

## Living With Pompe

By: Juan

I was born on Nov. 28, 1977 to Vic and Cynthia. Two of the most loving parents in the world. For the first 10 years of my life, things seemed pretty normal. I grew up just like any other kid. Enjoying time with family and the company of friends. Life was carefree and most of the time spent playing outside with the neighborhood kids.



We never had any inkling that something would change our lives forever. Sometime when I was 10 years old, my parents started noticing that I had difficulty keeping up with my friends physically. I was a slow runner and often stumbled.; this was especially apparent when we were playing games such as hide and seek. Riding a bike was also getting difficult. My parents decided to bring me to the family pediatrician. The pediatrician gave me a check up but found nothing except that I had flat feet. So he decided to just ask me to buy an insert for my shoes and to exercise in order to combat the effects of being flatfooted.

Life went on as usual. Days were still spent either studying or playing with friends. I especially loved swimming with my friends. As time went on my parents still noticed that I lacked the physical capabilities of other kids my age. It was also apparent that my condition was getting worse. It was definitely not my flatfeet that were causing these problems. My parents decided to look for doctors that could figure out what was going on.

They eventually found a doctor at a university hospital. The doctor prescribed some tests for me and eventually concluded that I had muscular dystrophy. Due to the lack of technology available at that time here in the Philippines, muscular dystrophy became the suspect. The prognosis for muscular dystrophy at that time was bleak. They could not do much for me except give me physical therapy to help combat the deterioration of my muscles.

At the age of fourteen my family had the chance to go to San Francisco to see one of the experts in Muscular Dystrophy. We went to California Pacific Children's Hospital to see Dr. Becker. With just one look at me the Dr. said that he thinks it's not Muscular Dystrophy but he will run some tests to make sure that his assumption was correct. A muscle biopsy followed my doctor's visit.

A few months after the visit, the test results were mailed to us back in the Philippines, the doctor's assumption was correct. I did not have Muscular Dystrophy. Instead we now had a confirmed diagnosis that I was suffering from Pompe disease, a rare metabolic disorder that shared similar symptoms to Muscular Dystrophy.

A long period of searching for doctor's that knew anything about Pompe ensued after the confirmation of the diagnosis. Living in the Philippines it was not easy to find medical help. There were no local doctors that knew much about Pompe. It was merely a footnote in some obscure medical book to them. With patience and perseverance my father was able to find a medical researcher in NY doing work on Pompe disease. Frank Martiniuk was studying the genetic mutations of Pompe disease he also knew of a doctor that specialized in helping Pompe patients manage the disease better. Frank pointed us in the right direction and introduced us to Dr. Alfred Slonim. Months of correspondence followed after the initial introduction, which resulted in an invitation by Dr. Slonim for my parents to fly me to NY. He would gladly see what he could do for me if my parents found a way to get us to NY.

By the time we were ready to leave for NY I was already 19 years old. Several years have passed by since the initial diagnosis of Pompe disease. I was 5'7" and weighed like a feather at 60 lbs. I was also in poor shape. I had severe pulmonary infection that resulted in a terrible cough that would not go away. My mom even pleaded with my dad to postpone the trip. My dad said that we might not get another chance to do this and that it's worth the risk.

Although it was an eventful trip to NY the results made up for it. Dr. Slonim upon seeing me prescribed a high fat, high protein diet. He also advised me to go on a Bi-Pap machine to aide in my respiratory problems. He said that as a result of Pompe's my lung muscles were not strong enough to expel carbon dioxide properly, which was causing all sorts of respiratory complications. Not to mention the chance of having a heart failure due to the excessive amount of carbon dioxide in my system.

Months after the visit to Dr. Slonim, my weight saw a drastic change for the better. I was able to gain 50 lbs that brought my weight up to a high of 110 lbs. The heaviest weight I ever attained in my entire life. As a result, my physical appearance improved as well as my self-esteem. I also had more energy to accomplish my daily tasks. I was able to finish college as a result.

The years that went by saw the disease progressed much slower although it still continued to progress. While my appearance seemed better my strength still deteriorated. From being able to propel myself in the wheelchair and doing transfers by myself I slowly had trouble with it, until I needed an aide to help me with daily activities.

My breathing also became much more difficult. After the initial visit to Dr. Slonim I used the Bi-Pap during the night while I slept. As the years went by I saw myself using it more and more. First during break times at noon until my present state now wherein I use the Bi-Pap all of the time, save for the time I take a bath. But even then an aide assists my breathing by making a fist and pumping my stomach to push air in and out of my lungs.

While there has been an improvement because of the diet, Pompe is still slowly edging it's way and taking more and more of my freedom. Currently I am totally dependent on an aide for all my daily activities. I also depend on a Bi-Pap machine for my respiratory needs.

My life has been severely affected by Pompe. I am now 28 years old, working in a business I've put up with my brother, trying to make the most of what I have. It's a hard and difficult life. A lot of liberties have been taken away. I only have a semblance of a normal life because of the love and support my family have given me. With out them I would not be able to do most of the things I do now.

I together with the other Pompe patients have been eagerly awaiting a treatment. And at December 2005 my day had come. My first infusion day was a very special day. My parents, brother, aunts, best friends and physicians were with me to share the special moment when I would receive enzyme replacement therapy with Myozyme for the first time in my life. I can't tell you how special it felt when my mother pushed the start button of the infusion pump and Myozyme started to flow in my body.

About one month after I started treatment I started to feel better. Somehow it felt as if I did have more energy. A couple of months later I noticed that my grip has strengthened. I also noticed that the deterioration, that I knew so well during the past years, had stopped. All in all I am slowly improving

I feel grateful and happy that Pompe patients do have a treatment now. That we can go back to a life that is normal. We have a lot to offer this world. We are talented, bright and creative individuals but we need the strength to make a difference in this world. The treatment will give us a chance to make our marks in this world.

Juan



Celebrating my first infusion together with my family and friends

