

TRUTH-TELLING AND TURNER SYNDROME: THE IMPORTANCE OF DIAGNOSTIC DISCLOSURE

ERICA J. SUTTON, MA, JESSICA YOUNG, AIDEEN McINERNEY-LEO, MSc, CAROLYN A. BONDY, MD, SARAH E. GOLLUST, BA, AND BARBARA B. BIESECKER, MSc

Objective A targeted analysis with transcript data from previous research was designed to study the perceived effects of secret-keeping on individuals with Turner syndrome (TS).

Study design Girls and women (n = 97) and 21 parents participated in the initial interview study. Transcripts were coded and analyzed for constructs related to secret-keeping.

Results Thirty percent of participants spontaneously mentioned that their health care providers (HCP) or parents had withheld all or part of their TS diagnosis. Of those, 15 individuals were not informed of the infertility component of their diagnosis. Individuals reporting secret-keeping were more likely to have had a negative perception of the HCP's role in the disclosure process compared with those participants who did not report that a secret had been kept ($P < .025$).

Conclusion The prevalence of secret-keeping within this sample population suggests it is likely an existing concern in the greater TS population. How HCPs disclose a TS diagnosis may affect whether secrets are kept. Conversely, secret-keeping may result in a negative disclosure experience. These observations suggest the need for interventions aimed at helping HCPs disclose health-related information to parents and their children in a timely, caring, and sensitive manner. (*J Pediatr* 2006;148:102-7)

Turner syndrome (TS) is a sex chromosome anomaly that occurs in 1/2500 live female births. This condition is most often caused by the loss of 1 sex chromosome, resulting in a 45,X karyotype. Typically, women with TS are infertile and short in stature. The time of diagnosis varies from the second trimester of pregnancy to adulthood, although most girls are diagnosed with the condition by their teenage years.¹ Before the 1970s, health care providers (HCPs) assumed a more paternalistic model of health care.²⁻⁴ Some HCPs believed that disclosing diagnoses might cause their patients, especially children, greater harm than good. Consequently, professional appeals to the "do no harm" principle resulted in nondisclosure or only partial disclosure of health-related information.^{2,3} HCPs believed they should protect pediatric patients from their diagnoses and prognoses, maintaining that young children could not comprehend concepts such as death, serious illness, or medical treatment.^{4,5} A paradigm shift in medicine toward greater patient autonomy and shared decision-making led to a more open disclosure policy with children in the 1970s to 1980s.^{2,4,5} Studies indicated that young children can achieve basic understanding of complex medical phenomena.^{2,4,5}

Graveholt et al⁶ conducted a study that assessed the characteristics and risk factors of bone fractures in women with TS. These researchers unexpectedly found that 45/322 participants were unaware of their TS diagnosis.⁶ However, few studies have explored the disclosure process as experienced by girls and women with TS and their parents or the effects of secret-keeping on the girls and women with this condition. The initial objective of this study was to ascertain the social, psychological, and medical concerns and challenges experienced by girls and women affected with TS.⁷ The analysis revealed that a number of participants spontaneously stated that some aspect of the TS diagnosis had been kept secret from them. Noting the unexpected frequency of such remarks, we conducted a second

See related article, p 95.

From the National Human Genome Research Institute and the National Institute of Child Health and Human Development, National Institutes of Health, Bethesda, Md.

Supported by the NHGRI Intramural Research Program and the NICHD Intramural Research Program.

Submitted for publication Jan 5, 2005; last revision received May 19, 2005; accepted Aug 1, 2005.

Reprint requests: Barbara Biesecker, MSc, NHGRI/NIH, 10 Center Dr (10/10C101), Bethesda, MD 20892-1852. E-mail: barbarab@mail.nih.gov. 0022-3476/\$ - see front matter

Copyright © 2006 Elsevier Inc. All rights reserved.

10.1016/j.jpeds.2005.08.022

HCP	Health care provider	TS	Turner syndrome
-----	----------------------	----	-----------------

analysis to describe the perceived effects of both secret-keeping on girls and women with TS and diagnostic disclosure on family communication.

METHODS

This qualitative research study was conducted in conjunction with an ongoing IRB-approved study at the National Institute of Child Health and Development entitled, "Turner Syndrome: Genotype-Phenotype." Individuals who were pregnant, had androgen treatments in the past 6 months, had coexisting autosomal chromosomal conditions, or had severe physical or mental challenges precluding study cooperation were excluded from both studies. Between January 2001 and May 2003, 97 girls and women with TS and 21 parents were invited to participate in this interview study. All 107 individuals approached provided their consent. Because of the young age of a number of the girls, 4 parents were interviewed on behalf of their young daughters. Seven parents were interviewed conjointly with their daughters, and 10 parents of older adolescents were interviewed separately.

Two interviewers (AM and BB) conducted most of the semistructured, in-person interviews, with fewer than 10 interviews conducted by 4 additional interviewers with AM or BB present. The interview guide included open-ended questions on the following topics: the diagnosis narrative, greatest concerns and challenges associated with TS, sources of information and support, suggestions for HCPs on how to improve the health care experiences of girls and women diagnosed with TS, and advice for newly diagnosed girls and their parents about how to address a TS diagnosis. Interviews ranged from 60 to 90 minutes in length. All interviews were audio taped and transcribed verbatim.

Data Analysis

Transcripts were coded using standard qualitative research methods.⁸ A numeric codebook was created on the basis of the re-reading of 9 transcripts where secret-keeping was described and on particular factors related to secret-keeping and family health communication. All transcripts were double-coded by 2 independent coders. One coder (ES) applied the initial codes. A second coder (JY) re-read the transcripts in their entirety and reviewed the initial codes to ensure their consistent application. Disagreements in the coding were addressed, discussed, and reconciled by the 2 coders. One coder (JY) entered the codes into NUD*IST, a qualitative research database. NUD*IST output was analyzed: themes were identified, frequencies were ascertained, and quotations capturing those themes were selected.

RESULTS

Sample Demographics

The 97 girls and women ranged from 7 to 59 years of age. The average age at diagnosis was 12.8 years, and the average year of diagnosis was 1979. The average adult height was 4'9", and 26% had taken human growth hormone. The

most common karyotype was 45,X. Eighty-four percent of participants between the ages of 20 and 59 had an undergraduate or graduate degree. Of the adults 18 years of age and older, 32% were married and 15% were divorced. Six women had children, 5 through adoption and 1 through in vitro fertilization.

Themes

Secret-keeping (defined as all or part of the TS diagnosis deliberately withheld from the participant), difficulty communicating an infertility diagnosis, and perceived negative experiences with HCPs at the time of diagnosis were the major themes resulting from this targeted analysis.

Secret-keeping and Infertility

During interview sessions broadly addressing concerns about living with TS, 30/97 participants spontaneously mentioned that all or part of their TS diagnosis had been withheld from them. Parents were the individuals most likely to keep the diagnosis from their daughters (13/30). In 6 instances HCPs withheld diagnostic information from their patients with TS. In 6 other cases parents and HCPs were allied in the decision to withhold diagnostic information from the participants. In the remaining 5 cases where interviewees stated important health-related information was withheld, the secret-keeper was unidentifiable from our data.

The types of secrets kept varied among participants. Fifteen girls and women from whom a secret had been kept knew every aspect of their diagnosis except the infertility component. Eleven girls and women were completely unaware they had a chromosomal condition, and 3 knew the characteristics associated with TS but did not know the clinical term for their condition. One woman was not told about the potential difficulties with sexual intercourse, despite having asked her doctor specific questions related to sexuality. These girls and women remained unaware of the full implications of their diagnosis for time periods ranging from a few days to 21 years. Although the secrets kept for protracted lengths of time are of greatest concern, that participants voluntarily mentioned having had their diagnosis withheld for only a few days suggests the lasting impact of what might otherwise be interpreted as initial parental adjustment to difficult information. Reports of secret-keeping were as prevalent among younger participants as they were among the older participants. Three parents interviewed on behalf of their daughters, ages 7, 10, and 13, revealed they had not yet disclosed the infertility diagnosis to their daughters.

Participants who had their diagnosis concealed from them reported learning of their condition in an undesirable manner. Most of the girls and women eventually learned of the secret through an HCP who inadvertently told them, usually in a blunt and matter-of-fact way, having assumed they already knew:

"The doctor finished his exam and I got dressed and went in. And he just kind of stopped, put his pen down and went, 'you know you're not having kids ever, don't you?' I went 'Excuse

me? No, I don't know that.' So that was the hardest time." (Diagnosed at 17, learned of infertility at 19.)

Some girls and women discovered their TS diagnosis by overhearing conversations, listening to diagnoses left on answering machines, confronting their parents, reading about their condition in school, or conducting their own research.

A number of participants reported that learning of their diagnosis unexpectedly or from strangers was traumatic. For a few individuals the discovery experience resulted in long-term consequences:

[When I found out,] I was really shaken up. And I don't even think I really got the opportunity to really emotionally cure that or get through that because then it went into this big denial thing. Of course [my parents] didn't feel good about lying, and all that kind of got put on to me, so then it wasn't real fun. So basically what happened was it really didn't get talked about. (Age 41)

Seventeen women spontaneously commented on their reactions to discovering a secret about their health had been kept from them. Twelve related feeling angry or upset:

"I did get upset with my mother [when I found out].... She... didn't explain. She said, 'Well, what's the big deal?' I said, 'I think it would have helped me to understand myself a little bit better. To tell—to fill in the gaps to tell you more of the story.'" (Diagnosed at 17, learned infertile at 28)

A handful of girls and women felt their family members had betrayed them:

There I am at this sterile place. And I found out that since the age of 9 to 16, everyone knew something I had no knowledge of about me that was pretty darn important. I was told [I was infertile] in such a horrendous way, "of course you know this." And all I knew was that the people that I trusted forever were willing to betray me. (Diagnosed at 9, learned infertile at 16)

For some women this realization resulted in a loss of familial trust.

Decisions to keep secrets about health-related conditions are complex and vary among families. Although we do not fully understand why parents or HCPs kept secrets about the TS diagnosis, our data from the parents' interviews suggest some justification from the perspective of the secret-keepers. Because most of the interviews were with girls and women with TS and not their parents, we captured primarily their perceptions of why their parents kept the diagnosis a secret. Parents in similar circumstances, not part of this study, might offer alternative explanations about why secret-keeping within this population occurs.

The main reason reported by parents in this study about why they kept the diagnosis a secret was because they felt ill-equipped to disclose the diagnosis to their daughters. Parents believed their HCPs did not provide them with enough information to answer their own questions about TS, much less their daughters' questions:

"And a couple of days later, we still hadn't told her because we weren't sure, like what do you say? What does this mean? We don't even know ourselves." (Parents of child diagnosed at 10)

A number of parents reported wanting to protect their child from the heartbreak of infertility, recognizing their daughters' desires to experience biological motherhood:

"So we haven't been able to share with her [the fact that she can't have children], because she's adamant on having children. The most important thing in her world is to have kids." (Parents of a 10-year-old, as of interview infertility remained a secret)

A few parents, according to the girls and women interviewed, had to accept and adjust to the diagnosis first before they could disclose the diagnosis to their daughters:

"[My mother told me] that she would have to deal with it. She said, 'I had to deal with not having the perfect child anymore.'" (Diagnosed at 16, learned diagnosis 3 weeks later)

Parents were also aware of the social stigma surrounding infertility:

"So there's a lot of difference between conceiving a child that's not really yours to—I mean, to be implanted with one in order to conceive is different." (Mother of a 10-year-old who remained unaware of her infertility at the time of interview)

In spite of some parents' efforts to shield their daughters from stigmatization, a number of women recalled enduring inappropriate comments about their infertility from parents, extended family, and friends; consequently, they believed themselves to be incomplete and inferior women.

To ascertain whether family communication contributed to secret-keeping, we compared statements made by participants who spontaneously mentioned a secret was kept and those who did not. Participants whose families or HCPs kept a secret indicated that communication about the TS diagnosis was absent in their homes more frequently than participants who did not state that a secret was kept. However, of the 30 girls and women who reported secret-keeping, 14 explicitly reported comfort with discussing TS with at least 1 member of their family once the diagnosis was disclosed. Although 10 reported a continuing reluctance to address TS, only 5 stated unequivocally that overall communication within the home was problematic. Six participants were not explicit regarding the communication dynamic at home.

The Disclosure Process

Participants frequently mentioned dissatisfaction with the manner in which HCPs disclosed the TS diagnosis to them or their parents. Of the 97 participants, 47 (of whom 21 had a secret kept and 26 made no mention of a secret) stated explicitly that they perceived the disclosure process negatively. Specifically, parents recalled that the HCPs did not provide enough information, displayed poor "bedside manner," lacked empathy, or did not prove supportive during this difficult time:

"I think the OBGYN that they sent me was even worse. He was really bad because the first thing he did—I think he did his whole exam. He thanked me for letting him see me, because I was a very interesting case and that they were sure—he was sure that I was a female...It took me years to recover from that." (Aged 37)

However, 10/97 participants (1 of whom had a secret kept and 9 who made no mention of a secret) reported perceiving the communication of the diagnosis positively. Specifically, their HCP was informative, compassionate, and sensitive to the difficulty associated with infertility.

A χ^2 analysis of the disclosure process suggests that women who had a secret kept were significantly more likely to perceive the manner in which HCPs disclosed the diagnosis negatively than women who did not mention that a secret was kept (Table).

Participants were also asked to give advice to parents of girls newly diagnosed with TS. Many girls and women, particularly those who mentioned that a secret was kept from them, expressed the importance of truth-telling and open communication within the family:

I know how much you truly want to protect your child. If you never had to tell them something, you would never tell them. But know that everyday you don't there are people out there in the world who may at some point, for any reason, say something that you will have no control over, because you don't...Eventually this person will know. (Age 41: Infertility kept secret for 7 years)

Participants explained that girls with TS are aware that something is not quite right physiologically, whether their awareness stems from multiple trips to the doctor or their failure to undergo the typical adolescent growth spurt or pubertal development:

"Don't lie to them. I knew I had had the test, so it's not like they'd just taken blood and I didn't know what was going on." (Age 25: secret kept for 3 weeks)

Participants also advocated for diagnostic disclosure at an age-appropriate time:

And don't keep things—you know, don't keep secrets. I mean, you don't have to tell a five year old she's not going to be able to have kids, but as subjects come up and as she's asking about things, be honest about it. You know, it's going to be a disappointment at whatever point that she finds out and you can't protect her from it. (Age 41: secret kept for 3 years)

DISCUSSION

Despite existing literature encouraging early and full disclosure of diagnoses to children with disabilities and illnesses and their parents,^{4,9-13} children are often uninformed about their health conditions.⁹ The study of Graveholt et al,^{14,15} coupled with the frequency with which our sample population spontaneously revealed that a secret had been kept, suggests that secret-keeping within the TS population occurs surprisingly often. The drawbacks of secret-keeping are many and can lead to patient depression, isolation, fear, and general mistrust of HCPs.^{14,15} In addition to denying girls and women with TS the opportunity to benefit from the advantages associated with an early and comprehensive disclosure,^{4,11-13} secret-keeping renders girls and women vulnerable to learning of their diagnosis in potentially traumatic, damaging ways. Furthermore, nondisclosure may delay the

Table. Perceived communication with HCP at the time of diagnosis (n = 97)

	Negative	Positive	Unclear	χ^2	D.F	P value
Secret kept	21	1	8	8.45	2	<.025
No mention of secret kept	26	9	32			

process of self-education toward becoming an advocate for one's own health care.

Disclosing the infertility component of TS was especially challenging for parents. One might argue that infertility is the hardest characteristic to disclose. In addition to parents perceiving themselves to lack sufficient knowledge to engage in such conversations with their daughters, infertility comes with the added burden of existing social stigma, their daughters' desires to become biological mothers, and the parental loss of biological grandchildren. The social emphasis placed on childbearing contributes to society's perception that infertility is an abnormal condition, counter to societal expectations and norms.¹⁶⁻¹⁸ Although some parents withheld infertility diagnoses from their daughters to protect them and delay the ensuing pain and stigmatization, the nature of an infertility diagnosis demands early and full disclosure.

An infertility diagnosis often sends women into identity crises.^{16,17} For many women, learning of their infertility generates a variety of emotions,^{17,19} as they are faced with the challenge of reinventing themselves to incorporate infertility into their changing sense of self.¹⁹ Disclosing the diagnosis at a young age enables girls to incorporate TS and its consequences for reproduction into their self-identity early in their development. By not telling young girls about their reproductive limitations, parents allow their daughters to create self-images that include biological motherhood. Slijper et al¹³ found that adults with androgen insensitivity syndrome had difficulty integrating correct health information into their self-image after a lifetime of being told inaccurate or false information. Similarly, girls with TS left ignorant of their infertility might have a harder time adapting to and coping with this significant ramification of their condition. Furthermore, girls and women with TS are receptive to adoption and in vitro fertilization even though the pain of lost reproductive capacity can persist throughout their life.⁷ Disclosing infertility early and engaging in open discussions enables parents to foster acceptance of alternative parenting means for their daughters, while simultaneously destigmatizing TS and, by extension, infertility.

A number of studies have explored parental reactions to and satisfaction with the disclosure process of their children's medical conditions.^{9-12,20,21} Scholars disagree about whether parents can distinguish their emotional reactions to the diagnosis and the manner in which the diagnosis was disclosed.^{9,11,12,22} That 10 participants fondly recalled the role of their HCP at the time of their diagnosis suggests satisfaction

with the disclosure process itself is attainable. The bulk of our data, however, corroborate the findings of Krahn et al⁹ and Hill et al,¹¹ who reported overall parental dissatisfaction with the process of disclosure, emphasizing the paucity or incomprehensibility of information offered by the HCP, the absence of empathy and support from the health care team, and the physician's unappealing bedside manner. Our analysis of the association between negative perceptions of the disclosure process and secret-keeping suggests that how a diagnosis is delivered may affect whether a secret is kept and that secret-keeping may ultimately lead to negative disclosure experiences. Medical students, residents, nurses, and physicians might benefit from additional coursework or training sessions that focus on the importance of HCP-patient communication with an emphasis on how to disclose medical diagnoses in a sensitive and compassionate manner.

Our study data support the following recommendations for HCPs caring for patients with TS. Parents are sensitive to the demeanor of the health care team, sense when their child is not well, and recognize the importance of the timing, location, and substance of the disclosure process.^{4,9,11,12} However, not all parents want the same amount of information. At the time of diagnostic testing, HCPs should ask the patient and her parents how much information they would like to receive throughout the process before obtaining a confirmed diagnosis.⁷

HCPs should deliver diagnostic information in person and anticipate that patients and their parents will have numerous questions once they learn of their condition. HCPs should schedule the disclosure appointment for a time when they can spend enough time with the patient to explain the diagnosis properly and fully answer the patient's questions.

When a diagnosis is first delivered, patients and their parents may have difficulty understanding and retaining important information pertaining to the condition.^{10,12,21} Some researchers suggest arranging 2 separate consultations: one for the disclosure of the diagnosis and the second to relate additional information about the condition.^{11,15} To ensure that patients and their parents are sufficiently informed about TS, at the time of the disclosure HCPs might consider providing a written, easy-to-understand explanation of what TS is, basic implications with respect to the health of an individual with TS, general standards of care for girls with TS, guidelines for parents on how to disclose a diagnosis, and contact information for the Turner Syndrome Society.

HCPs and parents should be allied in their commitment to disclosing the TS diagnosis to young girls in an environment best suited for the patient and her family. Parents of children with disabilities and illnesses are typically the individuals responsible for disclosing diagnoses to their child.^{5,10,13} However, for parents to feel comfortable with this important task, they must have a solid grasp of the condition's cause, associated characteristics, and future health outcomes. HCPs should emphasize to parents that they are accessible for questions and willing to facilitate the disclosure process. How test results will be relayed to pediatric patients should be discussed with the parents before testing is initiated.

HCPs should convey to parents the importance of early disclosure and the benefit of open family communication and truth-telling within the home. Parents contemplating withholding the TS diagnosis from their daughters should be informed that children are perceptive to parental and physician affect and tend to sense when something is not right with their health.^{5,15,21} If parents expressly state they plan to delay disclosing the diagnosis to their daughters, HCPs should emphasize that an early and full disclosure is important for their child's psychosocial development and future quality of life.

The age of disclosure will vary for each patient depending on the cognitive development and severity of the child's condition. A developmental approach to disclosure should be adopted. When a girl with TS asks questions about issues concerning her health, her appearance, or issues related to TS, parents and HCPs should answer them honestly and completely. The amount of detail provided should be gauged according to the age of the girl; however, all parties should remember that young children can comprehend seemingly complex concepts related to health and development.

Special thanks to all of the girls and women with TS and their parents who participated in this study and shared their life experiences with us.

REFERENCES

1. Savendahl L, Davenport M. Delayed diagnoses of Turner's syndrome: proposed guidelines for change. *J Pediatr* 2000;137:455-9.
2. Tse C, Chong A, Fok S. Breaking bad news: a Chinese perspective. *Palliat Med* 2003;17:339-43.
3. Beauchamp T, Childress J. Principles of biomedical ethics. 5 ed. Oxford: Oxford University Press; 2001.
4. Lipson M. Disclosure of diagnosis to children with human immunodeficiency virus or acquired immunodeficiency syndrome. *J Dev Behav Pediatr* 1994;15:S61-5.
5. Purssell E. Telling children about their impending death. *Br J Nurs* 1994;3:119-20.
6. Gravholt C, Vestergaard P, Hermann A, Mosekilde L, Brixen K, Christiansen J. Increased fracture rates in Turner's syndrome: a nationwide questionnaire survey. *Clin Endocrinol (Oxf)* 2003;59:89-96.
7. Sutton E, McInerney-Leo A, Bondy C, Gollust S, King D, Biesecker B. Turner Syndrome: Four Challenges Across the Lifespan. *Am J Med Genet* (in press).
8. Coffey A, Atkinson P. Concepts and coding. Making sense of qualitative data: complementary research strategies. Thousand Oaks: Sage Publications; 1996. p. 26-53.
9. Hill V, Sahhar M, Aitken M, Savarirayan R, Metcalfe S. Experiences at the time of diagnosis of parents who have a child with a bone dysplasia resulting in short stature. *Am J Med Genet* 2003;122A:100-7.
10. Carmichael P, Ransley P. Telling children about a physical intersex condition. *Dialogues in Pediatric Urology* 2002;25:7-8.
11. Krahn G, Hallum A, Kime C. Are there good ways to give "bad news"? *Pediatrics* 1993;91:578-82.
12. Gath A. Parental reactions to loss and disappointment: the diagnosis of Down's syndrome. *Dev Med Child Neurol* 1985;27:392-400.
13. Slijper F, Frets P, Boehmer A, Drop S, Niermeijer M. Androgen insensitivity syndrome (AIS): emotional reactions of parents and adult patients to the clinical diagnosis of AIS and its confirmation by androgen receptor gene mutation analysis. *Horm Res* 2000;53:9-15.
14. Freedman B. The roles and responsibilities of the ethics consultant. Hagerstown: University Publishing Group; 2000.

15. Keeley D. Telling children about a parent's cancer [Editorial]. *BMJ* 2000;321:462-3.
16. Whiteford L, Gonzales L. Stigma: The Hidden Burden of Infertility. *Soc Sci Med* 1995;40:27-36.
17. Gonzalez L. Infertility as a transformational process: a framework for psychotherapeutic support of infertile women. *issues Ment Health Nurs* 2000;21:619-33.
18. Cain M. *The childless revolution: What it means to be childless today*. Cambridge: Perseus Publishing; 2001.
19. Lalos A. Breaking bad news concerning fertility [Opinion]. *Hum Reprod* 1999;14:581-5.
20. Starke M, Möller A. Parents' needs for knowledge concerning the medical diagnosis of their children. *J Child Health Care* 2002;6:245-57.
21. Wilson B, Reiner W. Management of intersex: a shifting paradigm. *J Clin Ethics* 1998;9:360-9.
22. Hasnat M, Graves P. Disclosure of developmental disability: a study of parent satisfaction and the determinants of satisfaction. *J Pediatr Child Health* 2000;36:32-5.