PLUS: UNITED STATES MILITARY SECTION

EXCEPTIONAL PARENT MAGAZINE
JULY 2017
$7.95

SPECIAL HOUSING FEATURES:

CREATIVE OPTIONS *for* INDEPENDENT LIVING

AND:

HOUSING *for* LOVED ONES *with* PSYCHIATRIC DISABILITIES

INSIDE:

MANAGING EXPECTATIONS *with a* HOME REMODELING PROJECT

ACCESSIBLE HOMES

The Annual Issue

PLUS: SUSANNAH ROSEN

AN URGENT NEED *for* ACCELERATED KIF1A RESEARCH

SAFETY FOR A CHILD WITH A SEVERE MOVEMENT DISORDER

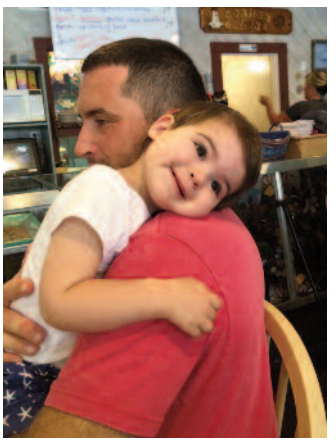
BY LUKE ROSEN

No matter how relentless we are about making our home accessible, Susannah's movement disorder is responsible for teaching her where to go when she feels the danger of her changing coordination. I'm thankful to her disease for that — for doing my job in some strange way. For somehow teaching her to be safe.

Keeping our kids safe is every parent's number one job. Even as an adult I associate protection and safety with my childhood home and my loving parents. It's a great relief when our children finally learn where to go and who to find when they're in danger. More than a relief, it's a proud parental achievement worth celebrating: "We're super parents! Our kids know where to go when they sense danger!" Mission accomplished, big exhale.

When your kid has a rare neurodegenerative disease causing her balance to worsen every day, the safety of your home is constantly being put to the test. There is no relief, there is no deep exhale. There is only constant heartache with every face-first fall into the brick wall that she once used to pull herself up.

When Susannah was a baby she would flop onto my shoulder and rest quietly for minutes at a time – it was heaven. I live for those shoulder flops. Just after her second birthday we were told this spontaneous cuddling was a symptom of her genetic condition that would eventually cause her to fall. A lot.



SAFE FOR NOW: *(Previous page)* The author with daughter Susannah as she dozes during an electroencephalogram; *(Above)* "The progressive nature of Susannah's disease might steal away her speech and steps, but she will never lose the sincere and purposeful kindness that is cause for those quiet shoulder flops."

Around the time of her diagnosis, we learned that our daughter's seemingly intentional maneuver of affection was just another implication of her increasingly low muscle tone, or hypotonia. As she grows taller and her gait becomes severely ataxic, constant tumbles are a regular part of our new normal. Not even three years old and her daily bumps and bruises resemble those of a hockey player.

I'm the luckiest guy in the world, for so many reasons. I'm blessed to have a thoughtful, funny, intelligent and kind daughter. The progressive nature of Susannah's disease might steal away her speech and steps, but she will never lose the sincere and purposeful kindness that is cause for those quiet shoulder flops. I have complete faith in science; however, there are two things I am certain medicine has wrong: that there is no cure for Susannah's condition, and that those divine shoulder flops have a clinical explanation like "low muscle tone." She flops onto my shoulder because she loves me. She's safe there, and she knows it.

This morning one of Susannah's daily acts of kindness warmed my soul to the core. I was about to walk out the front door when she extended her hand and led me into the bedroom. When we arrived at her bed, she flopped herself onto the mattress and said my favorite words, "Daddy cuddle me." Before the tears made it from the pit of my stomach up to my eyes, she pulled me onto the bed and we cuddled. One of the best cuddles ever.

PHOTOS PROVIDED BY LUKE ROSEN

As kids get older, hugs become less frequent. Not with Susannah.

NOT ALL SUPERHEROES WEAR CAPES: *(Clockwise from top left)* Susannah celebrates her third birthday; with her mom Sally at NewYork-PresbyterianChildren's Hospital; and with her brother Nat at the baseball field, wearing her braces which help her get around.

After a few minutes Susannah rolled over and said "Daddy I come to bed because it's safe." Crushing. The tears returned to the pit of my stomach and became a nauseating feeling of paternal helplessness. Is her bed the only place in our home where she won't fall and get injured? We haven't taught Susannah where to go when she feels danger, her disease has. Does she only feel safe in the bottom level of a New York City-sized bunk bed designed to keep her from falling out at night?

There's no great parental achievement here, no deep exhale celebrating the accomplishment of that essential hurdle: teaching our kids where to go when they're scared. That badge of honor doesn't belong to us because we didn't earn it, her degenerative condition did. When she's having an especially wobbly day, Susannah knows she immediately needs to return to her bed, or into my arms. The labyrinth of our living room and its old, uneven New York floors is overwhelming.

No matter how relentless we are about making our home accessible, Susannah's movement disorder is responsible for teaching her where to go when she feels the danger of her changing coordination. I'm thankful to her disease for that — for doing my job in some strange way. For somehow teaching her to be safe. But I'd like to meet KIF1A in a dark alley and destroy that monster for stealing away our daughter's sense of independence.

It's natural, as kids get older, for those precious extended hugs to become less frequent. That's one of the deliciously sad parts about watching our children grow up. Not with Susannah. Her genetic mutation is cause for one of my most coveted parts of the day. Selfish, for sure, but if Susannah flops into my arms and rests her head on my shoulder because of a rare genetic anomaly, I'll accept that. But that's not the reason. You see, my daughter is a trickster. The real reason she leads me into her bed for a safe cuddle is because she inherited her mother's beautiful tenderness. She's more generous than she is cautious. She knows how amazing it makes me feel when she flops on my shoulder. Susannah is a superhero who can fly around the room — and I thank her every single day for deciding to land on my shoulder. And for staying there.•

PLAYING THE RARE DISEASE CARD

BY LUKE ROSEN

Parenting a kid with special needs requires a lot of writing. It seems like I'm constantly writing, which is ok because... well, I'm a writer. Sitting at my desk typing a blog post is far less stress-inducing than the painful daily scribble that comes along with a rare genetic disorder.

Early Intervention requests; reminders that doses changed because of a seizure yesterday; letters to the mayor about the ramp our building is stalling on; financial aid applications; thank you notes to therapists; Christmas cards in February; invoices from the Tooth Fairy; camp registration and so on. By far the most difficult rare disease writing assignment is filling out new doctor registration forms.

I hate that damn clipboard. That pile of pages handed over by the sweet lady who always gives us an extra smile... Like so many diseases, these registration forms have a progressive course and deteriorate rapidly. Every epic page hurts even more.

Page One is harmless: Date of birth. Address. Phone number. Name of pediatrician. Emergency contact.

Page Two gets harder: Insurance. Siblings. Allergies. School... Thank god DOB was on Page One because I'm already so stressed I can't remember our daughter's birthday – Forget about her social security number.

Page Three makes me sweat profusely. The most difficult section: "Pain on a scale of one to 10."

I hope it's closer to a four today. I know it's not a one. Two would be nice but she's having tiny seizures and her eyes hurt. Last night she woke up in a spastic episode, so this morning her legs are cramped. She was vomiting so her chest is sore and she had a bad fall this morning... I don't know, seven?

If your kid has a rare disease associated with 50 different horrible conditions, you need a card. A Rare Disease Card. I am armed with a KIF1A card everywhere I go.

It's excruciating to imagine our daughter's pain. Luckily she's way stronger than me. She's a superhero. Like all superheroes, Susannah just smiles and waves at every passing baby. Sitting on the puffy, vinyl waiting room chair she looks like a movie star riding on a float in the Thanksgiving Day Parade.

Page Four: Infuriating. Two lines underneath the question "What brings you here today?" Then another two lines at the bottom for me to list "Other medical problems." TWO LINES? If your kid has a rare disease associated with 50 different horrible conditions, you need a card. A Rare Disease Card. I am armed with a KIF1A card everywhere I go. I am a card-carrying member of KIF1A.

These cards are amazing. I'm a happier person because of my card.

Secretaries don't hate me and ER visits don't take five hours thanks to my shiny new card. First Responders will save my kid's life because that card explains the neuropathy making her insensitive to pain. A laminated card (written by somebody far more eloquent and informed than I am) explains everything one needs to know about a rare genetic mutation and what to do when things get real.

It takes 15 minutes for me to briefly explain Susannah's rare disease. Usually I can't get through the conversation without crying – which adds another five minutes to my incoherent rambling. It's difficult to articulate her condition because the genetic anomaly stopping Susannah from walking is an elusive mystery without a name. Her condition is referred to as a KIF1A-related disorder caused by a mutation in her KIF1A gene. Susannah's previously unreported genetic mutation is rarer than rare.

Thank god for my KIF1A card! I love my card so much that I carry five in my wallet, 12 in my bag and Susannah always has one in her pocket. Every family member, teacher, babysitter, friend – they all have Susannah's Rare Disease Card. When I walk into a new doctor's office and that secretary hands me a clipboard, I just snap my Rare Disease Card onto the form and hand it right back. New school or camp? No problem. My consent form isn't an illegible novel – it's a prewritten baseball card stapled to the front. In the middle of the night when her body is as stiff as a board and I'm carrying her into a crowded Emergency Room, that little card lets me massage her legs instead of filling out a form.

Thanks to my Rare Disease Card, Susannah won't see the desperation in my face when I stumble through a foggy explanation of her neurodegenerative condition. Silly as it sounds, my ever-present fear goes away for one fleeting moment as I scoop her up, kiss her face and hand somebody that card. Thanks again, card.

Our incredible social worker is there for us every single day. She walked us through Medicaid and helped us get Susannah into school. She spent hours on the phone with insurance companies so we could take a breath and remember to sleep. When I have no idea what to do next (daily) she answers the call. She is our generous navigator and Susannah's relentless advocate. And she gave us a KIF1A Card. A card that protects Susannah from seeing how terrified I am whenever I talk about the disease with no name. When somebody asks what's wrong with my daughter, I say "Absolutely nothing, she's a superhero" and hand them the card •

EVERY DAY MATTERS: DR. WENDY CHUNG ADDRESSES THE URGENT NEED FOR ACCELERATED KIF1A RESEARCH

Our mission is to save the life of every child affected by KIF1A. The only way to discover treatment is to immediately accelerate research by convening the world's leading scientists in childhood genetic and neurodegenerative disorders. This collaboration will lead to treatment and increase the quality of life of those affected by KIF1A. It is vital for us to drive science, fund immediate research and find a way to stop the progressive nature of this disease.

Dr. Wendy Chung is a board certified clinical geneticist with a PhD in molecular genetics. She is director of the clinical genetics program at Columbia University and co-director of the molecular genetics diagnostics lab there. In a brief video explaining the work she and her team are doing on KIF1A, she talks about her role in its treatment and research.

"KIF1A is just one of many unfortunately rare genetic conditions. I happen to be a doctor for children and individuals with rare genetic conditions, and my job is kind of to be their Sherpa and to help them along this process to understand what club they're a member of, in other words what their underlying genetic is, and ultimately help them with the care until we get to the point of a cure," She says. "In other words, to be able to keep them as strong and as healthy as possible while, at the same time, working towards understanding what's causing their disease so we can come up with better treatments for them."

Dr. Chung emphasizes the crucial factor of time in the fight to help her patients. "It's a ticking clock in the back of my head and in the back of the head of all the families, because we do know that we have a window of opportunity that I hope is something like 5 -10 years in terms of when the time a child is diagnosed until we can really be able to stabilize this and give them meaningful life, but every day matters, and that's why we're working so hard to be able to come up with a cure for this."

View the video at: http://www.kif1a.org/research.html