

Annotation

Investigation of sleep disorders

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Abstract: Polysomnography or sleep study is the main investigation for paediatric sleep disorders. It involves the continuous and simultaneous recording of multiple physiological parameters evaluating sleep and respiration. It is most commonly used to diagnose obstructive sleep apnoea and to monitor nocturnal non-invasive ventilation requirements of children. Its role in other sleep related breathing disorders, narcolepsy and parasomnias is discussed.

Key words: excessive daytime sleepiness; hypoventilation; obstructive sleep apnoea; polysomnography; primary snoring; rapid eye movement sleep; sleep study.

Concern about a child's sleep pattern occurs in about one third of all families and in nearly two thirds of families with children who have developmental disabilities.¹ The majority of sleep disorders can be diagnosed by a thorough sleep history and sleep diary in addition to a routine paediatric history and physical examination and do not require investigations. Polysomnography (PSG) or sleep study is the most important investigation for sleep disorders, and is most commonly used for identification of obstructive sleep apnoea (OSA). However PSG should also be considered when symptoms or signs are suggestive of sleep-disordered breathing, excessive daytime sleepiness or unexplained nighttime wakings.

WHAT IS A SLEEP STUDY?

A sleep study involves the continuous and simultaneous recording of multiple physiological parameters evaluating sleep and respiration. Sleep is assessed by the electroencephalogram (EEG), electrooculogram (EOG), and electromyogram (EMG). Respiration is assessed by measuring nasal and oral air flow, respiratory effort (by movement transducers on chest and abdomen, and/or respiratory muscle EMG), arterial oxygen (by oximetry), and carbon dioxide (by transcutaneous and/or end tidal CO₂). An electrocardiogram is recorded. Other parameters that can be monitored are body position, leg movements (by anterior tibialis surface EMG) and snoring volume. The patient may be monitored by continuous infrared or low-light video.

Age should be no barrier to undergoing a sleep study if there are appropriately trained paediatric staff in a child friendly laboratory and a parent participates. There is little clinically significant night-to-night variability with paediatric PSG and thus a single night should be sufficient for diagnosis of significant sleep disordered breathing in children.² The PSG should be performed without sedation or sleep deprivation.³

A paediatric sleep study is labour-intensive, in both the acquisition and interpretation of the data. It can take one trained staff member up to an hour to place all the electrodes on a

cooperative child. The leads need to be connected to amplifiers and signal quality checked. Both the child and the physiological data being recorded need to be closely monitored throughout the night, so that signals which are lost or are of poor quality because of electrode displacement or interference can be corrected. The development of digital polygraphs has helped in the storage and display of data but as yet, in paediatrics, there is no reliable computer-aided analysis. Analysis is still performed manually by visual interpretation of all the physiological parameters which adds considerably to the time taken to report individual studies.

WHAT INFORMATION DOES A SLEEP STUDY PROVIDE?

Sleep is made up of two sleep states defined as rapid eye movement (REM) and non-REM (NREM) sleep. The PSG identifies the sleep states throughout the night (sleep architecture) and shows the frequency and aetiology of sleep disruption (sleep quality).

Respiratory physiology changes during sleep. REM sleep is associated with generalized muscle hypotonia, and breathing becomes dependent on the diaphragm due to inhibition of the intercostal and accessory muscles. The hypoxic and hypercapnic ventilatory drives are decreased in REM compared to NREM sleep and the awake state. Thus REM sleep provides a 'physiological stress' which can unmask potential respiratory difficulties that may not be apparent when the child is awake. As most REM sleep occurs in the second half of the night, the parents may not have observed these difficulties. It is important to note that children also have a greater proportion of REM sleep compared to adults.

Respiratory events can be defined as obstructive or central. In children an *obstructive apnoea* is defined as cessation of oronasal airflow in the presence of continuing respiratory effort.³ (Fig. 1) A partial obstruction or *hypopnoea* is defined as a 50% or greater decrease in the amplitude of the oronasal airflow accompanied by hypoxaemia and/or an

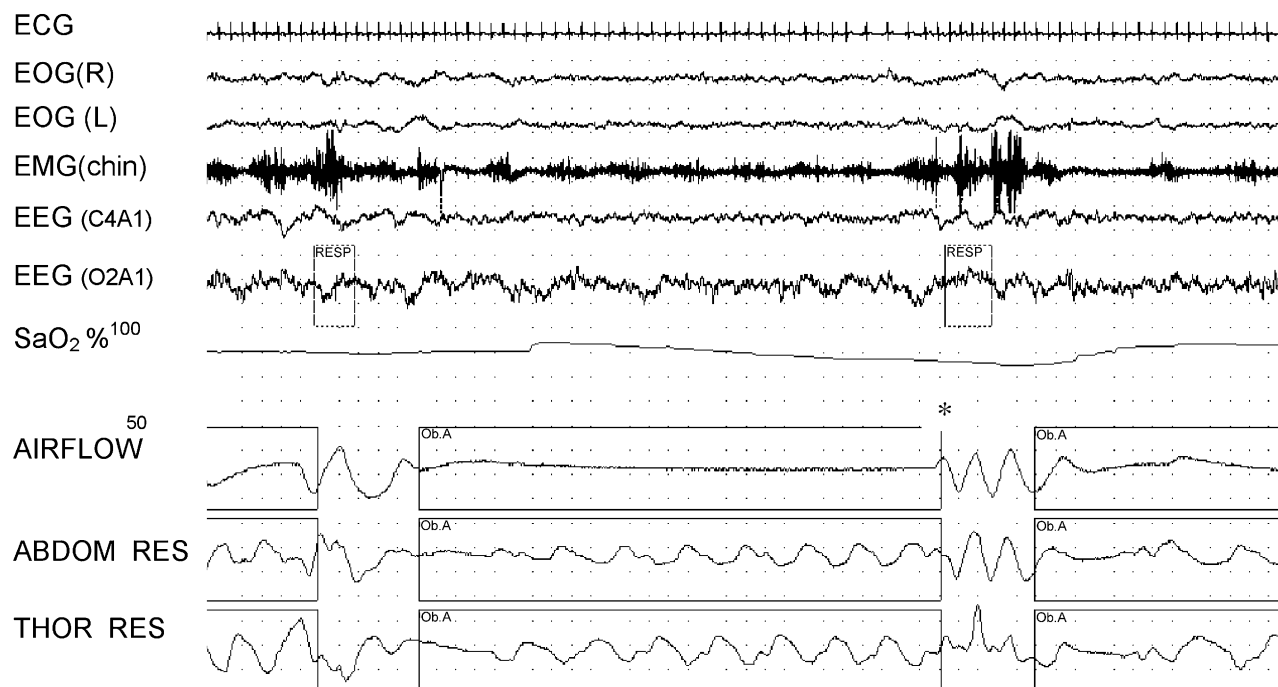


Fig. 1 Obstructive apnoeas in a child during rapid eye movement (REM) sleep where there is absent oronasal airflow and continued respiratory effort with paradoxical inward chest movement during inspiration. Inspiration is the upward deflection on the airflow trace. These events are accompanied by oxygen desaturation and an arousal as seen in both the ECG (increase in heart rate) and the EMG (increase in amplitude) channels. Time base 1 min.*Obstructive apnoea 28.4 s in length resulting in a 13% drop in SpO₂–82%. ABDO, abdominal movements; ECG, electrocardiogram; EEG, electroencephalogram central and occipital; EMG, chin submental electromyogram; EOG electrooculogram; airflow detected by thermistor; THOR, thoracic movements.

arousal.³ In children hypopnoeas are common and frank obstructive apnoeas rare. Most obstructive apnoeas, hypopnoeas and hypoventilation occur during REM sleep.⁴ A *central apnoea* is defined as absence of oronasal airflow with cessation of all respiratory effort.³ (Fig. 2) Central apnoeas are found in all normal children but are generally less than 20 s duration, and may be associated with transient oxygen desaturations. Some respiratory events contain both central and obstructive components and are referred to as *mixed apnoeas*. Mixed apnoeas are more common in younger children. Unlike adults, where 10 s is the minimum duration of events scored, the length of a scored respiratory event in children is equivalent to two respiratory cycles. This takes into account the change in respiratory rates with increasing age. Children often desaturate with apnoeas less than 10 s in length as they have a lower functional residual capacity and faster respiratory rates than adults.³

SLEEP RELATED BREATHING DISORDERS

Obstructive sleep apnoea

Obstructive sleep apnoea (OSA) in children is characterized by recurrent events of partial or complete upper airway obstruction during sleep, which disrupt normal ventilation and sleep patterns.³ This leads to hypoxaemia, hypercarbia and sleep disturbance. Morbidity of untreated OSA includes growth failure⁵ pulmonary hypertension and cor pulmonale and neurocognitive sequelae.^{6,7} Over the last decade there has been increasing interest in the effects of milder OSA in children, and its

deleterious effects on learning, behaviour and neurocognitive function including attention, memory and executive function.⁸ These are thought to be a function of the intermittent hypoxia and sleep fragmentation seen in OSA. Snoring is the most common night-time symptom in children with OSA. Children who snore but who do not have associated ventilatory abnormalities or sleep disturbance have *primary snoring*. As the first-line treatment for uncomplicated OSA is adenotonsillectomy^{7,9} it is important to differentiate between these two groups of children.

It is estimated that 10% of children have primary snoring and another 1–3% have OSA, with equal sex distribution.¹⁰ The peak incidence of OSA is at 2–6 years due to large adenoids and tonsils relative to the upper airway size. Symptoms include snoring, difficulty breathing, apnoeas, mouth breathing, excessive sweating and restless or disturbed sleep. Daytime symptoms include irritability, poor concentration, hyperactivity, and less commonly, excessive daytime sleepiness and failure to thrive. It is difficult from the parental history or questionnaire responses to diagnose those children who have OSA rather than primary snoring.¹¹ Physical examination can be suggestive but not diagnostic of OSA. Certain groups of children are at increased risk of developing OSA. These include children with problems interfering with the size of the upper airway such as maxillo-mandibular skeletal deformities (mid-face hypoplasia syndromes, Pierre Robin syndrome); metabolic or genetic defects leading to macroglossia or soft tissue deposition (Down syndrome, mucopolysaccharide storage diseases); neurological conditions and syndromes causing hypotonia, hypertonia and muscle weakness (neuromuscular diseases, cerebral palsy, Prader-Willi syndrome); and obesity.

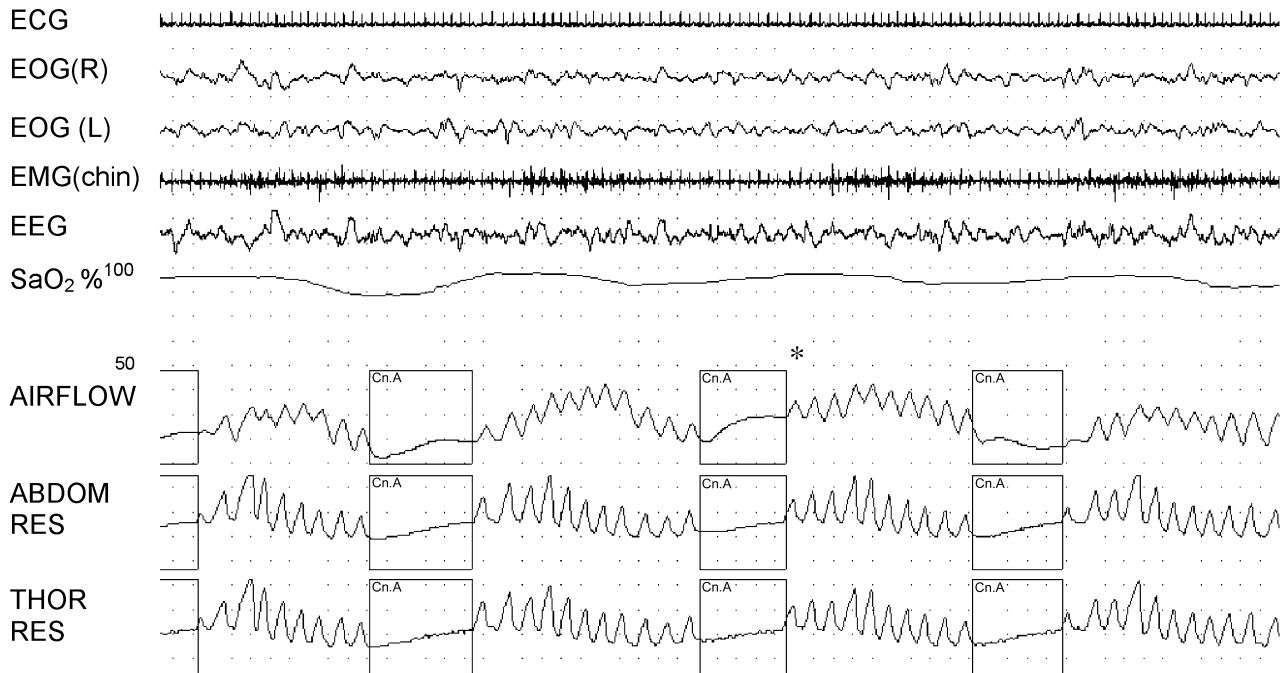


Fig. 2 Section from an infant sleep study showing recurrent central apnoeas where there is loss of oronasal airflow accompanied by absent respiratory effort and oxygen desaturation. Time base 1 min*. Central apnoea 6 s in length resulting in a 6% drop in SpO₂–91%. ABDO, abdominal movements; ECG, electrocardiogram; EEG, electroencephalogram central and occipital; EMG, chin submental electromyogram; EOG electro-oculogram; airflow detected by thermistor; THOR, thoracic movements.

Various screening tools have been evaluated including audiotapes, nocturnal videos and pulse oximetry. Oximetry has been found to be the most useful with one study showing that abnormal nocturnal oximetry in a child suspected of having OSA has a positive predictive value of 97%, but a negative predictive value of only 47%.¹² Oximetry is diagnostic if positive, but, if negative, does not exclude OSA so a PSG is required.¹² Oximetry does not assess the severity of OSA and provides no information on the type of events associated with oxygen desaturations, or on obstructive hypoventilation, work of breathing or sleep disruption. However, given that paediatric sleep studies are limited to large teaching hospitals and that there may be delays in accessing services, a normal oximetry while not excluding OSA, may reassure the clinician and parents that the child is unlikely to have severe OSA, while waiting for further assessment.

In summary, the history and examination are poor at discriminating between primary snoring and OSA. Screening tests such as oximetry are diagnostic if positive but not if negative. At present the only diagnostic test is PSG. PSG should be used in children less than 3 years of age, in children with significant clinical sequelae of OSA, in high risk groups, and in all children when the history is unclear. Severe OSA on PSG has been shown to be a risk factor for post operative respiratory complications following adenotonsillectomy.¹³

Ventilation requirements

Polysomnography should be performed annually in children requiring nocturnal ventilation. PSG monitors nocturnal ventilation requirements that may change with growth or disease

progression, and ensures normalization of sleep architecture and sleep quality. Increasingly non-invasive ventilation by either continuous positive airway pressure (CPAP) or bi-level positive airway pressure (biPAP) is being used to treat children who require nocturnal ventilatory support. In children with OSA, when surgery is contra-indicated, or when there are persistent symptoms despite surgery, non-invasive mask ventilation by either CPAP biPAP is the next treatment option.^{14,15}

Neuromuscular disease

Children with neuromuscular disease are at high risk of developing both central and obstructive apnoea due to a combination of respiratory muscle weakness, impairment of central ventilatory control, and decreased upper airway tone. During REM sleep with its associated muscle hypotonia and decrease in ventilation, this group is further at risk of nocturnal hypoventilation. Nocturnal hypoventilation may be exacerbated by increasing muscle fatigue as the night progresses, along with intercurrent upper respiratory tract infections.

Symptoms of nocturnal respiratory failure can be broad and include daytime sleepiness, fatigue, morning headache, daytime behavioral changes, difficulty sleeping and the need for frequent repositioning during the night.¹⁶ Inadequate nocturnal ventilation can occur despite normal daytime respiratory function. PSG helps to define whether abnormal night-time ventilation and associated sleep disruption are contributing to impaired daytime functioning, as compared to progression of the underlying disease process. Treatment is with nocturnal ventilation.

Central hypoventilation

Central hypoventilation syndrome (CHS) is characterized by adequate ventilation during wakefulness and hypoventilation during sleep. CHS is due to deficient respiratory control centres in the brainstem which affect automatic respiratory control when the patient is asleep. CHS may be congenital or acquired, the latter seen with trauma, infection, asphyxia, infarction or tumour.¹⁷ A transient form may be seen with severe obstructive sleep apnoea. Children with CHS have absent ventilatory responses to hypercarbia and variable responses to hypoxia when asleep. The PSG reveals shallow breathing (reduced tidal volume) and normal respiratory and heart rates despite significant hypoxia and hypercarbia. Initially this is evident during NREM sleep but if the condition progresses these changes are seen during REM sleep and may occur during wakefulness.¹⁸ Treatment is with nocturnal ventilation.

OTHER INDICATIONS FOR SLEEP STUDY

Narcolepsy and excessive daytime sleepiness

Excessive daytime sleepiness (EDS) has multiple aetiologies including insufficient sleep, sleep disruption (OSA, nocturnal seizures), structural brain lesions, depression, medications and drugs. Narcolepsy is a primary disorder of REM sleep regulation with EDS as its main symptom. The other main symptom is cataplexy which is the sudden onset of muscular weakness brought on by excitement or emotion and occurs in 70% of adults with narcolepsy.¹⁹ Sleep paralysis and hypnagogic hallucinations, along with EDS and cataplexy make up the classic narcolepsy tetrad. Although uncommon, it is not rare, with prevalence 2–6 per 10 000 and a peak onset in adolescence.¹⁹

Diagnosis of narcolepsy can be difficult with reported delays of up to 15 years from onset of symptoms to diagnosis.¹⁹ A careful history, along with human leucocyte antigen (HLA) typing and PSG may enable early diagnosis. There is an association with the human leukocyte antigen HLA DR2, and a tighter association with HLA-DQB1*0602, with over 90% of all narcoleptic patients positive for this marker.²⁰ However, around 25% of the normal population carry this marker and do not develop narcolepsy. The diagnostic test for narcolepsy is overnight PSG and a multiple sleep latency test (MSLT). The MSLT is a standardized objective measure of daytime sleepiness which consists of five scheduled naps every 2 h on the day following the PSG.²¹ In narcolepsy the PSG shows reduced time to fall asleep and enter REM sleep (reduced sleep latency and reduced REM sleep latency). A positive MSLT shows a mean sleep latency of less than 5 min, with two or more REM sleep onsets.^{19,21} The MSLT has a sensitivity of 70% and a specificity of 97%.²² Objectively assessing daytime sleepiness in a young child is difficult and the MSLT has only been standardized for children aged 8 years or older.²¹

Parasomnias

Parasomnias are the disruptive behaviours that disturb sleep. In children the most frequent parasomnias are those associated with impaired arousal from deep sleep (stage 3 and 4 NREM sleep) and include confusional arousals, sleep walking and night terrors. These events all share common characteristics of skeletal and autonomic nervous system activation, confusion and disorientation and no memory of the event the next day.²³ As these events occur in healthy children and disappear with

adolescence they are regarded as benign. History and examination are usually sufficient to make the diagnosis. PSG is indicated if events are atypical, violent or result in significant injury, or there is significant night-time disruption and daytime sleepiness.²⁴ The most common differential diagnosis is epilepsy, but OSA is occasionally identified as a precipitant of frequent recurrent events.²⁵ If seizure activity is suspected, a full EEG montage of 16 pairs of electrodes should be used rather than the three pairs used to stage sleep. In frontal lobe epilepsy, the ictal and interictal EEG may be normal, along with the PSG, so prolonged video-EEG should be pursued if the history is suggestive.²⁶

CONCLUSION

Polysomnography is currently the main investigation used to evaluate paediatric sleep disorders, the majority of which are due to OSA. The use of PSG in paediatrics is limited by its expense and the small number of paediatric sleep laboratories. OSA in children is common and associated with significant morbidity. Over the last decade research suggests that children at the milder end of the spectrum of OSA have impairment of neurocognitive function which is reversible with adenotonsillectomy.²⁷ Given the inability of the history and examination to distinguish between primary snoring and OSA¹¹ and screening tests such as oximetry being helpful only if positive¹² the challenge for the future is to develop diagnostic tests that are more portable and cheaper than PSG to identify children with sleep disordered breathing. However, at this point in time, PSG remains the 'gold standard' in diagnosing OSA and nocturnal hypoventilation in children and should be used routinely to monitor nocturnal ventilatory requirements in children. It is also used in the assessment of non-respiratory related sleep disorders such as narcolepsy and seizures.

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