Pediatric RESEARCH

INSIGHTS

Family reflections: tuberous sclerosis complex (TSC)

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My name is Shelly Meitzler, and I am the mom of three amazing children. Two of my three children have tuberous sclerosis complex (TSC). Ashlin is 19 years and Mason is 6 years. Because of recent research discoveries, their hopes for the future are entirely different.

Ashlin experienced her first seizure at 4 months old. At the local hospital, a computed tomographic scan was done and "spots" were found on her brain. I was accused of Shaken Baby Syndrome. In the local medical community, there was no other possibility, even though further testing showed no indication of abuse. She was ripped out of my custody, and I demanded a second opinion since I instinctively knew something was seriously wrong with my daughter.

Four agonizing weeks later, a second opinion at a children's hospital confirmed with a magnetic resonance imaging of the brain that Ashlin had TSC. The doctor who properly diagnosed Ashlin was nothing short of a miracle to my family. Further testing showed that Ashlin had rhabdomyomas in her heart and a wood lamp examination showed the hypopigmented spots and a shagreen patch. Ironically, those spots were noticed the morning of her first seizure at her pediatrician appointment and were dismissed as birth mark. I took Ashlin home with some printouts on TSC and two prescriptions for seizure medication and Diastat. I was angry, scared, heartbroken, isolated, and struggled to understand what all of this meant for the future of my daughter.

I felt defeated as multiple seizures, hospitalizations, life flights, countless failed medications, and endless testing dictated daily life. Status epilepticus when Ashlin was 2 1/2 years old ripped away a piece of my child forever. She came home after a 10-day hospital stay with right-sided paralysis, no vocabulary, the inability to feed herself, sit up, crawl, or walk. She now sees 10 different specialists, receives in-home therapy 5 days a week, will require assisted care for the duration of her life, and takes 18 doses of 7 different medications to treat the varying manifestations of TSC. Ashlin has involvement in her brain, eyes, heart, kidneys and liver. She takes Afinitor to stabilize the growth of the angiomyolipomas in her kidneys. She has autism, a mood disorder, attention-deficit hyperactivity disorder (ADHD), intellectual disabilities, language delays, and right-side hemi paralysis.

Mason was diagnosed with TSC in April 2014 at 7 months old. The reoccurrence of TSC in my family is due to germline mosaicism. He was promptly enrolled in a research study at Boston Children's Hospital, which has been invaluable to Mason's developmental progress. When his infantile spasms began, vigabatrin was started within 6 days, and he did not experience another infantile spasm. Mason experienced status epilepticus in March 2015 and required so much rescue medication, a code blue was called to resuscitate him. Fortunately, he recovered with no major setbacks. We added an additional seizure medication and I am pleased to report today that Mason takes one medication twice a day and has maintained seizure freedom for 3 years. While thriving developmentally, Mason has TSC manifestations in his brain, eyes, and kidneys. Currently, he does not need medication for his kidney involvement. Epilepsy and autism spectrum disorder are his main challenges.

The continued struggle with the behavioral manifestations, now called TSC-associated neuropsychiatric disorders (TAND), has been the hardest. Ashlin's autism, mood disorder, and ADHD compounds with her frustration, intellectual disabilities, language delays, and anxiety. It manifests into self-injurious and disruptive behaviors. Mason, while not self-injurious, with a diagnosis of autism spectrum disorder, struggles with extreme anxiety. As a caregiver, this continues to be the biggest symptom and effect on quality of life in TSC that is not easily remedied. The intensity of these behaviors and daily struggles have wreaked havoc on our entire family. We have in home behavior supports for both children, but it still renders a family helpless and isolated.

The urgent priority is proper diagnosis of TSC and the type(s) of seizures occurring as early as possible. The ability to prevent seizures and the devastation it does to a young developing brain could change the development of that child and the trajectory of the disease manifesting. Concurrently, the emergent issues of behavioral manifestations start young in our population. A concentration on the mental health components: depression, anxiety, and effective co-therapy of behavior services is needed. This may mean working on repurposing drug treatment options already on the market as well as identifying new treatments.

The TSC consensus treatment guidelines recommend the proper monitoring and time frame of the multiorgan involvement and to treat as symptoms occur. The long-term need is to prevent these manifestations before onset. The medication options currently available leave questions of what the long-term effects could be from lifetime use. We have made huge progress in terms of research and new treatments, but we have more work to be done and more answers to find.

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Over the past 17 years, both of my children have participated in research opportunities to help provide a better understanding for the future of TSC and are fortunate to have some of the best TSC specialists on their team. I've witnessed so much progress, and because of additional options now available, I have so much more hope for Mason's future.

I'm thankful for the support and information I've received over the years from the Tuberous Sclerosis Alliance, which is the only US-based nonprofit organization dedicated to finding a cure and better treatments for TSC. They have been instrumental in helping and supporting my children as well as working to make sure TSC research truly focuses on the needs of families like mine.

Just as important, I struggle to articulate how grateful families like mine are for such a strong, compassionate, motivated, and dedicated community of researchers and clinicians. Your tireless and unwavering commitment to understanding the pathogenesis and manifestations of TSC has a profound impact. The knowledge gained in the accomplishments of your work will yield a better quality of life for TSC patients present and future. The definition of hope is the feeling of wanting something to happen and thinking that it could happen. Individually and collaboratively you have provided hope.

TSC has redefined my responsibilities as a parent and led me on a journey I never planned or expected. I am, however, armed with knowledge, endless support in the TSC community, and invaluable insight.

Please accept my sincere gratitude and appreciation on behalf of the entire TSC community.