

RCPCH Working Party on Sleep Physiology and Respiratory Control Disorders in Childhood

Lay Summary 5 – Obstructive Sleep Apnoea in children with craniofacial abnormalities

What is Obstructive Sleep Apnoea?

Obstructive sleep apnoea/hypopnoea syndrome (OSA) is a condition in which a person stops breathing for a short time when they are asleep because of closing or narrowing of the throat. This can happen many times during the night, and causes the person to wake up for very short periods to allow normal breathing to restart (although usually the person often won't remember waking up). A child with sleep apnoea often snores, may be unusually sleepy or hyperactive during the day and may have problems in concentrating because of lack of sleep.

Which craniofacial abnormalities are risk factors?

There are a number of conditions which cause problems in development of the skull and face (craniofacial area) and may result in narrowing of the airway, either because of under-development of the jaw, or of narrowing of the structures behind the nose.

Conditions which often affect the airway include achondroplasia, Pierre Robin Sequence, Treacher Collins, Crouzon, Apert, Saethre-Chotzen, Nager, Pfeiffer and Stickler syndromes. In addition, mucopolysaccharidoses such as Hunter, Hurler and Hurler-Scheie syndromes may cause increasing upper airway obstruction.

How common is OSA in children with craniofacial abnormalities?

The risk of OSA depends on the type and severity of the abnormality. However, the congenital developmental problems differ from other children with OSA as symptoms may be present even in infancy.

What are the risks of OSA?

OSA can cause impaired growth and development, and is associated with poorer academic performance. It can also put extra strain on the heart, and occasionally causes pulmonary hypertension, a very dangerous condition with elevated blood pressure in the lungs.

How can OSA be detected in children without underlying problems?

In addition to snoring, symptoms of OSA in children may include the following:

<i>Symptoms and signs associated with OSA.</i>	
During sleep	In the day
Snoring or snorts	Behaviour problems
Gasping or laboured breathing	Poor concentration
Witnessed pauses in breathing	Excessive tiredness (symptoms may be subtle)
Odd sleeping positions	Poor growth
Sweating	Morning headaches
Bedwetting	Mouth breathing and nasal speech
	Misshapen chest

Because of the risk of early OSA in children with syndromes involving craniofacial problems, we recommend that all such children are assessed with oximetry (a measure of blood oxygen levels using a soft probe wrapped around a finger or toe), preferably in combination with a measure of carbon dioxide levels during sleep (see Lay Summary 1). This should be performed urgently if they have any signs of airway obstruction, and within 4 weeks of birth in any event.

Children with Pierre Robin Sequence may have worsening obstruction between 4 and 8 weeks of age, and a repeat assessment may be needed during this period if symptoms increase.

Reassessment should otherwise be performed at 3-6 monthly intervals during the first year of life, and thereafter according to clinical symptoms.

The role of screening in children with achondroplasia and with mucopolysaccharidoses is discussed below.

What is the treatment of OSA in children with craniofacial abnormalities?

In infants with Pierre Robin Sequence the initial treatment will involve the insertion of a tube through the nostril and into the throat to hold the airway open. This can be maintained by parents for several months until the airway improves. If this fails then Continuous Positive Airway Pressure (CPAP) should be tried, where a device is fitted to the nose during sleep to prevent the collapse of the airway. In refractory cases (cases difficult to treat), tracheostomy (a tube inserted into the throat to give access to the windpipe) may be necessary. Surgery to advance and elongate the jaw has also been successful and may avoid the need for tracheostomy.

In infants with poor mid-facial growth, CPAP should be tried as the initial treatment of OSA. If this is unsuccessful, then a tracheostomy may be needed. The role of craniofacial surgery in avoiding the need for tracheostomy needs further evaluation.

In children with achondroplasia screening for OSA should be offered in the first year of life, and again every 6-12 months in the next 5 years. Further screening should probably depend on clinical symptoms if negative until the age of 5 years. If OSA is discovered then surgery to remove the tonsils and adenoids should be carried out. If this does not correct the problem, then a trial of CPAP should be considered.

In children with mucopolysaccharidoses such as Hurler, Hurler-Scheie and Hunter syndromes, the clinician and parents should discuss the risks and benefits of screening for OSA as the condition progresses. If OSA is detected then surgery to remove the tonsils and adenoids should be considered. CPAP may be an option if the symptoms persist after adenotonsillar surgery, although the pros and cons of therapy need to be carefully weighed up.