Barbara Pusey

ALPHA-1 ANTITRYPSIN DEFICIENCY

"I have been so amazed at the positive attitude of patients in this community. They have taught me to cope, and they inspire me to keep going."

I live in Fort Collins, Colorado, with my husband. I am surrounded by my family of six children and eight grandchildren. I have a bachelor's degree in nursing and a master's degree in education. I spent most of my nursing career in hospitals working in the ICU or ER. I also taught nursing for several years. I started four urgent care/occupational medicine clinics, and I am most proud of starting a proprietary school to train paraprofessionals.

I was diagnosed with alpha-1 antitrypsin deficiency in 2003. I had never heard of this diagnosis –whether through our teaching curriculum or in the emergency room. I was totally blindsided. I remember driving home to look up alpha-1 in my nursing textbooks and found no information there. This was the pre-Google era.

I had a personal health history of bronchitis at least twice a year, which I either ignored or handled with antibiotics. In late 2002, I had pneumonia, and in early 2003 I had more severe pneumonia.

The doctor I worked with sent me to a hospital for a CT scan. When the radiologist called and stated I had emphysema, I defiantly told him he had the wrong films. I had never smoked.

It all went downhill from there. Once tested, I learned I was a ZZ. (Alpha-1 is caused by mutations in the SERPINA1 gene, and a ZZ is an individual with two copies of the Z allele.) I received a recommendation to start augmentation therapy, but I was in so much denial that I didn't. After about six months, I went to a Denver Education Day and observed other alphas. I decided I should help myself and began therapy. Since, I have only had three upper respiratory infections.

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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

On the one hand, getting the diagnosis was awful. On the other, the diagnosis helped answer a lot of questions in the back of my mind: Why could I tap dance for two hours but get out of breath going up two flights of stairs to the studio? Why could I ride my bike for miles until I came to a hill and have to get off and walk? Why did I feel out of breath when going up to Copper Mountain to ski?"

Living with alpha-1 means I can no longer do all the things I want to do. Some of my daily activities are a real struggle. I wish I had been tested and diagnosed earlier in life so I could have made better choices. Most alphas estimate that it takes seven years and three doctors to receive a proper diagnosis.

When I finally came to grips with my diagnosis and began therapy, I also took a position with AlphaNet as a coordinator. Founded in 1995, AlphaNet's mission is "Alphas Helping Alphas." The group employs over 40 coordinators with alpha-1. These coordinators make monthly calls to patients and help implement disease management programs. I have been so amazed at the positive attitude of patients in this community. They have taught me to cope, and they inspire me to keep going.