

Cynthia's brother's story

This story started many years ago, well not really; maybe I suppose its really about 30 or so years in all. But it just seems forever when you look back at certain things. Well, I noticed that my sister Cynthia seemed stranger than usual when walking. She was married to Graham, had two beautiful boys; a nice house and things were looking all right for her.

I was younger, just starting out with my career, busy with girls and I think in a world of my own. Too involved with my own life to see maybe what I should have seen earlier.

Cynthia gave birth to the first boy, Ben back in '77 and I was up like a flash to see my first nephew. I suppose that I spent more time with Ben than with my own sister. Still busy as hell, but a little time made for Ben. You don't notice what you should when you are busy.

The next nephew came along in '79. Troy. Again I was rapt. This baby business is great. This time I was married, still too busy, but still spent some time for the nephews. I should have spent more time with my sister.

About this time, I noticed that Cynthia had developed this quite strange little walk. She hid it well, I must admit, but I noticed it just the same. In hindsight, I noticed it all right, but probably years later than I should have. My older brother, Steve, has said, in hindsight I suppose, that when Cynthia was about 11 or 12 years old, she was in school sports day running races. She ran the race of her life and came 'stone motherless last'. Steve says, that it wasn't coming last that was the problem; it was the way that she ran. He says it was like a duck running but slower.

I have all the excuses in the book. She went off and got married when I was in my mid teens, and I was much too busy studying at school, to notice. She was busy, I was busy, and so I am blaming everybody else for me not really seeing things that I should have.

She was always as a kid very active. She worked, played tennis, and was involved with the army, boys. There seemed no end to her activities. Marriage, sons, housework,. Always on the go for this young mother of two. I was impressed. You would get whiplash watching her up and down. Later in life the tennis thing was an issue. She would just stand in the court and you would have to hit the ball to her, because she used to say I am not running for it'. Because she said she couldn't.

Yes this walk was weird. I tell you. I am thinking what the hell is going on. I put it down to old age; after all she was only in her late 20's. Visited often to the See the boys. See Cynthia, not too much seeing mind you. Anyway this walk was odd. I could not describe it. Typical male. Put it down to having babies, and being generally unfit.

I was so full of being Mr. Right, that I gave my sister a weight-training boot. Yes lets solve the problem. You need to work out. Pump some iron. Have legs like mine.

We have a problem. This once active girl is getting worse. What the hell is going on??? There has to be a reason. Diet? Fitness? Bad posture? The Ozone layer? Who knows?

Now the thing is, when you have some sort of medical problem, and you don't know what the hell it is, some people try to hide it, and compensate for whatever ails them. Yes, we can suffer in lots of ways. There's the physical thing that's wrong and coupled with that is the stress, the emotional hang up and the embarrassment.

Now this horrible problem it sneaks up on you. It started very slowly, and who knows when it actually did begin. And it painstakingly gets worse and worse and much worse. After a while, the truth comes out and she can't hide it anymore. Yes, there is something wrong with me. I can't walk much, I can't bend over, and I have trouble picking things up. I don't know what the hell it is.

Now this strange walk is getting real bad. I would hate to describe it. Couple it with the upper torso swing like an upside down pendulum, and the arms going left right and everywhere.

So she goes to see a Doctor, and after a while, seeing lots of experts with no idea of what the hell is going on, she gets diagnosed with this Pompes disease. It must be a huge problem, because they

gave it other names as well. And the good news is.... well; there is no good news. It's all bad. The worse thing you want to hear.

Now back a few paragraphs ago, when I wrote when you have a medical problem and you don't know what it is and some people try to hide it and compensate for it, etc, well the thing is when you also have a medical problem and you know what it is some people try to hide it and compensate for it.

This is a real bad side of any disease; you don't want people to notice you. It's embarrassing, not being able to walk properly. It's embarrassing being different. People start to treat you different and that can be annoying. There are lots of different pains that people who are ill have to put up with.

With Pompe; your muscles go. Cynthia's muscles are going, going, going.... all but gone. It makes you wince when I see her even try to get up out of a chair. She can't. She has to even sleep with a damn machine. Every organ is packing up and dying. It's just not fair. What do you do?

Now the thing I love about my sister is the attitude. She is stubborn, determined, and resolute pain in the backside you have to admire and love the spirit of someone faced with the news that she has the dreaded Pompe, but still makes the most of everything she has.

She has a wonderful support group. Graham her husband, now the full time carer is there. Her two boys Ben and Troy are there for her. Her friends on the net, who are facing the same future, are there. Her family, her Mum and her Dad, when he was alive, her brothers, and of course me especially, her future daughter in law, her other in-laws, all of whom have been terrific.

This support group was evident recently, well a year or so ago, when Cynthia got real bad and had to go to the Royal Adelaide Hospital and the Hampstead Rehabilitatee Centre. She made the big time and stayed in the Intensive Care Unit at the Royal Adelaide. Good care. Good people.

Now there is no more hiding what ails her Cynthia is now stuck with a walking frame with stupid bears on it. She also has a wheelchair, that goes really fast, but I am not allowed to do wheel stands with her in it. She has a respirator to breathe at night.

Now the next phase of this horrible malady is happening. Well two phases really. One phase is the future of Cynthia and the disease without any treatment. Who knows what will happen there. Well I suppose we all know what will happen, just don't know when. But the other phase is interesting and gives cause for some sort of hope. Enzyme Replacement Therapy. Treatment. Designed to make Pompe sufferers better, I hope. We will see what this brings. I am not holding my breath for a miracle, but it would be nice, because I have a younger brother with the horrible Pompe as well. So I hope like hell this ERT is the answer for Cynthia, my little brother Anthony and for all the Pompe sufferers the world over.

Cynthia's brother

I (Cynthia) started enzyme replacement therapy with Myozyme at March 23rd 2006. I have received 20 lots of Myozyme now. I am not sure how it's really going now...with Pompe disease everyone gradually loses parts without knowing. Then when those parts come back it's not until someone else points it out that you then realize that..and think 'Hey, they are right'.

I keep being told by friends that I am looking well...my colour is good, my eyes are sparkling and I look vibrant. When they tell me that I wonder how I must have looked before. I feel better in myself and am doing little things. My balance is better. I feel like I could walk further, but the frame is sometimes a drag. However I am not complaining, because I know some people would love to have that complaint. I am so lucky that I am still able to cook meals, do dishes, shower myself and able to partly dress myself. My husband only helps me with the lower part like putting on socks, shoes and putting my pants over my feet...then I do the rest. I still eat well and I can clean my teeth and brush my hair. I only use my ventilator during the night for about 8 to 10 hours.

My doctor has noticed that my neck strength has improved and that my arms are stronger.

Cynthia



Cynthia receiving her infusion with Myozyme