Down's Syndrome Association
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Annual Health Check Information for GPs

Sleep Problems

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Below is Down's syndrome specific information. The information is for GPs and is to be used alongside DSA's Adult Health Book.

Introduction

Sleep problems are common in adults with intellectual disability (ID), with estimated prevalence rates of 9-34%[1]. Around 50% of children with Down's syndrome (DS) report problems with sleep[2], but the prevalence of sleep disorders in adults with DS is unclear. There are currently no specific guidelines for management of sleep disorders in adults with DS and much of the available evidence base is derived from studies in the general population.

Insomnia and behavioural sleep problems

Many adults with DS report poor sleep. Disorders of initiation and maintenance of sleep (DIMS) are common.
A recent systematic review of sleep problems in adults with ID[1] estimates that settling problems are evident in 9-27% and night wakening in 11-34%. A study of 186 adults with DS[3] reports initial insomnia in 9%, broken sleep in 7% and early morning wakening in 7%, with 6% reporting a significant sleep problem. Rates of reported sleep problems increased with severity of ID.

Poor sleep can lead to daytime sleepiness, impaired daytime function, mood disturbances, irritability and challenging behaviour. However, these symptoms may be overlooked attributed to the DS itself.

There are few studies of treatment of sleep problems in adults with ID or DS, although the available data[1,4,5] suggests that non-pharmacological treatments, including cognitive behavioural therapy, sleep hygiene, optimal scheduling stimulus control and light therapy, can be effective. Although availability of these services within the NHS is limited, support can be accessed via third sector organisations (see Useful Contacts).

Adults with DS who report sleep problems should be investigated by a sleep specialist to exclude underlying causes such as sleep-disordered breathing or parasomnia.

Sleep-disordered breathing

Sleep-disordered breathing (SDB) is characterised by repeated pauses in breathing during sleep. Muscle relaxation during sleep leads to a partial (hypopnoea) or complete blockage (apnoea) of the airway, resulting in a repetitive cycle of airway obstruction, followed by resumption of breathing. Snoring can arise from vibration of the narrowed airway. Factors exacerbating this include the effects of gravity whilst lying on the back, fat deposits around the neck, anatomical features reducing the size of the pharynx and reduced muscle tone due to alcohol, sedatives or rapid eye movement (REM) sleep. A clinically-significant number of hypopnoeas or apnoeas during sleep is known as obstructive sleep apnoea (OSA), and the term obstructive sleep apnoea/hypopnoea syndrome (OSAHS) is used when OSA is associated with significant symptoms. Symptoms of OSAHS are outlines in Table 1.

Table 1: Nocturnal and Diurnal symptoms of OSAHS

Nocturnal symptoms	**Diurnal symptoms**
Snoring	Excessive daytime sleepiness (EDS)
Witnessed apnoeas	Morning headache
Gasping or choking episodes	Dry mouth
Restlessness	Poor concentration/memory
Frequent awakenings	Mood disturbances
Nocturia	Behavioural disturbances

In children, sleepiness can manifest as hyperactivity or behavioural problems, and it is possible that this is also the case in adults with DS; one study[6] has shown that self-reported breathing pauses and snoring are significantly associated with higher scores on scales measuring emotional and behavioural disturbance.

Sleep-disordered breathing is common in the general adult population, with SDB present in around 20% and 2-4% suffering from OSAHS[7]. Adults with DS are predisposed to SDB since many of the risk factors for SDB overlap with anatomical and physiological features associated with DS (see Table 2).

Table 2: Anatomical and physiological features predisposing adults with DS to SDB

Obesity/weight gain
Midface hypoplasia
Thick neck
Gothic palate
Adenotonsillar hypertrophy
Relative macroglossia
Hypotonia
Increased mucosal secretions

Two population studies[8,9] estimate the prevalence of OSA in adults with DS at 83-88%, although participant numbers were very small (n=6 and n=16 respectively). A recent study[10] of over 1000 adults with DS estimates an OSAHS prevalence of around 40% - 10 times higher than in the general population.

First-line treatment for individuals with OSAHS is continuous positive airway pressure (CPAP) therapy. Surgery is not recommended in adults, although removal of tonsils and/or adenoids may be effective in individuals with adenotonsillar hypertrophy. Current national guidelines[11] recommend treatment for all symptomatic patients in the moderate/severe category (≥15 apnoeas/hypopnoeas per hour of sleep). However, these guidelines are based on studies in the general (mainly male, middle-aged) population, and do not take into account specific populations such as people with DS.

In the non-DS population, untreated OSAHS can result in cognitive impairment, mood disturbances and a reduced quality of life. Greater OSAHS severity is linked with poorer cognitive performance in adults in the general population[12], and SDB has been shown to contribute to cognitive deficits in adults with DS[13]. All-cause mortality is significantly increased in non-DS adults with OSAHS; however, in patients treated with CPAP, mortality levels are comparable to those without OSAHS[14]. A recent randomised, controlled trial of CPAP in adults with DS and OSAHS demonstrated sustained and significant improvements in subjective sleepiness, behaviour and cognitive function with 12 months of CPAP use[18].

Given the increased risk of SDB in adults with DS, a thorough sleep history should be taken as part of an annual health check. While not designed specifically for use in adults with DS, a standard instrument to assess daytime sleepiness, such as the pictorial Epworth Sleepiness Scale, may be useful[15,16]. Adults with DS who exhibit symptoms of SDB should be referred to a sleep specialist for investigation and treatment; the British Sleep Society (see Useful Contacts) holds a register of all UK sleep services.

Parasomnias

Parasomnias are disorders characterised by unusual behaviours arising from sleep. They are categorised by the stage of sleep from which they arise, with sleepwalking, night terrors and confusional arousals occurring during slow wave sleep, and REM behaviour disorder or nightmares occurring during REM sleep. Some parasomnias, such as rhythmic movement disorders (head banging, head rolling, body rocking), sleeptalking and bruxism (tooth-grinding) can occur in any sleep stage.

In a study[17] of over 200 adults with ID, 36 of whom had DS, parasomnias were reported by 14% of the population (there was no significant difference between adults with DS and other forms of ID). However, literature is scant and further studies are required.

Adults with DS who exhibit parasomnias should be referred to a sleep specialist for investigation. Pharmacological treatment can be effective in the general population, although there is currently no evidence base for this in adults with DS.

Conclusion

Sleep problems are common in adults with DS, and a sleep history should form part of the annual health check for these individuals. Treatment can often be effective, so poor sleep should not be overlooked as "just part of the syndrome". Further research into sleep problems in adults with DS is required to allow provision of evidence-based management of sleep problems in this patient group.

References

1. van de Wouw E, Evenhuis HM, Echteld MA. Prevalence, associated factors and treatment of sleep problems in adults with intellectual disability: A systematic review. *Research in Developmental Disabilities.* 2012; 33: 1310-1332.
2. Down's Syndrome Association. Managing sleep problems in children with Down's syndrome. Down's Syndrome Association Medical Series. www.downs-syndrome.org.uk. 2008.
3. Boyle A, Melville CA, Morrison J, Allan L, Smiley E, Espie CA, Cooper S-A. A cohort study of prevalence of sleep problems in adults with intellectual disabilities. *Journal of Sleep Research*. 2010; 19: 42-53.
4. Gunning MJ, Espie CA. Psychological treatment of reported sleep disorder in adults with intellectual disability using a multiple baseline design. *Journal of Intellectual Disability Research*. 2003; 47(3) 191-202
5. Hylkema T, Vlaskamp C. Significant inprovement in sleep in people with intellectual disabilities living in residential settings by non-pharmaceutical interventions. *Journal of Intellectual Disability Research*. 2009; 53(8): 695-703.
6. Hill EA, Fairley D, Van Putten S, Cooper S, Forbes JF, Williams L, Riha RL. Prevalence of sleep apnoea, sleepiness and behavioural/emotional disturbances in adults with Down's syndrome in Scotland. *European Respiratory Journal.* 2012; 40 (suppl.56): 325s.
7. Young T, Palta M, Dempsey J, Skatrud J, Webber S, Bader S. The occurrence of sleep-disordered breathing among middle-aged adults. *New England Journal Of Medicine.* 1993; 328: 1230-35.
8. Resta O, Foschino Barbaro MP, Giliberti T, Caratozzolo G, Cagnazzo MG, Scarpelli F, Nocerino MC. Sleep related breathing disorders in adults with Down syndrome. *Down Syndrome Research and Practice*. 2003; 8(3): 115-120.
9. Trois MS, Capone GT, Lutz JA, Melendres MC, Schwartz AR, Collop NA, Marcus CL. Obstructive sleep apnea in adults with Down syndrome. *Journal of Clinical Sleep Medicine*. 2009; 5:317–23.
10. Hill EA, Fairley D, Van Putten S, Cooper S, Forbes JF, Williams L, Riha RL. Sleep-disordered breathing in adults with Down syndrome: a cross-sectional prevalence study. *In submission.*
11. SIGN (Scottish Intercollegiate Guideline Network). SIGN73: Management of Obstructive Sleep Apnoea/Hypopnoea Syndrome in Adults. *www.sign.ac.uk.* 2003.
12. Engleman HM, Douglas NJ. Sleep. 4: Sleepiness, cognitive function, and quality of life in obstructive sleep apnoea/hypopnoea syndrome. *Thorax*. 2004; 59: 618–622.
13. Andreou G, Galanopoulou C, Gourgoulianis K, Karapetsas A, Molyvdas P. Cognitive status in Down syndrome individuals with sleep disordered breathing deficits (SDB). *Brain and Cognition.* 2002; 50(1):145-9.

14. Marti S, Sampol G, Munoz X, Torres F, Roca A, Lloberes P, Sagalés T, Quesada P, Morell F. Mortality in severe sleep apnoea/hypopnoea syndrome patients: impact of treatment. *European Respiratory Journal.* 2002; 20: 1511–1518.
15. Ghiassi R, Murphy K, Cummin A, Partridge MR. Developing a pictorial Epworth Sleepiness Scale. *Thorax*. 2011; 66(2): 97-100.
16. Hill EA, Fairley D, Van Putten S, Cooper S, Forbes JF, Williams L, Riha RL. Use of the pictorial Epworth Sleepiness Scale in adults with Down's Syndrome. *Journal of Sleep Research* 2012; 21 (suppl.1): 291.
17. Brylewski JE, Wiggs L. A questionnaire survey of sleep and night-time behaviour in a community-based sample of adults with intellectual disability. *Journal of Intellectual Disability Research*. 1998; 42(2): 154-162.
18. Hill E A, Fairley D, Van Putten S, Cooper S, Williams L, Riha RL. A prospective, randomised, controlled trial of CPAP in adults with Down syndrome. In submission

Useful contacts

British Sleep Society
www.sleepsociety.org.uk

Down's Syndrome Association
www.downs-syndrome.org.uk

Down's Syndrome Scotland
www.dsscotland.org.uk

Sleep Scotland
www.sleepscotland.org

Sleep Research Unit, University of Edinburgh
www.ed.ac.uk/clinical-sciences/sleep-research

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