Learn About PCD

Primary ciliary dyskinesia (PCD) is a rare genetic (inherited) condition that can lead to chronic ear, sinus, pulmonary disease (bronchitis/bronchiectasis), reversed or flipped organs (situs inversus), and fertility issues.

Key Facts
- PCD is a genetic condition.
- People with this condition have mutations in genes that control the structure and function of cilia.
- When cilia do not beat properly, the affected individual is prone to infections in the ears (otitis), sinuses (sinusitis), and the lung (bronchitis).
- Eventually, bronchiectasis develops in the lungs.

What Is PCD?
PCD is passed from healthy parents to their children through their genes. People who inherit PCD lack certain building block proteins in cilia (tiny hair-like structures that beat constantly to “clean” the lining tissue of the ears, nose, and bronchial tubes, as well performing important functions in other tissues). This causes the cilia to not beat properly, and they fail to clear inhaled particles and bacteria from the lung. This leads to infection and inflammation.

How PCD Affects Your Body
In people with PCD, cilia do not beat properly, allowing mucus to build up and making them prone to infections of the airways (a chronic “wet” cough, bronchitis, pneumonia) from birth, as well as chronic, reoccurring ear, and sinus infections. Lung disease may progress over time and is worsened by environmental exposures such as smoke. This leads to bronchiectasis, where the airways are irreversibly damaged, and harbor excessive, sticky mucus, which is an ideal environment for bacteria.

Other symptoms may include reduced hearing from chronic damage to the middle ear and a chronic runny nose, or pain, and stuffed sinuses. Situs inversus sometimes known as “Kartagener’s syndrome” (reversed organs: heart on right, liver on left, and so on) is usually silent (sometimes people are diagnosed with PCD when situs inversus is noticed on an X-ray). Less commonly, babies may be born with more complex organ placement, a condition known as “heterotaxy,” which may include congenital heart disease due to abnormal valves or blood vessels. Women may experience difficulty getting pregnant or have tubal pregnancies, and almost all men are infertile as their sperm do not swim properly (like cilia, the sperm tails don’t beat).

How Serious Is PCD?
PCD is estimated to occur in 1 about every 15,000 to 20,000 individuals, occurring worldwide, although the disease is underrecognized. Although there are many symptoms associated with PCD, the most serious complication is bronchiectasis, which, in some individuals, may cause serious obstructive lung disease, and even respiratory failure. These people may need to depend on oxygen, breathing machines, or even sometimes lung transplantation.
- PCD is a cause of bronchiectasis and is usually underrecognized until it is relatively severe.
- It is often first diagnosed as asthma, bronchitis, or bronchiectasis of unknown cause (idiopathic).
With good care, people with PCD remain relatively healthy throughout their lives despite having some impairment of quality of life, mainly with chest infections. Although research is ongoing, and many questions remain to be answered, it is highly likely that early diagnosis, good preventive health-care, early treatment of infections, and avoiding other risk factors, such as cigarette smoking, can help prevent PCD from progressing to becoming very serious.

**PCD Symptoms, Causes, and Risk Factors**

PCD may first be diagnosed as asthma, chronic bronchitis, smoking-related COPD, or just bronchiectasis of unknown cause. Since PCD is an inherited condition, it is important to consider your family history of lung, sinus, or ear disease as a possible indication of PCD, or a family history of reversed organs.

**What Are the Symptoms of PCD?**

Typically, symptoms occur very early in life, either shortly after birth (“neonatal” pneumonia, for example), or in early childhood. Symptoms persist throughout life.

Symptoms of PCD can be classified as symptoms involving the ears, sinuses, and lung, and symptoms involving organs outside the respiratory tract.

The most common symptoms of PCD respiratory disease are:

- A “wet” cough from birth or infancy, with or without mucus production that lasts for a long time
- Shortness of breath
- Wheezing or a whistling sound while you breathe
- Recurring chest colds
- Asthma that does not respond to standard therapy
- Middle ear infections
- Chronic nose infections, thick nasal drainage, or sinusitis

Symptoms of lung disease usually may occur in newborns, very young children, or young adults, but it is not uncommon for the diagnosis to be delayed until adulthood. Ear symptoms may be particularly difficult in young children until the ear drainage tubes grow with age.

**What Causes PCD?**

PCD is an inherited disorder. PCD is caused by having two abnormal copies of the PCD gene due to inheriting an abnormal copy from both parents. You cannot catch or acquire PCD. It can occur in people who have no known family history of the disease because people with one abnormal PCD gene (called “carriers”) are usually healthy. When a person has two abnormal copies of the gene, they do not make one of a number of the proteins that allow cilia to beat normally. It is important to know that not everyone who inherits PCD will have the same symptoms or severity or have situs inversus (reversed organs). In fact, only half of people with PCD have situs inversus. Some gene mutations are more common than others, and one's ethnic background may influence what gene mutations occur in a given patient or family. Researchers are working to learn more about what causes PCD. If a patient, family, or clinician wish to know about gene testing for PCD, it is best to contact a specialized center for the latest news on how to get that done.

**What Are Risk Factors?**

- **Genes:** The genetic status of a person's parents determines whether they are at risk for PCD. A person is only at risk if both parents have one or more PCD gene abnormalities. However, because this is often not known, anyone with significant recurrent lung infections, or bronchiectasis of unknown cause with organ-sided issues, should consider PCD testing.
Early diagnosis: Early diagnosis of PCD is important because early treatment is considered essential to help slow the progression of PCD lung disease.

Environment and lifestyle: As with most diseases, it’s not all about genes. There are other factors that are involved that may influence the progress of the disease, especially environmental influences. These include:
- Not smoking
- Avoiding direct contact with people who have a cold or chest infection
- Receiving appropriate vaccines like the flu shot
- Eating well and staying active
- Practicing good “airway clearance”

Age: The course of PCD over time is unpredictable, as some patients do very well for a full lifetime, while others have serious lung disease by the time they are young adults.

When to See Your Doctor
If you have a family history of PCD, have been diagnosed with bronchiectasis or sinus inversus, or experience any of the above symptoms or other symptoms, you should consult your doctor and think about seeing a lung specialist.

Diagnosing and Treating PCD
Although there is no proof that early diagnosis is important, it is likely to be highly beneficial, as it is with other chronic diseases. It stands to reason that early intervention should help to prevent early and long-term deterioration.

What to Expect
Generally, a careful history obtained by an experienced clinician is the first step in diagnosing PCD, followed by a careful examination and targeted tests. Such tests should include basic blood work, sputum tests to examine for the typical bacteria seen in PCD, as well as a chest X-ray and probably a CT scan of the chest. Breathing tests (pulmonary function tests [PFTs]) will help the clinician assess for disease of the airway (obstructive lung disease). Often, at the end of these preliminary tests, the clinician should be in a position to say whether PCD is likely or not, although a specific diagnosis may not be possible at this point. More specific tests will be necessary to try to make the diagnosis.

How PCD Is Diagnosed
Since about the mid-1970s, the diagnosis of PCD relied on getting samples of the cells that contain cilia at their surface – either from the nose or the airways (windpipe or bronchial tubes). The cilia can be analyzed using special microscopes for the presence or absence of the proteins that help cilia beat normally. However, more recently, other tests have become available, such as measuring air extracted from the nasal passages for levels of nitric oxide. For reasons that are not clear, people with PCD have very low levels of nitric oxide gas in their nasal air. Although testing nitric oxide levels is a very easy test, it is only available at hospitals and a few outpatient facilities that have the equipment. Other tests include genetic tests, usually on blood samples. The number of genetic tests for PCD is steadily increasing, and it is likely that in the near future, more genetic tests will become available to
diagnose the people who have PCD. Commercial genetic testing companies have begun to offer testing for some of the known genetic mutations; however, there is a cost for this type of testing.

How PCD Is Treated
Although there is no specific treatment that helps the cilia work properly, there are treatments for the effects on patients of having PCD. These treatments include:

- Antibiotics to address lung or sinus infections.
- Airway clearance techniques. These include breathing and coughing techniques, usually with assistance of physical therapy or airway clearance devices. These techniques need to be done frequently to help the lung stay clear.
- New medicines are being tested; it is important to know whether PCD or another disease is the cause of bronchiectasis; usually patients with PCD are included in such studies.

Probably the most important aspects of PCD are to:

- See the doctor regularly.
- Have tests of breathing and sputum (for infections).
- Practice regular airway clearance.
- Keep good health practices and follow the doctor’s advice.

Living With PCD
For the most part, people with PCD who are diagnosed relatively early and have proper care should live a long life with good quality.

What to Expect
PCD usually requires daily management, like most chronic disorders. The severity of PCD can vary greatly from person to person. The key is deciding whether you will manage or disease or the disease will manage you.

Managing the PCD
Recurrent infections tend to be unpredictable, and usually need antibiotic treatment. In severe cases you might need antibiotics through an IV for a few weeks at a time. This can sometimes be done at home, but might mean a stay in the hospital. When in hospital, you will likely receive airway clearance with physical therapy. How often this occurs during a lifetime with PCD is unpredictable and varies from one person to another – this could be once every few years with mild PCD, to a few times with year with more severe disease.

It might be necessary to see an ear, nose, and throat (ENT) doctor if sinus and ear problems are difficult to manage. Then, the doctor might recommend cleaning methods for the nose and sinuses (“nasal rinses”) or even surgery. Some doctors recommend inserting tubes to drain the ear passages, but this is controversial, even among experts. They are not usually needed in adults with PCD.

Occasionally, even with the best of care, PCD may lead to severe lung disease that needs more intense treatment. Oxygen treatment might be needed at night or during exercise. With very severe disease, breathing assist devices might be needed to help with breathing, and if the lungs are failing badly, doctors might suggest an assessment for a lung transplantation. While few patients with PCD go on to transplant, it can be effective with very severe diseases.
Finding Support
The Lung Association recommends patients and caregivers join our Living with Lung Disease Support Community to connect with others facing this disease. You can also call the Lung Association’s Lung HelpLine at 1-800-LUNGUSA to talk to a trained respiratory professional who can help answer your questions and connect you with additional support.

Ask your health-care provider about lung disease support groups in your area, or look online for a Better Breathers Club near you.

Questions to Ask Your Doctor About PCD
Making notes before your visit, as well as taking along a trusted family member or friend, can help you through the first appointment with your doctor.

- Could I have PCD?
- Why do you think I could have PCD?
- Do I need special tests for PCD?
- What do I need to do to keep myself as healthy as possible for as long as possible, especially my lungs?
- Do I have any infections that might need treatment?
- Do I need, or will I need, intravenous (drip) antibiotics?
- Will I need a lung transplant at any stage?
- How about having children? Will I be able? Will they have PCD?
- Do I need to take any special precautions to stay healthy?