CONDITION

Primary alveolar hypoventilation

Primary alveolar hypoventilation is impaired automatic breathing that causes carbon dioxide to build up without another identified cause.

WHAT IT IS
Reduced automatic breathing without another identified cause

BODY SYSTEM
Brain and breathing system

MAIN SYMPTOMS
Morning headaches, daytime sleepiness, unrefreshing sleep, poor concentration

WHAT CAUSES IT
Inherited in congenital cases; otherwise often unknown

HOW IT IS DIAGNOSED
Blood gases, sleep study, and exclusion of other causes

MAIN TREATMENT
Long-term assisted ventilation, mainly during sleep

CAN IT BE CURED?
No - breathing control usually remains impaired

IS IT AN EMERGENCY?
Sometimes - confusion, blue lips, or severe breathing difficulty

LONG-TERM OUTLOOK
Often manageable with consistent ventilation and specialist follow-up

Overview

Primary alveolar hypoventilation is a rare condition in which breathing is too shallow or too slow to remove enough carbon dioxide from the body. The alveoli are the tiny air sacs in the lungs where oxygen enters the blood and carbon dioxide leaves it.

Breathing is partly under conscious control when you are awake. It is also controlled automatically by the brain, including while you sleep. In primary alveolar hypoventilation, this automatic response is impaired. Breathing may be most affected during sleep, when conscious control is absent, but it can also become inadequate while awake.

Carbon dioxide can then build up in the blood. This is called hypercapnia . Blood oxygen may also fall. Persistent changes in these gases can affect sleep, thinking, energy levels, the heart, and other organs.

"Primary" means the hypoventilation is not explained by another condition such as severe lung disease, obesity hypoventilation syndrome, a chest-wall disorder, muscle weakness, or a medicine that suppresses breathing. The term may include inherited congenital central hypoventilation and cases that begin later without an identified cause. Specialists may use more specific names after testing.

The condition is different from ordinary snoring or uncomplicated obstructive sleep apnea. It needs specialist assessment because a person can have high carbon dioxide without feeling obviously short of breath.

Symptoms

Symptoms often appear or become worse during sleep. They can be subtle and may resemble poor sleep, stress, or another medical problem.

Possible symptoms include:

Some people have few symptoms despite a significant rise in carbon dioxide. A family member may instead notice shallow breathing, long pauses, unusual sleepiness, or a change in alertness.

In congenital central hypoventilation syndrome, symptoms often begin in infancy or childhood. Some milder inherited forms are not recognised until later, sometimes after an infection, anaesthesia, or use of a medicine that slows breathing.

- Morning headaches
- Daytime sleepiness or unusual tiredness
- Unrefreshing or restless sleep
- Poor concentration, memory problems, or slower thinking
- Shortness of breath, especially during sleep or illness
- Reduced exercise tolerance
- Frequent waking or difficulty staying asleep
- Bluish or grey lips or fingertips when oxygen is very low

Causes and risk factors

Automatic breathing is regulated by networks in the brainstem, the lower part of the brain connected to the spinal cord. These networks respond to rising carbon dioxide and falling

oxygen by increasing breathing. In primary alveolar hypoventilation, that response is weaker than expected.

Congenital central hypoventilation syndrome is usually caused by a change in the PHOX2B gene. This gene helps the automatic nervous system develop. That system controls functions you do not have to think about, such as breathing, heart rate, and digestion. The genetic change is often new in the affected person, although it can be inherited.

Some cases begin in adulthood and have no identifiable genetic or structural cause. These may be described as idiopathic central alveolar hypoventilation. "Idiopathic" means the cause remains unknown after appropriate investigation.

Before calling the condition primary, the healthcare team must look for other causes of hypoventilation. These include:

These are alternative causes rather than risk factors for the primary condition.

- Lung, airway, chest-wall, nerve, or muscle disorders
- Severe obesity with sleep-related breathing failure
- Brainstem injury, inflammation, or a mass
- Low thyroid hormone or another metabolic disorder
- Opioids like oxycodone or morphine , sedatives, or other substances that suppress breathing

Diagnosis

Diagnosis usually involves a respiratory or sleep specialist. No single symptom can confirm primary alveolar hypoventilation.

A blood gas test measures carbon dioxide, oxygen, and the blood's acid balance. Blood may be taken from an artery or, in some settings, from a warmed capillary sample. Persistently raised carbon dioxide supports a diagnosis of hypoventilation.

An overnight sleep study, called polysomnography , records breathing, oxygen levels, heart rate, sleep stages, and body movements. Carbon dioxide should also be monitored, because an oxygen reading alone can miss inadequate ventilation. The study can show whether hypoventilation occurs only during sleep or also while awake. It also helps distinguish the condition from obstructive or central sleep apnea.

Other tests are used to rule out secondary causes. They may include lung function testing, chest or brain imaging, blood tests, a medicine review, and tests of nerve or muscle function.

PHOX2B genetic testing is important when congenital central hypoventilation is suspected, including in some people whose symptoms first become clear later in life. Genetic counselling can help explain the result and what it may mean for relatives.

Because this is a diagnosis of exclusion, an unexplained high carbon dioxide result is not enough on its own. The full pattern and possible alternative causes must be assessed.

Treatment and management

Treatment supports breathing so the body receives enough oxygen and removes enough carbon dioxide. It does not usually restore normal automatic breathing control.

Many people use non-invasive ventilation while sleeping. A fitted mask is connected to a machine that provides breaths or pressure support. The machine often has a backup rate, allowing it to deliver a breath if the person does not breathe often enough.

People with more severe impairment may need ventilation for longer periods, including while awake. Some require ventilation through a tracheostomy, which is an opening made at the front of the neck into the windpipe. The most suitable method depends on age, severity, daily activities, and whether ventilation is needed only during sleep.

Diaphragm pacing may be considered in selected people. This uses electrical stimulation to move the diaphragm, the main breathing muscle. It requires specialist assessment and surgery and is not appropriate for everyone.

Supplemental oxygen may be added in particular circumstances, but oxygen alone does not correct poor ventilation or remove carbon dioxide. Using oxygen without monitoring can hide worsening hypoventilation.

Ongoing management commonly includes:

Alcohol, opioids, sleeping tablets, and sedating medicines can further reduce breathing. They should be discussed with the prescribing healthcare professional rather than stopped suddenly without advice.

- Regular checks of carbon dioxide and oxygen
- Repeat sleep studies or overnight monitoring
- Review of the ventilator, mask, and stored machine data
- Heart assessment when clinically needed
- Planning for illness, surgery, anaesthesia, travel, and power failure

Complications

Untreated or inadequately supported hypoventilation can cause persistently low oxygen and high carbon dioxide.

Over time, this may lead to pulmonary hypertension, meaning high pressure in the blood vessels of the lungs. The right side of the heart may then have to work harder and can eventually weaken. Heart rhythm problems may also occur.

High carbon dioxide can cause headaches, reduced alertness, confusion, and impaired concentration. Severe rises can lead to respiratory failure or loss of consciousness.

Repeated low oxygen may increase the number of red blood cells, making the blood thicker. Poor sleep and daytime sleepiness can affect learning, work, driving, and emotional wellbeing.

People with PHOX2B-related congenital central hypoventilation can have other problems involving automatic body functions. These may include altered heart-rate control, digestive difficulties, changes in pain or temperature awareness, and, in some cases, tumours arising from developing nerve tissue. The risks differ between genetic variants, so follow-up is tailored to the confirmed diagnosis.

Prevention

Primary alveolar hypoventilation usually cannot be prevented. In inherited cases, the genetic change is not caused by anything a parent did during pregnancy. Cases with no identified cause also have no established prevention method.

Genetic counselling can explain inheritance, testing for relatives, and reproductive options when a PHOX2B variant is found. This does not prevent the condition in someone who already has it.

Complications may be reduced through consistent ventilatory support and regular monitoring. Having a plan for respiratory infections, anaesthesia, sedating medicines, travel, and equipment failure can help avoid periods of inadequate breathing.

Living with it and what to expect

Primary alveolar hypoventilation is generally a long-term condition. Breathing support may be needed every night and sometimes during the day. The underlying breathing-control problem does not usually disappear, even when treatment controls carbon dioxide and oxygen well.

Needs may change over time. An infection, surgery, pregnancy, weight change, or sedating medicine can increase the amount of support required. Children need repeated equipment adjustments as they grow. Adults may also need mask or ventilator changes to maintain comfort and effective treatment.

With reliable ventilation and specialist follow-up, many people study, work, exercise, travel, and take part in family life. Daily routines need to include equipment care and planning. Backup power, spare supplies, and clear emergency information are especially important for anyone who depends heavily on a ventilator.

Follow-up continues even when you feel well, because carbon dioxide can rise without obvious symptoms. Appointments may review sleep quality, alertness, heart health, equipment performance, and any effects of an inherited autonomic condition.

The long-term outlook is influenced by how much ventilation is needed, whether treatment is used consistently, and whether heart or nervous-system complications are present. Your own course cannot be predicted from the condition's name alone.

When to get medical help

Arrange routine follow-up if you develop more morning headaches, daytime sleepiness, poor concentration, breathlessness, or reduced exercise tolerance. Also seek advice if a mask leaks, the ventilator feels uncomfortable, alarms recur, or you are no longer able to use the equipment for the planned amount of time.

Contact your specialist team before planned surgery or sedation. The anaesthesia team needs to know about the condition and any ventilatory support you use. Ask for advice before taking a new opioid, sleeping medicine, or other sedating drug.

Seek urgent medical assessment if you become unusually drowsy, confused, hard to wake, or noticeably more short of breath. A respiratory infection accompanied by worsening sleepiness or ventilator problems also needs prompt attention.

Call emergency services for severe breathing difficulty, blue or grey lips, collapse, a seizure, loss of consciousness, or failure of essential ventilator equipment when there is no effective backup. Use the emergency plan provided by your specialist service while help is coming.

Common questions

Is primary alveolar hypoventilation the same as sleep apnea?

No. Sleep apnea causes repeated pauses or reductions in airflow. Hypoventilation means overall breathing is not deep or frequent enough to remove carbon dioxide. The two problems can occur together, which is why sleep testing includes several breathing measurements.

Can I have high carbon dioxide without feeling breathless?

Yes. A gradual rise may cause headache, sleepiness, poor concentration, or no clear symptom. Breathlessness is not a reliable measure of carbon dioxide, so blood gas and overnight monitoring are important.

Will oxygen fix primary alveolar hypoventilation?

Oxygen can raise the blood oxygen level, but it does not make breathing deeper or remove excess carbon dioxide. Assisted ventilation is the main form of breathing support. Oxygen is used only when the healthcare team determines that it is also needed.

Is primary alveolar hypoventilation inherited?

Some forms are. Congenital central hypoventilation syndrome is usually linked to a PHOX2B gene change. Other cases have no identified genetic cause. Genetic testing and counselling can clarify whether relatives may be affected.

Can primary alveolar hypoventilation be cured?

There is currently no treatment that reliably restores normal automatic breathing control. Ventilation can manage the abnormal gas levels and reduce complications, but it is usually needed long term.

Can I travel with ventilatory support?

Many people can travel with careful preparation. Plans may include backup equipment, batteries, power adapters, medical documentation, and arrangements with the transport provider. High altitude and air travel can affect oxygen levels, so individual planning with the specialist team is important.

Sources

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