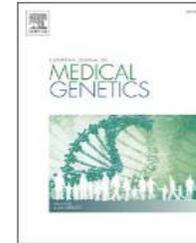




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Consensus recommendations on sleeping problems in Phelan-McDermid syndrome

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PD Dr. Sarah Jesse



What is normal sleep ?

Young babies will be expected to spend most of the day sleeping

~ 10–12 weeks of age, sleep will naturally become more nocturnal

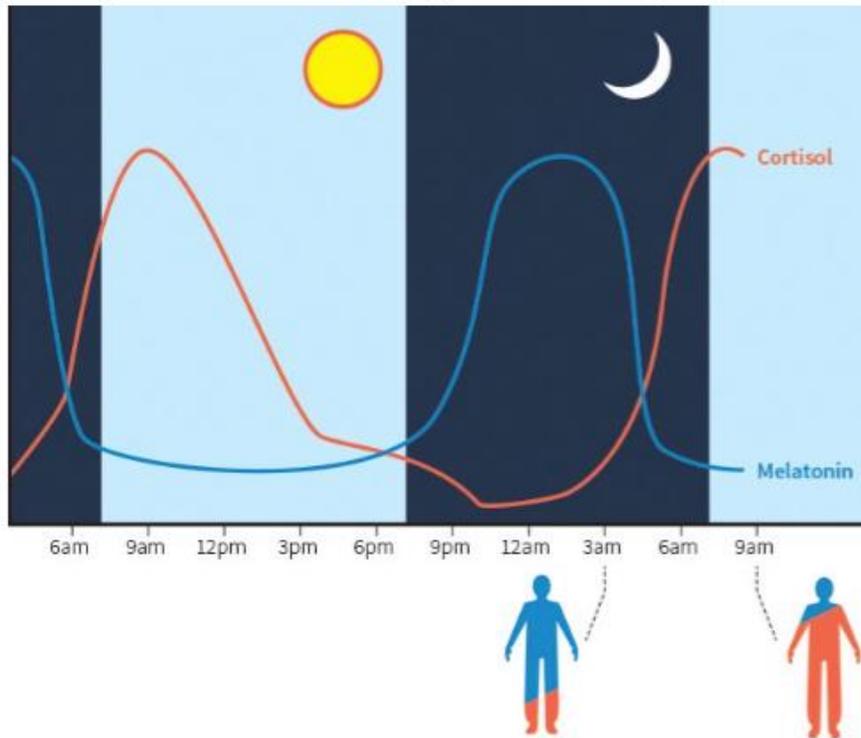
Until the age of 4, day napping will occur to achieve the needed sleep time

By the age of 5, day and night-time sleeping begins to decrease

Galland et al. Normal sleep patterns in infants and children: a systematic review of observational studies
Sleep Rev Med 2012 Jun;16(3):213-22.

What happens in sleep ?

Figure 1: Variation in melatonin and cortisol throughout the day



The guideline

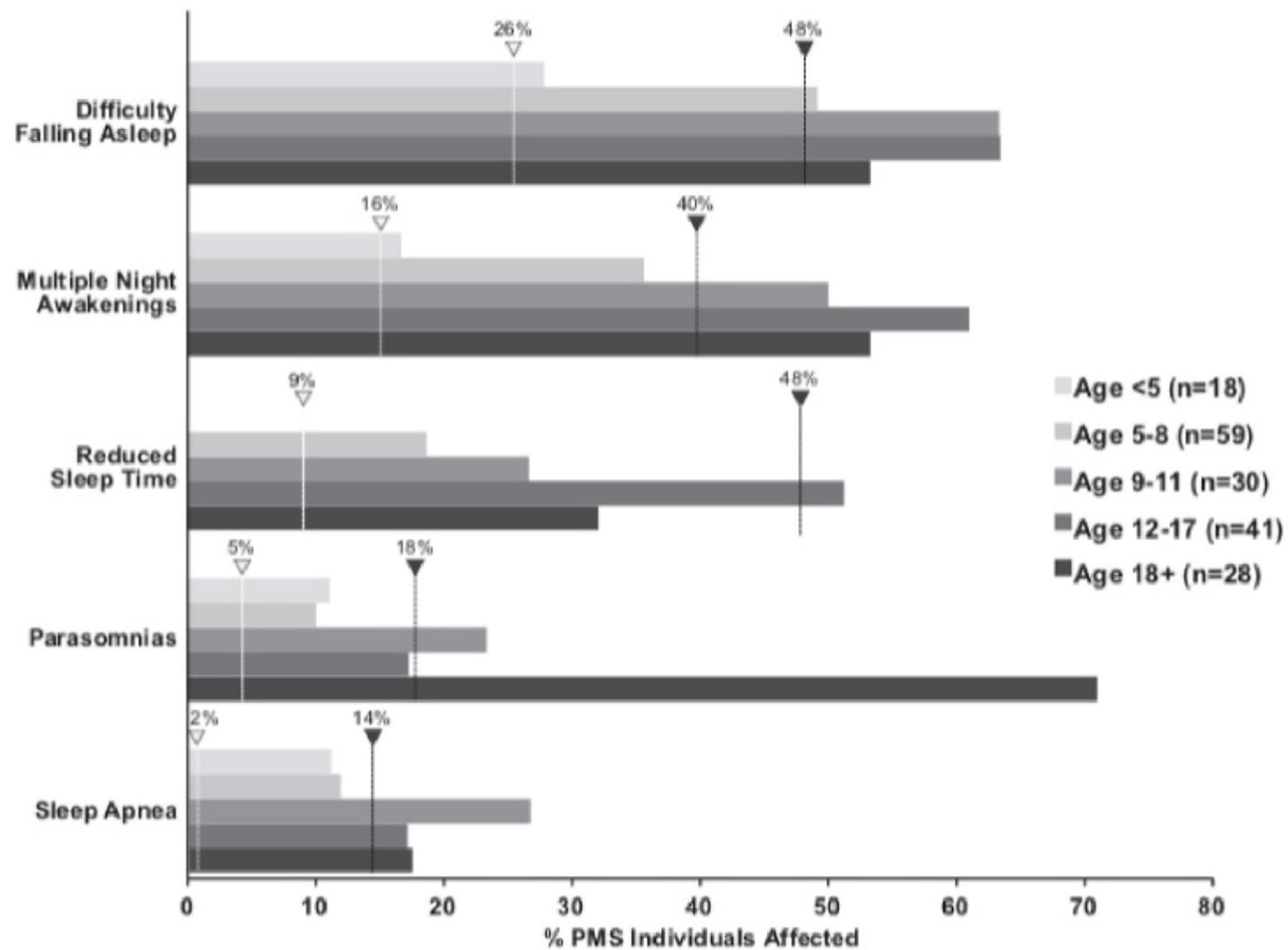
1. in-depth **literature search** found only four articles specifically focused on sleeping problems in PMS
 - prevalence rates ranged between 24% and 46%

2. a **world-wide survey** was created and distributed amongst parents
 - survey reported a prevalence of 59%

Landlust et al. Parental perspectives on Phelan-McDermid syndrome: Results of a worldwide survey
Europ J Med Gen 2023 Jul;66(7):104771

types of sleep disorder

difficulty falling asleep
numerous night awakenings
being restless in sleep
night-time incontinence
tooth grinding
sleep anxiety
parasomnias



Comorbidities and associations

- significant association between epilepsy and sleeping problems: 73% of individuals with epilepsy had sleeping problems compared to 54% of individuals without epilepsy

Landlust et al. Parental perspectives on Phelan-McDermid syndrome: Results of a worldwide survey
Europ J Med Gen 2023 Jul;66(7):104771

- somatic conditions like reflux, diabetes, asthma, pain or rheumatic disorders have been associated with sleep problems in the general population

Lazaratou et al. Medical comorbidity of sleep disorders in children and adolescents. Curr. Opin. Psychiatr. 25 (5), 391–397

- bipolar disorder, manic episodes, or mood dysregulation

Kohlenberg et al. Psychiatric illness and regression in individuals with Phelan-McDermid syndrome. 2020 J. Neurodev. Disord. 12 (1), 7

- Medication for comorbidities

Van Balkom et al. Consensus recommendations on mental health issues in Phelan-McDermid syndrome. Europ J Med Gen 2023

an increase in sleep disorders in children correlates with an increase in
sleep disorders and daytime sleepiness in parents/caregivers

Boergers et al. Child sleep disorders: associations with parental sleep duration and daytime sleepiness
J Fam Psychol 2007 Mar;21(1):88-94

Assessment of sleep problems

keeping a sleep diary for a minimum of 2 weeks

- Children's Sleep Habits Questionnaire up to 11 years of age
- Sleep Disturbances Scale for Children 6–15 years of age
 - Epworth Sleepiness Scale for adults

Non-pharmacological options

Environmental, sensory, and behavioral modifications

have an active, varied day with sufficient daylight
phase out mental and physical activities in the evening,

Create a consistent bedtime routine

Keeping regular bedtimes and meals can also help in establishing routines

use the same objects, photos, or pictures

fixed and appropriate bedtime

use a dark, quiet non-stimulating sleeping environment and dimmed nightlight

Light therapy (minimum 3000 lux, minimum 15 minutes) can be

used to influence the body's own melatonin production

Non-pharmacological options

Investigate possible (neuro)physiological reasons

Medication for somatic or psychiatric comorbidities can cause
or enhance sleep problems

Ruling out somatic causes

rule out restless legs syndrome

serum ferritin levels should be checked

levels that are too low (<50 ng/ml) should be treated with iron
supplementation for at least 3 months

Some epileptiform discharges that may only happen at night

Non-pharmacological options

Investigate possible mental health issues

Rule out psychiatric causes like manic episodes or hyperactive

Catatonia

Investigate changes in mood, such as anxieties, depression, etc.

attention deficit hyperactivity disorder (ADHD)

Pharmacological treatment

Melatonin is generally the most frequently prescribed sleep medication

Clonidine

sedating antidepressants

atypical neuroleptics

antihistamines

Bruni et al. Pharmacotherapeutic management of sleep disorders in children with neurodevelopmental disorders.
2019 Expert Opin. Pharmacother. 20 (18), 2257–2271

Summary



Every individual with PMS and sleep problems should be **evaluated for somatic** and/or **environmental** and/or **neuropsychiatric causes**

Mental health **conditions co-occurring** with sleep problems in individuals with PMS **need to be investigated and treated**

In individuals with PMS with sleep problems, sleep hygiene should be evaluated, and caregivers should be supported in establishing a structured approach (**behavioral interventions**)

If sleep problems persist despite appropriate interventions, the individual should be **referred to a specialist** experienced in sleep problems