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Advocating for Acid Sphingomyelinase Deficiency: A Rare Disease Story

JAMES MOORE | November 5, 2020

Purnell was Taylor and Sam Sabky's first child, and they couldn't have been more excited for his arrival and the pregnancy and delivery happened without any major issues. For the first six months of life, Purnell developed healthily and normally. However, Purnell suddenly began to have difficulty with sitting upright, a significant milestone in development. When his doctor noticed that his liver was enlarged, Purnell began to undergo genetic testing. The family was left in a lurch of uncertainty for months. However, they would soon start getting answers when they took him to see Dr. Melissa Wasserstein. Ultimately, he would be diagnosed with Niemann-Pick disease type A, also known as acid sphingomyelinase deficiency (ASMD).

> *"We got the diagnosis the Friday before Mother's Day… not wonderful timing. By then, Purnell was starting to have trouble with solid foods."*

About Acid Sphingomyelinase Deficiency

Acid sphingomyelinase deficiency (ASMD) was previously thought to be two diseases: Niemann-Pick disease type A and Niemann-Pick disease type B. More recent research has determined that these two illnesses were in fact extreme spectrums of a single illness. What was previously known as type A is a very severe disease that begins causing

problems in infancy; patients experience failure to thrive, jaundice, enlarged liver, and progressive degeneration of the nervous system. Most patients don't live beyond 3 years of age. What was previously called type B is less severe. This form can present anywhere from early childhood to later on in life. Symptoms include enlarged liver and spleen, problems with lung function, lung infections, inhibited growth, low platelet count, and abnormal cholesterol and lipid levels. Unlike type A, the central nervous system is usually spared in type B. Nevertheless, life-threatening complications still appear. There are no disease-altering therapies available for ASMD. To learn more about this disease, click here.

Purnell continued to progress, but the Sabky's had few options available as there is no known treatment for Niemann-Pick disease type A. There were drugs being tested in trials but what impact they would have on him was unclear. Additionally, there was no way that they would get approved in Purnell's limited lifetime.

However, Taylor and Sam weren't just going to give up and began searching for any possible options to intervene. She eventually connected with the Wylder Nation Foundation, a nonprofit dedicated to engaging patient families and finding treatments for Niemann-Pick disease and other lysosomal storage disorders. Taylor learned about a potential gene therapy that could help Purnell in time; however, there was a lack of funding.

The family was able to raise $750,000 on GoFundMe for the therapy, but it wasn't ready in time and is still being developed. However, Taylor is thankful that they got involved and helped make progress that could one day save lives.

"They encountered some challenges with the gene therapy that they are still working on but the money we raised potentially allowed to them uncover this earlier than if they had to wait for traditional funding sources."

Purnell was treated with another experimental drug that seemed to slow the progression of his disease. He was monitored on a quarterly basis, which allowed a detailed understanding of how the disease could progress.

During this time, Taylor started a Facebook page which wound up being a major source of support for the family and they have continued updating the page to keep up with their network and to spread awareness and advocacy.

"It has been very therapeutic in a way to keep it going and continue Purnell's legacy."

Taylor says that while Sam and her took a proactive approach in an attempt to extend or save Purnell's life, she understands that not all families would necessarily want to take the same path:

> *"It really depends on what they want and what they are capable of. We had to jump through a lot of hoops in order to get help for Purnell and it wasn't easy…we had to get through a lot of difficulties with health care providers and insurance companies. One of my goals in advocacy is to try to make those aspects easier for families."*

Taylor says that one of the most critical aspects of the entire situation was having a network of support and reaching out to organizations like the National Niemann-Pick Disease Foundation for information and opportunities to connect with other families.

> *"I can't stress enough the value of having support from other people. For us that support was very public with the Facebook page. Don't be afraid to share your story."*

Check out the Facebook page here.

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