Living with Pompe

Wilma Treur, September 2011

Written in November 2005

When I was asked if I could write about what it means to live with Pompe Disease - a muscle disorder - I realised that I had never really thought about it in that sense. I do run into all sorts of limitations, but I have never thought about putting everything in a row. So, a nice task!

A good year ago, on the 4th August 2004, I was told that it was 100% certain that I had Pompe Disease. The road to this diagnosis was long. Not long in the hospital, but long for myself. Who am I? I am a single mother, 42 years old, with a beautiful daughter.

Looking back now, I can recognise a lot of things, but at the time I had no idea. Now I can say that I noticed the first signs of muscle weakness when I was around 28 years old. Especially with a sport such as diving. I could not come out of the water easily whilst wearing and carrying full diving gear. But hey, I could do it.
During my pregnancy things became very noticable. I was 33 years old. Getting up from a chair or walking up stairs was very difficult. But I had an excuse ..... I was pregnant. And in that sense I mean that it was a long road.

Wilma with her daughter

One can, for a long time, find all sorts of reasons why walking up stairs is not so easy anymore, or that you are now walking with a rocking gait, or that you are noticing that you are short of breath.Out of condition or maybe pelvic instability? Tired!! Ah, things are very busy at work and in the meantime you have also been divorced. But at some time all that stops. And so it did with me. Becasue it was not pelvic instability and after doing sports twice a week things did not get better.

Then the medical circuit started. Really just by chance. A small test at the doctor's office to check if I had Pfeiffer's Disease. This was in connection with the constant fatigue. Well, it wasn't Pfeiffer's. However liver tests showed abnormal results. Yes, I know, now you are thinking 'Ahh, a silent drinker'. Well, I really do love a glass of wine ..... about three times a year!!!. No, that wasn't it, but the investigations did point out that I had elevated CK levels. And anyone with a little knowledge of muscle diseases, knows that this is not good. So to the neurologist, but he cannot find anything, and yet you know that this is not good. But you push it away, you don't want to know. You hope that you have a deficiency in vitamins and minerals; one bottle of pills and you'll be as good as new. But deep in my heart I knew there was something wrong. Because getting up from a chair without using my hands doesn't work, standing up from squatting down is impossible, climbing stairs is only possible if I use my hands for coordination, and I can no longer lift my daughter off the ground. These problems have been there for a long time already, but your mind and body work together in such perfect harmony that you automatically find other ways of doing things.
Until someone points it out to you and says: "Can you stand up without putting your hands on your knees?". The questions from friends and colleagues - if I have a sports injury or problems with my back. It seems that I walk funny. As if I have to push myself. Everyone means well but all this makes me very nervous.
What is wrong with me? I cannot explain it to anyone, because I don't know myself. I felt stupid and often thought:'Don't be so ridiculous, come on, get up those stairs!'. It makes you terribly unsure. Not that others would have noticed much of that. That's not in my nature. I will manage OK, I am a strong woman who can handle anything. At least for the outside world.

And if I was not like that, what was there to tell? Moaning about being so tired?? From what?? That you want t go to bed early, because you just can't stay up any longer? I am 40 years old, in the prime of my life. Others can do it. Why not me?

Ultimately I took the initiative myself, by again getting my CK levels tested. These were now further elevated. Not good. Back to the hospital. I go to the Internet, searching the muscle diseases, and there it is: **POMPE DISEASE. That's it!!!**

So recognizable. Finally an explanation for all my physical symptoms. It's all about a muscle disease whereby my musclecells slowly fill up with glycogen. I don't make enough alpha-glucosidase (a special enzyme which breaks down the glycogen in the muscle cells). This is the cause of the muscle weakness. When this disease manifests itself at a young age (as in babies), it is a fatal illness. The later this illness presents itself, the better the prognosis. The larger trunk muscles are often the first to be affected. This often leads to problems with climbing stairs or getting up from a chair. It is a progressive muscle disease and that means that I will get worse in the future. But no one can tell me how bad or how fast. But is all that right? Do I have this? Ultimately the diagnosis was made fairly quick, with the help of a muscle biopsy and, later, DNA investigation. It is certain, I have POMPE DISEASE.

Relief shot through me when the diagnosis was made. That may sound weird, but I was really relieved. Now I can explain to people what is wrong with me and the problems I have. This counts especially for support agencies and organisations. Because no diagnosis, no help! I immediately organised a few things such as a chair-lift, raised toilets and handgrips. And help for in the house. I work for 30 hours a week and I would like to keep doing that. That means chasing and organising things myself and searching through a maze of different organisations who can offer help. If I was not so tired already, then I surely would be when I was finished. But much is possible, you just have to know the ways to get there.

Since the diagnosis I feel like I am being tossed backwards and forwards between hope and fear. Fear in the sense that since I know, I have the feeling that I am getting worse faster. I don't know if that is really so, but that is how I experience it. Problems more often with tired, heavy legs. Picking up a book off the floor is nearly impossible. I still do some sport (fitness - under instruction), but in my eyes that does not count for much. I know. I cannot let the fact that I have a muscle disease overwhelm me. I can still walk, so why should I moan. But you look back at what you used to be able to do, and what you can do now. And the constant tiredness!! After shopping for an hour or doing an ordinary day's work I am totally exhausted, and I will often lie down for a while. That's not always nice for my 8 year-old daughter. "Mama is tired, Mama is going to lie down for an hour. Go and watch TV for a wile." But that is happening more often now. What do I organise for the future? I was told that I have a shorter lifespan than the average Dutch person. What does that mean? Shall I continue to make extra contributions towards my pension? For whom? Is it worth it? Many questions, but few answers. It is not real clear how this illness will affect everybody. Seems that everyone is different. Some sit in a wheelchair, some are on oxygen, some both. Where do I belong? But through all the stress that floods over me there is one thing that I hold onto ..... There is hope!

The Pompe Centrum, together with the Genzyme Company have been able to make the enzyme, recombinant human acid alpha-glucosidase. However, at the moment it is only available for people who are taking part in a Clinical Trial, and I am still too good for that. But it is effective. That has been shown in babies and children with Pompe. The effect is not the same for everyone, but one thing is clear - the sooner you are treated , the better the results. The application for approval by the European Authorities has been made. But that takes a long time. 12 to 18 months. And then there is the big question, who is going to pay for it?? It's not cheap! Am I worth it?

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In 2005 I wrote the upper part and it is now 2011. A lot has happened over the past 6 years. I had to wait till 2007 to receive the medicine and when I am honest I must admit I deteriorated in the time I had to wait for treatment. The thought that the medicine was within my reach kept me going and in November 2007 I started the treatment in the Erasmus Medical Centre. It is difficult to say what my thoughts were at that time as, depending on the day, my feelings about the effect of treatment varied. After the first infusion I thought I felt better, while after 3 infusions I felt less well.

My experiences in the hospital are very positive and I learned to know several very nice people; nurses, physicians and other patients. I also found a new friend who received treatment together with me in the first year. Unfortunately Lyda had to stop treatment due to allergic reactions. This was really sad, but we are still friends.

After 2 years of travelling to the hospital in Rotterdam every fortnight, I was allowed to receive my infusions at home. This is really wonderful! It saves me a lot of travel time and I have a nice nurse who works accurate.

I receive treatment for 4 years now and I can say that my condition has stabilized. I feel good and am busy with my work (I work 15 hours a week as advisor of Social Benefits at the Community of Haarlemmermeer), my beautiful daughter and my voluntary work. I am part of the Pompe Group of the Dutch Association of Neuromuscular Diseases (VSN) and additionally I am also board member of the International Pompe Association (IPA). In the past months I also participated in scientific research on the possible effect of intensive physical exercises on muscle fibres in Pompe patients. I followed a training program of 2 hours 3 times a week under the supervision of a physical therapist. It was heavy, but I enjoyed being physically active again en to contribute actively to my own physical condition. The treatment has contributed significantly to my quality of life and my future outlook. When I look at the scientific developments regarding the improvement of our medicine, I expect our future to improve further. It is unbelievable, but right now 3 pharmaceutical companies are doing research to find the best solution for Pompe disease.

If I ever had thought that I wouldn't become old, these thoughts definitely are gone now. I am going to become old, perhaps not walking, but I rather prefer to look at what I still am able to do, than what I am not able to do.

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