

Yves Story

Lately, me and 2 of my friend were here in Toronto to visit Maryland Park. When I saw the dolphin show, I was excited but at the same time I was kind of sad because it reminded me of my childhood. My name is Yves and I'm 32 years old.

Yes I did have happy childhood. I was normal like every 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 used to play hockey and did some windsurfing! Yes, I admit that sports was playing an important role in my life when I was young and I do miss it very much.

During the winter 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 a lot of appointments with my doctor all my tests were normal he said. He told me to go to the gym to rebuilt my muscles. So I decide to change doctors and after this decision my new doctor told me to go to CHUS in neurology. Few months after, I was diagnosed with Pompe disease after I got a muscular biopsy in my left biceps. This was in 1997 and I was 24 years. It was really difficult to learn about the diagnosis, but at the same time I finally knew what was wrong with me. I'm sure that it was devastating for my family to hear this and that there was no treatment for this kind of disease.

Yes it was really hard, but I learned my parents not to be guilty about this situation. That it can happen in life! We learn every day about this disease. Can you believe that I had more chances to win the lottery then to receive the diagnosis Pompe disease!

How does this disease change my physical ability? In my situation the disease changes slowly, but too fast for me. Time goes too fast. We observed the limitations of my capacity. One day I was able to do something and a month later I realized that I'm not able to do it anymore. My lower back, my shoulders and my shoulder plates were really weak. From this moment, each time that I loose strength, it coming harder and harder. The physical therapist is impressed that I'm still able to walk short distances. However it's really hard to walk outside, because there are a lot of obstacles on the floor. A little rock, the wind, the snow, water, sand.... can make me fall. When you fall almost 6 feet, you will never forget that !! Ouch. I do fall very frequently, my equilibrium is precarious.

I'm so lucky to stay with my parents. They help me a lot. They are always there for me. I know that not everyone does have this chance. At the day that my health becomes good, I promised my mother that I will wash the dishes for the rest of my life. I think that then it might be good to employ somebody!!!

In November 2004, I went to North Carolina with a friend to meet different physicians at Duke Medical Center to participate in a study on the natural history of juvenile and adult onset Pompe disease. I did meet Dwight Koeberl, a physician. I have past a lot of physical tests. It was source of motivation. I did like participating in this kind of a study.

On August the 30th, I went to the biomedical centre of Pittsburgh to participate in the screening study protocol sponsored by Genzyme. I did meet Dr Clement and 2 other patients with the late onset Pompe disease. One of them had 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 met people with the same disease. It was really fun and I had good time. My only deception was that I didn't meet Mario Lemieux and Sidney Crosby at the Mellon Arena !!!

Professionally, I never had any problem to make my way. I got my degree in Administration in 1997. I always have worked, but in May 2005 I had to quit my job, because physically it became too difficult for me. I worked as an accountant for 7 years and I did like it a lot.

Even now my quality of life very affected, I keep smiling and I keep up hope. I know that I have exceeded my limit. Yes I'm scared, but life is too beautiful.

I have younger sister. She is in good health, she is married and has two young children. By the way, I'm proud to be a god father of her eldest child. When my nephew took my hand to go playing, it broke my heart, because I can't because of my handicap.

To give an example to my physical situation, imagine that you are making a card castle, a little vibration, a short breath and everything will collapse. It's really simple, when you take a look at me you will understand that all the simple moves are really hard and ask a lot of energy.

It's difficult to understand that a simple deficiency of an enzyme, that isn't able to break down glycogen in all muscles can cause such a lot of problems. When I saw a picture of my left bicep biopsy, I did understand that. 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 patients with infantile and adult onset disease. I would not be surprised if I would hear later that there are more patients, but that they weren't diagnosed earlier.

Finally, I will turn 33 years on October 21st and I think that everybody knows my biggest wish.

Yves



Yves



Yves and his nephew Kas



Yves and his nephew Sam