

# WHAT TO EXPECT AT YOUR PRIMARY CILIARY DYSKINESIA EVALUATION

## WHAT IS PRIMARY CILIARY DYSKINESIA (PCD)?

PCD is a rare inherited disorder caused by defects in the structure and/or function of cilia. Cilia are tiny hair-like structures that move fluids and particles in various parts of the body, including airways such as the nose, sinuses, ears and lungs. If there are defects in the cilia lining the airways, the body cannot expel foreign material and mucus. This can lead to pulmonary complications, including frequent infections of the nose, sinuses, ears and lungs.

## WHAT CAN I EXPECT BEFORE MY FIRST VISIT TO CHOP'S PCD CENTER?

About 1-2 weeks before your child's scheduled visit with the Primary Ciliary Dyskinesia Center at Children's Hospital of Philadelphia (CHOP), you will receive a call from a PCD registered nurse to go over some pre-visit questions as well as answer any questions you may have about the visit.

## WHAT CAN I EXPECT DURING MY FIRST VISIT TO CHOP'S PCD CENTER?

### **Patients under age 5:**

Your child will meet with a PCD provider and, possibly, a pulmonary physical therapist and/or registered dietician.

### **Patients age 5 or older:**

Before meeting with our multidisciplinary team, your child will have to complete some breathing tests. First is nasal nitric oxide (NO) testing – performed under a research study – to look for low nasal NO levels, which PCD patients often have. Next is pulmonary function testing (PFT), which measures how well your child's lungs work. Once testing is complete, your child will meet with a PCD provider and might also meet with a pulmonary physical therapist and/or registered dietician.

## WHY IS A PHYSICAL THERAPY VISIT ADDED TO MY PCD APPOINTMENT?

Our physical therapists are experts in a variety of airway clearance techniques and are a vital part of the PCD team. They will assess your child's existing airway clearance plan and modify as needed, or they will provide your child with a personalized plan if they do not already have one.

## WHY IS A NUTRITION VISIT ADDED TO MY PCD APPOINTMENT?

The registered dietitians (RDs) at our PCD Center are dedicated to your child's nutritional needs. Research suggests that lower body mass index has been associated with worsening lung function. Depending on your child's needs, the RDs will work with you to design nutrition plans that will prioritize lung health.

## WHY IS SOCIAL WORK A PART OF MY NEW PCD APPOINTMENT?

Every patient who receives care from CHOP's Division of Pulmonary and Sleep Medicine is assigned a social worker for support. Social workers are a part of the interdisciplinary team and work with doctors, physical therapists, nurses, dietitians and other staff members to support your family's health and wellbeing. They can assist with school, transportation, insurance, mental health, housing, utilities, finances, food instability, emotional support and/or any safety issues.

## WILL MY CHILD RECEIVE GENETIC TESTING?

Since PCD is an inherited disorder, it can often be diagnosed with genetic testing. Over the last few years, genetic testing for PCD has expanded as more and more genes have been found to be responsible for causing PCD. Genetic testing can be useful in diagnosing PCD; however, a negative genetic test cannot rule it out entirely. Your provider will consider whether your child may benefit from genetic testing and may ask a genetic counselor to speak with you during your visit. Most commonly, genetic testing can be a blood test or a cheek swab.

### KEY TAKEAWAY

Diagnosing PCD can be challenging. Our team is here to support you throughout the process. While you may not necessarily leave your first visit with an official diagnosis, you will leave with a plan and next steps.

## RESEARCH OPPORTUNITIES

Clinical research is an important part of learning more about childhood lung diseases and helping to improve outcomes for children, adolescents and teens with PCD. A member of the clinical research team may reach out to discuss current research studies, either during or before your visit. If you would like to learn more about what research study opportunities are available through CHOP's robust research program, contact the PCD research team at [PulmonaryResearch@chop.edu](mailto:PulmonaryResearch@chop.edu).

## RESOURCES FOR PCD:

PCD Foundation

[pcdfoundation.org](http://pcdfoundation.org)

CHOP PCD Center

[chop.edu/PCD](http://chop.edu/PCD)

