

# THE CLINICAL AND LABORATORY ASSESSMENT OF CHILDHOOD SLEEP-WAKE PROBLEMS

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## Introduction

The evaluation of childhood sleep-wake problems requires the sleep history, a focused medical examination, and sleep laboratory testing when indicated. There are continuous maturational changes in sleep from infancy through adolescence; hence the need to be familiar with the age-related normal values of the relevant physiologic parameters.

## History

It is useful to guide the parent or patient through events and activities from the time of awakening through the night. Teens with depression, anxiety or delayed sleep phase syndrome may exhibit difficulty awakening in the morning. Daytime sleepiness may appear under the guise of “fatigue,” inattentiveness, hyperactivity, frank napping, automatic behaviors, or decline in academic performance. Cataplexy consists of episodes of muscle weakness that are triggered by laughter, surprise, or the anticipation of reward. Cataplexy can be fairly subtle, characterized only by transient mouth opening or a head roll. One needs to inquire about the frequency of physical exercise, significant weight gain, medications that impact sleep (prescription or over-the-counter), use of caffeine, alcohol and illicit drugs. Relevant factors around bedtime include the sleeping environment, use of electronic devices, urge to move the limbs as in restless legs syndrome, intrusive thoughts or worries and obsessive compulsive behaviors. After sleep onset, information about snoring, bedwetting, restless sleep, night awakenings, nocturnal events, etc. becomes relevant. One also needs to also inquire if the sleep problem is affecting the quality of life, about psychosocial issues, and family dynamics.

## The Sleep Related Examination

Components of the general physical, respiratory, and nose and throat examinations are utilized. The patient is examined for craniofacial anomalies such as maxillary hypoplasia (Down syndrome, Crouzon syndrome), mid-face hypoplasia, micrognathia (Pierre Robin sequence), macroglossia (Down syndrome, mucopolysaccharidoses) and the neurological exam become relevant. The height, weight, body mass index, and blood pressure are important in obstructive sleep apnea (OSA) and narcolepsy. Deviated nasal septum, swollen inferior turbinates, tonsillar hypertrophy and mouth breathing may accompany OSA. The Chiari type II malformation can be associated with snoring, hoarseness of the voice, decreased gag reflex, and altitudinal changes in the amplitude of tendon reflexes. Neuromuscular disorders like myotonic dystrophy, congenital muscular dystrophy, congenital non-progressive myopathies, and spinal muscular atrophy show chronic obstructive hypoventilation due to a combination of pharyngeal muscle and chest wall weakness. The kyphoscoliosis that commonly accompanies neuromuscular disorders can also restrict chest and abdominal movement, with consequent hypercarbia and early morning headache and daytime somnolence. For the assessment of episodic phenomena like cataplexy and parasomnias, a review of home video clips can be priceless.

## **Common Survey Instruments for Sleep-Wake Function**

### *1. Sleep Diary*

This is maintained by the parent or patient. It records sleep-wake function from over the preceding 2-3 weeks. Recording of bed onset time (week nights and weekends), sleep onset time, the number of night awakenings and their length, morning wake-up times on school days and on weekends, frequency and length of day time naps, how alert one feels during the day (on a 1-5 sliding scale), frequency of exercise, and the use of caffeinated beverages and medications is documented. Several examples of sleep diaries are available on the Internet.

### *2. Epworth Sleepiness Scale*

The Epworth Sleepiness Scale (ESS) is a widely utilized, self-administered questionnaire for older teens and adults. Eight questions estimate the chances of dozing off on a 0-3 scale during common activities such as while sitting and reading, watching television, sitting inactively in a public place, as a passenger in a car, lying down in the afternoon, sitting and talking to somebody, after lunch and while driving after stopping for a few minutes in a traffic light. The maximum score is 24, values < 10 are physiologic, and those above 12 correlate with pathologic daytime sleepiness. As the conventional ESS contains questions pertaining to the use of alcohol or driving, a modified pediatric version of ESS is utilized.

### *3. Children's Sleep Habits Questionnaire (CSHQ)*

This parent questionnaire is designed for use in children aged 4 to 12 years. There are 45 questions pertaining to eight domains of sleep-wake function: bedtime resistance, sleep onset delay, sleep duration, sleep anxiety, night awakenings, parasomnias, breathing disturbance, and daytime sleepiness. Responses are rated as *rarely*, *sometimes*, or *usually*. The higher the score, the more disturbed the sleep: scores of 41 or greater correlate with presence of a sleep disorder. Shortcomings of the CSHQ include its retrospective nature and bias introduced by parental proxy. Nevertheless, it is widely used, with the total score providing a reliable cutoff for presence or absence of a sleep disorder.

## **Diagnostic Tests**

### *Actigraphy*

The actigraph is a relatively inexpensive, wrist watch-shaped device for sleep-wake assessment in the home environment over a 1-2 week period. It is conventionally strapped to the left wrist. It works on the premise that muscle activity is frequent during wakefulness and infrequent in sleep. The device measures linear acceleration from muscle activity and translates physical motion into a numeric representation that is sampled frequently, e.g. every 0.1 second, and is aggregated at a constant interval or epoch, to be then displayed graphically. Other wearable devices like the *Fit Bit* work on a similar principle. The total time in bed, total sleep time, number of night awakenings, and sleep efficiency can be reliably estimated. Wrist actigraphy is useful for assessing insomnia, circadian rhythm sleep disorders, daytime sleepiness, and restless legs syndrome. It provides longitudinal, semi-quantitative information about sleep in the home environment. It is unable to distinguish wakefulness from sleep when the patient is awake and immobile. Further, actigraphy is also not able to distinguish between the various stages of sleep.

### *Overnight oximetry*

Overnight oximetry is a screening tool for sleep disordered breathing as apneas and hypopneas may be associated with drops in oxygen saturation. The mean value of the lowest overnight oxygen saturation in healthy children is 92%. There is no difference in saturation values obtained from applying the sensors over the ear or the finger tip, but recorded values may be lower from over the toes. Merits of oximetry include its low cost, non-invasive nature and applicability in the home environment. A disadvantage is that it is relatively insensitive, being abnormal only in about 25 % of patients with OSA. When the oximetry study is abnormal, the positive predictive value for OSA is 97%. A normal oximetry test does not however exclude the possibility of mild or moderate OSA.

### *Nocturnal polysomnography (PSG)*

This is the “gold standard” test that requires simultaneous monitoring of multiple physiological parameters in the sleep laboratory, such as EEG, eye movements, chin and leg electromyogram, oronasal airflow, nasal pressure, ECG, oxygen saturation, and end tidal CO<sub>2</sub>. PSG is indicated for assessing disorders of vigilance such as narcolepsy and idiopathic hypersomnia, OSA, hypoventilation syndromes, and central sleep apnea. In disorders of excessive sleepiness such as narcolepsy, the PSG helps exclude other disorders that may lead to sleepiness, such as OSA. Patients with narcolepsy generally show sleep-onset REM periods (SOREMP), which are defined as REM sleep within 15 minutes of sleep onset. Another finding in narcolepsy-cataplexy is the persistence of muscle tone during REM sleep (REM sleep without atonia). PSG may also be utilized for investigating nocturnal spells such as seizures and parasomnias. Patients with obesity, neuromuscular disorders, Down syndrome and Prader Willi syndrome can manifest hypoventilation, defined as end tidal > 50 mm of mercury for > 25% of the total sleep time. The merits of PSG include its reliability and in-depth information. Disadvantages include its expense, need for testing in the sleep laboratory environment which may be inconvenient for children and parents.

### *Multiple Sleep Latency Test (MSLT)*

When used in conjunction with the PSG, the MSLT is the “gold standard” for the assessing daytime sleepiness. The strengths of the test lie in its intuitive design (sleepy individuals are likely to fall asleep more quickly than those who are not sleepy), its reliability, and the availability of normative data across various ages. It is indicated in the investigation of suspected narcolepsy or idiopathic hypersomnia. A PSG is conducted the night before the MSLT in order to exclude confounding disorders like OSA. The MSLT is valid in children older than 5 years. Medications that impact sleep such as benzodiazepines, psycho-stimulants, antidepressants and barbiturates are stopped two weeks prior to the MSLT. The test consists of the provision of 4-5 nap opportunities, each of 20-minute length at two hourly intervals, e.g. 0900 hours, 1100 hours, 1300 hours and 1500 hours. The test measures the speed with which the patient falls asleep, and also the nature of the transition from wakefulness to sleep, whether it is into REM or NREM sleep. The time interval between “lights out” and electroencephalographic sleep onset is designated the sleep latency. In narcolepsy, the mean sleep latency derived from averaging the sleep latency of the 4-5 nap opportunities is < 8 minutes, and there is REM-onset sleep in 2 or more naps. The diagnostic sensitivity of the MSLT for the diagnosis of narcolepsy has been estimated around 61% while the diagnostic specificity when 2 or more SOREMPs are present is around 94%.

## Suggested Reading

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