

## Our daughter Benthe

### Benthe, November 2011, The Netherlands

Benthe was born in Hengelo hospital on 2<sup>nd</sup> November 2004. On first appearance everything seemed normal. She was allowed to come home with us. The next morning Benthe looked blue-grey and we immediately went to the hospital. She needed oxygen, her saturation level was around 50. She couldn't drink by herself and so was fed via a tube. An echo was made. Her heart looked a little enlarged but it was not alarming. During her birth Benthe had swallowed amniotic fluid so people thought it was an infection. Even after a course of antibiotics she didn't improve. The oxygen support remained necessary.



*Benthe in the incubator at November 3 2004.*

On Sunday 7<sup>th</sup> November 2004 the paediatrician decided to take another echo. Her heart was much bigger than that of a healthy baby, The paediatrician in Hengelo was not sure what was wrong and needed to discuss her case with a paediatric cardiologist at the Academic Hospital Groningen.

The paediatric cardiologist thought it necessary that Benthe should go to Groningen for an examination. She was then flown by helicopter to Groningen. That evening we were told that Benthe's heart muscle was much too big. This could be the result of different things. All sorts of tests were performed. The chance of survival was slim, not more than 2 to 3 weeks.



*Benthe receives her 1st infusion with the enzyme human alpha-glucosidase in Rotterdam at November 18 2004. Exact 16 days after her birth.*

On 9<sup>th</sup> November 2004, when Benthe was just a week old the diagnosis was confirmed. Benthe had Pompe disease. As there were people in Sophia Children's hospital specializing in Pompe disease, Benthe was flown there on 10<sup>th</sup> November 2004. Again here all sorts of tests were performed. The next day we had an appointment with Dr. van der Ploeg. She informed us that there was a therapy available for Pompe disease.

On 18<sup>th</sup> November 2004, Benthe received her first infusion and after a number of treatments we noticed that she was becoming stronger/sturdier. Her heart muscle became smaller and her ventilation support could be slowly decreased with the result that on 17<sup>th</sup> December 2004 she no longer required artificial ventilation. Slowly but surely her feeding improved. On the 19<sup>th</sup> of April 2005 the feeding tube was no longer necessary and Benthe could drink and empty her own bottle.

If we look back to how she was shortly after her birth to how she is now, we can really appreciate how sick she was. She was a "floppy baby" with very rapid breathing.



*Benthe pushes herself up.*

At the age of one year, she developed as any healthy baby would. She could roll over. She crawled, tried to sit down and a few times she could stand on her own. She did what any normal baby does. With 16 months Benthe showed us that she could walk normally as well. For us it was a clear sign that Benthe was doing well and that the enzyme replacement therapy was working.



*Benthe tries to walk.*



*Benthe on her rocking horse*

Benthe grew up and made the step from baby to toddler. She stood steady on her legs, could drive her toy car and could even run. She loved to swim and to play in the play garden where she enjoyed the slide the most.

At the age of two years and six months Benthe went to Kindergarten. To us it was a moment where we could compare Benthe with her peers. With bouncing and climbing stairs Benthe had just a little bit more difficulty than the other children. She just was less fast. But that really was the only thing. We also heard positive remarks from her teacher about her development.

When Benthe reached the age of 4 years, she was allowed to go to primary school. She felt she was a big girl. With her small bag pack and her princess pink breadbox and drinking cup in it, she walked to school. She enjoyed her class. After summer holiday she started in group one. They received 'voluntary' and 'mandatory' tasks. Her

huge persistence made that most often her 'mandatory' tasks were done first.

Once every fortnight Benthe couldn't go to school, while she had to receive her infusion at the Sophia Children hospital in Rotterdam. Benthe even went to school in the hospital for a couple of months when she received her infusion. In the morning from 9.00 to 11.00 she then sat together with the teacher and other children, busy drawing and doing other activities.



*Benthe enjoys the glide.*

In October 2009 it finally happened... Benthe was able to receive her infusion at home. This not only was more easy for the family, but also for Benthe. She now only has to miss two hours from school. Once every fortnight a specialised nurse of a home care company comes to our home and she takes care of Benthe her infusion.

In 2009, when Benthe just turned 5 years, she started to take swimming lessons. Soon we noticed that she really enjoyed it. The water wings were replaced by cork arm floats and within months they were unnecessary. When the corks around her waist were removed she had to stay above water on her own.

This turned out not to be a problem at all and in April 2011 Benthe received her swimming certificate A. However she thought she wasn't ready yet and wanted like all other children to get the swimming certificate B. In October 2011 she received that one too. She gained much self-confidence now she got both swimming certificates. She now tells everyone that she can swim independently and without supervision in the swimming pool.

Benthe is now 7 years and now learns to read, write and calculate. Since her home infusions went very well we requested if it was possible for her to receive her infusion at school. For her development it is important that she is able to be at school as much as possible among her classmates and doesn't need to miss lessons. Since a few weeks Benthe receives her infusions at school in her classroom and now she doesn't need to miss out on anything.



*Benthe plays during her infusion in the hospital.*



*Benthe in the swimming pool.*

**Jeroen and Mirjam, parents of Benthe, Nederland**