Parenting a child with a rare disease

Raising a child with a rare disease can feel overwhelming and isolating, but many parents end up being extremely proud of their children’s achievements.

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When Steven Striegel was three months old, his parents noticed he was having spasms. “To us, it looked like he was doing sit-ups. And to the tune of maybe 100 or 200 a day,” said Steven’s father, Scott Striegel. “Within a day or two we figured out something was wrong so we took him to our pediatrician who had been a pediatric neurologist.”

It was determined that Steven had tuberous sclerosis complex (TSC), a rare genetic disorder that may cause potentially life-threatening non-cancerous tumors to grow in major parts of the body like the kidneys, skin, heart and brain and can also result in epilepsy or developmental delays. TSC is estimated to affect 1 million people worldwide. “When Steven first got diagnosed with TSC we were relieved because we had an answer to his infantile spasms,” said Mandy Striegel, his mother. “It was scary because we didn’t know anything about TSC.”

Scott admits he struggled in the beginning. The fact that TSC is so rare and most doctors didn’t know how to treat it added to his stress. “You don't have a direction to turn to because you don't know what to do to fix it. The people that are the experts are telling you they don't know anything about it other than this is what it is,” he said. “Where do you go from there? It's kind of an empty feeling.”

Ryan Groves, another individual living with TSC, is the youngest of three sons, and his parents felt overwhelmed initially as well. “The whole family was impacted with Ryan's diagnosis, you know, first with just the shock and the uncertainty of what the disease would do to him and how it would affect his life,” said Ryan’s father, Bob Groves. “Because he was diagnosed at about a year old, we didn't know developmentally what it would mean.” Ryan’s diagnosis came with added challenge of being pre-Internet, so the Groves had limited research to work with, beyond what doctors told them.

The symptoms of TSC often change over time. As a kid, Ryan experienced some behavioral issues and neurological symptoms such as seizures that can impact those with TSC. Now at age 27, Ryan is impacted by non-cancerous kidney tumors associated with the disease, which are regularly monitored by his physicians. When he was 17, Ryan started participating in the Special Olympics and competed in the National Special Olympics in 2010. In 2013, he won gold for the softball throw at the Michigan State Special Olympics.

Steven, now 20, still goes to the doctor a lot to ensure his condition is regularly monitored, but also enjoys playing video games and spending time with his family. Last November he took part in the Greater Fort Worth Special Olympics Bowling Competition in Texas. “It means a lot to me when I'm bowling. I just keep an eye on my own lane, get the best score and win,” he said. “I practice a lot every Saturday.”

Today both the Streigels and the Groves also focus on their sons’ independence and happiness. “Our goal is that hopefully before he is 30, he will be able to be in his own apartment with support,” said Kathy Groves. “We spend time teaching him to do laundry and to cook and to clean, and I can safely say he cleans his room as well as any 27-year-old male does so.”

Mandy Streigel says she is incredibly proud of how her son has handled living with TSC. “Steven inspires me a great deal because he does not let anything get him down. He's always happy. He always has a smile on his face,” she said. “He does not let his diagnosis with TSC and the threat of daily seizures get in the way of him enjoying life and doing the things that he likes.”

Kathy also focuses on the positive. “My hopes and dreams for Ryan are the same as they are for my older sons,” she said. “That he is successful in his life, that he has a level of independence that matches his skills and abilities, and that he is contributing to society. Most importantly is that he finds enjoyment and happiness in his life in some way every day.”

Although a diagnosis of a child’s rare disease can be frightening for parents, many of them have a wiser outlook on life because of it. “If I had a message to send to anybody that had someone diagnosed with TSC, it would be to always keep looking forward. It opens up avenues to things that you never expect,” said Scott. “Steven taught me patience.”

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