

Our Life with MCT8 Deficiency:

Carter and Christina



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In October 2019, my son, Carter was born. He was a tiny bundle of joy, seemingly in perfect health. For three precious months, my world revolved around his coos, smiles, giggles, and cuddles; back then and still now, he is my bundle of warmth, especially when we hug.

As the months passed, as a first-time mother, I didn't notice that Carter wasn't hitting some of his regular milestones as he grew, until my sisters shared their concerns with me. Carter was never a fragile baby, but the physical strength wasn't there. His head control showed weakness, and his ability to control his body muscles resulted in jerking motions or what I now call spasms. Not knowing what was happening, I was scared.

Unfortunately, the COVID-19 pandemic was beginning, which caused many problems in accessing care due to numerous shutdowns in doctors' offices. Carter was three months old when his symptoms started, but it was six months before his first virtual consultation with a neurologist. We saw his first neurologist in person when he was about ten months old. As his symptoms persisted, we were referred to another specialist and met them when Carter was about a year old, following his first MRI.

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Carter's pediatrician was a huge help; because of her dedication, Carter started occupational therapy by 9 months through an early intervention program, and his developmental delays were undeniable. The old and new neurologists suspected something beyond cerebral palsy. The path led us to a geneticist. It took six to nine months to wait for the first genetics panels. The saliva swab test came back. During this painful period of uncertainty, Carter was inexplicably removed from his vital occupational and physical therapy. He was deemed ineligible without a solid diagnosis, which felt incredibly unfair and set us back.

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A flicker of hope emerged with the initial diagnosis of Angelman syndrome, but his traits were too few to confirm it as his official diagnosis, and his doctors doubted it, too. So, more waiting, additional bloodwork, and another panel of genetic swab tests stretched this process into several more months. Carter also started having seizures.

Then, as Carter turned three years old, the diagnosis arrived: MCT8 deficiency, also known as Allan-Herndon-Dudley Syndrome (AHDS). I had to Google the diagnosis to help me understand it better. I learned that MCT8 deficiency is a rare genetic disorder affecting the brain and multiple other organs, resulting in developmental delays and other related issues. Feeding difficulties, poor muscle tone, fast heart rate, trouble sleeping, and frequent infections are common issues with this disorder, and it is caused by mutations in the *SLC16A2* gene, which encodes the MCT8 protein - a transporter needed to carry thyroid hormones (T3 and T4) into brain cells. Affected individuals often have abnormal thyroid hormone levels (high T3, low T4, normal or slightly elevated TSH). There is no cure.

I cried. They also said that if I had another son, there is a 50% chance he could have the same diagnosis. This put fear in me about having more children, and I blame myself repeatedly because of my genetic disposition.

Since Carter was born, he has radiated pure joy. His beautiful smile lights up every room he enters, especially when he's fully immersed in an activity. Daycare, therapy, family events—as well as trips and weekend sports tailored for children with disabilities—he embraces them all. Currently, Carter enjoys attending his school, which is dedicated entirely to children with all types of disabilities.

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This is our life with MCT8 deficiency.

As I look at my smiling son, I cherish the present moment, appreciating each laugh and small victory while navigating the uncharted waters of our journey with unwavering love and fierce determination.