Exploring the Interaction between Autism Spectrum Disorder and the Symptoms of Hyperphagia in Individuals with Bardet-Biedl Syndrome

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Background: Bardet-Biedl syndrome (BBS) is an autosomal recessive genetic disorder that affects several body systems. Common clinical characteristics include obesity, retinal dystrophy, hypogonadism, renal dysfunction, polydactyly, and learning difficulties/cognitive impairment. 72% to 92% of individuals experience obesity beginning in early childhood due to hyperphagia, an insatiable hunger drive caused by impairment in the MC4R brain pathway. In addition to hyperphagia, some individuals with BBS experience autism spectrum disorder (ASD) and related symptoms. Rapid and excessive eating is a problem observed in many individuals with developmental disabilities. For example, the prevalence of obesity is significantly greater among those with ASD compared to the general population. However, there is a lack of research exploring the specific relationship between hyperphagia, ASD, and BBS. The primary objective of this study was to examine how ASD-related symptomology is associated with hyperphagia manifestations in patients with BBS.

Methods: This study used data from the Social Communication Questionnaire (SCQ), the Hyperphagia Questionnaire (HQ), and the Symptoms of Hyperphagia Self Report Questionnaire (SOH) collected from participants in the Clinical Registry Investigating Bardet-Biedl Syndrome (CRIBBS). Individuals were split into three groups: children who have a caregiver fill out surveys (children HQ), adults who have a caregiver fill out surveys (adults HQ), and adults who can self-report (adults SOH). Participants or their caregivers self-report a diagnosis of ASD. Spearman's correlation and Wilcoxon rank sum tests were used to evaluate associations between SCQ scores, and various hyperphagia related symptoms.

Results: Spearman's correlation analyses indicate significant correlation between total HQ scores and SCQ scores, both in the total sample ($r=0.31$ $p = 0.0001$) and in children HQ ($r=0.30$ $p = 0.0018$). Wilcoxon analyses indicate hyperphagia scores were significantly higher in adults SOH with an ASD diagnosis ($n=23$ $p=0.037$) as compared to those without an ASD diagnosis. This association was similar in children ($n=106$, $p=0.0066$) with an ASD diagnosis, but not in adults whose caregivers completed the HQ ($n=47$, $p=0.2269$).

Conclusions: Study findings suggest more pronounced hyperphagia symptoms in BBS participants with concomitant ASD. This association could be bidirectional as well, thus future studies should confirm how both hyperphagia and ASD symptoms present and interact over the life course. In addition, a better understating is needed on how increasingly common hyperphagia/obesity treatments, including medications like setmelanotide and GLP1-receptor agonists, impact hyperphagia expression, and potentially the severity of ASD symptoms, in patients with BBS.