

## **Advocate Like a Mother**

Both of my children were born with a rare neuromuscular disease called SELENON related myopathy. To date, there are no treatments and no cure for their disease. We can only treat the symptoms which include muscle weakness in the neck and core, severe scoliosis, reduced respiratory function, and poor flexibility.

We had no idea this disease was lurking in our genes when we decided to become parents. So little is known about the disease that a genetic cause was not discovered until about 10 years ago by a research lab at Boston Children's. This means we weren't able to access a genetic diagnosis until my sons were 5 and 8 years old. This lab is the only place where SELENON research is happening in the United States, so the odds are low that a treatment or cure will be developed soon.

Besides this long and expensive diagnostic journey, our family has faced challenges that are far too common among people affected by rare disease. We've had to fight to have life saving medical equipment and other medically necessary services covered by our health insurance. Doctors, who aren't on the wonderful team we have built for our sons, often have never heard of our sons' disease because it is so rare.

We got our first taste of ableism when our youngest son was dismissed from a private school because of his disability at age two and have experienced it many times since. Most recently, we listened as the lives of our children, who are at high risk for any respiratory illness, were routinely disregarded in the midst of a global pandemic.

I never intended to get involved in politics or to become an advocate, but given these experiences, how could I not?

In 2019, I traveled to Washington D.C for a rare disease legislative conference and met with my Members of Congress for the first time. Through this experience, I became empowered to tell our family's story and how the challenges we face can be easier through research, legislation, opportunity, and collaboration. I also discovered that rare disease isn't rare. Over 30 million Americans are affected by a rare disease, sharing similar journeys despite the differences in disease. Through my advocacy, I have met a community of rare disease families that are hopeful, passionate, and devoted.

So I will dig my heels in and continue to fight, because this is not the best the world has to offer those affected by rare disease. I will never stop speaking up for and advocating for my sons. I can't imagine being their mother any other way.