Mary Kitlowski & Rebekah Giannakos

PRIMARY CILIARY DYSKINESIA

"My sister, Rebekah, and I both had constant runny noses and ear infections, and I had a persistent cough."

My goal is to participate in a race in every state. In October 2014 I founded Running On Air to raise awareness of lung diseases, including the rare genetic disorder primary ciliary dyskinesia, which my sister and I have been diagnosed with since we were teens. With an FEV1 of 40 percent, I get through each race wearing a portable oxygen concentrator on my back.

The road to advocacy for PCD started at a young age. My sister, Rebekah, and I both had constant runny noses and ear infections, and I had a persistent cough. Our parents took us to see an otolaryngologist. We both had a series of surgeries to remove our tonsils and adenoids, and then had tubes inserted in our ears from the ages of 4-12. Rebekah developed pneumonia at the ages of three and nine. At the age of five I started seeing an allergist and receiving regular allergy shots. Rebekah soon followed with allergy shots at age 11. After rounds of tests, we were given the diagnosis of severe allergies and allergic rhinitis. But despite treatment, the coughing and congestion only seemed to get worse.

As a teenager I started having coughing fits at night. My mom would eventually wake me and make hot tea. At first these episodes were sporadic, every couple of months or so.

When I was 17 I started a part-time job working for my allergist. I was coughing so much that patients made jokes like, "You should find a good allergist to help you with that cough." My allergist tried different medications, but nothing helped.

At the same time, my dad insisted that I get a chest X-ray. It showed that I had bronchiectasis. All of my doctors were surprised. My allergist said to me "Every time you come in to see me I ask you if you're coughing more than normal." I replied, "I'm not coughing more than normal. This is how much I always cough." I was referred to a pulmonologist and began testing for the cause. Initial tests were inconclusive.

PRIMARY CILIARY DYSKINESIA

Primary ciliary dyskinesia (PCD) is an inherited disorder of motile (moving) cilia. PCD is also sometimes referred to as Kartagener syndrome (PCD with situs inversus) or immotile cilia syndrome.

A person with PCD experiences chronic, recurrent infections in the lungs, ears and sinuses due to the loss of ciliary activity in those areas.

PCD is a genetic disorder, meaning it is inherited from one's parents and cannot be acquired from the environment. PCD is most often passed in what is called an 'autosomal recessive' pattern of inheritance, in which the disease is only expressed when a child inherits two copies of a mutated gene—one from each parent.

Other conditions that may be associated with PCD are:

- Situs Inversus and Organ Orientation
- Polysplenia
- Congenital heart defects
- Hydrocephalus
- Fertility Issues

Learn more from ATS Public Advisory Roundtable member the Primary Ciliary Dyskinesia Foundation. pcdfoundation.org