The Scope of Paediatric Sleep Medicine

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Abstract

Despite apparent similarities to adult sleep medicine, the disorders of paediatric sleep medicine have a distinct epidemiology and pathophysiology. During childhood, the physiology of sleep develops and matures, resulting in changing patterns of normal behaviours and of sleep disorders. Through a fictional case scenario, this article aims to convey the range and complexity of disorders that may be encountered and the various investigations and treatments available to the paediatric sleep physician.

Key words: Child, Circadian rhythm, Neuromuscular diseases, Obstructive sleep apnoea, Restless leg syndrome

Introduction

Despite apparent similarities to adult sleep medicine, the disorders of paediatric sleep medicine have a distinct epidemiology and pathophysiology. During childhood, the physiology of sleep develops and matures, resulting in changing patterns of normal behaviours and of sleep disorders. An awareness of these changing patterns is essential in reviewing paediatric patients with sleep problems. Although it is not possible to completely review the scope of paediatric sleep medicine, this article aims to convey the range and complexity of disorders that may be encountered through a fictional case scenario.

Case Scenario

A 12-year-old boy presented to a paediatric clinic with a history of snoring, some pauses in his breathing, restless sleep and complains of cramps and an urge to move his legs at night. He goes to bed at 9 pm but has difficulty falling asleep until 11 pm. He reads, text-messages his friends and listens to music in bed. He has to wake up for school at 6 am and feels very tired and unrefreshed. At school his teachers are concerned that his academic performance seems to be less than his potential. They complain that he looks tired in class, fidgets, talks out of turn, is impulsive at times and seems to have difficulty sitting still. He is also distractible and seems very forgetful. On the weekends he sleeps in till 11 am given the chance.

Discussion

Perhaps 20 years ago an assessment by a clinician might formulate the following problems in this boy: attention deficit disorder, leaning disability, behavioural sleep problems, “typical teenager”, “growing pains” and so forth.

The first sleep studies (polysomnography or PSG) in children in Australia were performed in the early 1990s. Since then there has been a rapid increase in the number of sleep studies.1,2 This parallels an increasing understanding of the role of sleep and sleep disorders in child health and disease.

Development of Sleep and Circadian Rhythms

The normal circadian rhythm of waking during the daytime and sleeping at night is governed by neurohormonal influences and can be entrained by light and external stimuli.3 These processes are also affected by bedtime routines and meals.3 The amount of sleep and the proportions of the different stages of sleep vary across childhood. Newborn infants sleep more than 16 hours per 24 hours with more than 50% of total sleep being comprised of rapid eye movement (REM) sleep.4 The proportion of REM sleep and slow wave sleep gradually decrease with age. Prior to 4 months of age, infants do not have a natural circadian rhythm and sleep is spaced throughout the day.5 Infants gradually consolidate sleep into a long overnight sleep with shorter daytime sleeps. By 18 months most infants are sleeping 11.5 hours overnight with one to two 1-hour to 1.5-hour naps in the day.3,6
The process of consolidation continues into childhood with most children dropping the last daytime nap by 5 years of age, except in cultures where a “siesta” is culturally normal. There is a wide range of reported normal sleep at each age and individual children may vary by 1 to 2 hours around the mean for age. There is also evidence in Australia that the duration of sleep in school-aged children has declined by around 30 minutes over the last generation. Many children do not receive the required amount of sleep, particularly in adolescence, resulting in decreased academic performance. This has been related to extra curricular activities, increased homework, computer activity and television viewing but is also dependent on individual cultural and home environment influences.

**Sleeplessness in Normal Children**

Sleep problems are common in childhood with a reported prevalence in community surveys of 25% to 50% with bedtime resistance being the most commonly reported problem in young children. Undesired bedtime behaviours cannot be addressed in isolation to problems of daytime behaviour and limit setting and usually both need to be managed simultaneously.

Nocturnal sleeplessness in children and adolescents can be divided into sleep-onset insomnia, sleep maintenance insomnia, and sleep phase delay. In young children, the terms bedtime problems, night-time fears, and night-wakings are often used. Sleep-onset insomnia, manifested as bedtime resistance, is generally a behavioural or limit-setting disorder. Inappropriate sleep onset associations such as bedtime bottles, music, computers, or television viewing can make it more difficult rather than easier to fall asleep. Managing bedtime problems includes the removal of stimulating activities and associations in the hour prior to sleep.

Sleep maintenance problems or night-waking may be a consequence of sleep onset difficulties because children wake several times in the night and require the same sleep associations to settle themselves. Sleep maintenance problems in the absence of sleep onset difficulties are of more concern as this indicates that something is waking the child. This is likely to be a medical problem and a careful assessment should be made for causes of night waking such as itch from eczema, discomfort from gastro-oesophageal reflux, periodic limb movements (PLMs) or obstructive sleep apnoea (OSA).

In our case scenario, there is an element of sleep phase delay. Adolescence is a period characterised by important changes in cognitive, behavioural, social, and emotional functioning attributable to biological development (i.e. puberty) and to new roles and demands in the familial and social milieu (e.g. decreased parental involvement, increased academic requirements). There are also marked changes in sleep/wake patterns during adolescence, including a decrease in sleep duration, a delay in the timing of sleep, and an increasingly large discrepancy between weekday and weekend sleep patterns. Sleep quality is reduced as well. Many adolescents have their bedrooms set up as multimedia entertainment centres, making it difficult to associate going to bed with sleep onset.

The formal diagnosis of sleep phase delay (“delayed sleep phase type” or DSPT) requires 1 to 2 weeks of objective actigraphy and/or sleep diary data. The diagnosis of sleep phase delay should include exclusion of other causes of disrupted sleep such as PLMs and OSA.

Treatment of sleep phase delay or bedtime problems should begin with environmental and sleep hygiene measures, as in sleep-onset insomnia. A positive routine of 20 minutes of quiet activity is established prior to going to bed, the next step being “fading” or gradual advancement of the bedtime, advancing the bedtime by 15 minutes every few days until it is at the desired bedtime. An alternative strategy, using chronotherapy, is to delay bedtime and waketime by 3 hours each 3 days until the child has advanced around the clock. Melatonin is helpful as an adjunct for treating sleep-phase delay.

**Sleeplessness with Associated Problems**

Sleeplessness is commonly associated with other behavioural or developmental problems such as autistic spectrum disorder, cerebral palsy, cortical blindness, attention deficit hyperactivity disorder (ADHD), or other psychiatric problems. PSG may not be possible in these disorders where behaviour is a problem and the physician may be reliant on clinical history alone. As an example, children with autism have an incidence of reported sleep problems of 50% to 70%. Children with autism may have difficulty getting to sleep, night waking (prolonged waking between midnight and 0200h), short sleep, early morning waking and daytime tiredness. Treatment often includes a range of behavioural measures in combination with pharmacotherapy, many of which are given for daytime problems as well, such as clonidine and stimulants.

**Excessive Daytime Sleepiness (EDS)**

Excess daytime sleepiness (EDS) is the propensity to fall asleep in commonly encountered daytime situations. Parents of children who have poor sleep patterns often present early as their own sleep is disrupted. On the other hand, the child who sleeps too much often escapes early attention. However, childhood EDS can signify an important medical problem and when treatment is delayed or absent, can lead to serious consequences. A hypersonomolent child may present with inattention at school, restlessness, and emotional lability or can be silent, suboptimally perform academically and be described as “daydreaming”, lazy or...
forgetful. There is considerable overlap with the symptoms of excessive sleepiness and attention deficit disorder, especially of the inattentive subtype. It is very important to tease out the differences in the symptoms of tiredness, excessive sleepiness and lethargy. Broadly speaking, true excessive sleepiness in a child can be caused by anything that decreases the amount of sleep compared with the child's needs (e.g. delayed sleep phase), that decreases the quality of sleep [sleep-disordered breathing (SDB)], or increases the need for sleep beyond that required for other children of the same age (e.g. narcolepsy).

A careful history and physical examination, including a detailed sleep history, will help elucidate the possible causes of EDS. Specialised sleep related investigations such as PSG and a multiple sleep latency test (MSLT) might be needed. Actigraphy is also helpful in establishing the timing and duration of sleep. These tests often have to be interpreted with caution and sometimes repeated for children. For a practical review of EDS in children, please see a recent review.

Parasomnias

The word “parasomnia” comes from “para-” (around) and “somnia” (sleep). This is a type of disorder characterised by abnormal behavioural or physiological events occurring in association with sleep, specific sleep stages or sleep-wake transitions. Parasomnias in children are common, affecting up to 17% of children. Parasomnias can be divided into 3 subcategories, namely those that are associated with arousals from REM sleep (commonly nightmares, for example, and uncommonly REM sleep behaviour disorder), those that arise from non-REM sleep (e.g. sleepwalking and night terrors), and lastly, less stage specific parasomnias like enuresis and cataplexy.

Parasomnias can often be precipitated by an underlying sleep disorder, mainly SDB or periodic limb movement disorder (PLMD).

The treatment focuses on the underlying cause. Safety in the home is of paramount importance especially in the case where children are displaced from their beds (e.g. sleep walking and confusional arousals). Mostly parents can be reassured that many parasomnias are age-limited and that in the vast majority, there are no long-term neurocognitive or psychological consequences. Apart from pharmacotherapy, hypnotherapy has been reported to be of some success in the primary parasomnia.

Periodic Leg Movements and the Restless Leg Syndrome

PLMs are repetitive involuntary arm or leg movements occurring in sleep. When they are associated with clinical criteria of unpleasant sensations in the legs or an urge to move the legs which is worse at rest, or in the evening, and partial or complete relief is obtained by moving or stretching the legs then this is termed the restless leg syndrome (RLS). Children may have a PLMD diagnosed on PSG with or without the features of RLS. As in the case scenario, PLMs may also be found in association with SDB. In either case, PLMs can disrupt sleep, resulting in daytime tiredness. PLMs may be associated with iron deficiency in children and children may respond to maintaining the serum ferritin level above 40 mcg/L.

Obstructive Sleep Apnoea

The obstructive sleep apnoea syndrome (OSAS) in children is characterised by prolonged partial or intermittent complete upper airway obstruction resulting in intermittent hypoxaemia, hypercapnia, recurrent arousals and sleep fragmentation. OSAS is part of the spectrum of SDB which comprised primary snoring, upper airway resistance syndrome (UARS) and OSAS. Habitual snoring is reported in 7% to 12% of children. OSAS, on the other hand, occurs in 1% to 3% of children. OSAS in childhood may occur at any age but its peak incidence is between 2 and 6 years of age, which coincides with the period that the tonsils are at their largest size relative to the pharyngeal airway. As well as adenotonsillar hypertrophy, OSAS may also be caused by structural airway abnormalities, by hypotonia of the airway musculature, or by incoordination of the airway and respiratory muscles.

The symptoms of OSAS may be divided into nocturnal and daytime symptoms. While snoring is the principal nocturnal symptom of OSAS, mouth breathing, increased work of breathing or difficulty breathing in sleep, witnessed obstructive events, excessive sweating in sleep, restlessness, frequent wakenings, unusual sleep positions, or just parental concern about the child’s breathing in sleep may be reported.

Daytime symptoms or consequences of OSAS include excess daytime sleepiness, hyperactive behaviour, learning difficulties, and cognitive problems. The more severe consequences of OSAS such as failure to thrive, systemic hypertension, cor pulmonale, and death are reported less frequently with an increasing awareness of the disorder and access to diagnostic and treatment facilities. However, even mild OSAS and primary snoring may be associated with cognitive deficits.

The diagnosis of OSAS in children is by clinical assessment and PSG. Clinical assessment of OSAS has been found to be inaccurate in predicting OSAS detected by PSG and probably over-diagnoses OSAS. None-the-less, clinical assessment is essential to allocate and prioritise scarce PSG resources, to detect co-morbidities or underlying conditions, and to exclude non-OSA causes of breathing difficulties in sleep. PSG is not widely available and, even where available, a review of practice found that only 10% of children undergoing adenotonsillectomy had...
a PSG performed. Nevertheless, the use of PSG prior to adenotonsillectomy should still be considered, as children with severe OSAS prior to surgery should undergo repeat postoperative assessment for persistent disease. PSG can also directly perioperative management.

Treatment options for OSAS in children include adenotonsillectomy, nasal mask continuous positive airway pressure (CPAP), and corticosteroids or other anti-inflammator. Adenotonsillectomy is curative in 83% of children and is the primary treatment for OSAS in children. As there is a 15-20% rate of residual OSAS, postoperative clinical assessment is essential. Adenotonsillectomy is not without risk, with an overall mortality between 1 in 4000 and 1 in 27,000, and morbidity reported from 5-10% to 18-34%. The most frequent postoperative complication is respiratory failure secondary to upper airway oedema/obstruction or pulmonary oedema. The risk of postoperative complications is increased in children <3 years of age, OSAS, obesity, asthma, craniofacial or other comorbid conditions. These children should be observed overnight postoperatively. Children with an obstructive apnoea-hypopnoea index (OAHI) >20 events/h are at even greater risk of postoperative compromise and should be observed in a high dependency setting.68

Due to the possible risks of surgery, medical therapies have been investigated. Intranasal corticosteroids are effective in reducing the OAHI but may not resolve OSAS completely. Montelukast is an orally bioavailable cysteinyl leukotriene (LT) receptor antagonist which is a selective LT1-R blocker. There is some early evidence that montelukast might be effective in some children with OSA, particularly in children with residual problems post-adenotonsillectomy and in combination with intranasal steroids.72,73 At this stage it is unclear how long these effects will last.

CPAP is suitable for children with persistent OSAS post-adenotonsillectomy or with contraindications to surgery.66 CPAP may also be useful in avoiding tracheostomy or complex surgery in children with structural abnormalities of the face and airway. CPAP is a generally safe treatment but requires a period of parental training.74,75 Possible adverse effects of CPAP are pressure areas on the face, irritation of the eyes due to air leak, drying of the nose and mouth, epistaxis, and midface hypoplasia due to prolonged use. Some centres employ heated humidification as an adjunct to improve comfort and compliance of CPAP, either routinely or for individual children who are experiencing problems tolerating treatment. However, as yet there are no randomised studies of heated humidification in children and there is a need for evidence to support or refute this practice.

Neuromuscular Disease
The neuromuscular diseases are a group of disorders of varying aetiology characterised by muscle weakness and wasting. Due to involvement of the respiratory muscles, children with these disorders often go on to develop symptoms of respiratory failure such as headaches and excessive daytime sleepiness.

SDB in neuromuscular disease is primarily regarded as a disorder of hypoventilation. However, some authors have found evidence of obstructive apnoea and hypopnoea prior to the onset of hypoventilation. This is due to the failure of the upper airway musculature to support the airway during inspiration. The onset of respiratory failure can be predicted by spirometry and other measures of respiratory function and regular assessment is strongly recommended.80 There is evidence from long-term follow-up studies that the use of non-invasive ventilation (NIV) in neuromuscular disease is associated with improved survival in adults, particularly in patients with daytime hypercapnoea. Studies in children, including one RCT, have shown improved ventilation on NIV and a reduced rate of chest infection.

Central Hypoventilation
Central hypoventilation in children may be congenital or acquired in origin. Congenital central hypoventilation syndrome (CCHS), formerly known as Ondine’s curse, is a syndrome of primary persistent hypoventilation in sleep in the absence of primary pulmonary or neuromuscular disease and with onset in the first year of life.77 The gene for CCHS has been identified as a paired-like homeobox (PHOX2B) located on chromosome 4p12. Most cases of CCHS are heterozygous for thePHOX2B polyalanine repeat expansion mutation, with alternative mutations in PHOX2B in the other cases.9 CCHS is often treated with tracheostomy and invasive ventilation, and sometimes diaphragmatic pacing. However some infants can be managed with NIV.

Case Scenario: Conclusion
This patient underwent PSG which demonstrated an OAHI of 22 events/h. He also had a prolonged sleep latency and 8 PLMs per hour. He was referred for adenotonsillectomy and was treated with oral iron for 3 months and CPAP during sleep while he was on the waiting list. The night after surgery he had close monitoring of his oxygen saturation. At follow-up 8 weeks after surgery, he had resolution of obstructive apnoea and hypopnoea prior to surgery should undergo repeat postoperative assessment for persistent disease. PSG can also directly perioperative management.

Discussion
This article has attempted to convey the range of sleep disorders in children and the role they play in a wide range of so-called “daytime problems”. The child in the case...
scenario presented with snoring, witnessed apnoea, daytime sleepiness, poor school performance, inattention and impulsivity. These symptoms are consistent with OSAS, however, there were also features of sleep phase delay and PLMs which may be contributing to his daytime symptoms and which complicate his management. A child such as this is likely to benefit from adenotonsillectomy but may also require behavioural therapy and treatment with iron. PSG will help delineate the contribution of each of these problems, thus guiding management, and will also determine if close perioperative monitoring is indicated. Postoperative clinical review is essential in order to assess him for persistent OSAS and for management of his other problems.

Appropriate management has the potential to improve his school performance and quality of life. Paediatric sleep medicine is a relatively new and developing specialty. A complex case such as this illustrates the importance of taking a good sleep history and rationalising a range of investigations and management strategies.

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