Primary Ciliary Dyskinesia (PCD)

Patient Handout

What is Primary Ciliary Dyskinesia?

Primary Ciliary Dyskinesia (PCD) is a rare lung disease. It affects the small hair-like structures that line your nose, ears, and lungs called cilia. Cilia move back and forth to move germs, mucus, and dust out of your airway. If they don't work well, germs can stay in your airway leading to frequent infections and trouble breathing.

How do you get it?

PCD is a genetic disorder, meaning that it is passed from parents to children through genes. Mutated (faulty) genes can cause the cilia to be the wrong size, shape, or move in the wrong way. If a child receives faulty genes from both parents, they can get PCD. PCD happens in around 1 in every 10,000 to 30,000 births.

Symptoms

Symptoms vary from person to person – they can include:

- Daily runny nose
- Daily wet cough, often present from early age
- Frequent episodes of pneumonia
- Recurrent sinus infections
- Hearing loss
- Internal organs in different places in the body
- Recurrent ear infections
- Respiratory distress at birth

Diagnosis

If your doctor thinks that your child has PCD, they may recommend certain tests to confirm the disease.

Genetic Testing – Genetic testing is performed to screen for known disease-causing mutations. This is done with a blood sample.

Ciliary biopsy – a sample of the cilia from your nose or airway is taken. The sample is then viewed under a microscope to assess the structure of the cilia.

Nasal nitric oxide testing – This test measures the level of a certain gas, nitric oxide, in your sinuses. In people with PCD, they have lower levels of nitric oxide compared to other people.

Treatment

There is no cure for PCD. The main goals of care include:

(1) preventing infection
(2) removing mucus from the lungs

There are several ways to reach these goals:

Chest Physical Therapy (CPT)

CPT involves clapping over your chest to loosen mucus. This is often done by another person. There are also several devices to help such as vests or a hand-held device you breathe out through that rattles your chest.

Medicines

- **Antibiotics** are the main treatment for infections. These can be taken by mouth or for more serious infections through your blood by an IV.
- **Inhaled therapies** such as albuterol and hypertonic saline (salt water) may be used to help open the airway and loosen mucus in the airways.

People with PCD need care from multiple specialists including pulmonologists, respiratory therapists, and ear, nose, and throat doctors. Vanderbilt Children's is an accredited PCD Center, meaning we have the diagnostic tools and specialists needed to diagnosis and care for children with this rare disease.

Contact Us:

The Rare Lung Disease Program

Monroe Carell Jr. Children's Hospital at Vanderbilt

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PCD Center Website

Additional Resources

For more information about PCD, please visit: www.pcdfoundation.org