

Abstract – Paige Preston

Chronotype, Sleep Study Results, and Sleep Quality in Patients with Bardet-Biedl Syndrome

Background: Bardet-Biedl Syndrome (BBS) is a rare inherited ciliopathy, which causes early-onset obesity, retinal degeneration, polydactyly, cognitive impairment and dental crowding, among many other features. Sleep is closely intertwined with many aspects of physical well-being. Previous findings suggest that insufficient sleep is common in BBS, yet the relationship between aspects of sleep and well-being is unknown. This study characterizes sleep chronotype, abnormal sleep study results, and quality of life in patient with BBS.

Methods: A cross-sectional analysis was performed using data from the Clinical Registry Investigating Bardet-Biedl Syndrome (CRIBBS), the largest world-wide registry of patients with BBS as well as a previous accelerometry study involving CRIBBS participants. Accelerometry data was used to categorize participants by sleep chronotype. Quality of life, sleep study findings and recommendations, and vision were studied using data from the CRIBBS registry. Associations between sleep measures and other outcomes were analyzed using nonparametric tests.

Results: Accelerometer data revealed a high prevalence of inconsistent/mixed chronotype. Of the 175 participants for which accelerometry data was available, 111 (63%) had a mixed chronotype. Of the participants who had a sleep study, 161 (63%) reported an abnormal result. An abnormal sleep study was associated with a non-random difference in responses regarding feeling depressed compared to a normal sleep study.

Conclusions: This study adds to the understanding of sleep in BBS and identifies areas that are targets of intervention to improve health and well-being. Future research into interventions is needed to address inadequate sleep duration and an inconsistent chronotype for patients with BBS.