

Diagnoses and Characteristics of Autism Spectrum Disorder in Adults with Prader-Willi Syndrome

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Genetic mechanisms associated with Prader-Willi Syndrome (PWS) are often used to study biological models of Autism Spectrum Disorder (ASD). Data are limited, however, on the rates and characteristics of ASD in PWS, particularly so for adults. Previous estimates are predominantly based on autism screeners given to parents and may not be accurate. Additionally, less is known about adulthood than children in this population. 60 adults aged 17 to 55 recruited for a national longitudinal study on behavior and development in PWS were assessed with the Autism Diagnostic Observation Schedule-2. An expert clinical team made best-estimate ASD diagnoses based on ADOS-2 videotapes, developmental histories, and indices of current functioning. Adults were also administered the Kaufman Brief Intelligence Test-2; parents completed the Child Behavior Checklist and Vineland Adaptive Behavior Scales-Second Edition. Scores were compared across adults with PWS+ASD versus PWS only. Diagnoses were made in 7 adults, the majority of whom were the mUPD PWS genetic subtype. Adults with PWS+ASD did not differ from those with PWS alone in cognitive or adaptive functioning. Autism screeners should not be the sole index of probable ASD in PWS as rates through single measures alone may be overinflated. Diagnosis within genetic syndromes requires direct observation and consideration of developmental context. A review of adults with PWS both with and without comorbid ASD indicates the need for social activity, employment, and supports.

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