Sleep characterization in rare monogenetic groups associated with NDDs

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Background

Sleep plays a strong biological role in supporting the brain and behavior for all individuals at every stage of life. Individuals with Neurodevelopmental Disorders (NDDs) often have issues with sleep quality and sleep behaviors (Esbensen & Schwichtenberg, 2016). However, there is a limited understanding of whether rare monogenetic groups share similar sleep problems.

Objective

Here, we seek to understand more about the sleep habits and problematic behaviors surrounding sleep seen in individuals with a rare disruptive genetic mutation linked to neurodevelopmental disorders: *CSNK2A1*, *HIVEP2*, *MED13L*, and *SETBP1*.

Methods

Chronologically age-appropriate measures were used:

- **Adolescent Sleep Hygiene Scale** (ASHS; de Bruin, van Kampen, van Kooten, & Meijer, 2014) provides 8 subscale scores and an overall total sleep hygiene score.
- **Child Sleep Habits Questionnaire** (CSHQ; Fava, Tomba, & Sonino, 2012*):* CSHQ provides a total average score and eight subscale scores, reflecting key sleep domains that encompass the major medical and behavioral sleep disorders in this age group.

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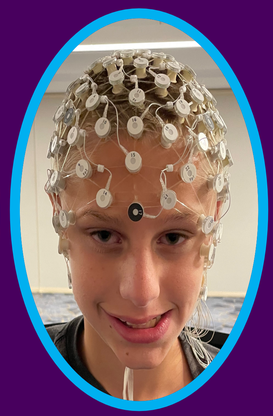
References on website

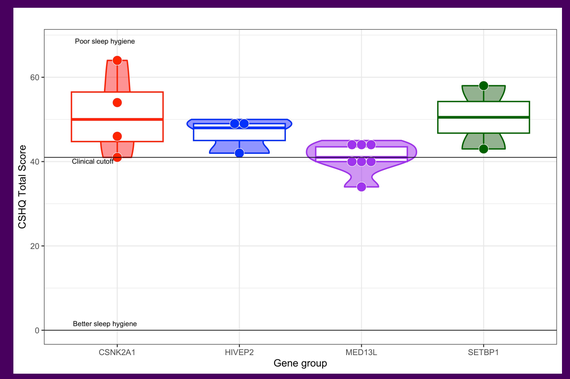
MAIN TAKEAWAY

We found **elevated sleep problems** for most individuals within these **4 rare genetic subgroups associated with neurodevelopment**. However, 3 adolescents did not exhibit frequent sleep problems (2 MED13L, 1 HIVEP2). There were no statistically significant group differences in sleep problems between these genetic subgroups.



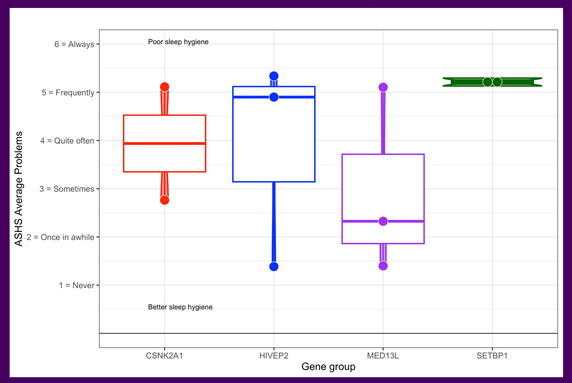
	CSNK2A1	HIVEP2	MED13L	SETBP1
N CSHQ ASHS	6 3 F : 1 M 2 F : 0 M *5 additional with no sleep data*	6 2 F : 1 M 0 F : 3 M *3 additional with no sleep data*	10 4 F : 3 M 2 F : 1 M *4 additional with no sleep data*	4 1 F : 1 M 0 F : 2 M *3 additional with no sleep data*
Clinical symptoms	• Intellectual disability • ASD • ADHD • Learning disability • Communication disorders • Tic disorders • Motor disorders	• Intellectual disability • ASD • Hyperactivity • Anxiety • Abnormal brain structure • Impaired speech • Hypotonia • Facial dysmorphisms	• Intellectual disability • Impaired speech • Facial dysmorphisms	• Intellectual disability • Hyperactivity • Attention or concentration deficits • Impaired speech • Hypotonia • Motor development delay • Vision impairment
Gene Function	• Casein kinase: controls cell cycle, & circadian rhythms • Wnt signaling pathway (*Casein kinase*)	• Enables DNA binding (Zinc-finger containing transcription factors) • Brain growth and development (*Hivep2 Gene: Medlineplus genetics*)	• Enables DNA binding (Transcription factor: RNA polymerase) • Brain growth and development (*MED13L gene: Medlineplus Genetics*)	• Enables DNA binding (protein binding) (*SETBP1 gene: Medlineplus genetics*)

Child Sleep Habits Questionnaire



Group differences:
$F(3,13) = 1.24$, $p = .34$

Adolescent Sleep Hygiene Scale



Group differences:
$F(3,6) = .63$, $p = .62$