PCD FOUNDATION
PRIMARY CILIARY DYSKINESIA

PCD Fact Sheet

What is Primary Ciliary Dyskinesia (PCD)?

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), reproductive organs, and 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. This important biological system is called 'mucociliary clearance.' Defects of mucociliary clearance can lead to profound illness. Cystic fibrosis, another genetic disorder of mucociliary clearance, 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 occurs in approximately 50% of the population. Situs inversus totalis, with complete reversal of all visceral organs is the most common presentation. Heterotaxy and situs ambiguus occurs in 12-16% of the population and may manifest as polysplenia (multiplespleens), asplenia (no spleen), and congenital heart defects.

Impaired ciliary activity from PCD contributes to lifelong respiratory disease with chronic, debilitating infections of the lungs, sinuses and ears. Over time, chronic infection results in bronchiectasis-- permanent damage to the airways. Progressive bronchiectasis can lead to respiratory failure and the need for lung transplant in some patients.

Adults with PCD may experience infertility (most males) or subfertility (some females) due to impaired activity of the sperm tails and of cilia in the Fallopian tubes.

COMMON FEATURES OF PCD

-Neonatal respiratory problems, in rare cases leading to infant death

-Organ placement/structural abnormalities

-Chronic cough; usually present from birth or early infancy

-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 Diagnosis

Because there is no single method of diagnosis that reliably picks up all cases of PCD, a matrix approach is often used, incorporating multiple diagnostic modalities, including:

- Detailed history suggestive of PCD
- Analysis of ciliary ultrastructure via transmission electron microscope. Requires technical expertise. Analysis can be challenging. Will miss 30% of PCD mutations that have no ultrastructural correlate.
- Genetic testing. Widely available comprehensive panels will pick up approximately 65-70% of all cases.
- Measurement of nasal nitricoxide (nNO) via chemiluminescent analyzer. Note: Not FDA approved for diagnosis. Very useful for screening when done under a research protocol.
- High-speed video microscopy (HSVM). Must be done at expert center. Commonly used in Europe, less so in North America.

PCD 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.

Strategies to decrease frequency and severity of respiratory exacerbations include: limit exposure to individuals with acute respiratory illnesses, get immunizations against respiratory pathogens (influenza, pneumococcus, COVID-19), clean nebulizers and other respiratory equipment, and avoid of airway irritants such as tobacco smoke, vaping, or toxic vapors/fumes.

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, sixth decade or even seventh decade. However, PCD 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 (PCDF), in collaboration with our research partners, established the PCDF Clinical & Research Centers 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 find a cure for PCD.

For More Information

PCD Foundation:
www.pcdfoundation.org

Genetic Disorders of Mucociliary Clearance Research Consortia:
https://www.rarediseasesnetwork.org/node/131

University of North Carolina, Chapel Hill PCD Site:
https://www.med.unc.edu/medicine/pulmonary/patient-care/pulmonary-subspecialty-care/primary-ciliary-dyskinesia-pcd/