# Primary Ciliary Dyskinesia Screening

Monroe Carell Jr. Children's Hospital at Vanderbilt

## Finding the right diagnosis for the right care.

### What is Primary ciliary dyskinesia?

Primary ciliary dyskinesia (PCD) is an autosomal recessive disorder resulting in impaired structure and function of cilia. Ciliary dysfunction prevents the clearance of mucus from the lungs, nasal sinuses, and ears leading to frequent infections. These recurring infections can lead to irreversible scarring and damage in the lungs (bronchiectasis), as well as other clinical problems such as sinus infections, hearing loss, and infertility. Movement of cilia is also important in organ placement in the developing embryo. Approximately 50% of individuals with PCD have an organ laterality defect in which the internal organs including the heart, liver, spleen, gallbladder, and stomach are in different positions in the body.

### Prevalence

PCD occurs in approximately 1 in every 10,000 – 30,000 births. The PCD Foundation estimates there are around 25,000 people in the United States with PCD; however, fewer than 2,000 people know that they have it. Some PCD symptoms like chronic cough, runny nose, and ear infections are common in children, making early diagnosis difficult. For that reason, patients often go a long time misdiagnosed or undiagnosed which can lead to poor outcomes.

### Specialty Care and Testing

That's why coming to a center with expertise in diagnosing this rare disease is so important! The Monroe Carell Jr. Children's Hospital at Vanderbilt is a newly accredited Primary Ciliary Dyskinesia Center with the tools and team needed to evaluate, diagnosis, and care for children with PCD. **But, we need your help as providers in the community in identifying these kids!**

### Who should be tested?

We suggest evaluating children with two or more of the following characteristics:

- Respiratory distress at birth for unknown reasons
- Daily runny nose
- Daily cough, usually wet in nature from birth or very young age
- Organ laterality defects

### How do we diagnosis PCD?

Primary ciliary dyskinesia is diagnosed through ciliary biopsy and genetic testing for known disease-causing mutations. Screening for levels of nasal nitric oxide can also be helpful in diagnosing patients, as people with PCD have lower levels of nitric oxide in their sinuses.

If you have patients that fit the clinical picture above, please refer them to our center for further evaluation.

Please contact our patient coordinator Kathy Russell at 615-875-6901 – or fax the attached form.

# Thank you for your referral.

Primary Ciliary Dyskinesia Clinic

Monroe Carell Jr. Children's Hospital at Vanderbilt

Date of Referral: _____

## Patient Information

| | |
|---|---|
| Patient Name: | Date of Birth: |
| Parent/Guardian Name: | Phone Numbers<br>Home:<br>Cell: |
| Home Address, City, State: | |
| Primary Language: | Are Interpretation Services Required? Y N |
| Primary Care Provider (MD/NP/PA): | |
| PCP Phone Number: | PCP Fax: |

## Referring Medical Provider (MD/NP/PA) (*if different from above*)

| | |
|---|---|
| Referring medical professional: | Last appointment: |
| Referring professional phone: | Referring professional fax: |
| Referring Provider's Signature: | |

**Please select which symptoms led to this patient's referral:**

- ☐ Daily runny nose
- ☐ Daily wet cough
- ☐ Organ laterality defects
- ☐ Recurrent ear infections
- ☐ Recurrent episodes of pneumonia
- ☐ Recurrent sinusitis
- ☐ Respiratory Distress at Birth

Has this patient had imaging (chest CTs, chest x-rays, sinus imaging, etc.) performed? Y N

**If yes, please fax reports along with this referral form.**

Thank you again for your referral – if you have any questions, please contact us at 615-875-6901.

**Please fax this completed form to Kathy Russell at 615-322-7553.**