Yves Story

Yves Leblond, September 2011

Yves with his dog

Pompe disease, acid maltase deficiency, glycogen storage disease, alpha-glucosidase! Different words, but with the same effect on the body. My name is Yves Leblond and I am 39 years old.

Yes I had a happy childhood. I was like any other kid. I used to practice a lot of sports and I was good at it. I was not the fastest one but I had good vision of the game. Who can believe that I use to play hockey and do some windsurfing?! Yes I admit that sports took a large part of my time when I was young and I do miss it very much.

During the winter of when I was 20 years old, I was running outside and suddenly, I fell on the ground. I realized that something was wrong with me. After many appointments with my doctor all my tests came back normal and my doctor advised me to go to the gym to rebuilt my muscles. So I decide to go to another doctor and after this decision my new doctor told me to go to CHUS at the department of neurology. A few months later, I was diagnosed with Pompe disease after I got a muscular biopsy in my left biceps. It was in 1997. I was 24 at that time. It was really difficult to get such a message, but at the same time it was good to know what wrong with me. I am sure that it was devastating for my family to hear such a news and that there was no treatment for this kind of disease.

Yes it was really tuff, but I told them to not be guilty about this situation. That this is life ! We learn every day about this disease. Can you believe that I had more chances to win to the lotery compared to receive the diagnosis of pompe disease !

How did this disease change my life? In my case the disease changes slowly, but too fast for me. The time goes too fast. I think that what we mostly observe is a capacity limit. One day we are able to do something and a month later I realize that I'm not able to do it anymore. My lower back, my shoulders and my shoulder plates are really weak. From this moment on, each time that I loose strength, it becomes harder and harder. The physical therapist is impressed that I am still able to walk short distances. Outside it is really difficult to walk, because there are a lot of obstacles on the floor. A little rock, the wind, the snow, water, sand.... This all can easily make me fall. When fell almost 6 feet, you will never forget that !! Ouch. Falling happens frequently, my equiliber is precarious. Since the age of 37, I am not able to walk anymore and I had to use a wheelchair to move around.

I am lucky to be able to stay with my parents. They help me a lot. They are always there for me. I know that not everyone has this opportunity. If one day my health will be improved, I promised my mother to wash the dishes for the rest of my life. When I think about it I might need to employ somebody!!!

In November 2004, I went to North Carolina with a friend to consult a physician at Duke Medical Center. There they performed a clinical study on juvenile and adult onset Pompe patients. I meet Dwight Koeberl, a doctor. I had to do a lot of physical tests. It was source of motivation. I enjoyed to participate in this study.

In August 30 2005, I went to the Biomedical Center of Pittsburgh to participated in the screening study protocol sponsored by Genzyme. I met Dr. Clement and 2 other patients with the late onset Pompe disease. One of them received the diagnosis in 2004. He was 38 years old. I can figure his reaction when he saw me in my present condition. I was like him when I was 20 years old. I know that Pompe disease

can occur at different ages. It was the first time that I was in touch with some people with the same disease. It was really fun and I had good time. My only deception was to not meet Mario Lemieux and Sidney Crosby at the Mellon Arena !!!

Professionaly, I never had problem to make my way. I graduated for my degree in 1997 in administration. I always worked but in May 2005, I quit my job because it was physically too heavy for me. I have been working as an accountant for 7 years and I liked it a lot.

In January 2006 I met the criteria to participate in the study done at Pittsburgh Medical Center and I was admitted to this protocol. I had to stay inside until at the end of these 18 months.

Even if my life quality is very affected, I keep smiling, I keep having hope. I know that I exceeded my limit. Yes I'm scared, but life is too beautiful.

I have a little sister. She is in good health, married and has two young children. By the way, I proud to be the God father of the oldest. When my nephew takes my hand for playing and I can't, because of my handicap, It breaks my heart.

To give a example to my physical situation, imagine that you are making a card castle, a little vibration, a short breath and everything collapses. It is realy simple, you take a look at me and you understand that all the easy movements are really difficult and ask a lot of energy.

It is difficult to understand that a simple deficienct enzyme that is not able to break down sugar or glycogen in all muscles can cause a lot of problems. When I see pictures of my left bicep biopsy, I undertand. When you take a look at it every part of the body is very important, even a small one. Here in Canada, I heard that there are 13 other people with infantile and adult onset Pompe disease.

Since 2010, I use a bip-pap during the night and my sleeping improved very much. I feel much better during the day and I never feel tired anymore. My fatigue completely disappeared. I still receive infusion with Myozyme in a hospital closer to my home every two weeks.

Keep hope and take care.

Yves Leblond, Canada