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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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.