Title: Assessment of Sleep Behaviors in Chromosome 15q11.2-13.1 Duplication (Dup15q Syndrome)

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Introduction: Duplication of chromosome 15q11.2-q13.1 (dup15q) results in a neurodevelopmental disability characterized by hypotonia, variable intellectual disability (ID), autism spectrum disorder (ASD), and epilepsy. Duplications include an extra, isodicentric chromosome 15 (idic) or interstitial duplication of 15q11-q13 (int). While a recent study documented abnormalities in overnight electroencephalography (EEG) in dup15q syndrome1, less is known about behavioral patterns of sleep in this population. Recently, the Children’s Sleep Habits Questionnaire (CSHQ), the most commonly used caregiver questionnaire to assess sleep disturbance in children2, was investigated for use in ASD3. Given the low endorsement of items more medical in nature, a modified questionnaire was proposed. It remains unclear as to whether the modified or original CSHQ is feasible for use in a population in which more severe cognitive, language, and/or motor impairment impedes item applicability. Thus, the present study had two primary aims: 1) to investigate the use of the CSHQ to examine sleep patterns in a neurodevelopmental disability characterized by cognitive, language, and motor impairments; and 2) to characterize sleep patterns in individuals with dup15q syndrome in an effort to monitor clinical practice.

Method: Participants included 42 individuals ranging from age 4 to 42 with a confirmed genetic diagnosis of dup15q syndrome (10 int(15), 32 int(15)). The CSHQ consists of 33-items and 8-subscales and is intended for use as a screening tool to characterize patterns of sleep contributing to common medical and behavioral sleep difficulties in children. Caregivers were instructed to skip items or mark “n/a” if the question was not applicable due to their child’s degree of impairment. As valid scoring requires a response to all items, participants were split into “fully completed” and “partially completed” for analyses. Total and subscale scores were computed for both the original as well as the ASD-specific revised 23-item, 4-subscale version excluding questions related to medical etiologies of sleep disturbance. Group comparisons were completed by duplication type (int(15) versus idic(15)) and epilepsy status within the idic(15) group.

Results: Overall, 56% of caregivers did not skip any questions and the distribution of duplication type and epilepsy status did not differ between the “fully completed” and “partially completed” groups. The “fully completed” group had significantly higher cognitive abilities (full scale, verbal, and nonverbal) than the “partially completed” group. Questions most frequently skipped relied on language skills (e.g., complains about sleep problems, reports body pains at night). The revised scoring did not yield more completed questionnaires and medically-related sleep disturbances were equally endorsed. The overall average score on the CSHQ was above the normative cut-off of 41. No statistically significant differences emerged between duplication type in the total scores for either the original or revised questionnaire, or when comparing idic(15) participants with and without epilepsy. Idic(15) individuals with epilepsy had lower scores (indicating fewer symptoms) on the Bedtime Resistance and Anxiety subscales of the original questionnaire.

Discussion: Results suggest that while the CSHQ is feasible for use in a population with syndromic ID, clinicians must use caution when assessing individuals with more profound cognitive impairment. Moreover, the revised questionnaire intended for use in ASD is not appropriate for this population given the high endorsement of medically-related sleep disturbances. The overall high endorsement of sleep disturbance by caregivers of individuals with dup15q syndrome highlights the importance of screening for sleep-related disorders in this population. Results also highlight the importance of considering seizure activity, as epilepsy uniquely affected caregiver ratings of sleep behaviors in addition to impacting physiological findings of sleep in the study by Arkilo and colleagues. Future studies with a larger sample size are needed to further investigate the psychometric properties of the CSHQ in severe-profound ID and to link behavioral sleep patterns with sleep EEG.

References:

1 The Help Group/University of California Los Angeles
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