

Our Life with MCT8 Deficiency:

Gemma, Aaron, and Their Four Boys



“My husband and I knew we had to be our son’s best advocates. Our nursing training told us that cerebral palsy didn’t typically explain what we were seeing.”

We’re Gemma and Aaron, and together we have six children in total: five boys and one girl. Four of our boys—including our almost two-and-a-half-year-old identical twins—have MCT8 deficiency, also known as Allan-Herndon-Dudley syndrome (AHDS), a rare, highly debilitating disorder with no approved treatments in the US. This X-linked disorder causes neurodevelopmental challenges, metabolic and cardiovascular issues, and a shortened lifespan. Our daughter, age 16, and oldest son, age 14, are typically developing teenagers who have become wonderful co-supporters, helping their siblings with MCT8 deficiency in every possible way.

We met while serving in the U.S. Army, and we were both nurses, but we have since stepped away from the profession. Out of necessity, our careers shifted to be more flexible around the needs of our children and Gemma transitioned to being the full-time caregiver for our boys, which, in many ways, has felt like continuing the nursing work we trained for. Yet, even with our medical backgrounds and understanding of the healthcare system, the road to getting our sons diagnosed and supported has been one of the hardest things we’ve ever faced.

Our oldest son with MCT8 deficiency—now age 10—was born in El Paso, Texas. After birth, and as the weeks passed, we noticed he wasn’t holding his head up the way his older siblings and other babies did. As his first symptoms emerged—failing to meet key developmental milestones, muscle weakness, and feeding challenges—doctors began to suspect cerebral palsy. Some questioned whether something had gone wrong during Gemma’s pregnancy or delivery; an accusation that haunted her. Even as a nurse, knowing she had done everything right, Gemma felt the weight of that suspicion settle on her as a mother. We moved to Columbia, Missouri shortly after, and by four months old, our son was labeled “failure to thrive.”

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We knew we had to be our son’s best advocates. Our nursing training told us that cerebral palsy didn’t typically explain what we were seeing. We pushed for answers for two long years, traveling from specialist to specialist, including a neurologist, endocrinologist, cardiologist, orthopedic surgeon, dietician, gastroenterologist, and multiple therapists for physical therapy, occupational therapy, and speech. Finally, a physical therapist said gently, “I don’t think we have the answers for you here. You need to go to a hospital in the bigger city.” That meant a nearly three-hour drive each way to St. Louis.

When we were finally able to be seen by a geneticist, he looked at our son for a moment and said, “I know what this is.” We were stunned; we had moved mountains for years trying to find a diagnosis, so how could this new specialist know just by looking at our son? The genetic counselor then came in and explained that they had recently diagnosed another child with the same condition at their clinic. We were encouraged to undergo genetic testing not only for our son, but also for Gemma, because the condition is X-linked, meaning it’s inherited from the mother.

A few weeks later, genetic testing and a thyroid hormone panel that included triiodothyronine (T3) testing confirmed the geneticist’s suspicion: MCT8 deficiency. We learned the disorder is caused by mutations in a gene called SLC16A2, which provides the code for a protein called monocarboxylate transporter 8 (MCT8). This protein allows thyroid hormones — especially T3 — to enter cells in the brain, where T3 is critical for development, and cells in other tissues, where it supports important metabolic processes. MCT8 is the major T3 transporter in the brain, so when it isn’t working properly, T3 can’t get into brain cells during development in the womb and after birth, and brain levels are too low. In other tissues, T3 enters through “back-up” transporters, so levels end up too high.

The same testing confirmed that Gemma was the genetic carrier. Even though she knew she hadn’t done anything wrong — that it was just her genes — she felt the worst kind of guilt. She was angry, thinking, “Why me? How could I do this to my child?” Perhaps the hardest part was explaining the diagnosis to Gemma’s mother and relatives from Mexico, who mistakenly believed the diagnosis was due to a “curse” or something she had consumed during her pregnancy. Confronting those cultural misconceptions and misinformation, alongside the emotional and physical toll of the diagnosis, was overwhelming.

At the same time, we had always wanted to have more children, but our son’s diagnosis brought up many fears. However, after a few years, we realized that being parents and bringing children into the world is the greatest gift we could receive in this life.

Almost three years later, we had another son—now 8 years old—and then our set of twin boys, all now living with the same diagnosis of MCT8 deficiency. While some people may view them as having special needs, we see those “needs” as hidden blessings and are dedicated to loving, caring for, and supporting them in every way we can.

In the MCT8 deficiency community, we often hear that no two boys with MCT8 deficiency are alike—well, our four boys sure do prove that. Despite sharing the same genetic mutation, each boy presents differently. Our 10-year-old son has more significant physical limitations than his 8-year-old younger brother, who can manipulate toys and communicate with a few words. The twins are different, too. Right now, at almost two and a half, they can sit up on their own, dance to music with their hands raised, and are getting closer to walking with support. Some of these differences we attribute to our growing knowledge of MCT8 deficiency and being able to obtain earlier support and services, which seems to have mattered enormously. But every one of our boys is his own person, and each has needed his own path of care.

Today, our family lives in a small rural town in Missouri, surrounded by cornfields. The isolation is real. Our boys had been making strong progress on eye-gaze communication devices before we moved here, but our rural school doesn't have the same expertise, and they have regressed. Equipment designed for special needs kids often isn't built for older, larger children. We've had to change our growing boys in the back of our special-needs van because we couldn't find an accessible changing surface big enough when we were out. We tell their therapists all the time: it doesn't seem like the people designing this equipment ever talked to a real parent. Living in such a rural area also means having to travel two hours to St. Louis just to get blood draws and checkups.

The financial strain has also been significant. Proactive, optimized multidisciplinary care is essential throughout our boys' lives. In the early years, under Aaron's private insurance, the testing, specialists, and appointments drove us into bankruptcy. We lost our house in El Paso, plus the special needs van we were still paying the loan on. There are programs designed to support caregivers like us, including state-funded retreats, but most assume you can leave your child somewhere—and there is no "somewhere" for kids like ours. Who cares for our sons if we travel? We need support built around the real lives of families caring for loved ones with MCT8 deficiency, not assumptions that don't fit.

In tandem, there needs to be greater recognition of MCT8 deficiency symptoms to support earlier diagnosis. Once we found the online support group for MCT8 deficiency families, something shifted for us. Suddenly, there were other parents like us who understood our realities. We learned about emerging therapies, adaptive equipment, and research opportunities. Today, we try to give back by sharing what we have learned, documenting our family's journey on social media, and ensuring new families don't feel as alone as we once did. It's a reminder that awareness of rare diseases doesn't grow only from research centers and big hospitals — it also grows from families like ours, telling our stories wherever we live.

Our boys have given us a master course in patience, gratitude, and living in the moment; shaping us into the parents and fierce advocates that we are today. They have also strengthened our faith, encouraging us to educate our cultural and religious communities to correct misinformation about disability and the root cause of rare, genetic conditions like MCT8 deficiency. Being seen and understood by your community matters almost as much as being heard by your doctors. Whatever the road ahead looks like, we will keep fighting for our boys, sharing what we learn, and walking alongside other families on this journey.



This is our life with MCT8 deficiency.

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