

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*, heterotaxy syndromes, congenital heart 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



Where Can I Get More Information?

Diagnoses and treatment of 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>

Genetic Diseases of Mucociliary Clearance Consortium:

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

UNC Pediatric PCD Site:

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

GeneTests PCD Review:

<http://www.geneclinics.or/query?dz=pcd>

Leicester University PCD Site:

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

UK PCD Family Support Group

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

German PCD Group

http://www.kartagener-syndrom.org/html_deutsch/sonstiges_weltweit.html

Children's Hospital of Eastern Ontario

<http://www.cheo.on.ca/english/9302a.shtml>

PCD Foundation
29252 N. 22nd Lane
Phoenix, Arizona 85085
612-396-1179
www.pcdfoundation.org
info@pcdfoundation.org

The PCD Foundation

Facts About Primary Ciliary Dyskinesia (PCD)

Kartagener Syndrome
Immotile Cilia Syndrome
Ciliary Aplasia

What is PCD?

PCD stands for **primary ciliary dyskinesia**. The term PCD is used to describe inherited disorders of motile (moving) cilia, including 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 15-20,000 Americans have PCD.

What are Motile Cilia and What Do They Do?

Motile 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. Collectively, these unusual organ arrangements are known as *situs ambiguus* (ambiguous) or heterotaxy.

What Happens in PCD?

PCD is an inherited defect of the structure or function of motile 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 a form of permanent lung damage called bronchiectasis may develop at an early age. Delays in proper diagnosis and treatment may increase the risk of developing end-stage lung disease.

PCD is also associated with female subfertility and male infertility. Other complications include complex congenital heart defects, pectus deformities and asplenia/polysplenia syndromes. Rare reports of hydrocephalus and biliary atresia have been published.

How is PCD Diagnosed?

Currently, the diagnosis of PCD 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.

Is Genetic Testing Available for PCD?

Recently, progress in identifying the genes responsible for PCD has resulted in the development of the first molecular (genetic) test for *DNAI1* and *DNAH5* mutations associated with PCD. Current genetic testing may only find mutations in 1/4 to 1/3 of PCD patients, so it will not replace ultrastructural studies for the majority of patients at this time. However, this testing may be useful as an aid to diagnosis of PCD or in testing family members of patients with known mutations. As more mutations are identified, the test will be modified to reflect new genetic information.

How Can I Get Tested for PCD?

PCD diagnostic testing must be arranged through a medical professional familiar with the disorder and with other conditions that may mimic the symptoms of PCD. The PCD Foundation can help you locate a physician with PCD expertise. Because PCD is a rare and difficult to diagnose disorder, travel to a large academic or specialized center for diagnosis may be required.

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.

Are There Barriers to Treatment?

Lack of awareness about PCD and limited access to qualified centers for diagnosis and treatment are a big challenge for individuals affected by PCD. The PCD Foundation was started by a dedicated group of patients, parents and health care professionals to raise the level of awareness about inherited motile ciliary disorders and to ensure access to prompt and appropriate diagnosis and treatment.

What is the Status of Research in PCD?

PCD is one of three disorders currently being studied through the Genetic Disorders of Mucociliary Clearance Consortium (GDMCC), a multi-center effort sponsored by the Office of Rare Diseases (ORD) and supported by the National Institutes of Health (NIH). For more information or to enroll in the research registry contact the PCD Foundation or see the GDMCC website (<http://rarediseasesnetwork.epi.usf.edu/gdmcc/index.htm>)