ANXIETY IN TURNER SYNDROME: ENGAGING COMMUNITY TO ADDRESS BARRIERS AND FACILITATORS TO DIAGNOSIS AND CARE

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Purpose of Study: Turner syndrome (TS), a genetic condition caused by complete or partial loss of the second sex chromosome, is associated with complex medical manifestations. The TS community identifies anxiety as a major contributor to reduced quality of life. This study aimed to improve our understanding of anxiety symptomatology in individuals with TS (IWTS) and to identify barriers and facilitators to diagnosis and care.

Methods Used: A mixed methods design integrated community engagement, including community leaders as decision-making co-Is and a collaborative community advisory board. The wider TS community was engaged through an online survey (N=135; Caregivers (CG)=61, IWTS=74) followed by in-depth interviews (CG=5, IWTS=5). Descriptive statistics, linear mixed models, logistic regression, and fisher's exact tests were conducted for quantitative analysis and a rapid team-based approach was used for qualitative analysis.

Summary of Results: The sample ranged in age from 3-64 years (CG: 12y±6; IWTS: 26y±12). Majority of respondents said anxiety symptoms occur two or more days per week (CG=65%, IWTS=87%) and regularly impact activities of daily life, with IWTS reporting more frequent symptoms than CG (p=0.03). IWTS reported feeling anxious more often at school/work (71%), while both rater groups reported anxiety related behaviors were most likely to be expressed at home (CG 61%, IWTS 78%). Insomnia was the most common symptom of anxiety endorsed frequently across age and rater groups (>70% per group). Some symptoms showed significant (p<.05) linear relationships with age: physical aggression (CG OR: 0.91 95%: 0.83-0.99), crying/tantrums (CG OR: 0.90 95%CI: 0.83-0.97), clinging (IWTS OR: 0.96, 95%: 0.92-0.99), and diarrhea (IWTS OR: 1.07 95%:1.03 1.12) decreased with age, while body tension (IWTS OR: 1.04 95%: 1.01-1.08) increased with age. Some anxiety triggers increased significantly (p<.05) with age: 'dealing with conflict' (CG OR 1.09, 95%CI 1.00-1.18), 'managing expectations' (CG OR 1.16, 95%CI, 1.07-1.28; IWTS OR 1.05, 95%CI 1.01-1.10), and 'recalling negative events' (CG OR 1.08, 95%CI 1.00-1.18). Non-linear relationships with age included 'dealing with organizational tasks' as a frequent anxiety trigger in school-aged children (p=.037), and 'new friendships' in adolescence (p<.001). Therapy and medication for anxiety were reported as helpful by most respondents who had tried these strategies. Qualitative themes included: 'Triggers for anxiety are related to TS' (e.g., executive functioning deficits, medical procedures), 'Anxiety impacts the whole family' (e.g., high burden on mothers, separation anxiety), and 'Opportunities for early identification and intervention' (e.g., missed early signs, appreciation for early conversations).

Conclusions: Anxiety is common in TS; however, symptoms and triggers can change with age and may go undiagnosed or untreated during childhood. Families carry the burden of managing and accommodating for daily anxiety symptoms in the home.

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