

Marius' Story

My son Marius was born in Amsterdam on February 19, 1999. I am a single mother and Marius is my only child. Marius and I live in Amsterdam. During his first two years of life, Marius appeared to be a healthy child, until one morning in October 2001, when he woke up and he suddenly could not walk anymore. As a mother, I was shocked and took him immediately to the hospital to have a neurologist have a look at him. The neurologist could not find any clear reason for my son not being able to walk and decided to take a blood sample for further investigation. After a couple of hours, Marius could walk again. However, the blood sample did show some potentially alarming deviations in the sense that his CK (Creatine Phosphokinase) and several liver values were abnormally high. Even though, Marius recovered completely from his episode of immobility, his CK and liver values remained high during the months following the first measurement. To become more clearness about the possible cause of the elevated CK values, in March 2002 a muscle biopsy was taken. The proper diagnosis on the basis of the muscle biopsy took a very long time. However, finally, in June 2002 the diagnosis was a fact: Marius suffered from an acid-maltase deficiency: his muscle tissues already showed signs of serious degeneration in the sense that the tissue showed vacuoles and glycogen storage. A skin-tissue sample confirmed the diagnosis: Marius had an acid maltase deficiency, Pompe's disease.

The diagnosis was devastating for me (Marius did not really understand what was going on: he was only three years old!). Even though Marius did not show any signs of any illness (except maybe some small weakness in his proximal muscles), he actually suffered from a very serious disease that, untreated, would lead to progressive immobility, respiration difficulties, and an early death.

Like every parent, the thing I wanted most was the happiness of my child. By understanding the implications of Marius' disease, I felt that happiness would be far out of reach for my son. I realized he would have a very hard life. To have a serious disease is one thing you may cope with, however, to have a serious *progressive* disease seems to me something impossible to live with.

Almost immediately after hearing the terrible diagnosis, I learned from internet that even though Pompe's disease was officially still an incurable disease, the results of recent studies had shown very very promising results with a small group of patients receiving treatment in the form of an Enzyme Replacement Therapy (ERT). A group of researchers in Rotterdam (most importantly, dr. Ans van der Ploeg and dr. Arnold Reuser) had in collaboration with the pharmaceutical industry (Pharming) developed a new therapy for Pompe's disease that really seemed to work!

To provide the best possible opportunities for my son, I made an appointment with dr. Ans van der Ploeg in Rotterdam. She extensively examined Marius and decided to keep track of his further development on a regular basis. Moreover, she provided me with answers to my many questions concerning the disease of my son. The regular visits to Rotterdam were reassuring, however, Marius was growing older and with the months passing by it became increasingly clear that he was growing

weaker and weaker. He started to have difficulties to run, difficulties to climb stairs, and problems to stand up from a lying position. In addition, he was continuously tired, a clear signal that his muscles were getting increasingly weaker.

During that period, it became clear to me that if Marius was to have any chance to lead a relatively normal life, he should receive the Enzyme Replacement Therapy as soon as possible. Muscle fibers that are destroyed cannot regenerate anymore. I realized that any deterioration that Marius showed might be irreversible. It was a terrible period of time. I knew of the existence of a therapy for Pompe's disease, a therapy that could prevent my son from becoming increasingly more disabled in his mobility. At the same time, there seemed to be nothing I could do to have my son receiving this treatment because the treatment was still in an experimental phase. It was just unbearable.

However, things changed very rapidly on one evening in December 2004 when I received a telephone call from dr. Ans van der Ploeg. In that call, she told me that possibly Marius could participate in a small study on the effectiveness and safety of Myozyme, the trade-name of the ERT as developed by Genzyme (the pharmaceutical company that took over the production of the enzyme for Pompe's disease). The study was planned to start in February 2005. She asked me whether we would agree to participate in such a study. I was delighted! Finally, after a time period in which I felt completely helpless, Marius' and my life started to have some perspective.

Marius is now (September 2005) receiving Myozyme on a two-weekly basis. The study in which he participates started on February 2, 2005. Participation in the study implies that we have to go to the Sophia Children Hospital (Rotterdam) every second week on Wednesday. During such a day, Marius is receiving Myozyme via an infusion (during approximately 4 hours). The frequent hospital stays in combination with the uncertainty about how Marius is going to react to the medicine are sometimes difficult to deal with. However, these unpleasant consequences are minor compared to the fact that Marius is doing extremely well since he is receiving this medicine. His motor capabilities are really better than they were at the beginning of the therapy. Marius is able to run again. He can now easily stand up from a lying position and is, like most 6-year old children not continuously tired anymore. Besides, Marius takes everything relatively easy. He even sort of likes it to go to the hospital to receive his infusion! These developments in combination with the realization that Marius might even be able to lead a relatively normal life, makes my life so much easier than it was before. Nevertheless, I am still worried: worried because Marius' life is now dependent on the timely administration of Myozyme, worried because I don't know what the long-term effects of the administration of Myozyme are, and worried because I am not sure whether future supplies of the medicine will and can be guaranteed. I realize though, that a worryless existence is not a real possibility anymore when you have to deal with a disease as devastating as Pompe's disease. Provided the fact that Marius suffers from this disease, currently, things seem to work out in the best possible way.

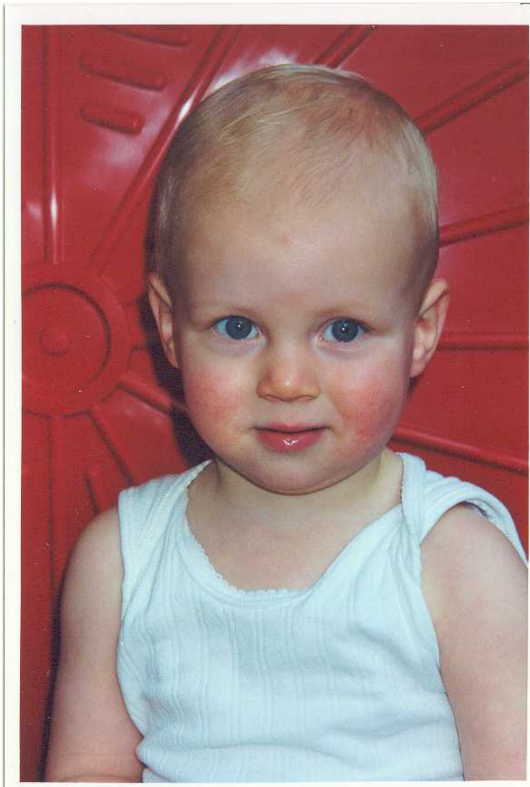
Mieke
Amsterdam, 27-9-2005



Marius and me (Mai 1999)



Marius (6 months old)



Marius (18 months old)



Marius (2,5 years old)



Marius (3,5 years old)



Marius (almost 5 years old)



Marius (almost 6 years old receiving his first infusion of Myozyme)



Marius (6 years old)



Marius (6,5 years old)