Robert’s Experience
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Robert

I was asked to put my experiences with Pompe and Myozyme on paper. And while I realised that this could become a rather long story, I thought “let’s try it”.

I was just nine years old when my parents took me to a hospital to try and determine what the problems were with me. My muscle development was always slower than other kids my age. ‘That’s not such a problem’ one would think, because this would only be noticed with gymnastics etc. Of course also with just playng outside. However, I made sure that certain activities such as running and football were carefully avoided. And because I wasn’t good in those things I certainly didn’t go looking for them. For the rest one could notice little difference in me.

Things changed for me when I was 14, because then I felt that things were getting worse for me when I exerted myself. You don’t realise that straight away because we alll have off days, or days where your staying power is less than it should be.

In the coming years I lost more ground and it seemed that there was no end to the negative experiences. During that period of my life I was very unhappy and I did not know how to put an end to those feelings so that I could pick up my life again and move on.
In a very short period of time my spine began to grow out of shape (scoliosis) and I had to start wearing a corset. My breathing became worse again and within eighteen months I was using nighttime ventilation.

Yes ......, I know. Things were moving very rapidly at that time. However, when you have a lot of bad luck, you tend to think that it has to change at some time.
During the following years I lost more ground and by the time that I was twenty I was in a wheelchair. It was a fold-up chair that I could push myself, and a few years later I moved to the electric version. But before that happened I got a job after leaving school. My first job was totally voluntary. That didn’t matter at that time as I was still living at home and I looked at it as a nice stepping stone to the next job, where I would get paid.
You can see that, even at that time, I was always hoping for something better.
One must never forget to dream!
After that I got a job with a small firm who rented rooms to students.. Back then it was a small foundation, today it is a commercial business.

I worked there for four years and found myself getting worse every year. Slowly I began to notice that I became more and more breathless which took an awful lot of my energy.The more breathless I was, the less work I could do. During the last year I only worked half days. Mainly because I just couldn’t cope with any more than that. But even with that situation I could not cope for long. When I drove home in the afternoons in my custom-fitted car I was so breathless, that I would have a bad headache and I was sitting behind the steering wheel retching as I drove into the street where I lived. I lasted there another three months and from then I spent most of the time on sick-leave.

It took me a month to recuperate and then, after two weeks back at work, I went down again and back on sick-leave with the same problems.
Then I got severe pneumonia, which only made me feel yet more breathless.
After that I got pneumonia again and then, with great difficulty for me, I had to give up my job. However difficult it was to give up work, it was a great relief for my body. I had no illness for at least fifteen months and the rest helped me to recuperate and build up my stamina and resistance to illness.

For the best part of my life I have fought against going backwards and losing gound, and when i look back it seems to me that it only worked to my disadvantage. However, you only see these things much more clearly as you get older. When you think that throughout your life there are certain things which you could have done different, then I think there must be at least óne thing which could have made my life easier.

By the time I was 27, I was so breathless that I could only sit on a chair and concentrate on breathing. I did not answer the phone anymore because no one could understand me. Moving around in my wheelchair also became too difficult so I mainly sat quietly in the corner of the room. I had to ventilate regularly throughout the day and usually I laid down to do that. There was little pleasure in life as I was mainly so tired. Finally I went to full-time ventilation. For me that was a great loss on one side, because I lost the battle against the muscle disease. On the other side there was also gain as I got something back. Namely some freedom, because at that time I also got an electric wheelchair. My ventilator could also be carried on that chair, so that gave me enormous freedom. Later I thought "I should have done this years ago!" But I didn't .... and looking back on what could have been was pointless.


Robert and his daughter in the zoo

The next step in my life also had an enormous influence. After a lot of serious thought my wife and I decided to get pregnant. But first we had long discussions with a doctor. I had to know if there was a chance that the baby could also have my illness. The chances of that happening were absolutely minimal, so we went for it. It was to take eighteen months before she became pregnant.

In the beginning I found it a scary idea to have a baby, knowing that you have an illness which will ultimately claim your life. But then .... no one lives forever, and while you can you should live and enjoy and get out of life what you can. Nine months later we had a beautiful and healthy daughter. At that moment much changes in your life. You start a new phase and start to think very differently about a lot of things in your life. I can well remember that my wife never used her seatbelt in the car because she didn't feel comfortable with it. Since we have our daughter she no longer "forgets".

In the beginning I noticed that I had problems, that I was fearful of holding my daughter. She was so small and fragile and I was scared that I would drop her. We found a solution to that. But still ...... I did not find it easy.


Robert taking the tram

But with a few adaptions and adjustments many problems can be solved. When all is said and done you just have to get on with your life and take it as it comes. I was overjoyed with the little one and am still very happy. Anyone who wants to start a family and has doubts ......I can surely recommend it. Of course, there are always ups and downs in your life. As the little one grew up, I lost more strength. The only way to get through the day was to regularly lie down and let the muscles relax for a while, so that later they could hold the body up again.

The ventilator on the wheelchair was a good move. Also totally necessary because I could no longer do without the ventilation. The headrest on the wheelchair got used more and more so that my neck was supported to hold my head up. As I was in my thirties, sitting in my wheelchair and moving the chair became a big task. Because it became more difficult for me I went outside less and less. And so it became more difficult to just sit, even though the chair was customized.

I also became ill more often even though I let my body have plenty of rest. In the end I was in bed nineteen hours a day and that is a lot of time to think. I got myself in a knot and you could say that I was in a depression even though I had my wife and little one at home with me. This was not a nice time for me and not at all nice for my wife. I tend to be a worrier and keep it all inside.

Then one day there was a phonecall from the hospital to ask if I would try something. That had happened before, but this time it was different. It seemed that a manufacturer was behind this. They had invested large amounts of money and they believed in this medicine.
Some time passed before I could start, but it was a very exciting time wondering if this medicine was going to do anything for me. There were lots of things to sign before the event, but I didn't care about that, I had nothing to lose.

The beginning was a little worrying as no one really knew whether or not I could tolerate this medication. But everything went well, apart from a small skin problem and cold.


Enjoying a day out

The first year I noticed little difference, except that I was less tired than before. The second year the medicine was better mixed and within three months I could nearly stay up all day. Because i had more time and energy I wanted to do all sorts of things. And the very first day that I was able to go without rest was really a milestone. I was tired, but I did it!
It seems very strange to get better after being sick for such a long time. It is a real challenge to start doing things again. Also, I did not get sick so much anymore because I was not so tired at the end of the day.
These days I don't even have a flu injection anymore because I feel that I can easily do without it. But I still use ventilation, I never stopped doing that. I also still use my electric wheelchair.

The years when I was so ill caused such a lot of damage to my body that it will never completely heal. The biggest benefit that I have gained is that I no longer have to go to bed during the day. As well as that I am no longer as tired as I was in the time that I spent nineteen hours a day in bed. The fact that I can be up and about, that I can be with my family, and that I have a life is enough for me. I saw enough of the other side to know how it can be. Especially the fact and the knowledge that I can see my daughter grow up is a fantastic experience for me. So, for me, the glass is half full, instead of half empty. Also having a family is a great driving force, and I think that I could not have done all this without them.

For me Myozyme means a great deal. And the fact that now no one will ever have to be as sick as I have been, heals a lot of wounds for me.

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