

Pompe Disease

Pamela, September 2011

I was born on July 19, 1986 with a very normal delivery, after a pregnancy without any problem. My first years have been those of a normal child, and although, I never had an appetite, my growth was normal and my motor development certainly did not lead to whatever happened after.

In the month of February 1990 I was admitted to the hospital of Figline Valdarno for a bad bronchial pulmonary. During the same year happened 4 more episodes of pulmonary infections, less serious than the first therefore they were taken care at home.

The pediatrician, in agreement with my family, decided to investigate both at the level of admission and as outpatient in order to research the cause of the infections at the department of immunology of the Pediatric Clinic Terza-Pediatric Hospital Meyer of Florence, directed by Prof. Alberto Vierucci. It is thought about allergies because all the episodes of bronchial pulmonary infection were characterized by a sudden beginning. The tests show that as far as allergies everything is negative, but the transaminases were increased, therefore they think about the liver intoxicated by the medicines given to take care of the pulmonary infections. The doctors advised to keep the situation under control waiting for the situation to be stabilize. In the meantime I lead a normal life, going to the day care center, then to school. I practiced dancing and I was growing up normally, although once in a while the same problems happened.



Pamela in the Summer of 1998. Before the worsening of Pompe Disease

In 1995, because the situation was not changing, the doctors decided to let me take more deeper analysis, and then it became relevant an increase of the muscular enzyme creatine kinase (CPK). For this reason is born the suspicion of a muscular illness and I was sent to Hospital Meyer of Florence, section Neurometabolics and Neuromuscular for a consultation with Prof. Enrico Zammarchi. After two years of further controls and analysis, they decided to do a muscular biopsy, arriving to the definite diagnosis of Glycogens type 2 late-infantile.



July 1999. My 13th birthday in the hospital with friends. One of the first exits from the reanimation ward of the Meyer hospital.

The doctors told my family that the disease has a progressive course, but very slow and an American Pharmaceutical Company was already developing a therapy. In the mean time I was supposed to conduct a normal life, above all to practice a lot of soft physical activities so to keep the muscles active and elastic, and to follow a diet with very high proteins.

In the month of March 1999, because of a very acute respiratory infection, I was admitted to the Hospital Meyer, Reanimation Department in need of respiratory assistance. During this very long hospital stay,

5 months, my muscular situation became very bad. I could not breath by myself or to walk. With the help of the hospital and the ASO 10 of Florence, a system was activated so it could be done at home what was done in the hospital, using a portable breathing ventilator thanks to which I could go to school and to the physiotherapy sessions.



July 2004, celebrating my 18th birthday. I, while removing wrapping paper from presents and with my father and mother blowing out the candles 8 months after enzymatic therapy.

On June 8, 2003, because of pulmonary edema, I was admitted again in the Meyer Hospital for 15 days; at this time relative and friends started spreading my story using newspaper and leaflets; they organized a torchlight procession to which many, many people for the area and from nearby towns participated.

All this noise arrived to the authorities who were able to reach the pharmaceutical company to get the medicine. On November 6, 2003 the first infusion was given to me. From here the walk of hope begins for all my family to be able to go back to life as before.

In these two years I had not had any more pulmonary infections, the respiratory acts have diminished (they have gone from 60-50 to the minimum of 20), weight and appetite have increased. Although there have been improvement I still am unable to detach myself from the breathing machine and I do not know if it is a psychological block or physical problems. Anyhow I am doing breathing exercises and lately I can put my feet on the standing that helps me strengthen the muscles of my legs, chest, neck and arms.



My graduation party with my family and friends at July 21, 2010.

Since I started the therapy I gained strength and the pulmonary infections have lessened, almost gone. In the winter I get normal colds which no longer lead to infections and I recover much quicker. I have managed to decrease the use of the respirator, but not completely. Many people tell me that the help I get from the respirator now is minimal, but I am very scared and cannot completely stop using it. Thanks to the therapy I feel much less tired during the day and compared to seven years ago I don't need a nap in the afternoons any more.

In 2010 I graduated from a three-year course at the Faculty of Psychology, and am currently taking a specialist degree course at the same Faculty. I wasn't able to graduate standing on my own two legs as I had hoped but it was still an emotional moment; without the help from the therapy I probably wouldn't have been strong enough to attend classes, prepare a thesis and graduate. I may not even have known what university was without the help of the drug.

In January 2011 I had a light pulmonary infection and as a precaution I was admitted to the Meyer Hospital in Florence. I was better within three days and just had to take antibiotics for a week.

In April 2011 I had an operation to lengthening my Achilles heel as I had a club foot, and thanks to this, after a year of rest, I am now back on my feet. The operation went very well and I got through the general anesthetic without any post-operative complications.

I am trying to do the best I can in everything I do, but it isn't easy. Luckily I have my parents and my two brothers who are always with me and give me constant support and help.

Pamela Pieralli, Italy