



# Gunky Lungs & Bungee Boogers:

## The Kid's Guide to PCD

Primary Ciliary Dyskinesia  
Kartagener Syndrome  
Immotile Cilia Syndrome

A Publication of the  
PCD Foundation

*This booklet is dedicated to children with PCD and their families and friends. It provides a brief overview of the causes and symptoms of PCD and some things that can be done to help cope with some of these symptoms. This book is not intended to replace medical advice. Please consult your physician before undertaking any treatment program. Children should get their parents permission before reading this booklet.*

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*Medical Review provided by*

Margaret Leigh, MD, University of North Carolina School of Medicine

Joseph Sisson, MD, University of Nebraska Medical Center



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## Part I

# What is PCD?

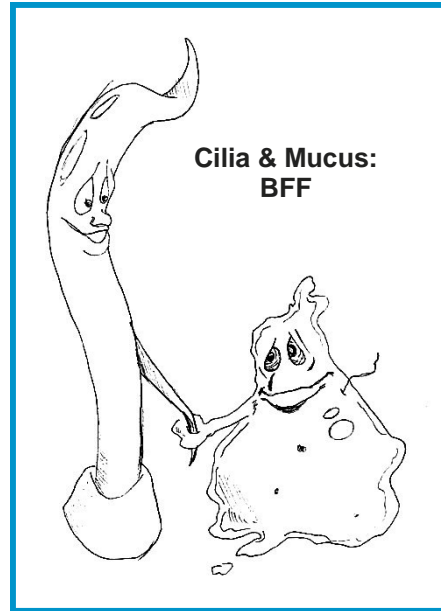
- Mucus
- Cilia
- Mucociliary Clearance

# WHAT IS PCD?

If you are reading this booklet, chances are you either have PCD (primary ciliary dyskinesia) or know someone who does. To understand what happens in PCD, it's important to understand how certain parts of the body work. Let's start with the thing that seems to cause the most trouble in PCD: Mucus!

## THE "M" WORD: MUCUS

Let's face it, not many people really want to talk about mucus.\* But if you are a kid with PCD mucus is a part of your life that just can't be ignored. There's lots of it in your lungs that can rattle and gurgle, there's plenty in your nose pretty much all the time. Sometimes you might feel like you are drowning in the stuff! So what is the deal with mucus, anyway? Why is there so much of it in people with PCD? Is there any way to control it?



## MUCUS IS YOUR FRIEND

First of all, it's important to know that—believe it or not—*mucus is a good thing*. In fact, when there is just the right amount of it and it is just the right “thickness,” it is a **VERY** good thing. This is because mucus, working closely with **cilia** (**SIL-ee-ah**), does an important job in the body. Many parts of your body, especially parts that have tubes or open spaces, need mucus and cilia to keep them clean and healthy. We will see how cilia and mucus working together keep these areas clean 24 hours every day.

## WHAT IS MUCUS?

Everyone knows what mucus is, right? The slimy, wet stuff found in certain parts of your body. But did you know that mucus is actually made up of both liquids *and* solids and that it forms layers? Each layer has to be perfectly formed if the mucus is to work effectively with cilia.

\*Mucus is spelled *mucous* in England, Canada and some European countries, but is pronounced the same. You may have your own name for mucus: gunk, goo, snot, loogies, phlegm, sputum, etc.

## WHAT ARE CILIA ?

Cilia are tiny (you need a microscope to see them), long, thin “organelles” (little organs) that come up from the surface of tissues that line certain parts of the body. Some cilia, called motile cilia, beat or wave in a consistent pattern. This motion is very important, as we will see later. Cilia may look a little like hair (in fact the word “cilia” means eyelashes), but they have nothing in common with actual hair. Their structure and function in the body is completely different.

There are billions of moving (motile) cilia in your airways, sinuses and ears, as well as some other parts of the body. For kids with PCD, problems with motile cilia in the lungs, sinuses and ears cause the most trouble.

Cilia and mucus must work together so they can perform their important job called **mucociliary** (myu-ko-SIL-ee-air-ee) clearance.

## MUCOCILIARY WHA...?

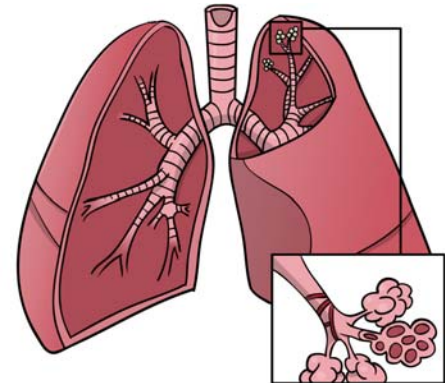
Our bodies need oxygen to survive. The best way to get oxygen from the outside atmosphere is to breathe in large amounts of air through the nose and mouth so it can get to the lungs and travel through the bronchial tubes to the alveoli. Unfortunately, outside air also contains things we don't want in our lungs, like pollutants, germs and dust. These unwanted particles are breathed in, as well. Sometimes these particles can be dangerous, so the body has developed a system for removing unwanted particles called **mucociliary clearance**. Here's how it works:

Mucus of just the right amount and the right “stickiness” lays over the surfaces of airways, ears and sinuses like a blanket. The mucus rests on a “carpet” of moving cilia. Particles that do not belong in these areas get trapped by the sticky mucus. Working together, the cilia lying under the mucus blanket beat very fast and in the same direction. The motion of the cilia moves the mucus toward the nose and throat where it can be coughed out or swallowed.

## WHAT PARTS OF MY BODY USE MUCOCILIARY CLEARANCE?

### LUNGS

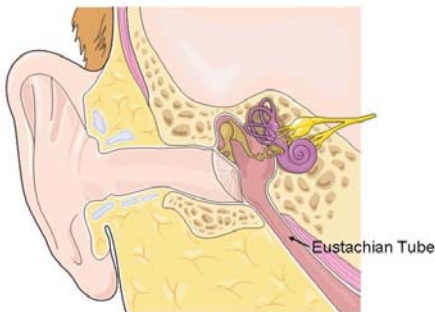
Every part of your body needs oxygen to live. Your lungs are responsible for getting oxygen from the outside air into your body. They do this with a system of branching tubes called **airways** or **bronchial (bron-KEY-ul) tubes**. They look a little like this picture. The air you breathe in passes through the airways to little sacs called **alveoli (al-VEE-oh-lie)** where oxygen is absorbed for the rest of your body to use.



Source: Rory Manion

It is important to keep the airways clear so oxygen can get to the alveoli. As we have seen, the airways are lined with cilia and mucus that work together to keep the airways clean.

### EARS



Source: Servier Medical Art

Your ears have tubes, too. One, called the **Eustachian (you-STAY-shun) tube**, is lined with cilia and fluid. The Eustachian tube helps control the pressure in your ears and keeps your middle ear healthy.

If your Eustachian tubes aren't doing their job, you may have the sense of plugged or full ears and you may not hear as well as you should. Here is a drawing of what the inside of your ear looks like. Can you find the Eustachian tube?

### SINUSES

You may not want to hear this, but guess what? **YOU HAVE HOLES IN YOUR HEAD!!** It's okay though, because so does everybody else. These holes are called **sinuses (SIGH-ness-ez)**. They may look like big empty craters, but they aren't. Sinuses are lined with living tissue. Cilia and mucus are important for keeping the sinuses clean, too. This drawing gives you an idea of where your sinuses are found.



Source: Servier Medical Art

# SO WHAT IS PCD (Primary Ciliary Dyskinesia)?

Now you know that mucociliary clearance is happening all day, every day in your body even though you are not aware of it, and that for it to work, both cilia and mucus need to do their part. If the mucus is too thick and sticky, the cilia may not be able to beat effectively and the mucus may get stuck in the airways. This happens in a disorder called **cystic fibrosis (CF)**. If the mucus is just right, but the cilia don't beat correctly mucociliary clearance won't work, either. Without the beating of the cilia, even normal mucus may get stuck in the airways. This is what happens in **primary ciliary dyskinesia (PCD)\***.

Both CF and PCD are inherited disorders, meaning passed down from parents to children, and both are disorders of mucociliary clearance. You cannot catch PCD or CF from another person and you cannot give PCD or CF to another person. Since this is a booklet about PCD, we will not talk about CF in detail. However, you may have heard PCD compared to CF and may see a doctor from a CF clinic. This is because they both are disorders of mucociliary clearance that require similar treatments. If you would like to know more about CF, please ask your parents or your doctor.

\*Primary here means "inherited." Ciliary means "related to cilia" and dyskinesia means "impaired movement." So PCD literally means inherited impaired movement of the cilia.



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## Part II

# So, My Cilia Don't Work... Now What?

- What Happens in PCD?
- How Is PCD Treated?

# WHAT HAPPENS IN PCD?

## Sometimes I Think PCD Should Stand for PRACTICALLY CONSTANT DRAINAGE!

When mucociliary clearance is impaired as it is in PCD, your body tries all sorts of tricks to get rid of unwanted particles. Producing more mucus is one of those tricks. You can think of it as your body trying to “flush” out the unwanted particles. Extra mucus or mucus that is trapped can lead to some of the most common problems experienced by people with PCD:

- Runny nose
- Thick, sticky nasal discharge (aka “bungee boogers”)
- Sinus infections and headaches
- Chronic cough
- Noisy breathing (aka “gunky lungs”)
- Lots of chest infections
- Plugged or painful ears
- Drainage from the ears
- Lots of ear infections

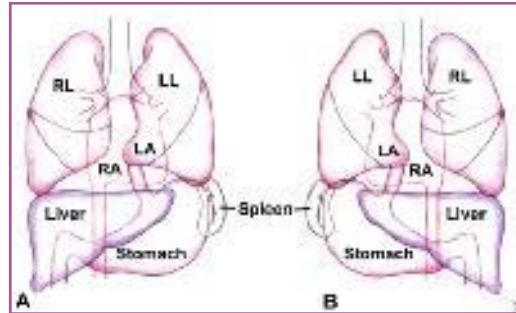
Not all people with PCD have all of these symptoms and some people with PCD may have more symptoms than others. If you have PCD, though, there is a good chance you have had many of them.



## IS MY HEART IN THE RIGHT PLACE?

Cilia are amazing little things. Believe or not, the way cilia move is also important in deciding where your organs will end up inside of you before you are born. Because of this, some people with PCD may have their insides arranged differently than other people. Organ placement inside your body is known as *situs* (**SITE-us**).

Most people have their heart a little to the left in their chest, their spleen on the left in their abdomen (tummy area) and liver on right. This is called *situs solitus* (**SITE-us SOL-it-us**). Because most people have *situs solitus*, this is the organ arrangement the doctor is expecting to find when he or she examines you, but there are other perfectly acceptable organ arrangements. If everything is completely reversed (heart and spleen on right, liver on the left), it is called *situs inversus totalis* or just *situs inversus* (**SITE-us in-VER-sus**). About half of all people with PCD have *situs inversus*.



*Situs solitus and situs inversus. Wilhelm, et al Emedicine. Used with permission*

*Situs inversus* alone is NOT a disorder. It is just a term to describe organ placement. When *situs inversus* is present in a person with PCD, it is called “Kartagener syndrome.” People with Kartagener syndrome and people with PCD have the same disorder—PCD. The term Kartagener syndrome just lets doctors know that *situs inversus* is present.

Sometimes (but not very often) people with PCD do not have complete *situs inversus*, but do not have *situs solitus*, either. They may have only one or two organs that have developed in an interesting way or are in an unusual place. For example, just the heart may be reversed (dextrocardia) or they may have multiple spleens (polysplenia) or no spleen at all (asplenia). Any unusual organ placement that is not *situs inversus totalis* or *situs solitus* is known as *situs ambiguus* (**SITE-us am-BIG-you-us**).

## Will My Organs Still Work if They are in the Wrong Place?

Most people with PCD have internal organs that work just like everyone else's, no matter where they are. For some people with PCD, though, unusual organ placement or development can cause problems. The most serious problem seen in PCD is known as **congenital (kon-JEN-it-al) heart disease (CHD)**. Congenital means "present at birth," although sometimes problems that are actually present at birth may not be noticed until much later. People with PCD and CHD need specialized care from a doctor familiar with heart problems, in addition to their regular doctor and their lung and ear/sinus doctors. Some people with CHD will need to have surgery to fix their hearts, others may just need medicine. Some may need no treatment at all.

No matter where your heart (and everything else) is, it is important to remember that it is in exactly the right spot for you! Usually, there are no extra problems for PCD kids with different organ arrangements, but talk to your parents and your doctor if you are concerned about this.



# HOW IS PCD TREATED?

As we learned earlier, people with PCD have problems with mucociliary clearance. When mucociliary clearance does not work effectively, too much mucus or fluid may collect in the airways, sinuses and ears. Most of the treatments used in PCD are aimed at reducing the problems caused by ineffective mucociliary clearance, like infections.

## ***Ways to Help Clear the Airways (Lungs)***

There are several types of treatment to help clear mucus from the airways and keep them healthy and free from infection:

- Airway clearance therapy (ACT)
- Medication to open the airways
- Medication to thin the mucus
- Medication to treat lung infections

### ***Airway Clearance Therapy (ACT)***

ACT is a very important part of daily care for kids with PCD. There are many kinds of ACT. Most involve physical action that is done to help mucociliary clearance. Sometimes the physical action includes shaking or pounding the chest to remove excess mucus. Some examples of this type of ACT are:

- \*Vest devices
- \*Chest physiotherapy (CPT)



*Example of a Vest Device: Smart-Vest® by Electromed*

Sometimes the physical action might involve positioning the body to help the lungs drain. This is called **postural (pos-CHUR-al) drainage**. Specialized breathing techniques may also be used.

Other ACT activities include blowing against a resistor. This is called positive expiratory pressure or PEP. Here are some examples of PEP devices:

- \*Flutter®
- \*Quake®
- \*Acapella®

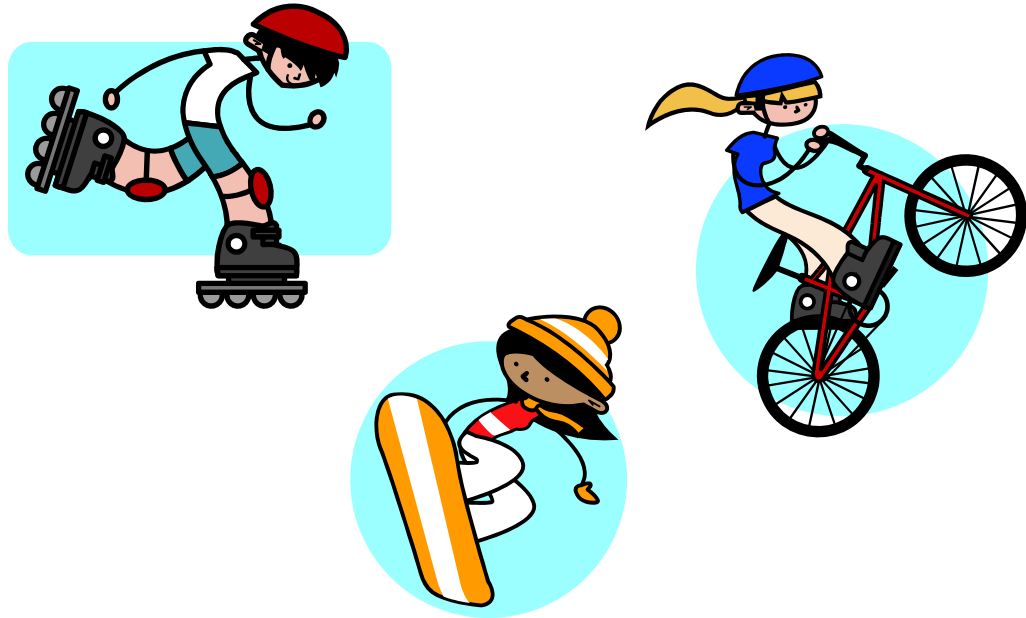


*Acapella® Airway Clearance Devices*

Remember, PEP devices (and any other device that comes into contact with mucus) must be thoroughly cleaned using the method your doctor recommends.

### ***Airway Clearance and Exercise***

Vigorous exercise is a very important part of airway clearance. Exercise is a “two-fer” because it helps to open up your airways *and* it helps to move the mucus out, so be as physically active as you can be!



### ***But Airway Clearance Makes Me Cough!***

Excellent! That’s exactly what it’s supposed to do. What all good airway clearance therapies have in common is that they make you cough. Coughing is the single most important thing kids with PCD can do to help clear their airways, so let it out! Of course, it is always a good idea to cover your mouth when you cough.

### **What You Can Do To Help With Airway Clearance**

For ACT therapy to be the most effective, you have to commit to doing it every day. Work with your doctor and your parents to come up with a personal ACT plan that will work for you and then stick with it!

### ***Medications to Help Open the Airways***

When mucus collects in the airways, it can make the airways irritated, causing them to tighten up and swell. There are medications that can help to relax tightened airways. These drugs are called **bronchodilators (bron-ko-DIE-laters)**. Other medications can help to reduce irritation and swelling. These drugs are called **steroids (STARE-oyds)**. Bronchodilators and steroids may be used separately or together and very often are given by puffer” (also called metered-dose inhaler or MDI), nebulizer or other inhaled method.

### ***Medications to Help Thin Mucus***

The mucus in most people with PCD is not overly sticky—at least it doesn’t start out that way. But mucus that sits in the airways can begin to thicken and plug the airway, causing more mucus to get stuck. Some people with PCD find that medicines that help to thin their mucus can help them with airway clearance. These medicines are called **mucolytics (myu-co-LIT-icks)**. Hypertonic saline and Pulmozyme® (DNase) are examples of mucolytic drugs. Most mucolytics are inhaled using a nebulizer.

### ***Medications to Help Fight Infection***

Sometimes the particles that get stuck in the airways are germs, like viruses and bacteria. When germs try to set up housekeeping in your airways, the result is called an **infection (in-FEK-shun)**. Infections that are caused by germs called bacteria can be treated using medicines called **antibiotics (AN-ti-by-OT-icks)**. Antibiotics do not work for all types of infection and only certain antibiotics work against certain bacteria, so it is important to get a sample of your mucus, called a **culture (KUL-chur)**, so your doctor can see exactly what bacteria are growing in your lungs and use the right antibiotic to treat your infection.

There are many different types of antibiotics and they may be given in different ways. The most common way is by swallowing them either as a liquid or in pill form. Some antibiotics can now be inhaled directly into your lungs. Some antibiotics need to be given directly into your vein. These IV (**intravenous**) antibiotics require that a tube be inserted into your arm or other area. Some IV tubes are in only for a day or two and some stay in longer (**PICC lines and port-a-caths**). It is never fun to need an IV, but the worst part is the needle stick to get the tube in place. Once in your vein, most IVs do not hurt.

## ***Ways to Help Clear the Sinuses***

Pain and stuffiness in the sinuses can be very uncomfortable for people with PCD. There are several things you can do to help with these problems:

- Sinus rinsing/irrigation
- Medication
- Surgery
- Sinus suctioning

### ***Nasal and Sinus Rinsing/Irrigation***

If your mucociliary clearance system is not working, mucus can get stuck in the sinuses. One way to help move this mucus out is to “wash” the nose and sinus areas. Washing (also called rinsing or ‘irrigation’) helps in several ways. First, it thins the mucus making it easier to remove, and second, it flushes away particles like germs and dust from the nasal passages before they can get stuck in the sinuses.

### ***Medications to Help the Sinuses***

Sometimes stuffiness in the sinuses is an indication of infection. Sinus infections are treated the same way that infections in the lungs are—with antibiotics. Medications that treat the symptoms of allergies can help to dry up nasal and sinus mucus and help you feel better for a little while. Dry mucus is even harder to remove than lots of sticky mucus, so some of these medicines can actually make you feel worse over time. Be sure to check with your parents and your doctor before using medicine that dries your sinuses.

### ***Surgery for Sinus Problems***

When nasal rinsing and antibiotics aren’t enough, sometimes people with PCD may have surgery to treat their sinuses. Some surgeries are done to help drain and rinse the sinuses one time. Some surgeries are done to create more permanent openings for drainage.

### ***Sinus Suctioning***

Extra mucus can be vacuumed or ‘suctioned’ from the nose and sinuses either at home using a device called the Wiva-Vac™ or similar device attached to your vacuum cleaner (yes, really!) or at the doctor’s office.

## ***Ways to Help Clear the Ears***

People with PCD may have pain, drainage or infection in their ears from time to time. Here are some ideas for things you can do to help with these problems:

- Medications
- Suctioning
- Hearing aids
- Surgery

### ***Medications to Help Your Ears***

Infections in the ears and sinuses are treated the same way that infections in the lungs and sinuses are—with antibiotics. Antibiotics and other medicines for ear problems can be given by mouth (liquid or pills) or can be in the form of drops that go directly in your ears.

### ***Suctioning***

Sometimes kids with PCD have more fluid in their ears than the Eustachian tube can manage. Extra fluid can lead to discomfort and hearing loss. Sometimes, the extra fluid may drain outside of the ear instead of down the Eustachian tube. If there is a lot of fluid, it can be suctioned—usually in the doctor’s office, but sometimes at home if you/your parents are trained to do it and have the right equipment.

### ***Hearing Aids***

For kids with PCD, chronic problems with their ears can lead to problems with hearing. Sometimes these problems may fix themselves in time or may be easy to fix with medications or surgery (see below). But sometimes, if the problems are longer lasting or if medication or surgery do not fix the problem, hearing aids can be very helpful to improve hearing. There are many types of hearing aids. Talk with your parents and your doctors to decide if hearing aids are the best choice for you.

### ***Surgery for Ear Problems***

Some of the ear problems in PCD can be fixed with simple surgeries. The most common of these surgeries is called placement of myringotomy tubes or just “tubes.” In tube surgery, a tiny opening is made in the eardrum and an actual little tube is inserted in the opening. This tube allows for the fluid behind the eardrum to drain out to relieve the pressure of extra fluid on the eardrum and to improve hearing.

## ANY OTHER PROBLEMS IN PCD?

Problems with lungs, ears and sinuses usually cause the most trouble for people with PCD, but there are some other things you should know about, too.

### ***MY TUMMY HURTS***

Some people with PCD have trouble with their stomachs. This may be due to medications taken for PCD or a condition called **reflux (RE-flucks)** that can be made worse when people cough a lot, or for other reasons we just don't understand yet. If your stomach is upset a lot, be sure to talk to your parents and your doctor about it.

### ***I HAVE A HEADACHE***

Kids with PCD may have more problems with headaches than other kids. This may be due to sinus infection or stuffiness, or it may be due to other causes. If you are having trouble with headaches, be sure you let your parents and your doctor know.

## WILL I HAVE PCD MY WHOLE LIFE?

PCD is a lifelong disease. However, most people with PCD live long and happy lives, even though they may have to treat infections from time to time. So what can people with PCD do? They can go to college, get married and have families just like everyone else. People with PCD can pursue hobbies and jobs that interest them. There are people with PCD who are horse trainers, nurses, lawyers, doctors, computer programmers, you name it! Everybody's health is different, so discuss your goals and plans with your parents. By taking care of your health and living an active life now, you can increase the chance that you will have a healthy and happy adulthood.



## **For More Information About PCD:**

It can be hard to find good, accurate information about rare disorders like PCD. Sometimes what is published on the Internet is either incomplete or it is just plain wrong! Here are some good websites you can visit to learn more about PCD.\* Please check with your parents before you visit these sites.

### **The PCD Foundation: [www.pcdfoundation.org](http://www.pcdfoundation.org)**

The PCD Foundation (PCDF) is the only North American patient education, research and advocacy organization dedicated to PCD. The PCDF website has information about diagnosis and treatment, upcoming events and a patient forum where affected individuals and their families can communicate with each other. There is a special forum site just for children, teens and young adults.

### **The University of North Carolina, Chapel Hill, PCD Research and Treatment Center: [www.med.unc.edu/cystfib/PCD.htm](http://www.med.unc.edu/cystfib/PCD.htm)**

The University of North Carolina (UNC), Chapel Hill, is the primary site for PCD research in the United States. This site provides information about PCD and links to individual researcher web pages and other resources.

### **UNC Pediatric PCD Site: <http://pediatrics.med.unc.edu/div/infectdi/pcd/>**

An additional site provided by a PCD researcher at UNC, Chapel Hill. This site has really cool videos of ciliary activity in people with normal ciliary activity and people with PCD

### **Children's Hospital of Eastern Ontario (CHEO) Immotile Cilia Syndrome Page:**

This site provides an excellent overview of PCD (remember 'immotile cilia syndrome' is a term sometimes used in place of PCD).

### **GeneReviews PCD: <http://www.ncbi.nlm.nih.gov/bookshelf/br.fcgi?book=gene&partid=1122>**

This site, provides in-depth, up-to-date and accurate information about genetic disorders. The information is a little technical, but if you are interested in learning all you can about PCD and feel up to the challenge, it is definitely worth checking out!

\*If you don't have Internet access, please call or send a letter requesting more information to:

**PCDF Info Request  
C/O Lynn Erhne  
61 Lake Meadow Drive  
Rochester, NY 14612  
585-621-8915**

