

Nico's story

Nico, our younger son, was born in 1997. During his first months everything seemed normal – he learned to turn over and he started to crawl. It was only shortly before his 1st birthday when we noticed that something looked different than in his elder brother. He did not learn to stand, although he tried very hard to push himself up while standing on his toes. When we told his pediatrician about our concerns, he told us that we should just give him time. But we could see that it was not a matter of time but that there was something wrong and that it was **not** an orthopaedic problem. First of all we went to see a physiotherapist. After a couple of weeks, she suggested to consult his pediatrician again to examine him for metabolic or neurological diseases. In the blood test, Nico had an elevated CK-level, which meant that he had some form of muscular disease. We went to the neurological department of the children's university hospital for a diagnosis. It took almost one year and quite a number of examinations until a muscle biopsy and a skin biopsy lead to the diagnosis. The doctors told us that our son had the late onset form of Pompe's Disease or Acid Maltase Deficiency, a progressive disease, that had no treatment and that he probably would reach his twentieth birthday. I can still feel the shock now as I write this report. We had to cope with the fact that our son's life would be very different from what every parent hopes. We were told that Pompe's Disease is a rare neuromuscular disease where a genetic defect prevents the body from breaking down the glycogen that is stored in the body cells. This leads to the muscle weakness (or an enlarged liver and heart). But the doctors could not give us much information about the progression of the disease so we did not know what we had to expect.

We were told, that the only thing we could do was physiotherapy, in order to slow down the weakening of his muscles. When Nico was 2 ½ years old, he got a wheelchair which gave him more independence – and he enjoyed this very much!

When Nico was about 3 years old we noticed that he began to develop contractures in his hips, knees and ankles which, in addition to the muscle weakness in his hips and legs, also prevented him from standing. When he was four years old, an orthopaedist found out accidentally that he had a subluxation in both hips. His hips had not developed properly as a consequence of the fact that he couldn't stand (the hip bones need the pressure when the children stand to develop properly) and because of the contractures. The orthopaedist proposed a complete reconstruction of his hip bones. As this is a severe and long surgery which would have meant many weeks in hospital, unable to move and more fragile hip bones, we were afraid that this would make him lose the little strength he had in his leg muscles. When we went to see a second orthopaedist, he suggested to tenotomize (extend) some muscles and ligaments in knees and hips. 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. We decided to "try" the tenotomy. Several weeks after the operation everybody was very surprised when the muscles in his hips, legs and knees seemed to get a little stronger and about 1 year after the operation he was able to walk short distances - something nobody had expected. It does not seem much but it made a very big difference for him. He always had felt the difference between himself and other children very much and was often sad or frustrated when he saw other children play soccer or run around when he couldn't.

Now, at the age of 8 years, he still has a subluxation in his right hip whereas it almost "disappeared" in the other one. He still can walk short distances of about 50 metres in the house and uses a wheelchair outside the house. But his muscular condition is gradually getting worse and he has developed a scoliosis over the last year. Three months ago, we went to the hospital to the sleep lab because we noticed that he has often nightmares that wake him up, that he comes home from school very tired and his lung function is deteriorating. The sleep test showed that he has hypoventilation and apnoeas during the night but that he does not need mechanical ventilation yet.

As to his nutrition, we try to make him eat as many proteins as possible which is not easy as he is a poor eater and loves especially pasta and bread. But he drinks a glass of milk with high-protein powder twice a day and we give him L-alanine 3-4 times a day.

Since last year he goes to a special school for physically handicapped children and he is really happy there. They have small classes (5 - 8 children) and they offer a variety of activities he really enjoys: they have computer classes, horse-riding, swimming classes, a circus project, a wheel-chair sports group etc. Once a week, he has physiotherapy in school and once a week he is seeing his local physiotherapist who knows him for 7 years now.

We very much hope that enzyme replacement therapy will come soon because this is the only chance to stop or slow down the progression of the disease.

Birgit

Nico's mother



Nico in wheelchair



Nico standing