"My Life Story"
by Ricardo

My name is Ricardo, I am 36 years old, and I live in Buenos Aires, Argentina. At 16 years of age I was diagnosed with "Pompe's disease". This is a brief overview of my life, how it has proceeded, and how this rare disease has affected me, from the very beginning when the first symptoms of weakness appeared until the present day. I want to emphasise how difficult it was for me and my family to endure this disease and to live with it, not only because of the physical consequences, with all that they involve, but also as a result of unfamiliarity and the little information that we had at our disposal (at least in the first instance). For everything that is done to provide more familiarity with this disease is a good thing.

(At 12 years of age)

My story begins in childhood. I was always very thin and short for my age, and my growth did not follow a trend similar to that of my friends from school. In spite of that I had no symptoms of weakness until my 11/12th year, and I developed and played football like any other normal child.
But there was something that was not good. Not a single doctor knew what was the matter with me, and they found nothing abnormal. At the age of 14/15 I was 1.40 m tall and weighed 27 kg.
I always had little appetite, and I felt very tired. At that time, however, I was diagnosed with "Anorexia nervosa". After an intensive treatment I succeeded in putting on weight, and I weighed 33 kg. From that moment on, my condition deteriorated rapidly. At night I did not sleep well and woke up constantly. In addition I did not breathe well, I had a very rapid pulse, and I was much too tired.
Later I began to feel weak, especially in the legs. I could no longer climb stairs or run. I had to drop out of physical activities at school.
The situation worsened to such an extent that I had to be hospitalised at age 16. That same night artificial respiration had to be applied. My diaphragm was functioning only 25 to 30%. Five days later a tracheostomy was performed, and a biopsy of the muscles was taken and sent to the United States for analysis. The diagnosis read: acid maltase deficiency, also known as "Pompe's disease", a rare progressive genetic disease. It is caused by the absence of an enzyme called "acid alpha-glucosidase". This enzyme is responsible for the breaking down of glycogen, a complex molecule formed by many units of glucose, in cellular compartments that are called lysosomes. In this disease this process is absent, and the accumulation of glycogen affects the function of the muscles. After three months of being hospitalised, I recovered from the crisis to the extent that I could go home again. I only used the breathing apparatus at night.
Shortly thereafter I was found to have scoliosis, which drastically worsened over the years. When I was 20 I began to use a wheelchair because I could no longer stand normally on my legs. I usually needed it at various times in the day.
During the last five years my situation worsened considerably, and as a result I need respiration practically 24 hours a day, even at meals.

As I have already mentioned, it was and is very difficult for me to endure this disease. I have had to do without many things, including things which everyone dreams of in life and has as a goal, such as studying at university, working, and forming a family of one's own.
I no longer had very many friends, and I saw them less and less. For the last few years, because of the worsening of my situation, I could not go on holiday, and going outside for a stroll became too tiring. So I became increasingly isolated, felt lonely, and had no information about was now the matter with me. I lost heart. After I turned 30 I fell into a deep depressive crisis and had to resort to psychological help, which I had had to do much earlier. The therapy helped enormously. There was, however, something that would help me even more to overcome the depression.

At that time I became acquainted with the Internet and thereby a gateway to the world. The first thing that I did was to search for information about my disease, and my life began to change again. I met people from other countries with the same problem and formed a nice friendship with them. I also became acquainted with organisations that dealt with this disease. Thus I was able to get information, and I no longer felt so lonely. Then I also heard that work was being done to develop a treatment of Pompe's disease and that trials were being performed on children with remarkable results. That all caused me to have hope again and to continue fighting with more vigor and perseverance than ever.

(At 34 years of age)

A few months ago I began with the enzyme-replacement therapy (ERT) that was developed by Genzyme. I still remember the day of the first infusion and how emotional it was for me when the Myozyme began to flow into me. At that moment I knew that I had been given a new chance in my life and that certain functions would surely be improved with the treatment. I am very hopeful that it will be so.

I want to thank APELRA (Association for Lysosomal Diseases of the Republic of Argentina), whose president is Mr. Carlos Gandolfo, for its great help in obtaining the treatment for me. Thanks also to FESEN (Foundation for the Study of Neurometabolic Diseases), which at the time was chaired by Dr. Chamoles, who also helped us tremendously. Special thanks also to the Genzyme Laboratory for treating me and giving me a new chance in my life. A very sincere thanks to all!

Ricardo