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## Turner Syndrome: Four Challenges Across the Lifespan

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### Abstract

Turner syndrome (TS) is a sex chromosome condition that occurs in approximately 1/2500 live female births. Despite the prevalence of this chromosomal condition, the challenges these women face throughout their lives are not fully understood. This qualitative research study aimed to characterize the subjective experiences of individuals with Turner syndrome throughout their lifespan, to investigate their concerns and obstacles, and to offer insight into the strengths and weaknesses of health care delivery, as they perceived them.

Ninety-seven girls and women with TS and 21 parents consented to participate in this interview study. Interviews were semi-structured and open-ended in design. Questions sought to elicit responses relating to existing concerns associated with their condition and positive and negative health care experiences. Participants were divided into four age categories (childhood, adolescence, adulthood, and mature adulthood) to facilitate a comparative analysis across the age spectrum.

Regardless of age, infertility was the most frequently cited concern followed closely by short stature. Sexual development and function and general health were also viewed as challenges by a number of participants in each age group.

Although the relative weight of these four concerns tended to shift based upon the individual's age and life experiences, all four issues remained significant throughout the lifespan. Enhanced awareness of the evolving physical and psychological challenges faced by girls and women with TS may help health care providers improve the quality of life for these individuals.

### Keywords

Turner syndrome; infertility; short stature; sexual development; health care providers

## INTRODUCTION

Turner syndrome is a relatively common condition due to the full or partial deletion of the second sex chromosome, resulting in a 45,X karyotype and a female phenotype. Some individuals with TS have a mosaic 46,XX/45,X (or less frequently 45,X/46,XY) karyotype, and others demonstrate the phenotype due to a major X-chromosome deletion (46,X del (X)). Physical features associated with a TS karyotype may include: ovarian failure with associated infertility, short stature, cardiovascular and renal malformations, hypertension, diabetes, hearing loss, skeletal abnormalities and neck webbing [Lippe 1991; Sybert 2001; Cunniff 2002; Elsheikh et al. 2002]. Girls and women with TS generally have a normal range of cognitive abilities but may have learning difficulties, particularly related to nonverbal skills such as visual-spatial abilities, motor coordination, and mathematics [Skuse 1987; Sylven et al. 1993; Saenger 1996; Lagrou et al. 1998; van Borsel et al. 1999; McCauley et al. 2001; Ross 2001; Sybert 2001; Elsheikh et al. 2002; Frias and Davenport, 2003].

A number of studies on Turner syndrome provide recommendations to health care providers on how to improve the overall physical and psychological health of girls and women with TS [Rieser 1992; Sylven et al. 1993; Saenger 1996; Rosenfeld 2000; Savendahl and Davenport 2000; Elsheikh et al. 2002; Frias and Davenport 2003]. Published timelines for administering various tests and explanations of symptoms and complications of TS serve as references for health care providers [Sylven et al. 1993; Saenger 1996; Savendahl and Davenport 2000; Ross 2001; Sas and de Muinck Keizer-Schrama 2001; Cunniff 2002; Elsheikh et al. 2002; Frias and Davenport 2003]. Although general information addressing future health concerns of women with TS exists, such as osteoporosis, hearing loss, and cardiovascular disease, the majority of women with TS are not monitored adequately for these late-onset diseases and conditions [Ross 2001; Elsheikh et al. 2002]. Consequently, less is understood about the overall morbidity and mortality of women with TS and many women are left with unanswered questions about their future [Sybert 2001]. In addition to the relative lack of research into the consequences of TS on affected adults, few studies present the challenges associated with TS and areas of needed improvement in health care as articulated by girls and women living with the condition [Sylven et al. 1993].

The goal of this qualitative research study was to explore concerns experienced by girls and women with Turner syndrome across the lifespan, specifically from childhood to mature adulthood. To the best of our knowledge this is the largest data collection of its kind within the Turner syndrome population. A comparative analysis of this data allowed both an exploration of the challenges affecting girls and women with TS across the ages, as voiced by the girls and women themselves, and an opportunity to identify whether age distinctions exist among commonly cited TS concerns. A secondary objective of this study was to offer girls and women with TS, as well as their parents, an opportunity to reflect and comment on their past and present health care experiences and propose suggestions as to how providers might improve the future delivery of health care to other individuals with Turner syndrome.

## METHODS

### Study Design

This qualitative research study was conducted in conjunction with the IRB approved National Institute of Child Health and Development's (NICHD) Turner Syndrome: Genotype and Phenotype study. Individuals who were pregnant, had co-existing autosomal chromosomal anomalies, had been on androgen treatments within the past 6 months, and/or had severe physical or mental disabilities preventing informed and consenting participation in the research were ineligible for the NICHD study.

From January 2001 to May 2003, 97 girls and women, ranging in age from 7–59 years, participating in the NICHD parent study agreed to participate in this IRB approved interview study. Twenty-one parents of children and adolescents also consented to participate: of these, 4 were interviewed on behalf of their young daughters who were too young to participate, 7 were interviewed in conjunction with their young daughters, and the remaining 10 were interviewed separately from their adolescent daughters. No one invited to participate declined participation in this study. For the purpose of analysis we used the parent interviews of the four youngest girls who were too young to be interviewed as the source of their data. This was noted accordingly in the reporting of results.

In-person interviews were conducted primarily by two genetic counselors (AM & BB), were semi-structured in design, audio-taped, and lasted approximately 60–90 minutes. The open-ended questions addressed the time of diagnosis, various past and present health conditions, concerns and challenges of living with TS, sources of information and support, advantages of living with TS, and suggestions for health care providers (HCPs) on how to improve the health care experiences of girls and women living with TS.

## Data Analysis

The audiotaped interviews were transcribed verbatim to facilitate the qualitative analysis. The initial codebook was designed based on the reading of the first few transcripts, listing topics underscored throughout the interview sessions. This analysis focused on the concerns and challenges of living with TS and suggestions for HCPs.

Five independent coders (ES, AM, SG, DK, BB) applied the codes to the transcripts using commonly accepted coding techniques [Coffey and Atkinson 1996]. Codes were then entered into the NUD\*IST database, a software package for qualitative data management, to aid in sorting the data and identifying themes. Illustrative quotations were then culled to convey those themes. Given the large number of coders, several steps were taken to ensure coding consistency. Frequent group meetings were conducted throughout the coding process to discuss how the codes were being applied and to reconcile any differences. Two coders (ES, DK) were responsible for reviewing the NUD\*IST output to ensure codes were applied correctly and obvious miscodings were corrected.

To capture the evolution of life challenges and concerns as experienced by girls and women with TS, participants were divided into the following four categories according to age: Childhood (7–13 years of age), Adolescence (14–19 years of age), Adulthood (20–39 years of age), and Mature Adulthood (40–59 years of age). The age division was based upon efforts to distribute participants evenly amongst groups and closely approximates the psychosocial stages of development as forwarded by theorist, Erik Erikson [Erikson 1963; Corey 2001]. The prevalence of each challenge associated with TS was determined by the frequency with which participants in each group cited the various concerns.

## RESULTS

### Sample Characteristics

A total of 97 girls and women with Turner syndrome and 21 parents participated. Table I depicts the demographics of the study population. A majority of participants were Caucasian and ranged from 7–59 years of age. The average age of diagnosis was 12.8 years and the average height of women 18 years of age and older was 4'9". Of these adults, 26% took human growth hormone at some point during their youth. The majority of the women interviewed had obtained either an undergraduate or graduate degree. The most common TS karyotype of the women in our study was 45,X. Of the 68 women between 20–59 years of age, approximately 32% were married and 15% were divorced. Six women had children, 5

through adoption and 1 as a result of *in vitro* fertilization (IVF). The women in this study underwent neurocognitive and neuropsychiatric testing as part of the NICHD parent study. Depression was assessed by the administration of the DSM IV Structured Clinical Interview and self-esteem was measured using the Rosenberg self-esteem scale [Rosenberg 1965;First et al. 1995]. Results revealed that women with TS had a higher rate of lifetime depression than the general population [Cardoso et al. 2004]. With respect to self-esteem, results showed that women rated lower on Rosenberg's scale than the normal control group yet exhibited similar levels of self-esteem as 46,XX women with infertility due to premature ovarian failure [Cardoso et al. 2004].

### Lifelong Concerns

In all four age categories the girls and women in this study, regardless of their karyotype, most frequently cited infertility as their primary concern associated with TS. Short stature was the second largest challenge. Interestingly, sexual development and function and health status were, comparatively, only ancillary concerns, yet prevailed over other concerns mentioned by participants during the open-ended questioning.

**Infertility**—Infertility emerged as the major concern for the majority of our participants. Thirty-six percent of girls aged 7–13 were, according to their parents, distressed upon learning of their inability to become biological mothers. Fifty-six percent of adolescents, aged 14–19, 74% of women between the ages of 20 and 39, and 62% of mature adults, ages 40–59, described feeling devastated when they received their infertility diagnosis. Although many women have adapted to their infertile state, their fertility diagnosis remains a sensitive issue. Table II provides a summary of the sentiments expressed by girls and women with respect to their infertility.

Contrary to initial expectations, girls diagnosed with Turner syndrome during childhood were, in many cases, still deeply affected by their infertility. Parents interviewed on behalf of their young daughters and older study participants reflecting on their childhood diagnoses recalled shattered dreams of motherhood. Adolescents also cited infertility as either a current or future concern. The inability to bear biological children was the most prevalent and painful challenge endured by most of the adult women interviewed, as they were continually reminded of their infertility as family and friends began to procreate. Although many women from our mature adult sample were beyond childbearing years, the majority of these women remained saddened by their infertility and felt deprived of an important part of womanhood and reproductive choice. Interestingly, even though the majority of participants embraced adoption and new reproductive technologies as legitimate options, their inability to have a biological child remained, for many, a source of great sadness. A minority of adults and mature adults, however, specifically mentioned that infertility was not an issue, predominantly because they were neither married nor in long-term relationships.

A number of women disclosed that their infertility had a significant impact on how they interacted with men, particularly within intimate relationships. Recognizing that their diagnosis of infertility would, in most cases, extend to their life partners, questions of when and how to disclose their infertility to their partner, concerns of being accepted despite their diagnosis, and fears of being abandoned as a result of their inability to have children preyed on the minds of these women.

**Stature**—Short stature was the second most cited concern. Sixty-one participants mentioned that their height bothered them at some point in their lives. Thirty six percent of children, 44% of adolescents, 44% of adults, and 55% of mature adults described being currently bothered by their short stature. Of those girls and women, 63% were less than 4'9"

and 40% were greater than or equal to 4'9". An analysis evaluating comments on short stature by women aged 18 years and older yielded the following results: 47% of the women less than 4'9" and 37% greater than or equal to 4'9" expressed being currently concerned about their height.

Another interesting finding was the accuracy with which the girls and women reported their height in the interviews. Even though each participant had undergone a physical examination prior to our interview, of the 69 girls and women who mentioned their height, 40 participants (59%) claimed to be taller than they actually were. The majority (29) rounded up their height anywhere from a fraction of an inch to an inch. However, 11 women exaggerated their height by one and a half to four inches. Table III provides a bulleted account of the findings addressing concerns with short stature.

Short stature affected almost all of the women in our study, either because they endured teasing over the years or were discouraged or frustrated by their dependence on other people to perform tasks such as driving or reaching things in high places. According to the girls and women interviewed, much of the teasing stemmed from the fact that their physical appearance differed from that of their peers and/or colleagues. For a number of women, height emerged during adolescence and adulthood as a greater obstacle than infertility. Fueling this concern was the belief that men would not view them as potential dating partners because they were shorter than most women. Desires to achieve a more balanced weight, a feat facilitated, they believed, by a few more inches, also contributed to their dissatisfaction with their height. Many women also related their ongoing frustrations with the rude and thoughtless comments of strangers, work colleagues, teachers, and even health care providers. Although only 12 participants of various ages explicitly stated height was not a concern, a handful of girls and women dissatisfied with their height did confess that being short had some advantages. Receiving positive attention from others, looking younger and cuter longer, being able to act like a kid, traveling comfortably, and the advantages of short stature in certain professions and sports were reported among the benefits of being short.

**Social Aspects of Sexual Development and Function**—A subgroup of participants (n=20) articulated that sexual development was a sensitive issue, particularly during adolescence. Much of the concern stemmed from delayed development, lack of sexual arousal and/or difficulty during intercourse. Table IV highlights the salient concerns of these women with respect to their sexual development and sexual functioning.

Adolescence was the time when adults and mature adults recalled being most concerned with their sexual development. A couple of adolescents expressed feeling different because they had not yet reached puberty and were, consequently, bothered by their immature figures. A number of adults and mature adults noted sexual development as a particular concern during their teenage years, especially when reflecting on their interactions with members of the opposite sex. A handful of adult and mature adult participants also expressed concerns surrounding issues of sexual identity—not feeling feminine, low sexual libido, and/or a general disinterest in intercourse. One forty-year-old woman viewed sex as a means to a childbearing-end and consequently, an activity that served as a constant and painful reminder of her infertility.

**Health**—Despite the medical complications experienced by many of these girls and women throughout their lives, few specified health as a primary concern. Sentiments pertaining to the uncertainty of aging with TS and the fears associated with the long-term risks of human growth hormone (HGH) and hormone replacement therapy (HRT) are exemplified in Table V.

A small number of children and adolescents briefly noted their increased susceptibility, as girls with TS, for diabetes and osteoporosis. Many participants acknowledged the potential for future health complications yet were not overly concerned, provided they continued to take care of themselves by eating well, exercising, and addressing medical issues in a timely fashion. The overwhelming majority of comments relating to health reflected women's feelings of uncertainty, particularly as they age, as less is known about the specific future manifestations of their condition. A number of participants expressed worries surrounding their mortality, questioning specifically whether TS results in a reduced lifespan. Others wished they had more information on aging with TS in addition to more specific medical guidance on when to cease using HRT.

**Parental Concerns and Challenges**—Parents of children and adolescents diagnosed with Turner syndrome were included in this study both to represent the voices of those girls too young to participate in the study and to offer additional information pertaining to the experiences of their daughters who were interviewed. In addition to conveying the concerns of their children, data showed that parents of affected children and adolescents have independent worries of which health care providers should be aware. Although health issues did not overly concern the girls and women living with this condition, parental fears were predominantly health and health-care oriented. Parents of older children and teens currently making decisions regarding HGH and/or HRT disclosed anxieties regarding making a decision now that could come to affect their daughters adversely in the future. Another finding unique to parents was the concern surrounding the financial expense of this condition. Table VI highlights parental concerns pertaining to their daughter's condition.

**Suggestions for Health Care Providers**—During the interviews women were asked about their health care experiences and were encouraged to offer recommendations for HCPs. Based on their positive and negative interactions with a number of different physicians and nurses over the years, the girls and women, as well as their parents, offered useful suggestions for HCPs regarding how to care for other girls and women with TS. Suggestions included to: diagnose TS at an early age, offer HGH and HRT interventions, ensure that all parties involved are educated and up to date on TS, continue to refine physician-patient communication strategies, and improve the sensitivity, compassion, and encouragement invoked during the diagnosis of the condition. Table VII, below, includes a list of these suggestions, with accompanying quotations, as informed by the past experiences of the girls and women in our study. The list is organized in descending order from the most frequently cited recommendations to the least cited.

## DISCUSSION

Interestingly, the age of the participants did not impact the perceived prevalence of the four most frequently cited concerns. Infertility was the primary concern and challenge in each age category followed by short stature, sexual development and function, and general health respectively. Although different challenges affected each individual or age group slightly differently, the major themes remained the same across the lifespan for girls and women with TS. In fact, a number of women reported increased grief due to their infertility as they aged and recounted with trepidation the great number of unknown variables associated with aging, reduced lifespan, and the possible health conditions that may afflict them as they mature, suggesting a need for more comprehensive health care of adult women with TS.

The inability to have biological children remained the central source of pain and hardship for women of all ages diagnosed with TS. Most women could recall their initial reactions of devastation to the infertility diagnosis. Also mentioned was how their diagnosis later interfered with dating, as some women were hesitant to enter into relationships out of fear of

being rejected once their partners learned of their inability to reproduce. These findings are supported by Sylvén and colleagues' research study of 22 middle-aged women with TS, which revealed infertility as the most difficult and distressing consequence of their condition and acknowledged similar sentiments surrounding infertility and intimate relationships [Sylvén et al. 1993]. Given the lifelong sadness associated with infertility, creating a family environment that does not define a woman or a woman's successes by her ability to bear biological children could help girls and women with TS view themselves beyond their reproductive capacities. Parents might encourage their daughters to embrace alternative means to becoming a parent, such as adoption. In addition, parents should remind their daughters that children can remain an important part of their lives in a variety of capacities, even if biological childbearing is not an option. Many participants described how much they enjoyed opportunities to interact with children, either through relationships with nieces and nephews or babysitting. Such experiences often reinforced their desires to parent or otherwise remain involved in the lives of children. Most importantly, how young children and adolescents perceive their condition will inevitably reflect the worries and concerns of their parents. Therefore, parents exhibiting protracted grief with an affected daughter, particularly about her infertility, could lead to deleterious long-term consequences and a delay in the healing and adaptation processes.

Our findings related to the psychological impact of infertility on the lives of participants should be considered in the context of the depression and self-esteem findings of the cohort. Participants' depressed mood state may be associated with their reports of the negative impact of infertility. Indeed the level of decreased self-esteem was consistent with that of other infertile women. This data further suggests the negative impact of infertility, or rather, their negative psychological well-being may be coloring participants' perceptions. Additional research is needed to understand the relationship between mood states and perceptions of the condition in women with Turner syndrome.

Women with Turner syndrome often struggle with their short stature both from a functional and social standpoint. The dependence on other people and objects to reach things irritated a number of the participants on a daily basis. Similarly, for a number of adolescents and adults, being forced to use public transportation, whether due to the general discomfort of large vehicles or their inability to get a driver's license due to spatial perception difficulties, also resonated as lost independence. A number of parents in our study elaborated on their efforts to increase their daughters' independence either by lowering counters, toilets, light switches, and clothing bars or creating a pseudo-apartment within the home for their daughters. Parents and adults with TS might consider implementing such strategies within their homes both to alleviate some of the frustration and increase independence. The Turner Syndrome Society and Little People of America provide resources and web-links, such as [www.short-stature.com](http://www.short-stature.com) and [www.jameron.com](http://www.jameron.com), for organizations that sell devices designed specifically to help short-statured individuals regain their autonomy.

Teasing is arguably the most distressing aspect surrounding height for many of the women with TS. The existing social stigma associated with short-statured people was felt strongly by women of all ages in our study. Short stature served as the main source of ridicule and torment for many participants during their formative years and did not fully disappear by adulthood. Equipped with comebacks, ignoring tactics, or a sense of humor, these girls and women encounter the daily scrutiny of others. However, few have forgotten the cruel and thoughtless nicknames and pranks endured over the course of their lives. Within this context, height emerged more as a social problem than a clinical one, as such challenges extend to most short-statured individuals and are not specific to girls and women with TS [Stace and Danks 1981a; Sawisch 1986; Skuse 1987; Rieser 1992; Skuse et al. 1994;

Sandberg and Voss 2002; Gollust et al. 2003]. The teasing that occurs during one's youth can be extremely damaging to a child's confidence and self-esteem [Gollust et al. 2003].

Since teasing is a real threat, girls and women with TS should know how to handle hurtful and/or unappreciated remarks before such experiences occur. Ignoring strategies were cited by a number of participants as their preferred coping mechanism. Rickert and colleagues argue, however, that adopting ignoring tactics might not be the best response, as this method teaches children, who in many cases already lack assertiveness and social skills [Skuse 1987; Siegel et al. 1998; Ross 2001; Sybert 2001; Elsheikh et al. 2002; Frias and Davenport 2003], to walk away from confrontation and isolate themselves further from their classmates to avoid being teased [Rickert et al. 1996]. The girls in our study who seemed unprovoked by their peers' efforts to hurt their feelings were the ones aware that they might be the target of hurtful words and were equipped with quick comebacks for each situation. Parents, school teachers, and even HCPs might consider teaching these girls how to respond assertively and effectively to bullying and teasing at an early age to ensure preservation of their self-esteem and self-confidence. Hopefully, as critical role models demonstrate respect for and comfort within the company of short girls and women with TS, including treating these individuals in an age appropriate manner irrespective of their size or physical appearance [Rieser 1992; Skuse et al. 1994; Ross 2001], the amount of teasing endured by these girls will diminish.

Overestimating one's height is a phenomenon not unique to the TS population [Giles and Hutchinson 1991; Tienboon et al. 1992; Imrhan et al. 1996; Jacobson and DeBock 2001; Engstrom et al. 2003]. A number of researchers have examined the accuracy of self-reporting height in the general population. In a literature review of 35 studies assessing the self-reporting abilities of women, Engstrom and colleagues concluded that women significantly overestimate their height with frequency [Engstrom et al. 2003]. In a study of male participants who were told in advance that their height would be measured later, the men reported more accurate self-measurements [Imrhan et al. 1996]. Although Tienboon and colleagues' research concluded that an individual's size did not significantly affect their propensity to overestimate their height [Tienboon et al. 1992], Giles and Hutchison found that men under 6 feet tall did overestimate their height more than taller men [Giles and Hutchinson 1991]. Research clearly supports the frequent tendency, by men and women, whether tall or short in stature, to over-report their height.

Concerns with sexual development and functioning are very private issues for most girls and women, and further study, with more directed information gathering, is required to understand fully the sensitive issues touched upon here. A number of adolescents with TS reported shyness and embarrassment concerning their underdeveloped figures. In some instances these views reflected an intentional delay of pubertal induction to allow more time for statural growth on HGH treatment. In other instances, the views may reflect unrealistic expectations or body image issues not very different from feelings experienced by normal girls undergoing puberty. In any case, girls with TS need to be involved in discussions with parents and pediatricians about the timing and pace of pubertal induction, and the teens' expectations and perceptions need to be elicited and understood by all.

A subgroup of adults and mature adults voiced spontaneous concerns about femininity and lack of interest in or enjoyment of sexual activity. This aspect of the adult experience of TS needs further, more focused investigation. Many factors could influence the perception of femininity and attitudes towards sex in women with TS, including, feelings of loss and inadequacy resulting from infertility. In addition, short stature and some other physical attributes of TS make some women feel unattractive and shy away from sexual encounters. Girls and women with TS take estrogen with the initial goal of feminization. Estrogen is



continued during adult years, with the addition of a progestin to protect the uterus, to maintain the feminine body habitus and protect against osteoporosis. The normal ovary produces very large amounts of testosterone, in addition to estrogen and progesterone, but androgen is not usually “replaced” in the hormone replacement regimens used to treat ovarian failure. In fact, women with TS are androgen deficient, reflecting the lack of ovarian function [Hojbjerg et al. 1999]. Since androgens appear to be important for libido and sexual satisfaction in women [Sherwin and Gelfand 1987; Goldstat et al. 2003], this relative androgen deficiency may also contribute to impaired sexuality in women with TS. Health care providers are becoming increasingly aware of the various psychosocial challenges faced by girls and women with TS [Saenger 1996; Rosenfeld 2000; Ross 2001; Sas and de Muinck Keizer-Schrama 2001; Elsheikh et al. 2002; Frias and Davenport 2003]. Parents and HCPs need to remain vigilant in monitoring their daughters and patients respectively, and provide the means through which girls may acquire the necessary skills to enter adulthood as confident, self-assured women.

## CLINICAL IMPLICATIONS

The Turner syndrome-related concerns and health care suggestions voiced by the girls and women in this study both corroborated and extended the findings and recommendations of previously conducted TS research. Although positive experiences with HCPs transpired, reporting the frequently cited negative experiences and accompanying constructive criticism may lead to further enhancement of physician-patient relationships.

The request mentioned most frequently by the girls, women, and their parents, was for an early diagnosis of the condition. The remarkable variability and sometimes subtle physical appearance of individuals with TS often contribute to a delay in diagnosis for many of these girls [Savendahl and Davenport 2000; Sas and de Muinck Keizer-Schrama 2001; Cunniff 2002; Frias and Davenport 2003]. Even if classic TS features are evident at birth, missed diagnoses are common [Savendahl and Davenport 2000]. Approximately seventy-five percent of all girls with TS remain undiagnosed for many years, learning of their condition only when causes for their short stature or pubertal delay are explored [Lippe 1991; Rosenfeld 2000; Savendahl and Davenport 2000; Cunniff 2002; Frias and Davenport 2003]. Ten percent of women with TS, particularly those who experience spontaneous menses, do not receive a diagnosis until adulthood [Elsheikh et al. 2002]. Recognizing this diagnostic delay, multiple clinicians have also called for an early diagnosis of TS, not only for the medical benefits but also for the opportunity to ensure psychological well-being [Saenger 1996; Rosenfeld 2000; Sas and de Muinck Keizer-Schrama 2001; Elsheikh et al. 2002].

Our study results, reinforced by the existing literature, highlight the potential deleterious effects of short stature and delayed sexual development on the adolescent girls’ body image, sense of identity, and social integration with peers [Sylvén et al. 1993; Skuse et al. 1994; Pavlidis et al. 1995; Lagrou et al. 1998; Rosenfeld 2000; Sas and de Muinck Keizer-Schrama 2001; Cunniff 2002; Elsheikh et al. 2002]. Although the physical and psychological benefits of HGH are widely debated, delayed diagnoses deny girls and their parents the opportunity to make decisions about HGH treatment and HRT.

Other recommendations made by participants encouraged a reevaluation of the communication methods used by some of their physicians. Considering the sensitive subject matter surrounding infertility and height, HCPs were encouraged to adopt an attentive, empathic, and more optimistic approach when delivering a diagnosis of TS. A number of women recalled that their health care providers just blurted out that they were infertile and proceeded to list off the unfortunate consequences of such a diagnosis. In some cases physicians were reportedly glib and adopted “humor” to address questions, concerns, and

fears. Neither approach resonated well with these women. Rather than expressing excessively negative and/or potentially inaccurate prognoses, given the highly variable nature of the condition, HCPs might alternatively provide patients and their parents with a thorough and balanced disclosure of the condition, offering positive depictions of life with TS. Physicians who both acknowledged the pain of infertility and what such a diagnosis meant for their patients' futures and positively portrayed adoption and IVF as possible alternative parenting options, were remembered fondly and appreciated for their compassion.

Parents, as well as a number of girls and women, requested that health care providers ensure parents fully understand the cause of TS, as some mothers experience deep feelings of guilt and grief, believing that their behavior while pregnant caused their daughter's condition. Also, given the majority of women in our study were generally receptive to alternative reproductive methods, discussing alternative childbearing options, adoption being the most widely available, might prove extremely beneficial for patients newly diagnosed with TS. As outlined in articles authored by Sybert, Cunniff, and Palvidis and colleagues, such discussions should additionally include an emphasis on the ability for women with TS to sustain long-term relationships, marry, have families, and experience sexual gratification [Saenger 1996; Sybert 2001; Cunniff 2002].

Timing is also essential when communicating a diagnosis and will differ for each patient. Most of the participants in our study, not unlike those in Starke's study [Starke et al. 2002], complained of being "left in the dark" while tests were being done and would have appreciated open communication throughout the entire diagnostic process. However, one girl and her mother were furious at being given worst-case scenarios pertaining to an unconfirmed diagnosis. Similarly, a few of the participants in Starke's study felt overwhelmed with the information provided [Starke et al. 2002]. Consequently, the amount of information desired by patients prior to a confirmed diagnosis should be discussed before the testing process begins [Starke et al. 2002]. If patients opt to hear possible causes for their short stature, pubertal delay, or other presenting features, HCPs might contemplate presenting the information as carefully as if they were disclosing a confirmed diagnosis. Preliminary discussions will inevitably set the tone for how the patients and/or their parents ultimately react in the event of a positive diagnosis. Participants did express their ability to appreciate the tentative nature of unconfirmed or suspected diagnoses.

Physicians might also bear in mind the girl's age and current environment prior to disclosing or withholding information related to the diagnosis. The level of comprehension of each patient and her readiness to hear certain information is also crucial to the disclosure process. Optimally, the information sharing process should, as suggested by participants, proceed in an open and honest, but developmentally appropriate, manner. Conducting regular follow-up sessions to continue the education process with these girls and women as they develop was repeatedly requested by our participants and is an important service not to be neglected. Given that girls may reach puberty at a young age, discussions about sexual development and reproduction might need to occur earlier than in former years.

An important and somewhat neglected issue requiring more focused clinical attention concerns the perceptions and experiences of girls and women with TS regarding sexual development and functioning. Adolescents and adults with TS in this study reported having numerous questions relating to sexuality and sexual performance growing up, yet did not feel they could discuss their concerns with anyone. These women repeatedly emphasized how they appreciated, or would have appreciated, candid responses to their direct and intimate questions. Since many women turn to their HCPs to discuss these personal issues,

HCPs should remain open to and encourage such important discussions and create a comfortable environment within which such conversations can occur.

Few physicians are experts on Turner syndrome despite the condition being relatively well known. Consequently, some adult women seeking care may feel lost and frustrated. As more research is published and as adult endocrinologists, gynecologists, and internists become more knowledgeable about the condition, questions such as, “when should I stop taking HRT?” and “how long can I expect to live?” will become easier to answer. In the meantime, physicians who expressed a willingness to research and learn about Turner syndrome were genuinely appreciated by the girls and women in this study and defined, in most cases, their most positive experiences with health care providers.

## STUDY LIMITATIONS

A number of study limitations should be considered. Although the sample size is large for a study of this type, our findings are not generalizable to all girls and women with TS. This group was highly educated compared to the US population, and those with lesser educational attainment may have different attitudes and concerns. Also, women with TS who did not participate in our study may have endured different challenges and/or had different responses to the four participant-articulated concerns of living with TS outlined throughout this paper. Given the wide age range of our participants, the interview process offered much opportunity for the women to reflect on past experiences, thereby rendering the data susceptible to recall bias. Similarly, the experiences relayed by these girls and women about their interactions with health care providers are from their personal perspective and providers themselves might have perceived the interactions differently [Gooding et al. 2002; Manber et al. 2003; Ryan et al. 2003]. However, how individuals perceive events and interactions with their providers remains important, as such assessments inform how they internalize their condition and illuminate possible areas of disconnect occurring during physician-patient communications that could be easily remedied. Finally, given the participants were primarily Caucasian, our results are limited to a specific ethnic group as well.

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**TABLE I**

## Characteristics of Participants with Turner Syndrome

		Number of Participants
<b>Age</b>	7–13	11
	14–19	18
	20–39	39
	40–59	29
	Note: 4/11 girls aged 7–13 were represented by their parents	
<b>Race</b>	Caucasian	93
	Minority	4
<b>Age at Diagnosis</b>	Birth	15
	1–10	25
	11–20	49
	21+	9
<b>Average Height (Age 18–59: n=74)</b>		145.99cm
<b>Percent Of Adults Who Took Growth Hormone</b>		26%
<b>Cardiac Status</b>	Coarctation	3
	Valve problems	6
	Other complications	16
<b>Education</b>	Elementary/Middle School	20
	High School	20
	Undergraduate	36
	Graduate	21
<b>Karyotype</b>	45X	44
	45X, 46,X,i(X)q10	10
	46X, del(X)	7
	45X, 46XX	7
	46X, i(Xq)	4
	45X, 46Xr(X)	5
	45X, 47XXX	3
	45X, 46X, indic(X)	3
	45X, 46X, del(X)	3
	Other	11
<b>Psychiatric Diagnosis*</b>	Depression	6/77
<b>Self-Esteem Evaluation*</b>	Mean	31.145
	Standard Deviation	5.243
	Range of Scores Obtained	18–40

		Number of Participants
<b>Marital Status</b> (Ages 20–59: n=68)	Married	32%
	Divorced	15%
<b>Women with Children</b>	Adopted Children	5
	Successful IVF	1

\* Psychiatric diagnosis evaluated according to the Structural Clinical Interview for the Diagnostic and Statistical Manual for Mental Disorders IV (SCID-DSM IV)

\* Self-esteem diagnosis was evaluated using the Rosenberg self-esteem scale.

TABLE II

## Concerns About Infertility

Infertility	
<b>Childhood</b> (Ages 7–13) n=7+8 parents	<ul style="list-style-type: none"> <li>Half of the parents relayed their daughters' current difficulties in accepting infertility. "I've told her that she can't have kids, but she won't accept that. 'When I grow up I'm going to have a boy and a girl.'" (Parent)</li> <li>A number of adolescents, adults, and mature adults diagnosed between the age of 7 and 13 mentioned that they were devastated upon learning of their infertility.</li> </ul>
<b>Adolescence</b> (Ages 14–19) n=18+10 parents	<ul style="list-style-type: none"> <li>The majority of adolescents who cited infertility as a concern mentioned being extremely upset and disappointed at the time of diagnosis, while a handful perceived childbearing as a future concern and/or anticipated increased grief as they aged. <i>"I think the hardest thing to deal with, as always being a person who loves kids, was the infertility issues ... At the time I was 12, yeah, I was told that I wouldn't be able to have kids. But I don't think it hit me then. I think it came on me more gradually as I realized, oh my God, I'm getting older, I have a serious boyfriend since I was diagnosed."</i> (Age 19)</li> <li>11/18 teenagers in the study viewed adoption and in vitro fertilization (IVF) as positive alternatives to deal with infertility. <i>"...[I]t kind of still bugs me that I can't have kids, but I figured out that I can adopt and so that doesn't bug me anymore that much."</i> (Age 19)</li> </ul>
<b>Adulthood</b> (Ages 20–39) n=39	<ul style="list-style-type: none"> <li>Infertility posed the biggest challenge for this age group, especially once family and friends started having babies. <i>"...[E]very once in a while, you know, when you're holding a kid and they're snuggling up to your neck, I really thought, you know, I wish I could have kids. I wish I had the choice."</i> (Age 31)</li> <li>A couple of adults made the distinction between infertility as solely a personal difficulty versus an obstacle that must be overcome by their partners. <i>"[A]ll of a sudden, this person I wanted to spend my life with was going to have to go without seeing a baby that was like him. And that was just devastating. I mean it was very, very, very, devastating."</i> (Age 24)</li> <li>27/39 adults viewed adoption and IVF as viable alternatives, yet their inability to have a biological child remained a source of sadness.</li> </ul>
<b>Mature Adulthood</b> (Ages 40–59) n=29	<ul style="list-style-type: none"> <li>The majority of women in this age group remained deeply affected by their infertility, feeling they missed out on an important part of being a woman. <i>"[Women]...get flowers and they get honored because they're a mother, but I don't --you know, I feel, does that make them a complete woman because they were able to have children? And I couldn't, so I must be incomplete."</i> (Age 40)</li> <li>A minority of mature adults claimed infertility was not a concern since they were either not married, did not want the responsibility of children, or did not feel childbearing was ever a guarantee.</li> </ul>



TABLE III

## Concerns Surrounding Short Stature

Stature	
<b>Childhood</b> (Ages 7–13) n=7+8 parents	<ul style="list-style-type: none"> <li>4/8 parents noted that their child was teased at school due to short stature and a physical appearance that differed from their peers. “[S]ome of the boys from the classroom, they [say], you are short, you are fat, you are not as normal as we are.” (Parent quoting 12- year-old daughter)</li> <li>More than half of the women reflecting on their past recalled cruel teasing during elementary and/or middle school.</li> </ul>
<b>Adolescence</b> (Ages 14–19) n=18+10 parents	<ul style="list-style-type: none"> <li>7/18 adolescents cited short stature among their largest concerns. “<i>Trying to reach things and always having to ask people. That can get really boring and tiring. Also, when it comes to, like... dating and stuff. It might hinder it a little bit.</i>” (Age 15)</li> <li>6/18 teens mentioned they currently endure teasing. Most either ignored the jibes or fought back, described the banter as playful teasing or maintained they simply do not care what is said. “I personally don’t have a problem with people’s comments on my height, but some people would. But I don’t have a problem with that because I know I’m short for my age and I don’t really mind it.” (Age 14)</li> <li>13/44 women recalled being teased during all or parts of high school and college.</li> </ul>
<b>Adulthood</b> (Ages 20–39) n=39	<ul style="list-style-type: none"> <li>9 adult women addressed stature as a primary concern and greater obstacle than infertility, citing reasons which included: the fear that men would perceive them differently because they were short or a personal desire to achieve a balanced weight. “I think the hardest thing is probably the height thing, because, you know, you always get a little self-conscious, you know. Is a man going to like me because I’m shorter?” (Age 25)</li> <li>Adult women perceived height either as a functional difficulty or source of social stigmatization. “<i>As an adult, I guess being short does interfere with doing certain things, like even just little things around the house...I don’t think as an adult its really interfered with my relating to people a lot.</i>” (Age 35)</li> </ul>
<b>Mature Adulthood</b> (Ages 40–59) n=29	<ul style="list-style-type: none"> <li>Short stature also concerned the mature adults, as they claimed their height challenged them daily. “I’m really terrified of being in big crowds because I’m scared I’m going to be trampled.” (Age 42)</li> <li>Ongoing frustrations at rude comments made by children, adults, and even health care providers were also relayed. “[A doctor at work] says to me, you know where you came from? I don’t know if he was asking me to respond, what he was doing. Finally, after silence, I said, yeah, my mother. He said, no, you came from munchkin land.” (Age 56)</li> </ul>

TABLE IV

## Concerns About Sexual Development And Function

Sexual Development and Function	
<b>Childhood</b> (Ages 7–13) n=7+8 parents	<ul style="list-style-type: none"> <li>A small number of parents anticipated sexual development as a future concern.</li> <li>One 4th grader had already begun to think about and discuss figure development, as many of her classmates had already undergone puberty.</li> </ul>
<b>Adolescence</b> (Ages 14–19) n=18+10 parents	<ul style="list-style-type: none"> <li>Sexual development was a concern during adolescence.</li> <li>Some girls felt different/“not normal” because they had not gone through puberty. <i>“Sometimes...you feel like you’re a little girl...because I’m short and, you know, the development, I mean, you feel like you’re not normal to some degree.” (Age 18).</i></li> <li>A minority of adults and mature adults reflecting on their past expressed feeling less feminine due to their lack of sexual maturity.</li> </ul>
<b>Adulthood</b> (Ages 20–39) n=39	<ul style="list-style-type: none"> <li>Sexual development did not seem to concern adult participants beyond their adolescent experiences.</li> <li>Women concerned about dating worried that their height or their infertility would cause men to reject them, not their underdeveloped figure.</li> </ul>
<b>Mature Adulthood</b> (Ages 40–59) n=29	<ul style="list-style-type: none"> <li>The few comments regarding sexuality beyond adolescent memories addressed both psychological and physical difficulties with sex. <i>“... he doesn’t understand where I come from as far as, you know, why I don’t think sex is important to me, or I don’t put that priority on my list, saying that I have to do it all the time. To me, it’s not a sign of love. That’s because--well, maybe if I was able to have children, I’d probably feel totally different, because there’d be a reason for having it, because I love that man and I want his children.” (Age 40)</i></li> </ul>

**TABLE V**

## Concerns Regarding Health and Aging with Turner Syndrome

<b>Health</b>	
<b>Childhood</b> (Ages 7–13) n=7+8 parents	<ul style="list-style-type: none"> <li>Children did not mention health as a concern.</li> </ul>
<b>Adolescence</b> (Ages 14–19) n=18+10 parents	<ul style="list-style-type: none"> <li>A small number of adolescents were concerned about their health.</li> </ul>
<b>Adulthood</b> (Ages 20–39) n=39	<ul style="list-style-type: none"> <li>In this age group health concerns were more prevalent. Osteoporosis, hearing loss, and uncertainty about future health were mentioned specifically.</li> </ul>
<b>Mature Adulthood</b> (Ages 40–59) n=29	<ul style="list-style-type: none"> <li>Given the number of health issues that can affect women with Turner syndrome throughout their lives, mature adults expressed a growing concern surrounding the uncertainty of their future lifespan and the lack of available information on aging with Turner syndrome.  “We’re the only female population that, for a long time, it was recommended that you start [hormone replacement therapy] at 12 and end it at, gee, gal, we don’t know, you can do it until you go six feet under if you want.” (Age 48)</li> </ul>

**TABLE VI**

## Parental Concerns Pertaining To Their Daughter's Diagnosis

Parents n=18	
<b>Infertility</b>	<ul style="list-style-type: none"> <li>A few mothers recalled feelings of sadness for their daughters when they learned of the infertility diagnosis. "I realized I hurt for her, because I felt she couldn't have children." (Parent)</li> <li>A couple of mothers related their initial disappointment. <i>"I also had to grieve my grandchildren. You know, because I had thought I was going...to have a house full of grandchildren."</i> (Parent)</li> </ul>
<b>Height</b>	<ul style="list-style-type: none"> <li>A number of parents noted that their daughters' desire to be taller would not be realized, even without Turner syndrome, because their families were short. <i>"[Our family's not] a couple of giants. You know?...your sister is 4'10". I'm 4'11"...So when she wasn't really that tall it was like, well, look at your aunt. You ain't going anywhere."</i> (Parent)</li> </ul>
<b>Sexual Development and Functioning</b>	<ul style="list-style-type: none"> <li>One mother questioned whether her daughter would have desire. "I worried initially about intimacy, what her parts were, would she have desire." (Parent)</li> <li>Some parents relayed their daughters' sentiments, particularly their efforts to understand their bodies.</li> </ul>
<b>Health</b>	<ul style="list-style-type: none"> <li>The majority of parents expressed compromised health as their greatest concern.</li> <li>A number of parents feared potential negative side effects of human growth hormone and hormone replacement therapy. "I'm always afraid of what it can--something that we give to her to cure one thing, what would that be as far as to deteriorate another."</li> <li>A handful of parents cited the huge expense of doctor visits, medical treatments and insurance coverage as an ongoing source of concern.</li> </ul>

**Table VII**

## Participants' Suggestions for Health Care Providers Caring for Girls and Women with Turner Syndrome

<b>Suggestions</b>	<b>Quotations</b>
<b>Strive to diagnose early</b>	<i>"I mean, I guess it's good to know right offhand because then it's not a big shock and you can, you know, not necessarily totally get over it, but deal with it and become comfortable with it and, you know, not fret over it." (Age 20)</i>
<b>Improve both understanding of Turner syndrome and how to care for women diagnosed with this condition</b>	<i>"I think the main thing is, keep yourself informed, especially if you have a patient with Turner syndrome. You know, get informed. I think we need to have a lot more awareness. Like I said, once you get my age, there's nothing as far as medical care for Turner syndrome." (Age 45)</i>
<b>Keep patients and their parents up to date with information about Turner syndrome</b>	<i>"But I would definitely do the research for the person and give them a lot more information, whatever is out there, as much as possible. Give them any options that are there, have resources there available where basically I did—you know, I found the resources, I did my own research. But as a physician, I think you should be able to get that for that person. I mean, I know Turner syndrome is not that common, but it's also not that rare." (Age 33)</i>
<b>Be more attentive, sensitive, positive, and encouraging</b>	<i>"To listen. Listen thoroughly before they try to give an answer, you know. Listen to what that girl with Turner's, or that woman with Turner's is saying, and then answer and talk about it. Don't jump to an answer right away, like my doctors did, you know. Hear them out. If an examination is needed, then do an examination, you know." (Age 47)</i>
<b>Engage in open and honest communication with patients and their parents</b>	<i>"I wish they would have just told me, this is why we're testing your blood. This could be the case or this could be the case. This is what we're doing...So that people get a little time to let it sink in before they actually get the results." (Age 32)</i>
<b>Offer girls diagnosed with Turner syndrome human growth hormone and hormone replacement therapy at the appropriate times</b>	<i>"But I think, for those that they can get growth hormone to help stay somewhat an average height with their peers, along with taking the hormones so they develop at a similar rate, that can help a lot with not feeling quite as different growing up." (Age 37)</i>
<b>Educate patients in a developmentally appropriate manner</b>	<i>"Well, when I first—I think knowing about Turner's was like the first thing I remember being told, you know, that's why you're going to be shorter than other kids. Then sort of like as I grew in my ability to understand and my parents told me more about it." (Age 39)</i>
<b>Put girls and women diagnosed with TS in contact with the Turner Syndrome Society</b>	<i>"So I went over to the clinic that I had been as a kid and gave the doctors both notes with my name and phone number, and said, we have this society. If you have any patients, feel free to refer them, kind of thing." (Age 37)</i>
<b>Offer Counseling</b>	<i>"...[T]he one thing they offered that was real helpful was a lot of literature about it. They offered, like, counseling for Turner Syndrome kids, support groups, you know." (Age 27)</i>
<b>Involve the parents</b>	<i>"Make sure the parents are armed with enough information to help the child." (Age 19)</i>