

# Our Life with MCT8 Deficiency:

## Tom, Meghan, and Jack



“Our pediatrician kept saying, *“Oh, we haven’t seen that before,”* which wasn’t comforting—it just made us more confused.”

I never imagined that becoming a dad would mean stepping into a world of medical terms, hospitals, sleepless nights, and decisions that feel impossibly heavy. When my son Jack was born—tiny, premature, but perfect to us—we thought the hardest part would be getting him out of the NICU and home. We had no idea what the next two years would bring.

Because Jack was a preemie, early intervention was part of our routine from the start. When he was between three and six months old, developmental specialists began noticing he wasn’t meeting key milestones. We didn’t want to see it at first. He has a bunch of cousins around his age, and we’d watch them do things—rolling, sitting, reaching—that Jack simply couldn’t yet do. But when you’re a new parent, you cling to the belief that every child develops at their own pace.

Our visit to developmental pediatrics was one of the hardest days of my life. Nothing really prepares you to sit across from a doctor who lists everything your child can’t do—hearing all his missed milestones framed as deficits felt like a punch to the stomach. The worst part was not having any answers. No direction. No guidance. Just fear.

For most of Jack’s first year, we believed he had spastic quadriplegic cerebral palsy (CP). His symptoms seemed to fit, so we moved forward with an MRI to confirm the diagnosis. When the MRI didn’t confirm CP, we were relieved—but also terrified, because we were suddenly back to square one without answers. That uncertainty was incredibly heavy, and we endured several setbacks before our care team referred us to genetics, which ultimately led us to the diagnosis we had been searching for: MCT8 deficiency, also called Allan-Herndon-Dudley syndrome. I had never heard of it in my life, and neither had my wife, Meghan. It took us a long time to even understand what it meant, let alone accept it.

We dove into research—as much as parents can without a medical degree—and what we found was crushing. People with MCT8 deficiency have a genetic mutation that disrupts the monocarboxylate transporter 8 (MCT8). MCT8 helps transport thyroid hormones in and out of cells throughout

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the body. This includes transporting the thyroid hormone T3 into the brain. It is a rare disorder, and currently, there are no approved pharmacologic treatments in the US.

My wife didn't accept that there were no options. She fell into what I call "warrior-mode," deep diving into medical literature, connecting with specialists, and seeking out families who had walked this path before us. He's still significantly delayed—but we celebrate every gain, every little step forward.

Caring for Jack is a 24/7 job, one that Meghan and I share, but for long stretches, she shouldered the heaviest burden. We faced ongoing challenges due to feeding difficulties, limited mobility, and sleep disturbances, requiring constant care and support. She left her job to become Jack's full-time caregiver: nurse, therapist, chauffeur—roles with no off switch. Daycare wasn't an option as Jack is medically fragile and spent six consecutive months sick. As a result, Meghan has spent most days isolated at home, except for Jack's weekly therapy appointments.

I eventually left my job as well. I tried juggling 50–60 hours a week working as a paralegal while coming home to meet Jack's intense needs—checking his feeding tube, monitoring his oxygen levels, and preventing aspiration. It became a vicious cycle: burnout at work, guilt at home, and exhaustion everywhere. Eventually, I had to choose, and I chose Jack.

Most days, it feels like Meghan and I are the only support system. Meghan is very active in the MCT8 deficiency community, and through it, she met another family — a family who 'gets it' in a way few people ever will. When we visited them, it was one of the best days we've had since Jack was born. Seeing their 8-year-old son included in everything showed us a different kind of future. Not just surviving with MCT8 deficiency, but thriving with it. That family embraced us immediately. It was one of the first times I felt like we weren't alone.

Meghan has also found a sense of purpose by sharing Jack's journey and advocating for other caregivers. She has supported legislative efforts in our state that would enable parents to be paid caregivers—a policy that could significantly affect lives like ours. She's also helped raise awareness of MCT8 deficiency and the urgent need for new therapies among regulators and other key decision makers.

“As Meghan says, “We don't work with milestones. We work with inchstones.”



## **This is our life with MCT8 deficiency.**

**As a dad—especially a dad of a medically complex child—I've learned you must talk about your feelings. I grew up believing you should keep that stuff inside, but if you do, it will crush you in this world. You need to talk to your spouse, your friends, and other parents. Despite everything—the grief, trauma, and uncertainty—Jack brings so much joy into our lives. I wouldn't change him for a second. The journey hasn't become easier, but it has become clearer. We understand what Jack needs, and we fight for it every day. Our lives aren't the ones we imagined, but they're ours, and Jack has taught us more about resilience, love, and purpose than we ever could have learned any other way.**

*Learn more about MCT8 deficiency and read more community stories at [www.lifewithmct8deficiency.com](http://www.lifewithmct8deficiency.com).*