Experiences with Pompe Disease

Frank van de Zand, July 2011

My name is Frank and I was born at April 1968. I am married and father of two children (Roy: 1995 and Kim: 1996). At December 2004 the diagnosis Pompe Disease was made.

At June 2003 I went for the first time to my General Practitioner (GP) due to lower back pain. I also went as many others asked me if I had back pain, because of my strange way of walking. My GP referred me to a specialised physical therapist. After one year the therapist noted that I failed to do be able to do some exercises more often. For example, I was not able to bend through my knees and to get up again. My GP then referred me to a neurologist in a local hospital in Zevenaar.

Frank with his two children.

I became a EMG (Electromyography) examination in this hospital In august 2004. Halfway the examination, it became clear that the values were not well. They told us (my wife and me) that I had to go to the University Hospital in Nijmegen and that we had to be prepared for a neuromuscular disease.

Since I could not believe that I had a neuromuscular disease and since I could not accept it, I started to work out very fanatically in the sports school. I had been working out since eight months. I started to do “spinning” during one hour a week. “Spinning” implies riding a bicycle in different tempos on music. During “spinning” you may sit or stand up. Now, I also understand why I was used to lean on my arms more than others: probably because I simply could not get the strength from my legs.

One week later, during our vacation in Austria it became clear to me that something was seriously wrong. During a walk we had to go uphill. This was almost impossible for me. I realized the strength in my legs was much less than it used to be. Back home, I experienced the same while I had to walk stairs or had to lift something from the floor such as a laundry basket or a crate or gardening.

In October 2004 I had several consults with the neurologists at the University hospital St. Radboud in Nijmegen. I got the results on December 2004, the same day at which the EMG examination was done. The diagnosis was Pompe Disease. Afterwards I realized, the diagnosis was made very soon after the first time I went to the hospital in Zevenaar.

Via the University hospital St. Radboud and later the UMC in Utrecht where also some examinations took place, I went to the Erasmus Medical Centre in Rotterdam. Since 2005 I have been participating in a study on Pompe Disease that takes place every 6 months. Every time again I am very curious to see if there is any deterioration to detect. During the muscle power tests and lung function tests I always give 100% to get a clear picture for myself. Unfortunately, in spite of my full commitment, deterioration takes place.

Right now it is more easy for me to deal with Pompe Disease than in the beginning, as I couldn’t accept it at first. Some small adaptions were made at home such as parallel banisters, a higher bed, a freezer with drawers instead of a big freezer box. I still work full time. I always bike to my work (a distance of 11 kilometre), but since a couple of years I use a bike with electronical support. At first I didn’t want to hear about it. Now I am very glad with it, also that my wife convinced me to try such a bike. I now don’t have muscle aches in my legs anymore.

At July 5 2007 I started enzyme replacement therapy. We were very happy that I was allowed to start and I still am!!! The first 2½ years we travelled to Rotterdam every 2 weeks (260 km v.v.). This was never annoying to me. How many people should drive this distance every day for their work?

Since November 2009 I receive treatment in the local hospital in Zevenaar. This is ideal as it only takes me 10 minutes to get there by bike and I am also able to do some work before or after my infusion. My employer and I adapted my working days. On non-infusion days I do work more hours, so I can take a day off at my infusion day. This solution works very good to me and my boss and it makes that I don't have trouble with social institutions.

Since May 2008 I also work at a gym together with my physical therapist Anne. She knows what I am able to do and let me do exercises to keep my muscles in shape and to trigger them once and a while to do a bit more. My arm strength is still good and we assume they will stay fine. My leg muscles are less good and we train them to keep them as good as possible. Every improvement is important.

In my experience Pompe Disease is mainly inconvenient. I don't feel sick and I have not much pain. Luckily I don't have any side effects from the enzyme replacement therapy. I do notice that I have more energy and in general I can say that my condition has stabilized, perhaps with a slight decline.
I experience receiving enzyme replacement therapy as a gift. If my condition stays as it is now, I will be able to continue to work and to do things I want to do, with some limitations here and there.

We can and always could talk easily with others on Pompe Disease. I even received a nickname. Because of my waggling gait my wife and children call me 'Penguin'. Via the International Pompe Association I have also come in contact with other 'Penguins'. Hans Teeuwen (note: a Dutch comedian) once said: 'If you look they waggle, when you don't look they walk normally'. Perhaps we do the same?

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