

# Why Awareness Days Matter to Me as a Parent of a Kid With a Rare Disease



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If you're on social media, you may have noticed that October is a very popular month to raise awareness. There are messages, ribbons, profile picture frames, and appeals all seeking to raise awareness for various issues or illnesses. Everything from pregnancy/ infant loss and domestic violence to breast cancer and liver disease. Tennessee even recognizes October as Elephant Awareness month. For our family, October means Niemann-Pick Disease Awareness month. There are three commonly recognized types of Niemann-Pick disease, and for our family, we especially know Niemann-Pick Type C (NPC). NPC only occurs in about one in every 100,000–120,000 live births, making it an ultra-rare disease. It is genetic, neurodegenerative, progressive and fatal. Currently, there is no cure.

For us, the one in 120,000 is our 6-year old son, Liam. He is a bright, goofy child with an old soul. He's a brave warrior who has had this insidious disease since before he was even born. He's undergone a liver transplant as well as roughly 13 other surgeries and receives invasive lumbar punctures every other week. He pushes himself in physical and occupational therapy even though it leaves him exhausted. Our son is fighting NPC every second of every day as it tries to take his life. Every October we join with the National Niemann Pick Disease Foundation, the Ara Parseghian Medical Research Fund, private family foundations, and the members of this strong and determined community to help raise awareness about this rare disease.

In the midst of it all — the social media posts, the speaking engagements, the ribbons, the cause bracelets and the fundraisers, I realize that it's not just about raising awareness, but being clear on why awareness is so important. Why do we share our deeply personal stories with the masses? Why do we seek to educate our families, friends, doctors, researchers, pharmaceutical companies, and even complete strangers? For our family, raising awareness is about earlier diagnosis, encouraging more research, raising money to fund that research, and decreasing the isolation that often comes with the Niemann-Pick diagnosis.

There are over 7,000 rare diseases and an estimated 25 million Americans have some form of a rare disease. It would be naïve to believe that any one doctor would know the symptoms of all

7,000 of those diseases and be able to accurately diagnose every patient right away. For an individual with NPC the average time to diagnosis is five years. That's five years of countless specialist visits, unanswered questions, batteries of tests, misdiagnoses, and constant worry. That's five precious years not exploring treatment options or participating in clinical trials, years not spent meeting other families that can help to support and guide you through the challenge that is rare disease. Awareness means that doctors recognize that jaundice and an enlarged spleen in an infant, a teenager with symptoms of bipolar disorder, or an adult that has difficulty walking and slurred speech could all have NPC.

Awareness means that a parent who is desperately searching for answers on why their child is losing abilities they once had may be told to research NPC because their friend saw a social media post or a story on the news about another child with similar struggles. Raising awareness means getting states to include NPC as part of their newborn screening panels so that life extending treatments can be started immediately and not five or 10 years from now.

Although Niemann-Pick disease is a difficult diagnosis, the families who face it are not without hope. There are promising clinical trials and research is advancing every day. Studies are being conducted and advances are being made in gene therapy. There's no cure yet, but it feels like we're standing on the precipice of one. It's like the old carnival game where there are a small fortune of coins resting on a ledge and each coin that you add to the machine gets added to the pile. You don't know when that tiny fortune will rain down, but with each coin you add to the machine your chances of pushing it over the edge increase. Raising awareness for NPC means gaining the attention of more researchers and clinicians and attracting more money through fundraising efforts. I see each and every penny that's donated, each research project started, as one more coin pushing that pile that will eventually bring us the jackpot of a lifetime — a cure.

Awareness also means breaking through the isolation that can come for those with a rare disease and for their caregivers. It means understanding children and young adults that live with NPC because not enough people know what the disease looks like or the effects it has. It's acknowledging the isolation that young kids may feel when they're not able to keep up with their peers on the playground or for young adults who start to lose their friends as they lose their ability to attend social functions. It's about the isolation for caregivers who can't leave the house as often as they used to or who have to spend weeks on end in the hospital.

As each family's journey is different. I can only speak on behalf of ours when I say that sharing our personal experiences with rare disease can be difficult and uncomfortable at times. But we do it because awareness is so incredibly important. We do it so that our son and those like him may be better understood. We do it so that a parent desperately seeking answers may find them and we do it because we so desperately want to push those coins over the edge and find a cure.

This October, or any of the other 11 months of the year, if you see a friend or acquaintance seeking to raise awareness, support them. You can help by simply asking questions to learn more, looking up information online, sharing their social media post or donating to their fundraiser. You never know — you might be the one that helps to break their isolation, that recognizes the symptoms in another individual or whose \$25 donation helps a family struggling to buy medical supplies. You can be the coin that helps to push that jackpot over the edge.