Conor & Carey Kauffman

Primary Ciliary Dyskinesia

Conor was born a full-term, 9-pound 4 ounce “healthy” baby boy. His birth was very quick, so his inability to get rid of a tiny bit of congestion seemed, according to the doctor, to be related to some residual amniotic fluid in his system that may not have been ‘squeezed out,’ but nothing to be alarmed about. After staying with us the first night, we were concerned about a grunting noise he was making when trying to breathe. Just as a precaution, he was taken to the nursery to check his oxygen levels. We were told not to worry, and that lots of babies start life with some breathing and oxygen problems. They assured us that it almost always works itself out with no trouble.

Upon discovering his low oxygen level, they took him to the neonatal intensive care unit (NICU) and ordered the chest x-ray that revealed that all of Conor’s organs were completely reversed – a condition called situs inversus. After the

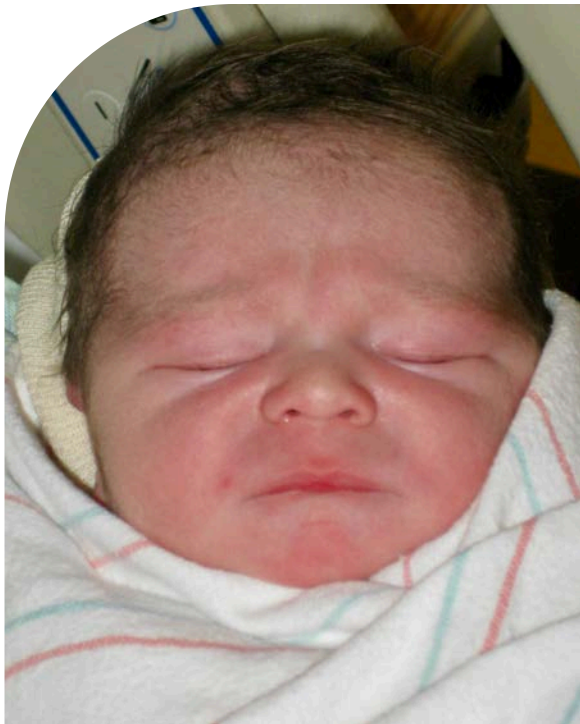
doctors told us that they had never seen a case before, they informed us that there was a 25 percent chance that Conor had an underlying condition called Kartagener Syndrome, which we now know is a subtype of primary ciliary dyskinesia (PCD). From that point, we heard a number of comments from the doctors that were concerning, if not alarming, in light of what would happen a few hours later:

- ***"Even if he has it, you won't need to worry about it until later in his life. It doesn't manifest itself until later."***

- ***"Donny Osmond has situs inversus totalis – and he's just fine!"***

- ***"Don't worry – he'll just a have a snottier nose than the other kids. All kids have runny noses."***

Once we had a name for what he might have, we started frantically researching anything we could find on our phones. From even the little information we could find, we realized that what we were hearing about PCD was not the reality of the disease. It wasn't even close. It was clear to us that Conor could be on a lifelong mission to prevent lung destruction aside from other PCD-related complications. He clearly wouldn't just be another "snotty nose kid."



In fact, I remember my husband saying, “Oh my God – this could be more like cystic fibrosis.” It was a very cold, isolating yet very confusing feeling to realize that these very experienced doctors in a state-of-the-art hospital might not know what we are dealing with. Unfortunately, we were right. Conor passed away the next day, shocking every doctor and nurse involved in his care. His lungs collapsed, causing his heart to shut down, and they couldn’t save him.

The doctors said that they had never seen or heard of anything like it – a perfectly healthy, robust baby boy rapidly deteriorating to the point where he couldn’t be saved - and couldn’t tell us what happened.

“Do you want an autopsy?” they asked. “Yes. And I want him tested for PCD! That’s what he had, I know it,” I replied. “PCD wouldn’t be seen as a ‘cause of death,’” the neonatologist replied, but agreed to do it because I was so serious about it. The autopsy came back citing acute pneumonia which again shocked the doctors who had taken multiple clear x-rays of his chest and had been unable to grow bacteria on tracheal samples when he was alive. They had also given him two antibiotics that would have addressed 90 percent of any infections out there. He tested positive for PCD.

Ultimately, we can get our minds around the cause of death. However, if your cilia don't work, they don't work from day one. ***Conor went into a fight with one arm swinging and he lost.*** He couldn't fight the infection like a 'normal,' healthy baby. We aren't doctors and have no medical training, but that made sense to us.

He's now the first baby on-record having died this way in the world, but we know he's not the first, and not the last. We cannot track what we do not measure.

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I do know that Conor's situation is uncommon, and I'm not sure he could have been saved – maybe he had a more severe form of PCD? Or maybe the pneumonia he got would have been too much for any child? Or maybe if he had gotten through the first couple of days, he'd be here right now slugging away with his daily treatments? But we don't really know, and never will.

Through the PCDF and the voices and talents of everyone touched by this disorder, doctors can be more informed, patients can live healthier lives and perhaps one day PCD will no longer need to be on any radar anywhere in the world.

Primary Ciliary Dyskinesia

Primary ciliary dyskinesia (PCD) is a rare, inherited, genetic disorder of motile (moving) cilia. Cilia are tiny hairlike structures on the cells in the body. Motile cilia perform an important role in the nose, ears, and airways within the lungs, working to remove unwanted inhaled particles and germs. PCD causes frequent respiratory infections starting at a very early age that result in lifelong, progressive lung, sinus and ear disease. People with PCD benefit from early diagnosis and treatment to hopefully limit permanent lung damage.

- PCD is an inherited disorder, meaning that people born with the disease receive a mutated (abnormal) gene from both parents. In PCD, mutations in the genes responsible for building cilia and controlling their function result in cilia that do not work effectively.
- Dyskinesia, or impaired movement, is the most common ciliary defect seen in PCD. Other defects may lead to not having enough cilia on each airway cell, which can also cause the clinical symptoms seen in PCD.

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ATS Patient Education Series