

GROWING UP WITH TURNER SYNDROME: EXPERIENCES AND PERCEIVED FAMILY IMPACT

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ABSTRACT

Background: Across the United States, more than 71,000 women and girls are living with Turner syndrome (TS), a chromosomal abnormality in which one of the two X chromosomes normally found in females is missing or incomplete.

Objective: The present study aimed to explore the life experience of women who have TS and the impact it has had on their families. This is an area lacking in the current literature, which is mostly focused on the medical complications and academic issues associated with TS.

Methods: This study employed a qualitative, phenomenological approach. Twelve adult women were interviewed to assess the impact TS has had on their daily lives and the lives of their family members. Data were analyzed using Moustakas' modified Van Kaam method.

Results: In general, participants believed they grew up like "normal" children, with the exception of frequent doctor appointments and following up with potential medical complications from the syndrome. They did not think TS greatly affected their family members. Participants emphasized that women with TS can lead happy, healthy lives. They also offered advice for family members, such as seeking out resources from the Turner Syndrome Society of the United States.

Conclusions: Participants did not believe that, overall, their life has been negatively affected or limited as a result of having TS. Girls and women with TS should be given any needed support and assistance to pursue activities and careers they enjoy, and should not be limited or stigmatized by the diagnosis. Family members should also receive any needed assistance and support. Health care professionals should be educated about the syndrome and refer

BACKGROUND

Turner syndrome (TS) is a chromosomal abnormality in which one of the two X chromosomes normally found in females is missing or incomplete. Affecting roughly 1 in 2500 live female births, more than 71,000 women and girls are living with TS in the United States.¹ Features of the syndrome may include short stature, gonadal dysgenesis, infertility, congenital heart defects, renal abnormalities, hearing loss, obesity, and nonverbal, visual-spatial processing learning disabilities.¹⁻⁵ Those with TS may also have challenges interpreting nonverbal communication, which can lead to decreased socialization skills.³ They have intelligence levels similar to the general population.¹ As a result of the physical and social challenges girls and women with TS experience, they may have difficulty forming interpersonal relationships and a having a positive body image, leading to psychological complications.⁶ These may include decreased self-esteem.

Although the medical ramifications of the syndrome have been studied, little research has been conducted that explores the impact TS has on family members and the overall experience girls with TS have growing up. In the present study, a qualitative, phenomenological research approach was taken to explore such issues. Adult women were interviewed to discuss their experiences growing up with TS, obtain their perspective on how the syndrome affects family members, and learn what recommendations they had for girls/women with TS and their families.

METHODS

The aforementioned research approach received institutional review board approval from Winston State University in North Carolina. Participants were recruited after presenting the study at a meeting of the North Carolina Chapter of the Turner Syndrome Society of the United States (TSSUS), and then sending an e-mail to TSSUS members. Those interested in participating

contacted the researcher to determine eligibility and to arrange an interview time. Eligibility requirements included being at least 18 years of age and having received a diagnosis of TS. Ultimately, 12 adult women with TS consented to participate in the study and were interviewed individually at a location of their choice, such as their home or at a restaurant (2 participants were interviewed via phone due to geographic distance between the researcher and participant).

Participant demographics are shown in the table. Interviews were informal and included discussing the impact TS has had on their family members as well as recommendations for those involved in the life of someone with TS. Questions asked during the interview aimed to gain a better understanding of the participants' experiences with the syndrome. They included "What has your experience with Turner syndrome been like?" "What challenges have you faced as a result of having Turner syndrome?" and "How has Turner Syndrome impacted your family?" A set list of questions was asked of each participant, and follow-up and clarification questions were asked as needed and appropriate.

Interviews were recorded and then transcribed by a medical transcriptionist. Moustakas' modified Van Kaam method was utilized throughout the data analysis process.⁷ This included reading transcripts from the interviews and highlighting the major concepts and ideas

related to the experience and impact of TS. The resulting concepts were then continually reviewed and grouped. Redundant information, or information specific to an individual case (eg, one participant discussed the adoption of her son in depth), was eliminated. A follow-up interview was conducted over the phone with each participant to review individual themes as a form of member checking, and to see if participants had anything further to add or clarify. Second interviews, also recorded and transcribed, were then analyzed separately. Once themes from each participant's interviews were combined, themes for all 12 participants were reviewed and combined to obtain the final findings. Each participant's combined themes were e-mailed to her for feedback as a form of data triangulation. No changes were requested by participants.

RESULTS

Three main themes arose from the present study examining the life experience of women who have TS and the impact it has had on their childhood and family members. Quotes from participants (identified by numbers) are included to further support the findings.

A Normal Childhood

When talking about growing up with TS, most participants said they were treated like other children and remarked that they "grew up as a normal child" (6).

Participant	Age Diagnosed	Current Age	Education Level	Relationship Status	Occupation
1	8-10 Years	Not Provided	High School	Married	Receptionist
2	Infant	36	Bachelor's Degree	Married	Substitute Teacher
3	18-19 Years	Not Provided	Master's Degree	Single	Teaching & News Writing
4	6 Years	Not Provided	Master's Degree	Single	Computer Programmer Analyst
5	Birth	37	Clinical Doctorate	Married	Pharmacist
6	Birth	35	Bachelor's Degree	Boyfriend	Dietician
7	4 Months	30	Bachelor's Degree	Single	Customer Service
8	Birth	27	Bachelor's Degree	Single	Unemployed
9	1 ½ Years	22	Attending Community College	Single	Unemployed
10	21 Years	61	GED	Single	Volunteers
11	Birth	35	Doctoral Degree	Single	Teaches at University
12	11	27	Master's Degree	Married	Social Worker

They were grateful for being treated the same as other siblings in their family, and did not think they received any special treatment. Participants stated that, "My family...they know who I am. Turner syndrome is a part of that..." (4). They reported that their parents "have always been supportive" (9), and that "any help I need, they will help me" (10). Growing up, participants were told by their parents that they could do whatever they wanted in life, and that they would stand behind them. The participants surveyed were also supported and encouraged in other ways, including being taken to medical appointments, receiving necessary medical care, having tutoring for academic areas like math, and getting advice and assistance with social skills.

When asked how the syndrome was discussed when they were children, many participants reported that TS felt very private and like a secret in their family. "In some ways, my family...would act like it was something to be embarrassed about...They would say 'You don't want to tell people, they might make fun of you' " (11). This led to some participants and family members not disclosing the TS diagnosis to others, including extended family.

Family Impact

When exploring how TS has affected their families, participants expressed that family members primarily had to deal with managing the effects of TS. Parents needed to learn how to handle both the physical and social aspects of the syndrome (eg, the difficulty some girls and women with TS may have making friends). There was a learning curve for many parents, which included researching TS, what it medically involves, how to deal with potential problem areas and challenges (eg, social issues and spatial awareness issues), and how to keep up with medical concerns. The infertility aspect of TS, and not being able to have biological children, was something that some participants believed was "...definitely something that they think about and are aware of" (7). Frequent doctors' visits also affected family dynamics, as it is not typical for children to have to visit medical professionals so frequently. Finally, some participants thought that "finances were probably the hardest part of the whole thing..." (9). Medical bills, such as those for growth hormones and doctors' visits, added to the typical financial stresses families may face. Participants reported that their families found financial assistance through organizations like the MAGIC Foundation and local churches.

When specifically discussing the impact TS has had on their mothers, participants frequently reported that "she was the one who went through everything with me the most" (12). This included going to doctors' appointments and TSSUS meetings. Part of the reason for this, according to participants, may have been that their fathers were usually working more than their mothers. Going to appointments and meetings often provided a time for participants to bond with their mothers. Participants believed that receiving the diagnosis of TS helped answer some of their mothers' questions and concerns, such as lack of development, which physicians previously might have attributed to the daughter being a "late bloomer." Participants reported that their mothers did not always know how to reveal to them that they had TS, possibly because their mothers did not have a lot of information on the condition. They reported that it was difficult for their mothers to learn more about TS and to help them through problems like teasing. Some participants stated that, "it took a little bit of a toll on her, but she was always there for me and it has definitely gotten better since I was younger..." (8). While participants felt that their mothers were sad and needed a grieving period because they could not have children, they reported their mothers were "...just like, I'm going to treat her like a normal kid" (2). They "kind of saw it as something that made me unique and...loved me all the more for it" (12). TS made the participants special, unique, and stronger in their mothers' eyes.

When discussing the impact of TS on fathers, some participants believed that their fathers' were skeptical about the diagnosis. This was largely because tests that were run to check for complications common in those with TS might have come back normal, and the participants might not have experienced any of the difficulties frequently found in women with TS (eg, some were at the top of their class academically). This did not keep fathers from assisting with things when needed, like giving growth hormone shots and going to doctors' appointments and other procedures. Although participants thought their fathers were not as involved as their mothers, they reported that their fathers have "always been supportive" (5).

Participants believed that their diagnosis of TS had no real impact on their siblings. TS is not something they generally discussed together, and participants reported that their siblings might not understand TS or know the

name of the condition. Participants conveyed that their siblings frequently stood up for them when they were younger, and that they continue to offer encouragement now as adults. Participants did report that it was difficult when their sisters had children, knowing that they couldn't have biological children themselves.

Know That the Girl With TS Is Like Any Other Child

Based on their experiences, participants had advice for families affected by TS. They recommended that families stay informed and learn as much as possible about the syndrome so they can better know what to expect. Overall, participants believed this would increase the family's ability to help the girl/woman with TS address any issues that might come up, such as medical challenges or difficulty with spatial relations in math. Participants thought that staying current with TS can also enable families to educate doctors who might not know what to look for and address in people who have TS. Participants further recommended that families work with the individual's strengths to optimize her success, offering support and encouragement when needed. Support might include following up with physicians and addressing any medical concerns, or getting Math and English tutors.

In addition to making sure the individual with TS receives appropriate care, participants stressed that families should not treat girls/women with the syndrome any differently from others. They recommended that families "be aware of what to look for...[but] just be a parent... Don't be seeking out things that aren't there...Don't be putting limitations just from a diagnosis" (6). They felt that putting limitations on a girl/woman with TS can lead to a self-fulfilling prophecy such that she might not reach her maximal potential. Along similar lines, participants stressed that parents should not lower their expectations for their daughter, because she can have a normal, healthy life. They stated to simply love her, and not to "expect any more or less of your daughter because she has Turner syndrome than you would of any other daughter...She can live a normal life...It really isn't so much different from being a parent of any other daughter...Every kid is different!" (4).

Participants believed that, while there may be a need to address some challenges, in general a woman with TS can be as healthy, smart, and capable as anyone else. They stated that families must find that balance between talking about TS, staying informed, and putting things in perspective while not limiting the girl/woman with TS or

making a big deal out of having the syndrome. Participants wanted families to know that TS is "...not the only thing about them or maybe the most important thing about them" (12). They thought that it was important for families to be comfortable talking about TS, including breaking things down to a level where their daughter can understand what is going on, and to be honest about how they feel about infertility. Such honesty can help the daughter deal with the issue as well. To get support for both the girl/woman with TS and other family members, participants recommended that families connect with others affected by the syndrome and seek resources, which included organizations like the TSSUS.

DISCUSSION

Based on the findings of the current study, health care professionals need to offer support and education to girls/women with TS and their family members. This includes helping family members realize the individual's strengths, as well as the potential challenges she may experience, and how to best support her. This could involve providing them with education on TS, what areas might need to be evaluated and addressed, tutoring resources, assistive technology options, their legal rights such as special accommodations at school, support groups, and resources such as the TSSUS. Areas that might need to be addressed include visual-motor, visual-spatial, visual attention, executive functioning, planning, and problem-solving skills.³ Families can be educated on how to assist the girl/woman with TS in compensating for and working around challenging areas.³ For example, providing strategies for the family to help them assist their daughter with social challenges and with developing skills might be beneficial. This can include how to effectively and safely respond to bullying and teasing, as well as preparing and assisting with the transition to adulthood (eg, obtaining accommodations for college, entering the workforce, and maintaining a home).⁸ Addressing self-image, self-esteem, and body image concerns, as well as the challenges with making friends that those with TS may have (areas of concern mentioned by participants in the present and other studies), is also important.^{2,9,10} This might be accomplished by encouraging girls/women with TS to sign up for activities that interest them in an effort to have more positive social interactions.¹⁰ As discussed by participants, with the appropriate intervention, "girls and women with Turner syndrome now more than ever have the capability of achieving their full potential." (11)

Despite potential challenges, the importance of treating the girl with TS like any other child, and understanding that she can lead a fulfilling life, should be stressed to family members. This was very important to participants in the current study, and included realizing that limits should not be placed on someone just because she has TS. The importance of being open to talking about the syndrome, and not making the topic feel taboo or like a shameful secret should also be reiterated to families. This might help reinforce that she can live a normal and happy life. Furthermore, health care professionals can provide suggestions on how to divulge and discuss TS with the individual who has it, as well as with others, to make such conversations easier.

It is also necessary for both family members and individuals with TS to learn how to advocate for themselves so their needs are met. This can range from locating financial support for medical bills to obtaining resources for academic success and medical care. Parents and other family members affected by TS need to know how important it is to get the support they themselves need, both in learning about the syndrome and getting emotional support and encouragement. This includes honestly exploring feelings about TS, especially the infertility aspect of the condition. Frequently discussed by participants in the present study, Sutton et al also found that the issue of infertility was “the most prevalent and painful challenge endured by most of the adult women interviewed, as they were continually reminded of their infertility as family members and friends began to procreate.”¹¹ In addition, mothers in their study reported feelings of sadness and disappointment for their daughters. Encouraging women with TS to explore other options, such as adoption, and helping them understand that children can still play an integral role in their lives in other ways, may help with this challenging issue.¹¹ Discussing and receiving support for such emotional and significant areas in life can help all involved better cope with and handle the complications of TS. Finally, it is important to consider how dealing with the concerns and complications of TS might affect family dynamics (an area of concern participants in the present study mentioned), and to ensure as much as possible that a healthy balance is maintained in the household. This will help guarantee that the needs of all children and adults in the household are being met. Ensuring that family members affected by TS have the support they need will help them as well as helping their child or sibling with TS.

Limitations

The present study had 12 participants, all from the southeast region of the United States. This may have affected which resources and support were available to them compared with other regions of the country and elsewhere (eg, the medical care and support groups available may be different across the various regions of the United States and in other countries). Furthermore, participants were recruited through the TSSUS. Women who are not members of the TSSUS most likely did not hear about the study, and may have different views and experiences compared with the 12 participants. It is also important to note that participants in this study may have been more open to discussing their experiences with TS compared with other women who have the syndrome but did not participate in the study. In addition, while women with TS have intelligence levels similar to the general population, the educational level of participants might be considered higher than the average educational level of women with TS.¹ Finally, the phenomenological, qualitative approach of this study makes generalizability difficult.

CONCLUSIONS

The findings of the present study indicate that most women with TS believe they have had a relatively “normal” childhood and that they were not treated differently or hindered from having a happy childhood by the condition. From their perspective, TS affected their family members in terms of having to cope with frequent doctors’ visits and assisting with common issues girls/women with TS may experience, such as difficulty with social and spatial skills. Participants believed that their mothers were more involved and affected by TS than were other immediate family members. When looking at recommendations participants had for family members, the common themes were to treat the individual with TS like any other child, to recognize that she can live a normal and happy life, and to get any needed support and assistance.

Suggestions for future studies include interviewing parents with TS to better understand the experience of raising a daughter with the syndrome. This could include discussing the challenges they have experienced and resources they have found, as well as recommendations they may have for health care professionals.

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