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.
## Sleep Difficulties

**Factors expected to contribute to sleep difficulties**

<table>
<thead>
<tr>
<th>Facet of sleep</th>
<th>CHARGE</th>
<th>Mechanism</th>
</tr>
</thead>
<tbody>
<tr>
<td>Circadian Rhythm</td>
<td>Visual impairment</td>
<td>Disruption to Melatonin cycle</td>
</tr>
<tr>
<td>Airway</td>
<td>Choanal atresia</td>
<td>Nasal and airway obstruction</td>
</tr>
<tr>
<td>Sleep disruption</td>
<td>Recurrent otitis media</td>
<td>Pain causing sleep fragmentation</td>
</tr>
<tr>
<td></td>
<td>Gastro-oesophageal reflux</td>
<td></td>
</tr>
<tr>
<td>Behaviour</td>
<td>Recurrent hospitalisation</td>
<td>Difficulty with self-soothing and sleep initiation and maintenance</td>
</tr>
<tr>
<td></td>
<td>Anxiety</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Hyperactivity</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Sensory issues</td>
<td></td>
</tr>
</tbody>
</table>

**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**


