

Maryze's story

I was born on August the 30th 1970. I had feeding problems from the beginning. I wasn't able to drink from her 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 to drink enough, but I simply couldn't.



Bottle feeding (age 2 months)

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.

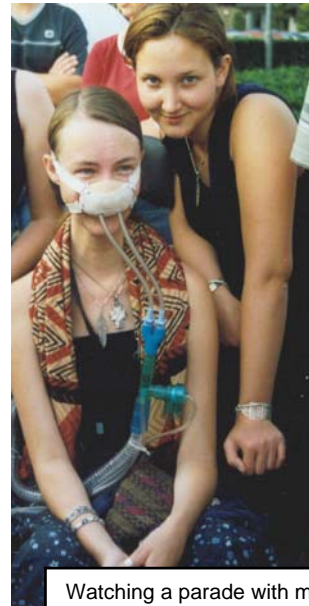
When I had physical exercises in the morning and came home to have lunch, I always suffered from serious stomach aches afterwards. Sometimes the pain was so severe that I wasn't able to attend the lessons in the afternoon. Later we understood that the combination of intense physical exercises combined with eating afterwards, was just too much for my body to cope with resulting in the severe stomach aches.

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 Academic Hospital 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 acid maltase deficiency also known as 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. For my parents the period after the diagnosis was a difficult one. The fact that their oldest child was so sick, suffering from a serious disease with an uncertain future was difficult to cope with.

After the diagnosis, my ability to run worsened and I wasn't allowed to play catch with the kids in the street anymore. I was very upset and sad that they didn't allow me to participate. My mother explained the other children what was wrong with me and why I couldn't run that fast. Now they knew, they decided that I was allowed to participate, but purely decorative for the show. I didn't care, the main thing was that I could join and have fun with the other children. At school my classmates also accepted my disease. I already took swimming lessons before I was diagnosed and no one understood why I failed my 1st swimming exam. All children passed, only I Failed. I didn't understand it at all, because I was doing my utmost best. According to the swimming teacher I did swim 'doggy style' and didn't close my legs and arms properly. It was only after another try and pushing myself to the limit that I passed. I learned typing at the age of 9 years, so I was able to keep up with dictation when writing became more and more difficult.

After I graduated from high school I 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. My GP referred me to the University Medical Center in Utrecht at the Center of Home Mechanical Ventilation. In my 2nd study year in 1990, I was admitted into the Intensive Care Unit to receive my first ventilator to solve my breathing problems. From that time on I wasn't able to travel with the train anymore, because it was impossible for me to carry my ventilator and suitcase. I already had a lot of difficulties traveling by train. I almost fell down the stairs of the train once when I tried to get of the train. Luckily a lady was able to grab me at my shoulders on time. From that time on we knew it was time to get a car. Until I got my own car, my parents and Anton took turns driving me every week to Leiden and to pick me up again for the weekend, so I could continue my study. Later my grandfather decided to give me his car on loan until I got my own car. He said: 'You need my car much more than I do'. Meanwhile my friends and brother took care for me by cooking meals, cleaning my room or picking me up from the floor when I had fallen on the floor. Thanks to my grandfather, brother, parents, Anton and friends I could finish my study.

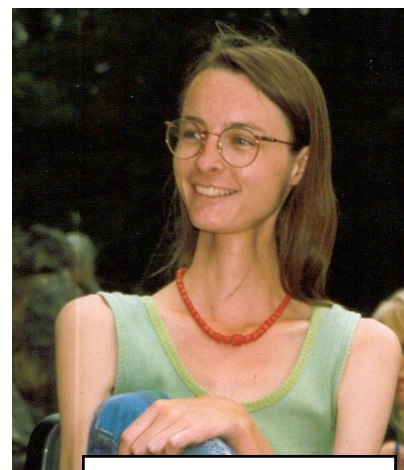


Watching a parade with my sister while I am in my wheelchair and use my ventilator (aged 30)

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 6 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.

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. 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 Anton, who then takes care of me.



At the age of 28 years I had become very thin and was malnourished.

Meanwhile my arms deteriorated as well. Now my helpers, Anton, parents and sometimes my sister needed to help me 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. I cried a lot because I knew I was going to loose my arm functions too. Actually I felt I was in a continuing mourning process, slowly saying farewell to my body.

In 2003 Genzyme started the expanded access program. I had been selected to receive enzyme replacement therapy! On December the 11th my parents and I left early on a cold morning. Stars were sparkling in the sky and when we arrived in Rotterdam a beautiful sun appeared at the sky. Maybe this was a sign? In the hospital we went to the ICU and I was put in bed. 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.



Getting enzyme replacement therapy via an infusion

Since that day I drive to Rotterdam every two weeks to receive the treatment. 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. I wasn't tired at all. Suddenly I realized that I wasn't tired, a new experience that slowly had

entered my life. The day after, I decided to try and not to go to bed. I felt fine and I had a good evening! 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 maintain my weight with normal nutrition. I also noticed that my breathing has 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, so I can ventilate myself manually. Now we are able to visit cities like



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

Venice, Florence etc. for a whole day. It is wonderful to notice that after such a day I wasn't exhausted, and that I am just tired like everyone else. I also started to use a mouthpiece 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. Besides this I also do physical exercises like arm and leg training with the aim to keep my body in condition and to improve my muscle functions. Enzyme replacement therapy has benefited me a lot already. I am able to live an active life again and to participate in society. I now do have a future.

Dealing with Pompe disease is a team effort that affects everyone close to me. I am so fortunate to receive enzyme replacement therapy already. Thanks to Myozyme I can continue my paid job, my voluntary work and can I give presentations at conferences and teach students without using the ventilator.

Unfortunately Pompe disease has been in my life for a long time, a lot of my muscles have been destroyed already, but I can tell you from my own experience that enzyme replacement therapy for Pompe disease does work!



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