A few milligrams happiness for Miguel.

Miguel was born in September 1994 and everything seemed to be normal. On my birthday in December he began to walk. For a father, there is no better present. What I didn't know was that in is case the ability to walk would end so soon. When he was 3 ½ years old following a muscle biopsy, we had the definite diagnosis. Miguel had Pompe disease.

We did not receive adequate and insufficient information to prepare us for the rapid progression of the disease, which still came as a surprise. Eventually a respiratory infection resulted in the necessity of a tracheostomy and breathing apparatus practically 24 hours a day.

From this moment a new life began for all 3 of us (for his mother, Miguel and me). Following months of hospitalization. Miguel could come home for Christmas. The thought of him walking was exactly 3 years ago. It was difficult to accept such a drastic change, but then came one of the first breakthroughs, namely there was a patient association that helped patients with information about the disease and that helped other families and patients come through this dramatic situation. It has been one of the best decisions in our lives. The continuing concern for Miguel was linked also to the concern of other patients with glycogen storage disease. This double function has dominated our daily life and that of our zoon, for seven years.

He frequently asks after other patients with a alpha-glucosidase deficiency . We have tried to inform others, and we have learned so much from many people that have seen the disease but have not been affected.

How much Miguel worsened was our main obsession and to know if there was a treatment, an also through the help of Dr. Loirent Acosta, from whom we had heard about a reliable initiative taken by the company Pharming.

For a long tine there was a photo on the main page of the website from the Dutch company where you could read "acid –alfaglucosidase". I logged on many times every day just to see this.

In the summer of 1999 I could participate in the International Pompe Conference which was held by the International Pompe Association in Naarden, the Netherlands. I felt a positive sensation that the expectations were positive and finally met people that had the same sickness as my son. In a certain sense I felt as they became a part of my family. I also felt that it was worth the effort to continue with the struggle to find a treatment. The only thing that held me back was the language! I didn't hesitate for a moment and started English lessons the same summer.

Exactly a year later (July 2000) while reading the Murca newspaper (the Spanish province where we lived at the time), my eyes automatically were drawn to the title: "Research into the effectiveness of rabbit's milk against muscle diseases". The article

was about Pompe disease and it was very important on a higher level, that a metaphorical indication that a treatment was coming closer. Many family members and friends had also read it and called us to offer congratulations and solidarity. The article has been framed.

Between shock, travel, telephone calls and so on. Miguel stayed focused on his study but his health deteriorated with increasing speed. After much adversity came the moment in November 2003, when our son received his first infusion of Myozyme®, the product that finally being produced by the company Genzyme.

From that day to this and with the existing parameters of the assess to the enzyme, we can propose that we at least reached a few degrees of normality. Miguel can now occasionally come off the breathing apparatus for a few hours at a time, and as given us a freedom and quality of life. Something 2 years ago would not have been possible.

We can go to the beach and walk without haste. We go to the museum, the cinema and the library. We can make visits and receive visitors and go out for dinner with out having to take the respirator with us. A few months ago Miguel went to school by himself where he was registered, (although where he couldn't follow class), to finally meet the other students. It was an unforgettable day for us all.

And if that wasn't enough, his mother and I have developed to become experts in the manoeuvering a wheelchair through a badly developed city for people with a handicap.

Together with the noticeable positive changes especially with his breathing, Myozyme had unmistakably helped him to gain weight and to improve his muscle tone (but in the range he remained clearly weak).

I would like to end by saying that Miguel will shortly be able to describe himself (in English) about what he feels, how he lives with his disease and what it means to him to receive this hope-giving therapy.

Antonio