Our Life with MCT8 Deficiency:

Rebecca and Elijah

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Elijah, whom I call the “joyful hero,” has a smile that can light up any room; however, his life didn’t start that way. His early years with his biological parent were tough. He didn’t receive consistent medical care, and although he was initially diagnosed with cerebral palsy, Elijah lived in unstable conditions, mostly bedridden, sleeping 17 to 18 hours a day, and often distressed.

In Fall 2022, when he was twelve, everything changed. After an extended hundred-day hospital stay, the initial call was for a foster home that could provide nursing and even hospice care. It was a delicate path, suggesting Elijah’s time might be limited. That’s when I heard Elijah’s story. I was a nurse who had closed my foster license but reopened it with a heart full of hope, ready to care for Elijah. Elijah came to our home officially in December 2022. With the help of the local fire department, we were able to safely transport him into our home and begin the journey of fostering Elijah.

We started with the basics: regular medication, breathing treatments, and just unwavering attention. Slowly, small but significant improvements started to emerge. Seeing these glimmers of progress ignited a fierce advocacy in me. I just felt that Elijah’s journey wasn’t anywhere near its end.

A truly pivotal moment arrived when a neurologist reviewed an updated brain MRI. He noted a stark contrast between the scan and how Elijah presented physically, questioning that initial diagnosis of spastic quadriplegic cerebral palsy. That led to further investigation and advanced genetic screening. We waited for two long months for the results. Finally, in December 2024, when Elijah was fourteen, we received his true diagnosis: MCT8 deficiency, also known as Allan-Herndon-Dudley Syndrome (AHDS), a rare, severely debilitating, and life-threatening disorder.

Learning this diagnosis resonated deeply; it was something I had instinctively felt all along, a missing piece finally found. Reflecting on how late the diagnosis came, I couldn’t help but wonder what earlier intervention might have meant for him. An early diagnosis of MCT8 deficiency is essential to providing the patient with supportive care, which can greatly enhance the quality of life of patients and caregivers. Symptoms typically emerge early and often mimic those of other disorders.

Driven by my nursing background and my unwavering support for Elijah, I dove deep into research the moment I learned the diagnosis, seeking every possible avenue for resources and treatment, including exploring participation in clinical trials.

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“Enlightening” is the word I would choose to describe our journey with MCT8 deficiency. I remain hopeful for continued progress while also staying grounded in the reality of his condition. Elijah thrives when we’re outdoors. Thanks to some amazing fundraising efforts, we have a hiking chair and a special needs tricycle, allowing me to help him explore the beautiful Colorado landscape he now calls home. He loves watching football and baseball games and truly appreciates any opportunity to be in the company of others – it brings him so much joy. Elijah and I sometimes participate in local special needs outings, often alongside others from the cerebral palsy community, which feels familiar given his initial diagnosis. Elijah also loves to participate in regular therapies such as physical therapy, occupational therapy, speech therapy, and feeding therapy. He loves to move his body and is learning to navigate an eye gaze device so that he can one day communicate his needs.

Due to the rarity of MCT8 deficiency, connecting with other caregivers, especially within the MCT8 community, has been challenging. While online peer communities offer support, establishing genuine peer-to-peer connections has proven to be difficult.

As someone who isn’t Elijah’s biological parent and is relatively new to the broader special needs world, I sometimes feel like an outsider in those larger support groups. Recently, however, after watching a documentary on Lifetime TV called “Behind the Mystery,” which focused on MCT8 deficiency, local connections have begun to flourish serendipitously. Another family with a child who shares Elijah’s diagnosis has recently moved to our town. Tragically, this family has already lost children to the same disease. United by our shared experiences, we’ve found mutual support, but it has also highlighted the urgent need for a stronger peer-to-peer caregiving network within the MCT8 deficiency community.

Caring for individuals with MCT8 deficiency presents significant challenges due to the complex and severe symptoms of this rare genetic disorder. When people ask about the challenges, I often mention the unpredictable muscle tone issues, Elijah’s inability to move or speak independently, and the significant difficulties we face in understanding when he is distressed. Seizures, dystonia, and disrupted sleep patterns add so many layers of complexity to his care. Being his sole caregiver without consistent nursing support means I face challenging circumstances daily, requiring constant vigilance.

But I remain steadfast. I am determined to advocate for the importance of fostering kids like Elijah. Recognizing and understanding the unique challenges faced by caregivers of individuals with MCT8 deficiency is essential for creating effective support systems, interventions, and treatments that genuinely meet the needs of both patients and caregivers. Increased awareness among healthcare professionals is also vital; it could lead to earlier diagnoses and better access to appropriate care and resources for families like ours.

This is our life with MCT8 deficiency.

Despite all the hurdles, Elijah’s progress is inspiring. Hope for potential treatment options remains strong, not only for Elijah but also for other families navigating the complexities of MCT8 deficiency. This journey is one of constant learning, fierce advocacy, faith in God, and profound, unwavering affection for a boy whose joyful spirit and infectious smile shine brightly through everything.