

I Have What?

My name is Lindsay and my story begins when I was 16. Basketball practice started as any other practice –drills, running, etc. - until my foot went numb. I couldn't feel the ball or heel of my foot. My trainer didn't have answers, so my dad was called and he took me to the emergency room at Cardinal Glennon Children's Medical Center for an evaluation. During my visit, many blood tests were run. The results showed my platelet count was extremely low and my white blood count also low - I was sent home with the plan to do more blood work in 2 days. The blood work remained the same. Concern over what the diagnosis might be was evident in the practitioners – Leukemia was the main concern. I was referred to the Costas center at Cardinal Glennon where more bloodwork, a bone marrow biopsy and an ultrasound of my spleen led me to see Dr. Jeff Teckman. Dr. Teckman entered the exam room, examined me, asked questions, and ordered more blood work – I felt like a pin cushion. Dr. Teckman mentioned the possibility of alpha 1 antitrypsin deficiency. I was relieved to know that I did not have leukemia, but created more questions over this disease that I had never heard of. The test results gave a positive diagnoses of Alpha 1 antitrypsin deficiency – **"I have what?"** Alpha-1 is an uncommon, inherited disorder that can affect liver function and/or lung function. It may progress to cirrhosis and liver failure. Alpha-1 antitrypsin, also called AAT, is a protein made in the liver. Normally, the protein travels through the bloodstream. It helps protect the body's organs from the harmful effects of other proteins. The lungs are one of the main organs that the AAT protein protects. AAT deficiency occurs if the AAT proteins made in the liver aren't the right shape. They get stuck inside liver cells and can't get into the bloodstream. As a result, not enough AAT proteins travel to the lungs to protect them. This increases the risk of lung disease. Also, because too many AAT proteins are stuck in the liver, liver disease can develop.

After Dr. Teckman discussed the disease with us he recommended that I give up basketball, a sport that I loved, due to my enlarged spleen. I attempted to play for a season with padding, but found it hindered my playing ability. So, I made a very difficult decision – I gave up basketball a decision that completely broke my heart, but I found volleyball. I played volleyball during high school, and club volleyball in the off season. Upon graduation, I was invited to play on the volleyball team at Webster University for the 4 years of my college career.

Dr. Teckman, followed my bloodwork, ultrasounds and exams every 6 months. I recently transitioned to an adult hepatologist, Dr. Bruce Bacon. I have not allowed the disease to hinder my ambitions.

I'm now 22 years old and strive to maintain a healthy lifestyle which consists of exercise, healthy diet and the drive to succeed in anything I choose. I have recently graduated from Webster University with a degree in Computer Science, a minor in web development and a certificate in web design. I have recently accepted a job as an Implementation Specialist. I have also been accepted to the graduate program at Webster University, seeking an MBA. I also volunteer for the Friends of SLU Liver Center.

Currently, the only cure for Alpha-1-Antitrypsin Deficiency is a liver transplant. Research continues in the hope that a cure will be found in the near future. How can you help? I ask that you please consider volunteer opportunities with the Friends of Saint Louis University Liver Center or please make a donation on the Friends website to support research. Thank you for your support!

