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

PCD FOUNDATION
primary ciliary dyskinesia

Where Can I Get More Information?

Diagnosis and treatment of PCD requires special expertise. Here are some online resources for additional information:

The PCD Foundation:
www.pcdfoundation.org

Genetic Disorders of Mucociliary Clearance Consortium (GDMCC)

www.rarediseasenetwork.org/gdmcc/

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The PCD Foundation

Facts About
Primary Ciliary Dyskinesia (PCD)

Kartagener Syndrome

'Immotile Cilia Syndrome'

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* and *immotile cilia syndrome*.

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. 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 motion of specialized cilia that form during embryonic development is important in determining organ placement. Roughly half of affected individuals have laterality defects including *situs inversus*, and *situs ambiguus.*

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 bronchiectasis may develop—even at an early age. Delays in proper diagnose and treatment may increase the risk of developing end-stage lung disease. Some patients progress to lung transplant

Chronic runny nose, pansinusitus and recurrent otitis media are common upper respiratory complications of PCD. The severity of these symptoms can debilitating and result in significant quality of life deficits.

How is PCD Diagnosed?

There is no single test that can reliably diagnosis all cases of PCD , so accurate diagnosis often requires multiple tests in addition to a careful medical history. Diagnostic workup at centers with PCD expertise and access to advanced testing approaches like nasal nitric oxide measurement and comprehensive genetic testing offer the best alternative for accurate diagnosis. nasal nitric oxide are also aids in diagnosis.

Genetic Testing for PCD

PCD is a genetically complex disorder and new genes are identified at a rapid pace. There are numerous options for PCD genetic testing from both commercial and academic providers. Check with your physician or the PCD Foundation for more information.

PCD Treatment

The main goal of treatment in PCD is to minimize 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.

Because PCD is a chronic, progressive disorder, affected individuals are strongly encouraged to be followed at a center with PCD or cystic fibrosis expertise.

PCD Research

Working closely with our research partners, the PCD Foundation is strongly committed to accelerating clinical research in PCD. Please contact the PCD Foundation or visit our website for information on current research opportunities.