Solving a 40-Year Medical Mystery

By Tara Voogel

Imagine you have a very active life as a mother and professional. Then suddenly, inexplicably, something changes. You become weak. Everyday tasks become harder to complete. Moving becomes difficult. Multiple trips to the doctor do not provide immediate answers or solutions. And to make matters even worse, your siblings display the same symptoms. This is my story.

My family's medical journey began more than 40 years ago. Over the course of that time, my siblings and I suffered from the same symptoms. We set up appointments with our physicians and participated in several tests and studies in search of a diagnosis. The question no one could accurately answer: why is this happening?

The years passed. Our symptoms became progressively worse. My oldest sister received a diagnosis of Limb-Girdle Muscular Dystrophy, which turned out to be incorrect. In a heartbreaking turn of events, she passed away. Then in 2010, finally, I received an answer to the question that had been plaguing us all of these years. I was finally diagnosed with GNE myopathy, which is also commonly known as hereditary inclusion body myopathy (HIBM), distal myopathy with rimmed vacuoles (DMRV), or Nonaka myopathy.

GNE myopathy is a rare, progressive muscle disease caused by mutations in the GNE gene that affects the production of sialic acid. GNE affects the lower and upper extremities, sparing the quadriceps. People with GNE myopathy typically show muscle weakness around 20 to 30 years of age and progressively lose muscle over the course of a lifetime.

Because GNE myopathy is such a rare disease, it is not typically known or understood among the general public or even within the medical community. It is often misdiagnosed for other conditions with similar symptoms, such as Limb-Girdle Muscular Dystrophy, Charcot-Marie-Tooth disease or Miyoshi myopathy.

What began as a diagnosis set a new course of direction in my life. At 58, I'm now an advocate for speedier GNE myopathy testing and diagnosis. My focus is to educate the public about this condition through my personal blog (taratalksgnemyopathy.blogspot.com), social media, and through a global online community for other patients living with GNE myopathy. People need to know about this disease and understand that support is available.

Some of the most common symptoms of GNE myopathy include:

- Foot drop: this happens when the muscles in charge of flexing the ankle and toes are weakened in such a way that the person needs to drag the front of the foot and bend the knee to lift the foot higher than usual.
- Waddling gait: due to the weakness of the pelvic muscles, the person uses the torso to help move the legs, resulting in an exaggerated waddling movement of the body.
- Loss of balance
- Difficulty walking up/down stairs

If you are experiencing any of these symptoms, I urge you to talk to your doctor and ask about GNE myopathy. You can learn more at www.gne-myopathy.org.

Additionally, free genetic testing is available to help patients understand how the disease affects them. For more information, please visit testingGNEM.com.

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