KCNT1 EPILEPSY FOUNDATION

Honoring Our Founders Five Years Later

Samantha MacMechan

Every parent of a child with a rare disease knows the conflux of emotions that accompanies diagnosis: shock, disbelief, fear, confusion and so much more.

“Sitting in that hospital, receiving that diagnosis and not knowing what to do with that information, “ Samantha MacMechan recalls of her daughter Charlotte’s KCNT1-related epilepsy diagnosis. “And, for me being a first-time mom, receiving that diagnosis and feeling like the world was going to end.

“But then saying how do we get through this? How do we go one day at a time, one hour at a time, and get through this? How do we take this horrible situation and turn into something beautiful that is going to inspire and help others. How do we create a legacy for our children who have received this devastating diagnosis?”

For Samantha, the answer to those questions was connecting with other parents and caregivers, including Seth and Susan Greenblott, to build the KCNT1 Epilepsy Foundation.

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“[I recall] feeling like there was nobody else out there going through this. And, then realizing that there were and that we needed to connect,” she adds. “A treatment. That’s what we all wanted but we knew the first step to getting there was connecting with other families. We’re not going to further clinical research without connecting, connecting with other families and creating community.”

While the impetus for building community was finding a treatment, sharing information, resources and support has not only improved our children’s quality of life, it has been a source of hope for parents and caregivers.

That support system, just to know there is someone out there, someone with accurate information,” Samantha says. “It was about creating hope for our community, for myself, hope for my daughter.”

In addition to being Charlotte’s mom, Samantha works as an executive with an international skin care company and credits her business acumen for providing a roadmap for the Foundation and complementing the legal and medical expertise of other founders.

“My career helped me understand how we needed to organize things—taking these pieces and determining how we take this to the next level, who we need to be meeting with, who needs to be at the table,” she explains. “I was able to lean into those skill sets and use them to our advantage.”eans more to these families than they could ever know.”

The Foundation's digital natural history study—a preplanned observational study intended to track the course of the disease—is among Samantha’s proudest moments: “Realizing how truly connected we were and how many parents were now coming to the table saying ‘I want to be a part of this.’ Being able to pull that off with no attrition. It demonstrates our commitment.”

More recently, she participated in the KCNT1-related epilepsy FDA Patient Listening Session organized by the Foundation. “The FDA Patient Listening Session was a very proud moment,” she says. “We’re being recognized. This little group of individuals whose children have an ultra rare disease have a voice and seat at the table.”

As proud as Samantha is of the Foundation and KCNT1 community, she is also open about the reality of this disease. “It’s tough because others have lost children. My work with the Foundation has made that more of a reality,” she explains. “It makes us realize how important this work is. It’s beautiful, it’s messy, it’s challenging, it's fun, and it’s inspiring. It’s given purpose in something far bigger than me, far bigger than all of us. I think it saved me from where I was in that [initial diagnosis] moment.

There are things we go through on this journey that no one can prepare you for, she adds. “Charlotte makes me better. This community makes me better. I am a better person because I have been exposed to this and, while it’s hard—hard is the greatest understatement—we are more resilient than we realize, we're stronger, we are more capable than we realize. We can truly make a difference.”

What does Samantha want supporters to know?

“That a small group can be mighty. A small group can move mountains. That this is an incredible community and these kids deserve a chance.”

She adds, “These kids are incredible. They are inspiring and they are amazing. What they go through everyday breaks my heart but it also inspires me. It is the fuel that lights my fire and it can light others’ too… If they only knew, if they only saw they are part of something so special, so rare, so incredible. Their support means more to these families than they could ever know.”