My Experience with Pompe Disease and Enzyme Replacement Therapy

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First problems (1970)

Bottle feeding (age 2 months)

I was born on August the 30th 1970. I had feeding problems from the beginning. I wasn't able to drink from the breast…I just sucked for a minute and fell back to sleep again. This continued for a few days in which my mother was only busy trying to feed me. The nurses of the hospital said that I was just a lazy baby and left my mother alone with my feeding problems. At home again, my mother decided to start bottle feeding in the hope that this would work better. Unfortunately this didn't work either. My mother became so desperate that she decided to take a pair of scissors to cut a larger hole in the rubber teat of the bottle. This worked…Finally I was able to drink the amounts of milk I needed. Later we understood why I couldn't drink…drinking was just taking too much energy, so I was tired easily and needed my sleep again even when I was still hungry. I wasn't a lazy baby at all, on the contrary I had struggled hard to drink enough, but I simply couldn't.

Diagnosis (1979)

After my mother solved my nursing problems, I developed normally. I started crawling, I walked at the age of 13 months and learned to climb stairs. At the age of 5 it was again my mother who noticed some unusual things. I always had an elevated body temperature, I fell down often, couldn't keep up with other children and I was often sick. My GP told her that 'every child was different ' and that these problems would disappear as I got older.

However the many questions remained: Why was I sick so often? Why couldn't I keep up with other children? Why couldn't I bike like other children? Why did I run that funny? Why did I fall down so often? Again my mother took me to the GP, who then decided to take some blood samples. A few days later the laboratory did find such disturbing lab results that they were alarmed. Further examination was needed. I was admitted into the University Medical Center in Nijmegen just after New Years day in 1979. There the children's neurologist Prof. Gabreëls did tests like an EMG, blood tests and a muscle biopsy. Soon after, my parents and I were invited to meet him. Then he told me and my parents that I suffered from Pompe disease, a rare hereditary progressive neuromuscular disorder. Prof. Gabreëls explained to us what was wrong with me. He explained to my parents and me that there was nothing he could do, that the best thing we could do was make the very best out of it.

Still being able to walk in 1989 while on holiday in Czechoslowakia.

Continuation of my life and disease progress (1979 – 1996)

After the diagnosis, my body deteriorated further: running became impossible, my walking was uneasy with frequent falls. In spite of these problems I graduated from high school normally and started to study Cultural Anthropology at Leiden University in 1989. I moved to a student home, because it took a 3 hours travel by train to Leiden. In my first year of study I started to have problems like: morning headaches, night mares, crying without reason, not knowing myself anymore, having serious concentration problems, not being able to fall asleep, feeling sleepy during the day and being exhausted etc. It was in my 2nd study year (1990) that I was admitted into the Intensive Care Unit to receive my first ventilator to solve my breathing problems. I received my manual wheelchair and scoot mobile in 1993 at the age of 22 years. Now I could go to my faculty and the library easier, go shopping and visit friends without any help.

After I graduated in the Summer of 1995 I decided to return to my parents again. At that time I knew that my body would only deteriorate further and I knew that living independently would put me in a difficult position. Going home was just the best solution for me at that time. 6 months later, in 1996, I started to work with my current employer. A job of 4 hours a week, that fitted perfectly what I wanted: working with migrant and refugee women from Turkey and the Middle East as a community worker.

New medical developments (1996)

At the age of 28 years I had become very thin and was malnourished. My weight was 30 kilo's.

Since November the 14th 1996, we, Pompe patients, know that enzyme replacement therapy has been developed for us. This day had changed my life in every way. I now had hope for the future. Though I did know that it would take time to get the treatment, I realized that now I wouldn't die at the age of 40.

While I was waiting for the treatment that was being developed. I slowly needed more ventilation and I became very thin. I couldn't eat enough food anymore, because I was simply too tired to eat. I needed to go to bed in the afternoon to rest. In 1998, at the age of 28 years my weight was 30 kilo's and I was already in the danger zone. Two weeks later, I went into surgery to get a G-tube. Getting a G-tube was difficult for me, because I felt I was losing everything: I couldn't breathe without support anymore, I needed help with everything and now I was also losing my ability to eat. The good thing was that the G-tube did it's job and my weight increased to 42 kilo's. In 1998 I needed home care, because it simply was too much for my parents to combine work with taking care of me 24 hours a day. In the weekends I live together with my partner Anton, who then takes care of me. While he works during the week.

Meanwhile my arms deteriorated as well. Now I needed help with almost everything: getting me dressed and undressed, showering me, washing me, lifting me to the toilet, lifting me in/out of the wheelchair, bringing me to bed, combing my hair etc. It's very painful to loose these abilities. Actually I felt I was in a continuing mourning process, slowly saying farewell to my body.

Expanded Access Program (2002)

Getting enzyme replacement therapy via an infusion.

In 2002 Genzyme started the expanded access program. I had been selected to receive enzyme replacement therapy! On **December the 11th 2002** I was admitted to the ICU at the Erasmus Medical Center in Rotterdam. At start of treatment my VC sp was 540 about 15% of normal and my FEV1 was 540 and 17% of normal. I was put in bed to monitor me closely, because I would become the 4th late onset Pompe patient in the world to receive enzyme replacement therapy. My parents, Anton, a good friend of mine and my physicians were present. When I received my first infusion I felt happy, grateful and I knew I had a future again. Since that day I drove to Rotterdam every two weeks to receive the enzyme replacement therapy.

Improvements (2003 - ….)

In October 2003, about 10 months after I started enzyme replacement therapy, I did notice a change. Normally I went to bed in the afternoon, something that I had been doing for a couple of years, because I was tired. However one day, I was laying on my bed and wondered what I was doing there, because I suddenly realized that I wasn't tired at all! From that time on I didn't go to bed in the afternoon anymore. Slowly I also experienced that I could eat better. I was less tired, so my swallowing and eating improved too. I stopped using my G-tube while I was able to gain and maintain my weight with normal nutrition on 60 kilo's. I also noticed that my breathing improved. I don't panic anymore when I am without ventilator for a while. On holidays I can be without my mechanical ventilator for quite some time. To be able to take deep breaths now and then I take my balloon (ambubag) with me to ventilate myself manually. Now I have been able to travel to countries like Italy, Baltic States, Poland, Jordan, USA, France, Turkey, India, Lebanon, Hungary, Portugal, UK etc. again. It is wonderful to notice that I am not exhausted after a travel and able to enjoy everything fully.

Together with my sister in Jordan at Mount Nebo.

Being on holiday in France (aged 34) without the constant need of ventilation.

In Spring 2005 I also started to use mouthpiece ventilation during the day. In September 2006 I started to take singing lessons to train and improve my diaphragm. This works out well. I wouldn't be able to sing before treatment. At July 5th 2007 my lung function test showed a VC sp was 650 / 19% of normal and my FEV1 was 600 / 20% of normal. These scores were repeated on June 4th 2008 where I had a VC sp of 630 / 18% of normal and a FEV1 of 610 / 20% of normal. My respiratory specialists at the Utrecht Medical Center were happy and impressed to see these results of small improvement with huge impact, while before treatment my lung function steadily declined continuously. My lung function remained stable since then.

I also do physical exercises like arm and leg training with the aim to keep my body in condition.

Home Treatment (2007)

After exact 5 years of bi-weekly treatments in Rotterdam my physicians offered me the opportunity to get my bi-weekly treatments at home. In the Netherlands a good home treatment infrastructure does exist and it's legal to have certain treatments at home when it does meet the safety criteria. In my situation the home treatment means 6 hours less travel time every two weeks. I can now also continue my work while receiving treatment. During the treatment a specialized and trained nurse is present continuously.

Working at home while receiving my enzyme replacement therapy.

My life and my future (2011 - ….)

Dealing with Pompe disease is a team effort that affects everyone close to me.

I know that thanks to enzyme replacement therapy I - most probably - will also become an old lady like most people in the Netherlands. Experiencing that this would most probably happen I realized that I had to take care of my old age pension. Before the treatment with enzyme replacement therapy I knew from medical journals and my bad physical condition at that time that I wouldn't become older than 40 years, but now my future perspective has changed totally. To be able to take care of my future and my old age pension I started my own consultancy in health and care. My company is called Patient Centered Solutions (www.pacesworld.com). Besides my consultancy I am active in other jobs too including voluntary work.

Enzyme replacement therapy has benefited me a lot. I am able to live an active life again and to participate in society. I now do have a future!

Now I am receiving ERT I am able to give presentations at conferences worldwide without using my ventilator.

Traveling light from Paris to Rotterdam with two ventilators, spare parts, dry battery and one suitcase.

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