

PAMELA

Glycogens type 2

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.



Summer 1998.
Before the worsening of
the illness.

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.

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.

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

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.



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



Thanks to the therapy I gained strength and I was able to study and to get my diploma from the Scientific Lyceum. Presently I am enrolled and attend courses of Psychology at the University of Florence, accompanied by my mother and a tutor. It is my wish to be able to obtain my self sufficiency and to be able to get my master degree with my own legs.

I sincerely hope that the therapy arrived to everyone who need it before the disease advances so they can live a life as normal as possible and they do not have to put in motion all what has been done in Florence for me.