Title: Characterizing Sleep Problems in Infants and Toddlers with Neurogenetic Syndromes: A Cross-Group Comparison

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Introduction: Children with neurogenetic disorders are high-risk for developing sleep problems, including increased sleep latency, frequent and prolonged night wakeings, and short sleep duration (Robinson-Shelton & Malow, 2015). While researchers have explored syndrome-specific sleep problems in pre-school and school-aged children with neurogenetic disorders (e.g., Cotton & Richdale, 2006), it is unclear when and how these problems may emerge in infancy and early childhood. As an initial step to characterizing sleep trajectories and treatment targets, the current study evaluated parent-reported sleep problems in infants and toddlers with Williams Syndrome (WS), Prader-Willi Syndrome (PWS), and Angelman Syndrome (AS). Our goals were to evaluate the proportion of children with severe sleep problems in each group, as well as characterize initial cross-syndrome patterns of infant and toddler sleep behaviors.

Method: Participants included 57 infants and toddlers ages 6-47 months with WS (n = 16), PWS (n = 19), and AS (n = 14). Mothers completed the Brief Infant Sleep Questionnaire (BISQ; Sadeh, 2004), a screening measure for daytime and nighttime sleep duration, frequency and duration of night waking, sleep onset/latency, and infant sleep preferences. We first examined categorical frequencies of sleep problems using two metrics: (1) whether parents endorsed sleep as a “very serious concern” on the BISQ, and (2) Sadeh’s clinical sleep cutoffs (Shahid et al., 2012), which are met if the child wakes more than three times each night, spends more than one hour awake each night, or receives less than nine hours of sleep per 24-hour period. To isolate the specific sources of sleep difficulties in our sample, we also examined group differences in sleep duration and onset latencies. We analyzed group differences using nonparametric Fisher Exact Tests (categorical variables) and Kruskal Wallis and Wilcoxon-Mann-Whitney Tests (continuous variables). Age was similar across groups (Kruskal-Wallis $\chi^2 (2)=4.89, p=.09$).

Results: Sleep concerns were relatively common in our sample, with 30% of parents reporting sleep was a “small problem,” (n = 15; 56% WS, 16% PWS, 21% AS) and only 10% of parents reporting sleep was a “very serious problem” (n = 5; 6% WS, 0% PWS, 29% AS). Twenty-two percent of parents (n=11; 13% WS, 5% PWS, 57% AS) reported clinically significant sleep problems using Sadeh’s clinical sleep cutoffs. Both parent-endorsed (Fisher’s $p=.01$) and clinical ($p=.001$) sleep problems differed across groups, with higher frequencies of “very serious” sleep problems in AS versus PWS ($p=.03$; AS vs. WS $p=.16$; WS vs. PWS $p=.46$) and higher clinical levels of sleep problems in AS compared to WS ($p=.02$) and PWS ($p=.002$; WS vs PWS $p=.58$).

Groups also differed in specific characteristics of sleep, including amount of sleep per 24 hours (Kruskal-Wallis $\chi^2 (2)=7.92, p=.02$) and sleep latency ($\chi^2 (2)=10.53, p =.003$). Overall, children slept for an average of 11 hours and 40 minutes per 24-hour period, with an average nighttime sleep latency of 22 minutes. Relative to children with PWS, parents reported shorter sleep duration in WS ($z=1.99, p<.05$) and AS ($z=2.41, p<.01$) groups. Similarly, longer sleep latencies were observed in AS ($z=2.34, p =.02$) and WS ($z=3.20, p =.001$), relative to the PWS group. The AS and WS groups did not differ in sleep duration ($z=1.39, p=.16$) or latency ($z=6.9, p=.49$). Final analyses will examine additional indices of sleep quality (e.g. number and frequency of night wakeings) and age effects in an expanded cohort.

Discussion: Although minor sleep concerns were relatively common across the sample, parents of children with AS reported particularly elevated sleep problems, suggesting pervasive symptoms that emerge early in development. Sleep problems were most mild in the PWS group, potentially reflecting the onset of features of hypersomnia (e.g. excessive daytime sleepiness, narcolepsy, and sleep apnea) identified in older children and adults with PWS (Robinson-Shelton & Malow, 2016). Future directions include comparing syndromic data to an age-matched comparison sample and assessing longitudinal sleep trajectories across early development. As sleep is a modifiable risk factor for developmental progress, it is critical for children with neurogenetic syndromes to receive early sleep screenings and targeted, syndrome-sensitive treatment.

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References/Citations: