

## **I have Fabry disease and here is my story. Cheryl B. (United States)**

Hi I am a wife, a mother, and a grandmother, I love our family Sunday dinners together, I am a full time employee with a school district in New York. I am active in my church, and do volunteer work. I LOVE cruising and I'm a Fabry patient.

At 16, I was diagnosed as a carrier of Fabry. That is what they said years ago, girls and women were only carriers.

I had many symptoms, growing up, but never attributed it to Fabry.

I had a hard time, especially in the summer with the heat. I am so glad that we had a pool; otherwise I believe I would have suffered with more symptoms.

Growing up, we were an active family. It was often difficult for me to keep up with them.

I remember one summer day, I was looking forward to going to a parade. I was so excited; I couldn't wait for Dad to get home from work, so that we could go. It was a very hot day; Mom didn't want me in the pool anymore because we had to be ready when he got home.

Prior to Dad coming home, I remember lying on the couch on our back porch not feeling well. My hands and feet burnt, it felt like they were on fire. Mom took my temp and it was 104. I had no other symptoms. All that day I had felt fine. Just a few hours before, I was in the pool having a great time with my friends, and by the time my father came home, I was too sick to go to the parade.

The fevers did happen frequently, and what we thought, were for no reason at all. I was a sickly child. Let me put it this way I never made the list of perfect attendance in school. LOL!

As a young adult, I would have fatigue, headaches, fevers and TIA's. I had neurological work ups with MRI's, spinal taps, EEG's, and other tests.. After each MRI, and after every TIA the MRI would show more white matter on the brain. I went to numerous specialists. I also went to Mt Sinai in NYC, and saw Dr. Desnick. I would have routine visits and tests there. I went every other year for a very long time. Each time I was asked the same question, "Do you have any pain?" I would say, "No, I feel great, heck I was in NYC!" I am not sure how many years went by, and the same question was asked... one visit there, a resident doctor asked me again, I gave him the usual answer, he then asked, "So you don't have pain in your hands or feet when you have a fever?" I said, well yes, everybody does." I still remember the look on his face. Calmly and with great empathy he said "No, not everybody." This was a light bulb moment for me. Oh my ... so this is Fabry – But wait, I was JUST A CARRIER I didn't understand.

There really was no one who I knew of, that was suffering with Fabry. If you were a female, it just wasn't acknowledged, you were just a carrier and not affected. I believe it is our responsibility to take charge, and to educate ourselves to be our own advocate and if need be, an extended family advocate. I familiarize myself with the test schedules, how often should I have an EKG, Echo's, blood work, MRI's, how often do I need to have a 24 hour urine? Is it time to wear that 3 day heart monitor? What other tests do I need?

My advocacy started, after one of the trips to NYC several years ago in the 90's. I was talking to my Mother one evening and she asked me to please contact my younger sisters and talk with them about Fabry. She asked me to help them better understand the process of screening and testing, let them know the importance of getting their children tested. She wanted me to try and persuade them to look into, maybe contacting Dr. Desnick for a complete work up. It had been years since they were diagnosed "as carriers". My mother said that they thought they were non symptomatic and that there really wasn't much that could be done. They didn't see the point of dealing with something that they thought they didn't have much control of.

I knew I just couldn't call them and tell them to get themselves to a research center, or have their kids tested, who would listen to that? I thought about it for a few days. How could I approach this in a nonintrusive way? I understood that they might not be ready to hear that they, or their children may have to deal with this chronic, progressive disease. In the 90's, the medical literature was just starting to recognize that females may be affected.

After a few days, I did call each sister. I talked with them about my experiences. I let them know that my son didn't have the disease, but my daughter was a "carrier". I told them what I had been doing to better understand Fabry. I could tell that they really weren't ready to talk on the subject right then, but they did listen to me. I would make it a point and call them every now and then. After each conversation I would put a little bug in their ear about the research. I told them I would be available to talk any time. I gently persuaded them to have their children tested. It took a while, but they started to call and ask me questions about my trips with Dr. Desnick. I am happy to say today, that both sisters are on treatment; two of my nephews have been tested and are negative. They are blessed and won't carry the gene further. My other sister's two children, twins, a niece and nephew, were split with the decision to be tested. My niece was tested and is not affected. My nephew is newly married, and he and his wife are not ready yet to see if he has the gene. I pray that he does not.

I am a firm believer on going to as many support groups as possible. Also if you have the chance, please think about going to Victory Junction, as a volunteer or family member. We just had our third annual family weekend. You will be in awe on how many people are there! We

are a great community of people. It is very reassuring to see throughout the years, more and more people at these meetings . We don't feel as isolated. , we may see doctors who specialize in Fabry. We share how we are feeling, what treatments we are receiving. We also discuss what clinical trials may be going on. We ask how family members are, what similar problems each are having or not. People share all kinds of information: How to deal with the weather conditions. What tricks work for what different symptom. We have one day where we have a conference in a hotel in Greensboro, there are a lot of meetings on different subjects that are very informative. The kids have their own meetings based on age groups, if the child has Fabry, or a sibling. Each children's group, have many activities, along with talking about Fabry, how it affects their lives living with Fabry or how it affects the siblings lives that don't have Fabry. After the meetings, we all go to Victory Junction, and forget all about Fabry, we have Family FUN the rest of the weekend! We do archery, swim in a really cool water park, get all dolled up, coloring our hair , there is bowling, fishing, wood working, arts and crafts, horseback riding. There are many kick ball games, especially with the Hill family! There are so many more activities there. We eat a lot, and dance after every meal! We have a Blast!

I also attend area local meetings, they are a great network place. They gave me the courage to find an appropriate doctor. I was at a standstill with my primary doctor years ago; I felt he really wasn't interested in Fabry, he thought that I was a just carrier, because that is what the medical literature told him. He didn't want to educate himself or take interest in Fabry. It was time to change doctors, but didn't know where to go? In 2003 I went to a meeting in Buffalo just like this, and talked with a couple of people from the Rochester area. I asked what doctors they saw. One person only saw a nephrologists', he didn't have a regular doctor. The other gave me his doctor's name, who he had just started going to and felt comfortable with. So that next Monday, I called and asked to make an appointment. I have been with him ever since, we are learning together, what works for me and what doesn't. He listens to me, he is totally supportive, and acknowledges my symptoms and treats them. We are aware that not all of my symptoms are Fabry. We work as a team on my health care.

A geneticist may have diagnosed some of us, others by a neurologist or other specialist. When I first saw my new primary doctor, in 2004, he asked me who prescribed the enzyme. I told him my neurologist, but hadn't seen him in a while. He then asked me, if I would like him to prescribe it,. That way we could keep up with the schedule for testing. Our physicians are learning with us, if you haven't seen your primary physician because things are going well you may not have all of the tests that you may need. I sign a HIPPA form with all my doctors so that they can share the information. They can network together when possible.

At these meetings, we may share tips on how to handle different problems that we are having. I found this tip out at one meeting. If my hands are burning from the neuropathy I put them under warm running water. It really works for me. It doesn't last long, but I can have temporary relief, and get the dishes done all in one sweep!

We all have busy lives today; I believe though, that we may need to get involved in our extended family with Fabry. We can be a great support to each other. We may think they are under treatment so they are fine, let's not talk about it ... Yes, let's not talk about Fabry every time we see or talk to our relative, but sometimes mention what your physician said, what meds seem to be working. Talk to them about that pain crisis that just knocked you for a loop. Let them know when you are having a few good days, weeks, or months, that your Fabry seems stable for right now. Ask that family member how they are handling their Fabry. What physicians have they gone to lately, what test have they had or going to have. This information is important to share, because not all health care providers keep up with the routine testing schedule. We need to make sure that we are getting the appropriate testing, and ask for tests when we are not being requested to have them

I am very fortunate and blessed to have great family, friends, and coworkers. I know that some families along with coworkers and friends want to deny the effects of Fabry. I am sure all of you have heard this, you are under treatment? Why do you take so many sick days? "you don't look sick!" or you're tired ... I know how you feel. Sometimes I want to say, "No you don't" But then know that they can't know how I am feeling,

Many people just don't understand how fatigued we may be at times, or how we hurt the way that we do.

Ten years ago, when I chose to start treatment. I was very nervous to go to my employer and ask for a half-day off every other week. They promptly said don't worry about us, take care of you. I had my infusions for approximately two years in an infusion center. Thankfully I still have that strong support at work and at home.

If you just haven't found that support network yet for yourself, get to know your case managers. They are a great support. They educate us on Fabry itself and the treatments available. They are proactive about our care and treatment, and they genuinely do care. I have had the same case manager for years. Every once in a while I will think of her and shoot her an email. I know she has done the same with me. Or we will give a quick call to each other. I tell her the good, bad and ugly on what is going on. I have called her in tears of joy and sorrow. She has always been there for me. We have a lot of giggles together and I consider her a good friend.

I come to these meetings, for new information. I take notes, and talk to my doctor about it, when it pertains to my care. Every time I go to a meeting, there is always new information, especially these past few years, with what is going on today and what they are looking toward the future?

I have joined many patient support groups on the web. A couple come to mind, FSIG, NFDf and the Fabry Forum. On Google alerts, did you know you can have email updates on the topics of your choice? I put in Fabry in the subject area, and now whenever there is an article on the web that has the name Fabry, information will be sent right to my email.

I was one of the first females on enzyme treatment. I was told that this made me a pioneer for females with Fabry; it was October of 2003 and approximately 200 infusions ago. I consider my grandson Zach as the pioneer of our family. When my daughter Michelle was diagnosed, I was so nervous. I was frightened that the gene would live on, when and if she decided to have children. Remember she was only going to carry the gene, not be affected. Michelle is on enzyme replacement therapy and was accepted in a clinical research trial in September. She suffers very much with Fabry; I pray that she finds relief soon. Michelle blessed us with Zach 14 years ago, and had him tested for the gene as a toddler. When we had the confirmation that he had Fabry, I was in tears. A male; MY GRANDSON had this disease. I thought, we had no hope of treatment, in the near future. What would happen? I only heard of males with Fabry, and never saw the affects up close. I was devastated to say the least.

Now Zach's generation will be the first to have enzyme therapy as young children. We will see how enzyme replacement could lengthen his life and how it could minimize his symptoms. I am so very happy and relieved by this. I still worry about the gene continuing in our family, but when it comes to Zach having children, he will tell you, he is only going to have boys!

I have read up on many lysosomal diseases, and have seen many pompe, gauche, and MP3 patients, while in the infusion centers. If I had to pick any lysosomal disease, Fabry would be my pick. I know we all suffer differently, and some of us are worse than others. With some of the other diseases though, I have seen young children, unable to walk, have tracheotomies, and have weekly infusions, it is very hard to see, and my heart goes out to them. We are so fortunate that we now have the option of enzyme replacement therapy, that for most of us, we see a difference in our own health, since going on treatment. Eleven years ago we had no alternative but to live with Fabry. Today there is a choice to have treatment or not. I look forward to having a better future, and to see my daughter and grandson have the brightest of futures.

We all attend these meetings for our own personal reasons. I want to thank you all for listening, and encourage you to attend future meetings, in pursuit to maintain and control how Fabry Disease affects your lives.

Thank you, Cheryl