

Jared's story

My journey with Pompe disease began just after high school. A routine metabolic and chemistry blood test indicated that something was wrong. The results came back showing elevated liver enzymes: AST, ALT, and LDH. This prompted a major workup to determine the cause of my "liver condition". My abnormal blood chemistry continued for many years to mystify my doctors. At this time, I knew nothing about medicine or about the human body. I wasn't particularly fond of the life sciences either. My abnormal blood chemistry led to an intense campaign on my part to try and explain what was going on with my body. My doctors continued for years, sidetracked, thinking I had a liver condition. My efforts helped steer my doctors to a specific diagnosis. Nearly five years later, at the age of 25 I was given the diagnosis of Pompe disease. Sometimes I wonder if I have over-reacted to my diagnosis. After all, I am in pretty good health. I guess my shy, sensitive nature, and the feeling growing up that I was so different than the others caused me to look towards my diagnosis as an explanation for why I felt this way about myself.

When the blood work originally came back abnormal I feared the worst. I was a sick kid growing up. Even at birth, I had a difficult time coming into this world. Due to a blood compatibility problem, I was born jaundice and lacking oxygen. Due to good luck and very keen doctors at Loma Linda University Medical Center, my life was saved. A relatively unknown blood factor was responsible for my jaundice and hypoxia. A blood transfusion saved my life. Perhaps this early experience caused me to have subconscious fear of doctors and hospitals. While you may think this common, I can assure you it was not. One only needed to observe the crazed terror I exhibited, to realize how serious it was. I can remember lashing out like a wild caged animal toward the staff when I was only four years old as they attempted to put me under anesthesia for a tonsillectomy.

There were some subtle clues early on that I was not a normal child. As early as the third grade I was teased by a couple of students for the way I walked. Apparently, I shifted my hips when I walked. Also, I could never do sit-ups. I remember in the Fourth grade, "Field day" was a day I tried to avoid. I remember telling my mom that I was sick in order to get out of having to do sit-ups, one of the events during field day. Eventually, I had to make a decision. Either I run away from home or I return to school and "make-up" field day. I eventually survived it although I can't say that I came out unscathed.

The feeling of being abnormal, although not unique for kids that age, was particularly severe and prolonged in my case. I was very embarrassed about being overweight. I vividly remember having a rare good day when I wasn't ridiculed about my weight. These feeling although never really went away, came to the forefront during physical education class. I was always terrified I would be forced to do something that would cause my peers to ridicule me; this meant any sport which required that I run. I had a unique gait when I ran. I guess it must have looked like I was running through water, there was great effort, but not tremendous results. Thanks to the advent of Physical Fitness testing brought on by our "wonderful administration", I spent a portion of every year under intense anxiety. I hated being the worst in the class. The events that cause me the greatest anxiety were the rope climb, cross country, sprint, hurdles, and of course the dreaded sit-ups. My anxiety level was least during the softball throw,

basketball shooting, and pull-ups since a lot of kids couldn't do pull-ups. There was a time when I could come quite close to be able to do a sit-up. In fact, if used the mat to prop my back up a few inches I could do a few sit-ups. Of course it didn't take long for the coach to notice I was the only student with my butt on the floor and my back on the mat.

After not being able to do sit-ups, pull-ups or the rope climb, the PE teacher pulled me aside and questioned me about my poor athletic ability. The PE teacher asked me about my daily activities. I told her nothing was wrong. I was totally active and loved basketball. This was true after all. I gained confidence playing basketball. I had a court at home and spent hours and hours playing basketball and perfecting my shot. I became quite good actually. I lacked quickness and jumping ability, but this wasn't unusual for a slightly chubby white guy. There wasn't anyone I knew who could beat me in a game of horse. Guys would overlook my unorthodox running style when saw what a great shot I had. I was even quite capable in games of one on one thanks to my quick shot release and my sheer determination.

Starting in high school at about 15 or 16 I would experience episodes where I began to feel really rundown. It's hard to describe, but I would feel very tired and "headachy". Trying to push myself during one of these episodes would result in a severe headache which could only be eased with rest. I'm not sure what this is all about but I continue to feel this way from time to time. My instinct tells me this is related to Pompe.

My breathing ability is out of the dangerous zone, but my FVC has fallen from the low 90s to high 60s since my diagnosis about 7 years ago. I am still in good health, although I am not as active anymore and no longer enjoy playing basketball and racquetball. My ability to run has diminished. I no longer have the strength in my legs to support running. I continually feel run down and tired. Feeling this way has caused me a certain amount of depression.

I continue to work full time although my productivity feels like it has dropped a lot over the years. It takes all of my energy getting through a day of work and when I get home I have nothing left to do things I once enjoyed. Sleep and caffeine seem to be the only things that make me feel better. Getting a good night's sleep has become a very high priority for me.

While I feel very fortunate to be in as good of health as I am, I can't help but wonder how things might have been different if I were treated at a very young age. Would this have had a major impact on my self-esteem? Would I have pursued my ambitions to become a pilot? It's impossible to say. I look forward to the day that enzyme replacement therapy is available. I think my science background allows me to be realistic about the prospects of enzyme replacement therapy. I consider it a worthwhile treatment, not a panacea. I also realize that next generation products will only get better and better. Thanks to biotechnology we can all have hope for a wonderful future.

Jared