

# Two families, united by a rare disease, push for awareness and a cure

'She may have a disabled body, but I refuse for her to have a disabled spirit,' one mom says



By [Dana Hedgpeth](#)

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Three years ago, Katie Gregg's maternal instinct told her something was amiss. Her chubby-cheeked, blue-eyed daughter Lilly wasn't hitting all her milestones as an infant. Sitting up was hard. She'd topple over when crawling, and as she got closer to 1½ years old, she'd pull up on furniture but not take steps.

Doctors told her and her husband, who live in Oakton, not to worry and advised them to wait, saying she would develop. Lilly underwent MRIs, and one neurologist suspected she had cerebral palsy. But Gregg wasn't convinced. She pushed for genetic testing. The results: Lilly has a rare genetic disease called hereditary spastic paraplegia (HSP) that affects her mobility, causing abnormal tone, stiffness and paralysis in the muscles of her legs. The disease is likely to get worse, moving into her upper limbs and possibly affecting her speech and cognition. There is no treatment and no cure.

The geneticist also told them that a few miles from their home, another girl close to Lilly's age had recently been diagnosed with the same gene mutation. After getting permission from both families, the geneticist connected the Greggs with the Loreks and their daughter Blair.

"Getting a life-altering diagnosis for your child was super scary, but then it was also unbelievable to learn there was another child with the same diagnosis nearby," said Gregg, 37.

The Loreks, too, had gone through an untold number of doctors' appointments, scans and other tests before Blair received her diagnosis.

The two moms connected by email in August 2020, and amid pandemic caution, the families met a month later on a Zoom call. They became fast friends. Both dads like to golf, the moms are avid runners, and they have other children who are close in age.

But above all, it was the coincidence of their experience around their daughters' disease — as well as the serendipitous proximity of their homes — that truly bonded them. And they desperately wanted to raise awareness about their daughters' specific type of HSP and support research to find treatment and a cure. So the two families in January established the [Lilly and Blair Foundation](#).

The girls' specific gene mutation — known as de novo SPG4 — is extremely rare. "De novo" indicates that Lilly and Blair, who are now 4 and 5 years old, respectively, are the first in their families to have the SPG4 mutation. More commonly, SPG4 is inherited. Because SPG4 affects so few people, experts said, pharmaceutical companies typically don't want to invest in the long, expensive process of developing and testing drugs or other devices to help those who have it.

"They have an ultra-rare disease, and that puts you in a very lonely place," said Darius Ebrahimi-Fakhari, a pediatric neurologist at Boston Children's Hospital who works with Blair and Lilly. "There's not much known. There's not a lot of research done. There's not a community of people that go through this who have advice to share."

Enter the Loreks and the Greggs: Foundations such as theirs are often the best way to jump-start research and drug testing for such rare diseases, experts said.

Chris Lorek, Blair's dad, said he feels a sense of urgency to advocate for the girls in finding a cure: "I want to give my daughter the best chance at living the fullest life possible."

For the two families, having each other has given them an emotional support system. They share their fears, tips and triumphs in navigating their daughters' conditions. They talk about the latest research, the daily therapy and doctors' appointments, the expensive equipment, as well as the battles with health insurance companies.

"There is nobody in the world who could get it more than them," Gregg said of the Loreks. "They're in the exact same situation as us, and to have them living down the road is incredible."

Both girls need to be lifted in and out of the wheelchairs and walkers they use at home and school. Blair needs help getting on the floor to sit cross-legged to play. And Lilly's mom works with her on exercises several times a day to deal with the tightness in her leg muscles — which sometimes, Gregg said, are "like an iron rod."

The Greggs and Loreks also discuss how they balance their careers and other children.

Gregg, who left her marketing job about a year and a half ago to take care of her two children, sometimes feels like she “should be out kicking a soccer ball with my son, but I can’t leave Lilly’s side.” Her husband, Michael, 36, works as a development manager for Peterson Companies, a real estate firm in Northern Virginia.

Both families have adjusted their homes for their girls. The Greggs lowered a sink, toilet and clothing rack in Lilly’s bedroom so she can brush her teeth and pick out her own outfits. Some rooms in their home are sparsely furnished so Lilly can get around in her stand-up wheelchair, and there are padded floor mats so she can also crawl.

The Loreks — Stephanie, 42, who works as a senior policy analyst for the Federal Deposit Insurance Corporation, and Chris, 41, a senior account executive for Amazon Web Services — have three children and moved last year to Ashburn so they could have a larger home where an elevator could be installed if needed. For now, Chris carries Blair up and down the stairs.

Several times a year, both families pack up and go to Richmond so Blair and Lilly can do a few weeks of intensive, daily physical and occupational therapy at a specialized center. Although both families have insurance, they spend up to tens of thousands of dollars a year in out-of-pocket medical costs.

Sometimes, the moms get sad about how they have to carry their daughters to the top of slides at parks while also playing with their other kids. And they commiserate over the declined birthday party invitations to bounce houses and climbing walls because their girls can’t participate.

Both families worry about how fast and when the disease will progress. Still, they’re learning from their “warrior” daughters’ strength, determination and upbeat spirits.

Blair — who’s called “Blair Bear” by friends, family and teachers — is said to be the happiest kid in her class. She likes her horse and swim therapy sessions, and she enjoys going outside and having someone collect leaves or rocks with her.

“Whenever we feel challenged or it’s tough going and then we see Blair smile, it clears everything away,” said Blair’s dad, Chris. “Her happiness is independent of her abilities.”

Her mom, Stephanie, said she sometimes wishes she could “be Blair for a day” just to “know how hard it is for her.” But when she watches her daughter, she realizes “she doesn’t know any other way.”

“She’s a happy, easygoing kid,” Stephanie said. “She has a great disposition. She never complains.”

Lilly, who’s kind, determined and opinionated, loves to sing, dance and bake with her little brother. As she has gotten older, she has become more aware of her challenges. She recently told her mom, “When I grow up, I’m going to walk,” Katie Gregg said. Neither will her family be discouraged.

“She may have a disabled body,” Gregg said, “but I refuse for her to have a disabled spirit.”