An Emotional Journey: Parent Experiences of Raising Young Daughters with Prenatally Identified Trisomy X

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BACKGROUND

- Trisomy X has an estimated incidence of 1/1,000
- Broad spectrum of phenotypic variability, including medical & neurodevelopmental features
- Increased prenatal diagnoses with NIPT
- Providers need efficient ways to relate reliable information about what parents could expect when receiving a prenatal diagnosis
- Prior research shows parents want more syndrome-specific, balanced, and holistic information to accompany the SCA diagnosis
- Qualitative methods highlight the voices of participants, and inductively capture the perspectives and priorities of the trisomy X community

METHODS

- Parents of young girls with prenatally identified trisomy X
- Recruited from the eXtraordinarY Babies study
- N=11 (10 mothers, 1 father)
- Interview topics: 1) diagnostic process, 2) parenting experience, 3) parenting stress and challenges, 4) coping mechanisms, 5) support systems
- Interview length: 27 to 64 minutes (M=49.1±5.1; range, N(91)
- Phenomenological thematic analysis

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CONCLUSIONS

- Parents reported negative diagnostic experiences characterized by early stress and ongoing uncertainty
- Parents also described healthy coping strategies and immense relief, pride, and joy when speaking of their daughters at present
- Early childhood years were focused on bonding with their daughters, researching trisomy X, connecting with others, and addressing risks through proactive medical care and early intervention

ACKNOWLEDGEMENTS

We would like to thank the parents who participated in the study for sharing their personal life experiences.

RESULTS

Diagnostic Experience

Poor Timing & Delivery
“An 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?” (Danielle, mother of 26-month-old)

Tainted Pregnancy
“The whole pregnancy in general to me was a debacle.” (Laura, mother of 19-month-old)

Differences Between Partners
“My husband was pretty angry. And at some point, I said to him, ‘I hope you don’t continue to be just angry, you know. Because this is what it is, and we have to digest it.’ It scared me almost to hear him so upset and mad at the diagnosis.” (Abby, mother of 11-month-old)

Early Childhood

Pride & Joy for Daughters
“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 snuggling and full of love.” (Kelly, mother of 29-month-old)

Appreciation of Early Diagnosis
“I feel almost empowered by [the diagnosis] and am trying to be her number one advocate, obviously, and just use my resources… That’s why I like her diagnosis. I’m empowered and have the ability to get help, because I have learned about it. Without [the diagnosis], you don’t know anything.” (Rebecca, mother of 6-month-old)

Uncertainty

What’s the Prognosis?
“I want to know what I guess everybody wants to know… What are the odds? Yeah, what are the odds, doctor? But that was something I felt was very lacking in [trisomy X literature], it was more of a list of possibilities.” (Peter, father of 23-month-old)

How to Parent?
“I would assume we push her a lot harder than we would if she didn’t have the genetic condition. Because we just know we want her to be the best she can be.” (Lori, mother of 44-month-old)

Disclosing the Diagnosis
“Family wise, it was a little hard just because you don’t want to disclose too much and get everybody concerned. And so, in that sense you kind of feel unsupported, that it’s just you and [your partner] dealing with it.” (Nora, mother of 26-month-old)

CLINICAL IMPLICATIONS

- Carefully consider word choice and timing of diagnosis
- Provide expectant parents with current research specific to trisomy X
- Facilitate connections with other parents
- Be prepared to support families with a range of emotional responses to the diagnosis and in decisions about disclosure