

PCD: Clues to Diagnosis

- Chronic ear, nose and pulmonary infections
- Neonatal respiratory distress of unknown cause
- *Situs inversus* and other organ anomalies (e.g. *situs ambiguus*, splenic, biliary and cardiac defects)
- Early Onset Bronchiectasis
- Hearing Loss
- Presence of Unusual Pathogens in Respiratory Cultures
- Subfertility or Infertility with a History of Respiratory Symptoms
- Negative Sweat Chloride Test and Immunodeficiency Studies



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The PCD Foundation

Facts About Primary Ciliary Dyskinesia (PCD)

Kartagener Syndrome
Immotile Cilia Syndrome
Ciliary Aplasia

What is PCD?

PCD is an acronym for primary ciliary dyskinesia. PCD is a unifying term used to describe a number of ciliary disorders including *Kartagener syndrome*, *immotile cilia syndrome*, and *ciliary aplasia*. The estimated incidence of inherited ciliary disorders ranges from 1:12,500 to 1:25,000. This means that roughly 20-25,000 Americans and as many as 400,000 people worldwide suffer from these disorders.

What are Cilia and What Do They Do?

Cilia are microscopic hair-like structures that line many internal body surfaces including the respiratory tract, sinuses, Eustachian tubes of the ear, ventricles of the brain, and the reproductive organs. There are approximately 10,000,000,000 cilia per square centimeter in the respiratory system, and they beat constantly at a rate of 5-50 beats or cycles per second. Cilia are an essential component of the mucociliary clearance activity required to sustain healthy respiratory tissue. The beating activity of the cilia moves debris-laden mucus out of areas vulnerable to infection or inflammation.

The beating motion of the cilia is also believed to be essential in determining organ placement during embryonic development. Roughly half the people affected by PCD will have a condition called *situs inversus*, in which their thoracic and abdominal organs are "flipped" to a mirror image position in the body. When *situs inversus* is present, the patient is diagnosed with *Kartagener syndrome*, a subcategory of PCD. Other abnormalities of the structure or function of abdominal/thoracic organs may be present in PCD, as well.

What Happens in PCD?

PCD is an inherited defect of the structure or function of cilia. The cilia in people with PCD do not function adequately (sometimes not at all). Respiratory difficulties are present almost from birth. Without functioning cilia, mucociliary clearance activity is profoundly impaired. Respiratory secretions begin to collect, thicken, and promote infection. Without aggressive treatment, permanent lung damage, such as bronchiectasis, develops at an early age. Without diagnosis and treatment, some PCD patients may require lung transplantation.

PCD is also associated with female subfertility and male infertility. Other (rare) complications include hydrocephalus, biliary atresia, complex congenital heart defects, and asplenia/polysplenia syndromes.

How is PCD Diagnosed?

There is no simple test for PCD. Diagnosis relies primarily on an assessment of ciliary ultrastructure done at a specialized laboratory or clinic. Clinical history and symptoms, and measurement of exhaled nasal nitric oxide are also aids in diagnosis.

How is PCD Treated?

The main goal of treatment in PCD is to minimize the damage caused by chronic infection and/or inflammation. Airway clearance therapy, including secretion removal and bronchodilation, and aggressive use of antibiotics are the most common forms of treatment. Other treatments are aimed at reducing or eliminating symptoms such as sinus pain and gastrointestinal upset.

Barriers to Treatment

When diagnosed early and treated appropriately, people with PCD can lead productive lives of nearly normal duration. However, lack of awareness about ciliary disorders frequently results in delayed diagnosis and unnecessary pulmonary damage. It is the goal of the PCD Foundation to raise the level of awareness about inherited ciliary disorders among healthcare professionals and patients, and to ensure access to prompt, appropriate diagnosis and treatment.

Where Can I Get More Information?

Diagnosing and treating PCD requires special expertise. Here are some online resources for additional information:

The PCD Foundation:

www.pcdfoundation.org

PCD Program at the University of North Carolina, Chapel Hill:

<http://pulmonary.med.unc.edu/PCD.htm>

UNC Pediatric PCD Site:

<http://pediatrics.med.unc.edu/div/infectdi/pcd/>

Children's Hospital of Eastern Ontario:

<http://www.cheo.on.ca/english/2013a3.html>

Leicester University PCD Site:

<http://www-micro.msb.le.ac.uk/mbchb/1c.html>

Genetic Diseases of Mucociliary Clearance:

<http://rdcm.epi.usf.edu/>

UK PCD Family Support Group

<http://www.pcdsupport.org.uk/>

German Kartagener/PCD Group

http://www.kartagener-syndrom.org/html_english/verein_wirueberuns.html