



# Congenital central hypoventilation syndrome: information for families

---

**Congenital Central Hypoventilation syndrome (CCHS) is a condition affecting how the autonomic nervous system manages breathing. This information sheet from Great Ormond Street Hospital (GOSH) explains the causes, symptoms and treatment of congenital central hypoventilation syndrome.**

The autonomic nervous system controls a number of bodily processes, such as heart rate, blood pressure, breathing, digestion of food and body temperature. These processes happen automatically without us having to think about them.

In CCHS, the normal safeguards, used by the autonomic nervous system to control breathing, are impaired. When breathing is shallow while asleep, the levels of carbon dioxide in the blood increase, which stimulates a breath. In CCHS, this stimulus does not occur and breathing can stop.

In the majority of people, this only happens while asleep, but for people with severe CCHS, it occurs all day every day and may be particularly evident when feeding (particularly in infancy) or when concentrating. If untreated, CCHS can lead to disability and death.

## What causes congenital central hypoventilation syndrome?

CCHS is a rare condition affecting around 1000 people worldwide. It is possible that this is an under-estimate as some cases of sudden unexplained death may in fact have been caused by CCHS.

CCHS is caused by a genetic mutation affecting a particular gene named PHOX2B. This gene is responsible for the development of nerves early in pregnancy to form specific types of nerve cells, especially in the autonomic nervous system. As the nerve cells do not work as they should, messages between the body and brain are not passed on correctly.

Most people with CCHS do not inherit the faulty gene; rather it develops for no reason. However, CCHS can be passed on from parent to child if a child inherits one copy of the faulty gene.

## What are the symptoms of congenital central hypoventilation syndrome?

Various signs and symptoms of CCHS tend to appear soon after birth, ranging from brief pauses in breathing (apnoea) or breath-holding spells, to complete failure to breathe after birth. The skin may develop a blue tinge (cyanosis) due to the reduced amount of oxygen circulating in the blood.

Approximately 20 per cent of children with CCHS will also have a condition named Hirschsprung's Disease. This is a rare disorder of the bowel, most commonly the large bowel (colon), which is related to the underdevelopment of the

autonomic nervous system for the gut and can lead to severe constipation and intestinal obstruction.

Other symptoms, such as a decreased response to light and pain and difficulties in controlling body temperature may also be seen. It has been reported that children with CCHS have a similar facial appearance. These are all related to the problems with the autonomic nervous system.

## **How is congenital central hypoventilation syndrome diagnosed?**

Diagnosis of CCHS depends on documenting that under-breathing, or 'hypoventilation', occurs during sleep, particularly in certain stages of sleep. Breathing difficulties while sleeping can be caused by other medical conditions so several diagnostic tests may be needed to confirm or rule out CCHS.

Sleep studies are used to investigate breathing abnormalities – these involve a child sleeping while various measurements, such as breathing rate, heart rate, carbon dioxide and oxygen levels (saturation), are recorded.

The diagnosis of CCHS is usually confirmed by a blood test to check for the mutation on the PHOX2B gene. As the mutation can sometimes be inherited, it is advised that close relatives also be tested and genetic counselling offered should the mutation be present.

## **How is congenital central hypoventilation syndrome treated?**

There is currently not a cure for CCHS but the symptoms can be managed. As CCHS can affect a number of systems in the body, management by several medical teams will be required

(multidisciplinary approach). Children with CCHS should be supervised by a specialist centre with experience in CCHS management.

Ventilatory support is used to assist with breathing and is almost essential for survival. In infants and young children this may require a tracheostomy (a direct opening into the windpipe), although using a mask or nasal prongs (also known as 'non-invasive ventilation') may be sufficient to stimulate breathing while asleep.

An alternative to ventilatory support, which may be more appropriate for some children, is the stimulation of the diaphragm by an electrical implant, stimulating the nerves going to the diaphragm to make the lungs take in air. The diaphragm is the curved muscle that separates the chest from the abdomen. It works in a similar way to a heart pacemaker causing regular contractions of the diaphragm whenever they do not occur naturally.

Children with Hirschsprung's disease in addition to CCHS will need surgery to remove the affected section of bowel.

## **What happens next?**

Normal day-to-day activities should be possible, although swimming and playing near water are best avoided. As children with CCHS grow, it is important that they understand the risks of alcohol and illicit drugs pose in further depressing the autonomic nervous system and increasing the risks of breathing difficulties.

Although CCHS is a life-long condition, complications can be avoided with early diagnosis and specialist management. Life expectancy for children with CCHS has improved greatly with technological advances, particularly in ventilatory support, and most will grow up to adulthood, with a near normal work and family life.

## **Further information and support**

In the UK, the CCHS Support Group can offer support and advice to anyone affected by CCHS. Call them on 01362 696 509 or visit their website at [www.cchssupport.co.uk](http://www.cchssupport.co.uk)

There is also a support organisation in the United States called the CCHS Family Network – visit their website at [www.cchsnetwork.org](http://www.cchsnetwork.org)