Alicia

Acute Hepatic Porphyria Ambassador

Alicia, AHP Patient and British Porphyria Association Volunteer

Have you ever heard the saying "If I knew then what I know now, oh how I would have changed things?" But what happens when the choice was not yours to make because you just did not have all the information, and you are left to deal with the consequences for the rest of your life?

When I was 19, seemingly out of nowhere, my health took a drastic turn. A simple chest infection that was treated with medication was a catalyst for tummy pains, sickness, and extreme fatigue. My health continued to get worse—I started having problems with my breathing and the pain in my tummy was crippling. One day while at work, I was rushed to the hospital, where I remained for 10 days. I was out of it, confused, hallucinating, unable to eat or sleep, and crying in pain.

No one knew what was wrong. No one knew that hiding inside my genetic history lurked a disease that would change my life.

Somewhere during the chaos, it was discovered that my dad's mum had a disease called porphyria. Porphyria...What's porphyria? No one at the hospital had been looking at porphyria as a possible cause of my severe symptoms, but they were desperately trying to help me.

"If I'd only been aware of my genetic predisposition for this disease, I could have done things differently."

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Acute Hepatic Porphyria

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While reviewing my medical history records, there it was, a brief reference buried in the natal screening notes—dad's mum, porphyria. I'd not been in contact with my dad's side of the family for many years, nor had my mum. Neither of us knew it was listed there because we'd never had a reason to check. It was extremely fortunate that it had been added to my medical history at all. I cannot begin to imagine what the outcome might have been if they hadn't noticed it.

Fortunately, we found a specialist who understood acute hepatic porphyria (AHP). He explained that my symptoms were a reaction to the build-up of toxins caused by a gene change (mutation)—my liver wasn't producing a certain enzyme. I was tested and within two days I had a specific diagnosis of AHP and a treatment plan. It was explained to us that it was very likely the medication I'd been prescribed for my chest condition had triggered my first attack. If it had been clear that I had a genetic predisposition to porphyria, my first attack might have been delayed or may have never happened.

This brings me to my family. While my dad was rarely in the picture, my family means everything to me. As I learned more about my AHP, as well as how it had been a part of my dad's family line, I realized my half-sister should be informed. I wanted to protect her. I encouraged her to have the genetic testing done, even though she'd had no symptoms. Genetic testing confirmed that she does have the gene and is therefore at risk of potentially developing AHP symptoms. If something good can come out of all of this, it is that my half-sister and our future generations will be tested as early as possible. Knowing the triggers can help keep the ball from rolling because once it does, it is hard to stop. Now that my half-sister has good information, she'll have a better chance of preventing her first attack, unlike I did, and will hopefully never be forced to endure the pain I have grown so used to.

In the end, it was actually a small reference buried in my earliest medical records from when I was a baby that led to my diagnosis. If I'd only been aware of my genetic potential for this disease, I could have done things differently. Maybe it was not soon enough for me, but at least I have been able to help others. At least knowing what I know now and sharing where I can, I might help my half-sister and others before it's too late.

AHP: Acute Hepatic Porphyria

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Click here for more information on “Genetic Testing for AHP”