Walking in Their Shoes: A Conversation About Living With Fabry Disease

an interview with **Lisa Bacon**

Often the best way to learn about living with a specific health issue is to go straight to the people who understand it best: those living with it. In this spirit, we sat down with Lisa Bacon to learn more about Fabry disease and its impact on her family.

When did you learn that Fabry disease was present in your family? We learned about Fabry disease in our family over ten years ago. My dad was diagnosed through his kidney doctor. We had long known something was wrong with his health, but it took a kidney biopsy to solve that mystery.

Since that day, how many people in your family have been diagnosed with Fabry disease? Seven people, including me and my dad.

When you first learned that you had Fabry disease, were you surprised? No, because we were told that if your father has it, then all the girls will definitely have it. Fabry disease is genetic, which means it runs in families. The changed gene that causes Fabry disease is on the X-chromosome, which means the gene will pass down, if the father has it, to all his daughters. When a mother has Fabry disease, there is a 50% chance for each of her offspring to inherit the gene.

What were the first symptoms of Fabry that you recognized in hindsight in your father? Growing up my dad would always have to stop at every rest stop to use the restroom. He also would have extreme fatigue. I noticed his purple angiokeratomas (lesions on the capillaries) at an early age, but never thought anything about it.

Have you ever had difficulties getting medical care for your Fabry disease because of the incorrect and out-of-date belief that women were "just carriers" not affected by Fabry disease? Not really because I am comfortable advocating for myself. However, my primary doctor doesn't seem to comprehend it all.

What is it like to be in the "sandwich" generation, with a parent and a child being affected by Fabry disease? Watching my dad's health get so bad at an early age definitely affected my son's outlook on his own life. We all try to live a healthy, active life and try to be involved in learning more about Fabry so that one day there will be a cure.

How did you decide when to start the FDA approved enzyme replacement therapy for Fabry that involves getting the medication through an IV twice a month? My son's test results left no doubt that he needed to start enzyme replacement therapy (ERT) at an early age. My goal is to get him the best care. For myself, I am working closely with my doctors, and when my results start showing symptoms, I will consider treatment as well.

What do you wish that you knew 15 years ago about Fabry disease? I think God's timing for our dad to be diagnosed was in His perfect time. Although my dad suffers from not being diagnosed earlier, my son received his diagnosis to receive ERT at a great time.

What do you think is important for people living on dialysis to know about Fabry disease? You can still live an active life being on dialysis. My dad still travels and functions fairly well.

Any other words of wisdom about living with Fabry disease? For a long time, I questioned every health concern as if it might be Fabry-related because there are no definite answers for a woman living with this disease. But now I understand that there are many things that may never be understood, and I'm okay with that. I try to eat healthy and stay active and if things do come up that I am concerned about, I seek the necessary answers and don't worry about it being related.

Any other words of wisdom about being a mother of a child living with Fabry disease? I treat my son as I do my other two children. He wouldn't want it any other way. It is hard thinking about your children having to live on this drug for the rest of their lives, but when you look around at many other children who have it far worse, we are thankful that he lives a fairly normal life that is only interrupted for a few hours every other week.