

## Nico

Nico, our second son, was born in October 1997. During the first months of his life everything seemed normal – he learned to turn over and began to crawl. It was only shortly before his first birthday that we noticed that something looked different from what we were used to watching when his elder brother grew up. Nico did not learn to stand, although he tried very hard to push himself up while standing on his toes. A blood test showed that Nico had an elevated CK-level. We were told that this meant that he had some form of muscular disease. At the neurological department of the University Children's hospital they performed a number of tests to exclude various forms of muscular disorders. A muscle biopsy finally led to the diagnosis of Pompe's Disease. The doctors told us that there was no treatment for Pompe's Disease and that very little was known about the progression of the disease. They also told us that Nico probably wouldn't reach his twentieth birthday and advised us to make the best out of the time we had. I can still feel the shock of this diagnosis as I am writing this report now.

As Nico didn't develop enough muscle strength to stand or walk, he got a wheelchair at the age of 2 ½ years. It gave him more freedom of movement and independence – and he enjoyed this very much!

When Nico was about three, we noticed that he began to develop contractures in his hips, knees and ankles which, in conjunction with the muscle weakness in his hips and legs, prevented him from standing. When he was four years old, an orthopaedist found out accidentally that he had a subluxation in both hips. His neurologist recommended that we "try" a tenotomy (extension) of some muscles and ligaments in knees and hips instead of a hip reconstruction. The idea was that this would allow him to straighten his knees and hips completely and maybe - with the use of a standing frame and the pressure on his hips - have a positive

effect on the development of his hips. Today, he still has a subluxation in his right hip, whereas the one in his left hip is almost gone.

Several weeks after the surgery, everyone was very happy and surprised to see that the muscles in his hips, legs and knees seemed to be getting a little stronger. About one year after the surgery Nico was able to walk short distances around the house - something nobody had expected. It may not seem much of an improvement, but it made a big difference for him. Outside the house or at school he now used an active wheelchair.



Nico, 6 years



At 8 years

In 2006 we started enzyme replacement therapy. During the first 6 months, Nico received his infusions in Mainz, one of the leading Pompe centers in Germany. He received his infusions at the same time as a friend, and the two boys used to play cards together, while we mothers played with them or enjoyed talking to each other. After 6 months we shifted to another hospital closer to home. Although we missed spending the long infusion hours with friends, it is more convenient and saves us a lot of time.

After a few months we noticed that Nico had a lot more energy throughout the day. The long hours at school were easier for him and he was more active and feeling stronger.

At present, Nico goes to an integrational school. His favourite subjects have always been PE, especially soccer (!), and maths. The school building is

completely wheelchair accessible, which he enjoys very much as it makes him independent.



Unfortunately, however, Nico developed a severe scoliosis which made him lose muscle function and lung capacity. The therapeutical recommendations ranged from spinal brace for the night and spinal brace 24/7 to scoliosis surgery. For almost two years we struggled with different types of spinal braces, which Nico didn't tolerate very well. He had problems with nausea, shortness of breath, pressure marks, etc. and as a consequence hated to wear the braces.

By the time Nico was turning 13, his scoliosis had deteriorated to the point where the doctors recommended surgery. He had lost his ability to walk because of the severity of the curvature of his spine which he could not compensate any more. He had surgery in January of this year (2011) and we went through some very tough weeks and months – both physically and emotionally. But now, 7 months later, Nico has more or less recovered. He still does not have all his energy back. But we are optimistic, since we know that recovery especially for Pompe patients is a slow process.

Birgit

Nico's mum