Karen Erickson

ALPHA-1 ANTITRYSPIN DEFICIENCY

"My doctor asked 'What matters most to you?,' making it clear that I had to take a lead role in defining and reaching my success."

Not a day goes by when I don't think back to two things: my donor and the gift he was willing to give, and the two-year prognosis that I was given 16 years ago. Both have stopped me in my tracks and both have also motivated me to continue this battle full force.

I've been on a long journey with Alpha-1 antitrypsin deficiency, and it's taken me from searching for a diagnosis to forging forward after receiving a bilateral lung transplant. At many points along the way, my partnership with providers (or lack thereof) has played a huge role in my success.

In the late '90s I was running a business, completing a degree, and training like crazy for triathlons. I was in a great place, but I started to notice that I was becoming more and more breathless. I had to rest during my busy days, the gains at the gym were becoming smaller and smaller until they were non-existent and I was always exhausted. I chalked it up to getting older and just a part of my busy lifestyle. But my symptoms worsened.

My doctors kept telling me the same thing. "Look at you sweetheart, you're fine. It's just exercise-induced asthma." Or, "You are just anxious about all the stuff you have going on. Here are some inhalers…take them in an emergency." Over the next year, I heard the same response over and over again. I was never given any other advice, testing or treatment plan, nor did I push the issue further. I came to expect it as my new normal.

Everything changed the day I was given my first pulmonary function test. The technician left the room, and the doctor came in. She told me I had the lungs of an 83 year-old and gave me a diagnosis of COPD. I was 33. Rather than digging in a bit more, she recommended that I stop smoking. I let her know that I wasn't a smoker. That statement was dismissed, and I left the office with a smoking cessation prescription.

I used and abused my inhalers to get me through my days and made frequent trips to urgent care, along my routes to and from work. On one of my trips in due

ALPHA-1 ANTITRYPSIN DEFICIENCY

Alpha-1 antitrypsin deficiency is an inherited form of emphysema. Patients with the disease do not have enough of a protein called Alpha-1 antitrypsin (AAT) in their blood. This protein is made in the liver, and it protects the lungs so they can work normally. Without enough AAT, the lungs can become damaged by emphysema. It also can cause liver damage. There is no cure for Alpha-1 antitrypsin deficiency.

Testing is recommended for those who have:

- Obstructive lung disease.
- Family history of Alpha-1.
- Early-onset emphysema (younger than 45 years old).
- Emphysema without an obvious risk factor.
- Emphysema that is worse at the bottom of the lungs.
- Chronic asthma (in adolescents and adults).
- Recurrent pneumonia or bronchitis.
- Unexplained liver disease.

Learn more: ATS Patient Education Series. "What is Alpha-1Antitrypsin Deficiency?" NewYork, NY. 2014. thoracic.org/patients/patient-resources/resources/alpha-1-antitrypsin.pdf

to breathlessness, a physician assistant asked if she could test me for a rare genetic disorder—although she was confident that I didn't have it. Blood was drawn, and I went about my business. Next thing I knew she called me into the office to discuss the test results.

The next morning the conversation went like this, "You have a genetic disease called Alpha-1 antitrypsin deficiency. Your body is basically eating your own lungs. There is no cure for it, but with treatment we think we can get you two years." The doctor went on to talk about referrals to a pulmonologist and other necessary steps, but I didn't hear a thing. My head was stuck on the 730 days I had left—my friends and family, my dogs, my job, everything…

I did see a pulmonologist, and he was excited to see me. He had only heard of Alpha-1 and was told never to expect to come across a patient. He admittedly knew very little about the disorder but managed to get me on a monthly augmentation therapy.

Uncharacteristically for me, I did not follow up with any research of my own. The news had rattled me enough, and I figured I had 730 days to live…. I distanced myself so much that I left for Ireland shortly after starting therapy.

It was there that everything changed again—but this time for the better. After months of infusing in a local hospital with no questions asked, a doctor came in to start my IV and asked what I was doing. I told him that I had AATD and it was replacement. I said "they" gave me two years to live. He laughed and quickly put that notion to bed. He let me know that if I died in the next two years, it wouldn't likely be from AATD. This doctor was doing research with an Alpha-1 expert at Royal University. My weekly visits soon became a spectacle. The team assured me that with proper exercise, nutrition, and compliance to medications, I could crush those 730 days.

The conversation turned me around. I headed back to the U.S. and hit the ground

running. I read everything I could, found a new doctor, brought materials to her, and we reviewed all the research updates we could find. I became involved with the Alpha-1 Foundation and met world-class AATD specialists. Several patient advocates had the same "two year" prognosis and had shattered it many times over.

My doctor asked "What matters most to you?," making it clear that I had to take a lead role in defining and reaching my success. With answer in hand, we worked out a treatment and follow-up plan, and as my disease progressed I was referred to a transplant center. I worked very closely with them, and we were a team. On March 26, 2013, I received the gift of life from a young man taken from his family too soon.

On the flip side of transplant, my team and I continue our regular and open communication. We use phones, emails, apps, electronic health record platforms, and meet face to face. They still cheer and motivate me, and they make me feel I am never alone in this battle.