Family of Young Resident Fights to Cure Rare Disorder

Doug and Kasey Woleben moved from North Dallas to Stonebridge Ranch in 2012 when their son, Will, was almost a year old. The young family settled into a craftsman-style house on a quiet cul-de-sac in the village of Freedom Park. Says Kasey, “We moved because we wanted a bigger house for our kids to grow up in. And we wanted a community with a family atmosphere and good schools. When we found Stonebridge Ranch, we knew this was home.”

Better yet, all the houses in their cul-de-sac—save one—were filled with young, friendly families like their own. The only exception happens to be the home of retirees Chuck and Barbara Hoffman, who act as de facto grandparents for all the cul-de-sac kids. “Everyone calls them Grandma Barb and Poppa Chuck,” Kasey says, and both of her young children smile and nod when they hear the names.

It was easy for the Wolebens to believe they’d hit the jack-pot in finding the perfect place to raise their growing family. But they had no idea just how much they’d come to depend on their neighbors, nor could they have anticipated the difficult circumstances that would bring them all closer together.

When Doug and Kasey’s second child, Lauren, was born in 2014, it was a time for celebrating new beginnings. But for some time, the young parents had felt a bit concerned about their son, Will. Though he was born healthy and had been a cheerful, active baby, as he neared age 2, he began to stumble and fall. His symptoms became so noticeable that his parents termed them “resting episodes” and began taking him from specialist to specialist to determine the cause. The doctors ruled out everything from digestive disorders to seizures and told the worried parents their child was fine.

But one afternoon when Will was barely two, Kasey rushed her son to the ER at Children's Medical Center Plano. He had fallen again and told his mom he couldn’t get up. She knew something was truly wrong by the look on his face. At the ER, a CT scan showed dark spots on Will’s brain. Suspecting a stroke, his doctors sent Will by ambulance to Children's Medical Center Dallas, where he underwent more tests, including an MRI.

The family got a definitive diagnosis later that week. A team of neurologists sat down with Doug and Kasey and showed them the MRI results and more dark spots on Will’s brain. The young parents learned then that Will has a rare genetic defect called Leigh’s Syndrome which affects approximately 1 out of every 100,000 children born worldwide. The disorder interferes with a single gene—Surf1—out of the 20,000 or so that make up the human genome.

As Doug stared back at the doctors in disbelief, he asked them what could be done to save his tiny son. “Nothing,” they said, noting that Will would eventually lose all his physical abilities. Historically, children born with Leigh’s Syndrome have rarely lived past age 10. Doug and Kasey were devastated.

Though some genes perform roles as simple as determining our eye color, the Surf1 gene is crucial to oxidative phosphorylation, the process by which our bodies’ cells convert the food we eat into energy. In Will’s case, his mitochondria are not able to perform this crucial step of energetic conversion, causing his cells to die. Will was barely past age two when the effects of the disorder started to become more pronounced. Though he remained a gregarious, curious toddler, he gradually began to lose the ability to walk, speak clearly, and even swallow.

Neither Doug nor Kasey's extended families live nearby, and the young parents quickly felt overwhelmed by the new way of life that came with Will's diagnosis. Lauren was only 7 ½ weeks old—still nursing and not sleeping through the night—when her parents first heard the words "Leigh's Syndrome". Will soon began to need more specialized care, including a feeding tube, and his parents faced the extra costs and time associated with taking him to multiple doctor and therapy appointments. Doug found himself relentlessly researching medical literature online every night, desperate to find some way to save his son from the fate described by his doctors.

It was at this critical time in the Woleben's life when their neighbor's stepped in to help. Upon learning the diagnosis, the families in their cul-de-sac set up a meal train to make sure the family had home-cooked meals. They often came over to help take care of baby Lauren so Doug and Kasey could take Will to appointments. Grandma Barb and Poppa Chuck even hosted a "Coffee on the Porch" fundraising event so the neighbors could help support the family financially, too. Says Kasey, "We'd made friends with our neighbors, but that's when they became our network, our support system. They have become our family."

In the four years since their son's diagnosis, his parents have not accepted that early pronouncement that "nothing" can be done for Will. They have chased down every lead they can to help find the cure he needs to live. The chase has kept them on a continual roller coaster of emotions. They've corresponded with multiple drug companies and flown hundreds of miles for experimental treatments. In 2014, they were able to get Will enrolled in a drug trial in Houston and drove him back and forth for the associated tests. The compound seemed to help stabilize Will's condition, and his parents were jubilant to see the result. But soon, the drug company halted the trial and pulled the drug, saying they didn't find enough evidence of efficacy.

Recalling the hard moments, Doug says, "You have to have hope. We choose to have hope." Kasey agrees: "You could, but you don't want to fall into a depression." The young mother can't help but look at her children when she says this. She watches Lauren beside her, now age four, who is intently coloring a page with neon markers. Then she turns and gives 6-year-old Will a drink of water, wiping his cheek. The mother and son grin intently at each other, twinkle-eyed.

From across the table, Doug asks his son, "What do you want to be when you grow up?" Six-year-old Will immediately arches in his wheel-chair and throws his arms in the air. His grin is so wide you can almost hear the cheer of an entire crowd behind him. "A goalie" he says with a quiet sound that his mom easily interprets as she runs her fingers through his golden hair. Notes Doug with a laugh: his son loves watching sports so much on TV, especially hockey, that he has converted his mother into a raving Pittsburgh Penguins fan. The two even have matching jerseys celebrating their favorite team. Kasey laughs out loud and says, "I know! Me? Hockey?! It's the craziest thing." Will grins back at his dad and nods his head when Doug says: "You're always the goalie, right, when we play hockey in the back yard? You always stop the puck!"

This past year has been perhaps the most hopeful for the family since Will's diagnosis. Doug and Kasey call it a miracle: of all the hospitals in all the world, doctors at UT Southwestern in Dallas are currently building what they hope will be one of the best (if not the premier) gene therapy center in the world. One faculty member in particular, Dr. Steven Gray, is intent on finding a cure for the Surf1 mutation, and he wants to be running human trials on a single-gene therapy within two years. The biggest obstacle now is finding the funds to pay the cost, which will eventually run into the millions.

Says Doug, “We’d heard about gene therapy before, but we never thought it would be ready in time to possibly save Will. We thought it could only happen many years in the future. But with the doctors at UT Southwestern, we’ve learned the science has caught up to the disease.” His voice grows more emphatic when he says: “Today, finding a cure *is* possible. And it has huge implications. Combined, genetic disorders are not rare. There are over 6,000 defined disorders, and all together, they affect more people than cancer. But it’s almost impossible to get funding for specific gene therapies because the drug companies don’t see how they can make money on them.”

He raises his hands and looks at them, and it’s easy to picture the young father sitting alone at his computer late at night, struggling with the rarity of what he and his family are facing, desperate for answers. “It does get overwhelming,” he says. “It’s the parents and the caretakers who have to deal with it on every level, from taking care of the child who has been diagnosed to fundraising to find the cure. But if we don’t do it and fight, it won’t happen. These kids deserve a chance at childhood and at life.”

Along with two other families whose children are affected by the Surf1 mutation, in 2018 the Wolebens started a nonprofit called the Cure Surf1 Foundation (https://www.curesurf1.org/). The foundation’s first mission: to raise $300,000 to fund research leading to an FDA-approved human trial of a single-gene replacement therapy for Surf1. Now that they’ve reached that first fundraising milestone, they are moving on to their overall mission to raise the $3.3 million needed to fund the research through human trails.

As they have been every step along the way, the Wolebens neighbors are in the thick of the fight with them. This September, Doug and Kasey organized a Top Golf Fundraising event with a silent auction to support the research at UT Southwestern. Grandma Barb and four other moms from their cul-de-sac provided invaluable help to make sure the event went off without a hitch. Says Kasey, “Our neighbors have been absolutely overwhelming in how hard they worked to get sponsorships and donations. They talked to so many businesses.” She adds with a laugh: “You should have seen Barbara’s house the week before. You’d walk in, and she had everything neatly organized—gift baskets on every table. All the raffle items had completely taken over her house.” Ultimately, the event netted $28,249 in donations.

The Wolebens are honest with themselves and others: they know that if Dr. Gray’s pre-clinical work does show promise, Will might not be accepted as a candidate for the human trials that could be paid for, in part, by their own fundraising. They know that time might run out for their son. But, says Kasey: “This is a risk we are absolutely willing to take. We believe this research could eventually help millions of people.” Doug agrees with a nod. “This is not just about our family. This is research that is going to change medical history.”

When asked to describe what a cure might look like for his son, Doug easily describes the process. “They will use a special virus that is safe for humans as a kind of packaging,” he says. “They’d insert a healthy copy of the Surf1 gene into the virus cells, and then they’d deliver those cells into Will’s body via a spinal tap.” Again his raises his hands, this time motioning as if he’s plunging the needle himself into his son’s spine. It’s that single moment he’s now fighting for every day of his son’s life. Kasey’s face lights up as she imagines the virus replacing the mutated gene throughout her son’s brain cells with healthy copies. “It would literally be trillions of cells,” she chimes in, “and he’d only need one shot.”

As she says wistfully, “It’s an absolute miracle,” it is impossible not imagine her young son—and many other kids like him—someday rising up…and taking a walk.