CHEO

PRIMARY CILIARY DYSKINESIA

Our airways, nose and sinuses are lined with microscopic, moving hair-like structures called cilia. These hairs beat in rhythm like "the wave" at a sports stadium. Cilia help move mucus (along with the bacteria, dust and pollutants trapped in it) to the mouth and nose where it can be swallowed or coughed out.

In children with primary ciliary dyskinesia (PCD), the cilia either don't move at all or move in an uncoordinated way. This leads to bacteria to getting stuck in the airways, leading to chronic bacterial infection and long-term airway damage.

Originally, it was thought that the cilia don't beat at all in all in those with PCD, so the condition used to be called immotile cilia syndrome.

What causes PCD?

PCD is a genetic disease so in order for it to happen, a child must get two PCD-causing genes—one from each parent. Cilia are complex structures and are made up of many different genes. As a result, there are many different genes associated with PCD, and more are being discovered every year.

PCD and the organs

When babies are developing, cilia inside the body make the organs go where they're supposed to. If the cilia don't beat, there's a 50% chance the organs will be on the other side of their body from where they usually are. Half of babies born with PCD have their heart on the opposite side (dextrocardia) along with the other reversed organs (situs inversus). When children have PCD and organs on the opposite side, it is called Kartagener's Disease. Sometimes, the organs end up in the middle of the body. This is often associated with congenital heart problems.

In men with PCD, the tails of sperm are closely related to cilia, and often also don't move. This often leads to fertility problems in men.

Diagnosing PCD

If someone in your family has PCD, or if you have a child with PCD, you can work with a genetic counselor to determine the odds your future children will have PCD.

Once a child is born, the process of diagnosing PCD isn't easy. It needs specialized techniques. The easiest method is collecting blood to look for PCD genes. Currently, we know the genes that cause PCD in about 70% of people, so genetic testing can be used to diagnose about 70% of people with PCD.

Another method involves scraping the inner lining of the nose (or inner lining of the airways, if an operation called a bronchoscopy is done) and looking at the cilia with an extremely powerful microscope. A few very specialized centers can look at live cilia with a video-microscope and see whether they're beating normally. Another technique involves collecting gas from the nose while the child breathes. A particular gas, called exhaled nasal nitric oxide, is noticeably decreased in PCD patients.

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Symptoms

Newborns with PCD often have:

- difficulty clearing mucus from the lungs after birth
- collapsed lungs (atelectasis)
- temporary difficulty breathing (known as respiratory distress) after birth

Children with PCD have bacteria in their airways all the time. Symptoms include:

- constant runny, congested or blocked nose
- severe recurrent ear infections
- draining ears
- reduced hearing
- chronic cough that sounds "wet"
- coughing up mucus (or sputum)

In older youth and during adulthood, the airways can become damaged from long-term bacterial infection. This leads them to become floppy and collapse when they cough, trapping mucus inside instead of coughing it out, perpetuating this process. This is called bronchiectasis.

What to watch for

If your child or youth catches a cold, their airway will have more mucus—a breeding ground for bacteria. This can lead to difficulty breathing, increased cough, fever, and/or wheezing. This is called a pulmonary exacerbation. Pulmonary exacerbations vary from mild to very serious and require immediate medical attention.

Signs of a severe pulmonary exacerbation include:

- severe shortness of breath, rapid or shallow breathing, and/or sucking in of the skin at the base of the neck, between the ribs, or just below the rib cage
- blue skin
- severe cough or wheezing
- inability to speak in full sentences because of difficulty breathing
- becoming tired or sleepy because of difficulty breathing

If your child or youth has these symptoms, seek medical attention right away.

How is PCD treated?

Because PCD is so rare, most treatments are based on therapy for cystic fibrosis, a similar condition.

Immunizations

Children with PCD should have all their immunizations to keep their lungs as healthy as possible. They should also get their flu shot every year.

Diet and exercise

A healthy diet is important, so the immune system has the energy it needs to fight chronic bacterial

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infection in the lungs and sinuses. Exercise is important to maintain fitness, and to stimulate cough, which helps clear mucus from the lungs.

Assisted airway clearance (chest physiotherapy)

Chest physiotherapy can help your child or youth to clear infected mucus. It's very important to start physiotherapy when your child is a baby so they get used to it as part of their daily lives (like brushing teeth). Some common methods are:

Percussion and postural drainage

This technique basically uses the "glass ketchup bottle principle." To get ketchup out of a glass bottle, you turn the bottle upside down and tap to get the ketchup out. During this technique your child is positioned so their airways are pointed downwards, and clapping on the chest helps loosen mucus so it can be coughed out.

Positive expiratory pressure (PEP)

This technique is for older children and youth who can use a mask or a handheld mouthpiece. It builds up pressure to keep the airways open and helps push mucus out of the mouth.

Physiotherapy vest

This technique uses an inflatable vest that compresses the chest a little and vibrates very fast to help loosen and liquefy mucus. Older children and youth can do this on their own, but the machines are expensive, and studies in cystic fibrosis suggest that PEP is more effective.

Medications

Antibiotics are used to treat worsening symptoms due to pulmonary exacerbation. They are chosen based on what bacteria was found on previous sputum cultures. Mild symptoms are treated with antibiotics given by mouth. Severe symptoms may require hospitalization for intravenous (IV) antibiotics.

Your child may be given an asthma inhaler if asthma is part of their lung disease. Salt water (saline) rinses or nasal corticosteroid sprays can be tried to clear nasal congestion.

Ear tubes

Severe, recurrent ear infections are sometimes treated by placing tubes in the eardrum. Some specialists feel that this leads to more draining ears, and others feel that it helps children.

Surgery

Children with extremely severe lung disease and very damaged lungs (bronchiectasis) may need a lung transplant.

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