Our Life with MCT8 Deficiency:

Jolene, Vini, and Sebastian

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I’m a single mother to two incredible boys—Vini, 13, and Sebastian, 7. Both of my sons have monocarboxylate transporter 8 (MCT8) deficiency—also known as Allan-Herndon-Dudley syndrome—a highly debilitating rare disease with no available treatment, but their journeys have been completely different. One of the biggest challenges of being a full-time mother, advocate, and caregiver to two children with MCT8 deficiency is managing their very different needs despite sharing the same diagnosis. I work as a professional family advocate and have served on hospital, state, and national advisory boards. Still, nothing has taught me more about the realities of rare disease than raising my boys.

My son, Vini, went undiagnosed for six long years. From the moment he was born, something felt off. He was labeled “failure to thrive,” and we were sent home too soon. He wouldn’t feed or latch. I was a new mom and didn’t realize he wasn’t meeting milestones until family members gently pointed it out.

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Eventually, a geneticist recommended a full genetic panel and T3 hormone testing, which finally set us on the path to understanding what was happening. But at that time, multidisciplinary care in my state was so fragmented that I was constantly traveling long distances to get Vini the evaluations he needed. It was exhausting, lonely, and overwhelming.

When I was pregnant with Sebastian—whom we lovingly call “Sebash”—doctors warned me about a high risk if the baby was male. Pre-natal genetic testing confirmed that he also carried the MCT8 mutation. Hearing that news was

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devastating, but I was determined to give both my boys the care, support, and love they deserved, even in the face of divorce and becoming a single parent caregiver.

Vini, once labeled “failure to thrive,” has made steady progress through years of physical and occupational therapy and hard work. He uses a wheelchair, stays cognitively engaged, and can say a few words. Watching him grow into himself—slowly, but surely—has been one of the greatest joys of my life.

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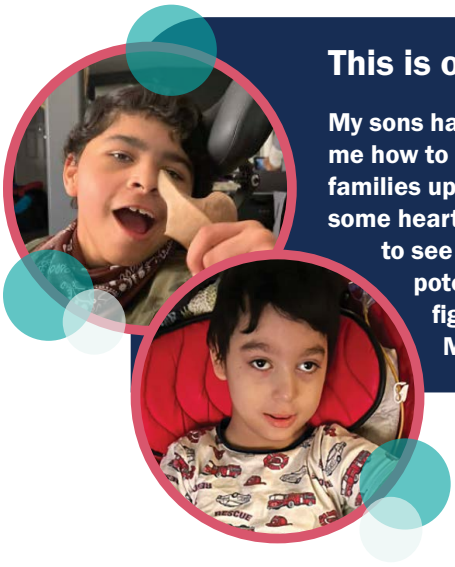
Sebash’s journey has been far more severe. In addition to MCT8 deficiency, he also has autism and Lennox-Gastaut Syndrome (LGS), a rare and debilitating form of epilepsy. I’ve noticed LGS in several other boys with similar MCT8 mutations, and it adds a layer of complexity most people can’t imagine. There were times we thought we were going to lose him. He was in hospice once. He’s been at the edge of life more than once. As a mother, nothing prepares you for that kind of fear.

If there is one thing that keeps me going, it’s the MCT8 deficiency community. I am deeply involved in supporting and sharing information with other MCT8 parents and caregivers. We regularly share updates on research, personal experiences, and practical tips. We just understand each other in a way no one else does. They are my lifeline.



This is our life with MCT8 deficiency.

My sons have shaped me into the advocate I am today. They’ve taught me how to fight for answers, push for better care, and lift other families up along the way. Our journey has been full of challenges—some heartbreaking—but it’s also been full of purpose. I am hopeful to see more investment in research for MCT8 deficiency and potential new treatments. No matter what, I will always keep fighting for Sebastian, Vini, and every family living with MCT8 deficiency. My boys deserve nothing less.

Learn more about MCT8 deficiency and read more community stories at www.lifewithmct8deficiency.com.