

Turner Syndrome Foundation

Affects 1 in 2000 Females

The Implications of Withholding a Diagnosis

Well-meaning parents looking to safeguard their daughter's childhood from difficult information sometimes grapple with the issue – How and when is a good time to share the diagnosis with my daughter? As evidenced in the white paper, "Truth-Telling and Turner Syndrome: The Importance of Diagnostic Disclosure" by Sutton et al, it appears the answer to this question is sooner rather than later.

This study was conducted to research the effects of secret keeping on individuals with Turner Syndrome, specifically pertaining to diagnosis and infertility. The researchers interviewed 97 girls and women and 21 parents. Thirty percent were not informed of their diagnosis and 15 individuals were not informed of the infertility component. This study directly explores how girls and women with TS experience the disclosure process with both parents and health care providers. The participants were interviewed on topics such as concerns and challenges associated with TS, suggestions for health care providers on how to improve health care experiences for those with TS, and suggestions for newly diagnosed girls and their parents on how to address the diagnosis.

The girls and women with TS who stated they were unaware of their diagnosis also reported learning of their condition in a negative way. Most learned of their diagnosis from their health care provider who assumed they already knew. One woman who learned of her infertility at age 19 shared her story, saying: "The doctor finished his exam and I got dressed and went it. 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."

Others learned of their diagnosis by confronting parents, doing their own research, reading about their condition in school, or overhearing conversations. Many of the women who found out unexpectedly noted that there were long-term consequences. Emotions the women described included anger, sadness, and betrayal. One woman who learned of her infertility 11 years after her diagnosis had this to say: "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 fill in the gaps to tell you more of the story."

The main reason parents decided to keep the diagnosis a secret was because they felt they didn't have the right tools to disclose the diagnosis. Parents felt that health care providers did not provide enough answers to their questions, let alone enough answers to help their daughters. Parents of one child described the situation in this way: "And a couple 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." Some parents stated that they were trying to

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protect their children from the heartbreak of infertility. Others needed time to adjust to the diagnosis themselves before sharing with their daughters.

After asking the girls and women for advice for those newly diagnosed, many noted the importance of truth-telling and open communication within the family. They explained that girls with TS are aware that something is not quite right so it's best to explain the situation instead of keeping them in the dark. Lastly, participants advocated for age-appropriate diagnosis disclosure. One woman who was unaware of her infertility for seven years gave this advice: "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."

Overall, this study suggests that withholding a TS diagnosis from the person affected can lead to depression isolation, fear, and mistrust of health care providers. The potential for learning of the diagnosis in a negative way also increases greatly. Full disclosure begins the process of self-education toward becoming an advocate for one's own health care. Many parents noted that the hardest part of disclosing a TS diagnosis is infertility. Though many wanted to protect their daughters from the social stigma often attached to infertility, disclosing this information allows girls to accept infertility as part of their self-identity early in development. Sharing this information early on allows for an open discussion and acceptance of alternative parenting methods as well.

For health care providers disclosing the diagnosis, here are some tips to make this easier for your patient. Begin by asking a parent how much information they would like to receive. Always deliver a diagnosis in person. Be aware that parents and patients will have questions. Schedule a secondary appointment when you will have the time to fully answer any questions and concerns. This will also give the patient and parent time to digest the diagnosis so they will be more prepared to listen. Understand that at the time of diagnostic disclosure a patient or parent may have trouble retaining information. Consider providing an easy to read document that explains what TS is, basic health implications, and general standards of care.

Ultimately, parents and health care providers need to work together to ensure timely diagnostic and infertility disclosure so girls with TS are able to develop psychologically and socially, and so women with TS may have the best quality of life. When a girl with TS asks about her condition, parents and health care providers should answer honestly and fully.