Neurodevelopmental and Mental Health Outcomes in a National Clinical Sample of Youth with Sex Chromosome Trisomies Compared with Matched Controls: A PEDSnet Study

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Abstract:
Objective To compare the prevalence of neurodevelopmental and mental health diagnoses in youth with sex chromosome trisomy (SCT) with matched controls.
Study design Using the PEDSnet database, patients with one or more outpatient encounters and a diagnosis code mapping to 47,XXY/Klinefelter syndrome (n=1,171), 47,XYY/Double Y syndrome (n=243), or 47,XXX/Trisomy X syndrome (n=262) were matched (1:4) with controls using propensity scores incorporating eight variables. Generalized estimating equations were used to compute odds ratios (OR) with 95% confidence intervals (CI) for the prevalence of Neurodevelopmental and Mental Health composite diagnoses, prescriptions for psychiatric medications, and encounters with behavioral health providers.
Results All SCT groups had higher odds of diagnoses within the Neurodevelopmental (OR 3.90, 95% CI 3.42, 4.45, P <0.0001) and Mental Health composites (OR 2.17, 95% CI 1.78, 2.65, P <0.0001), and one or more encounter with a behavioral health specialist (OR 3.86, 95%CI 3.39, 4.39, P <0.0001) than matched controls. Youth with XXY and a mental health diagnosis were more likely to have a prescription for a selective serotonin reuptake inhibitor (OR 1.88, 95% CI 1.38, 2.57, P<0.0001).
Conclusions Compared to matched controls, youth with SCT have greater odds of a Neurodevelopmental and Mental Health diagnoses, emphasizing the need for appropriate screening, evaluation and treatment in these populations.

Keywords: sex chromosome aneuploidies; Klinefelter syndrome; Trisomy X; Jacobs syndrome; 47,XXY; 47,XYY; 47,XXX