Pompe Disease Treatment

Pompe disease is a rare neuromuscular genetic disease in which certain enzymes are lacking. It can be treated with Myozyme®, an enzyme (alglucosidase alfa) replacement therapy from Genzyme Corporation that can be administered via a bi-weekly infusion. Individuals with Pompe disease relate their treatment outcomes below.

Maryze Schoneveld van der Linde

The Netherlands, maryze@xs4all.nl

I have received this treatment for some years now. When I began, my lung function capacity was 13%. As of July 2007, it is 20% capacity. I can't live without a ventilator, but now I can be without one for much longer. Last week I traveled to Budapest and I was able to go around with only an Ambu® resuscitation bag to ventilate myself intermittently. I have also regained my full energy. Lack of energy is a major problem with Pompe disease. With my increased energy and improved breathing ability, I am able to work fulltime and to travel again.

In my case, I don't think I will ever be able to be without a ventilator, but I do hope that others with Pompe disease who are able to start treatment in time (before the disease has impacted their diaphragmatic function) will not need a ventilator or a wheelchair. Even with a slight improvement in my situation, the impact of the treatment is huge.

Maryze is active in the International Pompe Association (www.worldpompe.org).

Luke Garrett

Glens Falls, New York, maryrick@verizon.net

"IVs? A port in my arm? I have to do this?" My first reactions to the news of receiving Myozyme treatment were a little nerve-wracking. A frequent worry that controlled my mind before starting the drug was the unknown. "What was this drug going to do to me? Is it worth the possibility of a reaction to the medicine?" However, these worries faded as I became accustomed to the treatments.

Now, more than four years of treatments later, the abundant improvement continues every day. Playing baseball and going to school on a part-time basis are now activities that I can easily participate in. The improvements do not end there. My energy levels have also drastically increased, and I push to become even stronger with the help of Myozyme. But it hasn't been an easy road when physical therapy, exercises every day and diligence are required.

Days aren't all schoolwork and exercise. I spend time on my favorite hobbies, such as playing baseball, listening to music, and playing rock n' roll with my father and brother Max. If Max hadn't started to play the guitar four years ago, my inspiration to learn would have never sprouted. I owe it all to him.

My independent breathing is slowly becoming easier, although I still need the ventilator 24 hours a day. Without the help of Myozyme, my diaphragm may have gotten weaker, and improvement of its strength would have reached a plateau unfortunately.

I am grateful for the treatments that continue to be provided, and I hope to keep improving. ▲

Luke Garrett, age 14

For more on Luke's story, go to www.ventusers.org/edu/valnews/val10-2b.html#liv and www.ventusers.org/edu/valnews/val17-1b.html#luk.