

Telling the Truth about Turner Syndrome: Disclosure of a diagnosis and infertility to a romantic partner

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ABSTRACT

Telling the Truth about Turner Syndrome: Disclosure of a diagnosis and infertility to a romantic partner

A thesis presented to the Graduate Program in Genetic Counseling

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Turner syndrome (TS) is a sex chromosome abnormality causing a variety of physical features, renal and cardiac defects, and infertility. TS occurs when one sex chromosome is partially or completely lost, resulting in a karyotype of 45,X, in true or mosaic form, in most affected females. Research has shown that women with TS often struggle with low self-esteem and have trouble forming social and romantic relationships. In addition, studies have identified infertility as the primary concern for these women, no matter their age. Little research however, has looked at the process and outcomes of disclosure of TS and infertility by these women, and research specifically on discussions with romantic partners is lacking. We created a qualitative study exploring the experiences of women with TS who disclosed their syndrome and infertility to romantic partners. This study involved semi-structured in-depth interviews with 10 women that focused on feelings about life with TS and infertility, disclosure to romantic partners and advice for other women. Our participants ranged in age from 23-38 years, and the number of partners they had ranged from one-10. We transcribed the interviews and subjected them to thematic analysis, which identified common themes among participants. We found that participants who

were more confident and accepting of TS had more positive experiences with disclosure to partners. Often, a strong sense of self originated from having supportive parents. Participants also felt that there is a benefit of being in control of the disclosure conversation, and that disclosure to romantic partners presents unique challenges. Most participants had positive attitudes about TS, and appreciated the opportunity to share their experiences and offer advice to others with TS. This advice included seeking guidance from women who have disclosed to partners, disclosing in a comfortable manner, being proud of yourself, and creating a strong support network. Based on our participants' experiences and advice, we hope to create a resource that health professionals, including genetic counselors, can share with other women with TS, to help them feel confident regarding disclosure.

Keywords: Turner syndrome, infertility, disclosure, romantic partners, genetic counseling, self-esteem, advice

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Introduction

Turner syndrome (TS) is a common sex chromosome abnormality, occurring in approximately one in every 2000 live female births (Frias & Davenport, 2003). TS occurs when there is a complete or partial loss of the second sex chromosome, resulting in a karyotype of 45,X in many affected females. Other karyotypes, including 45,X/46,XX, 45,X/47,XXX, 46Xi(Xq), and 45,X/46,XY, are the underlying etiology in as many as half of TS cases (Hook & Warburton, 2014). While clinical features of Turner syndrome vary significantly and relate, in part, to the underlying chromosomal constitution, affected women most frequently have short stature, a webbed neck, and cardiac and renal defects (Sutton et al., 2005). Inability to enter puberty, primary amenorrhea and infertility are also near universal issues for women with TS (Castelo-Branco, 2014).

Living with a diagnosis of TS can have a negative impact on the psychological well-being of affected individuals. Previous research has demonstrated a high prevalence of shyness, social anxiety and impaired self-esteem for women with TS (Schmidt et al., 2006). Danielewicz and Pisula (2005) also found that girls with TS have decreased self-esteem and are self-conscious of their physical features, health and physical abilities. In addition, studies have shown that dealing with the complication of infertility can be particularly challenging for women with TS. Sutton et al. (2005) found that infertility was the primary concern for all participants with TS. Many women in that study mentioned how having TS interfered with dating due to fear of rejection, and a large number felt worried about pursuing romantic relationships. This and other

studies led to the hypothesis that the anxiety that women with TS have about infertility may be associated with their difficulty forming relationships with romantic partners.

The disclosure of a genetic diagnosis to romantic partners is challenging because hereditary conditions have implications for one's own health, can cause worry about what the future holds for those affected, and can potentially be passed on to the next generation. Modi et al. (2010) concluded that it was easier to discuss information about a genetic disorder with family members than partners. Participants in that study found that disclosure to partners is unique because of expectations of marriage and children. In that study, negative responses to disclosure were rare, but were more frequent when disclosing to partners than to anyone else. Klitzman and Sweeney (2011) also studied disclosure of genetic conditions to partners and found that individuals worried about rejection and often avoided or postponed the disclosure, yet many felt they should disclose due to an expectation of trust in the relationship.

The limited research surrounding disclosure of a TS diagnosis has focused on discussions between healthcare professionals and parents, and the effects of parents sharing information with their daughters. Starke et al. (2002) found that parents, who receive information about TS in a clear manner from a provider, were more accepting of their child's condition. The same study explained that a lack of information about TS from a healthcare provider can cause anxiety, feelings of being different, and social withdrawal of the whole family. Studies examining disclosure from parent to an affected daughter have found that it is important to disclose a TS diagnosis in a thoughtful and considerate manner. Results from other research suggest that telling a girl about a TS diagnosis in a way that focuses on her strengths, is more likely to foster healthy psychological development (Kagan-Krieger, 2001). Therefore, it is likely that the way in which a girl first learns about her diagnosis of TS will affect her throughout her life.

It is reasonable to think that women with TS who recalled learning about their diagnosis as a positive experience, may be better adjusted in adulthood and more comfortable sharing information about TS and infertility with partners. At the same time, the increased frequency of low self-esteem (Schmidt et al., 2006), the universal concern in women with TS about infertility (Sutton et al., 2005), and the demonstration of the difficulty sharing any genetic diagnosis with partners (Klitzman & Sweeney, 2011) would suggest that disclosure would be especially challenging to women with TS. To date, no study has looked at the discussions women have had about their diagnosis of TS and related infertility with romantic partners.

The goal of our research study is to explore the experiences of women with TS who have disclosed both their diagnosis and the related infertility to romantic partners. We hope to use the insights gathered from this study to create a resource for other women with TS as they go through their own disclosure process. Finally, we hope the results of our project will provide guidance to the parents of girls with TS about how to broach the subject of disclosure with their daughters as they get older.

Methods

Brandeis University's Institutional Review Board approved this study.

Study Population

We recruited participants through the Turner Syndrome Society of the United States using a notice posted on their website and in their online newsletter. We also recruited participants through an email sent to women in the database of the Turner Syndrome Support Group of the Boston Area. We also recruited some participants by word of mouth. Specifically, a woman who edited a book called "Standing Tall with Turner Syndrome", mentioned the study to women she knows who have TS. Recruitment materials encouraged interested individuals to email the student researcher directly. We completed an eligibility assessment by phone for each potential participant. Subject inclusion criteria were that women must: be at least 18 years of age, speak fluent English, have a confirmed diagnosis of Turner syndrome, have been in a romantic relationship, be infertile, and have disclosed TS and infertility to a romantic partner. Thirty-three women contacted us expressing their interest in the study and we arranged eligibility screening calls for the first ten respondents. One woman was not eligible to participate, so we arranged a screening call with one additional respondent, to meet the maximum of ten participants.

Once we determined an individual was eligible, we emailed an informed consent document for her to read and sign. She then emailed the form back to the student researcher, who signed the document as well. We returned a copy of the informed consent document with both signatures back to the participant, and then scheduled phone interviews. We offered each participant a \$25 gift card as a token of appreciation for their time.

Interviews

We pursued a qualitative study design because this allowed for flexibility throughout data collection and provided a way to identify themes through in-depth analysis. We designed a semi-structured interview guide with open-ended questions. The three areas of focus in the interview guide were 1) Feelings about having Turner syndrome including experiences with the diagnosis and attitudes about living with TS and infertility, 2) Experiences with disclosure to partners including details of first and most recent disclosures and differences from disclosing to others and 3) Advice for others. We used the same interview guide for each participant, but the specifics of the questions depended on the issues that arose. Following the recorded section, which included questions in the three areas, we gathered demographic information on each participant and recorded this by hand. The interviews lasted between 35 and 68 minutes each.

Data management and Analysis

We recorded all interviews on a secure website called freeconferencecalling.com, and a professional confidential transcription company transcribed all of the interviews. To ensure the privacy of the participants, we de-identified the transcripts, and assigned each one a code number. Recordings and transcripts were stored on Brandeis University's secure website, box.com. We analyzed our data using ATLAS.ti (version 7.5.6) software. We coded information from the transcripts to condense the data into analyzable units. We assigned codes to sections of texts and paragraphs based on important and interesting findings that we noted in the transcripts. We further categorized the codes into code families, and then into broader themes.

Results

Characteristics of Study Population

Ten women participated in this study, ranging in age from 23-38 years. All participants were willing to share information about their lives with TS and experiences disclosing the syndrome and infertility to romantic partners. These women all identified as White/Caucasian, and all had at least some college education. Six of the women were currently married, two had long-term boyfriends, and two were single. Eight of the women received a diagnosis of TS at birth, while two occurred in childhood. Eight participants saw specialists for medical conditions related to TS, the most common being hypothyroidism and high blood pressure. The number of partners these women had ranged from one to 10. Seven women stated they disclosed their diagnosis and infertility to their first romantic partner, and three of the women were currently in relationships with the first partner to whom they disclosed. Seven first disclosed as young adults, two participants as teenagers, and one did not specify her age at the time of her first relationship or disclosure. Tables 1 and 2 display information about the participants' demographics and their disclosure experiences.

Themes

We designed our interview guide to focus on three key areas of discussion: (1) Feelings about having Turner syndrome and infertility, (2) Experiences with disclosure to romantic partners, and (3) Advice for others. Within each area we asked the participants questions, coded their remarks, and identified the themes that emerged. Table 3 displays the themes and subthemes within each area of discussion.

1. Feelings about having Turner syndrome and Infertility

The importance of sense of self and self-esteem

Many participants expressed that TS is just a part of who they are, and they do not define themselves by this condition. Women stated they often felt confident and capable of dealing with challenges surrounding TS because of their upbringing and learning to be true to themselves. Despite the fact that most women expressed that they grew up with supportive parents who helped them gain self-confidence, many also felt that having TS and infertility caused them to feel different from others, which created feelings of isolation.

a) Strong sense of self comes from supportive parents

Many participants expressed how supportive their parents were about having TS, and felt that this had a strong influence on their self-esteem. Participant 5, who had a diagnosis of TS since birth, explained,

“They (parents) weren't ashamed of it, and they never let me use it as an excuse. And they never babied me because of it. Like they would never let me say ‘oh, I can't do this because I have Turner,’ or, ‘I can't do this because I have Turner.’...and they encouraged me to try my hardest, and they were always proud of me.”

Participant 2, a woman who always told her friends about TS related issues, and casually mentioned TS in conversation with her partners, added,

“It's (TS) nothing to be ashamed of. And my mom treated it like that.”

Participant 8, whose parents made an effort not to treat her differently from the rest of the family, explained that she was always confident enough to be herself.

“And they (parents) were always really proud of me that I did my own thing; I didn't try to fit in and do what everyone else was doing just to be cool. I was very true to myself.”

b) Infertility component of TS caused a sense of isolation

A common finding among participants was that infertility made them feel different, and created a sense of isolation from other women. Some felt this at the typical time of puberty, while others experienced it when friends or family members began having children. Participant 9, who reported that she was teased due to her short stature and looking different from others, felt frustrated when she could not relate to her friends going through puberty. She stated,

“The girls in school are talking about, ‘Oh, I got my period today,’ that type of thing. And I felt like I was missing out kind of on a rite of passage. I kind of felt isolated from my friends who were going through normal changes in their bodies and growing up.”

Some women felt that the sense of isolation is strong when others were becoming pregnant.

Participant 8 indicated,

“So as well-meaning as my sister-in-law and my mom are, it always kind of felt like I was not in the club...it does make you feel a little out of the inner circle when you see everyone getting really excited that my sister-in-law is pregnant. I think the idea of having children is almost like a rite of passage of being a female.”

She also said,

“It can feel very alone. It can feel like even if you have a loving family that supports you, if they don't have Turner syndrome and they don't know what you're going through, it can feel very lonely.”

Participant 3, who discussed feeling like an outsider among her peers, expressed,

“Most little girls always want to be a mom. I remember playing dolls with my friends and pretending that they were our little baby girls. So I remember I was a teenager and I realized that's not going to happen, it made me kind of upset that it wouldn't happen.”

2. Experiences with Disclosure to Romantic Partners

Benefit of being in control of the conversation

We asked participants to explain the circumstances surrounding disclosure of TS and infertility to their romantic partners. Some women planned their disclosure conversation, and were able to decide ahead of time exactly what to say, while others disclosed because an event prompted the discussion. Those women who were able to be in control of the conversation felt

that the disclosure process was easier and less stressful than for those who disclosed because of an event. Participant 9 discussed the benefits of disclosing to her current partner on her own terms. She decided only to share the most relevant information with him at first. She explained that when he asked her why she was going to a cardiologist appointment, she answered,

"Well, I have a condition called Turner syndrome— part of the condition is that I have my heart condition. So that was like the first initial dipping your toe into the water, just feeling it out type of conversation. So with him (her current partner), I actually got to do it the way I should have and it was much less stressful and much less anxiety-inducing."

Participant 1, who disclosed for the first time to her husband, had a unique situation provide an opening for her disclosure. She knew that she wanted to initiate the conversation with her partner, but was looking for an opportunity to begin to disclose on her own terms.

"A friend of mine with Turner had actually just moved in with me, and she was actually starting to date another friend of ours at this point when I was still dating my husband. She had shared it with her boyfriend, and so, then I said, "Well, I do, too." I knew I wanted to share it with him, and that just presented the perfect opportunity to tell him."

In contrast, we found that women who disclosed due to prompted events when they were unprepared, felt more hesitant and awkward about their disclosure conversations than women who were better able to plan the initial discussions. Participant 9 discussed how her first disclosure occurred when her partner asked why she never complained about her monthly cycle. This led her into an explanation about TS and infertility at a time when she was not prepared to have that conversation. She said that she felt this unplanned disclosure was the reason that relationship ended.

"It was a very open and kind of abrupt beginning, but I'm like, okay, all right, well, here we go. I had thought about small ways to bring it up gradually, very slowly, so that it wasn't just a huge conversation all at once. I think that may have genuinely been the downfall to our relationship, was the fact that it kind of did end up in a lump sum marathon conversation, instead of slowly bringing it up over time...gradually introducing things on your own terms, or over time most likely would be a much better way to go."

Participant 10 had two separate disclosure experiences prompted by certain events, and she was not able to be in control of the process or plan out the conversation in either situation. A friend initiated her first disclosure experience. She explained that,

“My friend kind of, not forced me into it, but kind of did in a way. She was just like, “Well, why don't you tell him now?”...Oh, geez. Here we go. Yeah, that's basically why I told him, that's kind of what pushed me over the edge to finally tell him. I think I was just more hesitant just because I wasn't sure how to approach it.”

The second disclosure began after her partner questioned if she could be pregnant. She stated,

“That's when I realized that we really needed to have a talk about it, I realized that he didn't really have an understanding of what it was. And it was an awkward start to the conversation.”

Some of the women, who were not able to plan or prepare for their disclosure conversation, stated that they wished they had been able to, because it would have given them a chance to disclose in a more comfortable way. Participant 10 explained,

“Well, for me, I would want it to be about finding the right time, the right place, and the right way to say it. It's not a casual conversation that you can just have anywhere or you can just say whatever you want.”

Disclosure to partners presents unique challenges

We asked participants to compare disclosing their diagnosis to romantic partners to their discussions with others. All participants felt that disclosure to romantic partners was different than telling their friends or family, because they felt at risk of losing their partner, and were worried about the possible expectations a partner may have regarding starting a family. Some women we interviewed were also concerned about the medical issues related to TS, and how those would impact their partner's life.

a) Fear of losing their partner

Numerous participants expressed that they were nervous their partners would not be accepting of their infertility, and ultimately would end the relationship. Participant 1 stated:

“I felt more at ease about sharing it with a friend than I did with him initially, just because I was more scared. I was a little scared that, what if I tell him and I lose him.”

Participant 8 added,

“My bigger issue was, I was very worried about, in the end, having a guy tell me that he was totally fine with adopting and not being able to have kids, but five, seven years into marriage or three years into marriage, he changes his mind.”

Participant 2 also mentioned concerns of a partner feeling differently after disclosure, and

ultimately ending the relationship.

“You don't want them to break up with you, or not accept you or love you. You don't want to scare them away or think you've got something horribly wrong with you, or think you're defective.”

b) Concerns about the effects of TS and related infertility on romantic partners

A number of participants shared that another challenge specific to disclosing to romantic partners is worry over how their having TS and infertility directly affects their partner's life.

Four participants explained that by choosing to build a life with someone who has a medical condition and infertility, a partner is agreeing to have to deal with medical concerns, and to never have biological children as a couple. The statement made by participant 4 further explains this:

“Because in my thinking, you're part of a couple, and it's important that whoever you're with understands that you having Turner is going to affect his life as well. And if you stay together, you'll have to make choices as a couple that maybe other couples wouldn't...knowing that there may be some unexpected health crises that have to be dealt with, and understanding that it's going to have an impact on him as well.”

Participant 9 described the difference between the effect of TS on romantic partners and friends or family, saying,

“Friends and family, they love you unconditionally; they're going to be there to support you no matter what. But with a romantic partner, there's a completely different level of responsibility and a completely different level of understanding that has to happen, because obviously when you're in a romantic relationship, they're going to be the ones that you form your future family with. And so, it's either you have children together, or you don't have children together. You are going to have that conversation.”

Participant 10 added,

“When you're speaking to a romantic partner, you are speaking to someone who is going to not suffer, but who's going to have to deal with the side effects of Turner syndrome right along with you. Because your friends aren't planning on having children with you. They're not going to be tied to you in that way. Whereas a romantic partner, they are going to be forced to go through alternate methods with you. It's not just going to be you in that battle. And so, it's a little bit harder to talk to romantic partners about it in that way. Because they're going to be dealing directly with the side effects.”

Self-esteem affects comfort level with disclosure to partners

Among our study participants, there appeared to be a correlation between a participant's self-acceptance and confidence and the way she felt about sharing information about her diagnosis and infertility with a romantic partner. Participants who had a strong sense of self were less likely to struggle with the idea of disclosing to partners. For example, participant 5, who grew up aware of her diagnosis and infertility expressed,

“I really wasn't hesitant, because it was sort of like, okay, if he can't accept that, then obviously I'm not the right person for you.”

Participant 2, who has always been open about her diagnosis and accepting of who she is, stated,

“I feel like this is me and if you don't like me, then that's fine, whatever, accept me for who I am. I mean, I've always kind of been like that...if he's not going to love you because of this, then that's not the relationship you're meant to be in or the person you're supposed to be with.”

On the contrary, women who struggled with their sense of self or mentioned low self-esteem, generally felt more nervous and unsure about the process of disclosure. Participant 7 had never told any friends about her diagnosis of TS and infertility, and to this day worries about the possible negative assumptions people may make about her if they knew she had a chromosome abnormality. Regarding disclosure to partners, she stated,

“And I hesitated. And I think it was more, how is he going to react? Is he going to think that I'm some kind of crazy person? Is this a deal breaker, the infertility?”

Participant 4, who mentioned always feeling insecure, explained her thoughts after disclosing to a partner:

“I just doubted that he truly was okay with all of that. And I don't know how much was actually a vibe I was getting from him, or just my own insecurities. Like me just not trusting myself, trusting the fact that someone would actually want to be with someone like me, who has been through all this. Never trusting that somebody would actually just accept me for me...it makes it hard being in a relationship sometimes when you feel like you don't deserve or you can't fully trust someone, it makes it hard.”

Participant 10, a woman who had disclosed her diagnosis of TS and infertility to several partners starting as a young teenager, further explained,

“It's just always been the one thing that's made me...feel ashamed or more like a disappointment. I think, well I think personally, like because I'd wanted kids so badly, I automatically put more weight on that, because I feel like it's a bigger letdown for myself. But especially when it comes to partners. I guess that's always just made me feel like I'm less worthy as a romantic partner.”

3. Advice for others

We asked our participants to offer advice to others with TS, and all were appreciative of the chance to share their suggestions based on their experiences of disclosure to partners.

Seek guidance from women who have been there

A number of participants reflected on the lack of guidance they received before sharing their diagnosis with a partner, and felt that other women with TS who are thinking about disclosure to romantic partners need direction on how and when to begin these conversations.

For example, participant 4 said,

“I would definitely have liked to hear other women's stories. It definitely would have been helpful to have some sort of guidance. And I think that...would have been the most helpful is like, how much information to share the first time you talk about it. Because you don't want to inundate them with all this information.”

Participant 7, who discussed not having spoken to any women in similar situations, added,

“I think it would have helped me not feel so alone having this conversation. I think I had always kind of planned in my mind exactly what I wanted to say when I got to that point in a relationship. I just think I felt so alone. I'm like, nobody else has to have this conversation but me. And maybe would have helped with the timing, too. Instead of when do I have to have this conversation, what is the risk if I do it now versus later? What are the pros and cons?”

Be proud of who you are

Another piece of advice that emerged from discussions with a number of our participants was to be confident in what you have to offer a romantic relationship as you go into the process of disclosure to your partner. Participant 3 said,

“Don’t feel ashamed about infertility. I think that’s the big thing for me, is making sure that Turner girls don’t feel ashamed of not being able to have children.”

Participant 10 emphasized this further, adding,

“And I would just want to make sure that they knew that it doesn’t make them less of a woman, it doesn’t mean that you’re a worse choice than someone who doesn’t have Turner syndrome.”

Participant 7 stressed that women with TS are just as worthy partners as anyone else, stating,

“Don’t sell yourself short, because I’ve had several friends with Turner who wound up divorced already. I think that they’re settling for the first person that proposes to them or the first serious relationship that they get into, because they’re afraid that nobody else will accept them for who they are. I think they need to be comfortable with who they are before they get in a relationship.”

Participant 10 described the importance of not hiding the diagnosis and being honest during disclosure,

“Don’t sugarcoat anything. And don’t try to hide yourself. I just feel like if you try to hide behind it and you try to hide that aspect of your life, it’s not allowing you to grow in your relationships.”

A number of participants discussed how being proud of yourself also means understanding that the right partner will accept you for who you are. Participant 1 explained,

“I mean, if the person really cares about you, they’re going to love you and care about you for who you are, no matter what happens and no matter what’s going on with you.”

Disclose in a manner that is comfortable for you

Another common sentiment among participants was that there is no one correct way to disclose, and every woman needs to do so in a way that puts her most at ease. Many women did however, emphasize that disclosing slowly can be beneficial. Participant 9 explained the need only to discuss things as they come up. She said,

“Gradually introducing things as they come up, or over time most likely would be a much better way to go about it than just all of a sudden dropping everything in one conversation. Honestly, the biggest thing I could say to any woman going through disclosure is, start slowly.”

Participant 4 also expressed this idea, explaining,

“You don't have to share everything all in one fell swoop. You can take a breath and it'll be okay, and share on your own time. It can be hard to remember that.”

Create a strong support network

Almost every participant expressed the importance of creating a strong support network of others with TS. These women felt that having a support network and being close to others with TS is invaluable for dealing with all aspects of the syndrome.

Participant 5 stated,

“Building that support network, getting really involved with the Society. It's just been an amazing experience...Especially after going to (TS) camp that first year, I have always kind of viewed it as kind of more of a blessing than a hardship. The girls that I met at (TS) camp, they have been my best friends. They've been an amazing support network.”

Participant 10 added,

“It wasn't until I went to (TS) camp that I really started accepting it and embracing it. Once I did that, I was like, this is a part of who I am and if somebody doesn't like it, who needs it?”

Part of having a strong support network for most of our participants involved attending TS conferences. For some participants, these conferences were helpful in learning about disclosure, and hearing how others approached the process. Participant 1 mentioned,

“We do actually talk a lot about these types of things in some of the conference sessions. So I have had a little guidance ahead of time. There's one speaker that has come to the conference before, and she actually said...Turner syndrome is like a screening. It really allows us to kind of screen the people in our lives in the sense that once we share it, we can see how they react.”

Participant 5 added,

“The conferences, they actually offer a lot of times a workshop on how to share with a partner and the best ways, some advice.”

Table 1: Demographic information of the 10 study participants

Age Range	23-38
Race/Ethnicity	
White/Caucasian	10
Education	
Some college	2
Bachelor's degree	5
Master's degree	3
Karyotype	
45,X	7
Mosaic	2
Unknown	1
Ongoing Medical TS-related Management*	
Autoimmune disease	1
Cardiology	1
Endocrine	2
Hypertension	3
Thyroid	5
Current Relationship Status	
Single	2
Boyfriend	2
Married	6

*some women reported being followed for more than one medical condition

Table 2: Disclosure information

Participant	Number of Partners	Age of first partner	Disclosed to first partner?	First partner you disclosed to is current partner?	Have a current partner?
1	Two	High school	No	Yes	Yes
2	Few	High school	No	No	Yes
3	Few	20's	Yes	No	No
4	Few	29	Yes	No	No
5	Few	College	Yes	No	Yes
6	One	Unsure	Yes	Yes	Yes
7	Few	High school	No	Yes	Yes
8	Few	College	Yes	No	Yes
9	Few	College	Yes	No	Yes
10	Ten	13	Yes	No	Yes

Table 3: Summary of areas of discussion, themes, and subthemes

Area of Discussion	Theme	Subtheme
1. Feelings about having TS and Infertility	Importance of sense of self and self-esteem	Strong sense of self comes from supportive parents
		Infertility causes a sense of isolation
2. Experiences with Disclosure to Romantic Partners	Benefit of being in control of disclosure	
	Disclosure to partners presents unique challenges	Fear of losing partner
		Direct impact on a partner's life
	Self-esteem affects comfort level with disclosure to partners	
3. Advice for Others	Seek guidance from women who have been there	
	Be proud of who you are	
	Disclose in a comfortable manner	
	Create a strong support network	

Discussion

The 10 qualitative interviews conducted for this study provided a first look into the experiences and issues surrounding disclosure of TS and related infertility to romantic partners.

Feelings about having TS and Infertility

We first explored our participants' feelings regarding living with a diagnosis of TS and the related infertility. The key theme that emerged from this section was the importance of sense of self and self-esteem. Most of our participants had very positive senses of self, were confident, and were true to their own identity. This is a particularly interesting finding for women with TS, because studies have shown they often have difficulties in social situations, a low sense of self and problems forming relationships (Carel et al., 2006). Given this, we expected that participants would more often talk about struggles with self-esteem and insecurities, and how that influenced the disclosure process. While many women discussed self-esteem, most did so in a way that was different than we anticipated, with only a few women mentioning a lack of self-esteem having a negative effect on relationships. While our participants may not represent the TS population as a whole, our study demonstrates that not every woman with TS struggles with low self-esteem.

One subtheme that emerged in the discussions with participants about their sense of self and self-esteem was that those who felt more confident typically attributed this to their upbringing. For example, one woman's comments exemplified this when she discussed how supportive and encouraging her parents were, and stated that the way she grew up living with TS and how her parents dealt with the condition affected her in a positive way throughout life. In contrast, a few other participants expressed that their parents imposed strict limitations and

restrictions because they worried about their well-being or were over-protective and concerned about discussing TS issues with them. These women were more likely to state that they were insecure, had a lower sense of self or were fearful that others would see them as different. For our participants then, low self-esteem and insecurities may relate to their upbringing, and not just to the innate low social cognition and self-esteem struggles seen in previous research on women with TS. Our data suggest that an affected woman's parents influence how she views herself, and how parents deal with their daughters growing up with TS may explain the differences in self-esteem seen in affected women. Whether a person's self-esteem struggles are related to TS itself or to her upbringing, parents of young girls with TS may be able to learn from this finding and appreciate the need to be open in discussing TS related issues with their daughters, and to be encouraging and supportive about the condition and related implications.

A subtheme seen in the discussions with participants, which was independent of whether they had a strong sense of self or were insecure, was that infertility created feelings of isolation. These women noted that this feeling was particularly strong when one's friends began going through puberty, as well as when friends and family members became pregnant. Many participants discussed how they felt that having children was a female rite of passage that they would never be able to experience, or they expressed that because of their infertility they felt less worthy as romantic partners. Some participants also talked about how they felt unfit as women because they were not able to fulfill their evolutionary role of reproducing. Even women with a strong sense of self who discussed having supportive parents that openly discussed TS, stated they felt different from others because of infertility. It is possible then, that in the larger population of women with TS, those who have even lower self-esteem than that seen in our

participants, may find that the infertility component of their disorder creates an even stronger feeling of isolation and has an even greater impact on their sense of self.

These findings are significant for the TS community as well as for healthcare providers who care for these women. The fact that all participants expressed a sense of isolation, regardless of upbringing, highlights the importance of helping all women with TS realize they are not alone in feeling different due to infertility. Making parents of girls with TS and women aware of this at an early age may help them become more accepting of being infertile and more comfortable discussing their infertility with others. Healthcare professionals should discuss the potential impact of infertility with parents of affected girls, be involved in the discussions with their patients as they get older, approach the topic in a sensitive manner, and provide resources to their patients. Doing so may have a positive impact on girls and women with TS and help them feel more connected, supported and better understood by the medical community.

Experiences with Disclosure to Romantic Partners

The major goal of our study was to gain insight into the experiences of women with TS disclosing their diagnosis to romantic partners. We interviewed participants about the first time they shared their diagnosis and infertility with a partner and, if applicable, about their current or most recent disclosure experience. We asked about specific information that participants shared, their feelings during the process, and the reactions of their partners. We did not specifically analyze data gathered about partners' reactions, given that every partner, every relationship, and every conversation is different, and there is no way to predict a certain reaction from a partner.

A significant theme that emerged when exploring women's feelings about disclosure to romantic partners, was that there was a perceived benefit to being in control of the discussion and that there was a difference in the disclosure experience depending on whether it was a

spontaneous or planned conversation. The majority of our participants stated they were able to take control and plan when they wanted to disclose to their partners. These women generally felt that disclosure was easier when they were able to decide when to have the conversation, introduce topics at a comfortable pace, break down the information and share only what they wanted to. While the information initially shared with partners varied, almost all women did address the infertility component during the first conversation. Because previous studies have shown that disclosure to romantic partners was stressful and caused individuals with genetic conditions to worry about negative reactions from partners (Klitzman & Sweeney, 2011), it makes sense that our participants felt less anxious when they were able to take the lead in the conversation and plan the appropriate amount of information to discuss.

The disclosure process however, appeared to be more difficult for those who had to do so because of a prompted event, and this occurred with three participants. When discussing the disclosure conversations she had with two different partners, one participant noted the difference between being able to plan ahead and decide what information to provide during disclosure, compared to having the conversation occur spontaneously. This woman explained that, in a previous relationship, a partner asked her why she never complained about her monthly cycle, which made her feel pressured into having the conversation about TS in that moment, even though she was unprepared to do so. In contrast, when her current partner asked her why she was going to a cardiologist, her prior experience allowed her to assess how much information was necessary to share at the initial moment. Though prompted by a question from this partner as well, she was able to retain control of the conversation and felt comfortable just touching on the basics of TS and mentioning she has a heart condition. This made her feel less stressed and anxious about the process. Experiences such as this one may help others with TS recognize the

benefits of being in control of disclosure to romantic partners, and maximize their opportunity to feel confident about the initial conversations. Disclosure is a sensitive and complex process, and if other women with TS can see the value of planning out the conversation and only sharing what is comfortable, this may have a positive impact on their own disclosure experiences.

Another theme that emerged from our participants' comments was that disclosure to partners presents unique challenges. Most notably, women worried that telling a partner about infertility would be especially difficult because a partner may already be planning or hoping for a biological family with her. This is consistent with research by Modi et al. (2010), which found that sharing information about a genetic diagnosis with partners is a different experience than disclosure to anyone else, largely because of the expectation of having children. Our participants felt that disclosure of infertility would alter their partner's future plans much more directly than those of a friend or relative.

Most of the women in our study stated that one issue specific to their discussions with romantic partners was that there is a fear of losing a partner after disclosure. This is consistent with research by Klitzman and Sweeney (2011) that found individuals worried about rejection because of their genetic condition and either avoided or postponed disclosure to partners. What is unique to our study however, and to TS in general, is that participants' fear of losing a partner often stems from the worry that their partner would not want to continue a relationship with someone who is infertile. No matter how accepting a woman with TS is of her infertility, participants felt that having to share this information with someone who hopes to have a biological family, would potentially jeopardize their relationship.

Another subtheme that emerged in the comments from participants was concern about TS and infertility impacting a partner's life in multiple ways. Many of the women acknowledged

that by choosing to be in a relationship with a woman with TS, their partners were agreeing to never have biological children as a couple and this may include making choices as a couple such as adoption, or deciding not to have children. Their partners were also, by being in a relationship with these women, dealing with the potential health problems associated with TS, and some participants commented that a partner may fear that having TS shortens the woman's lifespan. Thus, participants felt that their diagnosis of TS has more far-reaching implications for a partner than for friends or family members.

Finally, we saw that the self-esteem and identity of a woman with TS affected her feelings about disclosure to romantic partners. Participants who were confident and accepting of TS, felt better about the disclosure experience than women who suffered from low self-esteem. Women who had positive self-identities generally felt more capable during disclosure and understood that even if a partner rejected them, they were strong enough to overcome that setback. In contrast, some participants expressed that, due to their own insecurities or feelings of being different due to TS, they were extremely hesitant during disclosure, or did not trust that their partner would ever accept them for who they are. One woman, who had significant doubts about herself admitted that it was due to her own insecurities that the relationship with her partner ended. These findings suggest that self-confidence is associated with a more positive disclosure experience. Healthcare providers and parents should focus on instilling acceptance and positive self-identities in girls with TS from a young age as this may allow them to be better able to face challenges in the future, such as discussing their diagnosis with romantic partners.

Advice for Others

Our last area of focus was asking participants to provide advice for other women with TS who have not yet gone through the disclosure process. Overall, participants were very excited to

have an opportunity to offer guidance and words of wisdom to the TS community. In addition, many mentioned how appreciative they were for the chance to discuss these issues, and how they hope that their stories will make a difference.

One universal sentiment among participants was that women should seek guidance from others who have been through the disclosure process. Our participants believed that, had they received help on how to approach disclosure to partners, they would have felt less alone in the process, and they would have had more of an idea of how much information to share. Some mentioned that they would have benefitted from advice about when to disclose, while others explained they would have enjoyed hearing what other women said to initiate the conversation. This lack of guidance may relate to the fact that, as a number of participants pointed out, their doctors and parents focused on medical aspects of TS, and spent less time discussing the psychosocial aspects. In addition, while a few women explained that some TS conferences included workshops about disclosure, the majority of our participants did not mention attending the sessions. Given participants' recommendation that others with TS seek guidance about disclosure to partners, providers need to be prepared to offer assistance to their patients regarding this topic. Genetic counselors can help women with TS who have not disclosed, by identifying resources and by connecting them to others with TS who can offer first-hand experiences.

The next piece of advice was for women with TS to be proud of who they are. Many participants expressed how important it is not to feel ashamed and stated that having TS and infertility does not make someone less of a woman or a less worthy partner. Being proud of oneself also means understanding that the right partner will accept someone for who she is. Participants often saw disclosure as a screening process, saying that if a partner is not accepting, then they are not the right partner. This is somewhat unexpected, given the documented self-

esteem issues that women who have TS struggle with, but may be due to the overall higher levels of self-confidence seen in our study participants. It is also possible that these women's disclosure experiences have actually made them stronger. Our participants' experiences and their strength, pride and confidence, may serve as a source of inspiration for other women who are going through the disclosure process and their advice may help other women to have positive experiences themselves.

Another suggestion from our study population was for women to disclose in a manner that is comfortable for them. The women we spoke to acknowledged that every partner and each situation is going to be different, and therefore there is no single way to go about the disclosure process. As a result, what is comfortable for one woman may not work for another. Participants felt strongly that there is no rush to explain all aspects of TS, advised women to take the disclosure process slowly, and to make their partner aware that they are open to discussing matters on an ongoing basis. Many women wanted others to know that they should be honest and open regarding what having TS and infertility means for oneself, one's partner, and their future together as a couple.

Being involved with a TS support organization was an important factor in helping the women in our study form their positive self-identities, gain confidence and view TS in a positive light. This leads to participants' final recommendation – create a strong support network of others with TS, or who know about TS. Several women talked about how their first time attending TS camp, or a TS conference, or meeting someone else with TS, was a turning point in their lives in terms of self-acceptance. This is not surprising given that the goal of disease specific organizations is to provide education and positive experiences for affected individuals. The women with TS in our study, who have created support systems through these interactions,

were more likely to be accepting of TS, which, in turn, helped them in feeling comfortable about themselves and about sharing information about their diagnosis with romantic partners.

The words of advice provided by our study participants could be motivational to other young women with TS. In fact, many participants mentioned how helpful it would have been for them to hear from others who had gone through the disclosure process. There is a need to explore the topic of disclosure with women and girls who have TS and to remind those thinking about disclosing to a partner that they are not alone. The TS community, being so well-established, can play an important role in providing a forum for discussing these issues and allowing women who have been through disclosure to share their experiences openly and honestly. To help facilitate this, we will use the advice we gathered to create a resource for women with TS who are hoping to find guidance about disclosure to partners. Our hope is that this resource will be published on the TSSUS website, or be available at TS conferences.

Limitations and Future Research

This was an exploratory qualitative study designed to learn about the experiences of women with TS who have disclosed their condition and related infertility to romantic partners. Our analysis focused on data from a small sample from a self-selected group of women. Participants all identified as White/Caucasian, had some college education, and those who had medical problems were generally mild. Our findings therefore, represent those of a limited demographic group, and may not be generalizable to women with TS as a whole.

Our method of recruitment, primarily through the TS Society and TS support group, may have created a bias in the data we gathered. People who are involved in such organizations or support groups may be more accustomed to talking openly, more likely to have friends with TS, and may feel more confident and adjusted to living with TS. In fact, the 10 women who

participated all generally had positive attitudes about TS and all but one felt good about at least one disclosure experience. We relied on the internet for most of our recruitment and communication with potential participants, which may have narrowed down potential participants by making it easier for people who are technologically savvy to enroll. The age range of participants supports this idea as there were no participants older than 38-years-old.

Recall bias is another important potential limitation of our study. All of the data collected was based on what participants said they could remember about certain events in their past and it is not possible to know whether their answers accurately reflect their past experiences. Finally, only the student researcher did the qualitative analysis and coding of the interview transcripts, which means there may have been subjectivity in the coding of the themes.

Even with the data collected from this study, there are many areas regarding disclosure of TS to partners that warrant further research. Gathering similar data on a larger sample size would allow us to determine whether the findings in this study are reflective of a larger population of women with TS. It would also be worthwhile to continue this study with a follow-up survey, including more detailed questions about the romantic relationships themselves and sexual experiences or intimacy. It would also be important to learn more about these sensitive topics from women with TS who come from different communities, ethnicities, and socioeconomic backgrounds. Finally, interviewing or surveying the partners of women with TS regarding the disclosure process would provide valuable insight on their experiences and reactions to learning about their partner's diagnosis of TS and related infertility.

Conclusion

The purpose of this study was to explore the experiences of women with TS who have disclosed the syndrome and the related infertility to romantic partners. A common theme we saw was the importance of sense of self and self-esteem, and we found that this correlated with how participants felt about disclosure. In our study, those women who were more confident and accepting of TS felt better about disclosing than those who struggled with low self-esteem. Many women indicated that positive self-esteem came, in large part, from growing up with supportive parents. However, regardless of one's upbringing, all participants explained that infertility caused isolation and feelings of being different from others. The women in our study felt there was a benefit to planning out the disclosure conversation ahead of time, and being able to disclose on one's own terms. Disclosure to romantic partners also presented unique challenges, because of the direct impact infertility has on a partner, and fear of a relationship ending following disclosure. When asked to offer advice to others with TS, many participants discussed the importance of seeking guidance, learning from women who have gone through the process, and disclosing in a comfortable manner. Finally, they encouraged others to not be ashamed of TS and to create a strong support network of others within the TS community.

Genetic counselors and other health professionals working with women who have TS or with parents of a newly diagnosed daughter, may use insights from our study to further discuss the issues that surround disclosure and suggest additional resources in the TS community, particularly women who have been through disclosure themselves. We hope that the results of

this study will allow providers to serve families or patients with TS better, and help these women when it comes time to have conversations about TS and infertility with their romantic partners.

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Appendix A: Letters of Permission

October 29, 2014

To Whom It May Concern,

It is my understanding that Niri Carroll's research project for her Master's thesis in genetic counseling involves an important social aspect of Turner Syndrome. As the Editor of *Standing Tall With Turner Syndrome* I have evaluated her project and feel that it is extremely worthwhile, and I am eager to help her accomplish her goals.

The subject of her research is how and when a woman with Turner syndrome tells her romantic partner, about having this condition, and the related issues of infertility that usually accompany this diagnosis. There are many adolescent girls and young women who struggle with how and whom to tell about their condition, and this project has potential to shed light on the issue in a way that will have practical benefits for women with Turner syndrome.

Therefore I plan to support Ms. Carroll in her work by helping her to recruit married women with Turner syndrome, and by remaining involved till the completion of the project in May of 2015.

Sincerely,

Claudette Beit-Aharon
617-916-5115-home
617-821-5043-cell
claudette.beitaharon@gmail.com

October 24, 2014

Ms. Patricia Collins
Leader
Turner Syndrome Support Group
Of The Boston Area
447 Cushing Street
Boston, MA 02043

Dear IRB:

I am very happy to assist and support Niri Carroll with her thesis project about women with Turner Syndrome and their experiences disclosing their syndrome and infertility to their boyfriends, husbands, and or partners.

I can help Ms. Carroll recruit participants by using the database of 130 members of the Support Group of The Boston Area as well as introduce her to married women I have spoken to about this project and who have already expressed their willingness to participate.

I understand Ms. Carroll's project will be on-going until the end of May 2015 and I am more than happy to assist her during the entire length of this project.

Please know that the Turner Syndrome Support Group is glad this issue is getting attention through a thesis paper being written about the subject. We hope this research and thesis may help all women with Turner Syndrome as well as helping all genetic counselors who will work with women who have Turner Syndrome during their careers.

Sincerely,

Patricia Collins
Leader
Turner Syndrome Support Group
of the Boston Area

Mara Gittess, M.A., L.P.C.
3400 Bissonnet – Suite 282
Houston, TX 77005
(713) 666-1354

10/29/14

To: Members of the IRB

I am a member of Niri Carroll's thesis committee. The goal of Ms. Carroll's qualitative research project is to describe/explore the experiences of women with Turner Syndrome who have shared their diagnosis, more specifically their infertility, with their romantic partner. As part of this study she will be interviewing women with TS who are in a relationship and have disclosed their diagnosis and infertility with their partner. In addition to being on Ms. Carroll's thesis committee, I am also a woman with Turner Syndrome and am close to various members of the community. Therefore, I will be aiding her with recruiting participants for her study.

Ms. Carroll would like to use the understanding obtained from the study to create written materials containing helpful advice for those women with Turner Syndrome who might be struggling with disclosing their diagnosis and infertility to a romantic partner. I believe that this research project will benefit many women with Turner Syndrome and am honored to aid her in any way I can with this research project.

Sincerely,

Mara Gittess, M.A., L.P.C.



To whom it may concern,

The Turner Syndrome Society of the United States will support the research of Niri Carroll. We will aid in recruitment for her study by posting the information seen on her flyer to those who receive our emails or visit our website.

We are pleased to aid Niri in her recruitment for her thesis study and are eager to hear of her progress and results.

Cindy Scurlock

Executive Director

Appendix B: Recruitment Notice

Copy of the email I am going to send to go along with the recruitment notice

Email Subject: Turner Syndrome Research Opportunity

My name is Niri Carroll and I am a graduate student in the Genetic Counseling Program at Brandeis University. As part of my graduate school training I am conducting a qualitative research project. I am currently seeking volunteers to participate in this project. The goal of this study is to explore the experiences of women who have shared their diagnosis of Turner syndrome and infertility with romantic partners. I hope that the knowledge gained from this study will be valuable to other women with Turner syndrome, genetic counselors and healthcare professionals.

I have attached the recruitment notice for the project to this email. This recruitment notice provides more information about the project, as well as information about who to contact if you are interested in learning more or participating in the research study.

Thank you very much for your time.

Sincerely,
Niri Carroll
ncarroll@brandeis.edu

Are you a woman living with Turner syndrome?

Have you been, or are you currently, in a romantic relationship?

I am a graduate student in the Genetic Counseling Program at Brandeis University. I am seeking volunteers to participate in a qualitative research project. The goal of this study is to explore the experiences of women who have shared their diagnosis of Turner syndrome and infertility with romantic partners. It is my hope that the knowledge gained from this study will be valuable to other women with Turner syndrome, genetic counselors and healthcare professionals.

Participation in this study is voluntary and open to women who meet all of the following criteria:

- Are 18 years of age or older
- Have Turner syndrome and infertility
- Have discussed their Turner syndrome diagnosis and infertility with romantic partners
- Speak fluent English

Following a brief phone questionnaire to assess eligibility, participants will be asked to take part in one audiotaped telephone interview that will last approximately 45 minutes. Participants will be given a \$25 gift card to Amazon.com as a token of appreciation for donating their time.

All identifying information of participants will be kept confidential and will be destroyed after completion of the study. Identifying details will be changed to protect the privacy of the participants.

If you are interested or you know someone who might be interested in learning more about the study, please email Niri Carroll at ncarroll@brandeis.edu with your name, phone number or email address, and the best time to contact you.

I look forward to hearing from you.

Thank you!

Niri Carroll
Genetic Counseling Graduate Student
Brandeis University
Waltham, MA

Appendix C: Eligibility Screening

Hello, my name is Niri Carroll. I am a genetic counseling student at Brandeis University. As part of my graduate school training, I am conducting a study with women who have Turner syndrome to learn about disclosure of their diagnosis to romantic partners, including sharing the infertility component of the syndrome. How did you hear about this study?

The data collected from this study will be used to help understand the experiences of women with Turner syndrome. My hope is to create resources that will be available to you and other women with Turner syndrome to help facilitate such conversations with romantic partners.

Taking part in this study is completely voluntary and you can stop at any time.

Would you be willing to answer a few questions to see if you are eligible to participate?

1. How old are you?

- Over 18 years Younger than 18 years

2. Do you have a confirmed diagnosis of Turner syndrome?

- yes no

3 What is your karyotype? *45X, 45X/46XX, 45X/46XY, ring X (rX), deletion (delX), isoXq, 45X/46XiXq, unsure, other*

4. Have you ever been in a romantic relationship?

- yes no

5. Have you discussed your diagnosis of TS with a past or current romantic partner?

- yes no

6. Have you shared the infertility component of having Turner syndrome with a romantic partner?

yes

no

7. Do you speak fluent English?

yes

no

Individuals younger than 18 years are ineligible to participate in the study. Individuals who have not been in a relationship will be ineligible to participate. Individuals who have not shared their diagnosis and infertility with a partner will be ineligible. Individuals who do not speak English fluently will be ineligible to participate.

If a participant is ineligible: *Thank you very much, however, for this study I am looking for women who meet very specific criteria which includes (the criteria that they do not meet).*

Informed Consent:

- I. Conduct informed consent process and discuss confidentiality
 - a. *The aim of the study is to learn more about the experiences of women sharing the diagnosis of Turner syndrome and related infertility with romantic partners. I will be asking you questions about when and how you disclosed your diagnosis to your partner, how you felt about this experience, advice you may have for other women, and if you received any guidance for the process. I will also be asking specific questions with regard to the infertility component of Turner syndrome. Any experiences you can share will be valuable.*
- II. Explain the presence and purpose of recording equipment
 - a. *With your consent, I will be audiotaping our interview so that I do not miss anything. Is it okay with you if I audiotape the interview?*

Afterward, the interview will be transcribed by a confidential transcription service and your name will not be associated with it. Any potential identifying information that might come up as we talk will be deleted from the transcript before data entry.

- b. Explain that the informed consent will be emailed to them

I will be emailing you a copy of the informed consent after this conversation. We ask that you read the consent form before the interview. You will be asked if you understood all the information in the informed consent form, whether you have any questions, and if you voluntarily agree to participate in this study, before we begin the interview.

- c. Any questions so far?

When would be a convenient time to schedule the interview?

I will email you the instructions for calling into the conference calling website.

I give my consent to have the phone interview be audio recorded _____ (Initial on the line)

Date and time for interview: _____

Appendix D: Informed Consent

BRANDEIS UNIVERSITY

DEPARTMENT OF BIOLOGY

GENETIC COUNSELING GRADUATE PROGRAM

Informed Consent to Participate in Research

**Telling the Truth about Turner Syndrome: Disclosure of a diagnosis and infertility to a
romantic partner**

Principal Investigator: GRETCHEN SCHNEIDER

Student Researcher: NIRI CARROLL

INTRODUCTION

This is a qualitative research study exploring the experiences of women who have Turner syndrome who have disclosed their diagnosis and related infertility to romantic partners. Niri Carroll, a student researcher and Masters candidate in the Genetic Counseling Program at Brandeis University will be conducting the interviews. Gretchen Schneider, the Co-Director of the Clinical Training for Brandeis University Master's Program in Genetic Counseling is providing oversight and guidance to the project.

You are invited to participate in this research study because you are a woman living with Turner syndrome who has discussed the syndrome and infertility with a romantic partner.

The decision to take part in this research study is completely your choice. You should not feel any pressure to participate. If you do decide to participate, you can decide to stop taking part in this research study at any time for any reason.

Please read all of the following information carefully. Ask any questions that you have about this research study. Do not sign this consent form unless you understand the information in it and have had all of your questions answered to your satisfaction.

If you decide to take part in this research study, you will be asked to sign and date this form. You will be given a copy of the form signed by you and by the student researcher as well. You should keep your copy for your records because it has information, including important names and telephone numbers, to which you may wish to refer in the future.

PURPOSE OF STUDY

The purpose of this research study is to learn about the experiences, feelings and thoughts of women with Turner syndrome who have disclosed their diagnosis and related infertility to romantic partners. We hope that what we learn from these interviews will provide valuable insight that we will be able to share with other women with Turner syndrome as well as professionals who provide services to them such as genetic counselors and other healthcare professionals.

PROCEDURES TO BE FOLLOWED

You will be asked to participate in a brief phone call to assess eligibility. After you have been deemed eligible to participate in the study, you will be asked to partake in a recorded phone interview. The entire phone call including the interview portion may take up to one hour. During this interview, you will be asked detailed questions asking you to relate stories of your

experiences sharing the diagnosis of Turner syndrome and related infertility with a romantic partner. The interview will be recorded, transcribed, and studied. The information collected from the interview will be analyzed and written about in the final thesis paper.

RISKS

Your participation in this study involves minimal risk. It is possible that taking part in the interview could cause you stress or upsetting thoughts and feelings. If at any point you feel uncomfortable or upset you may skip a question, stop telling a story, take a break from the interview or stop the interview entirely. Should you wish to speak with a professional about your feelings, Janet Rosenfield is available to provide additional resources and/or support. You may contact her at jmrosenf@brandeis.edu.

BENEFITS

There will be no direct benefit to you for your participation in the study. We hope that the information obtained from this study will help us gain a better understanding of the experiences of women disclosing their Turner syndrome diagnosis and infertility to partners.

ALTERNATIVES

An alternative is to choose not to participate in this research study.

PRIVACY AND CONFIDENTIALITY

All records and documents containing identifying information, such as names, email addresses, telephone numbers, and home or work addresses will be kept strictly confidential during the study. The signed informed consent will be kept in a locked file within the office of the Assistant Director of the Genetic Counseling Program, which is kept locked and is located within a suite of

offices that may only be accessed by electronic key cards. All study related documents (eligibility questionnaires, interview transcripts, audio files) will be kept in a secure location accessible only to the student researcher and the Principal Investigator. Transcripts, interview notes, and audio files will be labeled with a coded ID number, which will be assigned to you upon enrollment into the study. You will never be referred to by your real name or by any other identifying information in any reports based on the interview.

PAYMENT

You will receive a \$25 gift card to Amazon.com for participation in the research study as a gesture of appreciation for your time.

COST

There will be no cost to you to participate in the study, other than the time it takes to conduct the eligibility phone conversation and the interview.

WHOM TO CONTACT

If have any questions about the study or how to set up an appointment or an interview, please contact the student researcher, Niri Carroll, at ncarroll@brandeis.edu or (617)877-1027.

If you are having difficulty reaching Niri Carroll or encounter any problems related to study participation please contact the Principal Investigator for this project, Gretchen Schneider, at gretchen@brandeis.edu.

If you have questions about your rights as a research study subject, contact the Brandeis Committee for Protection of Human Subjects by email at irb@brandeis.edu, or by phone at 781-736-8133.

PARTICIPANT'S STATEMENT

I have read this consent form and have discussed with Niri Carroll the procedures described above. I have been given the opportunity to ask questions, which have been answered to my satisfaction. I understand that any questions that I might have will be answered verbally or, if I prefer, with a written statement.

I understand that my participation is voluntary. I understand that I may refuse to participate in this study. I also understand that if, for any reason, I wish to discontinue participation in this study at any time, I will be free to do so.

If I have any questions concerning my rights as a research subject in this study, I may contact the Brandeis Committee for Protection of Human Subjects by email at irb@brandeis.edu, or by phone at 781-736-8133.

I have been fully informed of the study with its risks and benefits, and I hereby consent to the procedures set forth above.

I understand that as a participant in this study my identity and data relating to this research study will be kept confidential.

Yes, I consent to having my phone interview be audio recorded (initial on the line) _____

Date

Participant's Signature

Date

Student Researcher Signature

Appendix E: Interview Guide

Interview Guide

Participant ID code:

Date:

Time:

Introduction

Hello, my name is Niri Carroll. I am a genetic counseling student at Brandeis University. As part of my graduate school training, I am conducting a study with women who have Turner syndrome. My study will explore the issues surrounding disclosing their diagnosis to romantic partners, with an emphasis on sharing the component of infertility.

******START TRANSCRIPTION******

This interview should last about 45 minutes. Is that amount of time okay with you? I am going to ask you some questions about your experiences living with Turner syndrome and telling others about your diagnosis and infertility. Each interview will be tape-recorded and transcribed (by a professional transcription company) so that I have written copies of my conversations with participants. I may also take notes while we are talking.

Neither the tape, notes, nor transcript of the interview will have your name on them. They will be labeled with a code that will be connected with your name on a separate document that will be kept separate from the interview information. The pages and files with your name and contact information will be destroyed when the study ends. I will not be sharing your individual answers or your personal information with anyone other than the researchers associated with this project.

The main risk of this interview is that you may become upset or anxious when talking about your experiences. If that happens and you would like to talk with a genetic counselor, I can provide you with a counselor's phone number. There are no direct benefits to you for participating in this study, but your answers may help others in similar situations. Whether or not you decide to do this interview is up to you. You may skip any questions that you do not want to answer, and you may stop participating at any time.

Do you have any questions about the informed consent form or anything else before we begin?

Are you willing to participate?

QUESTIONS

I am going to start by asking questions about your early experiences with Turner syndrome.

Early Experiences

1. What do you remember about when you first learned you had Turner syndrome?

How old were you?

Who told you?

What was your reaction?

2. Did you have any specific medical problems associated with TS? *Heart, hearing, ear infections, kidney, immune disorders, dental, vision, skeletal, thyroid*
3. What specialists did you see who had experience in managing individuals with Turner syndrome?

4. Please tell me about how and when you learned about the infertility related to Turner syndrome.

Describe your reaction to hearing about the infertility component.

At that point in your life, what were your thoughts regarding having children? Had you desired biological children?

Impact of TS diagnosis on your Life

5. Do you feel that having Turner syndrome has affected your life at all?

In good ways?

In bad ways?

Emotionally? Socially? Medically?

6. **If yes**, did your feelings about your diagnosis of TS change over time?

If so, in what way?

Discussions about TS with Other People

7. How did your parents react to your TS diagnosis?

Stress, anger, denial, acceptance, celebratory

8. What were conversations like between you and your parents regarding TS?

What was discussed?

How often were these conversations had?

9. Did you or your parents share your diagnosis with people other than family members?

If yes, with whom?

Was there a specific reason for you or your parents telling others about your diagnosis?

10. Please tell me about your experiences disclosing TS to your peers.

At what age did you start telling your peers?

What were their reactions?

How did you feel about these conversations?

11. Do you remember receiving any guidance about how to discuss the diagnosis with others? *From health professionals, from parents, support groups*

Experience with Disclosure of the Diagnosis to a Romantic Partner

12. When was your first romantic relationship?

Did you share your TS diagnosis with that partner?

*If no, “was there a particular reason you did not share with that partner?” **then go to question 13***

*If yes, ask the following, **then go to question 14***

Level of comfort, closeness, honesty, communication

How long into the relationship did you tell your partner?

What made you tell them?

Were there things that made you hesitant about sharing your diagnosis?

Can you walk me through the initial conversation you had with your partner when you disclosed your diagnosis to him/her?

What information about TS did you tell?

Did you specifically discuss infertility at that time?

Had you discussed having children with your partner prior to your disclosure?

Was it difficult to discuss the infertility component? Why/why not?

How did you feel before, during, after disclosure?

What were the reactions of your partner(s)?

What was the most challenging part of the disclosure process?

How did sharing your diagnosis affect your relationship(s)?

How long did the relationship last?

13. Please tell me about the *first* relationship in which you shared your diagnosis.

Level of comfort, closeness, honesty, communication in the relationship

How long into the relationship did you tell your partner?

What made you tell them?

Were there things that made you hesitant about sharing your diagnosis?

Can you walk me through the initial conversation you had with your partner when you disclosed your diagnosis to him/her?

What information about TS did you tell?

Did you specifically discuss infertility at that time?

Had you discussed having children with your partner prior to your disclosure?

Was it difficult to discuss the infertility component? Why/why not?

How did you feel before, during, after disclosure?

What were the reactions of your partner(s)?

What was the most challenging part of the disclosure process?

How did sharing your diagnosis affect your relationship(s)?

How long did the relationship last?

14. If you have disclosed your diagnosis of TS to more than one partner, please tell me about your current or most recent relationship in which you had these discussions.

Level of comfort, closeness, honesty, communication in the relationship

How long into the relationship did you tell?

What made you tell them?

Were there things that made you hesitant about sharing your diagnosis?

Can you walk me through the initial conversation you had with your partner when you disclosed your diagnosis to him/her?

What information about TS did you tell?

Did you specifically discuss infertility at that time?

Had you discussed having children with your partner prior to your disclosure?

Was it difficult to discuss the infertility component? Why/why not?

How did you feel before, during, after disclosure?

What were the reactions of your partner(s)?

What was the most challenging part of the disclosure process?

How did sharing your diagnosis affect your relationship(s)?

How long did the relationship last?

15. **For participants who disclosed to more than one partner:** What was different in your current/most recent relationship regarding the experience of disclosure compared to the first time you disclosed to a partner?
16. How was the experience of telling a partner(s) different from telling other people?
17. Who was the easiest person to tell?
- Peer, family member, friend, partner, coworker, other*
18. Would you have liked guidance on how to share your diagnosis with partners?
- If yes, what would have been most helpful to you?*
19. If you were to offer some advice to others with TS regarding disclosure to a partner, what would you say?

Wrap-up

We have reached the end of the tape-recorded portion of the interview. Do you have anything else you would like to add? Was there anything you thought I was going to ask but didn't? Is there anything that you wish I would have asked?

*****END TRANSCRIPTION*****

Demographics

1. How did you find out about our study?
 3. How old are you?
 4. What is your chromosomal make-up?
- 45X, 45X/46XX, 45X/46XY, ring X (rX), deletion (delX), isoXq, 45X/46XiXq, other, unsure*

5. What is your racial or ethnic background? *White, Hispanic or Latino, Black or African American, Native American or American Indian, Asian / Pacific Islander, Other*
6. What is your highest level of education? *Some high school, High school graduate, Some college credit, Trade/technical/vocational training, Associate degree, Bachelor's degree, Master's degree, Doctorate degree*
7. Are you currently employed? If so, what do you do for work?
8. Are you currently followed for any health problems?
9. What is your current marital or relationship status?
10. Did you grow up in a two-parent household?
11. Do/did you have any siblings?
12. What is your current living situation? (With parents, alone, friends, partner, etc.)

Closing comments

- a. Thank the participant
- b. Ask them how the interview experience was for them
- c. Remind them that Janet Rosenfield is available as a resource if they experience any emotional distress following the interview
- d. Reassure participant of confidentiality of responses
- e. Offer to send a copy of the abstract to the participant when the study is complete