

I have Fabry disease and here is my story. Bob L. (United States)

Over the last few years, I have had an unbelievable journey, and I am still here to tell the story. I hope this story will inspire everyone to never give up.

I woke up, I did not know where I was, oh yes, I just went through a major open heart surgery. Once I was out of recovery I went on to the Cardiac floor. I guess you are wondering how I ended up here. Well, several weeks earlier, I went to see my Nephrologist, you see, the disease has destroyed my kidney functions. My creatinine level, this is what tells you how well your kidney's are removing the toxins from your body, were increasing pretty rapidly. Little did I know that I was informed that I only had fifteen percent of my kidney function left, and I needed to get on the list. I asked what list, the national kidney donor list, she said. I was shocked, I knew my kidney's we're getting worse, I was getting sick on my way to work on a regular basis, but I guess I did not want to admit to myself that I was that bad.

The hospital suggested besides getting on the list I should solicit family, and friends to possibly be a donor. I called my wife to inform her of the bad news, without hesitation she said, I will get tested to possibly be a donor. I believe in fate, I have a strong faith, and low and behold my wife was tested and was a match.

Now it was my turn, I needed to have my heart functions checked out to make sure I could survive a major operation.

The procedure is called a cardiac catheterization, I was in the operating room having the procedure; and it did not last long. The doctor barely started and suddenly announced, okay we are done, I was thinking this is either really good, or really, not so good. The latter was the case, they found four blocked arteries. Immediately I was scheduled for the bi-pass surgery, and here I am in the hospital room. Unfortunately the stress of the operation caused my kidney's to fail. My creatinine level, which should be around one rose to twelve. I could hardly lift my head off of the pillow, they started dialysis to try to bring the levels down, I laid in bed as they hooked me up to the dialysis machine, I did not have the strength to sit up. Normally dialysis is completed every other day, three times a week, they were giving it to me every day to try and get the levels down. The levels did not go down, I was lying in bed early one morning and looked up and said "is this it?" No sooner did I say that, toxins started coming out of my mouth, I started feeling better and after sixteen days in the hospital I finally was able to go home.

Now I needed to recuperate from the heart surgery so that I could have the kidney transplant. Of course since my kidneys failed, I had to continue dialysis treatment three times a week, I have to say this is horrible, four hours hooked to a machine, like a washing machine cleaning your blood of the toxins, and then putting it back into your body again. This drained me, not only am I recouping from the heart surgery, but getting treatments to boot.

Three months later, the transplant takes place. The operation for both my wife and I went well, my wife was in a lot of pain due to the position she had to be in while they removed one of her kidneys. She was in the hospital for three days and then was able to go home, I was there for eight days and then homeward bound

My wife saved my life, and for that I will be eternally grateful.

Ah but there is a twist, when my wife was going through the testing process, they found out that she had something called CMV. This is a potentially deadly virus. She was actually born with this; thousands of people are. But if your immune system is stable you could live your whole life and not know you have it.

We were informed that they have performed many transplants with someone having this and it would be no problem. Well I guess you could imagine what happened next. I was given specific medications on the outside chance I contracted CMV, after six months clear and free they said it was ok to stop taking the medications, no sooner did I stop, I contracted CMV.

I have to say by far this was the worst pain I have had in my entire life. The diagnosis did not happen right away, I was in and out of the hospital the levels should be zero, mine were several million. Once diagnosed, I was given massive doses of medications to reduce the levels.

The only thing that would remotely reduce the pain was morphine, it got to the point where i was in so much pain I was begging the nurses for it, eventually the levels started to go down. I was able to go home.

A port was put in, my wife learned how to give me the medicine, everything was going well for a time.

Okay, I thought to myself things are going pretty well. Little did I know of what was about to take place. I awoke early one morning with blood all over my chest, something was very wrong with the port. I am yelling to my wife and daughter that the port is bleeding. Ah I forgot to mention that the CMV affected my blood platelets, my body now destroys my own platelets. Blood platelets help with the clotting process, the normal range is 150-200 thousand, my levels run around 30 thousand.

So needless to say if I am bleeding badly it could be a very serious situation. My daughter gets towels and puts on pressure to try to stop the bleeding. My wife gets the car started and off we go to the hospital.

I immediately went into the ER. The same doctor is there from my other visits. That is pretty sad when they know you by name. They were able to stop the bleeding. It was determined that I had a blood infection and the port needed to be removed. The doctor started to pull it out and it stuck, he said, I will be right back. I need to get a surgeon . That is the last thing I remember, I flatlined.

I later found out that my neighbor, who is a nurse at this particular hospital, was one of the group of nurses and doctors that ran to my room when STAT was initiated.

I am not sure how long I was out for, and to this day I have not asked our neighbor, I just do not want to know. I finally came back; I did not know where I was. I looked up my wife was there. She left right before they were going to remove the port. My daughter and sister-in law were

there. I said to my sister-in law, what are you doing here? There were doctors and nurses there; I was in the ICU. After I was stabilized, the helicopter from a major hospital was flown to the front parking lot of the local hospital I was in, and off I went.

I was in the ICU for eight days, then to the floor for a few days, and home again.

My thought now was, I hope this is the end of my challenges. Unfortunately that was not the case.

There had been a contamination at the plant that manufactures the medication that I receive intravenously every two weeks to replace the enzyme that I am missing. There was a need to ration the doses each patient was to receive. After a period of time of not getting the full dose, I became very ill and wound up in the hospital again. Fortunately they were able to make what they call a mercy plea and obtain a new drug to the United States. I was immediately started on this drug and after a period of time started feeling better.

You may think the story is over, but not so fast, I developed a Hernia from the Kidney transplant, also the new kidney is pushing on the walls of my stomach.

Well you say , get those things straightened out, ah I would in a heartbeat, except something else came into the picture.

One night as I was watching TV, I had a sharp pain in my head, it felt like someone was sticking a knife in my head. It would last a few seconds and then happen again, over and over. Now this lasted about twenty four hours. I notified my doctor and he ordered an MRI. So off I went. I needed to have my bi-weekly treatment as well, so I had the MRI and then went right to the infusion center. I was waiting to get the infusion and my doctor came in and said can you follow me. We went down a hall, as I was walking I was thinking to myself, this cannot be good. We went into an empty room and I sat down. He said, I have some bad news for you, the results show you have lesions on the right side of your brain, and an aneurysm on the left side.

I think I wanted to cry, how much more could I take. The decision was made to have an MRA completed, which is a more comprehensive image of the arteries.

The MRA went fine. I was very anxious to hear the results. Maybe the MRI was incorrect, not likely, but you never know. A few days later I received a call from the doctor, he asked if my wife and I could come down to see him.

I had a pretty good feeling, having two of my brothers who have passed away from this disease, that the news was not going to be good. My wife on the other hand, tried to stay positive.

We went into his small office and the doctor began showing us images from the MRI, and MRA. The buildup, GL3 I believe it is called, is in the arteries in my brain.

The arteries, the doctor explained are widening and weakening. The infusion treatments have no effect once it is in your brain.

My wife knew what this meant and began to cry. The doctor looked at me and said, you know because of your brothers, I shook my head , yes.

Needless to say the ride home was a silent one. The doctor could not give a timeframe as to when something would happen, due to the fact that they do not know when the buildup initially reached the brain.

I have had many what they call mini strokes since this news was broken to us, but not the big one yet.

I know you have to be saying to yourselves, this has to be the end of this story. I wish it was.

I really feel bad about this next incident, not that I could help it. We were just getting ready to go on a company paid cruise. We thought if we were ever going to go on a trip the cruise idea would be the best. Doctors and nurses were right there on the ship in the event that I may have something come up. We were so excited to have some time to relax, enjoy life a little after everything we have been through.

It was not to be. It was two days before we were ready to leave and I woke up with blood was coming out of me. It was like a CSI crime scene. This continued for what seemed like forever. Again I was brought to the local ER! Again the same doctor; in hindsight I suppose this is a good thing. My platelets finally did their job and I stopped bleeding, I had lost two pints of blood. I was given two transfusions. My hemoglobin level which should be around 16 was at 6 and my blood cell count which should be about 40 was at 20.

We both thought it actually was a blessing that it happened when it did. What if this happened on the ship! But we were still very disappointed. Back to the bleeding issue. After I was in the hospital for a few days we needed to find out what caused the bleeding in the first place. Off we went to have an Endoscopy. This procedure for most people is pretty routine, of course not in my case.

My wife and I were there for hours as they went back and forth deciding which way to proceed with the issue is the brain aneurism. Putting me out completely could be very dangerous to say the least. I could very well have a heart attack or stroke.

So after a lot of deliberation the decision was to put me out but to be at the ready to bring me back if needed. Well, all was going as planned. They were just getting ready to do a biopsy of my intestines and I started to lose oxygen to my brain. They immediately brought me back.

The doctor contacted me and informed me that the veins in my esophagus had burst due to the buildup, and that is what caused the bleeding.

I am finally almost at the end of my story, I hope!

The last incident just happened very recently. I was conducting what we call an “all employee

meeting” at our office. I had just completed my piece; I was the General Manager. Yes, I was working at least fifty hours a week in a very stressful job during all of this!

I was one of the supervisors who was conducting the training. I began to get an excruciating pain in my chest. It felt like someone stabbed me with a sword through my heart. Luckily I was standing right next to a desk, I held on as I began to fall to the floor. I managed to get out of the room and into the garage area where I sat down hoping that the pain would subside, but to no avail.

Some of the employees saw me and called 911. The ambulance came. The EMT did an EKG and stated, you are going to the ER! There I am in the back of this ambulance on the way to the hospital. This one strangely enough, I have never been to.

They were able to stabilize me and the nurse started to ask me about my medical history. She said, let's start with the head and go down to the toes, and in what seemed an eternity we were finally done!

So here I am, sitting on my deck, not feeling too good today with upper respiratory issues, yes that is another symptom of FABRY.

It is a beautiful day; it's pretty warm considering it is the beginning of October. I am glad to be alive to enjoy it. I was trying to decide how to end the story, but I think I just did.