

My Morbus Pompe

Nisa Karatekin, August 2011

My name Nisa means "brave woman" but my parents call me their little sunshine or mouse. I was born "healthy" in September 2007 but very soon my mother noticed something was wrong with me. She went to a pediatric and told him that I sleep a lot and I appear to be rather tired. He told her that that is just normal for newborns.

Also during the following months I barely moved. There were additional problems with the way I kept my head. I could not keep up my head normally. My parents started to be seriously worried about my health. They took me to the pediatric again and he redirected us to the physiotherapist who in turn sent us back to the pediatric and so forth. My mother repeatedly claimed she did not have a good feeling about my development. I was not able to normally keep up my head after 4 months. In February I got a cold and fever. My parents went to see the general practitioner, Dr. Maier, who redirected us to the hospital where I had to stay. In the hospital, they took a blood sample which was very painful!

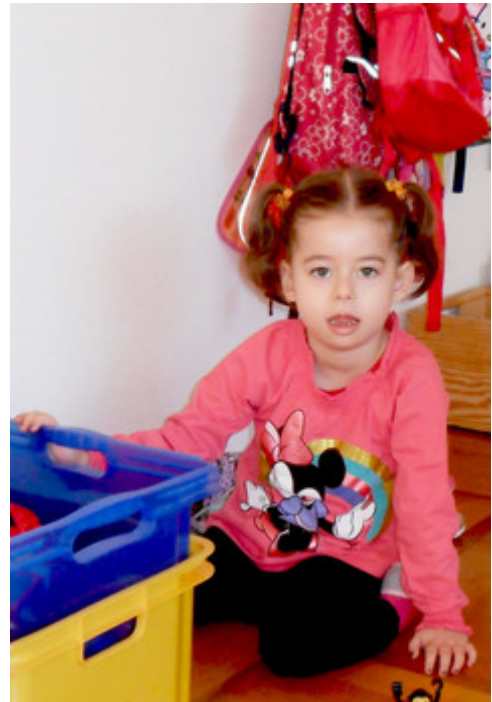
The doctors in the hospital soothed my parents by telling them that I probably got a bacterial infection of the bronchi, which would be easily treatable with antibiotics. I felt better the next day, however, my mother did not sleep during that night in the hospital: she was very worried about my wellbeing. My father did not stay in the hospital but came back in the early morning. During that day, I suddenly became a very high fever again. A lot of doctors visited me and quietly talked at the side of my bed. I was upset because I did not understand what they said. My parents were upset too and looked at each other in despair. The next day, a nurse took me to another location in order to make an X-Ray and a CT scan. My mother stayed with me all the time: she knows that I am afraid to be left alone.

The results came in during that same day. A female doctor told my mother that both my heart and my liver were enlarged. I heard my mother cry and felt that something was not right. The next day, another doctor appeared who wanted to take another blood sample. He told us that the blood sample was important for diagnosis purposes.

After a stay of 10 days, I was discharged from the hospital. I was very happy to be able to sleep in my own bed again. It was much more quiet at home than in the hospital and I did not need to be connected to any monitor or infusion. However, the atmosphere was weird: my parents looked at me in despair and talked quietly when I was in the room.

After about 10 days, we had an appointment with two doctors, a man and a female. The male doctor examined me again. He then took a seat and said: "Your child has an incurable metabolic disease, Morbus Pompe. She will have only a few months left to live". I did not understand what that meant "incurable". I saw however that my mother started to cry. My father, who does not understand German, did not understand what was going on. He asked my mother. However, during the first minutes, she could not explain to him that I was mortally ill. In the end, he was as shocked as my mother was. My parents tried to understand the incomprehensible fact that even though I looked healthy, I was very ill. I started to feel very anxious because I had never seen my parents so desperate. When my parents and me calmed down a little bit, the doctor told us that a doctor in Bregenz applies a therapy in the treatment of Morbus Pompe. Shortly thereafter, we had an appointment with this doctor, Dr. Huemer. She gave us some hope by telling us that I could make it.

March 22, 2008, I became my first Enzyme Replacement Therapy with Myozyme. To decrease the risk of infection, I was transferred from a regular unit to a unit for premature children. The nurses were very nice: they played with me and let me caress the little babies. My condition did not get better though during the



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first few months. Instead, my condition worsened a little bit. Dr. Huerner was a little bit worried about that. She decided that it was necessary to proceed with the therapy on a weekly instead of a two-weekly basis.

Even though my mother became a separate room at the childrens unit, my mother stayed with me during the night. She slept in the chair next to my bed which caused her muscles to ache during the mornings. Unfortunately, my father could not stay as much as my mother did because he had to work. My stay in the hospital was very hard for my mother: I had to do a lot of physiotherapy and had to go to multiple appointments with Dr. Fritz, the cardiologist who I really like. After examining me, he was often very worried. This lasted until Christmas 2008 when he called my parents and told my mother that the Enzyme Replacement Therapy appeared to work in the end. My heart started to shrink! My mother was extremely happy about that results. She was again crying and thanked the doctor for telling her.

I could sit up alone at the end of January 2009. I was better able to keep up my head. At the end of the year I was even able to crawl around through my room. I have taken a lot of hurdles since the start of the therapy: first I became a Porth-a-Cath, then I became a dental treatment, and in 2011 my tendons were lengthened which implied that I had to wear plaster casts during 9 weeks! My feet are really nice now, however, I can still not use them to walk around. I need more strength and practice to do that. Nevertheless, I am sure I will be able to walk one day which would make me and my parents extremely happy. I really try hard to do the best, in particular when I have to get my infusion while I'd rather play around with my friends.

I become the Enzyme Replacement Therapy at the practice of our general practitioner, Dr. Maier since the end of 2009. My heart is doing quite well since then. The doctors are very happy with the results. My cardiologist said: "Nisa, you're writing history". Also doctor Huerner never gave up. She was the one who gave my parents new hope and learnt us to live with this disease. I am really proud of my parents who never gave up. And when they look at me with this sad view in their eyes, I just throw them an enchanting smile which allows them to briefly forget to think about my unknown future.

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