Interview with Mrs. Brandi Brown

Mother of a daughter with ARSACS

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Who are you and where do you live?

My name is Brandi and I live in the state of Vermont in the United States of America. However, I was born in the southern geographic portion of the United States, as was my spouse, and our families have lived in that area of the country for many generations. We have no family or ancestors from Quebec.

What is your relationship with ARSACS?

In 2005 my daughter's pediatrician noted that she had an unusual gait and that she walked on her toes. From there we began what would become a 13-year journey to figure out what medical condition was affecting our daughter. We learned a few years later that she had atrophy in her cerebellum, particularly in her vermis. She underwent significant numbers of tests over the years, as the number of symptoms she experienced increased, with consideration being given to many different genetic disorders, including Friedreich's ataxia, Charcot Marie Tooth, and mitochondrial disease. Finally, in 2017 an ataxia panel was ordered by a new physician and they discovered that she had two variants in her SACS gene. After that, to determine whether we could confirm an ARSACS diagnosis, we consulted many different experts, thanks in large part to the help from Sonia Gobeil from the Ataxia Charlevoix-Saguenay Foundation and the assistance of our genetic counselor. We also asked our daughter's optometrist to administer an Optical Coherence Tomography (OCT) test, which determined that she had significant nerve fiber layer thickening. Subsequent genetic testing revealed that one variant came from me and one variant came from my spouse. No one in our families had ever had ARSACS and, until we did social media searches, we did not know anyone else in the United States with this condition. We now know several families who are affected, and we are working to create a community with those other families through use of social media.

How did you react to your daughter's diagnostic?

Honestly, I was a bit relieved at receiving the diagnosis. There was no doubt that my daughter had a genetic disorder that was progressive. We had known it for many years, without being able to determine exactly what it was and therefore what the future held. More importantly, not knowing

a diagnosis hampered us from being able to actively seek a cure or an effective treatment. Getting the diagnosis has now allowed us to direct our energy entirely towards care and, hopefully one day, a cure. For our daughter, the diagnosis was a relief as well. She could finally put a name on what was affecting her, so that she could share that information with her friends and loved ones. As a teenager, it was very hard for her to face the challenges she did without knowing why she had them.

If you have any advice to a parent whose child is affected with ARSACS, what would it be?

Don't remain discouraged or afraid, even if you are at first. Take all the negative emotions that getting a diagnosis could create and channel it into something positive. Become an advocate, become involved, share advice, become a storyteller. Tell your story so that other parents with children with neurodegenerative disorders can learn from it. There are many people out there who are trying to find answers. And there are many people out there who are going through a similar experience to you. Form a community and a support group. Make sure your child knows that they are not alone. Above all, do not ever give up hope.