



Factsheet 20

Sleep difficulties and CHARGE syndrome

HELEN HEUSSLER, MB, BS, FRACP, MRCPCH, DM, Conjoint Associate Professor,
University of Queensland

Sleep problems in any child have strong relationships to behaviour and function. Parents report that over half of children with CHARGE syndrome have a sleep problem (Hartshorne *et al.* 2009) this is not surprising given the difficulties these children face.

The sleep cycle

It is common for children with a significant visual impairment to have difficulty with sleep rhythm (Stores and Ramchandani, 1999). This is because melatonin is produced by the pineal gland in the brain in response to low light, and production is stopped by bright light exposure.

Where children are not able to visualise this, then melatonin secretion cycles tend to be disrupted. In these circumstances, using other methods such as strict adherence to routine, food, etc., it is important to help a child to get into a sleep cycle.

If a consistent routine does not enable a child to develop a regular sleep cycle then the use of melatonin has been advocated in children with visual impairment with reasonable success.

Expected contributions to sleep difficulties are shown in the chart on page 3.

Middle ear infections

A relationship between poor sleep and middle ear infections has also been found. However, it may be that these children had repaired clefts and choanal atresia, and so middle ear pathology remains (Samadi *et al.* 2003). It is not known whether the effect of ear infections on sleep is related to pain or to a longstanding effect on the upper airway.



Behaviour

Behavioural and sleep difficulties are related in people with CHARGE. It is hard to know how much sleep difficulties contribute to behaviour problems and vice versa – particularly settling a child at night.

Sleep difficulties in CHARGE have been most strongly associated with behavioural problems, self-absorbed behaviour, anxiety and social relating behaviours (Hartshorne *et al.* 2009). This is in contrast to the general population where sleep difficulties are most strongly related to hyperactivity, concentration and memory.

Craniofacial abnormalities

All children with craniofacial abnormalities, clefts and choanal atresia are at risk of sleep disruption due to obstructive sleep apnea.



The obstruction should be managed as early as possible to avoid some of the compounding effects on a child's learning and behaviour. Known effects of obstructive sleep apnea include poor concentration, hyperactivity, alterations in mood, impulsivity and other problems that affect learning and cognition.

Many of these effects can be ameliorated by treatment (Friedman *et al.* 2003). Obstructive sleep apnea can cause fragmented sleep and children may find it difficult to maintain sleep unless the obstruction is relieved. This alone may effect how a child functions during the day.

Sleep initiation and maintenance

The study by Hartshorne *et al.* (2009) shows that many children with CHARGE have problems going to sleep and staying asleep. These difficulties have also been commonly identified in children with autism (Johnson and Malow, 2008). Given the number of children with CHARGE who also exhibit autistic-like traits, this may not be surprising.

There are many theories as to why children with CHARGE may have problems with sleep initiation and maintenance. Commonly, it may relate to children with these traits finding it difficult to self-settle. High levels of anxiety or sensory processing difficulties will compound this (Shochat *et al.* 2009). These settling difficulties should be explored in depth – particularly where the child has difficulty in communicating.

Management

Where an obstruction is causing sleep difficulties, this should be treated. In early childhood this may include treatment for choanal stenosis. Also, surveillance for obstructive symptoms is needed in the early years when children are prone to enlarged tonsils and adenoids. This may also require early treatment including surgery.

Children should be encouraged into good sleep hygiene practices. These involve regular routine and settling practices with day/night contrast, e.g. activity/quiet and light/dark. Good sleep hygiene includes regular bed and wake times with appropriate sleep associations such as various comfort objects.

Exploring what helps a child to settle and relax is important – and can vary in children with CHARGE who may have different sensory experiences. It is important to know what settles the child and to be able to encourage this as part of the night settling routine. Sometimes this may involve heavy covers, temperature management, vibratory toys, etc.

It is important to try to avoid bright light at night and to try to maximise night-day (dark-light) contrast. Children who develop significant circadian scheduling problems relating to visual impairment or poor routines may benefit from a trial of melatonin (Jan and Freeman, 2004).

Sleep – one parent's perspective

**SIMON HOWARD – PARENT OF A DAUGHTER WITH CHARGE;
VICE CHAIR OF THE CHARGE FAMILY SUPPORT GROUP**

Night parties! The bane of many parents' lives who have children with CHARGE.

It is clear to me from conversations with other parents and from the various discussion forums/boards, that many parents choose to put up with these wakings. Some because they do not wish to add to the list of medications their children are taking, others because there is no obvious solution. The effect of this not only on the individual, but also the whole family, needs to be considered.

With the CHD7 gene having an impact throughout the body, I believe the brain cannot be excluded and there may be neurological reasons that contribute to sleep disruption.

In the context of sleep another big factor that gets overlooked is pain. There are the obvious causes such as ear infections and reflux but I believe there may be other reasons such as dental pain, migraines, constipation and other gastro intestinal problems.

**Created: November 2013
Review due: November 2015
www.sense.org.uk**





Factors expected to contribute to sleep difficulties

Facet of sleep	CHARGE	Mechanism
Circadian Rhythm	Visual impairment	Disruption to Melatonin cycle
Airway	Choanal atresia Abnormal airway Repaired cleft palate	Nasal and airway obstruction
Sleep disruption	Recurrent otitis media Gastro-oesophageal reflux	Pain causing sleep fragmentation
Behaviour	Recurrent hospitalisation Anxiety Hyperactivity Sensory issues	Difficulty with self-soothing and sleep initiation and maintenance

GLOSSARY

Choanal stenosis/atresia: a narrowing or blockage of the passageway between the nose and the pharynx by tissue.

Circadian rhythm: The 24-hour activity cycle. Sometimes referred to as the biological clock.

Gastro-oesophageal reflux: when liquid content of the stomach regurgitates (backs up or refluxes) into the oesophagus .

Otitis media: inflammation of the middle ear.

Sleep apnea: a sleep disorder characterised by abnormal pauses in breathing or instances of abnormally low breathing during sleep.

REFERENCES

Friedman B.C., Hendeles-Amitai A., Kozminsky E., Leiberman A., Friger M., Tarasiuk A., Tal, A. (2003) Adenotonsillectomy improves neurocognitive function in children with obstructive sleep apnea syndrome. *Sleep*. Dec 15, 26(8), pp. 999–1005.

Hartshorne, T.S., Heussler, H.S., Dailor, A.N., Williams, G.L., Papadopoulos, D., Brandt, K.K. (2009) Sleep Disturbances in CHARGE Syndrome. *Developmental Medicine and Child Neurology*. 51(2), pp. 143–150.

Jan, J.E. and Freeman, R.D. (2004) Melatonin therapy for circadian rhythm sleep disorders in children with multiple disabilities: What have we learned in the last

decade? *Developmental Medicine and Child Neurology*. 46, pp. 776–782.

Johnson, K.P. and Malow, B.A. (2008) Sleep in children with autism spectrum disorders. *Current Treatment Options in Neurology*. 10(5), pp. 350–9.

Quine, S. (2000) Sleep problems in primary school children: Comparison between mainstream and special school children. *Child Care Health Dev*. 27, pp. 201–211.

Samadi, D.S., Shah, U.K., Handler, S.D. (2003) Choanal atresia: a twenty-year review of medical comorbidities and surgical outcomes. *The Laryngoscope*. 113, pp. 254–258.

Shochat, T., Tzischinsky, O., Engel-Yeger, B. (2009) Sensory hypersensitivity as a contributing factor in the relationship between sleep and behavioural disorders in normal schoolchildren. *Behav Sleep Med*. 7(1), pp. 53–62.

Stores, G. and Ramchandani, B.M. (1999) Sleep disorders in visually impaired children. *Dev Med Child Neurol*. 41, pp. 348–352.

Wiggs, L. and Stores, G. (1996) Severe sleep disturbance and daytime challenging behaviour in children with severe learning disabilities. *J Intellect Disabil Res*. 40, pp. 518–528.