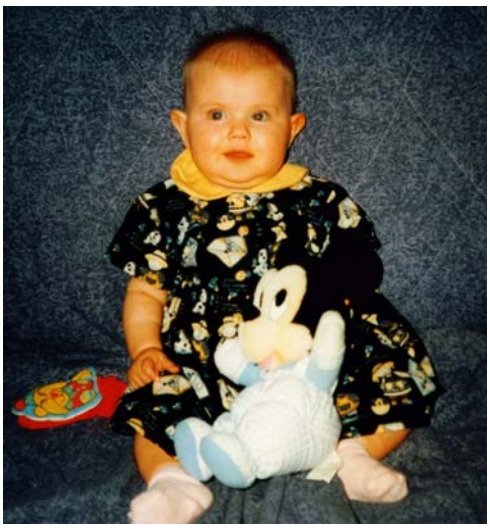
Karen and Pompe’s disease

Before the diagnosis



Karen, June 1996

On the 3rd March 1996 our little girl was born. Karen was bottle fed. She was a greedy baby and was always hungry. Because she vomited after every feed, she was changed over to Soya milk. But there was no change and it was decided to add Gaviscon to her bottles. This powder mixes with the milk and prevents or minimizes the vomiting.
However, the day before we were due to go home, Karen was rushed, by ambulance, to the University Hospital in Gasthuisberg, where she was diagnosed to have 'Gaviscon-bezoar'. The powder had an unknown side-effect. It dissolved completely in the milk, but when in the stomach it formed a solid mass, so it was quite understandable that she vomited after feeding. During an endoscopy this mass would be shattered and then a suitable food, which she could tolerate, would have to be found. She spent five days in the Neo-Natal Unit and looked like a giant among all the little premature babies. Then finally, on the 13th March, we were allowed to take her home. I can still hear my mother say: "She had a false start, but everything is in order now". We had no idea of what was yet to come.

Karen, April 1997

Karen's Pompe's Story started in December '96. She was then 9 months old.
My mother, who was our daily babysitter, thought that we should talk to a Paediatrician, because she did not think it was normal that a nine-months old baby should not want to try and stand on her feet and would just hang like a rag-doll when you held her up, holding her under the arms.
Karen became hysterical if we laid her on her tummy and, from lying on her back, she could not roll onto her tummy and vice versa.
To us she just seemed a bit lazy, so we just wanted to take things easy. After all, she could sit up! Karen is our first, and still only, child, so maybe we were not so experienced in what was 'normal'.
So we went to see a Paediatrician and she wanted to do some tests: EEG, CT-scan and blood tests. The first two tests were OK, but the blood tests showed an abnormally high CK level which, according to the Paediatrician, could point to a muscle disease.
We were referred to another specialist. In the meantime a start was also made with Physiotherapy twice a week. Everything had to be learned.

The diagnosis and then .....

Two weeks later we went to the University Hospital in Leuven for the first time, to Child-Neurology. More blood tests. In February '97 there was a muscle biopsy, and the final verdict came six weeks after the biopsy. Karen has an incurable metabolic illness, which has serious consequences for the large muscle groups: Glycogenosis Type ll, probably the juvenile form. Another name for this metabolic illness is Pompe's Disease.

In the meantime we didn't sit still and we searched for an illness that would fit Karen's symptoms - although we did read a lot about GSD types and thought that Type ll could well be the answer. We found some symptoms in our little girl, only in a much milder form than the descriptions in the articles which we had collected. And when we were finally told that our suspicions were true ..... at that moment our whole world just fell apart. There were no answers to the many questions we asked. They had to follow Karen's illness for a while to evaluate her first. There was no treatment or therapy. The message was: "There are research-teams who are working on developing a therapy, but that is still some years away. We just have to make sure that Karen stays as healthy as possible".

So we continued with the Physiotherapy. And it helped! Karen improved some and in view of these improvements we could, according to the doctor, cautiously expect her to start walking, although a little later than a healthy child would, but probably somewhere between the age of 2 and 3 years. We kept ourselves strong in the knowledge that, mentally, there was nothing amiss with our baby. She was, in fact, a little ahead of her age group. She could already say several words.

Karen was 15 months old when she began to crawl.

And then our most unforgettable moment: at 18 months, all by herself, she took her first steps. We will never forget the expression on the small face. It was just as if a whole new world had opened up for her. She smiled and couldn't get enough of it. Medically seen, 18 months is just inside the acceptable limits for a healthy child.

"Karen is doing exceptionally well", we were told. The illness seems to limit itself to the skeletal muscle, as they say so nicely, and is concentrated on the pelvis and below.
In the beginning of 1998 a slight enlargement of the liver was found, but not enough to cause us real worries. In 1998 she also had her first saturation test. Her breathing was not yet involved, neither was her heart.

Since 1998 Karen follows a protein-rich diet. At first this was supplemented with L-Carnitine, but in 2001 the L-Carnitine was replaced with L-Alanine. The protein-rich diet (starting with 20% protein and aiming for 25%) was quite easy. Karen seemed to instinctively reach for things that are good for her, and it is not so difficult to have to follow a diet like that, for a child who likes milk products and meat. We supplemented her normal food with a protein preparation. It is only in the last year that we are having some difficulties reaching the 25%.

Karen had a lot of problems with abdominal cramps and constipation, but these problems seemed to become less as we reduced the dose of protein preparation.
In comparison with other patients Karen has, up until now, experienced only a few problems from this disease. And as she got older things seemed to improve, it almost seemed that she was growing out of it .....

Maybe this was because we always made very sure that Karen followed her diet, and that she really did the exercises which the Physiotherapist gave her to do. This was not always easy, as there were times when Karen just did not want to exercise. There were times when the tears flowed and this then led to yet another discussion.

The Physiotherapy treatments became less as Karen improved. At the beginning it was twice a week, then once a week, then once every two weeks and now once every three weeks. However, exercise remains important because when there is a period of time that she does not exercise, we notice very quickly that she loses not only muscle strength, but also condition. Karen's biggest problems are loss of muscle weakness and problems with the balance. With gymnastics and swimming lessons she is not as good as other girls in her age group. Some exercises in gymnastics - especially if they require more muscle strength - she just cannot do. But that does not seem to worry her a great deal, because the teachers then ask her to do other things that she can do, and so Karen does not feel left out. Karen is also a member of a local youth group. Every Sunday afternoon she joins in the games and activities with other children of her age group. We think it's important that she can join her girlfriends in all sorts of activities.

Other recurring problems are the respiratory infections and frequent colds. During the whole school year we saw our local doctor every month or so for a cold which, without medication, would become another serious respiratory infection. During the pre-school period we would sometimes keep her home as a preventive measure, if we knew that there were classmates who had colds or infections. But since she has started school we cannot do that any more, as she would lose too much learning time.

When Karen was very small she already realised that she was different to other children. She was also very careful, or maybe unsure, when she was with other children; frightened of being pushed over, because of her poor balance. But she was just like other children who loved the play park and the slippery dip. It was a tremendous effort for her to climb to the top of the slippery dip, but she would keep on climbing back up, until she was just about ready to drop.

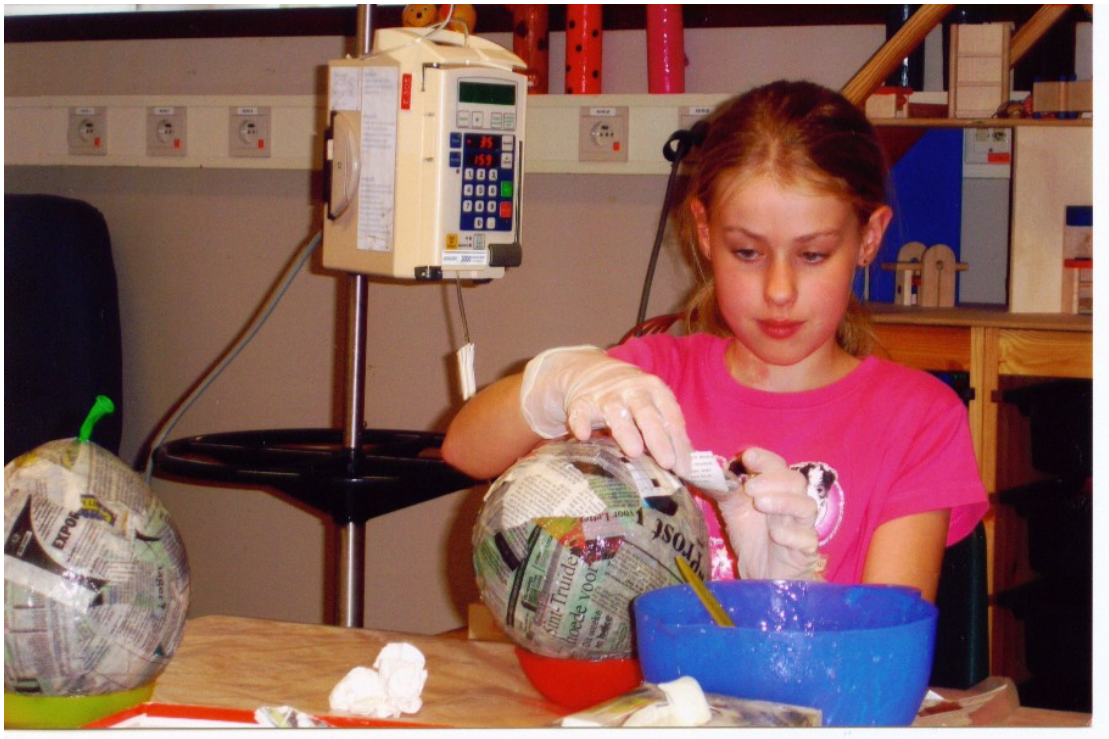
Karen, July 2001

Karen, 6 years old

Karen, 8 years old

Taking part in a Clinical Trial

Since February 2005 Karen has been receiving Enzyme Replacement Therapy. She is taking part in a Clinical Trial together with a number of other children. It took a lot of effort to convince Karen to take part in this trial. At first it seemed that she just did not want to do this, or was it because now was the first time that she was really confronted with the possible consequences of this disease?
Although the time she has been receiving treatment is relatively short to talk about spectacular improvements, we do notice that the therapy is having a positive effect. Also Karen notices the difference. She does not tire as quick, and during gymnastics or activities within the youth group she finds that she does not have to sit on the sidelines as often, to have a rest. On the contrary, now there are times when she comes home from an afternoon of full-on play with the youth group, and still wants to do some rope-skipping or bike-riding. For parents of a healthy child of this age, this is probably totally normal. However, we were used to Karen coming home dead-tired after such an afternoon. Not only has her condition improved, but we also notice improvement in her muscle strength. Also, Karen is not sick as often as she used to be.



Karen (9.5 years old) being creative while she receives enzyme replacement therapy

Before the therapy we would visit the doctor at least once a month for yet another cold which, without help, would progress to yet another stubborn respiratory infection. A few months ago we were there again, however now it was more than six months ago when she had the previous cold. She seems much less susceptible for all sorts of infections which do the rounds in her environment (school, family .....). The problems with the abdomen recur every now and then, but not as often as they did before.

And we, as parents

We also had to learn to live with our child being 'different'. Despite all the stresses, the anger. the pain and the sorrow, I feel that our relationship has grown stronger. With Karen's progress, the pain has also worn off a little. Sometimes we still think 'Why Karen, Why us?' But then, when we look at this busy bee, always cheerful, very active, with enormous perseverance ..... then we can sometimes forget that she will always be our worry-child.

It's not always easy to organise everything to fit in with our work. But thankfully, our employers have always shown us a great deal of understanding. Furthermore, we can always count on our parents.

Karen's future

With the therapy Karen's future has changed enormously. She will be spared from the fatal consequences of her illness. With therapy she can live a relatively normal life, just like the other children in her class. But of course it remains a big task ..... for the rest of your life, every two weeks an infusion, to ensure that your condition does not deteriorate. And although, for the outside world, it seems as if Karen handles all this with apparent ease, we know that she is not always so happy about it. More than once we have heard from people, who didn't know about Karen's illness, "But you can't see that there is anything wrong with Karen ....." We also do not find it necessary to tell everyone that Karen is a Pompe patient. We and Karen do not need the pity that is so often shown when you explain just what the actual illness is. We do want understanding for Karen's situation, and it is important to us to make sure that the adults who have an influence on Karen's life (teachers, youth leaders, eventual employers…) are well informed about her disease. Because we are convinced that someone like Karen, given the necessary understanding, can live a 'normal' life.

Dirk and Hilde,
Karen's parents.

2006