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Readiness for transition to adult care in adolescents and young adults with Turner syndrome

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Abstract

Objectives: Turner syndrome (TS) is a complex and chronic medical condition that requires lifelong subspecialty care. Effective transition preparation is needed for successful transfer from pediatric to adult care in order to avoid lapses in medical care, explore health issues such as fertility, and prepare caregivers as adolescents take over responsibility for their own care. The objective of this study was to evaluate accuracy of knowledge of personal medical history and screening guidelines in adolescents and young adults (AYA) with TS.

Methods: This was a prospective cross-sectional study of 35 AYA with TS of ages 13–22 years recruited from a tertiary care center. AYA completed questionnaires on personal medical history, knowledge of screening guidelines for TS, and the Transition Readiness Assessment Questionnaire (TRAQ).

Results: Eighty percent of AYA with TS were 100% accurate in reporting their personal medical history. Only one-third of AYA with TS were accurate about knowing screening guidelines for individuals with TS. Accuracy about knowing screening guidelines was significantly associated with TRAQ sum scores ($r = 0.45$, $p < 0.05$). However, there was no association between knowledge of personal medical history and TRAQ sum scores.

Conclusions: Transition readiness skills, TS-specific knowledge, and accurate awareness of health-care recommendations are related, yet distinct, constructs. Understanding of one's personal medical history is not an adequate surrogate for transition readiness. Validated tools for general transition, like the TRAQ, can be used but need to be complemented by TS-specific assessments and content. Providers are encouraged to identify opportunities for clinical and educational interventions well in advance of starting transfer to adult care.

Keywords: adolescent; continuity of patient care; female; health literacy; transition to adult care; Turner syndrome; young adult.

Introduction

Turner syndrome (TS) is a chromosome disorder of phenotypic females who have complete or partial absence of their second sex chromosome in association with one or more clinical manifestations [1]. The main characteristics include short stature, cardiovascular abnormalities (e.g., coarctation of the aorta, bicuspid aortic valve), premature ovarian failure, and infertility. Other features include hearing loss, recurrent ear infections, kidney anomalies, nonverbal learning and attention deficit hyperactivity disorders, and anxiety [1]. Youth with TS are primarily followed up by a pediatric endocrinologist for treatment of short stature, induction of puberty, and monitoring of autoimmune diseases and other comorbidities. Adult women with TS have specific lifelong health needs, but they are at risk of lapses in care or may be followed up by providers unfamiliar to comprehensive TS care guidelines

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[1, 2]. Therefore, women with TS benefit from knowledge of their condition and awareness of the rationale for ongoing TS-specific screening and prevention services to preserve optimal health [3].

Transition from pediatric to adult care is a developmentally normative task for all adolescents and young adults (AYA) [4]. However, for AYA with a chronic medical condition, especially one that may affect cognitive skills, the movement between family-oriented pediatric health systems to more independent adult-oriented health systems can present unique challenges [5]. The importance of objective tools to measure transition readiness over time is well documented in the chronic illness literature [6, 7]. These tools, such as the Transition Readiness Assessment Questionnaire (TRAQ), frequently emphasize patients' knowledge about their condition and capacity for self-management as central to transition readiness [8, 9].

Previous studies evaluating transition readiness across different chronic conditions identified that AYA with TS had significantly reduced TRAQ transition readiness scores compared to AYA without chronic medical conditions [5]. This remained true even after taking into account age, sex, ethnicity, maternal education, Individualized Education Plan status, and health literacy, indicating that the difference in transition readiness was likely due to unmeasured differences between groups, including specific characteristics of TS (see Table 1) [5]. With regard to specific transition readiness topics, AYA with TS demonstrated variability across different domains, scoring highest in daily independent activities such as preparing meals and going to the grocery store or pharmacy, but scoring the lowest in talking to their health-care provider [5]. Given that AYA with TS will require lifelong interfacing with health-care systems, a better understanding of TS condition–specific knowledge is needed. The purpose of this pilot study was to assess knowledge of personal health history and TS-specific screening guidelines in AYA with TS and to determine if this personal health and TS-specific knowledge predicts transition readiness.

Table 1: Descriptive statistics of AYA with TS.

Demographic	AYA with TS
N	35
Mean age (SD), years	16.92 (2.47)
Percentage (n), female sex	100% (35)
Percentage (n), minority	12% (4)
Percentage (n) with classic 45,X karyotype	48%(17)
Percentage (n), mothers with more than a high school diploma	66% (23)
Percentage (n), single-headed households	20% (7)
Percentage (n), AYA with an IEP	57% (20)
Mean health literacy (SD)	12.0 (1.63)
Mean TRAQ score (SD)	3.04 (0.79)

AYA = adolescents and young adults; TS = Turner syndrome; IEP = Individualized Education Plan; TRAQ = Transition Readiness Assessment Questionnaire; SD = standard deviation.

Methods

Participants

As part of a larger study [5], 35 AYA with TS of 13–22 years of age wererecruited during routine visits to the endocrinology clinic at Cincinnati Children's Hospital Medical Center. The AYA were eligible for the study if they were fluent in the English language, capable of answering questions independently, and able to provide consent or assent (where appropriate) for the study. Participants were excluded if they had an apparent moderate developmental disability (determined by nurse evaluation), were unable to answer questions independently, could not read at or above a third-grade level, or were already being cared for by an adult medical provider.

Procedures

Institutional review board approval was obtained for this study. Each eligible participant was given information about the study, and consent (>18 years of age) or consent and assent (<18 years of age) were obtained. All participants were asked to either complete questionnaires in the clinic or complete questionnaires at home and mail them back to the research coordinator. The participants were instructed to complete the questionnaires independently. Each of the participants was provided with a $15 gift card for their time and participation.

Measures

Transition readiness was assessed by the TRAQ 5.0 [8, 9]. The TRAQ 5.0 is a 20-item validated assessment of transition readiness. The TRAQ examines knowledge and health-related skills in the following areas: disease characteristics, medical history, medications, treatment history, health service resources, and navigation of the health-care system. Item responses are on a five-point Likert-type scale from 1 (No, I don't know) to 5 (Yes, I always do this when I need to). Responses are averaged to determine a total score ranging from 1 to 5, with one indicating low readiness to transition and five indicating high readiness to transition [9, 10].

The participants answered questionnaires about their personalmedical history, the frequency of various recommended screenings associated with TS, and knowledge of typical prevention guidelines for individuals with TS (see Appendix 1). These questions were derived

from existing screening guidelines [11, 12]. Chart reviews were conducted to compare participant responses with regard to medical history to the electronic medical record and scored for accuracy. Personal medical history was assessed with 14 items with yes, no, or I don't know response options. Accuracy was verified based on chart review by a research nurse. Recommended screening guidelines were assessed by 12 items with multiple-choice answer response for frequency of recommended screening: every visit, once a year, every 2–5 years, no need, or I don't know.

Statistical analysis

Descriptive statistics and accuracy rates were calculated for questions about personal medical history, screening recommendations, and prevention guidelines for individuals with TS. Pearson's correlations (r) were used to estimate the rank-order association between TRAQ scores and accuracy of personal medical history, screening recommendations, and prevention guidelines for individuals with TS. Correlations of r = 0.1–0.29 are considered small, r = 0.3–0.49 are considered medium, and r = 0.5 or higher are considered large effect sizes [13]. A p-value of <0.05 was used to determine statistical significance. All of the analyses were completed using SPSS 17.

Results

Thirty-five AYA with TS aged 13–22 years (mean = 17.9 years, standard deviation [SD] = +2.5 years) were enrolled in the study. Descriptive statistics are provided in Table 1.

Eighty percent (80%; n = 28) of AYA with TS were 100% accurate in reporting their own personal medical history (Figure 1) and what hormone therapies they had received thus far (e.g., growth hormone, thyroid hormone, estrogen therapy). However, only 33% of AYA with TS were accurate in their knowledge about the current screening recommendations and prevention guidelines for individuals with TS (e.g., frequency of heart imaging, blood pressure levels). Of note, accuracy among AYA with TS under the age of 16 years (n = 18) did not significantly differ from accuracy for

Table 2: Correlations (r) between TRAQ subscales and accuracy in AYA with TS.

TRAQ Subscale	Accuracy of personal medical history	Accuracy of personal treatment history	Accuracy of knowledge of TS guidelines
TRAQ sum score	−0.06	−0.02	0.36*
Medication subscale	−0.01	−0.02	0.41*
Appointment subscale	−0.01	0.03	0.39*
Health issue subscale	−0.05	−0.02	0.25
Provider subscale	−0.17	−0.10	0.09
Daily activity subscale	−0.09	−0.06	0.13

*p < 0.05.
AYA = adolescents and young adults; TS = Turner syndrome; TRAQ = Transition Readiness Assessment Questionnaire.

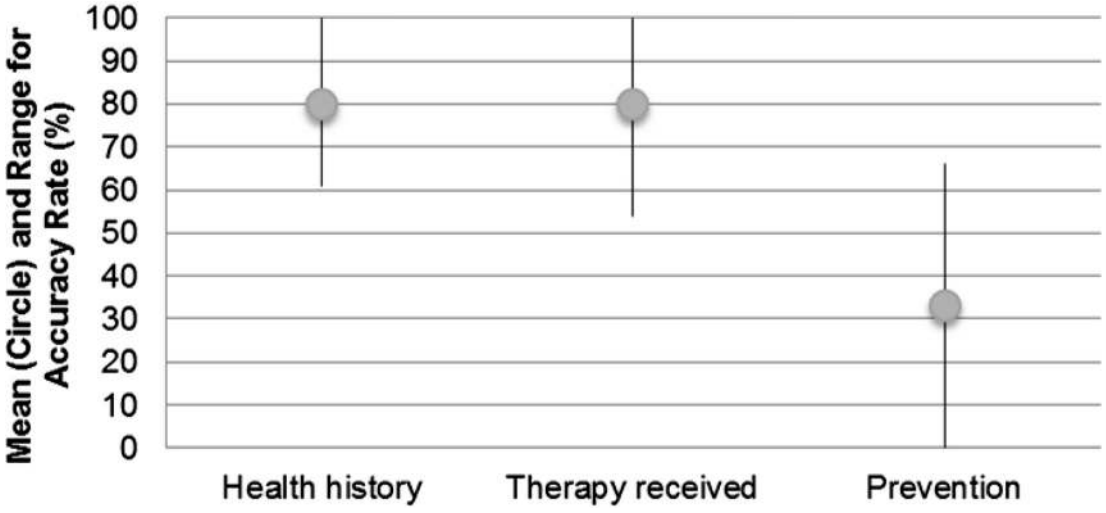

Figure 1: Accuracy in knowledge of personal medical history and screening guidelines in AYA with TS. AYA = adolescent and young adult; TS = Turner syndrome.

the full sample: personal history accuracy was 82.9% (SD = 9.9%), and accurate knowledge of guidelines for individuals with TS was 35% (SD = 18.9).

Accuracy about adult care guidelines (e.g., how often they should have heart imaging) was significantly associated with TRAQ sum scores ($r = 0.45$, $p < 0.05$), a large bivariate effect. The correlations between TRAQ scores and accuracy of personal medical history ($r = 0.04$, $p = 0.85$) indicate that TRAQ scores do not have a significant or clinically meaningful association with knowledge of personal medical history in this population.

Further analysis showed accuracy of knowledge about adult care guidelines was significantly associated with both the medication subscale of the TRAQ and the appointment subscale of the TRAQ ($rs = 0.41$ and 0.39, respectively, $ps < 0.05$). However, there was no significant correlation for accuracy of personal medical history or personal treatment history and the TRAQ subscale scores (see Table 2).

Discussion

TS is a complex and chronic medical condition that requires lifelong subspecialty care including (but not limited to) cardiology, gynecology, endocrinology, otolaryngology, and psychology. It is widely recognized that adult women with TS may have lapses in care with consequent unrecognized and undertreated health conditions as a result. Effective transition preparation is needed for successful transfer from pediatric to adult care and requires knowledge of personal history and future screening recommendations, as well as skills to navigate and communicate in health care. This pilot study of AYA with TS highlights opportunities to address gaps in health knowledge, adult screening guideline awareness, and health navigation skills to promote successful transition. The methodology we used in the present study can be applied to other complex chronic conditions.

This is the first study, to our knowledge, that evaluates accuracy of personal medical history among a cohort of AYA with TS. Participant responses indicated strong familiarity with personal medical history (e.g., if they have heart issues or kidney issues themselves) and what hormone therapies they have had (e.g., growth hormone, thyroid hormone, estrogen therapy). However, these same participants also showed less knowledge about future screening recommendations and prevention guidelines for individuals with TS. This is an important finding because the TS population is at high risk of increased morbidity and mortality (e.g., cardiac, metabolic, bone health complications), which can be mitigated by following appropriate screening and prevention guidelines [1, 2, 11, 12]. The current study points to an opportunity to improve education about the importance of continuity in medical care for AYA with TS, with a particular focus on ongoing health-care needs and what screenings are necessary in adulthood. In pediatrics, the pediatric endocrinologist often takes on the role of coordinating care for individuals with TS. The adult provider who will assume this role (typically a primary care provider, adult endocrinologist, or gynecologist) should be identified during the transition process as well so that screening and prevention guidelines can be shared with the receiving provider. Existing health navigation tools, such as the Endocrine Society TS transition guidelines (http://www.endocrinetransitions.org/turner-syndrome/) and a personal TS passport, are available to assist with transition preparation and education prior to transfer to adult-oriented care [12, 14].

Despite acceptable health literacy scores [5] and the finding that AYA with TS are accurate about personal health history, AYA with TS had midrange transition readiness scores, lower than their peers without any chronic medical conditions [5]. The lowest readiness to transition score for AYA with TS was in the area of communication with a health-care provider. An expanded definition of health literacy includes developing the "cognitive and social skills to articulate health needs and preferences" [15]. Findings from this pilot study demonstrate that although AYA with TS have good knowledge of their own health, they often lack necessary social communication skills (e.g., effective communication with a health-care provider) for transfer to adult care. There is a need for more comprehensive transition readiness preparation specifically designed for AYA with TS, such as in teaching health-care navigation, making and tracking appointments, and social communication and pragmatic language skills to effectively interact in a health-care setting. In the setting of a TS multidisciplinary team, it is important to clarify roles of the team members in the transition process. For example, the psychologist can assess and aid in the development of organizational, social, and communication skills for the TS individual, while a medical provider focuses more on adult health screening guidelines.

When examining the TRAQ subscales, questions related to medications (i.e., Do you take medication correctly on your own?) and appointments (i.e., Do you keep a calendar or list of medical appointments?) were significantly correlated with increased accuracy of TS-specific adult care guidelines. This highlights that AYA with TS who have higher TRAQ scores on domains where executive functioning and problem-solving skills are required may also better understand or have more awareness of TS guidelines. In practice, focusing TS-specific transition education on organization and problem-solving skills may aid in promoting transition readiness.

The findings of the present study should be interpreted in the context of several limitations. First, AYA with TS in this study get comprehensive care at Cincinnati Children's Hospital Medical Center, where they receive written and verbal information about their condition at each visit (as frequently as every four months). That 80% of young women had 100% accuracy in knowledge of personal medical history could reflect the robust role of the care and supportive materials provided at the center. Secondly, the pilot study was unable to analyze the effects of mental health or neurocognitive concerns, such as anxiety. It is possible that AYA with TS who have more anxiety may need more skills in order to communicate effectively with providers. Generalizability of results is limited by the single-center pilot study design and small sample size with a wide age range of 13–22 years. It was expected that younger AYA with TS would have lower levels of readiness, but they were included as the most recent clinical guidelines recommend starting transition counseling at the age of 12 years [7]. When we did analyze the data from AYA with TS who were less than 16 years, there was no significant difference in their knowledge of personal medical history or of the screening guidelines. Expansion to multiple centers to increase the sample size and reflect variation in practice patterns is an important consideration for future studies.

Overall, the findings from this pilot study support that transition readiness skills and TS-specific knowledge about their medical condition and health-care recommendations are related, yet distinct, constructs. Understanding of one's personal medical history, while important, is not an adequate surrogate for transition readiness. Providers should start involving young girls with TS in their own medical care discussions and decision-making early. This will help build their provider-patient communication skills and provide anticipatory guidance using strategies that enhance problem-solving and organizational skills about future adult care guidelines. Validated tools for general transition, such as the TRAQ, can be used in this population, but need to be complemented by TS-specific assessments and content such as fertility and risk of aortic dissection. Advanced health systems have successfully integrated a transition planning tool into an electronic medical record for an institution-wide approach to general transition readiness [16]. The results from our study suggest value in complementing general readiness skills with condition-specific content as an approach to support needs across a variety of subspecialty populations graduating into adult care settings. A useful tool for guiding these discussions is the patient-oriented summary of the updated International TS consensus guidelines (Clinical Practice Guidelines for the Care of Girls and Women with Turner Syndrome – Brief Synopsis for Turner Syndrome Girls and Women and for their Parents/Caregivers/Families). In summary, providers are encouraged to incorporate TS-specific resources into their transition preparation practices to consistently identify opportunities for clinical and educational interventions prior to starting the transfer to adult care.

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Informed consent: Informed consent was obtained from all individuals included in this study.

Ethical approval: Institutional review board approval was obtained for this study. Each eligible participant was given information about the study, and consent (>18 years of age) or consent and assent (<18 years of age) were obtained.

Data Availability: The data that support the findings of this study are available from the corresponding author upon reasonable request.

Appendix

	Yes	No	I Don't Know	Name of Product
Are you on estrogen therapy?				

Do you have any of the following?	Yes	No	I Don't Know	Medication (if needed)
Attention or learning problems				
Anxiety				
High blood pressure				
High cholesterol				
Diabetes (high blood sugar)				
Heart problems				
Hearing loss				
Kidney problems				
Liver problems				
Overweight				
Osteoporosis (weak bones)				
Thyroid disease				
Vision or eye problem				
Other (fill in)				

How often should the following be checked?	Every Visit	Each year	Every 2-5 years	No Need	I don't know
Mood/ Learning issues					
Blood Pressure					
Cholesterol					
Heart imaging (Echo or MRI)					
Hearing					
Kidney function					
Liver function					
Weight					
Bone health (DXA scan)					
Gynecology exam (Pap test)					
Thyroid hormone levels					
Vision/ eye exam					
Other (fill in)					

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