

Personal Experiences with Pompe Disease

Theo van der Voort, July 2011

At the beginning of the year 2006 I received the news that I was allowed to participate in a clinical study to determine the efficacy of enzyme replacement therapy. Some good results with enzyme replacement therapy were achieved already in a study with baby's affected with infantile onset Pompe Disease.

In March 2006 I received my first treatment at the Erasmus Medical Centre in Rotterdam. It was possible that I would receive a placebo, because it was a double blind study. Every two weeks my wife and I went to the Erasmus Medical Centre to receive the infusion. It was always a big trouble as my mobility was limited, but I felt it was worth it.

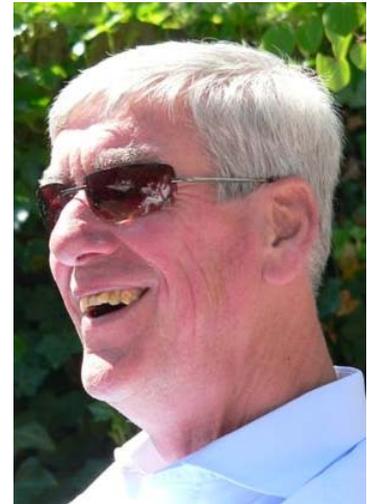
After about 2 months I noticed that my energy improved and that my movements went easier. I was able to comb my hair again and I could bring a cup to my mouth again. It was a great feeling. For the first time in my life I felt my body improving. This feeling can't be described, but it is one of deep happiness.

In February 2006 I was able to walk 235 meters behind a walker in 6 minutes. In August 2006 I was already able to walk 280 meters in 6 minutes and in June 2007 I walked 295 meters in 6 minutes. Then, however, I fell and broke both my upper arms. This meant that I had to be in bed for a couple of weeks, without being able to use my arms I couldn't use my walker and therefore couldn't get out of bed. In December 2007 I recovered so far that I could restart my walking exercises again. This led to a result of 200 meters in 6 minutes.

Unfortunately the improvement didn't return and in March 2009 I only could walk a distance of 140 meters. The deterioration continued and I now worry for my future again. Especially my back muscles worsened and this means that it is difficult for me to stabilize my upper body.

Since August 2008 I receive my fortnight infusion at home. This really is a great solution as I don't need to travel and to stay in a hospital bed for my infusions anymore. I now can continue to do the things at home as I am used to.

Currently my wife and I are busy to get equipment to allow me to function optimal as long as possible. Right now we are dealing with a 'pole' to be able to turn myself around in bed and a shower chair with an elevated seat. All in all I now have the following pieces of equipment: walker, manual wheelchair, electrical wheelchair, a standing up chair, a triple chair, a hoister and a nursing bed. It is very strange to need this equipment, but I am glad they are available. The first problem I need to solve now is my difficulty to turn around in bed I now use cords that are tight at both sides of my bed, so I can pull myself around. I experience that soon this will not be possible anymore. To have the feeling that I am actively fighting the disease, I do physical exercises 3 times a week to keep the least affected muscles as good as possible. If it helps, I don't know, but my feeling is good about it.



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