In 1986, at the age of 28, Todd began experiencing back pain while working on a horse farm. Two years later, his arms would “give out” while working on equipment above his head. He just thought he was out of shape, so he did not seek medical attention. In 1992, Todd was working as a high school shop teacher and was experiencing severe pain. He said his legs and hips were “killing him.” He took Tylenol® to help manage the pain. By 1997, he noted that he was often tripping while working or hiking. “My hips would give away, and it was like falling down in slow motion. There was no way I could stop myself,” he said. His wife, who was a nurse at the time, finally pushed him to see his family physician. Suspecting spinal disc problems, he referred Todd to a neurosurgeon, who ordered an X-ray and an MRI.

A diagnosis
The neurologist ordered bloodwork and an EMG, and had Todd perform a strength test. Todd’s serum CK level was 400 IU/L. The neurologist’s diagnosis was polymyositis, but he told Todd it was “not definite.” A few months later, Todd was experiencing difficulty breathing while lying on his back. The neurologist referred Todd to a respiratory specialist, who prescribed a BiPAP machine and referred him to another neurologist who specialized in myopathies. This neurologist ordered a second EMG and a muscle biopsy of Todd’s back.

Management
Todd was prescribed low-dose prednisone for the next year. He experienced weight-gain and double vision, but no improvement. The neurologist then prescribed Imuran® (azathioprine) for six months, but there was still no change in Todd’s condition. Next, he received monthly intravenous immunoglobulin (IVIG) infusions for a period of six months. There was still no improvement.

*Tylenol is a registered trademark of McNEIL-PPC, Inc. Imuran is a registered trademark of GlaxoSmithKline Inc.
**Muscle weakness**

By 2001, Todd’s arm and leg muscles were getting weaker, and his back pain was increasing. The second neurologist told Todd nothing more could be done for his condition except exercise. Todd did water aerobics twice per week, which he said helped him maintain his strength. For the next three years, Todd went back and forth between the two neurologists. His serum CK level remained between 400 and 500 IU/L. In 2004, he asked if there was another specialist he could see. He was referred to a third neurologist who specialized in the management of polymyositis. He suspected inclusion body myositis and ordered another muscle biopsy, this time on Todd’s shoulder.

**Still no resolution**

Todd had routine annual visits with his first neurologist over the next several years. His serum CK level remained between 400 and 500 IU/L. The neurologist consulted another physician in the practice and asked him to examine Todd. He suspected limb-girdle muscular dystrophy (LGMD) and told Todd, “Even if we can put a name to this, there isn’t anything we can do about it.” Frustrated, Todd decided to stop going to these neurologists. At the suggestion of his wife, Todd began to see a rheumatologist in 2010. The rheumatologist put him on a methotrexate regimen, but after six months, there was no improvement.

**A clinical trial**

In 2011, Todd read an article in Quest magazine — a Muscular Dystrophy Association publication — about a LGMD clinical trial at New York University (NYU). Study investigators were recruiting adult subjects with a difficult-to-classify form of muscular dystrophy or myopathy, speculating that some undiagnosed muscle conditions may be late-onset Pompe disease. Todd contacted them about participating. The study coordinator was not sure Todd met the criteria until he mentioned that he used a BiPAP machine at night. The study coordinator sent Todd the paperwork and a finger-prick blood test that Todd performed at home and mailed back to NYU.

**Conclusion**

Todd’s story illustrates why Pompe disease should be considered in the differential diagnosis when a myopathy is suspected in patients who present with progressive muscle weakness. You can screen for Pompe disease with a rapid and reliable blood test.¹ Ask your Genzyme representative about free testing options.

“I would get so frustrated. All these years I kept saying there’s got to be something else wrong with me.” – Todd


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