CONDITION

Primary ciliary dyskinesia

Primary ciliary dyskinesia is an inherited condition in which tiny airway hairs do not clear mucus properly.

WHAT IT IS
Inherited condition affecting mucus-clearing cilia

BODY SYSTEM
Lungs, airways, ears, sinuses, and reproductive system

MAIN SYMPTOMS
Wet cough, blocked nose, ear infections, breathing problems

WHAT CAUSES IT
Inherited gene changes

HOW IT IS DIAGNOSED
Genetic testing and specialized cilia structure or function tests

MAIN TREATMENT
Airway clearance, antibiotics, and specialist monitoring

CAN IT BE CURED?
No - inherited cilia changes remain lifelong

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

LONG-TERM OUTLOOK
Often manageable, but lung damage can progress over time

Overview

Primary ciliary dyskinesia, often shortened to PCD, is an inherited condition affecting microscopic structures called motile cilia. Cilia line the nose, sinuses, airways, middle ears, and some parts of the reproductive system.

Healthy cilia beat in a coordinated pattern. This moves mucus, germs, and inhaled particles out of the lungs and airways. In PCD, the cilia may not move, may move poorly, or may beat in an uncoordinated way. Mucus then builds up, making repeated infections and inflammation more likely.

PCD is present from birth, although it may not be recognized until later. Some babies have unexplained breathing problems soon after delivery. Children and adults often have a daily wet cough, long-lasting nasal congestion, repeated ear infections, or hearing problems.

Over time, repeated chest infections may damage and widen the airways. This is called bronchiectasis. Early diagnosis and consistent care can help protect lung function, manage symptoms, and reduce complications.

Symptoms

Symptoms often begin during infancy, but their pattern and severity differ between people. A cough caused by PCD is usually wet or mucus-producing and happens throughout the year, rather than only during a cold.

Common features include:

Many full-term babies with PCD develop rapid breathing or need oxygen shortly after birth. Because several conditions can cause these problems, this symptom alone does not confirm PCD.

Cilia also help position organs while an embryo develops. Around half of people with PCD have a mirror-image arrangement of some or all internal organs, called situs inversus. PCD with situs inversus and bronchiectasis has historically been called Kartagener syndrome.

A smaller group has a more complex organ arrangement called heterotaxy. This may occur with congenital heart, spleen, or intestinal differences that need separate assessment.

In adults, PCD may affect fertility. Sperm may not move normally. In the fallopian tubes, impaired cilia can make it harder for an egg to travel toward the uterus. Fertility problems do not affect everyone with PCD.

- A daily wet cough beginning in early childhood
- Constant or frequent nasal blockage and discharge
- Repeated sinus, chest, or middle-ear infections
- Hearing loss caused by fluid behind the eardrum
- Shortness of breath, wheezing, or reduced exercise tolerance
- Bronchiectasis developing during childhood or adulthood

Causes and risk factors

PCD is caused by changes in genes involved in building or controlling motile cilia. Many different genes can cause the condition.

Most forms follow an autosomal recessive inheritance pattern. This means a person inherits

one altered copy of the same gene from each parent. The parents are usually healthy carriers. Less commonly, PCD can follow X-linked or autosomal dominant inheritance patterns.

Having a close relative with PCD increases the chance of having the condition. However, a person can be the first diagnosed member of a family.

PCD is not caused by smoking, infections, parenting, or anything someone did during pregnancy. It is not contagious. Smoke and air pollution can still irritate the lungs and worsen respiratory symptoms.

Diagnosis

PCD can be difficult to diagnose because its symptoms overlap with asthma, cystic fibrosis, immune conditions, and ordinary childhood infections. There is no single test that identifies every case.

Assessment usually takes place at a center experienced in PCD. The process begins with a detailed history, including symptoms during the newborn period, daily cough or nasal congestion, repeated infections, hearing problems, and unusual organ positioning.

Tests may include:

Other tests may measure lung function, check hearing, culture mucus for bacteria, or look for bronchiectasis with a chest CT scan. A sweat test is often used to check for cystic fibrosis, which can cause similar symptoms.

A negative genetic test or normal-looking cilia sample does not always rule out PCD. Results need to be interpreted together by specialists.

- Nasal nitric oxide testing: People with PCD often have very low levels of this gas in the nose. The test is most reliable in people old enough to follow breathing instructions. A low result supports the diagnosis but does not confirm it by itself.
- Genetic testing: A blood or saliva sample can be checked for gene changes known to cause PCD. Finding disease-causing changes can confirm the condition, but current testing does not identify every genetic cause.
- Cilia structure testing: A small sample from inside the nose or airway is examined using electron microscopy, a powerful microscope. Some types of PCD cause recognizable structural changes, while others look normal under this test.
- Cilia movement testing: High-speed video microscopy can examine how sampled cilia beat. Infection can temporarily change cilia movement, so testing may need to be repeated.

- Immunofluorescence testing: Special stains can show whether important cilia proteins are present and in the correct place.

Treatment and management

There is no treatment that corrects the inherited cilia changes. Management focuses on clearing mucus, treating infections, protecting hearing, and limiting further lung damage.

Airway clearance is a central part of care. A respiratory physiotherapist can teach techniques that move mucus upward so it can be coughed out. These may include controlled breathing, positive expiratory pressure devices, or other chest physiotherapy methods. Regular physical activity can also help mobilize mucus, but it does not replace an individualized airway-clearance routine.

Chest infections are usually treated with antibiotics like amoxicillin or doxycycline . The choice and length of treatment are based on symptoms, previous mucus cultures, allergies, and local practice. Some people with frequent flare-ups may be offered long-term antibiotics after specialist review.

Mucus samples are checked for bacteria because PCD airways can develop long-term infection. Treatment may change if particular bacteria continue to appear.

Other parts of care can include:

Inhaled medicines used for asthma are not automatically helpful in PCD. They may be used when testing shows asthma or reversible airway narrowing. Any medicine or mucus-thinning treatment should be discussed with the PCD team because evidence and individual benefit differ.

- Keeping routine immunizations, including respiratory infection vaccines, up to date
- Saline nasal treatments for thick mucus or sinus symptoms
- Hearing tests and support for hearing loss
- Ear, nose, and throat assessment for persistent problems
- Fertility advice or assisted reproduction when needed
- Lung transplantation assessment in uncommon cases of advanced lung failure

Complications

Repeated infection and inflammation can permanently widen the airways, causing

bronchiectasis. Widened airways hold more mucus, which can lead to a cycle of infection and further damage. Lung function may gradually decline, although the rate is not the same for everyone.

Other possible complications include chronic sinus inflammation, nasal polyps, persistent middle-ear fluid, and hearing loss. Hearing difficulties during childhood can affect speech, learning, and classroom participation if they are not recognized.

Some people cough up blood from inflamed airways. Small streaks can occur during an infection, but larger or repeated amounts require prompt medical assessment.

PCD can also cause reduced fertility. People with fallopian tube involvement may have an increased risk of a pregnancy developing outside the uterus, known as an ectopic pregnancy. Early medical assessment is important when pregnancy occurs.

People with heterotaxy may have heart, spleen, or digestive tract differences unrelated to lung clearance. The risks depend on which organs are affected.

Prevention

PCD itself cannot currently be prevented because it is inherited.

Some infections and complications may be reduced through regular airway clearance, prompt assessment of worsening chest symptoms, recommended immunizations, and scheduled specialist follow-up. Avoiding smoking and secondhand smoke helps limit additional airway irritation.

Handwashing and avoiding close contact with people who have active respiratory infections may reduce exposure, although it is not possible to avoid every infection.

Genetic counselling can explain the inheritance pattern in a particular family. It may also discuss testing for relatives and reproductive options. This provides information and support; it does not mean anyone is responsible for passing on the condition.

Living with it and what to expect

PCD is lifelong. Daily cough and nasal symptoms often continue even when there is no active infection. Airway-clearance treatment usually becomes part of the everyday routine.

Symptoms may become more noticeable during respiratory infections. A flare-up may cause increased coughing, a change in mucus amount or color, fever, tiredness, breathlessness, or lower exercise tolerance. Having an agreed plan can make it easier to recognize and respond

to these changes.

Regular care commonly includes lung function testing, mucus cultures, hearing checks, and review of airway-clearance techniques. Follow-up continues even when symptoms seem stable because infection or lung changes may develop gradually.

Many people with PCD attend school, work, exercise, travel, and have families. Treatment routines, hearing problems, fatigue, and repeated appointments can still affect daily life. Schools or workplaces may be able to allow time for airway clearance, medical visits, and recovery from infections.

The long-term course cannot be predicted from symptoms alone. Some people maintain relatively stable lung function for years. Others develop progressive bronchiectasis or significant breathing limitations. Consistent specialist care aims to preserve lung health rather than remove the underlying condition.

When to get medical help

Arrange routine follow-up with the PCD team for ongoing cough, sinus symptoms, hearing concerns, treatment review, fertility questions, or changes in exercise ability.

Seek prompt medical advice if you have:

Call emergency services for severe difficulty breathing, blue or grey lips, confusion, fainting, or a large amount of coughed-up blood. These are emergency signs rather than routine features of stable PCD.

- A clear increase in cough or mucus
- Mucus that changes color, thickness, or amount
- Fever, chest pain, or worsening breathlessness
- Coughing up blood, even if the amount is small
- Symptoms that are not improving with an agreed treatment plan
- A positive pregnancy test with one-sided pelvic pain or bleeding

Common questions

Is primary ciliary dyskinesia the same as cystic fibrosis?

No. Both conditions can cause thick mucus, repeated chest infections, and bronchiectasis, but they have different genetic causes. Cystic fibrosis affects salt and water movement across

cells. PCD affects the structure or movement of motile cilia.

Is PCD contagious?

No. PCD is inherited and cannot spread between people. The viruses and bacteria causing respiratory infections can still be contagious.

Why are some people with PCD born with their organs reversed?

During early development, specialized cilia help establish the left and right sides of the body. If these cilia do not work normally, organs may develop in a mirror-image or mixed arrangement. A complete mirror-image arrangement may cause no problems by itself, although associated organ differences need assessment.

Can someone with PCD have children?

Yes. Some people conceive without help, while others have reduced fertility. Assisted reproductive treatments may be an option. Anyone with PCD who could become pregnant can discuss fertility and early pregnancy monitoring with an appropriate specialist.

Does PCD shorten life expectancy?

PCD can cause progressive lung damage, but its course differs widely. Better recognition, airway clearance, infection treatment, and specialist monitoring have improved care. An individual outlook is shaped mainly by lung health, infections, organ differences, and response to management.

Can exercise help PCD?

Exercise can improve fitness and help loosen airway mucus. It is generally used alongside, not instead of, regular airway-clearance treatment. The most suitable activities depend on current lung health and physical ability.

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