patientworthy®
RARE PATIENT NEWS. WELL DONE.

Dr. Justin Hopkin: Doctor, Parent, and Rare Disease Advocate

JAMES MOORE | OCTOBER 19, 2020

Dr. Justin Hopkin's son Garrett was born happy and healthy, but when he was around four to six months old, his parents began to notice some problems. Garrett was having trouble with feeding and was starting to miss important developmental milestones. He was treated for ear infection and acid reflux but didn't show signs of improvement; at his one-year checkup, Garrett's spleen was enlarged. **Due to abnormal platelet counts, doctors first checked for signs of cancer, but one of them had a hunch that the baby might have a rare lysosomal storage disease.**

After running a series of tests, Garrett was eventually diagnosed a few months later with acid sphingomyelinase deficiency. The combination of Garrett's more severe symptoms and informed doctors meant that the family got a correct diagnosis much more quickly than other rare disease patients. **Since then, Dr. Hopkin chose to take action and get involved with the rare community as a patient advocate.**

About Acid Sphingomyelinase Deficiency (ASMD)

Acid sphingomyelinase deficiency (ASMD) was previously thought to be two diseases: Niemann-Pick disease type A and Niemann-Pick disease type B. However, Dr. Hopkin says that 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**.

Seeking Community

For the first couple of years following the diagnosis, the Hopkin family did not make much effort to get involved in the broader Niemann-Pick and ASMD community. They were focused on caring for Garrett and getting a better understanding of how the disease could impact his future. **Things began to change when the family attended a gala-style event intended to raise money for research.**

Justin was impressed by the event, which had been organized by another patient family. **The experience helped him realize that he had the ability to make a difference, and he was motivated to start taking action.** Soon after, he was invited to become part of the board at the National Niemann-Pick Disease Foundation.

> *"There is an entire community of patients and families that is really incredible in trying to help bring treatments and therapies to their loved ones, and they also support each other throughout the entire disease process." – Dr. Justin Hopkin*

The family moved once Garrett, who is now ten years old, was able to get involved in an ASMD clinical trial.

> *"We have six FDA-approved Niemann-Pick clinical trials going on right now in the US. In the ASMD space we have one clinical trial. That study is evaluating an enzyme replacement therapy…it's an really exciting and hopeful time in the community."*

The National Niemann-Pick Disease Foundation has been around for around three decades and Dr. Hopkin credits the efforts of the original families that started the organization for a lot of the progress that's been made.

> *"It's an incredibly passionate and active community. They've really helped move the needle so that we're close to having access to therapies that can impact the course of these terrible diseases."*

Click here to learn more about Dr. Hopkin's story.

Originally Published: https://patientworthy.com/2020/10/19/dr-justin-hopkin-doctor-parent-and-rare-disease-advocate/