To Help and Be Helped

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It was in May 2017, two days before Mother's Day, shortly after her son Purnell's first birthday, that Taylor Sabky found out the boy had the ultra-rare disease Niemann-Pick type A.

When a doctor delivered his diagnosis, Sabky and her husband Sam were told that while there were therapies in development, none would be approved in time to treat Nell.

"We were told that it's extremely severe, uniformly fatal, and that there is no treatment for type A, and that no treatment would be available in Purnell's lifetime," Sabky said.

Nell began missing milestones at six months. He was unable to sit independently, but when a pediatrician noticed his liver was enlarged, he was sent to a specialist.

Niemann-Pick is a lysosomal storage disorder. The lack of an enzyme needed to break down fats in the body causes deposits to accumulate in cells throughout the body and cause progressive

damage. It can cause the liver or spleen to become enlarged, and create difficulties with motor skills, speech, swallowing and feeding, as well as cognitive impairment.

As the condition progressed in the case of Nell, he eventually needed support from a feeding tube and oxygen tank. As Sabky reached out to other families involved in the world of Niemann-Pick disease, she learned of therapies in development.

Sabky became a patient advocate after learning of a potential gene therapy that was being developed that might be able to treat Nell. She launched a GoFundMe campaign and a Facebook page and told her family's story.

"We can't do anything about the science, but we certainly can raise funds," she said. "And even though it was an extremely large amount of money, we said, 'This is our shot. Let's go for it.'"

She and her husband raised $750,000 to help advance the gene therapy to human clinical trials, but unfortunately delays prevented Nell from ever benefitting from it. But she also found that sharing her family's story prevented the isolation she felt from having a child with a terminal disease take too deep a hold on her. She felt support from others as they learned what she was going through and offered their help.

Though the family managed to get access to experimental therapies in development, one of which provided some benefit and appeared to extend Nell's life, the disease slowly robbed him of his abilities and he died at the end of 2019.

It would be easy to understand why a parent who lost a child to a rare disease would choose to move on with their lives and choose to stop living in the world that came to dominate so much of their daily lives, but like many others, Sabky didn't do that.

Perhaps it is part of the healing process. Perhaps it is knowing how much it means to those who come after of having someone who understands what they are going through and pay that forward. Perhaps it's a way of keeping Nell alive and seeing some good come from what they went through.

Sabky continues to maintain the Facebook page she created and expects to increase her advocacy work to address the difficulties parents of children with conditions like Niemann-Pick type A face in getting access to needed services and medical equipment.

In a recent post on the Sakby's SavePurnellToday Facebook page she posted a picture of notes and letters she has received. She spoke to why she continues to work to help other families with Niemann-Pick type A babies and what she has learned.

"When you help others, you receive a kind of help in return. This is evident as we revisit notes from members of Nell's circle of care—doctors, nurses, hospital staff, clinical research staff, home service coordinators, health care professionals, other rare families," she wrote in the post. "In their messages they reveal we were helping each other all along. One of my favorite parts of Nell's legacy is the glimpse he gave us into the goodness of humanity. Let us all continue to help and be helped."

Photo: Purnell Sabky with his parents

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