PCD FOUNDATION™

PRIMARY CILIARY DYSKINESIA

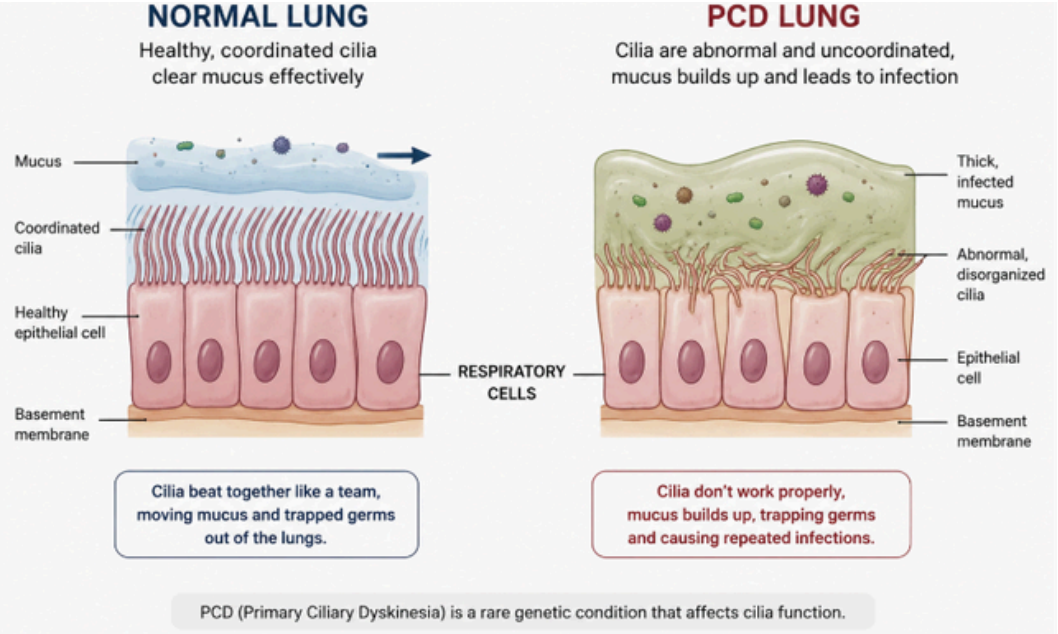
# Primary Ciliary Dyskinesia (PCD)

## The Basics

## What Happens in PCD?

PCD is a rare, genetic disease caused by changes (called “variants”) in genes responsible for ciliary structure or function. Changes in more than 60 different genes can cause PCD. In most cases, PCD is inherited, which means it’s passed down from a person’s biological parents. But in some rare cases, the gene changes happen for the first time (called ‘de novo’) in the person with PCD and were not inherited from their parents.

Cilia are microscopic ‘organelles’ (little organs) that beat in a coordinated fashion to help remove excess mucus and the dust, bacteria, and unwanted particles that we inhale every day from the airway. Functioning cilia are critical for effective mucociliary clearance.




## What is Mucociliary Clearance and Why is it Important?

When you breathe, you take in the oxygen your body needs, also unwanted dust, particles, and germs. To keep your airways healthy, your cilia move in a wave-like motion to push mucus, (which traps the unwanted stuff) out of your lungs and sinuses. This process is called mucociliary clearance.
This process depends on two main things:

1. Cilia that move well
2. Mucus that’s the right thickness and amount

In people with PCD, the cilia don’t move the right way. Because of this, mucus builds up, gets thicker, and becomes harder to clear out of the airways. This makes it easier for infections to happen. Over time, repeated infections can damage the lungs and airways, causing breathing problems and a loss of lung function.

## How Rare is PCD?

PCD is a rare disease. It affects about 1 in every 7,500 to 15,000 people. But the symptoms of PCD—like frequent sinus and lung infections—are common, so it can take a long time to get the right diagnosis. Many people with PCD see over 50 doctors before being diagnosed. Even though PCD starts at birth, it often isn't found until later in childhood or even adulthood.

## What Body Systems are Affected by PCD?

The lungs, sinuses, and ears are the areas most commonly affected by PCD, because they depend on cilia (to help clear mucus. Cilia also help with:

- Organ placement & development – Cilia are critical for directing organ placement & development in the embryo. Some people with PCD have organs that are not in their usual positions or that did not develop properly. Sometimes these changes affect only a single organ (e.g. dextrocardia) or they can affect many organs. When laterality ('sided-ness') defects are present, the affected person may be told they have PCD with situs inversus totalis (all organs in mirror-image, includes dextrocardia) or PCD with situs ambiguus/heterotaxy which describes any organ arrangement that is not typical (called situs solitus) or situs inversus totalis. The important thing to know about laterality/situs defects in PCD is that they are random. People with PCD may present with any of the three types of laterality. The issue is lack of ciliary motility caused by PCD.

- Reproductive health – Cilia are important for both the male and female reproductive systems., Many people with PCD may have trouble getting pregnant. Men often have infertility, and women may have reduced fertility (subfertility).

- Other rare effects – PCD may co-occur with other disorders, and in very rare cases, this may result in additional symptoms. These include a rare eye condition called retinitis pigmentosa (which can lead to vision loss) and hydrocephalus, a condition where fluid builds up in the brain, or enlarged brain ventricles.

VIEW MORE SCIENTIFIC PICTURES ON PCD:

HTTPS://WWW.ABOUTKIDSHEALTH.CA/PRIMARY-CILIARY-DYSKINESIA-PCD

VIEW MORE VIDEOS ON PCD:

HTTPS://WWW.PCDFOUNDATION.ORG/VIDEO-LIBRARY/

## What Are the Symptoms of PCD?

PCD is a lifelong condition. Symptoms often begin at birth and continue throughout life. There is no cure, so treatment focuses on preventing lung damage and managing daily symptoms.

### COMMON SYMPTOMS IN CHILDREN

1. Breathing problems as a newborn
2. Ongoing cough with noisy breathing (all year)
3. Ongoing nasal mucus (all year)
4. Frequent ear infections
5. Hearing problems
6. Pneumonia or bacterial bronchitis
7. Organ placement issues (laterality defects)

### COMMON SYMPTOMS IN ADULTS

1. Long-term cough with lots of mucus
2. Lung damage such as bronchiectasis
3. Worsening lung function
4. Frequent sinus and lung infections
5. Serious sinus disease (sometimes requiring surgery)
6. Ongoing ear and hearing problems
7. Use of oxygen or lung transplant in very advanced cases
8. History of infertility or trouble getting pregnant
9. Organ placement issues

## How is PCD Diagnosed?

Doctors begin by reviewing your medical history, symptoms, and family history. For children, diagnosis often includes at least 2 of these 4 signs:

1. Daily, year-round cough starting in infancy
2. Daily, year-round nasal congestion or chronic sinusitis
3. Organ placement differences (laterality defects)
4. Breathing problems at birth (neonatal respiratory distress)

About 80% of children with PCD also have recurrent ear infections, but these are common in many children, so they're not as helpful for diagnosis alone.
In adults, the same signs are looked at, along with:

1. Evidence of bronchiectasis
2. History of infertility or subfertility

If PCD is suspected, doctors will order several tests to confirm it, and you may need more than one.

## Main Tests to Help Diagnose PCD

1. Genetic Testing – This checks for known PCD gene variants. A positive test confirms PCD. A negative test doesn't rule it out.
2. Ciliary Biopsy with a Transmission Electron Microscope (TEM) – A small sample of cilia is taken (usually from the nose) and examined under a powerful microscope. This should be done by a center with PCD expertise.
3. Nasal Nitric Oxide (NNO) Test – Most people with PCD have very low nitric oxide levels. This test should be done at a specialized center.
4. Other specialized tests available only at PCD specialist centers, like high-speed videomicroscopy or immunofluorescence.

If all tests are negative but you still have symptoms, doctors may need to repeat tests or look for other causes.

## What Kind of Care do People with PCD Need?

People with PCD should be treated by a specialized care team* who knows how to manage the condition. This team may include:

- You and your family
- A clinical coordinator (usually a nurse) to organize care
- A lung doctor (pulmonologist)
- An ENT (ear, nose, throat) doctor and audiologist
- A respiratory or physical therapist to help with breathing treatments
- A social worker to help with daily life challenges
- Mental health providers to support emotional well-being
- A dietitian to help with healthy eating and growth

## AIRWAY CLEARANCE THERAPY

Most people with PCD have daily mucus build-up, so they need airway clearance therapy every day. This may include:

1. Inhaled medicines to thin mucus
2. Physical techniques to move mucus like:
   - Chest physiotherapy/ percussion therapy
   - "The Vest," wearable devices that creates high-frequency vibrations on the chest
   - Positive Expiratory Pressure (PEP) devices
   - Exercise
3. Cough – the most important thing people with PCD can do is cough frequently to help clear excess mucus

People often do treatments twice a day—once in the morning (to clear mucus from the night) and once at night (to clear what builds up during the day).
Taking care of PCD can feel like a full-time job. That's why it's important to have a plan and support team. Many clinics give patients a chart to keep track of visits, treatments, and long-term health checks.