

Autism-Like Symptoms in Children with Bardet-Biedl Syndrome



Sarah-Blessing Tetedje
University of Alabama

Sarah-Blessing Tetedje, Jeremy Pomeroy, PhD
Fritz Wenzel Center for Clinical Research

Background: Bardet-Biedl Syndrome (BBS) is a ciliopathy linked to both visual impairment and higher rates of autism. Vision problems in people with BBS typically present in middle childhood or adolescence, impacting the perception of nonverbal social cues. This early vision loss can obscure the recognition of autism-like symptoms in patients with BBS. The purpose of this study was to examine associations between autism diagnoses and autism-like symptoms scores in patients with BBS ages 4-8 years (before significant vision issues tend to occur).

Methods: We included participants from the Clinical Registry for BBS (CRIBBS) who were between the ages of 4-8 and had scores from both the Social Communication Questionnaire (SCQ), a 40-question autism screening, and the Adaptive Behavioral Assessment II (ABAS II), a standardized test of developmental function. Excluded participants were legally blind, had missing SCQ or ABAS II scores, or had scores more than 180 days apart. We ran summary statistics and stratified by autism diagnosis status at the time of their tests, the timing of their autism diagnosis, whether SCQ scores were above the cutoff of 15, and genotype.

Results: We ran Wilcoxon two-sample test and Kruskal-Wallis tests to determine significance. The final sample had 98 participants. The 81 participants not diagnosed with autism at the SCQ had a mean SCQ Total Score of 9, which is higher than averages reported in most studies but lower than the cutoff. The 15 children with an autism diagnosis by the time of their SCQ had significantly higher mean SCQ scores and lower ABAS II scores than those who were never diagnosed with autism (n=61) or those diagnosed later (n = 20). Of the children who reported never being diagnosed with autism, 13% had an SCQ total score at or above the cutoff. 40% of the children who were diagnosed after their SCQ were above the cutoff. We also identified unique intermediary groups. One group had SCQ scores above the cutoff and no autism diagnosis, and the other had children with an autism diagnosis and SCQ score below 15. The former group scored lower on ABAS II measures than all other groups.

Conclusions: Autism-like symptoms appeared in early to middle childhood, before the onset of vision loss. Additionally, the timing of autism diagnosis was associated with performance on the ABAS II and SCQ. Our findings support the importance of early recognition of autism-like symptoms and associated interventions in this population.