families who identify the presence of TS in early pregnancy are often faced with a significant decision regarding whether to terminate the pregnancy. Following, an overview of the medical aspects of TS; gender and cultural factors; prognosis; individual, couple, and family dynamics; and treatment considerations are presented. Finally, a case illustration is presented to demonstrate considerations for providing counseling services to families affected by TS. Because of recent increase in the number of children born with TS (Genetics Home Reference, 2012), an awareness of TS and the issues faced by individuals within this population, as well as their families, is necessary.

Medical Aspects of Turner Syndrome

In 1938, Dr. Henry Turner described what is now referred to as Turner syndrome (TS), a condition which he categorized by the presence of short stature, a webbed neck, broad chest, and infertility (Turner Syndrome Society of the United States [TSSUS], 2013). Since its introduction within the research literature, a lot of information has been discovered regarding the diagnosis and potential treatment of TS. While the majority of symptoms noted by Turner have inconsistently been found among the population of individuals with TS, research has pointed to short stature and infertility as consistent among this population (Boston Children’s Hospital, 2013), as TS is a condition caused by a chromosomal abnormality that alters normal development in females. Present in approximately 1 of every 2,500 live female births and approximately 1 in every 10 miscarriages or stillborn births, TS is caused by complete or partial absence of the second sex chromosome (National Institutes of Health [NIH], 2013).

According to the Genetics Home Reference (2012), various types of TS exist with two more prominent types. Monosomy X, which exists in about one half to three-fourths of all individuals diagnosed with TS, occurs when an individual possesses only one sex chromosome, resulting in a total of 45 chromosomes as opposed to the standard 46. Mosaic TS, on the other hand, which occurs in approximately 20% of individuals diagnosed with TS, involves partial or rearranged genes within the second sex chromosome. Although the cause of TS and the presentation of the disorder are unclear, research has demonstrated the potential importance of the short stature homeobox (SHOX) gene, located on both sex chromosomes in human growth and development (Genetics Home Reference, 2012).

TS is the most common genetic disorder in women. Early diagnosis and treatment of individuals with TS in the Western world has developed faster than in Eastern countries (Athar, Idiculla, John, Tilak, & Kapur, 2012). Diagnosis can occur prior to birth, but is often not confirmed until after the child is born due to the presence of specific physical features (i.e., extra skin around the neck, puffy hands and feet, inability to straighten the elbow joints, low hairline, and soft upturned nails) and lack of normal

development (i.e., failure to grow at normal rates and absence of puberty onset). According to a study conducted by Massa et al. (2005), diagnosis of monosomy X TS is often diagnosed earlier than the mosaic karyotype, with an average diagnosis occurring between the ages of 6 and 7 and over three fourths of individuals being diagnosed prior to the age of 12.

Diagnosis of TS is difficult due to the diversity of symptoms. Although diagnosis can occur during amniocentesis, which has only recently screened for the disorder, this procedure is not carried out for all pregnancies (NIH, 2013). Consequently, some individuals do not receive a diagnosis of TS until their childhood, adolescence, or in some cases, early adulthood. The most common early symptoms occur around the age of 3 when a girl's rate of growth slows significantly. Puberty serves as another stage in which symptoms of TS become more noticeable. According to NIH (2013), females with TS often do not develop periods or breasts and will not do so without hormone treatments. Additional medical issues, which have been linked to Turner syndrome, include chronic ear infections, which can result in hearing loss; congenital heart defects, including the presence of a heart murmur; hypertension; speech problems; dental problems; infertility; and obesity. Furthermore, as TSSUS (2013) noted, despite the majority of individuals with TS possessing cognitive abilities similar to the general population, higher rates of visual-spatial learning disabilities were noted.

Gender and Cultural Factors

Although the cause of TS is unknown, research has noted no correlation between an increase in the mother's age at the time of conception and the rate of TS pregnancies (Boston Children's Hospital, 2013). In fact, Wolff, Van Dyke, and Powell (2010) further identified that in addition to being found in pregnancies of diverse ages, instances of TS can be found in all ethnic groups and in all areas around the globe. Factors associated with the diverse occurrence of TS and lack of an established cause result in the potential for any woman to give birth to a child with TS. In fact, the only consistent cultural factor associated with TS is that TS only occurs in females.

Prognosis

According to Iyer et al. (2012), results of a 10-year study revealed that the number one cause of death among individuals with the TS karyotype was voluntary termination of pregnancy. Specifically, nearly two thirds of pregnancies involving children diagnosed with TS prenatally were terminated voluntarily. Of the 124 fetuses identified as having TS (either prenatally or postnatally), only one fourth of pregnancies resulted in live births (with two additional children dying in the first few days after birth; Iyer et al., 2012). Although results of this study indicate increased rates of live births for individuals with mosaic TS as well as those with less common karyotypes, the high rates of early termination, especially of fetuses diagnosed with monosomy X, result in difficulties of accurately assessing the disease's prognosis.

Although prognosis for successful delivery of a TS fetus is relatively poor, prognosis following a live birth is significantly better. In fact, the Boston Children's Hospital (2013) noted that "most girls with the disease go on to lead normal, happy lives"

(para. 2). While there is no cure, there are treatments to address the majority of significant health issues associated with TS. As a result of growth hormone therapy, estrogen replacement, and early preventive measures to treat symptoms associated with TS, studies report increased pregnancy rates (Abir et al., 2001), increased stature (TSSUS, 2013), and life expectancy rates similar to that of the general population (Genetics Home Reference, 2013). In fact, Naess, Bahr, and Gravholt (2010) noted that, with proper support, individuals with TS obtain similar rates of productivity in education and work environments. While promising, the infancy of these approaches when working with individuals with TS results in the lack of established long-term treatment effects.

Individual Dynamics of TS

Similar to the diversity in chromosome abnormalities and physical symptoms associated with the presence of TS, psychosocial dynamics of individuals with TS are likely to differ. Bondy (2007), for example, revealed increased psychological symptoms within the population of individuals with TS including anxiety, depression, and social withdrawal. The results of this study are not surprising, however, given the numerous studies indicating heightened bullying victimization among individuals with TS (Suzigan, De Paiva E Silva, Guerra-Junior, Marini, & Maciel-Guerra, 2011). According to Cragg and Lafreniere (2010), decreased self-esteem has been linked to decreased body image but has not been linked to decreased performance abilities. These results indicate potential social effects of physical differences from the general population.

Couple and Family Dynamics of TS

Increased rates of psychosocial symptoms, including poor self-esteem and body image, have the potential to extend beyond the individual to couples and even families. According to Pope, Murray, and Kemer (2013), positive self-esteem is associated with relationship satisfaction. Poor self-esteem with one member of the relationship, therefore, has the potential to limit overall relationship satisfaction. Another factor with potential implications for the couple dynamic includes the high propensity for infertility. Specifically, Peterson, Gold, and Feingold (2007) identified that issues of fertility have been demonstrated to decrease self-concept, while increasing individual and relationship stress, sexual issues, and marital dissatisfaction. These factors may lead to the overwhelming numbers of couples seeking medical treatments to increase the potential of conception. These procedures, however, may be highly costly and are not guaranteed to be efficacious. In addition, many of these treatment options significantly increase the potential for women to become pregnant with multiple fetuses, increasing the risk of birth defects, loss of pregnancy, and even death within the first year following birth (Pope et al., 2013). Decisions regarding how to approach these issues can be difficult and can serve to increase stress and conflict.

Although the presence of TS among women within relationships may be associated with increased marital dissatisfaction, the effects of the presence of TS within children can also be the source of issues within the family system. According to TSSUS (2013), 99% of pregnancies involving fetuses with monosomy TS are voluntarily terminated, end in miscarriage, or are stillborn. In cases where TS is discovered

prenatally, parents are faced with a decision regarding whether to terminate the pregnancy (Iyer et al., 2012). The decision for early termination may increase mental health symptoms and conflict within the family system (Lipp, 2009).

Despite evidence presented by Lightsey and Sweeney (2008) supporting the potential for the birth of a child with a disability to increase the strength of the family relationship, financial stressors may accompany raising a child with TS. According to Boston Children's Hospital (2013), the cost of hormone treatment for children with TS can place a significant strain on families. Fortunately, NIH (2012) noted that the majority of health care plans cover both preventative treatment and hormone treatment with children diagnosed with TS, reducing the potential financial stress.

Individual, Couple, and Family Considerations in Turner Syndrome

Counselors working with individual, couples, or family systems dealing with issues of TS must understand the uniqueness of each presented case. Variability in associated symptoms as well as age at diagnosis are likely to impact the treatment approach. In cases where discovery of TS occurs prenatally, for example, psychoeducation may be beneficial by providing clients the tools to make an informed decision. Having a strong team to support the family in making decisions is important (TSSUS, 2013). Counselors could provide support for the family in order for them to make important decisions about birth and equip the family with resources that they may need following birth.

Difficult decisions regarding care of individuals with TS do not end at birth, however. According to the Turner Syndrome Foundation (2014), individuals with TS and their families are often faced with decisions regarding the use of growth hormone therapy and estrogen replacement therapy. According to Tolt and Rehan (2014), families often elect to have adolescent girls with TS injected with growth hormones at an early age to increase their projected height from 4 feet to 4 feet 8 inches. Such injections are conducted several times per week, with potential side effects to include headaches, scoliosis, and even carpal tunnel syndrome. Whereas growth hormone therapy is intended to support in the development of bone growth, estrogen replacement therapy is intended to initiate puberty in the majority of individuals with TS who likely would not enter puberty without it (Turner Syndrome Foundation, 2014). Because estrogen replacement therapy cannot be introduced until growth hormone therapy is completed, individuals with TS are more likely to enter puberty later than their peers, resulting in increased social anxiety, isolation, and even bullying (TSSUS, 2013). Counselors involved with a family at this stage could help to normalize the experience that the parents feel while balancing the independent needs of the adolescent.

The effects related to the combination of decreased height and late onset puberty in adolescence must be considered in providing counseling services to individuals with TS and their families. According to Gonzalez and Witchel (2012), the notable differences in development between those individuals with TS and those without during the adolescent period further compound concerns with low self-esteem among those with TS. The development of support groups for individuals with TS should be considered due to their demonstrated efficacy (TSSUS, 2013). While the availability of these resources is growing, less than half of states have recognized TS chapters available.

The novelty in development of TS support groups within the country is mirrored by the presence of minimal research on addressing mental and behavioral health symptoms and even disjointed medical care (TSSUS, 2013). These factors further strengthen the importance of growing TS research as well as the need for increasing knowledge regarding the disease among medical and psychological providers. While no research to date examines the most effective approaches to working with individuals with TS or their families, results of previous studies on TS indicate various aspects to consider. For example, Christopoulos, Deligeoroglou, Laggari, Christogiorgos, and Creatsas (2008) found that the establishment of strengths within the individual and family system was associated with increased adjustment among women with TS. Donaldson, Gault, Tan, and Dunger (2006), on the other hand, identified the importance of early education and intervention in increasing adjustment among those affected by TS. Specifically, Donaldson et al. proposed that education and support for parents of a child with TS, including discussion of infertility during childhood, are beneficial in making a better transition into adulthood. Therefore, strength-focused, systemic approaches should be considered.

Case Illustration

Brenda is a 12-year-old female with TS. A well-adjusted child in elementary school, Brenda began showing signs of isolating herself from others when she entered middle school. While many of her peers experienced significant growth spurts, Brenda's growth has been significantly slower. If that was not enough to increase her feelings of being different, Brenda's peers have begun entering puberty. In fact, Brenda has noticed that she is the only girl who has not developed breasts. Whereas she used to be the girl everyone wanted to play with due to her personality, she has found that boys are now interested in the other girls. As a result, her self-esteem has greatly suffered.

Brenda's parents, Carl and Jean, are lost as to what to do to help their daughter. While they knew the day would come when their daughter's TS would be more pronounced, they had never prepared themselves to talk to Brenda about TS or the fact that she had been diagnosed with it during the initial amniocentesis. After years of unsuccessful attempts to have a child, Brenda was their miracle child. Maybe due in part to the feelings of Brenda being a miracle, Carl and Jean noted that they somehow thought Brenda already knew that she was different and had just adjusted well. After all, there were weekly appointments for growth hormone therapy, and they had always stressed how they thought that they would never be able to have children.

While the couple experiences guilt that they had not talked to their daughter sooner about the experiences she may face as a result of TS, Carl and Jean also experience guilt in their decision to extend the growth hormone treatment until now instead of initiating estrogen replacement therapy. They had wanted to give Brenda the opportunity to reach her maximum growth potential to increase her feelings of normalcy and believed that Brenda might be one of the small percentage of girls with TS who initiated puberty without the estrogen replacement therapy.

Feeling like failures as parents and worried about their daughter, Carl and Jean were encouraged to seek out the support of a counselor. They are referred to a counselor with knowledge of the medical aspects of TS who specializes in family therapy. Because

they are not aware of Brenda's understanding of her diagnosis, the couple chooses to work with the counselor without Brenda at first. In establishing this relationship, Carl and Jean establish three specific goals for therapy. The preliminary goal was set to increase the family's understanding of their daughter's diagnosis. Understanding the implications of Brenda's diagnosis, not only for Brenda but also for the family, was noted as important in improving Carl and Jean's perception of their parenting abilities and their faith in their capabilities to provide effective and informed decisions with her. Secondary to the goal of improving their understanding of Brenda's diagnosis, the couple established a goal to be able to communicate effectively with their daughter especially in regards to her diagnosis. In making this goal, the family identified the impact of ignorance regarding TS in guarding their daughter against the realities of the world and stated a desire to interact with their daughter based on her capabilities. Finally, Carl and Jean established what they noted as the most important goal for them, although the couple noted that they understood that they could not make much progress on meeting the final goal until they were able to work on the first two. In this goal, the family expressed a desire for their daughter to adapt positively to her environment in order to increase her self-esteem and feelings of belonging with her peer group.

Given the goals established in the session, the counselor must develop effective interventions focused on improving knowledge and awareness of TS, improved communication skills, and the development of self-esteem. In doing so, the counselor must be cognizant of the potential discrepancies between an individual with TS' overall processing and individual elements of social and emotional processing. While these differences are associated with many individuals diagnosed with TS, the counselor must also understand that the only symptom consistent among all individuals with TS is short stature. Therefore, differences in processing cannot be assumed.

Case Reflection

Given the variability of symptoms associated with TS and individuality among all individuals with TS, the counselor must assess the abilities of Brenda. It is only through this understanding of Brenda's capabilities that the counselor can lay the foundation for services. To start, both Brenda and her parents may benefit from participation in a TS support group. While TSSUS provides support for individuals with TS and their families nationally, 26 states also have local chapters for individuals dealing with issues associated with TS (TSSUS, 2013). A referral to this agency may be beneficial in increasing support as well as education. Because education and advocacy are both important elements of TS (TSSUS, 2013), treatment that integrates elements to empower the client and family will also be beneficial. Above all, prior to providing therapy to clients with TS or their families, counselors should recognize the advancements in treatment for TS and the potential for individuals with TS to live normal, fulfilling lives.

Conclusion

TS is an unexplained condition which occurs in a small percentage of live births both in the United States and across the world. Thanks to advancements in early screenings and treatment for individuals with this condition, the potential for early, ongoing interventions has increased the overall prognosis for individuals with this condition. Although TS is primarily a medical condition, treatment of TS must also integrate solid therapeutic care to address the social implications of the disorder. As such, counselors must increase their knowledge and awareness of the social aspects of TS in order to treat these individuals and their families effectively. Therefore, the current article serves only as a baseline foundation for what counselors should know.

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Note: This paper is part of the annual VISTAS project sponsored by the American Counseling Association. Find more information on the project at: <http://www.counseling.org/knowledge-center/vistas>