Angela Merkens

ALPHA-1 ANTITRYPSIN DEFICIENCY

Angela Merkens was a patient speaker at the ATS 2018 International Conference in San Diego, California.

When I was 30 years old, I lived in Flagstaff, Arizona and I loved to hike, bike ride, and run. I had my dream job, I was working for a biotech company doing research and product development in the lab. One day in the lab I had a chemical exposure to a severe lung irritant. The chemical exposure caused an inhalation injury, which led to a huge inflammatory response in my lungs.

From that day on, my life has never been the same. I now have a chronic cough, dyspnea and fatigue. The elevation of Flagstaff is 7,000 feet, and within four weeks, I had to move to a lower elevation because I couldn't breathe. My family had to come out from California to pack my apartment and move me down the mountain to Phoenix, where the elevation is closer to sea level. I was no longer able to work, and I eventually lost my job.

I became so deconditioned that I had to use an electric scooter at the grocery store. I had to choose whether to go grocery shopping or go to lunch with friends, because I couldn't do both in the same day. I was on a daily dose of prednisone (20-40 mg) for over two years, four nebulizers a day of Albuterol and several steroid inhalers. Nothing helped my dyspnea.

I was evaluated by five pulmonologists over two years and they all missed an Alpha-1 diagnosis. One pulmonologist suggested that I have an open lung biopsy, and another suggested that I go on a chemotherapy drug. Luckily, I did not do either. Two of the five said that there was nothing wrong with my lungs and attributed my shortness of breath to psychological issues.

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At 32 years old, I was diagnosed with COPD and I had become so deconditioned that my physician recommended that I attend a pulmonary rehab program. There, a respiratory therapist told me about a rare genetic disorder called Alpha-1 Antitrypsin Deficiency. I decided to take advantage of the testing the program offered. It was a simple blood test.

Two weeks later, I received a call from my pulmonologist. He confirmed the Alpha-1 diagnosis and explained that this was the missing piece as to why I was not improving. He went on to say that while there is a treatment to slow down the damage, Alpha-1 is a chronic progressive disorder, and there is no cure. In the end-stages, it can require a lung transplant.

I soon learned that the Alpha-1 protein, Alpha-1 Antitrypsin, is made in the liver and goes through the bloodstream to the lungs to turn off an inflammatory process, and that the Alpha-1 protein inhibits neutrophil elastase in the lungs. Alpha-1 is also known as the genetic form of COPD and/or liver disease and can cause panniculitis.

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The treatment for Alpha-1 is a weekly IV infusion for life of an FDA approved Alpha-1 proteinase inhibitor plasma product. I am now 38 years old, and I have been receiving weekly IV Alpha-1 proteinase inhibitor infusions for about 5 years. I am not as short of breath, I am able to exercise longer, I no longer use an electric scooter, and I do not get as many respiratory infections. However, I still use oxygen when I fly or go above 3,500 feet.

For the past four years, I have had the privilege of being a support group leader for two Alpha-1 Foundation Support Groups. The support groups are for patients with Alpha-1 and their family members in San Diego and Los Angeles, California. ■

Alpha-1 Antitrypsin Deficiency

Alpha-1 antitrypsin deficiency is an inherited form of emphysema. People with the condition, also known as AAT Deficiency or alpha-one antitrypsin deficiency, 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 chemical enzymes in the tissue that cause emphysema. Alpha-one antitrypsin deficiency also can also cause liver damage. Alpha-one antitrypsin deficiency testing is recommended for certain groups of people, including those who have:

- Family history of alpha-one antitrypsin deficiency.
- Early-onset emphysema (less than 45 years old).
- Emphysema without an obvious risk factor such as smoking or occupational exposure to a substance known to cause the disease.
- Emphysema that is worse at the bottom of the lungs.
- Difficult to control asthma.
- Recurrent pneumonia or bronchitis.
- Unexplained liver disease.
- Patients diagnosed with COPD should consult with their health care provider to see if they would benefit from being tested for alpha-one antitrypsin deficiency.

Learn more: ATS Patient Education Series What is Alpha-1 Antitrypsin Deficiency? New York, NY. ***www.thoracic.org/patients/patient-resources/resources/alpha-1-antitrypsin.pdf***