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ORIGINAL ARTICLE

# The emotional journey of adapting to prenatally identified trisomy X

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## Abstract

There is a paucity of research on the experiences of parents of children with trisomy X (47,XXX). Increased prenatal diagnoses associated with advances in noninvasive prenatal screening necessitate a better understanding of how trisomy X impacts family systems. This qualitative investigation aimed to describe the lived experience of parents of young daughters with prenatally identified trisomy X to guide genetic counseling. Semi-structured qualitative interviews were conducted via teleconferencing with parents ($n = 11$) of girls with trisomy X, ages 6–44 months. A descriptive phenomenological approach was used to code transcripts for significant statements and reduce data into themes describing the experience of receiving a diagnosis of trisomy X and the experience of early parenting in this population. Participants described an *emotional journey* of adapting to prenatally identified trisomy X. Four descriptive themes included two related, yet distinct, life stages: Negative Diagnostic Experience and a Hopeful Early Childhood, as well as two ongoing experiences: Persistent Ambiguity and Coping with and Adapting to Uncertainty. Results suggest providers should carefully consider word choice and timing in delivery of diagnosis, and genetic counseling should provide expectant parents with current research specific to trisomy X, facilitate connections with other parents of young girls with trisomy X, introduce developmental monitoring approaches, and be prepared to support families with a range of emotional responses to the diagnosis and decisions regarding disclosure.

**KEYWORDS**
genetic counseling, parents, phenomenology, prenatal diagnosis, sex chromosome aneuploidy, trisomy X

## 1 | INTRODUCTION

Trisomy X syndrome (47,XXX) is a sex chromosome aneuploidy (SCA) in which females have an extra X chromosome compared to the typical 46,XX. Trisomy X has an estimated incidence of 1/1000 live female births (Berglund et al., 2019) and is associated with a variety of physical, medical, psychological, and developmental features. Common physical features of trisomy X include tall stature, epicanthal folds, hypotonia, and clinodactyly (Wigby et al., 2016). There are increased risks for comorbid medical features including seizures, cardiac defects, renal and genitourinary abnormalities, and premature ovarian failure, although the majority do not have these conditions (Berglund et al., 2022). Speech and motor developmental delays are more common in children with trisomy X than in the general population (Bender et al., 1983; Linden et al., 1988; Pennington et al., 1980; Tartaglia et al., 2010). Children with trisomy X are also at increased risk for cognitive deficits, learning disabilities, attention deficits, anxiety, social

  

communication deficits, and depression (Freilinger et al., 2018; Pennington et al., 1980; van Rijn, 2019; Wigby et al., 2016). Adult studies have also shown compromised neurocognitive and social emotional features and decreased educational and socioeconomic outcomes for women with trisomy X (Otter et al., 2021, 2022; Stochholm et al., 2013). However, there is a broad spectrum of phenotypic variability, with some individuals only mildly affected or asymptomatic and other individuals with more significant physical, medical, and psychological features (Berglund et al., 2022; Tartaglia et al., 2010).

Historically, trisomy X has been underdiagnosed likely due to a lack of distinctive pathognomonic features. Additionally, many problems associated with trisomy X, such as anxiety and learning disabilities, are relatively common in the general population and are not typically indications for genetic testing (Tartaglia et al., 2010; Wigby et al., 2016). Previous research suggests that only 10% of individuals with trisomy X are clinically ascertained in their lifetime (Nielsen & Wohlert, 1991). Thus, prior research was affected by small sample size and/or ascertainment bias, with more severe cases being identified postnatally in a clinical setting, skewing the perception of phenotypic presentation and variability (Otter et al., 2010).

The onset of noninvasive prenatal screening (NIPS) utilizing cell-free fetal DNA circulating in maternal serum in 2013 was adopted as an obstetric standard of care in 2016 and 2020 in the United States (American College of Obstetricians and Gynecologists' Committee on Practice Bulletins—Obstetrics; Committee on Genetics; Society for Maternal-Fetal Medicine, 2020). This advancement provides a unique opportunity to better understand the phenotypic spectrum and natural history of trisomy X with fewer biases by evaluating those girls identified before or at birth, who may otherwise go undiagnosed. As a result of clinical adoption of NIPS, an increasing number of parents are faced with prenatal results for aneuploidies, including trisomy X (Gadsbøll et al., 2020). Medical providers and genetic counselors need efficient ways to relate reliable information about what parents could expect when receiving a prenatal result. This can be an especially difficult task considering the broad variability associated with the trisomy X phenotype (Lalatta & Tint, 2013) as well as the broad cultural differences in termination rates (Otter et al., 2010).

Research on the parent experience in SCAs has only recently begun to emerge, and there are few studies specific to trisomy X. Results from an Italian study (Lalatta et al., 2010) showed parents can adapt their initial anxiety related to the ambiguity of a trisomy X diagnosis, provided they receive adequate counseling. Furthermore, despite initially traumatic responses to the diagnosis, which the authors hypothesized might explain Italy's high termination rates, most parents who continued the pregnancy recognized the value in a prenatal diagnosis (Lalatta et al., 2010). Pieters et al. (2011) interviewed eight parent dyads of children with unforeseen fetal findings of SCA, including one with trisomy X and one with mosaic trisomy X. Overall, they found parents were mostly concerned with fertility and stature and had few concerns about psychosocial development. However, this was likely influenced by the sample, as these medical features are more common to Turner (45,X) and Klinefelter (47,XXY) syndromes which comprised the remaining six families interviewed for the study. One qualitative investigation of a sample of parents of children with SCAs, including 11 with trisomy X, produced two distinct thematic analyses of the dataset. First, Jaramillo et al. (2019) examined diagnostic experiences and found parents desired a more balanced portrayal of the SCA prognosis at the time of the diagnosis, and many parents described uninformed providers who lacked information about their child's specific SCA condition. Richardson et al. (2021) focusing on the parenting experience found parents had to advocate strongly to receive a diagnosis for their child and to secure adequate support services, and often assumed the role of expert on the genetic condition. In a mixed methods U.S. survey study, Riggan et al. (2020) found that parents receiving a prenatal SCA diagnosis reported more negative emotional experiences than those receiving a postnatal diagnosis, and a majority of parents of children with SCAs found resources provided by genetics and medical professionals to be outdated and overly medical or negative. Qualitative analysis of open-ended responses from survey respondents who had received a prenatal diagnosis indicated many were unprepared for the possibility of a prenatal SCA diagnosis, diagnostic experiences varied greatly, and parents were disappointed in the lack of useful resources (Riggan et al., 2021). Overall, studies suggest parents and their daughters with trisomy X have a unique lived experience, highly influenced by their systems of support and the timing of diagnosis (Attfield, 2020, 2021).

To date, most qualitative studies on parenting in SCAs (except for Lalatta et al., 2010) have included all SCA karyotypes with little differentiation between conditions in the interpretation of results. While there are certainly overlapping phenotypic presentations between the sex chromosome trisomies, there are some notable

### What is known about this topic

Trisomy X is a sex chromosome aneuploidy, occurring in approximately 1 in 1000 female births, and is associated with variable physical, mental, developmental features. Historically, it is a condition that has been underdiagnosed, but with the adoption of noninvasive prenatal screening, its diagnostic rate is increasing.

### What this paper adds to this topic

As a result of clinical adoption of noninvasive prenatal screening, an increasing number of parents are faced with an unexpected prenatal identification of trisomy X. This paper provides parent experiences of their child's diagnostic journey and early parenting with trisomy X, which can provide clinicians and genetic counselors with reliable information about what patients might experience when receiving prenatal results and a diagnosis.

differences (e.g., risks for behavior and learning problems, societal expectations related to gender, hormone treatments, cognitive profiles), and therefore, it is likely that the experiences and challenges parents face will differ by condition. Furthermore, prior study samples have included a range of ages from early childhood through adulthood, with a mixture of prenatal and postnatal ascertainment. Contextual factors differ significantly for parents of young children who receive a prenatal diagnosis as opposed to older children receiving a postnatal diagnosis secondary to medical and neurodevelopmental concerns. Therefore, as families report a desire for more syndrome-specific, balanced, and holistic information to accompany the SCA diagnosis (Jaramillo et al., 2019), there is a need for a more in-depth understanding of parent experiences specific to trisomy X and across the lifespan. This qualitative study aims to bolster genetic counseling in trisomy X through filling this gap in the literature by describing the lived experiences of parents of very young daughters with prenatally identified trisomy X, inductively capturing emergent thoughts and priorities regarding the diagnostic process, early parenting experience, stressors and challenges, and supportive resources.

## 2 | METHODS

### 2.1 | Recruitment and inclusion criteria

Participants were recruited from a sample of parents of children aged 2–60 months with prenatally identified SCAs who were participating in an ongoing natural history study (The eXtraordinarY Babies Study; NCT03396562) and had expressed an interest in future research opportunities. Individuals were invited by email to participate if they were parents or guardians of girls with prenatally identified trisomy X with diagnostic (pre- or postnatal) confirmation. All participants provided consent prior to any data collection and the study was approved by the Colorado Multiple Institutional Review Board (COMIRB # 20-1379). Recruitment efforts were ongoing simultaneous to data collection and interpretation, and persisted until thematic saturation was achieved (Creswell & Poth, 2017).

### 2.2 | Data collection

Semi-structured phenomenological interviews aimed to capture the lived experiences of parent participants, specifically in the areas of receiving the diagnosis, impact of diagnosis on their child and the family, parenting a child with trisomy X, and support resources. The interview guide (see Table S1) was developed through a literature review with input from a multidisciplinary team with extensive experience working with children with trisomy X and their families, including a psychologist, genetic counselor, genetic counseling graduate student, endocrinologist, and developmental pediatrician. Demographic data were also collected and managed with a brief electronic survey using REDCap, an electronic data capture tool hosted by the Colorado Clinical and Translational Sciences Institute (Harris et al., 2009).

Parents were interviewed in a private space in their homes using a HIPAA compliant, secure, password-protected video platform. All interviews were conducted by the second author, a female genetic counseling graduate student without a clinical relationship to the participants. The interviewer received training and ongoing supervision from the first author, licensed psychologist with expertise in qualitative methods and senior author, licensed genetic counselor. In-person interviews in conjunction with scheduled research visits were offered; however, all participants elected to do the interview through the online platform. Interviews lasted from 27 to 64 min ($M = 49.1 \pm 13.1$). Audio was recorded through the conferencing software and transcribed using an automatic transcription service and subsequently checked and edited for accuracy before being uploaded as de-identified files into qualitative analytic software for storage and organization (ATLAS.ti Mac) along with any analytic memos taken by the interviewer before, during, or after the interview.

### 2.3 | Data analysis

A descriptive phenomenological approach was used to analyze all interview data and to capture the essence of adapting to prenatally identified trisomy X (Giorgi, 2009). Two research team members (first and second authors) acted as primary coders, reading through all interview transcripts multiple times to immerse themselves in the data, bracketing personal subjectivities through reflexive journaling and discussion, and aiming to fully understand the overarching perspectives of the interviewees. Next, coders applied open codes (Saldana, 2015) to significant statements that embodied participants' lived experiences. The process was iterative; codes were discussed with the research team and renamed or redefined as needed throughout the coding process. Phenomenological data reduction was conducted collaboratively by the team through collapsing and merging redundant codes, categorizing codes into meaning units, and eventually developing broad common themes with detailed subthemes describing the lived experiences of parents of young girls with prenatally identified trisomy X. All thematic findings were discussed and corroborated by the larger research team. Methodological rigor was supported through reflexive journaling and the triangulation of data across transcripts and across multiple investigators with varied clinical and research expertise. Member checking of themes was conducted by presenting findings and sample quotes to a mother of a young daughter with prenatally diagnosed trisomy X who was not part of the study sample (Tracy, 2010). Feedback indicated themes were credible and transferable to others with similar lived experiences in the trisomy X community.

## 3 | RESULTS

### 3.1 | Sample

Of 22 invitations, 10 families (total of 11 parents) consented to participate. Both parents were invited to participate and provided the choice of interviewing separately or together. A total of 10 interviews were conducted, nine with mothers only and one dyadic interview with both mother and father. The demographics for participants were not entirely representative of the larger study (Table 1). The majority of participants were white, married, and had earned a college degree. Families lived across the Midwest, Southern, and Western United States with their young daughters with trisomy X (aged 6–44 months).

### 3.2 | Thematic findings: An emotional journey

One superordinate theme of an *emotional journey* encapsulated the essence of adapting to prenatal identification of Trisomy X (Figure 1).

**TABLE 1** Demographics.

| | M ± SD; range, *N* (%) |
|---|---|
| Total *N* | 11 |
| Age | |
| Child (months) | 22.3 (11.6); 6–44 |
| Parent (years) | 39.3 (5.1); 33–47 |
| Respondent | |
| Mother | 10 (91) |
| Father | 1 (9) |
| Highest education completed: Respondent | |
| Some college | 1 (9) |
| College degree or higher | 3 (27) |
| Advanced degree (Master's, PhD, MD) | 7 (64) |
| Gross annual family income | |
| <$100,000 | 2 (20) |
| $100,000–$250,000 | 6 (60) |
| >$250,000 | 2 (20) |
| Timing of confirmatory diagnosis | |
| Prenatal | 5 (50) |
| Postnatal | 5 (50) |
| Identified Trisomy X with NIPT | 9 (90) |
| Advanced maternal age | 9 (90) |
| Race/Ethnicity | |
| Caucasian | 10 (91) |
| Mixed race | 1 (9) |
| Hispanic/Latino | 1 (9) |
| U.S. Region | |
| West | 4 (40) |
| Midwest | 4 (40) |
| South | 2 (20) |

Participants described a range of emotional experiences; their initial alarm, frustration, and fear upon positive screening and/or diagnosis of trisomy X and the subsequent ups and downs that followed throughout their pregnancies as well as more hopeful early years of parenting. Four descriptive themes represent this emotional journey with two related, yet distinct, life stages: A Negative Diagnostic Experience and The Hope of Early Childhood, as well as two ongoing experiences: Persistent Ambiguity and Coping with and Adapting to Uncertainty (see Figure 1). Themes and subthemes are supported below with examples of illustrative parent quotes lightly edited for clarity.

#### 3.2.1 | A negative diagnostic experience

*Poor timing and delivery of the diagnosis*

All participants reported receiving the diagnosis was an alarming and stressful experience. Many expressed frustrations with the limited or incorrect information presented by providers. Several parents described unpleasant exchanges with insensitive word choice and generalizations. Multiple participants described their providers' struggle to communicate the trisomy X diagnosis, as it required divulging the child's biological sex when some families had intended not to learn this information prior to birth.

> The OB was very vague, and it wasn't super smooth. He was like, 'Oh, wait, do you want to know the gender? Do you want to not know the gender?' And then he said, 'Wait, you have to know the gender.' Like his dialogue was going on and it was very jumbled. That probably made me more stressed out hearing it that way, too.
>
> (Mother of 11-month-old)

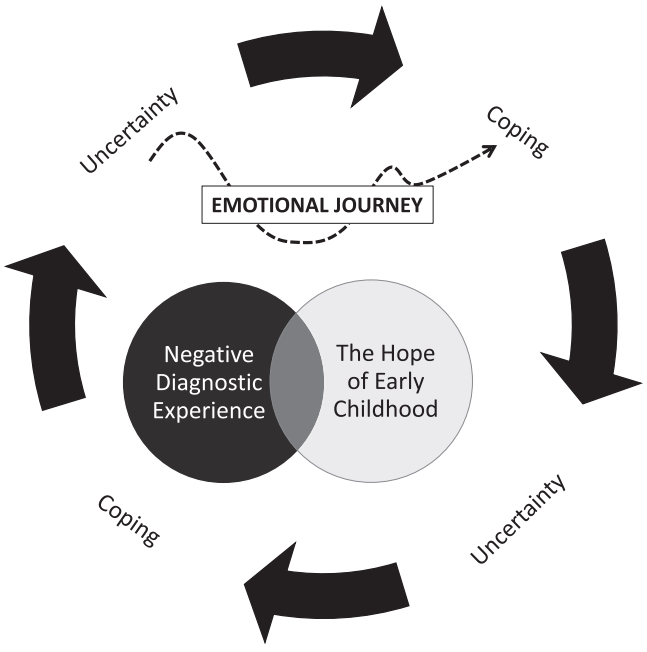


**FIGURE 1** Emotional journey of parenting a young daughter with prenatally identified trisomy X.

Some recalled presumptuous statements from providers about their daughter's limited future potential based on literature showing risks for reduced cognitive skills and educational outcomes in trisomy X.

> A doctor called us and he gave us the results. And, you know, right away, he told us, 'she's probably not gonna go to college'. And it's like, how do you say that? How do you tell families things like that? How do you know?
>
> (Mother of 26-month-old)

Several were provided with inaccurate information, such as providers confusing trisomy X with Turner syndrome (45,XO) and incorrectly telling parents their daughter had a 50/50 chance of bearing a child with Klinefelter syndrome (47,XXY).

Many described the timing as problematic, such as those who reported receiving the diagnosis by phone in the middle of a busy workday or immediately before a holiday. Most participants were informed of a positive result for trisomy X over the phone, and many experienced long wait times between the phone conversation and in-person post-test counseling. By the time parents met with genetic counselors, many had already begun their own unguided research. Several parents expressed frustration that the appointment with a genetic counselor or other provider was a wasted visit and felt the medical provider had nothing new to share. The extreme rarity of the condition meant many providers had never counseled or treated patients with trisomy X and showed little understanding of the meaning of the diagnosis for their child's future life. Overall, diagnostic experiences were described as difficult, insensitive, and uninformed, leaving expectant parents with fears and unanswered questions.

*Looming diagnosis weighs on pregnancy*

Parents repeatedly reported that learning about the risk for trisomy X diagnosis through NIPS added stress to the pregnancy.

> Yeah, I'm thinking of it now. It was scary. It was, you know, sad. It was hard. It was really hard. So, it was a sad time. And really, the pregnancy, it was sad, I was sad.
>
> (Mother of 26-month-old)

Several parents reported the trisomy X diagnosis added a layer of such intense distress that they were concerned about the impact on the baby.

> The stress of the pregnancy...To be honest, I think it kind of stuck with me [crying]. Still, I'm fine. You know, every day I'm not emotional like this. But I think that time period has stuck with me. You know, just thinking back. Wow, that was really stressful. And obviously stress isn't good for the baby. And there's only so much deep breathing and yoga and all that stuff if you even have enough time for it. You know, you can only do so much to keep yourself de-stressed.
>
> (Mother of 11-month-old)

Those who delayed confirmatory testing intensely re-experienced these negative emotions shortly after birth and their worries about the diagnosis weighed more heavily on the early days of parenting a newborn. Those with a prenatal confirmation of trisomy X reported processing the news more fully during pregnancy, and an ability to move past the looming diagnosis more quickly after their daughters were born.

### 3.2.2 | Persistent ambiguity

*The uncertainties of NIPS*

Uncertainty surrounded the NIPS experience, as the screeners were non-diagnostic and ambiguous and left parents with options for how and when to confirm (or not confirm) the findings. Our sample was split; half elected for prenatal confirmatory diagnoses and half waited to confirm until after birth. However, both subgroups similarly described their eventual genetic testing as a method to reduce ambiguity and begin to prepare for supporting their daughters. Those who waited until after birth all cited fears of harming the baby through invasive testing procedures, and a certainty that the results would not impact their decisions about continuing the pregnancy; therefore, the risk was unnecessary. For those who tested prenatally, there was more of an urgency to understand and prepare as early as possible. Multiple participants described that they understood the risks of miscarriage associated with CVS and amniocentesis, but their fears were overshadowed by the need to know.

> We pursued [amniocentesis] because we already had two kids at home. It wouldn't have changed what we did, like keeping her or any of that. But just wanting to prepare for our future and making sure that homelife was set up as needed. And if we needed to get in contact with specialists or educate ourselves more. We needed to know what we were looking at.
>
> (Mother of 26-month-old)

After taking the time to adjust to the confirmatory diagnoses (both pre- and postnatal), participants described feelings of relief and a sense that they could finally move into action, meeting with specialists and disclosing to friends and family for support.

*What is the prognosis?*

However, beyond the initial diagnosis, ambiguity returned repeatedly regarding the range of possible outcomes in trisomy X. With a long list of possible features, significant variability in the trisomy X phenotype, and a lack of recommendations for intervention, parents were unsure of what to expect. Primary features of concern were

social–emotional, learning, and communication problems parents had read were unique challenges for women with trisomy X. Many parents expressed a desire for more detailed and specific information about their daughter's prognosis and lamented the vague language in trisomy X literature.

> I want to know what I guess everybody wants to know with anything. What are the odds? Yeah, what are the odds, doctor? What are the odds? And I'm sure that's always a hard thing to do. But that was something I felt was very lacking in [trisomy X literature], it was more of a list of possibilities.
>
> (Father of 23-month-old)

While many had questions, some had never asked them of their daughters' medical teams because they doubted the availability of evidence-based answers. Several participants described what they perceived as an unfortunate discrepancy between current research and educational resources on trisomy X and another more prevalent supernumerary X condition, Klinefelter syndrome (47,XXY). Participants described that they were often counseled with, or directed to, resources for SCA conditions in general, which were almost exclusively focused on males with 47,XXY, and that this lack of information specific to their daughters' condition was distressing. Overall, the fact that trisomy X was rare, highly variable, and relatively understudied, left parents without a clear prognosis and this ambiguity led parents to doubt and question the expertise of their daughters' doctors.

*How to disclose the diagnosis?*

Parents spoke about ambiguity related to protecting their child's future autonomy and her right to choose if, and when, to disclose her invisible diagnosis to others. Multiple parents reported keeping the diagnosis of trisomy X from family members and friends, which added an additional challenge, as these parents reported inadequate social supports. In retrospect, some parents who chose not to disclose regretted keeping the information private, as it limited their support systems. Those who chose to disclose described the benefits of how open communication facilitated receiving support about their fears and needs related to the diagnosis. In addition to wondering about disclosure outside the immediate family, parents also wondered how and when they should eventually tell their daughters about the diagnosis.

> I've often wondered what having that [trisomy X] diagnosis would be like, and what is ideal for telling her about it. I think my husband and I vary a little bit on how we think we should handle it. He tends to have the perspective that 'This is no big deal at all, so why even tell her?' I think that we should tell her, and we don't need to make it sound negative. It's just what it is.
>
> (Mother of 22-month-old)

Some described a tension between wanting to allow their daughters to control the disclosure process at an older age, and their own desire to share the diagnosis with family and others to receive support.

### 3.2.3 | Coping with and adapting to uncertainty

*Research*

Parent participants reported researching trisomy X as a coping strategy after the diagnosis and well into their early months of parenting. A mother of a 44-month-old reported, "I probably read every trisomy X published paper before she was born." All parents had conducted their own informal research, especially in the weeks between the positive NIPS and meeting with a provider. Participants reported learning as much as possible about trisomy X through internet searches and reading educational materials on advocacy websites.

> I researched the hell out of everything. And I joined all the groups and all that kind of stuff. I'm generally a wallflower, but I suck up all the information.
>
> (Mother of 19-month-old)

Several participants described a desire to understand the science more deeply by reading academic manuscripts on trisomy X and making sense of the data directly from the source, rather than relying on parent-friendly translations of studies. Through research, parents became experts on trisomy X, often with what they believed was a more robust understanding than many of their medical providers. Research changed parents thinking about the diagnosis and contributed to an overall more optimistic perspective.

> Through doing my own research, I was finding that these [issues] aren't major. She can go through her whole life, and contribute to society, and these aren't going to debilitate her, hopefully.
>
> (Mother of 6-month-old)

*Reframing*

Reframing was a recurring theme throughout the interviews. Some participants reframed the diagnosis as something entirely positive.

> We don't really think much about [Trisomy X]. Occasionally my husband will say she has superpowers because she's so smart. She amazes us all the time. So, he calls it her superpower.
>
> (Mother of 29-month-old)

Parents discussed trisomy X in comparison to other genetic diagnoses with more severe outcomes, reframing initial fear into feelings of relief. Parents also frequently normalized the diagnosis, finding comfort in the fact that some women go their entire lives without ever knowing they have trisomy X, and that potential

neurodevelopmental problems associated with trisomy X (e.g., anxiety, mild gross motor delays, learning disabilities) are relatively common in the general population.

> I think the one thing that was kind of comforting is when [the counselor] said that a lot of people go through life and don't even know they have it…We likened it to, every child is going to have struggles, we just happen to know about hers earlier.
>
> (Mother of 6-month-old)

*Reaching out*

Reaching out for social supports was mentioned by all participants. Parents described how family members and friends helped them research trisomy X shortly after the diagnosis, offered a listening ear, helped with childcare, or accompanied them to medical appointments.

> There's been days where [My husband's parents] took her to her therapies because they just knew I needed a break from all that. So, they've been great. My parents, they're a long way away but they've been excellent. They always just remind us, she has a genetic condition, but it's not stopping her. As you see, she is one ambitious little girl.
>
> (Mother of 44-month-old)

Many expressed a desire to meet with other parents of girls with trisomy X to share experiences and support.

> I would really love to have a social group locally, where, especially as she grows, where she knows, there's people like me, and we're doing okay. Or, you know, if I have this trouble, just to feel like she has a peer group.
>
> (Mother of 19-month-old)

The rarity of trisomy X and inevitable geographic distance of families with similarly aged girls led parents to rely on private social media groups; however, opinions varied. While some appreciated the narrative and personal nature of the online support groups, others spoke about finding inaccurate information or feeling discouraged by other parents' negative experiences. In all, parents described immense benefits from finding authentic connections with others as they navigated both the diagnostic process and early parenting experiences.

### 3.2.4 | The hope of early childhood

*Pride and joy for daughters*

Parents described parenting as more enjoyable and less stressful than their pregnancies. Many of the concerns participants had about possible risks associated with trisomy X were not yet observed in their infant and toddler daughters. Parents described their daughters as beautiful with no notable physical features. With time to get to know their daughters and observe their personalities and strengths, parents reported a significant reduction in stress. Many reported strong feelings of love and pride toward their daughters at the time of the interviews, with positive emotions including adoration, gratitude, and joy. Parents frequently described their daughters as the perfect fit for their family system.

> She's just fun and spunky, full of energy and smart. Really smart. Yeah, she's just a joy. Honestly, she's such a joy to our family. She's so snuggly and full of love.
>
> (Mother of 29-month-old)

While some girls did show signs of early developmental and medical challenges, when asked to describe their daughters, all participants overwhelmingly focused on strengths and positive family relationships. Many participants in our study expressed a clear sense of relief after the stress of pregnancy. One mother expressed a message she wished she could back and tell herself at the time of the diagnosis.

> I would tell myself to not stress and not worry about things forever. You know, she's gonna come out perfect, and she's beautiful. And this is your little angel that you've been asking for. So, you know, everything is going to be fine.
>
> (Mother of 6-month-old)

*Appreciation of early screening and diagnosis*

In hindsight, most participants acknowledged the benefits of prenatal identification, such as the ability to prepare, research, accept the diagnosis, and to find support. Access to early intervention was frequently listed as a benefit of the early diagnosis, and multiple families reported ongoing preventative services, despite their daughter meeting developmental milestones on time. Others detailed early developmental delays and a need to advocate for therapies with multiple specialists (e.g., speech therapists, physical therapists). Many participants were grateful that the trisomy X diagnosis had opened doors for public early intervention programming that might have otherwise been denied and reported using the diagnosis to advocate for their daughters.

> I had things in place before I needed them. That's one of the things about the diagnosis, [it] sort of gives me ammunition. Instead of being a worried mom, I'm an appropriately concerned mom. I'm an advocate. But it's a lot of work. We have the developmental therapy, the physical therapy, now we're adding the speech therapy, and then on our own working with her.
>
> (Mother of 19-month-old)

Thus, in early childhood, the diagnosis was leveraged as a tool for gaining access to services and improving quality of life.

In addition to developmental supports, parents found comfort in knowing their daughters were being closely followed by medical specialists in infancy to address documented risks associated with the trisomy X diagnosis, including renal ultrasounds, echocardiograms, and audiology evaluations. Some participants were comforted by their daughter's lack of trisomy X-related health problems in early childhood. However, for participants whose daughters did have current medical problems, the genetic diagnosis had triggered early identification and ongoing specialist care.

> In some ways its almost at an advantage that we get all these resources for her because she gets these extra people who know what the heck they're doing helping raise her.
>
> (Mother of 22-month-old)

While the identification of heart murmurs and early speech delays were sources of concern and worry, participants reported strong feelings of hope about their daughters' prognoses due to the proactive nature of their medical care. Overall, participants expressed appreciation for the prenatal diagnosis, as it prompted close monitoring that might prevent more serious future medical problems.

## 4 | DISCUSSION

Our study informs genetic counseling through an in-depth illustration of the ongoing cycle of persistent ambiguity related to the diagnosis, and coping with uncertainty that parents experience as they receive a diagnosis and then parent a young daughter with trisomy X. Although parents described a wide spectrum of lived experiences, one common superordinate theme of an *emotional journey* emerged from the data, as illustrated in Figure 1, which depicts the ups and downs experienced by parents as they received the diagnosis, interacted with genetic counselors and other providers, navigated a pregnancy imbued with stress and worry, and then welcomed their daughters into their families. Our sample was limited to those participating in one of the largest and ongoing natural history studies in trisomy X. Therefore, results cannot be generalized to all families of children with trisomy X, many of whom receive more fractured care with fewer resources. However, our inductive qualitative approach provides unique perspectives on genetic counseling needs in the prenatal period and early childhood years of trisomy X, and elements of thematic findings are likely transferable to others experiencing the phenomenon.

Many parents found the initial diagnosis of trisomy X to be a distressing experience. While positive prenatal screening results have been well established to cause an increase in anxiety (Akbas et al., 2021; Kleinveld et al., 2006), our study results and other published reports specific to trisomy X show this parental distress is often attributed to provider unfamiliarity with the condition and the ambiguous prognosis (Lalatta et al., 2010; Vuorenlehto et al., 2021) and can have a lasting impact on the pregnancy. Parents in our study discussed a desire for more specific information to accompany the positive NIPS results, such as details about the prognosis, the level of risk for each associated feature, and suggestions to mitigate risks (e.g., early intervention). While previous research has been limited by small sample sizes and ascertainment bias, the current eXtraordinarY Babies natural history study of sex chromosome trisomies (Tartaglia et al., 2020) will answer many of these questions, as it currently follows the largest cohort of prenatally identified girls with trisomy X from infancy through childhood (as of 8/2023, $N = 67$). Dissemination materials from the study should be developed with the understanding that, as our participants described, families will be reading both family-friendly educational materials and academic medical journal articles. Genetic counselors can help families accommodate the news of an alarming diagnosis by facilitating their preparation for the early parenting life stage (Michie, 2020); including following the initial diagnosis with comprehensive resources specific to trisomy X, planning for ongoing clinical monitoring and assessment, recommending options for proactive early intervention, and facilitating the development of social supports.

As others have reported (Jaramillo et al., 2019), our participants described awkward and poorly timed diagnostic experiences. For some, this was complicated by the need to disclose the child's biological sex as part of delivering the trisomy X diagnosis, despite parents' original plans to delay learning the child's sex until birth. Our findings contrast with results from a recent survey on genetic counseling practices following positive NIPS for SCAs where the vast majority of genetic counselors reported they were somewhat *comfortable* or *extremely comfortable* discussing a diagnosis of trisomy X (Fleddermann, 2018). This discrepancy may suggest a disconnect between what prenatal genetic counselors are comfortable providing and what expectant parents find informative and beneficial to assist in their preparation. There is a need for additional resources with specific talking points contributing to this discrepancy so that counseling can be viewed as more effective for families (See Table S1).

Parents in our study ultimately recognized that the benefits of confirmatory genetic testing outweighed the stress the positive prenatal screening caused during pregnancy. After confirmation of the diagnosis, parents shifted into more proactive behaviors such as research, identification of appropriate resources and early interventions, and watching for early indicators of neurodevelopmental delays (Samango-Sprouse et al., 2020). These preparation benefits are particularly salient, as current research has documented increased special education services and poorer educational outcomes in trisomy X (Attfield, 2020, 2021; Berglund et al., 2020; Thompson et al., 2020; Thompson, Davis, Janusz, et al., 2022). Our findings also revealed that young girls with trisomy X may not demonstrate many obvious outward signs of their genetic difference at a young age, meaning the critical months of early childhood when the brain is most plastic and amenable to intervention could easily be a missed opportunity. The prenatal screen and subsequent confirmatory diagnosis of trisomy X can alert families to their child's potential risks for delays and, with

provider support and appropriate referrals, can help qualify children for preventative therapies in some US States (Barger et al., 2019).

Parents in our study described a variety of adaptive coping strategies to help them adapt to the trisomy X diagnosis. Coping is defined as the thoughts and behaviors used to manage events and experiences that feel beyond our control, and can be categorized as problem-focused, emotion-focused, or social support seeking (Lazarus & Folkman, 1984). Our findings indicate that from the initial diagnosis and throughout the formative early parenting months, parents of girls with prenatally identified trisomy X may engage in problem-focused coping by researching medical literature in order to answer questions their providers are unable to address. While this research may begin in early childhood as an effort to reduce stress from negative diagnostic experiences and uncertainty of prognosis, it may evolve into a more arduous role identified in parents of older children with SCA conditions who assume the role of medical expert and the burden of care coordination for a fragmented health care system (Richardson et al., 2021). Genetic counselors can also encourage social support seeking and emotion-focused coping strategies described by our participants, such as reaching out to the online community of trisomy X families and reframing initial grief into more positive and adaptive thought patterns, which may help to mitigate the emotional toll associated with being the default expert (Richardson et al., 2021).

Our study provides new insight about the hopeful and enjoyable aspects of early parenting experiences in trisomy X. Anxiety appeared to decrease from pregnancy to the early childhood phase as parents came to know their children and were reassured by their daughters' lack of obvious symptoms and their emerging strengths. This finding is reassuring in contrast to previous literature demonstrating the negative effects prenatal distress can have on parental postnatal adaptation (Dollberg et al., 2016), and expands upon research showing families of young children with prenatally identified SCAs believe that their children will have good quality of life (Pieters et al., 2011). Admittedly, our results may have been impacted by social desirability bias (Johnson & Van de Vijver, 2003), as studies show parents of children with disabilities may have a desire to present themselves in a more socially acceptable way (Goodman & Glenwick, 2012). However, prior research using a neurodiversity lens supports the presence of positive parenting experiences across a wide spectrum of genetic conditions and disabilities, highlighting the importance of genetic counseling for such conditions (Cost et al., 2021; Green, 2007; Hastings & Taunt, 2002). Parents of older children with trisomy X have reported numerous strengths of character, including kindness, perseverance, a love of learning, social intelligence, and creativity (Thompson, Davis, Takamatsu, et al., 2022). In the current study, parents enthusiastically pronounced their love and affection for their young daughters, whom they described as persistent, affectionate, fun, spunky, smart, beautiful, and perfect. Symptoms of trisomy X were limited and included mild medical and developmental findings (e.g., heart murmur, speech delays, and delayed motor milestones). While this may reflect that early childhood is a life stage of relative normal development in trisomy X, additional research is needed to describe the broad spectrum of early development in a prenatally identified sample. Participants also described an appreciation for the supportive care teams helping them to raise their daughters (e.g., research teams, early childhood therapists, medical providers) and the optimistic attitudes they had about early treatment and proactive care. These findings are supported by others reporting resiliency in parenting and adaptation achieved through various coping strategies, including emotion-focused coping with positive psychological reappraisal (Beighton & Wills, 2017; Pieters et al., 2011).

### 4.1 | Limitations

This study is limited by the sample, as our data were limited to the perspectives of parents who were proactively participating in a natural history study and were generally well educated with adequate social supports. The lived experiences of parents who do not receive regular care and supports from a specialty care clinic, through an ongoing research study, or those from more marginalized backgrounds such as racial and ethnic minorities, non-English speaking families, or families without adequate healthcare might be quite different from our participants and more research is needed in these diverse populations. Furthermore, our study only examined the earliest years of parenting in trisomy X. While we found significantly reduced stress and feelings of relief and hope in early childhood, many of the more concerning symptoms in trisomy X may not yet have emerged. It will be critical to follow these families over the life course, as parenting stress may increase again when their daughters begin to engage more outside the home, and risks associated with trisomy X (e.g., social skill deficits, anxiety, learning disabilities) become more evident and impairing (Freilinger et al., 2018; Otter et al., 2021; van Rijn, 2019). Additional input from the paternal perspective may also broaden findings, as mothers' reflections may not adequately express the lived experiences of fathers who have shown less emotional distress in prior SCA research (Pieters et al., 2011). Due to the inductive nature of our interview questions, we did not directly ask participants to report which specific provider delivered the diagnosis (e.g., OB, geneticist, genetic counselor), a factor which may have had a significant impact on the quality of the diagnostic experience and should be investigated in future studies. Despite these limitations, this study was patient-centered and highlighted the experiences of an underrepresented population in the literature. Findings revealed valuable insights from parents of daughters with trisomy X that could not be ascertained through a questionnaire. By using an inductive approach, this study was able to capture emergent themes that can support future, larger scale, hypothesis-driven studies.

### 4.2 | Conclusion

These findings suggest unique lived experiences for parents of daughters with a prenatally identified diagnosis of trisomy X during

the earliest years of life. Distinct features of the phenomenon include (1) initial interactions with providers who have never met a patient with trisomy X due to the rarity and historic underdiagnosis of the condition, (2) a relatively limited body of research and educational materials that mostly feature other more prevalent SCA conditions, (3) a variable phenotype with ambiguous prognosis and wide range of potential outcomes, (4) somewhat preserved early childhood development with concerns for later onset of more severe psychosocial and medical problems, and (5) the subsequent preventative focus of treatment in early childhood facilitating a more hopeful life stage. Results suggest providers should carefully consider word choice and timing in delivery of diagnosis, and genetic counseling should provide expectant parents with current research specific to trisomy X, facilitate connections with other parents of young girls with trisomy X, introduce developmental monitoring approaches, explain the benefits and risks of confirmatory testing, and be prepared to support families with a range of emotional responses to the diagnosis and decisions regarding disclosure.

This investigation prompts the benefits of expansion to include and compare findings with other aneuploidy conditions, including other sex chromosome trisomies (47,XXY& 47,XYY) and parents with discordant NIPS results that screened positive for a trisomy condition and subsequently revealed a tetrasomy condition (48,XXYY) after birth (Howell et al., 2022). Research should also continue to follow families, documenting potential changes in the lived experiences of families impacted by a trisomy X diagnosis over the lifespan, including entering the school system, adjustment to the diagnosis, and transitioning to adulthood. Understanding the evolution of needs for clinical care and supports as they change over time will inform a developmental model for genetic counseling in trisomy X and other SCAs.

## AUTHOR CONTRIBUTIONS

Authors Talia Thompson, Jessica Tisher, and Susan Howell confirm that they had full access to all the data in the study and take responsibility for the integrity of the data and the accuracy of the data analysis. All authors gave final approval of this version to be published and agree to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved.

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## CONFLICT OF INTEREST STATEMENT

Talia Thompson, Jessica Tisher, Shanlee Davis, Christina Miller, Jillian Kirk, Nicole Tartaglia, and Susan Howell have no known conflicts of interest to disclose.

## DATA AVAILABILITY STATEMENT

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

## ETHICS STATEMENT

Human studies and informed consent: Approval to conduct this human subjects research was obtained by the Colorado Institutional Review Board (COMIRB # 20-1379). All procedures followed were in accordance with the ethical standards of the responsible committee on human experimentation (institutional and national) and with the Helsinki Declaration of 1975, as revised in 2000. Informed consent was obtained from all patients for being included in the study.

Animal studies: No non-human animal studies were carried out by the authors for this article.

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## SUPPORTING INFORMATION

Additional supporting information can be found online in the Supporting Information section at the end of this article.

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