Our Life with MCT8 Deficiency:

My journey into motherhood began with pure joy: my firstborn, a healthy daughter, completed my world as a single mom. Ten years later, remarried, I faced heartbreak—losing my son Will six months into pregnancy. Trusting my instincts, I requested genetic testing but was initially denied and told it was just a fluke, and it wouldn't happen again.

Nicole and Liam

When I became pregnant again within a year, my doctor finally allowed genetic testing at four months. The results hinted at chromosomal issues behind Will's passing, casting a long, heavy shadow.

In 2011, Liam arrived, and his birth brought a renewed sense of hope. The initial tests showed a healthy baby boy, and my heart began to believe. However, a concerning decline started around 4-6 months. Liam struggled to hold his head up; he seemed to have no movement on his left side, and even feeding became difficult. We started therapy, and although a clear diagnosis was hard to determine, doctors labeled it cerebral palsy.

Liam's early years were marked by a relentless cycle of illness. RSV, the flu, monthly hospital stays, and frequent ER visits defined our complex reality. Doctors blamed his problems on "failure to thrive." My heart ached as I tried to feed him pureed foods, but Liam's small body fought back, gagging and vomiting. A warning from a feeding therapist confirmed my worst fears: Liam was aspirating. It seemed that food was becoming his enemy.

> "Liam's early years were marked by a relentless cycle of illness. RSV, the flu, monthly hospital stays, and frequent ER visits defined our complex reality. Doctors blamed his problems on 'failure to thrive.'"

A doctor recommended a gastrostomy tube, also known as a G-tube, which is a surgically inserted tube that provides access to the stomach for feeding, hydration, or medication delivery. Liam's responsiveness decreased, leading to a frightening emergency that led to G-tube surgery. I carefully documented every medical visit in a binder, a desperate effort to keep track of everything, and I voiced my important request for post-operative cortisone to the surgeon and nurses. One nurse initially dismissed my plea. After Liam was moved to recovery, and when I repeated my concern to a second nurse, I encountered the same resistance. Then, Liam began to code. The trauma of that moment etched itself into my memory, leaving me with PTSD.

Thankfully, the hospital took the incident seriously, and my voice, the voice of a fearful mother, echoed a universal truth: parents need to be heard. But the ordeal wasn't over. The day after surgery, I was overwhelmed by Liam's fragile state and numerous wires. While attempting to untangle an IV, the G-tube drainage bag accidentally dislodged. Though nurses reassured me, the guilt was immense. The reinsertion was done incorrectly, requiring a second immediate surgery, which was devastating. I was sent home with a brief tutorial on Liam's feeding pump, which operated around the clock. Despite my requests, I was denied at-home nursing assistance, leaving me to care for Liam day and night with little to no sleep.

> **"I knew I needed to learn as much as I could about what was happening to my son."**

I knew I needed to learn as much as I could about what was happening to my son. In 2013, we pursued genetic testing. Blood samples were taken from me, my ex-husband, and Liam, but we faced a two-year delay in receiving the results. In 2015, a new chapter began as we relocated from New York to Florida. My determination to find answers and better care for Liam only intensified. We saw a new gastroenterologist and a geneticist who was finally able to access our 2013 results—the news was devastating.

Liam's condition was not cerebral palsy but Allan-Herndon-Dudley Syndrome, also known as MCT8 deficiency. This rare, life-threatening disorder leads to neurodevelopmental impairments, systemic metabolic and cardiovascular disturbances, and increased morbidity and mortality. They explained its rarity, a cruel consequence of an X chromosome mutation that I did not know I had. I am the first one in my family. A bleak picture emerged: a shortened lifespan, a life bound to a wheelchair, devoid of speech, and the ability to eat. I felt utterly isolated, adrift on a lonely island of despair with my sweet boy.

However, a lifeline appeared when I was connected to an online group for MCT8 deficiency families. This was a turning point. Suddenly, I wasn't alone anymore. I made a silent promise to Liam, a fierce vow whispered into his soft hair: I would face this head-on, for him. I tackled each symptom with unwavering resolve, fighting for every therapy, every piece of equipment, and even involving state representatives when I encountered roadblocks. Comprehensive, lifelong care for MCT8 deficiency requires a multidisciplinary team, ensuring proactive coordination across disciplines to support individuals effectively affected by the condition. Liam began physical and occupational therapy and enrolled in a special needs school. The need for constant care meant that having a 24/7 nurse became essential, as he required numerous feedings around the clock. This time, the nursing request was approved.

Throughout Liam's journey, routine blood tests have been a constant. One day, Liam's health took a sharp and terrifying downturn. Tachycardia and seizures prompted a frantic rush to the hospital. An endocrinologist joined his care team and ultimately ordered a full thyroid panel. The results revealed alarmingly high T3 levels, a crucial piece of information that had never been shared with me before. I was always told that if your thyroid-stimulating hormone (TSH) is normal, then your thyroid is fine, but that isn't always true. He had no issues with his TSH. How could something so important be missed for so long? Liam seems to be doing okay in other aspects, but his thyroid levels are a rollercoaster. We have to make adjustments here and there as he is going through puberty, too.

Liam is now receiving palliative hospice care at home. I live with the constant, heavy fear that he might slip away in his sleep, a common and cruel complication of MCT8 deficiency. Yet, amidst these challenges, I focus on bringing joy to Liam's days.

This is our life with MCT8 deficiency.

I know, with every fiber of my being, that his happiness is what truly matters in this unexpected and challenging life we share. I will continue to fight for him, advocate for him, and cherish every single moment we have together.