Health & Wellness

Pine Brook Resident Advocates for Patients with Rare Muscle Disease

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MONTVILLE, NJ – When a family member is diagnosed with a debilitating condition, it’s natural to reach out to not only family, but others whose loved ones are going through the same thing.

However, when this happened to Pine Brook resident Sarah Foye, her family member’s condition was so rare that finding others to reach out to was like finding the proverbial needle in the haystack.

In her case, her family member had been diagnosed with a muscular disorder called centronuclear myopathy (CNM).

“We lived in fear during that time, because we knew very little about our family member’s condition,” Foye said.

Her career as an occupational therapist and her parents’ passion for learning led her to become an advocate for other families who have a loved one with this disease. Her husband is a physician, and his knowledge and expertise help support the advocacy as well, she said.

In 2006, they went to the Joshua Frase Foundation Gala in Boston and met the researchers the foundation was supporting. It was there that they met the Frase family and researcher Alan
Beggs. The Frase Foundation supports research in the field of CNM, as well as supporting patients and their families.

“We had no idea any of this was going on,” Foye said. “It opened our eyes to a whole new world of making a difference. When we saw that there were opportunities for making a difference and that things were being done, we started taking on more advocacy. It became my passion.”

Any disease is difficult to go through, but rare diseases are made more difficult because of the diagnosis mistakes doctors often make, Foye said. Patients with muscle diseases are misunderstood.

“Some people are diagnosed with depression, laziness, or fibromyalgia,” she said. “This is a major problem, especially with the adult-onset cases, because the symptoms include muscle weakness or pain. Our journey took 12 years. We had negative and normal results reported back to us in 13 tests. We have been to the NIH and doctors all over the area. We would go, draw blood, wait 4-6 weeks for results... This went on and on for years.”

Foye says they would repeat the process every time a gene was discovered, and on two occasions she and her husband collaborated on a grant to allow a new gene to be available for testing in a commercial laboratory.

Finally, with Beggs’ help, they entered a study and were able to find the diagnosis they had been waiting for: their family member was diagnosed with titin, which is the cause of more than 10 forms of muscle or heart diseases.

“It’s the largest protein in the human body and one of the reasons we were never able to get a diagnosis in the past,” Foye said. “Because the gene has 366 parts, genetic testing methods had to basically catch up to the ability to diagnose the disease. Most genes only have about 20 parts.”

Now, with diagnosis in hand, Foye has focused her efforts on advocating for other patients with problems due to titin mutations.

“My aim is to serve as a link between affected family members and other stakeholders,” she said. “We have families connected on social media. I host and manage an informational website. I provide patient and family education, and patient support. We’ve had online webinars and meetings. We had a team of collaborators write care guides for patients.”

Due to her work as a patient representative, Foye and Alison Frase were invited to a workshop sponsored by the European Neuromuscular Centre (ENMC) held in Heemskerk, the Netherlands at the end of April. The ENMC was founded “by a group of European patient associations dedicated to bringing leading researchers and clinicians from all over the world together,” according to the
ENMC website. The organization sponsors workshops where the professionals meet in person to collaborate.

“My role was to bring the patient and family role to these scientists, because a lot of them are in the lab and never have patient contact,” she said. “They love to have information that ‘what I’m doing in my lab affects this person.’”

Foye says the biggest result of the workshop was a database of clinical information. Each researcher might have ten cases, she said, but together, it pools into a bigger amount of information. The group is working to collect 100 cases into the database.

“Going to the workshop, I gave my whole heart,” Foye said. “All of my hopes, dreams and fears, I put it all on the table - that’s what I felt like. It was so amazing that I had a platform to participate in. It was really exciting. It was an honor to participate.”

In the meantime, Foye is also governing board president of the Congenital Muscle Disease International Registry. The organization has a biobank, a tissue repository, and a patient registry.

“We work with a lot of genetic testing centers,” she said. “If researchers need titin cells, they can’t just pull them out of the air. They may not be able to do outreach to families, but I can.”

Foye shares the researchers’ work with the families, which she says bring hope to the families. In turn, sometimes the families will support the research with funding or enrollment.

“Titin is complicated, research is slow, but our needs are urgent,” she said. “When you’re losing loved ones, it’s hard. I know it’s hard for the scientists too. Research is so incredibly slow.”

“Testing wasn’t finding titin before,” Foye said. “I do this [advocating] because my dream is that other families can get a diagnosis with a spit kit or a finger stick, instead of waiting 12 years or being told they’re mentally ill. Patients need to not only get a diagnosis early, but have a plan of care that they can follow to help prevent illness or even early death. That’s the vision that keeps me involved.”