

What is Primary Ciliary Dyskinesia?

- Primary ciliary dyskinesia, or PCD, is a rare genetic condition that affects tiny, hair-like structures that line the airways. These structures are called “cilia”.
- Cilia move together in wave-like motions. Cilia move mucus toward the mouth and nose to be coughed or sneezed out of the body. The mucus contains inhaled dust, bacteria, and other small particles.
- If the cilia don't work well, bacteria stay in the airways. This can cause breathing problems, infections, and other disorders.
- PCD mainly affects the sinuses, ears, and lungs. Some people who have PCD have breathing problems at birth, and others develop symptoms over time. People may have serious sinus, ear and lung infections. They often have year-round nasal congestion and cough. The symptoms and severity of PCD vary, and not all people have all of the symptoms.
- Some people with PCD have a mirror-image reversal of their heart and organs in their abdomen, called situs inversus totalis. Some men with PCD may have infertility.
- PCD is an inherited disease. "Inherited" means the condition is passed from parents to children through genes. Even though PCD is a genetic condition, individuals with PCD may have no other family members with PCD.
- We have two copies of most genes. In most cases, PCD is inherited from both parents who each “carry” one gene change in a gene that causes PCD. Carriers for PCD do not have PCD. When a child inherits two gene changes in the same gene for PCD—one from each parent—they are born with PCD.
- There are many different genes associated with PCD, with some genes still not yet known. These genes affect how cilia grow and work. Harmful genetic changes to the gene may cause the cilia to be different in size or shape or move differently. Sometimes the cilia are missing.
- Research studies help identify new genes that cause PCD. Knowing the genes that cause PCD can lead to better treatments. Participation in the PCD patient registry helps advance PCD research and the understanding of PCD.

