Sarah's Story

A three-year journey

I am 21 years old and first got sick when I was a sophomore in high school. I had lost 23 pounds and started developing weird neurologic symptoms. For example, my eyes stopped tracking left and right, I couldn't swallow well, and I began to pass out frequently. I was passed along from specialist to specialist so they could rule out the things in their field. I heard everything from brain tumor, cancer and untreatable autoimmune diseases to rare fatal genetic diseases. I went on the journey through specialists for about three years.

My world flipped upside down

In February 2013, my world was flipped upside down. I was a freshman in college and was living with my parents at the time. On Valentine's Day I woke up in the middle of the night and knew something wasn't right. My heart rate was over 160, and I was dizzy and shaking. I went downstairs to wake up my parents. My mom checked my heart rate and knew something was off, so she went to grab something. By the time she came back, I had passed out on her bathroom floor. When she finally got me to come to, I couldn't feel my left leg or arm, and I was extremely weak. They rushed me to the emergency room where it was eventually determined that I had a stroke-like episode. I was admitted to the hospital to determine the cause but no one could figure anything out. I was so weak and my left side wasn't working. I couldn't walk, shower, or move around without assistance. Five days later I was discharged from the hospital and my parents were told I would be in a wheelchair until they figured out what was wrong with me.

Finally a diagnosis

I went home in my wheelchair and started to adjust to my new life. It was so crazy to be an 18-year-old college student who was completely incapable of doing anything for herself. A couple of months later I had another episode. These episodes quickly led me to travel to see a neurogeneticist in Atlanta. After a few appointments and an unbelievable amount of testing, I was diagnosed with Lambert-Eaton Myasthenic Syndrome (LEMS) and another rare condition.

So we thought "what now?". I had a diagnosis but we knew nothing about the disease, how to treat it, or how to make me feel better.

A clinical trial, a potential new treatment

I found a support group on Facebook and that helped me understand it a little bit. I started to get infusions of a readily available medication once a month. It did help by restoring the feeling in my hands and feet, as well as and increasing my energy, strength and stamina.

I had learned to live my new life. I was walking again, doing a lot of things for myself, and had started going back to school. Then I heard about a LEMS conference in Orlando put on by the National Organization of Rare Disorders (NORD). My mom and I decided we should drive down there and see what it was all about. At that conference, I found out about a medication that had undergone clinical trials and was preparing to be submitted for FDA approval. I was so excited. I did not have much hope about living with this disease. But here we are in 2016 and I have so much more hope knowing about the possibility of a new FDA-approved medicine.