PCD FOUNDATION™

PRIMARY CILIARY DYSKINESIA

Primary Ciliary Dyskinesia (PCD)

Resource Guide

What is PCD?

PCD stands for primary ciliary dyskinesia.

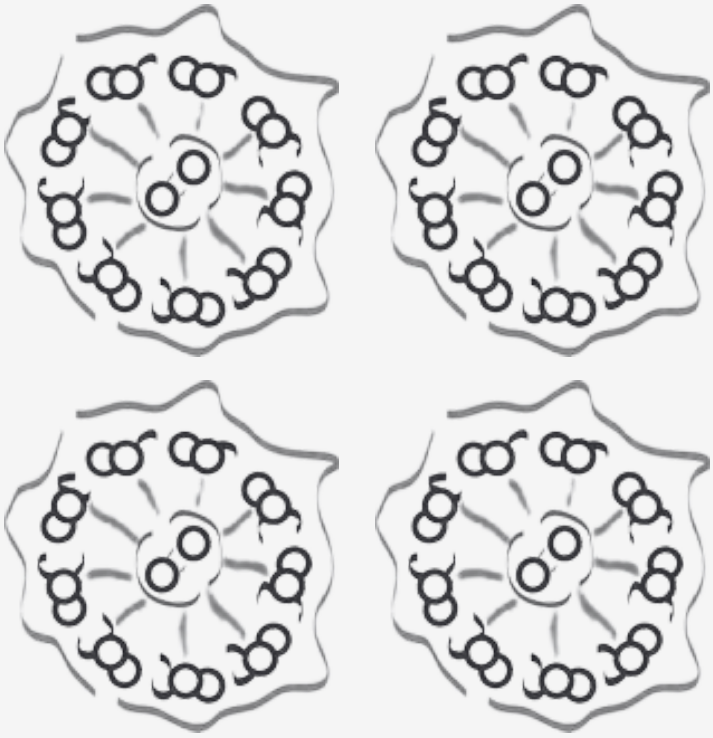

Image courtesy of Dr. Heymut Omran

The term PCD is used to describe inherited disorders of motile (moving) cilia. Synonyms for PCD include Kartagener syndrome and immotile cilia syndrome.

The estimated incidence of inherited ciliary disorders ranges from 1:7,500 to 1:15,000.

WHAT ARE MOTILE CILIA AND WHAT DO THEY DO?

Motile cilia are microscopic hair-like structures that line some internal body surfaces including the lower respiratory tract, sinuses, Eustachian tubes of the ear, ventricles of the brain, and the reproductive organs. Working with mucus, ciliary motion is an essential part of the mucociliary clearance activity required to sustain healthy lungs, ears and sinuses.

A unique type of motile cilia, nodal cilia, form during embryonic development and play an important role in determining organ placement and laterality (sided-ness). Roughly half of people with PCD have some type of laterality defect including situs inversus and situs ambiguus. Laterality issues in PCD are associated with congenital heart defects in a small percentage of patients.

What Happens in PCD?

PCD is an inherited defect of the structure or function of motile cilia. These defects result in cilia that do not function adequately, with consequent impairment in mucociliary clearance. Respiratory difficulties are present very early--often from birth. Respiratory secretions begin to collect, thicken, and promote infection. Without aggressive treatment, bronchiectasis - permanent scarring of the airway - develops, sometimes from a very early age. Delays in diagnosis and treatment may increase the risk of progression to end-stage lung disease. Some individuals with PCD will ultimately require lung transplant Chronic runny nose, sinusitus and recurrent otitis media (fluid behind the ear drum) are common upper respiratory complications of PCD. The severity of these symptoms can be debilitating and result in significant deficits in quality of life.

How is PCD Diagnosed?

There is no single test that can reliably diagnose all cases of PCD, so accurate diagnosis often requires multiple tests. The most important tool for diagnosis is 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 option for accurate diagnosis.

Genetic Testing for PCD

PCD is a genetically complex disorder and new genes are being identified at a rapid pace.

There are numerous options for PCD genetic testing from both commercial and academic provides. Check with your physician or the PCD Foundation for more information.

PCD: CLUES FOR DIAGNOSIS

·Respiratory problems in the neonatal period despite term birth

·Chronic year-round pulmonary, sinus and ear infections starting in the first year of life

·Chronic year-round rhinitis (runny nose) starting in the first year of life

·Situs inversus or other organ anomalies (situs ambiguus heterotaxy, congenital heart defects)

·Early onset bronchiectasis

·Hearing loss

·Presence of unusual pathogens in respiratory cultures

·Infertility/subfertility with a history of respiratory symptoms

·Negative sweat chloride and immunodeficiency studies

PCD Treatment

The main goal of treatment in PCD is to minimize damage caused by chronic infection and/or inflammation.

Airway clearance therapy to aid with mucus removal and help keep the airways open, coupled with aggressive treatment of infections are the most common forms of PCD therapy. 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 committed to accelerating clinical research in PCD to improve diagnosis, bring effective therapies to affected individuals, and ultimately find a cure for PCD. Please contact the PCD Foundation or visit our website for information on current research opportunities.

Where Can I Get More Information?

Primary ciliary dyskinesia (PCD) 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/cms/gdmcc

UK PCD Family Support Group
http://pcdsupport.org.uk/

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