WHAT IS PCD (Primary Ciliary Dyskinesia)?

Primary ciliary dyskinesia is an inherited disorder of microscopic, whip-like organelles (cilia) that line the upper and lower respiratory tract (including nasal passages, sinuses and lung, and eustachian tubes of the ear), the reproductive organs, and the ventricles of the brain. The activity of motile (moving) cilia, working in cooperation with airway mucus, provides a first line of defense for the airways, maintaining healthy lung tissue. This important biological system is known as '***mucociliary clearance***.' Defects of mucociliary clearance lead to profound illness. Cystic fibrosis is an example of another devastating genetic disorder of mucociliary clearance that shares many features with PCD.

Ciliary activity is also responsible for organ placement in the developing embryo. When ciliary function is impaired, congenital defects of the structure and/or placement of internal organs may occur. Complete reversal of the organs, a condition called *situs inversus totalis*, occurs in approximately 50% of all PCD patients. Other commonly seen organ abnormalities include polysplenia (multiple spleens), asplenia (no spleen), congenital heart defects, vascular abnormalities and cystic lesions in the kidneys, spleen, liver or pancreas.

In PCD, impaired ciliary activity results in lifelong respiratory disease with chronic, debilitating infections of the lungs, sinuses and ears. Over time, chronic infection results in permanent damage to these areas and, in the case of the lungs, possible respiratory failure. Adults with PCD experience infertility (males) or subfertility (some females).

COMMON FEATURES OF PCD	**CONDITIONS ASSCOCIATED WITH PCD**
Neonatal respiratory problems, sometimes resulting in infant death	Congenital heart defects—200X increased risk for those with PCD
Organ placement/structural abnormalities	Hydrocephalus (enlarged ventricles of the brain)—rare
Chronic cough; usually present from birth or early infancy	Retinitis pigmentosa (a form of progressive blindness)—rare
Excessive production of mucus and recurrent, debilitating lung infections	
Bronchiectasis (scarring and permanent damage to the airways)	
Sinusitus, unusually severe and from an early age	
Frequent, severe ear congestion/infections and hearing loss	
Male infertility, female subfertility or ectopic pregnancy	

PCD DISEASE MANAGEMENT

There is no cure for PCD. Management is focused on symptom relief and slowing the progression of lung damage. Daily interventions include airway clearance and aggressive treatment of respiratory inflammation and infection in the upper and lower respiratory tract. More strenuous intervention is required for disease exacerbations. The use of IV or inhaled antibiotics and supplemental oxygen may be required for acute symptoms. Repeated hospitalizations are common in PCD.

LONG-TERM OUTLOOK

People with PCD are sick from birth and experience a greatly diminished quality of life. Disease expression is variable with some succumbing to lung damage early in life. Others may live into the fifth or sixth decade, but the disorder is progressive and quality of life deficits result in a lifespan that is far from 'normal.'

Recognizing the urgent need for better diagnostic capability and improved access to research and care for individuals with PCD, the PCD Foundation, in collaboration with our research partners, established a PCD Clinical Center Network. This North American initiative provides patients and healthcare providers with state-of-the-art diagnostic and treatment options, while creating a path to clinical trials that will ultimately help us find a cure for PCD.

FOR MORE INFORMATION:

PCD Foundation, www.pcdfoundation.org

University of North Carolina, Chapel Hill PCD site: http://www.med.unc.edu/pulmonary/areas-and-programs/pcd-research-treatment-at-unc

Genetic Disorders of Mucociliary Clearance Research Consortia:
http://rarediseasesnetwork.epi.usf.edu/gdmcc/about/index.htm