

My Pompe Story

In my mid-thirties was when I started noticing a problem. My gait was off, it was getting harder to get up out of a chair and going up stairs was getting more and more difficult. This was in the early 1990's. After writing it off as a "bad back" for a few years, I finally decided to go see about it. This was in 1998. After being tested for ALS, AIDS and a few other diseases and after two muscle biopsies, I was diagnosed with Polymyositis. Well at least I had a diagnosis. I took medications for PM for about a year when I realized I was getting worse not better. I asked my rheumatologist if I can stop taking the meds, he agreed. So for the next 8 years, while I got worse and worse, I thought I had PM, until my troubled breathing is what brought me to the emergency room on March 24, 2007. I have not lived at home with my family since. My doctor in the ER told me if I waited one more day I would have either went into a coma or died from carbon dioxide poisoning. He put me on the bi-pap mask but it was not working for me. I had a tracheoscopy a day after going to the ER. It took a few more months and another biopsy to get the correct diagnosis of Pompe Disease.



During one of my rare visits home I am shortly together with my wife and son.

In December 2008, I was in touch with a doctor who told me he would take the trach out and I'd be home in two weeks. I transferred to his hospital, within an hour of being this doctor's patient, the trach was out. I was put on the bi-pap mask at night but found myself usually wearing it all day long. Two days later I had to be intubated(sp?). A week later we tried again but in a day or two I had to be intubated again. After 2 months the doctor realized I needed the trach. It was put back in and I was transferred back to the long-care facility where I am currently residing.

I've been away from home for over 4 years, away from my wife and 8 year-old son who was 4 years-old when I first was admitted to the hospital. I started on what was then called Myozyme in the summer of 2007 through a clinical trial started by Genzyme. I was on the trial for 3 years after which the drug was approved in the United States. I get up from my hospital bed to walk a little bit each day, but I need a walker. But I do feel the drug has kept me stable. I also have a lot of upper body strength. When I was first hospitalized I was in terrible shape, but now I am ready to go home and get on with my life and I think Myozyme has a lot to do with that.

After the drug was approved the hospital where I was having the infusion and the hospital I am residing in were at odds as to who would be giving me the drug. The battle went on for 3 months before the facility I'm in decided to administer the drug. Meanwhile I missed 6 treatments. After finally getting back on the drug for 3 months, the facility stopped my treatment because of non-payment from my insurance company. I just received the word (May 20, 2011) that Medicare will start paying for my treatments. I really hope that I will be able to receive my treatments soon again after not having one since January 2011.

While all this is going on, I've been fighting insurance companies to give me enough home care so I can get back to my family.

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