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

Alexandra Carl1, Marybel Good2, Erica Haag2, Tiana Grosskreuz1,3, Christa Hutaff-Lee1,4, Elizabeth Bennett1,5, Nicole Tartaglia1,5, Shanlee Davis1,6, Talia Thompson1,7

1. eXtraOrdinary Kids Clinic and Research Team, Children’s Hospital of Colorado, University of Colorado Anschutz Medical Campus, Aurora, CO | 2. Turner Syndrome Colorado | 3. Child Health Internship Program, Department of Pediatrics, University of Colorado School of Medicine, Aurora, Colorado | 4. Division of Neurology, Children’s Hospital Colorado, Aurora, Colorado, Department of Pediatrics, University of Colorado School of Medicine, Aurora, Colorado | 5. Division of Developmental Pediatrics, Department of Pediatrics, University of Colorado School of Medicine, Aurora, Colorado. | 6. Colorado Section of Endocrinology, Department of Pediatrics, University of Colorado Anschutz Medical Campus, Aurora, CO | 7. Child Health Biostatistics Core, University of Colorado School of Medicine, Aurora, Colorado.

Aim: to improve understanding of anxiety symptomatology in individuals with TS and to identify barriers and facilitators to reduced quality of life

**Background**
- Turner syndrome (TS) is a genetic condition caused by complete or partial loss of the second sex chromosome
- Despite complex medical manifestations, the TS community identifies anxiety as a major contributor to reduced quality of life

**Methods**
- Mixed methods study design integrated community engagement – community leaders as decision-making co-Is and collaborative and paid community advisory board
- TS community was engaged through an online survey (N=135) and in-depth interviews (Caregivers=5, Individuals with TS=5)
- Descriptive statistics summarized survey results
- Team-based rapid analysis synthesized interview findings, which academic partners and the CAB used to develop overarching themes

**Results**

### Demographics

<table>
<thead>
<tr>
<th></th>
<th>Self-advocates (n=74)</th>
<th>Caregivers (n=61)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Age (years)</td>
<td>26y±12</td>
<td>12y±6</td>
</tr>
<tr>
<td>Race (white)</td>
<td>64 (86.5%)</td>
<td>57 (93.4%)</td>
</tr>
<tr>
<td>Ethnicity (Hispanic)</td>
<td>6 (8.5%)</td>
<td>6 (9.8%)</td>
</tr>
<tr>
<td>Days per week with anxiety symptoms</td>
<td>mean=5.1</td>
<td>mean=4.2</td>
</tr>
</tbody>
</table>

### Triggers for anxiety

- Anxiety and TS Emotions
- Social Isolation
- School
- Adolescent
- Adulthood

### Symptoms of anxiety

- Early childhood
- School age
- Adolescence
- Adulthood

**Qualitative Thematics**

#### Anxiety impacts the whole family

- “Our family is to the point where, I hate to say, but we all know when we have to walk on eggshells around her.”
  - Parent of a teen with TS

- TS creates a unique anxiety experience
  - In certain basic social situations that wouldn’t cause anxiety for the average person, it might for me because of my Turner syndrome.
  - Adult with TS

- Opportunities for early identification & intervention
  - The behavior, especially when they are younger, we don’t recognize as anxiety. We see meltdowns, or trouble adjusting when things don’t go right, or perfectionism.
  - Parent of a teen with TS

**Setting and Population**
- Partnership between patient-advocacy group Turner Syndrome Colorado (TSCO) and the eXtraOrdinary Kids TS team at Children’s Hospitl Colorado (CHCO)
- Partnership has been actively addressing the challenge of limited resources and fractured care for youth with TS for 10 years
- Participants were recruited from a combination of sources:
  - CHCO Turner Syndrome Clinic
  - TSCO social media
  - INSIGHTS Registry

**Conclusions**
- Anxiety in TS presents differently across the lifespan and may necessitate a nuanced, TS-informed and family-systems approach to diagnosis and care
- Educational products to share our findings in development
- Majority responded that therapy and medication were helpful when tried
- Future research directions include
  - Adapting existing anxiety screening tools for the TS population
  - Further engagement with a more diverse community sample

**Acknowledgements**
- Funded by: NIH/NCATS Colorado CTSA Grant Number UL1 TR002535
- Thank you to all the individuals who participated in our survey and interviews!
- Thank you to our Community Advisory Board members: Marybel Good, Erica Haag, Laura Magal, Amanda Scerbeck, Jessica Bushus, Jayda Rice, Chandler Isem, Mary Rose Masters, Shanlee Davis, Elizabeth Bennett

---

- [Image](image)
- [Image](image)