Arno – 13 years old – Diagnosis: Morbus Pompe -


Arno, aged 13

Arno is our only child. He was born, two weeks early, on the 7th of September 1992, after a normal pregnancy and a short and normal delivery.

When he was born, Arno weighted 2950 gram and his length was 50 cm. Even though he was relatively small, he appeared to be healthy and strong. He slept a lot during the first weeks of his life. His sucking reflex appeared to be weak though resulting in an under stimulation of the milk production yielding breast feeding problematic. Nevertheless, he was very well capable to cry out loud when he was hungry. Breastfeeding was therefore supplemented with bottle feeding. Only after Arno was three months old, his sucking reflex was strong enough to breastfeed him without any additional bottle feeding during the next three months. Arno's development, including his motor development, was normal. Nevertheless, he appeared to be relatively relaxed in the sense that he did not show much initiative to move around and his muscle tone appeared to be weak.

When Arno was half a year old, he still failed to show much initiative to move. For example, he did not lift his legs towards a ball held above him, he did not seem to want to bounce up and down while standing on his feet, as babies normally do. He also failed to keep an object in his hands when pulled away. The paediatrician prescribed physiotherapy (after Bobath & Vojta). As parents we were taught several exercises Arno had to do once or twice on a daily basis. Because Arno, who usually appeared to be a very happy baby who barely cried, showed severe protest during his exercises, we decided to quit the Bobath Therapy after a brief period of time.

Arno's further development was supported by a moderate physiotherapy only. Even though he showed a slow motor development, he reached the normal milestones: When he was 10 months old he could sit, when he was 14 months old he could crawl and pull himself up. When he was 16 months old he could walk when supported, and when he was 18 months old he could walk freely.

During this time he became a febrile spasm *(*fever convulsion) due to bronchitis. After two days and a fever of 41 °C he was hospitalised in a child hospital. During his stay, his liver- and muscle-values (GOT, GPT, LDH und CK values) were determined and showed up to be too high (between 300 and 500 U/l). After one week, he was dismissed from the hospital. However, it was recommended to monitor the blood-values during the time to follow.

Half a year later, the blood values were still not normalized. We were referred to a university hospital to have a muscle biopsy taken. The diagnosis followed shortly thereafter: Arno was diagnosed with „Glycogen Storage Disorder Type II b". Since his diagnosis, he is physically screened in the children hospital on a yearly basis.

Arno was diagnosed when he was two years old. At that point in time he weighted 12 kg, and his length was 90 cm. His fine motor development as well as his coordination and language development were completely normal. In addition, he was physically healthy in the sense that he was not more often sick than other children of his age were. However, his gross motor development was severely delayed. For example, he had substantial difficulties to walk stairs or to bend through his knees and get up again. These difficulties became less when he grew

older. Nevertheless, he always appeared to have less strength reserves than normal children have. In addition, he always tends to spare his strengths in his activities.

During the years following, he used to have a severe bronchitis during spring time. In 1997 (when he was 4,5 years old) he was hospitalized again during 14 days because of a severe pneumonia. When he arrived in the hospital he was unable to lift his head. When he was dismissed from the hospital he was not able to climb the stairs anymore. He was very weak at that time and needed about 3 months to fully recover. Since that time, I gets a yearly anti-fever injection, and does not get sick as often anymore.

Winter 1999, Arno has Winter service duty

At the end of the summer in 2002, Arno, who was almost 10 years old, became his first acute Dyspnoe-attack. His condition improved fast when he received Betamimeticum inhalations. His lung capacity was however strongly reduced (FVC-value of 52%). During the next three months his lung capacity increased to about 80%. One year later he was again hospitalized during two days because of a second Dyspnoe (again caused by an obstructive bronchitis). He was diagnosed with asthmatic bronchitis caused by a dust mite allergy. Since that time he is durably receiving Betamimetic and Corticosteroids inhalations. His vital lung function has since been constant at about 80%. His lung function is checked every half a year.

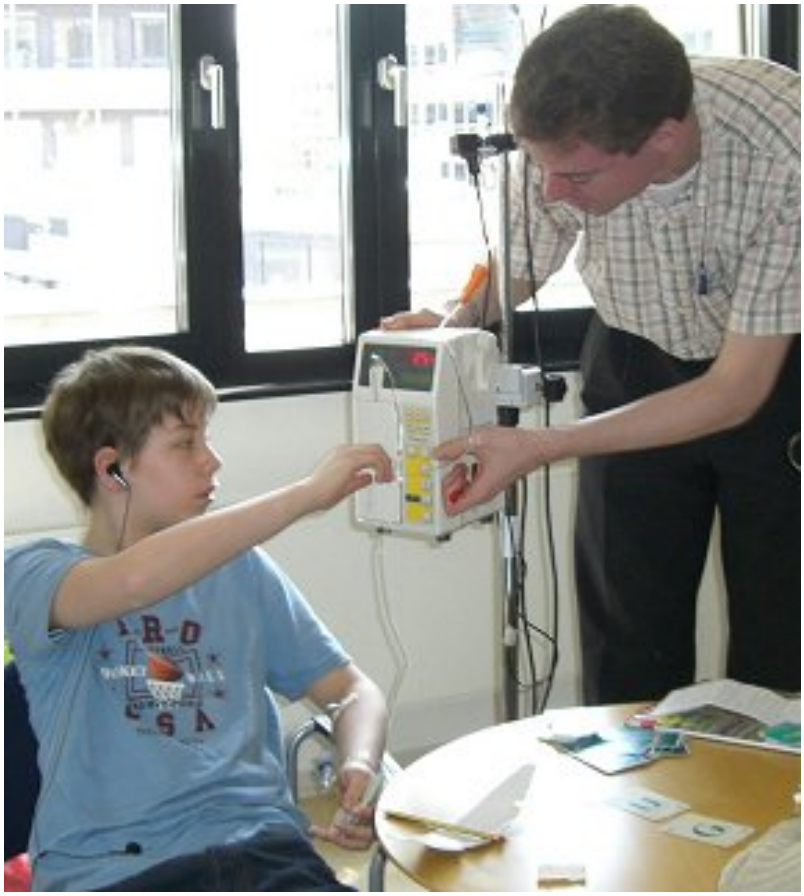
Arno always preferred a protein-rich diet. Since he has been diagnosed with M. Pompe he also receives protein supplements and L-Alanine. Like other Pompe patients, he has also been suffering from unspecific diarrhea, however, these problems have been largely alleviated by homeopathic medicines.

Summer 2005, holiday in Cornwell

Currently Arno weights about 53 kg and his length is 172 cm. His length is above that of the average 13-years old boy. Nevertheless, his muscle strength is much below normal. His muscles in his hips, tights, and shoulders are most severely affected. He is only able to ride a bike on even ground. He can only walk fast over about 1 to 2 km. He is not able to run. It takes him a lot of energy to climb stairs, and change his direction of movement. He is experiencing increasingly more difficulty to bend through his knees and stand up again. During standing up he uses many compensatory movements to remain in balance. He lifts his legs with his arms when he gets into a car. He supports one arm with the other arm when he is holding a cup. Arno has learned to cope with the effects of his disease. Even though he still perceives his disease as a “normal” part of his life, he is very conscious about the limitations imposed by his disease on his daily activities. Currently, he still takes part in various activities at school

and at home. For example he participates in the regular sport classes, he takes part in special events, like walking tours and parties. He has many friends in school and in the neighbourhood. He is very well integrated in his peer group. During sports, however, it becomes increasingly clear that he is unable to keep up with the others. As a result he only takes part to a limited extend. In addition to the regular sport classes, he is doing physiotherapy to prevent his Achilles tendons to shorten and to prevent his muscles from fast deterioration. Apart from this, he is physically not very active. He likes to lie down on his bed to read a book or play with his Play station.

Up till now Arno has been hoping that M. Pompe may some day be treatable. We are therefore very happy that he is one of the first to receive Myozyme since it has been approved by the authorities. We thank all those, who enabled the development of the Enzyme Replacement Therapy for M. Pompe patients, and hope that with Myozyme Arno may have an almost normal future.



3th of May 2006, Arno starts his first infusion