

TITLE: Anxiety in Turner syndrome: Engaging community to address barriers and facilitators to diagnosis and care

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Background. Turner syndrome (TS) is a genetic condition caused by complete or partial loss of the second sex chromosome, affecting 1 in 2000 females. Despite complex medical manifestations, the TS community identifies anxiety as a major contributor to reduced quality of life. This study aimed to improve understanding of anxiety symptomatology in individuals with TS and to identify barriers and facilitators to diagnosis and care.

Setting/Population. The current joint pilot study was conducted in partnership between patient-advocacy group Turner Syndrome Colorado (TSCO) and the eXtraOrdinary Kids TS team. This partnership has been actively addressing the challenge of limited resources and fractured care for youth with TS for 10 years.

Methods. A mixed methods study design integrated community engagement, including community leaders as decision-making co-Is and a collaborative and paid community advisory board. The wider TS community was engaged through an online survey (N=135) followed by in-depth interviews (Caregivers=5, Individuals with TS=5). Descriptive statistics were used to summarize survey results. Team-based rapid analysis synthesized interview findings, which academic partners and the CAB used to develop overarching themes.

Results. Participants with TS represented diverse ages (Caregiver survey: 12y±6; individual with TS survey: 26y±12) and geographical locations. Most identified as white (93.4%) and non-Hispanic (90.0%), and caregiver respondents had high educational attainment and annual income. Half of respondents reported experiencing anxiety symptoms 4 or more days per week, and caregivers and individuals reported anxiety affects their daily life (mean of 4.2 and 5.1 out of 10 respectively). Individuals with TS reported *feeling* anxious more often at school/work, while both caregivers and individuals reported anxiety *expression* increased at home. Insomnia was the most common symptom of anxiety endorsed across age and rater groups. Children were primarily triggered by stimulating environments and medical appointments and displayed aggression and hyperactivity as symptoms of anxiety. Perceived anxiety symptoms in adolescents included clinging and rumination and were triggered by conflict and increased expectations. Therapy and medication were rated as helpful when used, and use increased with age. Qualitative themes were: Anxiety impacts the whole family, TS creates a unique anxiety experience, and there are opportunities for early identification and intervention. Stakeholder comments supporting these themes will be presented.

Conclusions. Anxiety in TS presents differently across the lifespan and may necessitate a nuanced, TS-informed and family-systems approach to diagnosis and care. We are developing educational products to share our findings. Future research directions include adapting existing anxiety screening tools and interventions to improve utility for the TS population through engagement with a more diverse community sample.