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Reflections On The Current State Of Healthcare Transition for Young Adult Women With Turner Syndrome: Strategies For Facilitating Autonomy and Self-Management

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Reflections on the Current State of Healthcare Transition for Young Adult Women with
Turner Syndrome: Strategies for Facilitating Autonomy and Self-Management

by

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Bachelor of Science
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Abstract

The transition to adult-centered healthcare is a critical period for emerging adults, especially those with special healthcare needs (SHCNs). Considering the ongoing medical monitoring necessary for women with Turner syndrome (TS), it is essential that the transition process be comprehensive and well-coordinated. The aims of this study were to invite young women with TS to reflect on their healthcare transition experiences, to explore participants' perceived control of their medical management, and to identify ways in which genetic counselors can be involved in multidisciplinary healthcare teams. The hypotheses were that young women with TS are motivated to learn more about their diagnosis, benefit from counseling on how to develop self-management skills, and believe it would be useful to speak with a genetic counselor as adolescents.

Study participants included twenty-two women between the ages of 18 and 30 years with a diagnosis of TS. A mixed method study design was used, consisting of quantitative data collection via an online survey tool and follow-up qualitative interviews. Interviews were transcribed and reviewed for major themes using data driven coding and subsequent exploratory thematic analysis. Results demonstrated that participants who received counseling on the development of self-management skills as adolescents were more confident, independent, and satisfied with their transition to adult-centered medical care than participants who did not. Six major themes were identified in participant interviews. These included diagnosis disclosure, TS education, genetics knowledge, transition support, patient autonomy, and multidisciplinary care.

In conclusion, young adult women with TS are highly motivated to achieve autonomy in their healthcare and would appreciate being given additional information about their condition at a younger age. Healthcare providers should carefully consider patient preferences in order to provide patients with optimal support and appropriate resources. New recommendations for clinical practice should incorporate involvement of genetic counselors in the multidisciplinary healthcare teams of girls and women with TS.

Keywords: Turner syndrome, healthcare transition, emerging adulthood, patient autonomy, genetic counseling

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List of Abbreviations

AAP	American Academy of Pediatrics
ACGC	Accreditation Council for Genetic Counseling
KMO	Kaiser-Meyer-Olkin
LC	Learning collaborative
PCA(s)	Principal component analysis
PI.....	Principal investigator
SHCNs	Special health care needs
SPSS.....	Statistical Package for the Social Sciences
TRN	Turner Resource Network
TS.....	Turner syndrome
TSF.....	Turner Syndrome Foundation
TSSUS	Turner Syndrome Society of the United States

Chapter 1: Background

Increasingly more children with special healthcare needs (SHCNs) and various genetic conditions are living into adulthood and flourishing as independent individuals (Beal, Doty, Hernandez, Shea, & Davis, 2007). The majority of children with SHCNs receive excellent care in the pediatric setting, but our confidence in providing these individuals with the best quality care as they grow into adulthood is less well understood (Oswald et al., 2013; Romelczyk et al., 2015). One critical component of quality healthcare includes providers' abilities to encourage emerging adults to take responsibility for their medical management during the transition to adult-centered care. The healthcare transition process, as described by Blum et al. (1993), is considered "a multifaceted, active process that attends to the medical, psychosocial, and educational needs of adolescents as they move from the child-focused to the adult-focused health-care system" (p. 573). The process of transition has captured national attention, with various organizations and advocacy groups striving to determine how professionals can achieve the most comprehensive care for all emerging adults, especially those with SHCNs (American Academy of Pediatrics, American Academy of Family Physicians, American College of Physicians, & The Transitions Clinical Report Authoring Group, 2011).

Self-management and health literacy of emerging adults with SHCNs is another area of healthcare that has received increased attention over the past few years (Sattoe et al., 2014; Sebastian, Ramos, Stumbo, McGrath, & Fairbrother, 2014). Jeffrey Arnett, a developmental psychologist, describes emerging adulthood as a distinct developmental

stage from late adolescence to early adulthood, with an emphasis on ages 18 to 25, during which individuals are recipients of healthcare transition services (Arnett, 2000).

Throughout these critical years, individuals are exploring their identity, experiencing a great amount of life changes, and developing self-reliance skills. Therefore, emerging adulthood is a crucial time when healthcare providers should strive to educate and better engage adolescent patients, especially those with SHCNs, about their long-term medical management requirements.

Encouraging young people with SHCNs to develop self-management skills can be a daunting task, but fostering such skills in emerging young adults can greatly improve confidence and health-related behavior in adulthood (Harvey et al., 2008).

Unfortunately, the development of innovative services to address the lifestyle and psychosocial needs of emerging adults with SHCNs has not kept pace with other achievements in medicine that have afforded these individuals longer lives (Betz, Lobo, Nehring, & Bui, 2013). According to Betz et al. (2013), the voice of young adults with SHCNs has not been effectively addressed, and

few studies [have] examined the services and/or supports needed [by those] acquiring the health-related self-management knowledge and skills needed to become self-sufficient with special health care needs management... or learning the skills needed to access community-based transition and adult services and programs (p. 333).

According to the World Health Organization's revised definition of health in an article by Huber et al. (2011), health is no longer the "state of complete physical, mental, and social well-being" (p. 1) but rather "the ability to adapt and self-manage" (p. 3) in the

face of social, physical, and emotional challenges. The ability to self-manage has been described by Barlow, Wright, Sheasby, Turner, and Hainsworth (2002) as one's "ability to manage the symptoms and the consequences of living with a chronic condition, including treatment, physical, social and lifestyle changes" (p. 178) with the support of family, community, and healthcare professionals. Considering the impact of self-management on one's overall health, fostering these skills in emerging adults is critical and may encourage individuals to comply with follow-up appointments and management plans. This increased compliance could then improve long-term health outcomes.

The transition of adolescents with SHCNs into adulthood was first identified as an area for improvement by a consensus statement co-authored by various stakeholders in 2002 (American Academy of Pediatrics, American Academy of Family Physicians, American College of Physicians, & American Society of Internal Medicine, 2002). An update on this topic was provided in the most recent clinical report authored by the AAP et al. (2011). In this report, various professional organizations commented on the importance of patient medical homes and provided practicing medical professionals with an algorithmic protocol that is intended to help patients succeed in their transition to adult-centered care. Elements of this protocol include timely initiation of the transition planning process; the use of written, adaptable, and up-to-date policies to be discussed with youth and their families; thoughtful care coordination; education on the complexities of self-care; the use of readiness checklists to ensure all age-appropriate transition issues have been addressed; and lastly implementation of formal adult-centered healthcare (AAP et al., 2011).

Various organizations have tried to promote such comprehensive transition services for all youth and families, but high-quality care has not yet been realized. For instance, many pediatric practices are not initiating transition planning early in adolescence and many providers find that youths with SHCNs are often not prepared to be their own, independent health advocates (AAP et al., 2011). Peter, Forke, Ginsburg, and Schwarz (2009) explored internists' opinions regarding transition in a recent study and identified patient maturity, patient psychosocial needs, and seamless coordination of the transition process as major areas of concern. Such insight into the perspective of internists, who will be the primary medical care providers for many young adults with chronic conditions, highlights the elements of transition that require greater attention and underscores the significance of a smooth, effective transition (Peter et al., 2009).

Incorporation of high-quality transition support by both pediatric and adult primary care practices into the medical home is also emphasized in the AAP's most recent clinical report (AAP et al., 2011). Additionally, the AAP's clinical report (2011) intended to "provide a structure for training, continuing education, and research to further the understanding of best practices for transition of adolescents to adult care" (p. 182). By providing a well-delineated decision-making algorithm and clearly defining the various steps providers must take to achieve transitional success with patients, this report serves as a useful guide for providing appropriate transitional support to patients, regardless of their specific SHCNs (AAP et al., 2011).

One group of individuals who would benefit from an enhanced transition experience is young adult women with Turner syndrome (TS). TS is a chromosomal disorder in which a female is born with the complete or partial absence of one of the two

X chromosomes. TS affects approximately 1 in 2,500 females worldwide (Stochholm, Juul, Juel, Naeraa, & Gravholt, 2006). The American College of Medical Genetics recommends that a diagnosis of TS be made by identification of the complete or partial absence of an X chromosome in females by a standard 30-cell karyotype, which can detect a minimum of 10% mosaicism (Wolff, Van Dyke, & Powell, 2010). A 45,X karyotype (complete absence of the second X chromosome) is observed in approximately 1-2% of pregnancies, 10% of miscarriages, and 1% of stillbirth. Greater than 99% of 45,X pregnancies result in miscarriage, usually prior to 28 weeks gestation. About 45% of women with TS possess a 45,X karyotype, and the remainder of women with TS may have mosaic cell lines with or without structural abnormalities involving the X chromosome (Wolff et al., 2010). These cytogenetic results are always associated with short stature and often accompanied by other phenotypic features characteristic of TS, including endocrine deficiencies, kidney problems, cardiovascular issues, and reproductive challenges such as premature ovarian failure and infertility. Most girls and women with TS have normal intelligence, but developmental delays and nonverbal learning disabilities are possible and vary from person to person (Saenger, 1996; Sybert & McCauley, 2004).

Ongoing medical monitoring for women with TS is critical, as they are at risk for a plethora of medical issues including cardiovascular complications such as aortic dilation or dissection, atherosclerosis, and hypertension. Other comorbidities may include hypothyroidism, high cholesterol, vision and hearing loss, and osteoporosis (Bondy, 2007; Freriks et al., 2011).

Although fertility preservation in TS is an evolving area of research that may allow women to conceive with their own oocytes, women with TS who become pregnant may still encounter increased risk of various maternal and fetal complications (Bodri et al., 2006). Chevalier et al. (2011) suggest that these women are at an increased risk for cardiovascular events during pregnancy with oocyte donation, specifically hypertensive disorders. Other studies also show there is a 2% risk of pregnancy-associated death and aortic dissection in women with TS who have pursued pregnancy with assisted reproductive technologies (Karnis et al., 2003). Considering the significance of the associated long-term health problems women with TS may experience across their life span, it is critical that these women receive appropriate education about their health risks.

Carolyn Bondy (2007) authored a paper for the Turner Syndrome Consensus Study Group that discussed the proceedings of a multidisciplinary conference in April 2006. Various practice guidelines that were developed from peer-reviewed scientific publications and expert opinions are outlined in this paper. Specifically, issues regarding diagnosing TS, cardiovascular disease, growth and development, psychological and educational needs, and TS in adulthood are emphasized as areas in need of attention. This paper serves as the most up-to-date set of guidelines for clinical practice in the care of patients with TS throughout the lifespan and is considered a pivotal piece of literature for genetics practices.

The guidelines state that all patients with TS should receive screening for a variety of health concerns at the time of diagnosis, but that equally important are interval screenings for additional health issues that may develop with age. For example, all patients with TS should see an ENT and audiologist regularly, and older girls/adults

should be monitored for a variety of health conditions including, but not limited to, celiac disease, thyroid disease, liver disease, and psychosexual adjustment.

The guidelines also include recommended intervals for cardiac imaging. First, all patients should have a formal cardiac evaluation at the time of diagnosis: MRI and echocardiogram for adults/older girls and echocardiography for infants/young girls. Reevaluation, according to Bondy (2007), should occur at “timely occasions, *e.g.* at transition to adult clinic,” (p. 13) and at a minimum of every 5 to 10 years for those with apparently normal heart health. Patients with a personal history of heart disease should be treated and followed as determined by an experienced cardiologist (Bondy, 2007).

With regards to the healthcare transition process, Bondy (2007) recommends a protocol that consists of a gradual, staged process beginning around the age of 12. Additionally, Bondy (2007) states “the health care focus also shifts from maximizing height to inducing feminization, counseling the adolescent with TS about the evolving impact of her condition into adulthood, and promoting the development of independent self-care behaviors” (p. 18). The responsibility is placed on the pediatric endocrinologist to engage the emerging adult and to collaborate with her new providers in order to ensure the patient will continue to receive optimal care as an independent adult (Bondy, 2007).

Dr. Karen Rubin (2008) added another element to this discussion by elaborating on how the transition to adult-centered care should be a staged process initiated during early adolescence. At that time, the patient can voice her opinion to healthcare providers and her parents and develop confidence in her ability to be an equal partner in her own care. By developing a transition checklist and completing a “TS Transition Passport,” (p. 780), medical professionals can help better facilitate this process and provide patients

with customized and supportive resources (Rubin, 2008). Considering their need for lifelong care into adulthood, it is essential that women with TS have good relationships with their medical providers and a full understanding of their condition, as confidence in one's transition will undoubtedly improve long-term health and quality of life (Davies, 2010).

Despite the efforts to improve healthcare transition for all individuals with SHCNs, many patients, including women with TS, are lost to follow-up after being discharged from their pediatric care providers. In one study, assessment of adequacy of medical follow-up in a cohort of women with TS revealed that only 3.5% of patients had undergone medical follow-up in accordance with international recommendations (Devernay, Ecosse, Coste, & Carel, 2009). An Australian study of 39 women with TS revealed that only 63% of women received regular follow-up and only 44% of women had adequate health surveillance even though 87% of these women had one or more medical concerns in addition to their TS diagnosis (Pedreira, Hameed, Kanumakala, & Zacharin, 2006). Furthermore, a recent observational study at a multidisciplinary clinic for adult women with TS determined that one third of their study participants lacked any form of ongoing specialty care (Freriks et al., 2011). This study also recognized that multidisciplinary evaluation of adult women with TS identified previously undiagnosed morbidities in several women. Thus, women with TS are likely to benefit greatly from a seamless and comprehensive transition from pediatric to adult medical care with the help of a multidisciplinary healthcare team (Freriks et al., 2011).

While several studies have inquired about the perspective of providers and parents of children with SHCNs (Geenen, Powers, & Sells, 2003; McManus et al., 2013; Peter et

al., 2009), fewer studies have explored the direct opinions, preferences, and attitudes of these individuals. In a recent study conducted in the Netherlands, Jedeloo, van Staa, Latour, and van Exel (2010) performed semi-structured interviews with adolescents to learn more about their preferences for care delivery and self-management. Results identified four distinct preference profiles: (1) conscious and compliant, (2) backseat patient, (3) self-confident and autonomous, and (4) worried and insecure (Jedeloo et al., 2010). This study illustrated that not all adolescents with chronic conditions share common opinions about their medical management and that providers should not assume one type of transition plan fits all. Individualized transition planning is critically important and providers should take time to learn more about patients' preferences for parental and/or provider involvement, desire to learn more about their medical conditions, and ability to engage in healthcare management planning. Furthermore, all participants deeply valued the opportunity to have both a voice and choice in decisions regarding their care (Jedeloo et al., 2010). By recognizing these preference profiles, healthcare providers can achieve a more personalized approach to transition as well as better engage adolescent patients in the conversation about their long-term care needs.

With regards to emerging adult women with TS, much of the same principles can be applied to this patient population. Behm (2014) recently explored which factors influence behavior and psychosocial outcomes of emerging adult women with TS ages 16 to 26. Her study underscores the idea that many women frequently feel misunderstood by their healthcare team and experience insensitivity towards the emotional burden of coping with reproductive challenges. Additionally, healthcare professionals should

develop improved strategies that better address the psychosocial and cognitive needs of this cohort of women (Behm, 2014).

In July of 2014, the inaugural Turner Resource Network (TRN) Symposium brought together an assortment of stakeholders in TS research, including affected individuals and their families, advocacy leaders, clinicians, scientists, and other persons with interest in the wellbeing of women with TS (Backeljauw et al., 2015). The TRN was established following collaborative efforts between the Turner Syndrome Society of the United States (TSSUS) and the various professionals that comprise the TSSUS advisory board. Ultimately, it was decided that an association such as the TRN was necessary so that greater attention could be paid to the psychosocial and health issues facing women with TS, such as the increased susceptibility to common adult conditions. The TRN initiative is also tasked with trying to expand access to comprehensive healthcare among girls/women with TS and establish a patient-powered program that encourages interdisciplinary care and education for patients and their families (Backeljauw et al., 2015).

Several prominent stakeholders spoke on the aforementioned topics and current TS-related issues throughout the symposium. First, Steven Chernausek discussed future healthcare needs and the role of patient advocates, emphasizing that care should be delivered across the spectrum of ages and that a smooth transition to adult-care is paramount for maintaining appropriate surveillance for comorbidities and optimal delivery of care to women with TS (Backeljauw et al., 2015). David E. Sandberg spoke on the discrepancy between what is recommended versus what is actually achieved with regards to the coordination of multiple specialty services involved in the care of women

with TS. Gary Lorigan reviewed the results of his survey, which asked the question of what is needed to ensure care for patients with TS throughout the transition period. The results of his survey indicated the need for more research into issues and concerns related to TS and easy online access to information and resources for patients. Lastly, Philippe F. Backeljauw shed light on the various barriers for improving health and wellness for women with TS: lack of coordinated specialty care, challenges in identifying adult providers with experience working with patients with TS, and lack of uniform criteria for lifelong screening and care of women (Backeljauw et al., 2015). The plethora of discussions and educational sessions that took place throughout this symposium certainly underscored the importance of this research and illustrated that gaining a patient-perspective is essential and closely aligns with the mission of the TSSUS and other significant stakeholders in TS-related research.

One unique group of healthcare professionals that is well equipped to provide both educational and psychosocial support to these women is genetic counselors. The practice-based competencies for these medical professionals include a variety of domains relevant to support needed by emerging adults with TS: (1) genetics expertise and analysis, (2) interpersonal, psychosocial, and counseling skills, (3) education, and (4) professional development and practice (Accreditation Council for Genetic Counseling, 2013). Some of the more specific skills these professionals must possess include the ability to integrate knowledge of psychosocial aspects of genetic conditions to promote client well-being and to employ active listening and interviewing skills to identify, assess, and empathically respond to emerging concerns. Additionally, genetic counselors are able to effectively educate clients about a wide range of genetics and genomics

information based on patients' needs and indication for referral while also advocating for individuals, families, communities, and the genetic counseling profession (ACGC, 2013).

Considering the expertise and interest of the genetic counseling profession, these individuals should be considered as essential members of integrative healthcare teams for adolescents with SHCNs. As noted above, the literature shows that women with TS feel that they are often times misunderstood by their healthcare providers. Although the parents of many young adults with TS have met and extensively discussed the ramifications of a TS diagnosis with a genetic counselor, gaps in the literature exist concerning how individuals affected by TS have directly been counseled and educated about their condition by a genetic counselor and whether or not such individuals would appreciate having reliable fact sheets on TS written by genetics professionals to give to their future health care providers. Questions such as these as well as others that assess these women's confidence in their understanding and knowledge of TS and self-management skills are all important areas that should be addressed.

The purpose of this study was to find answers to several of these concerns and to invite young adult women with TS to reflect upon their healthcare transition experiences. Additionally, it explored the opinions of this population regarding meeting with healthcare professionals such as genetic counselors during the transition process and prior to their final transfer to adult care. By providing women with TS a voice through participation in this study, the principal investigator hoped to shed light on the various support needs of these women and their motivations for pursuing self-management and establishing themselves as autonomous individuals.

Chapter 2: Reflections on the Current State of Healthcare Transition for Young Adult Women with Turner Syndrome: Strategies for Facilitating Autonomy and Self-Management¹

2.1 Abstract

The transition to adult-centered healthcare is a critical period for emerging adults, especially those with special healthcare needs (SHCNs). Considering the ongoing medical monitoring necessary for women with Turner syndrome (TS), it is essential that the transition process be comprehensive and well-coordinated. The aims of this study were to invite young women with TS to reflect on their healthcare transition experiences, to explore participants' perceived control of their medical management, and to identify ways in which genetic counselors can be involved in multidisciplinary healthcare teams. The hypotheses were that young women with TS are motivated to learn more about their diagnosis, benefit from counseling on how to develop self-management skills, and believe it would be useful to speak with a genetic counselor as adolescents.

Study participants included twenty-two women between the ages of 18 and 30 years with a diagnosis of TS. A mixed method study design was used, consisting of quantitative data collection via an online survey tool and follow-up qualitative interviews. Interviews were transcribed and reviewed for major themes using data driven coding and subsequent exploratory thematic analysis. Results demonstrated that participants who received counseling on the development of self-management skills as adolescents were

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more confident, independent, and satisfied with their transition to adult-centered medical care than participants who did not. Six major themes were identified in participant interviews. These included diagnosis disclosure, TS education, genetics knowledge, transition support, patient autonomy, and multidisciplinary care.

In conclusion, young adult women with TS are highly motivated to achieve autonomy in their healthcare and would appreciate being given additional information about their condition at a younger age. Healthcare providers should carefully consider patient preferences in order to provide patients with optimal support and appropriate resources. New recommendations for clinical practice should incorporate involvement of genetic counselors in the multidisciplinary healthcare teams of girls and women with TS.

2.2 Introduction

Evidence-based recommendations for the quality and standard of care for women with TS are limited; thus, the majority of guidelines have developed from the opinions, perspectives, and judgments of experienced health professionals. Proper care for patients with TS is extensive, encompassing screening for and management of a great variety of organ system comorbidities including cardiovascular and renal complications, thyroid disease, hearing loss, metabolic bone disease, infertility, and reproductive issues, as well as the provision of growth hormone therapy, puberty induction, and psychosocial counseling (Bondy, 2007). Considering the complex nature of TS, an integrative, multidisciplinary approach to the healthcare of these women is most appropriate.

According to Boon, Verhoef, O'Hara, Findlay, and Majid (2004), the working definition of integrative healthcare emphasizes the process of utilizing a collaborative team approach guided by consensus building, mutual respect, and a shared vision of

healthcare. This ensures that each healthcare provider and the patient are able to contribute their particular knowledge and skills to patient healthcare plans (Boon et al., 2004). Since transition management is such a critical component of healthcare planning, an integrative healthcare approach should continue to be employed during transition.

It is recommended that the transition to adult-centered healthcare occur during late adolescence and as part of a staged process beginning around the age of 12 (AAP et al., 2011; Rubin, 2008). In the most recent report on clinical practice guidelines by Carolyn Bondy for the Turner Syndrome Consensus Study Group (2007), the importance of counseling the adolescent about her condition and its evolving impact on her health into adulthood is explicitly emphasized as an essential component of the healthcare transition process in order to promote self-management and autonomy. By having this conversation in early adolescence, emerging young adults, ages 18 to 30, should feel empowered and confident in their skills.

Given the TS Consensus Study Group's emphasis on having the emerging adult and healthcare providers collaborate in order to ensure careful, continual, and coordinated healthcare, there is a demand to address whether or not young adult women with TS feel confident in their healthcare and understanding of their specific medical needs (Bondy, 2007). Patient education is extremely important, especially during the transition to adult-centered care, as individuals must feel confident in their ability to self-manage and communicate with health professionals about their health status (Ackerman & Bamba, 2014). Additionally, helping emerging adults with TS develop trusting relationships with their healthcare providers as they transition to adult-centered care is a critical step that can enable these young women to feel more supported and better understood (Bondy,

2007). Studies have demonstrated that adolescents who receive effective counseling and support during their healthcare transition are more confident in their self-management skills (Londoño & Schulz, 2015; Taylor, Edwards, & Ku, 2006; Wells, McMorris, Horvath, Garwick, & Scal, 2012). However, the current lack of such a consistent and comprehensive healthcare transfer for women with TS is an issue that must be addressed, and patient needs must be identified accordingly.

The objectives of this study were to explore participants' perceived control of healthcare transition, to identify existing resources and educational needs, to evaluate access to genetic counseling, and to develop strategies for facilitating self-management of long-term healthcare needs in young adult women with TS. The study hypotheses were that young adult women with TS:

1. Are motivated to learn more about their diagnosis and specific long-term healthcare needs, especially during the transition process from pediatric to adult-centered medical care; and
2. Benefit from counseling on how to establish appropriate self-management skills as emerging young adults; and
3. Believe it would be useful to speak with a genetic counselor as adolescents in order to identify resources relevant to their healthcare needs and to discuss psychosocial issues of developing patient autonomy.

2.3 Materials and Methods

This study utilized a mixed methods approach involving a quantitative online survey tool and qualitative interviews for data collection. The University of South

Carolina Institutional Review Board approved this study prior to its implementation (see Appendix B).

2.3.1 Participants. Participants were women between the ages of 18 to 30 years who had a formal diagnosis of TS, were English speaking, and had access to the Internet. There were no restrictions regarding geographic location, ethnicity, marital status, or educational background. Participants were excluded if their age fell outside our specified age range and if they did not complete the majority of the survey. Participants were identified through a purposive sampling method, in which invitation letters were included in newsletters that were emailed to members of the Turner Syndrome Society of the United States (TSSUS) and posted on the research page of the Turner Syndrome Foundation (TSF) website. After reading the invitation letter, individuals self-selected to be study participants by accessing the online survey tool, answering the majority of our survey questions, and either agreeing or refusing future contact for a follow-up interview.

2.3.2 Pilot study. The principal investigator (PI) developed the survey tool using SurveyMonkey™ after identifying major themes in the literature on the topic of transition issues for women with TS. Because this was an original research project and the survey tool had not yet been validated, a brief pilot survey was included as part of the ongoing research process to evaluate the quality of survey questions asked. Five individuals with TS completed the pilot survey one month prior to the official data collection period. These women provided constructive feedback to the PI via email responses and phone conversations. Adjustments were made to clarify misunderstandings, remove repetitive questions, and eliminate confusing or insensitive language. The PI made the necessary adjustments to the survey tool, and the committee approved of the final version.

2.3.3 Quantitative data collection and statistical analysis. Participants completed a nationally distributed online survey tool that was generated through SurveyMonkey™ and was kept open for official participant recruitment for a total of five months from September, 2015, to January, 2016. Survey content included multiple-choice questions, open-ended questions, and questions that assessed level of agreement to various statements along a Likert measurement scale: Strongly Agree (6), Agree (5), Somewhat Agree (4), Somewhat Disagree (3), Disagree (2), and Strongly Disagree (1). Each response option was assigned a numerical value, as indicated by the number in parentheses, so that quantitative scores could be computed. A power analysis was performed before survey data collection in order to determine the sample size needed to reveal statistically significant results in the quantitative portion of the study. The goal sample size was 24 participants, but the PI adjusted the statistical analysis approach accordingly following the completion of data analysis.

All quantitative analyses were performed using Microsoft Office Excel 2013 software and Statistical Package for Social Sciences (SPSS) version 23.0. Considering the survey tool included a variety of questions that explored participants' opinions and feelings towards similar topics, principal component analyses (PCAs) were conducted in order to reduce the data into single 'artificial' variables that would account for the majority of variance in participant responses. More specifically, PCAs were performed to establish a Perceived Confidence Score (Table 2.1), Transition Coordination and Satisfaction Score (Table 2.2), and Level of Independence Score (Table 2.3) for each survey participant. Typically, a minimum of five to ten cases per variable is necessary for performing a PCA; considering this study's small sample size of 22 participants, it was

determined that a total of four variables would be factored together to represent each ‘artificial’ variable.

Questions #20, #21, #22, and #26 of the survey tool were used to establish Perceived Confidence Scores for participants. These data elements were determined to be factorable following identification of a Cronbach’s alpha of 0.769 that justified the reliability of these four variables, an overall Kaiser-Meyer-Olkin (KMO) measure of 0.71, and a statistically significant ($p < .0005$) Bartlett’s test of sphericity. Following inspection of the scree plot and component matrix generated by SPSS software, it was determined that one component would be appropriate to use.

Prior to conducting independent samples t-tests to evaluate for group differences, two additional PCAs were performed in order to establish a Level of Independence Score as well as a cumulative Transition Coordination and Satisfaction Score for participants. Similar methods as mentioned previously were employed to determine which factors (i.e. survey questions) could be reduced into a single, representative principal component for each of these new ‘artificial’ variables (i.e. Level of Independence Score and Transition Coordination and Satisfaction Score).

Questions #23, #24, #27, and #40 of the survey tool were used to establish a Level of Independence Score for participants. These elements were determined to be factorable following identification of a Cronbach’s Alpha of 0.769 that justified the reliability of these four variables, an overall KMO measure of 0.715, and a statistically significant ($p < .0005$) Bartlett’s test of sphericity. SPSS software generated a scree plot and component matrix and provided justification for a single principal component to be used as each participant’s Level of Independence score.

Questions #39, #31, #32, and #35 of the survey tool were used to generate a Transition Coordination and Satisfaction Score for participants. They were determined to be factorable following identification of a Cronbach's Alpha of 0.921 that justified the reliability of these four variables, an overall KMO measure of 0.74, and a statistically significant ($p < .0005$) Bartlett's test of sphericity. SPSS software generated a scree plot and component matrix and provided justification for a single principal component to be used as each participant's Transition Coordination and Satisfaction Score.

Group differences with regards to perceived confidence, satisfaction with the transition to adult-centered care, and level of independence were assessed according to whether or not participants received counseling on the development of self-management skills as adolescents. Independent samples t-tests were utilized to determine whether participants who received counseling on the development of self-management skills as adolescents (Table 2.8, question #68) reported significantly higher perceived confidence, level of independence, and satisfaction with how well they were transitioned to adult-centered healthcare than those who did not receive adolescent counseling. These variables were assessed on a continuous scale of agreement and represented by participants' Perceived Confidence Score, Level of Independence Score, and Transition Coordination and Satisfaction Score. Independence of observations was assumed since participants could only be placed in one category or the other – agreed or disagreed with receiving counseling as an adolescent. Other assumptions that were addressed when analyzing the data included handling any outliers in the two groups, establishing a normal distribution of dependent variables for each group, and evaluating homogeneity of variances. These assumptions were assessed using SPSS software techniques, including

checking for normal distributions with a Shapiro-Wilk's test, which is appropriate for smaller sample sizes, and exploring the nature of descriptive statistics by creating a boxplot that would check for any outliers.

All test assumptions for the investigation of counseling group differences in Level of Independence Scores were met except for the absence of significant outliers and homogeneity of variances. Levels of Independence Scores for each group were normally distributed, as assessed by Shapiro-Wilk's test ($p > 0.05$). Participant #1's Level of Independence Score of 1.75 was identified as an outlier upon inspection of a boxplot. In order to correct for this result in an appropriate manner, her score was adjusted and replaced with the next lowest Level of Independence Score, 2.25. Since the assumption of homogeneity of variances was violated, results were interpreted from a modified t-test that SPSS software produced. This modified t-test, or unequal variance t-test, is able to accommodate unequal variances while still delivering valid test results.

All test assumptions for the investigation of counseling group differences in Transition Satisfaction and Coordination Scores and Level of Independence Score were met, except for the homogeneity of variances. The two score measurements for each group were normally distributed, as assessed by Shapiro-Wilk's test ($p > 0.05$). Since the assumption of homogeneity of variances was violated, results were interpreted from a modified t-test for the two score measurement, which SPSS software produced.

Considering the exploratory nature of this study, various descriptive statistics extracted from survey responses were also incorporated in the data analysis process. The frequency of responses and percentages of participants who agreed or disagreed with

various elements of the survey tool provided valuable information regarding participant preferences' and opinions about their healthcare transition experiences.

2.3.4 Qualitative data collection and analysis. Following official completion of survey data collection, the PI implemented the qualitative portion of the study, which involved 20-minute interviews with participants who agreed to follow-up upon completion of the survey. Interviews took place during the month of February, 2016. The list of interview questions used to guide the direction of participant interviews has been included in Appendix A. Interviews were either conducted using the Adobe Connect web conferencing software or by phone at The University of South Carolina Specialty Clinic, which provided a secure, firewall-protected network to ensure study participants' confidentiality. A video component to these interviews was intended to enhance participants' engagement and commitment to the research project; however, only one participant agreed to this interview style. Participants were offered the alternative of having telephone conversations with the PI in lieu of virtual interviews if they were uncomfortable with or unable to use the video feature; this was the most used interview technique. Interviews were recorded so that the information exchanged between each participant and the PI could be transcribed for later analysis. Participants were not identified by name in these transcriptions to ensure the anonymity of their responses. All 12 participants who consented to later interviews were contacted to allow for an unbiased selection of interview participants and increased likelihood of reaching data saturation. The final qualitative sample consisted of five participants who were successfully interviewed using a semi-structured interview style, guided by questions derived from the literature and informed by preliminary quantitative survey results (Appendix A).

An inductive thematic analysis of qualitative interview data was performed using data driven coding in order to determine the relationship to a priori hypotheses. Subsequently, interviews were reviewed to determine the preference profile for care delivery and self-management for each interview participant. Four care-delivery and self-management preference profiles derived from the work of Jedeloo et al. (2010) were used in this segment of the qualitative data analysis: (1) conscious and compliant, (2) backseat patient, (3) self-confident and autonomous, and (4) worried and insecure. Data were kept organized, coded, and analyzed for exploratory key themes using Excel.

2.4 Results

2.4.1 Quantitative analysis.

2.4.1.1 Demographics. By the end of the survey data collection period, 35 women had completed the survey. Twenty-two of these women met all of the inclusion criteria for participation in the study. Participant demographics are illustrated in Table 2.4. Half of the participants were between the ages of 27 and 30 (50%, $n = 11$). The majority of participants identified as Caucasian (91%, $n = 20$) and most women reported their marital status as single, never married (82%, $n = 18$). Other noteworthy demographic elements included participants' reported time of receiving their diagnosis of TS: Before birth (14%, $n = 3$), soon after birth as a newborn (46%, $n = 10$), between the ages of one and five (9%, $n = 10$), between the ages of six and 11 (5%, $n = 1$), and between the ages of 12 and 18 (27%, $n = 6$). No participant had received her diagnosis of TS as an adult or after the age of 18. While four participants reported meeting with a geneticist and one participant reported being followed or seen by a genetic counselor in the past, no participants indicated they were currently being seen routinely by a genetics professional.

2.4.1.2 Participant's reported level of motivation. In order to assess the hypothesis that women with TS are highly motivated, several different survey questions that were intended to measure motivation were included in the survey tool. Seventeen participants agreed that they are highly motivated to manage their own healthcare and medical needs, while four participants disagreed ($n = 21$) (Table 2.6). Furthermore, all 22 participants agreed that they would like to learn more about TS and what implications their diagnosis might have for their future healthcare management (Table 2.6 and Table 2.8). With regards to the healthcare transition process, all 22 participants agreed with wanting additional resources and supports, and all participants also agreed that they wished they had additional resources to provide their healthcare providers regarding TS and associated medical needs as emerging adults transitioning to adult-centered care (Table 2.8). These results indicate our population of emerging adult women desire more information and are highly motivated to become independent, autonomous patients.

2.4.1.3 The impact of self-management counseling as an adolescent. With regards to having previously received counseling as an adolescent (between the ages of 12 and 18) on ways to promote independence and self-care behaviors, only 10 participants agreed with this statement while 11 participants disagreed (Table 2.8, question #68). Participants were grouped by whether they (1) agreed or (2) disagreed with having received such counseling in order to determine if there were group differences with regards to confidence, level of independence, and transition experiences.

There was a statistically significant difference in mean Perceived Confidence Scores between those who agreed with receiving adolescent counseling on developing self-management skills (score = 4.93, $n = 10$) and those who did not (score = 3.59, $n =$

11). Those who received counseling had higher scores than those who did not: $M = 1.34$, 95% CI [0.52 to 2.15], $t(17.29) = 3.446$, $p = 0.003$. In other words, participants who reported receiving adolescent counseling had scores that were 1.34 points higher than those who did not receive counseling, which suggests that these participants have higher levels of perceived confidence in their healthcare as adults (Figure 2.1).

There was a statistically significant difference in mean Transition Coordination and Satisfaction Scores between those who agreed with receiving counseling as adolescents (score = 3.92, $n = 9$) and those who did not (score = 2.73, $n = 11$), with those who received effective counseling having higher scores than those who did not: $M = 1.19$, 95% CI [0.02 to 2.41], $t(16.87) = 2.143$, $p = 0.047$. In other words, participants who reported receiving counseling as an adolescent had scores that were 1.19 points higher than those who did not receive counseling, indicating that these participants experienced greater satisfaction with the coordination of their transition process (Figure 2.2).

Lastly, there was a statistically significant difference in mean Level of Independence Scores between those who agreed with receiving adolescent counseling (score = 4.95, $n = 10$) and those who did not (score = 3.8, $n = 11$), with those who received counseling having higher scores than those who did not: $M = 1.15$, 95% CI [0.09 to 2.22], $t(16.62) = 2.285$, $p = 0.036$. In other words, participants who reported receiving counseling as an adolescent had scores that were 1.15 points higher than those who did not receive counseling, indicating that these participants have feelings of greater independence in their healthcare (Figure 2.3). A side-by-side comparison of participants' confidence, transition satisfaction and coordination, and level of independence scores can be found in Figure 2.4.

2.4.1.4 Existing support during the healthcare transition process. Another area of interest included participants' preferences and opinions regarding their support needs and access to resources during the transition to adult-centered care. While the majority of participants agreed that their healthcare transition was well coordinated ($n = 13$), eight participants disagreed, with four participants strongly disagreeing that their transition was well coordinated ($N = 22$). Furthermore, 10 participants reported that their healthcare providers began to shift their attention away from their parents and ask them questions directly after the age of 18, while only two participants were addressed directly at the ages of 12 and 13 ($N = 22$). Finally, approximately half of the participants ($n = 12$) reported being satisfied with how they were transitioned to adult-centered care. A summary of these findings as well as additional related results can be found in Table 2.5.

2.4.1.5 Healthcare transition education needs. Participants had mixed opinions regarding the extent of counseling and education they received as emerging adults. The large majority of participants ($n = 15$) reported being well educated about future health problems they may face as an adult with TS, yet 10 participants did not feel that they had been fully counseled about how their condition would affect them in adulthood ($n = 21$). Interestingly, nine participants had not yet been educated on the topics of fertility and reproductive options for women with TS at the time they completed the survey ($n = 21$). Additionally, the large majority of participants ($n = 20$) indicated that they wish to learn more about the genetic principles underlying TS, yet 18 participants had never been offered genetic counseling ($n = 21$). These findings as well as additional related results are summarized in Table 2.6.

2.4.1.6 Access to and involvement of multidisciplinary healthcare teams. With regards to feeling like their voice is being heard, a large majority of participants ($n = 18$) agreed that their healthcare providers actively listen and respond to their healthcare needs and concerns ($n = 21$). In reference to this particular study cohort, healthcare providers reportedly serve as advocates in the following areas: personal life ($n = 7$), health insurance ($n = 6$), school and education ($n = 4$), and work and employment ($n = 2$). Yet, 11 participants reported that their healthcare providers do not serve in areas outside of their immediate medical needs ($n = 21$). Fifteen participants appreciate receiving care from multiple specialists, and each of these participants agreed that by receiving multidisciplinary care, they were better prepared to transition to adult care ($n = 21$). While participants may be receiving multidisciplinary care, only eight agreed that their providers work as a team ($n = 21$). These findings as well as additional results are summarized in Table 2.7. Additional data collected through survey responses can also be found in Appendix A.

2.4.2 Qualitative analysis

Participants were asked a series of open-ended questions that expanded on the survey questionnaire topics specifically pertaining to the healthcare transition experience (see Appendix A). Data driven coding and subsequent exploratory thematic analysis identified themes as areas of concern to participants surrounding six key elements of the healthcare transition process specifically related to TS: (1) diagnosis discovery and disclosure, (2) diagnosis education and counseling needs, (3) exposure to and knowledge of genetics, (4) important elements of patient autonomy, (5) healthcare transition barriers and challenges, and (6) support needs and opinions of multidisciplinary care clinics.

2.4.2.1 Demographics. Of the 22 survey respondents that met inclusion criteria, 12 participants agreed to be contacted for a follow-up interview at a later date. Of the 12 participants who were contacted via email to be interviewed, five participants responded and completed the qualitative interviewing portion of the study. Of the five participants interviewed, one participant successfully completed a virtual interview, while the other four participants chose to be interviewed by phone.

2.4.2.2 Diagnosis discovery and disclosure. The majority of participants could not remember the exact age at which their diagnosis was disclosed to them or at what age they were first educated on what TS is and the reasons TS can happen. As one participant stated, *“I honestly don’t remember. Growing up, I always knew that I had TS. I don’t remember exactly how I found out the reason, or cause, behind TS. It’s just been something that I’ve always known.”* Three of the five participants were diagnosed as newborns and each explained how they have always been aware of their condition, yet none of these women could pinpoint when they were educated about TS. One participant said her understanding came with age: *“I would say that as I was getting older, I was explained more about it. So I guess at various stages, but I guess more as I was older and over time.”* Another participant expressed regret in not trying to learn more about TS at a younger age: *“That is one of my major regrets, not doing my research and not looking into it... It just never really mattered to me until I got to be like a teenager... it just started to be more noticeable.”*

Two participants who were 12 years old when they were diagnosed with TS commented on how their shorter stature or delayed pubertal development contributed to the discovery and subsequent disclosure of their diagnosis:

I was twelve. I didn't know anything was different about me until I reached the age when every other girl was developing and I wasn't right around the 7th grade. So that's kind of how my parents noticed it even though I was clueless. So they brought me to an endocrinologist and confirmed it at twelve.

The other participant also spoke about a similar experience:

I was 12 years old and hadn't gotten my period yet, so I went in to see a doctor and that's kind of how they found out because I was really short as well...that was kind of the red flag because from the outside you couldn't tell there was anything wrong.

With regards to how each of these participants was educated about TS and how their diagnosis was disclosed, each expressed similar feelings of not fully understanding. One woman stated, *"At 12 years old, I didn't know a whole lot. To be honest with you, when they explained the whole missing part of an X chromosome, I thought I was a dude. Like I really didn't understand at first."* Another participant commented that she felt overwhelmed at the time of her diagnosis and did not feel she was fully informed even though she could have handled the information: *"It was kind of a lot. I wouldn't say that I was educated as much as I should have been... I feel like I could have gotten more information, and I could have been involved more in the situation."*

2.4.2.3 Diagnosis education and counseling needs. Most participants expressed that it would have been helpful to receive a greater amount of education about TS at an earlier age. One participant stated,

Yeah, I think I would have benefited from [genetic counseling] more when I was younger. Because, as I said, I was so clueless when I was 12. I thought they

were trying to tell me I was somehow genetically a guy. It didn't register. I wish probably I had [a genetic counselor] at 12, but now I understand.

Another participant commented on how she feels confident in her understanding of TS at her current age (26-years-old) but that additional education at a younger age would have been valuable: *"I mean I feel like at this point... I do know a lot. It just would have been more important like during the growth process and like back when I could have had growth hormone and stuff."* She remarked that she would have appreciated having and could have comprehended additional information about TS *"...around age 8 or 9, before going through puberty."* A third participant, who also felt that she has a good understanding at her current age (20-years-old), explained, *"...it would have been really easy to sit down with a genetic counselor or someone to learn a lot more...I would be really interested to learn more specifics because I think my family didn't."* A fourth participant commented on how having a better understanding at a younger age would have helped her be more accepting of the diagnosis of TS: *"Yeah, I think it would have helped me be more accepting. I think it was hard to accept it because I didn't really understand [the diagnosis of TS]... I think with more knowledge, it would have been easier."*

Some participants also shared their opinions regarding their healthcare providers understanding and knowledge of TS. Participants seemed to have mixed feelings with regards to how much their providers understood about their diagnosis. One participant who expressed having to educate several of her healthcare providers stated,

We just kind of had to do our own research and say, people with TS have these things. But even then, no matter how good they were... most of them hadn't

[worked with TS]. And the major thing, with regards to cardiac anyway, is everyone needs a baseline cardiac MRI. And then every 5 years after that, so you have to explain that kind of stuff to a practitioner who has actually never worked with someone with TS.

In contrast, another participant, who was very appreciative of the care and knowledge her pediatric healthcare providers demonstrated, had the following to say about returning to see that same provider as a young adult: *“Yeah, he is awesome. And during the break, I didn’t go see him for maybe 3 or 4 years, but when I went back he was very supportive and knew exactly where to pick back up in my care.”*

2.4.2.4 Exposure to and knowledge of genetics. Participants also seemed to have a discrepant amount of exposure to providers who were knowledgeable about genetics and received little general education on the genetics of TS throughout their healthcare experiences. Those who admitted that they were unaware of the field of genetic counseling or who were not familiar with genetics seemed to lack an understanding of the etiology of TS (meiotic or mitotic nondisjunction rather than a mutation) and how their personal genetic make-up (genotype) influences how their condition manifests (phenotype). For example, one participant stated, *“They told me that I was a mosaic. Which, I guess, they define as like someone who doesn’t totally have all of the features... whatever that is supposed to mean?”* Another participant expressed, *“I guess that’s one thing that I would be curious about. I mean what causes the mutation?”* and later admitted that she was uncertain about what having the mosaic form of TS meant exactly: *“I don’t really know. I think it’s kind of like having an X and a half or like having one X and the other is curved or something. I don’t know;”* she also could not recall if genetic

counseling had been a service that was offered to her at a particular age. Another participant who was unfamiliar with the field of genetic counseling and had less of a genetics background had the following to say:

I feel like [genetic counseling] would have helped with my understanding of TS. I definitely felt very isolated growing up.... I think having a better understanding of why this was going on would have helped with those feelings of isolation at a younger age. Because I knew I had it, and that was about it.

One participant, who was more familiar with genetics and genetic counseling than some of the other participants, also stated that she strongly wished she had received such counseling about her specific condition:

We did want to follow up with a genetic counselor before I left [anonymous hospital system] but my mom made a call and they never got back... I think it's something we're still curious about. We want to learn about new technology and learn a couple of more things and more specifics, so we'll probably find someone outside of [anonymous hospital system].

2.4.2.5 Important elements of patient autonomy. A recurrent topic of conversation was overprotective parental involvement. One participant expressed wishing her parents would have 'let up' at the time of her diagnosis at age 12:

I can't really blame my mom and my dad for taking it as seriously as they took it and for being as overprotective as they were. But I guess if there were one thing I could say to parents it would be ease up. I mean let your daughter tell you what she wants because I mean even at that age I still knew what I wanted.

Another participant spoke of her mom's continual involvement in her healthcare, especially throughout the transition process: *"I mean other than my mom, yeah she was pretty much my transition coordinator if you will. She was pretty much the only person I had as far as that goes."* This participant also described her mother as her primary source for genetics education: *"My mom has tried to explain the mosaic part of things to me, you know, and that there are things in common with traditional TS and mosaic TS, but that there are also specific things for mosaics too."* A different participant expressed frustration with having her opinions about her treatment plan dismissed by her parents:

My parents would book me a doctor's appointment, and my mom asked me about [growth hormone]: 'What do you think? Do you want to do it?' So she was kind of asking for my input... but my parents didn't want me to have it [the growth hormone treatment].

Another key issue was how motivated participants were to be involved in their healthcare or how strong of a voice they would have appreciated having while they were transitioning to adult-centered healthcare. For instance, one participant commented,

I mean I wish they had talked to me instead of my mom and dad because at the time they were still with me. It wasn't until I was probably 18 or 19 that I started to go to those kinds of visits on my own and have a better comprehension of what was really going on and what the best options were for me.

Another participant expressed similar feelings towards wishing she had been included in shared-decision making regarding her healthcare: *"I wish I had been involved in the decision first and maybe I would have been able to learn a little more."*

Finally, several participants made comments regarding being their own self-advocate and how they needed to develop the necessary maturity and confidence that comes with acting more autonomously in one's own healthcare decisions. One participant provided the following personal perspective of the TS community:

We are some of the most resilient people you'll ever meet. That is absolutely what I've learned. Because I mean you have to be strong. I mean you are faced with a lot of heavy decisions at a pretty young age, most of us. So you have to mature kind of quickly to understand the best way to make them. That's probably the one word I would use is resilience. That is how I would describe how everyone going through this would have to feel after all that.

Another participant commented on feeling more empowered and independent with age and with adjusting to not having her parents in the room:

Having a more open conversation with my doctors has been nice. I know growing up I sometimes felt that with my parents in the room there were some things I might not have been able to ask. But as the adult in charge, I feel like I am able to ask more questions openly.

Yet another participant simply commented, *"Definitely. I enjoy having more autonomy."*

2.4.2.6 Healthcare transition barriers and challenges. All participants shared their opinions regarding healthcare transition barriers or other challenges they have faced throughout their healthcare encounters. Geographic location was identified as either inhibiting or facilitating the transition process. A few participants spoke highly about their geographic location. For instance, one participant stated, *"Looking back, I can't*

believe how lucky I was to be here in [anonymous city] with this clinic.” Another participant who was extremely grateful of her pediatric care in [anonymous city] stated,

[Anonymous city] has some of the best hospitals in the country... it was the best place that this could have happened to me because I was really surprised. I mean there were a lot of doctors. I had choices. They were all just as educated.

She later shared her appreciation for having a local TSSUS chapter in [anonymous city] and being able to meet other girls who also had TS. On the other hand, some participants shared stories of how their location hindered their healthcare transition and experiences.

One participant stated, *“Yeah, transition has never really been a big focus... especially in [anonymous state]. There wasn’t really any sort of transition, so that was sort of difficult for us.”* Lastly, one participant expressed frustration with having to move and find an adult-care provider in a new city:

I am looking for an adult-care provider. I left my pediatric provider just last year, but I moved into [anonymous city]. I grew up outside of [anonymous city], but now I live in [anonymous city], and I just haven’t been able to find one. Well I think I could find one, it’s just getting my medical records transferred here. It’s been such a pain... so I’ve been putting it on the back burner.

Other challenges that several participants commented on included social issues, feelings of isolation, and having difficulty accepting their diagnosis as adolescents. One participant shared that she never really talked about her diagnosis with her friends but eventually became more open about having TS after a friend watched her get a growth hormone shot:

I guess I didn't realize until maybe a friend had to see me get a shot. Normally I would try to do it downstairs. You know I'd be like 'Just call me down.' But one time I remember my dad forgot, and he was like, 'You know it's late, and it's past the time to get your shot.' So you know I had to explain why I had to get one.

Another participants expressed feelings of isolation as a young girl because of her diagnosis: *"I think when I was younger I knew about it, but I didn't go into detail or try to reach out to other girls or women with it. I definitely felt, I don't want to say alone, but more isolated."* A third participant discussed how she had difficulty with accepting her diagnosis of TS:

Yeah I would say I couldn't really wrap my head around it back then... I would say [it wasn't] until I started to go to counseling when I really started to be able to be more accepting and all that just because I didn't really have to think about it for so many years. And it just happened so fast and I wasn't really in the decision making process when they decided to remove my ovaries when I was 12, so it was just kind of like I suppressed it for a really time.

Difficulties in school, both with social interactions and academics, was another common challenge for many participants. One participant who also shared that she was diagnosed with a non-verbal learning disorder in grade school stated, *"Public school... Around 7th grade, I had a lot of problems... it [having TS] just started to be noticeable, and I had a lot of problems socially and in math."* Another participant who also reported having a non-verbal learning disorder said, *"...it made school, especially math, a little harder for me... I learn best when someone verbally explains a concept to me."*

Other transition issues included participants' uncertainty when making major medical decisions independently and the lack of sufficient support and provider education on the needs of girls and women with TS. One participant reported that, *"The hardest part was making a lot of decisions about my treatment on my own. I was kind of handed that at 17, 18 years old."* Another participant discussed her disappointment with the coordination of her healthcare transition:

I would say that it was kind of challenging. I think there was a phenomenal lack of the children's hospital supporting us and saying, 'These are the people who are most educated on TS and we feel that could help you.' It would have been fantastic just to have someone that the children's hospital referred us to and to have like a transition coordinator to work with us and make referrals to providers who have worked with patients with TS. There was no interprofessional communication as far as that went because it was difficult. We were pretty much having to start all over with regards to explaining what TS is and all of that to all of these doctors as we slowly found them and put them in place.

Finally, some participants mentioned that they wish they could have been more confident in their providers' understanding of their condition. For example, one participant stated, *"I think it's just been frustrating. You know I guess I wish that the person taking care of me would know something or just know a little bit more about it."*

2.4.2.7 Support needs and opinions of multidisciplinary care clinics. The last key element of the healthcare transition process in which themes were identified, dealt with participants' differing needs for support, as each participant had different support preferences. While some participants appreciated attending TSSUS events and receiving

support from other women with TS, others did not feel the need to attend support groups frequently nor did they want to be extremely involved with the TS community. One participant who holds a leadership position with her local TSSUS chapter stated that her involvement has been helpful:

I mean I didn't meet someone who had the same thing I had until I was maybe 17 years old. We did a walk for TS Awareness Month last February and had almost over 200 families come out, so that was a great experience to just to kind of learn from other people that are older than me but also help some of the littler ones, so I've really been enjoying being the leader.

Another participant had temporarily participated with the TSSUS but also received support from her parents and her husband, whom she met when she was 16 years old, when transitioning to adult-centered care: *"Yeah I think I had a variety of support, so I was pretty lucky."* With regards to her attendance at support group meetings, she felt that she was *"the only one who didn't live in the Turner syndrome world."* One other participant had a similar perspective with regards to not letting TS define her. She was also not involved with support groups growing up, nor had she met another woman with TS until she was about 23 years old:

So you know I really didn't know about any of that type of stuff... I didn't even know there were groups, so I attended my first meeting this summer, and it was good. I would say that I think between my three really good friends and my sister, they're enough, but meeting with other women with TS was really eye opening just hearing about their experiences. But at the same time, it is not something that I think defines me. So I don't feel the need to go to the groups and be

involved in so many events just because I don't want to be fully defined by that.

There is so much more to me than TS.

This participant also reiterated how not every girl with TS is the same, and since there is such a huge spectrum, providers should remain aware that not every girl or woman with TS will need or want the same kinds of support or treatment: *“Each case needs to be treated specifically and uniquely. Every girl is very different... so I think we need to get away from generalizations because there is such a large spectrum of girls with TS.”*

Another important topic that was brought up in conversation by several participants was multidisciplinary care and the utility of multidisciplinary clinics for girls and woman with TS. All participants who had been exposed to multidisciplinary care or who expressed that they were familiar with the concept spoke positively about it. For instance, one participant who received care at a multidisciplinary clinic as an emerging adult shared that this experience made her transition to adult-centered care easy:

[Anonymous hospital system] has a great clinic that helps with transition... I stopped going when I was in college just because I felt I was too busy for it. But when I went back after college as an adult, it was a very easy transition for me. I think it had something to do with the great clinic they have there. They have the audiology department, the cardiology department... So they were able to pick up right where I had left off in my care.

Another participant who is working hard to establish a multidisciplinary clinic at her local children's hospital expressed a similar appreciation for access to this type of care:

That is something that I would love to see, and I know whatever I might be able to do to help make that happen, I definitely will do to at least try to educate and

try to make this happen because it is so helpful. Like you said, you could have genetics, endocrinology, cardiology, and everyone come together and you know see patients... The thing is just getting the resources for that. I mean we're not really in the middle, but it's a large state and we would have enough space. But you know we need to the backing from the children's hospital here to do it.

Overall, participants' support preferences varied greatly, with some only needing support from immediate family and friends and others appreciating having a larger circle of support persons who they could turn to during their transition to adult-centered care. Participants were also given an opportunity to briefly comment on any additional supports not previously mentioned that they would have appreciated having during their transition to adult care, and these open-ended responses can be found in Table 2.5.

2.5 Discussion

The educational and support needs of emerging adulthood women with TS transitioning to adult-centered healthcare deserve greater attention. This study addressed such deficits in the literature by asking young adults with TS to reflect on their healthcare transition experiences, ability to communicate with healthcare providers, confidence in self-management skills, and knowledge of genetic principles underlying TS. This survey of 22 young adult women with TS as well as five interviews with participants collectively help shed light on the counseling needs of adolescents with TS. By exploring study participants' level of control and confidence in healthcare transition, existing support and resources for healthcare transition, educational needs during the transition process, and access to and involvement of multidisciplinary healthcare teams, this study brought important healthcare transition elements that need greater attention to the surface.

2.5.1 Elements of participant motivation during healthcare transition. Given that the large majority ($n = 17$) of our participants reported being motivated to manage their own healthcare and medical needs, healthcare providers should be challenged to develop strategies for empowering these girls and women so that they are able to obtain patient autonomy. However, almost half of the participants ($n = 10$) reported that they were not asked questions about their health directly until after the age of 18 years, which is consistent with past research that has shown most adolescents between the ages of 12 and 17 years of age do not receive time alone with their healthcare providers to receive routine screening and anticipatory guidance for health-related behaviors (Irwin, Adams, Park, & Newacheck, 2009). Additionally, all but one of the present study's participants reported that they wish to learn more about the genetic principles underlying TS, and all participants reported that they would have liked additional resources during the healthcare transition process. It is evident that these women are motivated to learn about their diagnosis of TS, including its genetic etiology, and appreciate knowing how to access resources and how to manage their medical needs as young adult women with TS.

In order to determine when emerging adult women with TS are ready to learn more about their diagnosis and take responsibility of their care, healthcare providers should assess when it is appropriate to begin the transition process. A recent editorial by McManus (2014) commented on a new transition readiness assessment tool that was developed by Got Transition, a federally funded program operated by the National Alliance to Advance Adolescent Health, as part of the Six Core Elements of Health Care Transition (2.0). These six core elements include 1) transition policy, 2) transition tracking and monitoring, 3) transition readiness, 4) transition planning, 5) transfer of care,

and 6) transition completion (Got Transition, 2014). Implementation of the Six Core Elements of Healthcare Transition and resulting improvements in quality of care were recently evaluated in a study by McManus et al. (2015), which demonstrated practices improved their transition planning, staff knowledge about transition, and transfer of care following a learning collaborative (LC) on healthcare transition. In fact after the LC, patients were being transferred to adult-centered care with updated medical summaries, completed transition readiness assessments, formal written transition plans, and chronic condition fact sheet as part of a comprehensive transition package (McManus et al., 2015).

By incorporating the use of such practices in the care of girls with TS, pediatric providers can identify the unique healthcare needs of each patient and individualize the transition process according to patient maturity and motivation to achieve autonomy. Furthermore, by taking formal steps to complete the transition process as recommended by the Got Transition protocol (2014) as well as the AAP's clinical report (2011), pediatric providers can better engage emerging adults with TS and help girls develop the confidence and self-care skills needed to effectively care for themselves as adults.

2.5.2 Adolescent counseling and educational needs. A major objective of this study was to identify if participants benefit from adolescent counseling on how to develop self-management skills. By comparing group differences between those who reported receiving counseling on how to develop self-care skills and those who do not, this study clearly illustrates that participants benefit from counseling. In particular, participants who received self-management counseling benefited in the areas of perceived control and confidence in their medical management (mean score = 1.24 points higher),

transition satisfaction and coordination (mean score = 1.15 points higher), and level of independence in their overall healthcare (mean score = 1.19 points higher). These improvements are consistent with an Australian study by Harvey et al. (2008), which demonstrated that patients who are viewed as *partners in health* and participate in self-care educational programs improve in their learning, knowledge, and health status. Furthermore, their study illustrated that patients with better self-management skills had better health outcomes with regards to improved health service utilization, general wellbeing, and fewer adverse impacts of their chronic illness (Harvey et al., 2008). The significance of enhanced health literacy in the context of one's own chronic condition in improving patient self-care abilities is also supported by the literature (Heijmans, Waverijn, Rademakers, van der Vaart, & Rijken, 2015).

Given the emphasis on education and counseling adolescents with TS about their condition throughout transition, the question is how health professionals will provide such services in an effective, consistent manner. As Rubin (2003) describes, utilization of a transition passport that provides a clinical summary, lists possible morbidities and risk factors, and delineates guidelines for adult patients with TS could be the key to improving transition outcomes for women with TS. Considering that the present study's participants desired better direction with regards to 1) which specialists they should see, 2) how to coordinate their care from multiple providers, 3) how to locate providers in their area most knowledgeable of the needs of women with TS, 4) ways to locate social and emotional supports, and 5) access to dedicated TS clinics (see data in Appendix A), developing formal transition passport programs, or so-called graduation clinics, in which

patients, parents, and providers collaboratively work to meet the individualized needs of each adolescent as they transfer to adult care, is imperative to ensure quality healthcare.

Fortunately, the large majority of study participants ($n = 15$) had been educated about the potential future health problems they may encounter as adults with TS. However, nine participants (40%) reported never having received education on the topics of fertility and reproductive options for women with TS. As more women with TS reach adulthood and research on reproductive options for women with TS continues to grow, it is critical that emerging adults with TS receive routine education on such topics. A study by Davies (2010) supports this practice initiative by promoting the necessity of utilizing annual appointments as educational opportunities to discuss sexual health and contraception for those with spontaneous periods. Considering that young women with TS require life-long care, it is critical that they have strong relationships with their providers and a full understanding of their condition and the impact it will have into adulthood, as these factors contribute to compliance (Davies, 2010).

2.5.3 Multidisciplinary care for girls and women with TS. Taking a multidisciplinary approach towards the lifetime care of patients with SHCNs is the most effective way to ensure a successful continuum of care through transition and yields the best health outcomes (Gravelle, Davidson, & Chilvers, 2012; Lucaccioni et al., 2015; Saenger, 2003). In this study, 17 participants reported that they receive care from multiple specialists. Of those, 15 participants felt this type of care better prepared them for their transition to adult-centered care. While these numbers are promising, only eight participants felt that their healthcare providers work as a team, utilizing each person's unique set of skills and expertise. Therefore, it is important that health professionals work

towards better interdisciplinary communication and care so that patients with SHCNs, specifically girls and women with TS, can receive well-coordinated, comprehensive care.

In addition to the challenge of interdisciplinary health professional involvement, many other barriers to multidisciplinary care exist for emerging adult women with TS. As described in several patient interviews, geographic location can be a physical limitation to accessing multidisciplinary care clinics. Given that not all girls and women with TS will be able to attend clinics dedicated to the multidisciplinary care of patients with TS, other strategies for delivering this type of care must be developed. Examples of alternative care delivery include telemedicine, satellite clinics, and/or traveling providers. Additionally, greater care should be taken when evaluating the need for multidisciplinary clinics at major academic or large children's hospitals. Multidisciplinary clinics that are dedicated to the care of children with Down syndrome, muscular dystrophy, and cystic fibrosis are becoming more universal across the nation, and the present study provides further supporting evidence that multidisciplinary care of girls and women with TS is equally critical for improving the healthcare transition process and associated health outcomes (Lucaccioni et al., 2015; Saenger, 2003).

2.5.4 The utility of support from genetic counselors. As mentioned before, genetic counselors are a group of healthcare professionals well equipped to educate adolescent females with TS about their diagnosis and to counsel them regarding the lasting impact of their condition into adulthood. However, the literature reveals a deficit in the amount of education available to assist emerging adult women with TS on how to cope with potential psychosocial challenges associated with their diagnosis (Behm, 2014). The present study provides supporting evidence that genetic counselors should be

involved as valued and essential members of integrative, multidisciplinary healthcare teams for young adult women with TS.

In the present study's population of young adult women with TS, 18 of 21 participants reported that they had not yet been offered genetic counseling. While it is difficult to measure precise outcomes of genetic counseling, a recent study by Zierhut, Shannon, Cragun, and Cohen (2016), identified twelve important genetic counseling outcome-related themes. These included psychosocial outcomes, adherence to or receipt of appropriate medical management, and patient and provider knowledge, each of which are extremely relevant to the counseling needs of young adult women with TS. The next most prevalent outcome-related themes included coordination and improvement of care, autonomous decision-making, and providing patient resources, which further support the utility of genetic counseling services during the healthcare transition process of emerging adult women with TS (Zierhut et al., 2016). Given the relevance of the outcome-related themes that were identified by this large cohort of genetic counselors, it is apparent that the genetic counseling profession is aware of the needs of patients with SHCNs and is well outfitted to provide counseling services to adolescent girls with TS.

Genetic counselors are uniquely skilled in providing age-appropriate counseling and adapting counseling agendas according to patient engagement, patient intellect, and patients' expressed objectives for their counseling appointment. Counseling or visual aids are highly valued tools in the profession and may be of great worth when educating adolescent females with TS. Given that 20 of 21 participants wished to learn more about the genetic principles underlying TS, and 18 of 21 participants were not familiar with the concept of haploinsufficiency, which is of extreme relevance to TS, a genetics education

is integral to the multidisciplinary care of adolescents with TS. Interview participants were also uncertain of the meaning of having the mosaic form of TS. The utility of genetic counseling among the adolescent population is supported by a recent study by Pichini et al. (2015), which identified adolescents' understanding of a genetic counselor's role, increasing perceived personal control, and adolescent-specific factors that impact adaptation to one's genetic condition as important elements of the adolescent perspective of genetic counseling. This study also provided suggestions, tools, and alternative strategies for developing an adolescent-focused framework to counseling adolescents experiencing the transition from pediatric to adult-centered care (Pichini et al., 2015). Genetic counselors are well trained in explaining genetic principles to those who do not have a background in genetics and are equipped with unique strategies to help younger individuals better understand complex principles related to their specific diagnosis.

Additionally, genetic counselors would be able to provide adolescents with fact sheets or a general summary of guidelines for the care of girls and women with TS, which could be used for one's own personal education or given to healthcare providers who may be less familiar with caring for girls and women with TS. Lastly, care coordination is a key skill of genetic counselors. Through incorporating genetic counselors into the TS care team, the overall health – emotional, social, and medical – of girls and women with TS would be addressed and this group of motivated patients could receive as much education and counseling as desired.

Upon learning what education genetic counselors can provide to girls and women with TS, the large majority of participants in the present study who were interviewed about their transition experiences agreed that such services would be useful. Furthermore,

a large proportion of these women felt that they would have benefited from counseling at a younger age. While the precise age of such counseling was not fully agreed upon, it was apparent that receiving counseling as early as at the onset of puberty and at the very least before the age of 18 years would be of great benefit to these women. Given that some women reported having a difficult time accepting their diagnosis because they did not fully understand it, it is essential that counseling services be available to women at an early age. For girls who are diagnosed around the time of puberty and vulnerable period of adolescence, genetic counseling should be an immediate resource that is offered so that these girls can better adjust to their diagnosis of TS as well as more confidently communicate their diagnosis of TS with both their peers and other healthcare providers. Genetic counseling services should thus be offered at a young age (e.g., age 12), as recommended in the AAP's clinical report (2011).

2.5.5 Differing support needs for girls and women with TS. Participants in this study expressed varying support needs throughout their transition to adult care. While several participants commented on their appreciation for the TSSUS and how attending annual conferences and/or local chapter meetings has helped their personal development and acceptance of their TS diagnosis, other participants did not have these same sentiments. For example, while some participants began attending society meetings and making friends with other girls with TS at a young age, other participants had not met someone else with TS until they were maybe in their 20s and felt that was all they needed to get a new perspective. Taking all of these differences into consideration, it is important for healthcare providers to understand that not all patients benefit from referrals to support groups; providers should be challenged to further inquire about patient support

preferences and their psychosocial support needs so that they can help patients locate and access more appropriate resources. Additionally, some participants appreciated their parents' involvement during the transition while others expressed that they wished they were given a louder voice and more direct input with regards to the direction of their healthcare as adolescents with TS. Therefore, it is essential that providers take time to learn about their patients' preferences for care and sit down with families in order to complete formal transition paperwork so that parents, patients, and providers expectations are equally valued and achieved throughout the transition to adult-care.

In the present study, participants' preferences for care delivery were assessed following their participation in short, 20-minute interviews. Although Jedeloo et al. (2010) described four patient care delivery preferences – conscious & compliant, backseat patient, self-confident & autonomous, and worried & insecure – none of the interview participants fit completely into the worried & insecure or the self-confident & autonomous categories. Rather, each of the participants' healthcare journeys reflected a shift in care preferences between the remaining two patient care delivery models over time. The comments from four out of the five participants aligned most closely with the conscious & compliant patient profile at the time of their interviews. These individuals want their doctors to talk to them and not to their parents. They know enough about their condition that they understand the importance of treatment adherence for their wellbeing. They also feel their diagnosis does not prevent them from living a normal life, that they are independent in their healthcare, and that they appreciate being treated as adults. The remaining participant's care delivery preferences was determined to be mostly in agreement with being a backseat patient, as she was not as expressive about taking charge

of her healthcare. While she commented on appreciating greater autonomy and being handed more responsibility in her healthcare as an adult with TS, her overall level of independence did not appear to be of as high priority as it did for other participants. Yet, she did seem to desire additional responsibility and was working towards becoming a more conscious & compliant patient as an adult with TS.

Although four of the five participants currently fall into the conscious & compliant category when they reflected back on their healthcare transition, they had been previously more aligned with the backseat patient, as they had relied on their parents to make decisions for them when they were younger and had not been given much of a voice in their healthcare choices. For example, one participant shared she was not involved in the discussion of when to remove her ovaries.

2.5.6 Study limitations and future research. One limitation of the present study is its small sample size of 22 participants and resulting limitations for generalizing these results to the larger TS community. Purposive sampling of women who were already involved with either the TSSUS and TSF might have also introduced a selection bias. Lastly, study participants also represent a fairly homogenous group of women, given 90% of participants identified as Caucasian and the large majority had a high level of education. The survey also did not provide a precise, operational definition for *counseling on self-management skills*; therefore, participants may have interpreted this question in different ways, which might have influenced the study's results. Had distinct elements of *counseling on self-management skills* been more clearly defined, statements of agreement or disagreement would be more explicit and consistent.

One additional aspect of healthcare transition that was not evaluated by this study is whether or not improved confidence in care and enhanced patient autonomy truly improve health outcomes. Future research on this subject should be undertaken so that it can be determined whether or not patients' perceived level of control, independence, and satisfaction with the healthcare transition process influence the health outcomes of women with TS. Larger studies that include more numerous patient interviews should be pursued so that women with TS can continue to have a voice in promoting their care preferences. Lastly, research that takes the opinions of adolescent girls with TS into account should be conducted so that health professionals and genetic counselors can better understand the needs and desires of this specific population more directly. Case-control studies that compare outcomes following genetic counseling interventions for adolescent girls with TS should also be pursued so that health professionals can continue to learn more about the support and educational needs of emerging adults with TS.

2.5.7 Practice implications. Given this collection of findings, it is apparent that care delivery preferences for girls and women with TS are likely to evolve over time, with age and increasing maturity. Therefore, it is critical that providers take action to modify the way in which they care for girls with TS as they move through adolescence and transition into adulthood. It is likely that emerging adult women with TS will want to be seen as *partners in health* and will appreciate learning more about their condition so that they can take greater responsibility for their healthcare. Considering the motivation of these patients to achieve autonomy, providers will want to formally counsel adolescent girls with TS on ways to develop self-management skills by using a staged process and should turn to the algorithmic protocols described in the AAP's clinical report (2011). Genetic

counselors are well positioned to provide such counseling in order to enhance patient autonomy and confidence in their healthcare, and their involvement in multidisciplinary clinics is highly warranted. Given the positive implications of providing a basic genetics education to emerging adult women with TS, practice guidelines should be written to reflect the value that genetic counselors would bring to the specialty care teams of girls and women with TS.

2.6 Conclusion

The healthcare transition process is a critical period for emerging adults, especially those with SHCNs. Considering the ongoing medical monitoring necessary for girls and women with TS, it is essential that the transition to adult-centered care be well-coordinated and comprehensive. For those who are motivated to learn more about their condition with age and develop self-management skills, encouragement throughout the transition process by a multidisciplinary healthcare team can help empower girls with TS to take full responsibility of their medical needs with utmost confidence.

Given that the participants in this study reported a high level of motivation to learn more about TS and the implications this disorder would have for them into their adult years, it is critical that health professionals follow algorithmic protocols and develop personalized transition plans for their own patients with TS. It is apparent that girls and women with TS can benefit from receiving counseling on how to develop appropriate self-efficacy skills, as participants in this study who received such education reported higher levels of perceived confidence, independence, and satisfaction with the healthcare transition process.

The large majority of study participants appreciated receiving care from multiple specialists and felt that by receiving well-rounded care they were better prepared for assuming responsibility for their healthcare as autonomous adults. Those participants who felt their voice was heard during their transition to adult-centered care were also more independent as adults with TS and celebrated this fact.

This study also highlights how an increased knowledge and understanding of the genetic etiology of TS may help girls and women to be more accepting of their diagnosis at an earlier age. Only a minority of the participants reported having been offered the opportunity to meet with a genetic counselor, and most participants were unfamiliar with the resources that these health professionals could provide. Given the benefit of counseling demonstrated by this study, genetic counselors are strongly encouraged to continue advocating for their involvement in the multidisciplinary healthcare teams of emerging adult women with TS. Genetic counselors should also be involved in conversations about and help guide recommendations for clinical practice, emphasizing that these guidelines reflect the utility of promoting self-efficacy in young adults with TS.

Table 2.1: Principal Component Analysis – Perceived Confidence Score

Perceived Confidence Score					
Question #20					
I am confident in my ability to explain my special healthcare needs to friends and family members.					
Strongly Agree	Agree	Somewhat Agree	Somewhat Disagree	Disagree	Strongly Disagree
Question #21					
I am confident in my ability to explain my diagnosis to my healthcare providers.					
Strongly Agree	Agree	Somewhat Agree	Somewhat Disagree	Disagree	Strongly Disagree
Question #22					
I am confident in my ability to communicate my medical needs to my healthcare providers.					
Strongly Agree	Agree	Somewhat Agree	Somewhat Disagree	Disagree	Strongly Disagree
Question #26					
I feel that my healthcare providers understand all aspects of my Turner syndrome diagnosis.					
Strongly Agree	Agree	Somewhat Agree	Somewhat Disagree	Disagree	Strongly Disagree

Table 2.2: Principal Component Analysis – Transition Coordination and Satisfaction Score

Transition Coordination and Satisfaction Score					
Question #29					
My transition to adult-centered healthcare was well coordinated.					
Strongly Agree	Agree	Somewhat Agree	Somewhat Disagree	Disagree	Strongly Disagree
Question #31					
I felt well supported throughout my transition to adult-centered healthcare.					
Strongly Agree	Agree	Somewhat Agree	Somewhat Disagree	Disagree	Strongly Disagree
Question #32					
I was able to easily identify adult-care providers that were knowledgeable of Turner syndrome during my healthcare transition.					
Strongly Agree	Agree	Somewhat Agree	Somewhat Disagree	Disagree	Strongly Disagree
Question #35					
I am satisfied with how I was transitioned to adult-centered healthcare.					
Strongly Agree	Agree	Somewhat Agree	Somewhat Disagree	Disagree	Strongly Disagree

Table 2.3: Principal Component Analysis – Level of Independence Score

Level of Independence								
Question #23								
I am able to locate any health-related information and paperwork quickly and easily.								
Strongly Agree	Agree	Somewhat Agree		Somewhat Disagree		Disagree	Strongly Disagree	
Question #24								
I know when and how to ask appropriate questions during my medical appointments.								
Strongly Agree	Agree	Somewhat Agree		Somewhat Disagree		Disagree	Strongly Disagree	
Question #27								
I feel that I have achieved complete independence in my healthcare.								
Strongly Agree	Agree	Somewhat Agree		Somewhat Disagree		Disagree	Strongly Disagree	
Question #40								
My ability to share in decisions made about the management of my condition is:								
8 (Very Good)	7	6	5	4 (Satisfactory)	3	2	1	0 (Very Poor)

Table 2.4: Patient Demographics (N = 22)

Group or Category	Percentage (n)	Group or Category	Percentage (n)
Age		Time of Diagnosis	
18-20	14% (3)	Before birth	14% (3)
21-23	5% (1)	Soon after birth (newborn)	46% (10)
24-26	32% (7)	Between the ages of 1 & 5	9% (2)
27-30	50% (11)	Between the ages of 6 & 11	5% (1)
Ethnicity		Between the ages of 12 & 18	27% (6)
White	91% (20)	After the age of 18	0% (0)
Asian	5% (1)	Diagnosing Physician	
Hispanic	5% (1)	Pediatrician	36% (8)
Geographic Location		Pediatric Endocrinologist	41% (9)
Northeast	27% (6)	Endocrinologist	5% (1)
Southeast	23% (5)	Internal Medicine	5% (1)
Midwest	14% (3)	Obstetrician	9% (2)
Northwest	5% (1)	Unknown	5% (1)
Southwest	23% (5)	Regular Specialty Care	
Not Reported	9% (2)	Heart	41% (9)
Marital Status		Kidney	0% (0)
Single (Never Married)	82% (18)	Ear, Nose, and Throat	9% (2)
Married or Domestic Partnership	14% (3)	Skin	18% (4)
Divorced	5% (1)	Endocrinologist	59% (13)
Highest Education		Geneticist	0% (0)
Graduated High School	18% (4)	Genetic Counselor	0% (0)
Some Postsecondary	32% (7)	Previous Specialty Care	
Vocational	5% (1)	Heart	86% (19)
Bachelor's Degree	23% (5)	Kidney	27% (6)
Some Postgraduate Education	5% (1)	Ear, Nose, and Throat	64% (14)
Master's Degree	14% (3)	Skin	41% (9)
Professional Degree	5% (1)	Endocrinologist	91% (20)
		Geneticist	18% (4)
		Genetic Counselor	5% (1)

Table 2.5: Existing Support and Resources for Healthcare Transition (N = 22)

Survey Question	Response Options	n	Survey Question	Response Options	n		
29. My transition to adult-centered healthcare was well coordinated. (n = 21)	Strongly Agree	1	30. My doctors began to shift their attention away from my parents and ask me questions directly between the ages of .	12 and 13	2		
	Agree	6		14 and 15	4		
	Somewhat Agree	6		16 and 18	6		
	Somewhat Disagree	2		After the age of 18	10		
	Disagree	2					
31. I felt well supported throughout my transition to adult-centered healthcare. (n = 21)	Strongly Disagree	4	32. I was able to easily identify adult providers that were knowledgeable of TS during my transition. (n = 21)	Strongly Agree	1		
	Disagree	2		Agree	3		
	Somewhat Disagree	4		Somewhat Agree	4		
	Somewhat Agree	4		Somewhat	4		
	Agree	7		Disagree			
33. I wish that I had access to more resources and supports throughout the healthcare process.	Disagree	2	35. I am satisfied with how I was transitioned to adult centered healthcare. (n = 21)	Disagree	4		
	Strongly Disagree	4		Strongly Disagree	5		
	Somewhat Disagree	0		Strongly Agree	0		
	Somewhat Agree	6		Agree	5		
	Agree	6		Somewhat Agree	7		
34. I would have appreciated more support in the following areas:	Somewhat	0		Somewhat	3		
	Disagree	0		Disagree			
	Disagree	0		Disagree	2		
	Strongly Disagree	0		Strongly Disagree	4		
				“Which specialists to see, how often to see them, and how to coordinate care between multiple healthcare professionals.”			
				“Psychology and Cardiology.”			
				“More doctors familiar with what TS is and how they can help.”			
				“Getting the health knowledge and knowing where the knowledgeable doctors are in the area.”			
				“Better transition and assistance from my pediatric endocrinologist into an adult endocrinologist.”			
				“I wish I knew how to find doctors in my area that were more knowledgeable, but I do know where to find resources if my doctors need education on TS.”			
				“My mother was my support and transition coordinator... My mom did a great job getting every specialist in place.”			
				“List of physicians by location who are knowledgeable about TS.”			
		“Identifying the main doctor best suited to coordinate all aspects of my healthcare.”					
		“Doctors telling you what steps you need to take for your medial care. All I learned was from other TS women and at medical conferences.”					
		“Help with my non-verbal learning disability.”					
		“Social and emotional support. More doctors that are aware of TS and that know what I should be getting care in.”					
		“I live in a very small town, so I wish I lived closer to a TS clinic.”					

Table 2.6: Exploration of Healthcare Transition Education Needs (*n* = 21)

Survey Question	Response Options	<i>n</i>	Survey Question	Response Options	<i>n</i>
38. I was well educated about and fully understand the potential future health problems I may face as an adult with TS	Strongly Agree	3	39. I was effectively counseled about how my condition might affect me in adulthood between ____.	12 and 13	3
	Agree	9		14 and 15	2
	Somewhat Agree	3		16 and 18	5
	Somewhat Disagree	2		18 and 23	0
	Disagree	4		After 23	1
	Strongly Disagree	0		I have not been fully counseled.	10
43. My healthcare providers listen to my opinion and personal preferences for an estrogen regimen...	Strongly Agree	4	44. I was educated on the topics of fertility and reproductive options for women with TS between ____.	12 and 13	4
	Agree	6		14 and 15	2
	Somewhat Agree	5		16 and 18	2
	Somewhat Disagree	2		18 and 23	3
	Disagree	3		After the age of 23	1
	Strongly Disagree	1		Never	9
41. I wish to learn more about the genetic principles underlying TS.	Strongly Agree	11	42. One of my healthcare providers has effectively explained the concept of haploinsufficiency in the context of my condition.	Yes	1
	Agree	4		No	2
	Somewhat Agree	5		I am not familiar with that concept	18
	Somewhat Disagree	1			
	Disagree	0			
	Strongly Disagree	0			
36. Has genetic counseling ever been offered to you? If yes, when were you presented with this option?	Yes	3	61. I am highly motivated to manage my own healthcare and medical needs.	Strongly Agree	4
	No	18		Agree	9
Between 12 and 13				Somewhat Agree	4
	Before the age of 12			Somewhat Disagree	1
				Disagree	2
				Strongly Disagree	1

Table 2.7: Involvement of Multidisciplinary Healthcare Teams (*n* = 21)

Survey Question	Response Options	<i>n</i>	Survey Question	Response Options	<i>n</i>
51. My healthcare providers actively listen and respond to my healthcare needs and concerns.	Strongly Agree	1	52. My healthcare providers empower me to take responsibility of my healthcare.	Strongly Agree	2
	Agree	12		Agree	9
	Somewhat Agree	5		Somewhat Agree	6
	Somewhat Disagree	2		Somewhat Disagree	2
	Disagree	1		Disagree	1
	Strongly Disagree	0		Strongly Disagree	1
53. My providers connected me to a sufficient number of supports as I prepared for my transition to adult-care. (N = 19)	Strongly Agree	1	54. I would have enjoyed meeting with a health professional in private as an adolescent to address my concerns about transition.	Strongly Agree	9
	Agree	3		Agree	6
	Somewhat Agree	7		Somewhat Agree	3
	Somewhat Disagree	1		Somewhat Disagree	3
	Disagree	3		Disagree	0
	Strongly Disagree	4		Strongly Disagree	0
55. My healthcare providers serve as advocates in the following areas of my life.	Work & Employment	2	56. I wish that my healthcare providers served as advocates in other areas of my life.	Yes	9
	Personal Life	7		No	12
	School & Education	4			
	Health Insurance	6			
	No Other Areas	11			
57. I appreciate receiving care from multiple specialists. I do not receive care from multiple health specialists	Strongly Agree	6	58. By receiving care from multiple specialists, I was more prepared to transition. I do not receive care from multiple health specialists	Strongly Agree	5
	Agree	7		Agree	6
	Somewhat Agree	2		Somewhat Agree	4
	Somewhat Disagree	1		Somewhat Disagree	1
	Disagree	1		Disagree	1
	Strongly Disagree	4		Strongly Disagree	4
62. My healthcare providers work as a team, utilizing each person's skills & expertise.	Strongly Agree	0	63. I feel that my healthcare is comprehensive.	Strongly Agree	1
	Agree	4		Agree	7
	Somewhat Agree	4		Somewhat Agree	8
	Somewhat Disagree	8		Somewhat Disagree	1
	Disagree	4		Disagree	1
	Strongly Disagree	1		Strongly Disagree	3
			Disagree		

Table 2.8: Development of Self-Management Skills ($n = 21$)

Survey Question	Level of Agreement	<i>n</i>	Survey Question	Level of Agreement	<i>n</i>
65. My healthcare providers prioritize my inclusion in my medical management	Strongly Agree	1	66. I wish to learn more about TS and what implications my diagnosis has for my future healthcare management. ($n = 20$)	Strongly Agree	7
	Agree	10		Agree	11
	Somewhat Agree	4		Somewhat Agree	2
	Somewhat Disagree	2		Somewhat Disagree	0
	Disagree	1		Disagree	0
	Disagree	3		Disagree	0
	Strongly Disagree	0		Strongly Disagree	0
67. I wish that I had more resources to provide to my healthcare providers regarding my diagnosis of TS.	Strongly Agree	9	68. I was counseled as an adolescent regarding ways to promote the development of independent, self-care behaviors.	Strongly Agree	4
	Agree	9		Agree	3
	Somewhat Agree	3		Somewhat Agree	3
	Somewhat Disagree	0		Somewhat Disagree	2
	Disagree	0		Disagree	6
	Strongly Disagree	0		Strongly Disagree	3

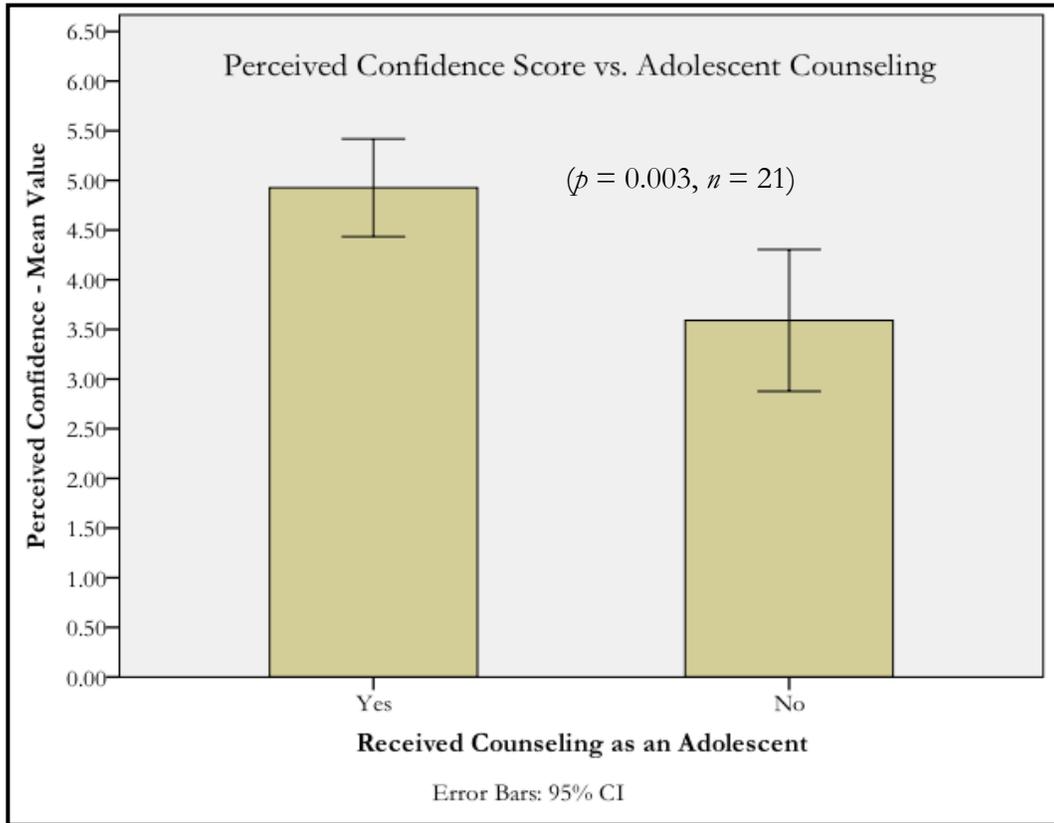


Figure 2.1: Mean Perceived Confidence Score vs. Reception of Adolescent Counseling on Development of Self-Management Skills

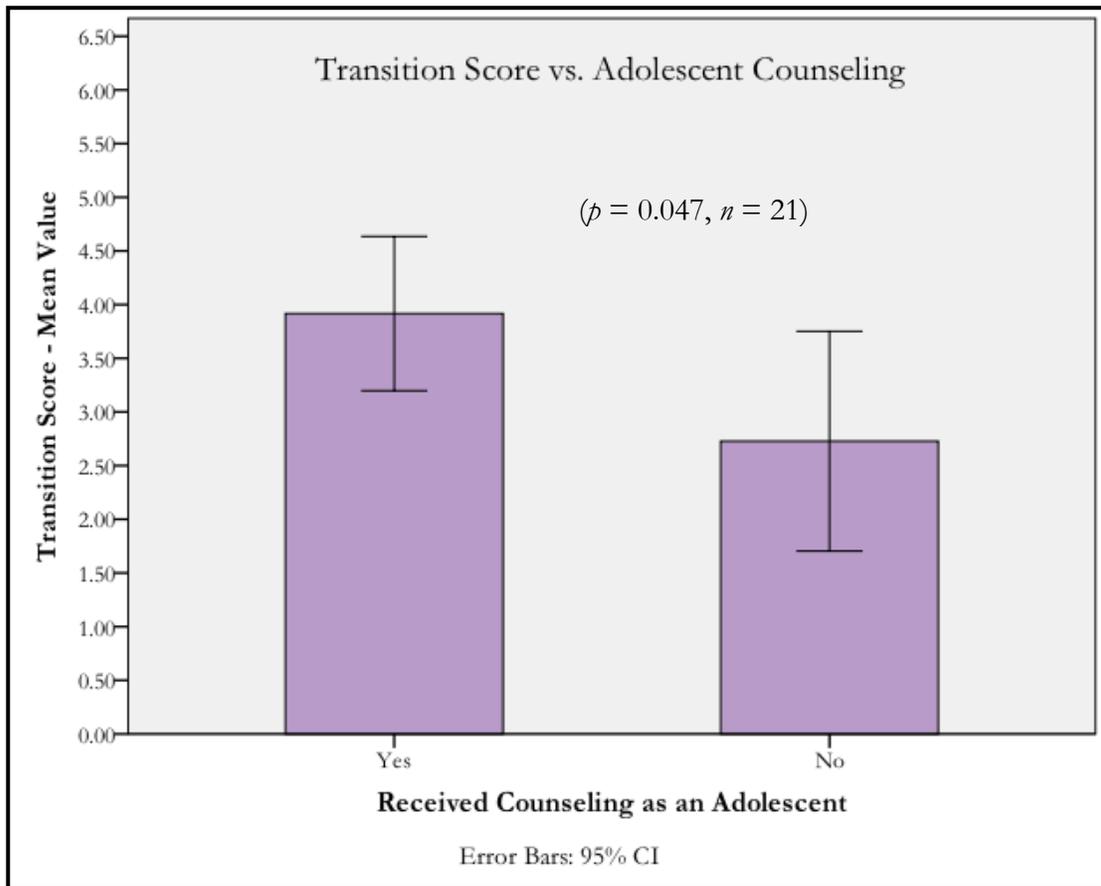


Figure 2.2: Mean Transition Score vs. Reception of Adolescent Counseling on Development of Self-Management Skills

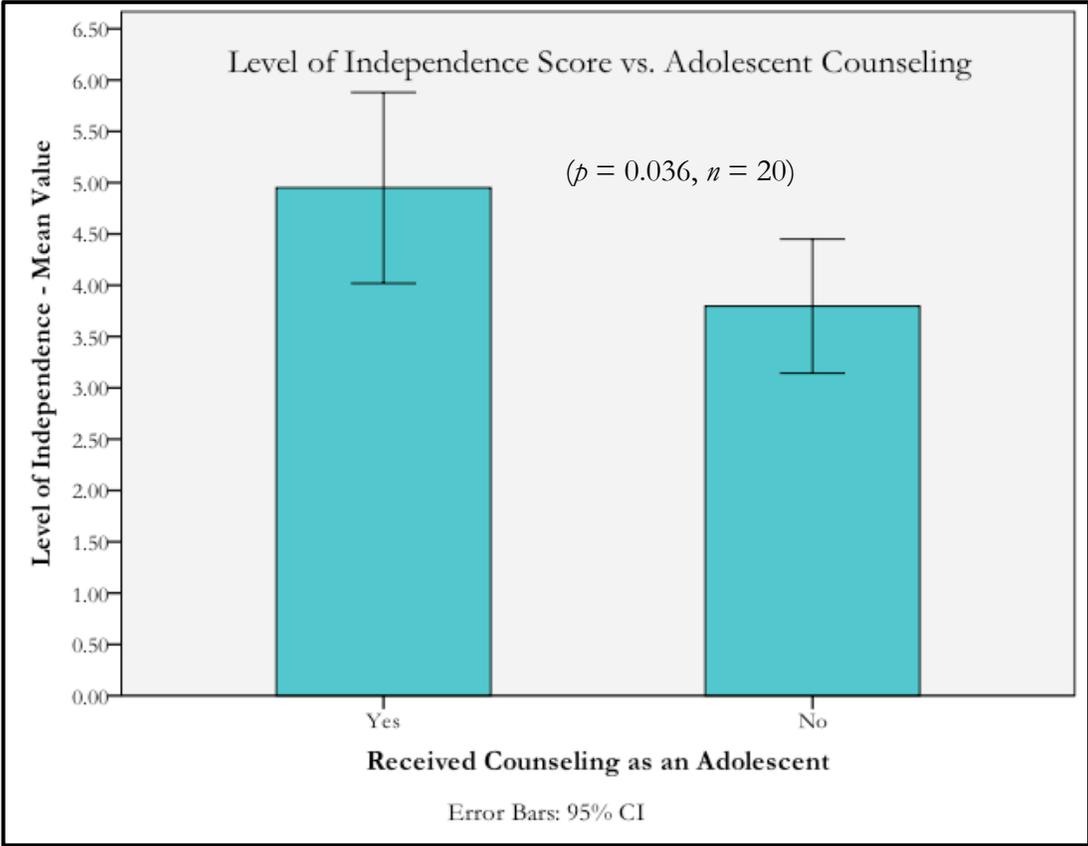


Figure 2.3: Mean Level of Independence Score vs. Reception of Adolescent Counseling on Development of Self-Management Skills

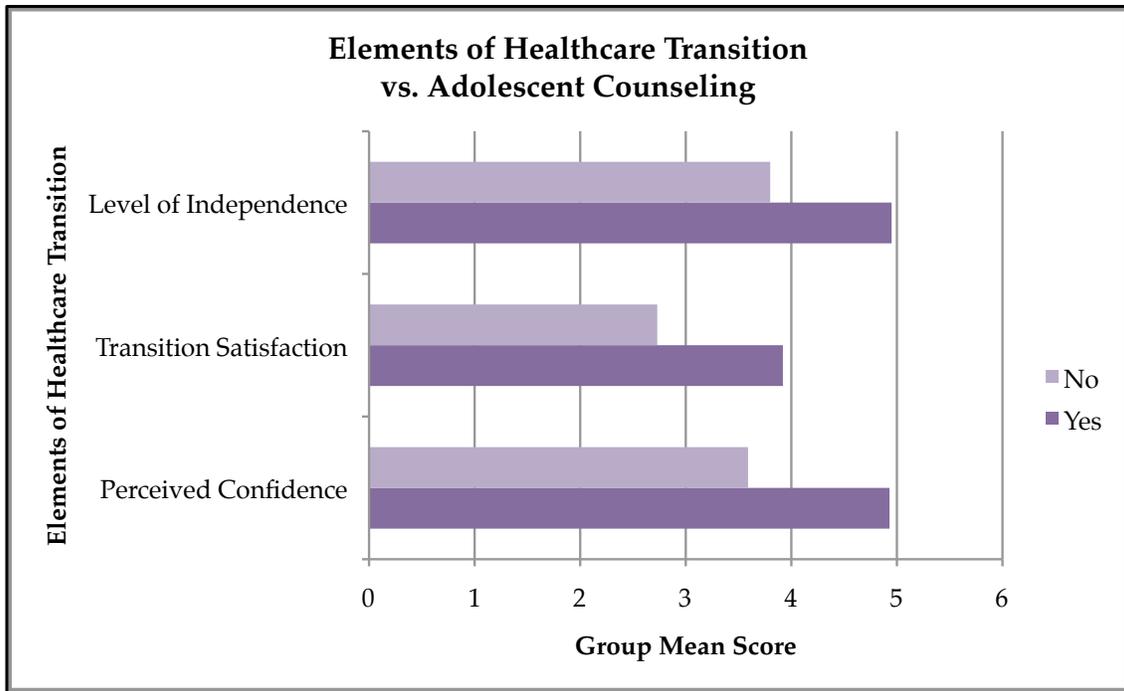


Figure 2.4: Comparison of Different Elements of Healthcare Transition

Chapter 3: Conclusions

The healthcare transition process is a critical period for emerging adults, especially those with SHCNs. Considering the ongoing medical monitoring necessary for girls and women with TS, it is essential that the transition to adult-centered care be well-coordinated and comprehensive. For those who are motivated to learn more about their condition with age and develop self-management skills, encouragement throughout the transition process by a multidisciplinary healthcare team can help empower girls with TS to take full responsibility of their medical needs with utmost confidence.

Given that the participants in this study reported a high level of motivation to learn more about TS and the implications this disorder would have for them into their adult years, it is critical that health professionals follow algorithmic protocols and develop personalized transition plans for their own patients with TS. It is apparent that girls and women with TS can benefit from receiving counseling on how to develop appropriate self-efficacy skills, as participants in this study who received such education reported higher levels of perceived confidence, independence, and satisfaction with the healthcare transition process.

The large majority of study participants appreciated receiving care from multiple specialists and felt that by receiving well-rounded care they were better prepared for assuming responsibility for their healthcare as autonomous adults. Those participants who felt their voice was heard during their transition to adult-centered care were also more independent as adults with TS and celebrated this fact.

This study also highlights how an increased knowledge and understanding of the genetic etiology of TS may help girls and women to be more accepting of their diagnosis at an earlier age. Only a minority of the participants reported having been offered the opportunity to meet with a genetic counselor, and most participants were unfamiliar with the resources that these health professionals could provide. Given the benefit of counseling demonstrated by this study, genetic counselors are strongly encouraged to continue advocating for their involvement in the multidisciplinary healthcare teams of emerging adult women with TS. Genetic counselors should also be involved in conversations about and help guide recommendations for clinical practice, emphasizing that these guidelines reflect the utility of promoting self-efficacy in young adults with TS.

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Appendix A: Supplemental Data

Table A.1: Open-Ended Responses in Comment Box on Survey Tool

<p>69. Please feel free to comment on any additional information and/or your feelings towards your transition to adult-centered care in the box below.</p>	<p>“I wish I had known I had mosaic TS or at least the possibility of it before I randomly just started developing. It was actually kind of scary and confusing because I was told this wasn't supposed to happen to me. My bones fused quicker with the onset of puberty, and I wound up even shorter than projected. I also wish the doctor had made more of an effort to include me more in discussions all along. It would have helped if they used simpler language for a child patient to understand instead of adult concepts and advanced medical terminology. I felt like a side note at the end of my own doctor’s appointment. They'd finally ask 'oh do you have any questions?' after talking over my head.”</p>
	<p>“I think you should have allowed for answers that included parents as my Mom did everything with transition and education not the doctors or children's hospital. I was happy with transition but not because the professionals helped.”</p>
	<p>“While I feel like I was educated on future problems I would face, it mainly came through my parents who I assume were educated by my pediatrician. I would have liked more direct counseling/education as an adolescent. Overall though, I live an independent life as an adult now. I saw a pediatric endocrinologist as a child/adolescent, and he managed my hormone therapy (both growth hormone as a child and estrogen as an adolescent) and my hypothyroidism. My primary care physician took over the responsibilities of managing my hypothyroidism after I was phased out due to age. Although I saw cardiologist every five years as a child/adolescent, I have not had any history of the heart conditions associated with TS, so I will probably not see a cardiologist again unless I decide to pursue pregnancy. I would probably also consider seeing a genetic counselor at that time as well.”</p>
	<p>“I feel that my doctor is amazing and what I have heard is adult doctors that help with TS are horrible and only worry about a little thing not the whole TS. I'm staying with my kid doctor till she kicks me out! Dr. [anonymous name] is the best. Spread the word to anyone who lives in [anonymous U.S. state].”</p>
	<p>“The pediatric clinic I visited as a minor seemed a little more ‘in tune’ to listen to and respond to my complaints. The adult one is only now getting better.”</p>

Table A.2: Participants' Level of Control and Confidence in Healthcare Transition (N = 22)

Survey Question	Response Options	n	Survey Question	Response Options	n
20. I am confident in my ability to explain my special healthcare needs to friends and family members	Strongly Agree	5	21. I am confident in my ability to explain my diagnosis to my healthcare providers.	Strongly Agree	4
	Agree	9		Agree	6
	Somewhat Agree	4		Somewhat Agree	7
	Somewhat Disagree	1		Somewhat Disagree	1
	Disagree	2		Disagree	3
	Strongly Disagree	1		Strongly Disagree	1
22. I am confident in my ability to communicate my medical needs to my healthcare providers.	Strongly Agree	5	23. I am able to locate any health-related information and paperwork quickly and easily.	Strongly Agree	3
	Agree	7		Agree	8
	Somewhat Agree	6		Somewhat Agree	8
	Somewhat Disagree	1		Somewhat Disagree	0
	Disagree	3		Disagree	1
	Strongly Disagree	0		Strongly Disagree	2
24. I know when and how to ask appropriate questions during my medical appointments	Strongly Agree	4	25. I feel that my healthcare providers understand all aspects of my TS diagnosis.	Strongly Agree	1
	Agree	6		Agree	3
	Somewhat Agree	7		Somewhat Agree	6
	Somewhat Disagree	1		Somewhat Disagree	6
	Disagree	2		Disagree	3
	Strongly Disagree	2		Strongly Disagree	3
26. I feel that I have sufficient knowledge on all aspects of TS.	Strongly Agree	2	27. I feel that I have achieved complete independence in my healthcare.	Strongly Agree	3
	Agree	6		Agree	6
	Somewhat Agree	10		Somewhat Agree	8
	Somewhat Disagree	2		Somewhat Disagree	1
	Disagree	1		Disagree	1
	Strongly Disagree	1		Strongly Disagree	3
28. I achieved full independence in my healthcare between the ages of _____.				16 and 17	1
				18 and 20	7
				21 and 25	4
				26 and 30	2
			I do not feel that I have achieved full independence in my healthcare.		8

Table A.3: Results from Miscellaneous Survey Questions

Survey Question	Response Options	<i>n</i>	Survey Question	Response Options	<i>n</i>
40. My ability to share in decisions made about the management of my condition is... (<i>n</i> = 21)	0 (Very Good)	3	45. I have experienced a cardiovascular complication and/or have a heart defect. (<i>n</i> = 21)	Yes	9
	1-3	9		No	12
	4 (Satisfactory)	7			
	5-7	2			
46. I carry medical information in my wallet or on a bracelet in order to alert... about my heart condition. (<i>n</i> = 9)	Yes	2	47. I have been educated about the associated risks of my heart condition and understand the need to keep medical appointments... (<i>n</i> = 9)	Strongly Agree	5
	No	7		Agree	3
				Somewhat Agree	0
				Somewhat Disagree	0
		Disagree		1	
48. I intend to pursue or have already pursued pregnancy as an adult woman with Turner syndrome. (<i>n</i> = 21)	Yes	5	49. I was educated about the medical complications and risks associated with pregnancy... between the ages of... (<i>n</i> = 5)	12 and 13	0
	No	16		14 and 15	0
				16 and 18	2
				18 and 23	0
				23 and over	1
50. I intend to pursue or have pursued other family planning options. (<i>n</i> = 16)	Yes	9	59. While I receive care from multiple specialists, I would appreciate more coordinated care. (<i>n</i> = 19)	Strongly Agree	8
	No	7		Agree	7
		Somewhat Agree		4	
		Somewhat Disagree		0	
		Disagree		0	
60. My healthcare providers express interest in my wellbeing. (<i>n</i> = 21).	Physical	21	61. My healthcare providers are sensitive to my diagnosis. (<i>n</i> = 21)	Strongly Agree	2
	Social	6		Agree	9
	Emotional	4		Somewhat Agree	7
				Somewhat Disagree	2
		Disagree		0	
17. The healthcare provider who manages most of my care is a... (<i>n</i> = 21)	Pediatric Endocrinologist	3	70. I am interested in being contacted for participation in a follow-up interview at a later date. (<i>n</i> = 21)	Yes	14
	Adult Endocrinologist	4		No	7
	Family Doctor	9			
	Nurse Practitioner	1			
	Internal Medicine	4			

Appendix B: Invitation to Participate

Dear Potential Participant,

You are invited to participate in a graduate research study exploring the current state of healthcare transition for young adult women with Turner syndrome. I am a graduate student at the University of South Carolina pursuing a master's degree in Genetic Counseling. My research aims are to learn more about how comfortable young adult women feel when explaining their condition to peers, family, and healthcare professionals. We also hope to identify ways in which genetic counselors might serve as advocates and help women develop self-management skills beginning in early adolescence.

The survey is intended for women between the ages of 18 and 30. If you fall outside of this age range, please feel free to complete the survey, but your responses will not be included in the study analysis.

As part of your participation, you will be asked to complete a brief 20 minute online survey that will ask you questions concerning your healthcare transition experiences and motivations for pursuing patient autonomy. All responses gathered will be kept anonymous and confidential. Upon completing the survey, you will be asked for your name, phone number, and email address for the purposes of contacting you for a virtual interview at a later date if interested. It is not required that you provide this information however. The results of our study may be published or presented at professional meetings, but participants will never be identified.

Your participation in our research is entirely voluntary. By completing the survey, you are consenting that you have read and understand our research objectives and methods. If at any point you no longer feel comfortable participating, you may withdraw from the study by not completing the survey. Furthermore, by providing your name and contact information at the end of the survey, you will be consenting for follow-up contact and an invitation to participate in a virtual interview that will afford you the opportunity to elaborate on challenges and strengths of your healthcare transition.

In order to access our survey tool, please insert the following URL into your web browser: <https://www.surveymonkey.com/r/uscggeneticsTS>. You will then be taken to SurveyMonkey™ and prompted to begin the survey at your convenience.

If you have any questions regarding this research, you may contact either myself or my faculty advisor, Richard Ferrante, PhD, directly using the contact information below. If you have any questions about your rights as a research participant, you may contact the Office of Research Compliance at the University of South Carolina at (803)-777-7095.

Thank you for your time and consideration to participate in this study. Your responses and input will help place genetic counselors in a unique position to foster independence and self-management skills in young adolescents with Turner syndrome transitioning to adult-centered healthcare.

Sincerely,
Molly Snyder

Appendix C: Survey Questions and Materials

Survey Instructions

Instructions for Completing this Survey Tool

Our survey will be administered through SurveyMonkey™, which is an online survey software. You will find that this survey tool includes a variety of different question types. The following explanations will serve as a guide to help you work through completing this survey. After completing each series of questions, you will be taken to the next set of questions by clicking the “Next” button at the bottom of the page. You may return to different sections of the survey at anytime by clicking back and forth between the “Previous” and “Next” buttons. Your survey is not officially complete until you select the “Submit” button after completing the final page of this survey. Additionally, I have included a list of important and useful definitions at the end of these instructions for you to refer to while completing this survey.

NOTE: The questions in this section are just examples. You do NOT have to answer the sample questions.

Rating Scale: The majority of survey questions will consist of a general statement followed by a series of scaled responses that reflect your extent of agreement with the statement. For example, if the general statement was, “I consider myself to be a friendly person,” you will select the response that most accurately reflects your agreement or disagreement with that statement by clicking on the circle.

1. I consider myself to be a friendly person.

Strongly Agree/Agree/Somewhat Agree/Somewhat Disagree/Disagree/Strongly Disagree

Yes or No Response: Another significant portion of our questions will have “yes” or “no” responses. These will resemble the following example and give you the option to click on the yes circle or no circle. Some questions might also include the option to select “unknown” when applicable.

2. I have a pet cat. Yes/No

Comment Box: Other questions will prompt you with a specific task or direction and include a comment box for you to enter your response in. These questions do not have formal word limits, so please feel free to add as little or as much information as you feel necessary to provide an answer.

3. Tell me more about your work experience.

Drop Down: A few questions include a drop down menu for you to select your response from. These questions will only allow you to select the single best answer from a list of options.

4. I have ____ sibling(s).

Multiple Choice – One Response: There will also be a few multiple choice questions. For questions that include “circles” next to the possible answers, you will only be able to select the single best response. For example, your response to the question below could only be 1 of the 4 provided options.

5. My favorite season is _____. Spring/Summer/Fall/Winter

Multiple Choice – More Than One Response: For other multiple choice questions, you will be able to select multiple responses. These questions will be marked with squares next to the list of possible responses. Please feel free to select as many or as few responses as you feel appropriate.

6. I enjoy listening to _____ music. Country/Hip-Hop/Pop/Bluegrass/Alternative/Rock

Important Definitions

You may find that not all terminology and vocabulary used in this survey is familiar to you. Please refer to this list of definitions if you are confused about what a certain question is asking or need help defining a specific word. Again, you may contact Molly Snyder at 704-564-3429 or snyderme@email.sc.edu with any questions.

Multidisciplinary Healthcare = The utilization of a number of healthcare professionals with specialized skills and expertise to provide collaborative and integrative care. This approach to health cares for the patient as a whole and addresses his/her emotional, social, and mental needs.

Empower = To motivate or make one stronger and more confident, especially in controlling one's life and claiming one's rights.

Haploinsufficiency = A situation in which having a single copy of the normal gene(s) does not provide the necessary quantity or quality of molecular/chemical/biological products.

Demographics and Background

7. I am ____ years old.

8. Ethnicity:

White - Hispanic or Latino/a - Black or African American - Native American or American Indian – Asian - Other (please specify)

9. I live in the _____ region of the United States. Southeast

Northeast/Midwest/Southwest/Northwest

10. I was diagnosed with Turner syndrome _____.

before birth/soon after birth (i.e. as a newborn)/between the ages of 1 and 5/between the ages of 6 and 11/between the ages of 12 and 18/after the age of 18

11. My _____ diagnosed me with Turner syndrome. Other (please specify)

12. What is your marital status? Single (never married)

Married or Domestic Partnership/Divorced/Widowed/Separated/Other (please specify)

13. Highest educational achievement:

No schooling completed

I have completed some grade school (i.e. elementary, middle, or high school)

I have graduated high school or earned my GED

I have completed some postsecondary education

I have completed some trade/technical/vocational training

I have graduated from a postsecondary education program, received a Bachelor's degree

I have completed some postgraduate education (i.e. graduate, medical, law school, etc.)

I have received a Master's degree

I have received a Professional degree

I have received a Doctorate degree

14. I am currently working as a _____. (i.e. student, stay-at-home mom, company manager, lawyer, physician, entertainer, volunteer coordinator, advocacy speaker, etc.)

15. _____ is/are who I go to for support. My parents/A close friend/My sibling(s)/A mentor figure/Other (please specify)

16. A genetic counselor was present when my parents were informed of my diagnosis.

I was informed of my diagnosis directly as an adult/Yes/No/Unknown

17. The healthcare provider who manages most of my care is a _____. Other (please specify)

18. What specialists do you visit regularly? Heart Doctor

Kidney Doctor/Ear, Nose, and Throat Doctor/Skin Doctor/Endocrinologist/Geneticist/Genetic Counselor/Other (please specify)

19. What specialists have you seen in the past? Heart Doctor

Kidney Doctor/Ear, Nose, and Throat Doctor/Skin Doctor/Endocrinologist/Geneticist/Genetic Counselor/Other (please specify)

Perceived Control and Confidence in Healthcare Transition

20. I am confident in my ability to explain my special healthcare needs to friends and family members.

Strongly Agree/Agree/Somewhat Agree/Somewhat Disagree/Disagree/Strongly Disagree

21. I am confident in my ability to explain my diagnosis to my healthcare providers.

Strongly Agree/Agree/Somewhat Agree/Somewhat Disagree/Disagree/Strongly Disagree

22. I am confident in my ability to communicate my medical needs to my healthcare providers.

Strongly Agree/Agree/Somewhat Agree/Somewhat Disagree/Disagree/Strongly Disagree

23. I am able to locate any health-related information and paperwork quickly and easily.

Strongly Agree/Agree/Somewhat Agree/Somewhat Disagree/Disagree/Strongly Disagree

24. I know when and how to ask appropriate questions during my medical appointments.

Strongly Agree/Agree/Somewhat Agree/Somewhat Disagree/Disagree/Strongly Disagree

25. I feel that my healthcare providers understand all aspects of my Turner syndrome diagnosis.

Strongly Agree/Agree/Somewhat Agree/Somewhat Disagree/Disagree/Strongly Disagree

26. I feel that I have sufficient knowledge on all aspects of Turner syndrome.

Strongly Agree/Agree/Somewhat Agree/Somewhat Disagree/Disagree/Strongly Disagree

27. I feel that I have achieved complete independence in my healthcare.

Strongly Agree/Agree/Somewhat Agree/Somewhat Disagree/Disagree/Strongly Disagree

28. I achieved full independence in my healthcare by the age of _____. 14-15

16-17/18-20/21-25/26-30/I do not feel that I have achieved full independence in my healthcare.

Existing Resources for Healthcare Transition

29. My transition to adult-centered healthcare was well coordinated.

Strongly Agree/Agree/Somewhat Agree/Somewhat Disagree/Disagree/Strongly Disagree

30. My doctors began to shift their attention away from my parents and ask me questions about my healthcare needs directly between the ages of _____:

12 and 13/14 and 15/16 and 18/After the age of 18

31. I felt well supported throughout my transition to adult-centered healthcare.

Strongly Agree/Agree/Somewhat Agree/Somewhat Disagree/Disagree/Strongly Disagree

32. I was able to easily identify adult-care providers that were knowledgeable of Turner syndrome during my healthcare transition.

Strongly Agree/Agree/Somewhat Agree/Somewhat Disagree/Disagree/Strongly Disagree

33. I wish that I had access to more resources and supports throughout the healthcare transition process.

Strongly Agree/Agree/Somewhat Agree/Somewhat Disagree/Disagree/Strongly Disagree

34. I would have appreciated more support in the following areas: (please list below)

35. I am satisfied with how I was transitioned to adult-centered healthcare.

Strongly Agree/Agree/Somewhat Agree/Somewhat Disagree/Disagree/Strongly Disagree

Healthcare Transition Educational Needs

36. Has genetic counseling ever been offered to you? If so, when were you presented with this option? Yes/No

37. I met with a genetic counselor about my diagnosis of Turner syndrome between the ages of _____.

Before 12/12 and 13/14 and 15/16 and 18/18 and 23/Over 23/I have never met with a genetic counselor

38. I was well educated about and fully understand the potential future health problems I may face as an adult with Turner syndrome.

Strongly Agree/Agree/Somewhat Agree/Somewhat Disagree/Disagree/Strongly Disagree

39. I was effectively counseled about how my condition might affect me in adulthood between the ages of _____.

12 and 13/14 and 15/16 and 18/18 and 23/After the age of 23/ I have not been counseled.

40. My ability to share in decisions made about the management of my condition is:

0 (Very Poor) 1 2 3 (Satisfactory) 5 6 7 8 (Very Good)

41. I wish to learn more about the genetic principles underlying Turner syndrome.

Strongly Agree/Agree/Somewhat Agree/Somewhat Disagree/Disagree/Strongly Disagree

42. One of my healthcare providers has effectively explained the concept of haploinsufficiency in the context of my condition, Turner syndrome.

Yes/No/I am not familiar with the concept of haploinsufficiency.

43. My healthcare providers listen to my opinion and personal preference for an estrogen regimen when making decisions about hormone replacement therapy in my adult years.

Strongly Agree/Agree/Somewhat Agree/Somewhat Disagree/Disagree/Strongly Disagree

44. I was educated on the topics of fertility and reproductive options for women with Turner syndrome between the ages of _____.

12 and 13/14 and 15/16 and 18/18 and 23/23 or above/Never

45. I have experienced a cardiovascular complication and/or have a heart defect. (Examples: hypertension, bicuspid aortic valve, coarctation of the aorta, aortic dilation/dissection, etc.) Yes/No

Cardiovascular Risks

46. I carry medical information in my wallet or on a bracelet in order to alert medical personnel to my heart condition.

Yes/No

47. I have been educated about the associated risks of my heart condition and understand the need to keep medical appointments and monitor my health.

Strongly Agree/Agree/Somewhat Agree/Somewhat Disagree/Disagree/Strongly Disagree

Pregnancy and Family Planning

48. I intend to pursue or have already pursued pregnancy as an adult woman with Turner syndrome. Yes/No

Pregnancy Education

49. I was educated about the medical complications and risks associated with pregnancy by a healthcare provider between the ages of _____.

12 and 13/14 and 15/16 and 18/18 and 23/23 and over/Never

Family Planning

50. I intend to pursue or have pursued other family planning options. Yes/No

Access and Involvement of Multidisciplinary Healthcare Teams

51. My healthcare providers actively listen and respond to my healthcare needs/concerns.

Strongly Agree/Agree/Somewhat Agree/Somewhat Disagree/Disagree/Strongly Disagree

52. My healthcare providers empower me to take responsibility of my healthcare.

Strongly Agree/Agree/Somewhat Agree/Somewhat Disagree/Disagree/Strongly Disagree

53. My healthcare providers connected me to a sufficient number of supports in the community as I prepared for my transition to adult-centered healthcare.

Strongly Agree/Agree/Somewhat Agree/Somewhat Disagree/Disagree/Strongly Disagree

54. I would have enjoyed meeting with a health professional in private as an adolescent (i.e. between 12 and 18) to address my concerns about the healthcare transition process.

Strongly Agree/Agree/Somewhat Agree/Somewhat Disagree/Disagree/Strongly Disagree

55. My healthcare providers serve as advocates in the following areas of my life: (select all that apply) Work and Employment

School and Education/Personal Life/Healthcare Insurance/None/Other (please specify)

56. I wish that my healthcare providers served as advocates in other areas of my life.

Yes/No

57. I appreciate receiving care from multiple health specialists.

Strongly Agree/Agree/Somewhat Agree/Somewhat Disagree/Disagree/Strongly Disagree

I do not receive care from multiple health specialists.

58. I believe that by receiving care from multiple specialists, I was more prepared to transition to adult-centered healthcare.

Strongly Agree/Agree/Somewhat Agree/Somewhat Disagree/Disagree/Strongly Disagree

I have not received care from multiple health specialists.

59. While I receive care from multiple specialists, I would appreciate more coordinated healthcare.

Strongly Agree/Agree/Somewhat Agree/Somewhat Disagree/Disagree/Strongly Disagree

Improving Health Outcomes and Satisfaction

60. My healthcare providers express interest in my _____ well being. Please select all answers that apply from the list of options.

Physical/Social/Emotional

61. My healthcare providers are sensitive to my diagnosis.

Strongly Agree/Agree/Somewhat Agree/Somewhat Disagree/Disagree/Strongly Disagree

62. My healthcare providers work as a team, utilizing each person's specialized skills and expertise.

Strongly Agree/Agree/Somewhat Agree/Somewhat Disagree/Disagree/Strongly Disagree

63. I feel that my healthcare is comprehensive.

Strongly Agree/Agree/Somewhat Agree/Somewhat Disagree/Disagree/Strongly Disagree

Development of Self-Management Skills

64. I am highly motivated to manage my own healthcare and medical needs.

Strongly Agree/Agree/Somewhat Agree/Somewhat Disagree/Disagree/Strongly Disagree

65. My healthcare providers prioritize my inclusion in my medical management.

Strongly Agree/Agree/Somewhat Agree/Somewhat Disagree/Disagree/Strongly Disagree

66. I wish to learn more about Turner syndrome and what implications my diagnosis has for my future healthcare management.

Strongly Agree/Agree/Somewhat Agree/Somewhat Disagree/Disagree/Strongly Disagree

67. I wish that I had more resources to provide to my healthcare providers regarding my diagnosis of Turner syndrome and associated medical needs.

Strongly Agree/Agree/Somewhat Agree/Somewhat Disagree/Disagree/Strongly Disagree

68. I was counseled as an adolescent (i.e. between the ages of 12 and 18) regarding ways to promote the development of independent, self-care behaviors.

Strongly Agree/Agree/Somewhat Agree/Somewhat Disagree/Disagree/Strongly Disagree

69. Please feel free to comment on any additional information and/or your feelings towards your transition to adult-centered care in the box below.

Agreement to Follow-Up Virtual Interview

70. As a second part of our study, we would like to interview participants about a few more details regarding their healthcare transition and confidence in their self-management skills. We plan to conduct these interviews via Adobe Connect, a secure server that will ensure interviews remain confidential. At the time of the interview, participants will receive an email that will transfer them to a safe connection with the principle study investigator. Thus, individuals must have access to a computer to participate. These interviews will take approximately 20 minutes and will include similarly themed question as those in our survey. If there are technical difficulties connecting to Adobe Connect for the interview, the primary researcher, Molly Snyder, will reach out to you with a second email in an attempt to correct the situation. Participants will also be provided with Molly's contact information if she has any questions or concerns about the interview.

If you would be interested in participating in a follow-up interview at a later date, then please select yes below. If you would not like to be contacted, please select no. If you are selected for an interview, you will be contacted via email by Molly Snyder at snyderme@email.sc.edu so that you can select a convenient date and time for your interview. While we do ask for your phone number and email address, the interviews will be interpreted anonymously and your survey responses will not be linked to your contact information.

Thank you for your time and consideration!

Yes/No

71. I would prefer to be contacted about a virtual interview first by phone at:

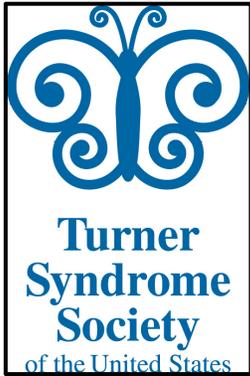
72. I would prefer to be contacted about a virtual interview first by email at:

Follow-Up Interview Questions:

1. What aspects of Turner syndrome would you like to learn more about?
2. How were you educated on the etiology (i.e. cause) of Turner syndrome as an adolescent?
3. Do you feel having a better understanding of Turner syndrome would help you feel more empowered? If so, how might it improve your self-management skills? If not, why?
4. What is the greatest healthcare challenge you've experienced?
5. In what areas did you lack support during your transition to adult-centered healthcare?
6. During which parts of your healthcare transition did you feel most supported?
7. How do you understand the roles and responsibilities of a genetic counselor?
8. What resources and supports did you find most helpful during the transition process?
9. What did you find most enjoyable about the healthcare transition process?
10. Is there anything you'd like to tell me that I haven't asked you about?

Study Participation Opportunity!

The University of South Carolina School of Medicine



Reflections on the Current State of
Healthcare Transition for Young
Adult Women with Turner
Syndrome: Strategies for Facilitating
Autonomy and Self-Management

Who: Women ages 18 to 30 with a formal diagnosis of Turner syndrome who speak English and have access to the Internet.

When: September 2015 to January 2016

Where: Online survey tool on SurveyMonkey™ (~20 minute time commitment) <https://www.surveymonkey.com/r/uscgeneTS>

Why: The purpose of this research study is to invite women to reflect on their healthcare experiences and to learn more about how comfortable young adult women feel when explaining their condition to peers, family, and healthcare professionals. We hope to identify ways in which genetic counselors might serve as advocates and help women develop self-management skills beginning in early adolescence.

If you are interested in learning more about our study, please call the principal investigator, Molly Snyder, or her thesis advisor, Dr. Richard Ferrante, PhD, at the contact information below.

Molly Snyder – Phone: (704)-564-3429

Email: snyderme@email.sc.edu

Dr. Richard Ferrante – Phone: (803)-414-0299

Email: Richard.Ferrante@uscmed.sc.edu

Appendix D: University Materials

INSTITUTIONAL REVIEW BOARD FOR HUMAN RESEARCH APPROVAL LETTER FOR EXEMPT REVIEW

This is to certify that the research proposal: **Pro00045298** – Entitled: *Reflections on the Current State of Healthcare Transition for Young Adult Women with Turner Syndrome: Practical Strategies for Facilitating Autonomy and Self-Management*

Submitted by:

Principal Investigator: Molly Snyder
College: School of Medicine
Department: Genetic Counseling – Columbia, SC 29203

was reviewed in accordance with 45 CFR 46.101(b)(2), the referenced study received an exemption from Human Research Subject Regulations on **6/15/2015**. No further action or Institutional Review Board (IRB) oversight is required, as long as the project remains the same. However, the Principal Investigator must inform the Office of Research Compliance of any changes in procedures involving human subjects. Changes to the current research protocol could result in a reclassification of the study and further review by the IRB.

Because this project was determined to be exempt from further IRB oversight, consent document(s), if applicable, are not stamped with an expiration date. Research related records should be retained for a minimum of three (3) years after termination of the study.

The Office of Research Compliance is an administrative office that supports the University of South Carolina Institutional Review Board (USC IRB). If you have questions, contact Arlene McWhorter at arlenem@sc.edu or (803) 777-7095.

Sincerely,

Lisa M. Johnson
IRB Manager