Jon Hagstrom

Alpha-1 Antitrypsin Deficiency

I have Alpha-1 Antitrypsin Deficiency, “Alpha-1” for short, a rare genetic disease that can affect the lungs and/or the liver. I want to share my own experience with this condition, as it highlights the human cost of late detection, which is all too common for people with my disease.

As the condition is genetic, I’ve had it since birth. But there is no newborn screening in place to catch it, and I made it through early childhood without any symptoms that might have precipitated an early diagnosis.

As was common in the late 60s and 70s, I grew up surrounded by second-hand smoke. Had we known about my condition, that might have been avoided, or maybe my parents would have discovered their own elevated risk of lung disease and quit smoking - they both carried a single allele for the disease, resulting in higher risk for lung and liver disease than normal, though far less

than I had with my two alleles. In my teenage years I added first-hand smoke to the mix, a habit I maintained through my 30s. That certainly would not have happened if I knew about the special risks I had as a person with Alpha-1.

When I was 13, I had blood work done which showed significantly elevated liver enzymes. Today that might have led to testing for my condition. In the early 80s there was even less awareness of the disease, and I went undetected.

In my 20s, an aunt who had never smoked died relatively young from lung issues. Under a more rigorous detection regime, that might have set off a cascade of testing in the family, warning me before it was too late.

Finally, in my late 30s my breathing was bad enough that I decided to see a pulmonologist. I then had lung function (FEV1) of only 29 percent of normal. Based on my age and the severity of lung damage I had, my pulmonologist tested me on my first visit. I remember him saying “There’s this odd disease you probably don’t have but I’m going to test you for it anyway…”

I was lucky to have been diagnosed on my first-ever visit to a pulmonologist. The average lung-affected Alpha sees three different medical professionals and goes six to seven years between first reporting lung issues and finally getting diagnosed.

Upon testing positive for Alpha-1, I started weekly intravenous infusions of the Alpha-1 Antitrypsin protein – then and now the only therapy approved for Alpha-1 lung disease. It does not cure the disease, and it doesn't reverse existing lung damage, but it can slow the further decline of lung function. But by the time I started it, too much damage had already been done. I can't know for sure whether augmentation slowed my decline but if so, it didn't slow it much - within six years of diagnosis I was referred for a lung transplant.

Such was my disease course - the bulk of my decline happened before I even knew I had the disease. Even if a cure had existed and had been given to me immediately upon diagnosis, at 29 percent lung function, I still would have lived a very constrained life, with many normal activities beyond my reach.

My story is not atypical. Indeed, most people with my condition never get diagnosed at all. Some liver-affected Alphas get diagnosed at autopsy. This is unacceptable considering testing has been available for decades, and technologies such as direct-to-consumer genetic testing is only making it easier.

This is a problem we need to fix. At the individual patient level, time is critical. This disease can move quite fast, as my own story confirms. In 2010, I had a high energy, demanding lifestyle, working at an investment bank, and splitting my time between London and New York. Nobody looking at my life from the outside would have ever thought of me as sick.

A mere three years later I was officially disabled. I no longer carried my passport and briefcase. Instead, I dragged an oxygen concentrator with me

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everywhere I went, including to my son's kindergarten orientation. He called it "Daddy's special air". I had to hire someone to play catch with him because I couldn't do it myself. That tremendous change happened in just three years, between 2010 and 2013. But it continues. Three years after that, my lung function was at 14 percent as I was wheeled into an operating room for a bi-lateral lung transplant. I walked out of the hospital with a six-year life expectancy and a seven-year-old boy waiting at home. We need to do better for the Alphas who come after me. And one of the ways to do that - earlier detection - is something we can do now.

Alpha-1 Antitrypsin Deficiency

Alpha-1 antitrypsin deficiency is a genetic condition that decreases lung protection resulting in an inherited form of emphysema. People with the condition, also known as AAT Deficiency or alpha-1 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.

- Alpha-one antitrypsin deficiency is an inherited condition. Every person inherits two AAT genes—one from each parent. Inheriting two abnormal AAT genes causes very low levels of AAT in the blood.
- AAT deficiency lung symptoms usually appear after age 30 but may emerge earlier or much later. The first symptom is usually shortness of breath during daily activities. Other symptoms include wheezing and decreased ability to exercise.
- AAT deficiency only can be detected through blood tests. One type of blood test measures the body's level of AAT. If the AAT level is lower than normal, your healthcare provider may order a genotype or a phenotype blood test.

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