

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

Lay Summary 7 – Congenital Central Hypoventilation Syndrome

What is CCHS?

Congenital Central Hypoventilation syndrome (CCHS) is a rare genetic condition of under-breathing involving the failure of automatic control of breathing in the brain. The breathing abnormalities are always present during sleep, but may occur to a milder degree when a child is awake. Thus the condition may range in severity from being relatively mild during quiet sleep, with normal breathing when awake, to completely stopping breathing during sleep and severe under-breathing when awake. The latter may be particularly evident when feeding (particularly in infancy) or when concentrating. Thus all children with CCHS will by definition have abnormalities of breathing during sleep. If untreated, these will lead to disability and death.

How is CCHS detected?

In most cases the child will present very early in life with a variety of symptoms, which range from brief apnoeas or breath-holding spells, to complete failure to breathe after birth. Certain conditions, such as Hirschprung's Syndrome, which affects the bowel, are associated with CCHS. The diagnosis of CCHS depends on documenting that under-breathing occurs during sleep, and that this is not due to a muscle, nerve or lung disease. Measurement of breathing will involve a cardiopulmonary sleep study, sometimes with detailed measurements of breathing volumes. (For more details about tests, please see Lay Summary 1). The diagnosis can be confirmed in over ninety-five per cent of cases by finding the genetic abnormality.

If there is a suspicion of CCHS we recommend that children are referred to a specialist centre for further assessment.

How is CCHS treated?

In children with CCHS ventilatory support (assistance with breathing) is almost always essential for survival. This will normally require a tracheostomy (a tube in the neck which allows access to the windpipe) in the first 6 years of life.

If a child with CCHS continues to need ventilatory support around the clock, diaphragmatic pacing should be considered. This involves the surgical insertion of wires and a control box which regularly stimulate the diaphragm and induce regular breaths.

The care and assessment of CCHS should be supervised by a specialist centre with experience of CCHS management.

Because children with CCHS do not have a normal ability to sense when their breathing is inadequate, levels of oxygen in the blood (oxygen saturation) should be monitored continuously during sleep as a safety measure. During acute illnesses children with CCHS require checks of oxygen saturation and carbon dioxide levels when awake.

Children with CCHS may not increase their breathing enough during exercise, and carers should be aware of this as a potential problem.

Alcohol or cannabis use carry particular dangers in children with CCHS.