# Psychosocial experiences of individuals and families with diagnoses of sex chromosome aneuploidy

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## Background

- Integration of sex chromosome aneuploidy (SCA) detection into prenatal genetic screening protocols is positioned to increase SCA diagnoses.
- Due to variable expressivity, SCAs are poorly characterized relative to other genetic conditions.
- The psychosocial impact of all SCAs remains challenging to anticipate, making clinical management and counseling difficult.

## Objectives

We are investigating the social and emotional experiences of individuals and parents who have received a genetic diagnosis of an X or Y variation.

## Methods

Data collection:

Semi-structured, qualitative interviews with 40+ individuals recruited through community-based support organizations

Participants:

Individuals (ages of 16-21) and/or their parents who have been diagnosed with a SCA, including: Trisomy X (XXX), Klinefelter (XXY), XYY, 48,XXYY, 48, XXXY, and other X/Y aneuploidies. Exceptions: Turner's Syndrome (XO) and Fragile X.

Data analysis:

Preliminary coding analysis of fist 20 interviews is reported

**Experiences internal to nuclear family, parental responses to diagnosis, emotional and financial burden of condition.**

"I've struggled all along, just wondering if I'm a horrible parent. Why is it so hard?"

"The counseling, the psychiatrist…very expensive…[our son] is very good at meeting the family deductible"

"The sperm bank. We paid for ten years up front. That was thousands of dollars. It's a ton of expenses."

**HOME**

**Decision-making around disclosure to extended family members and friends.**

"He's better behaved in a very close environment with family than he is outside of that environment. He obviously would go to day care…and he still has issues there."

"I only have shared it with my nuclear family on my side. I don't think my husband's told his parents…if people ask me I'll say, "[Child] has a hormone imbalance."

**SUPPORT**

**Context of diagnosis and symptom management**

'We've tried some antidepressants or antianxiety medicine and mood Stabilizers. He's on Vyvanse…now he's on testosterone therapy a few months after the diagnosis. "

"She has hypermobility…the defiance has been an issue… Because of the benign tremor, because of the speech delay… we saw a neurologist."

**MEDICAL**

**Public knowledge and opinions about SCAs**

"Once the public gets educated, then it's not as scary. I think it's gonna take some celebrity to have an XXY son for it to happen and for them to be honest about it."

**PUBLIC PERCEPTION**

"It's because it's relatively unknown (…) but I think it's gonna explode. Prenatal testing will become more prevalent and less invasive very soon. the diagnosis rate will go up dramatically in the future."

"I'm very quiet about advocating for my son. Advocating happens behind the scenes."

**Supplementary services, including occupational, speech, and physical therapy.**

"If you go in to the speech therapist and go "Oh, we need speech therapy" without telling the speech therapist the cause of the speech delay, I feel you're handicapping the speech therapist and the teacher and the dyslexia specialist…"

"You need to be able to support these families of these men or boys. Because they can only be as productive as we provide the assistance for them…"

**SERVICES**

**Experiences navigating the education system, including individual education planning and seeking accommodations.**

"Her teacher is just a really good fit for her…we're in IEP. I told them what kind of person she needs… so they put her with a great teacher."

"She's in a private preschool, and for privacy reasons…I'd like her to get into the public program, where they have these therapists and people in place…"

**EDUCATION**

## Discussion

- SCA diagnoses do not have simple or easily predicted implications, but fall on a spectrum of potential experiences.
- Through internet and social networks, families connect with others facing similar experiences and identify more information about their condition.
- Some individuals perceive a SCA diagnosis as a positive distinction from their peers.
- Inadequate knowledge about SCAs and their associated symptoms presents multiple challenges for diagnosed individuals and their families in accessing social and educational services.
- Limitations: Due to our recruitment mechanism, our participant pool may be skewed toward highly-functioning populations and/or families with high socioeconomic capacity to identify and obtain social services

## References

Tartaglia, N., et al. (2015). The eXtraordinarY Kids Clinic: an interdisciplinary model of care for children and adolescents with sex chromosome aneuploidy. *Journal of Multidisciplinary Healthcare*, *8*, 323–334.

Visootsak, J., Tartaglia, N. (2013) "Sex chromosome aneuploidy." *Autism and other neurodevelopmental disorders.*

*Gale, N., et al. (2013)* Using the framework method for the analysis of qualitative data in multi-disciplinary health research. *BMC Medical Research Methodology.13, 117.*

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