

Central congenital hypoventilation syndrome

[Health & Medicine](#), [Disease](#)



Central Congenital Hypoventilation Syndrome What is CCHS? Many people around the world stop breathing when they go to sleep. Why? It's because they have a syndrome called Central Congenital Hypoventilation Syndrome or Ondine's Curse. What is this? Congenital central hypoventilation syndrome (CCHS) is a rare syndrome, present from birth, and is defined as the failure of automatic control of breathing.. Some may only have apnea when asleep, but in some severe cases they may experience apnea while awake.

About 300 children worldwide have this syndrome. History: In a French folktale, a nymph named Ondine was a water goddess. Like all nymphs and mermaids, if they was to fall in love with a mortal and bears his child, then they would age like a mortal. Meaning they would lose their everlasting beauty. Ondine ends falling in love with a mortal, and a year into the marriage, she bears a son. Then Ondine finds out that the mortal has been unfaithful with his former fiance. Ondine puts a curse on him, saying for so long he is wake, he shall breathe.

But if he shall ever fall into sleep, breathe will desert him. This is where the name Ondine's curse is from. Causes: CCHS is caused by mutations of the PHOX2B gene. The PHOX2B gene provided instructions for making a protein that acts early in development to help promote the formation of nerve cells and regulate the process by which the neurons mature to carry out specific functions. 90 percent of cases of CCHS result from new mutations in the PHOX2B gene. Children who have CCHS there is no history in their family of this syndrome.

CCHS is exhibited typically as a congenital disorder, but in rare can result from severe brain or spinal trauma, which can result from automobile
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accident, stroke or as a complication of neurosurgery. In one case a woman was diagnosed with CCHS because of her chronic alcoholism. Symptoms: Some symptoms that victims may experience are: shallow breathing, night apnea, brain damage, hypoxemia, hypercapnia, acidosis, and dysphagia. It may cause hirschsprung disease or neuroblastoma.

Hypoxemia is an abnormally low concentration of oxygen in the blood. Hypercapnia is the physical condition of having the presence of an abnormally high level of carbon dioxide in the circulating blood. Acidosis is an excessively acid condition of the body fluids or tissues. Dysphagia is condition in which swallowing is difficult or painful. Hirschsprung disease involves an enlargement of the colon, caused by bowel obstruction. Neuroblastoma is a malignant tumor composed of neuroblasts, most commonly in the adrenal gland Treatment:

If an infant is suspected to have CCHS a sleep study is ordered and performed to determine how server the breathing difficulty is. Cardiac and neurological examinations are done to rule out any other type of disorder. Early diagnosis is prominent because if not caught the infant's oxygen level will keep decrease until the child cannot breathe on their own. One diagnosed the child will either be on a respirator (ventilator) while asleep or other may be on the respirator for 24 hours day. To use the respirator individuals will have to have a tracheotomy performed.

Tracheotomy is a surgical operation that creates an opening into the trachea with a tube inserted to provide a passage for air. Some infants may have a surgical implant in the diaphragm muscle can allow electrical stimulation of the muscle to control breathing. Mistakes are made: Many children are <https://assignbuster.com/central-congenital-hypoventilation-syndrome/>

diagnosed with a different disease or syndrome when they actually have CCHS. Infants with CCHS may be mistakenly thought to have heart defects. Some infants that have unrecognized CCHS may die and be thought to have SIDS or Sudden Infant Death Syndrome.

Children with CCHS: Children with CCHS can live a normal life. They can go to school, go out with friends, or hang out with family just like other kids. The only real precaution children with CCHS has to be supervised while there in the bathtub or pool. They may “ forget” to breathe while underwater. This may cause them to breathe in some water. Central Congenital Hypoventilation Syndrome is a lifelong syndrome. If children with CCHS is treated right their life expectancy will not be alter. They can live a full healthy life.