Title: Rates of Autism Spectrum Disorder in Adults with Prader-Willi Syndrome

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Introduction: Prader-Willi Syndrome (PWS) is a neurodevelopmental disorder caused by a lack of paternally derived imprinted genes on chromosome 15q11-q13. It is associated with mild to moderate intellectual disability, irritability, outbursts, rigid behavior, and hyperphagia which increases risk for obesity (Dykens & Roof, 2008). Those with PWS have an increased risk for psychiatric issues such as anxiety, attention deficit disorders, psychosis and autism spectrum disorder (ASD). However, there are methodological concerns about the rates of ASD in PWS as most research in this area has relied upon autism screeners or checklists instead of direct assessments combined with clinical reviews to establish ASD diagnoses. Previous studies also typically include a wide age range, introducing potential age effects and observer memory bias. Finally, the nature of repetitive behaviors in PWS may inflate or complicate ASD diagnoses. This study addressed these methodological concerns in well-characterized adults with PWS.

Method: Participants included 42 adults (19 male, 23 female) with PWS ranging in age from 22 to 55 years (M = 30.41 years, SD= 8.21). The Autism Diagnostic Observation Schedule (ADOS-2) Module 3 and Social Communication Questionnaire (SCQ) were used as two ASD assessments, with ASD diagnosis determined by clinician consensus and review of pertinent data. Module 3 of the ADOS-2 was used due to the delays typical of PWS and of the successful use of other ADOS-2 modules in severely delayed adults with intellectual disability (Sappok et al., 2013). Each participant was also administered the Kaufman Brief Intelligence Test-2 (KBIT-2), the Vineland Adaptive Behavior Scales-II, and the Repetitive Behavior Scale-Revised (RBS-R).

Results: 20 adults (47.6%) were identified as meeting ASD criteria based upon the recommended SCQ cutoff score of ≥15. 13 adults (30.9%) were identified as meeting autism spectrum criteria using ADOS-2 calibrated severity scores. In contrast, only 4 adults (9.5%) received an autism diagnosis when ADOS-2 data, videos, and other behavioral data were subjected to a thorough clinical review. 3 out of these 4 were of the maternal uniparental disomy (mUPD) genetic subtype of PWS. For the group as a whole, no significant correlations were found between ADOS-2 scores and gender, age, RBS-R or KBIT-2 scores. Moderate negative correlations were found between Vineland’s Communication, Daily Living Skills, and Socialization domain scores and ADOS-2 Overall Calibrated Severity Scores (r = -.49, -.63, -.44 respectively, p < .01) and Social Affect Severity Scores (r = -.44, -.57, -.35 respectively, p < .05).

Discussion: Both the SCQ and ADOS-2 alone suggested more than twice of the sample met ASD criteria as compared to diagnoses made using a review of data and clinical consensus. Future studies should not rely on ASD screeners alone to establish probable ASD in PWS. Most adults with PWS and ASD had the mUPD genetic subtype of PWS. Issues related to administering the ADOS-2 in adults will be discussed.

References/Citations: