

## Sean

THIS IS A PERSONAL STORY SHARED BY A FAMILY WHO HAS A CHILD WITH  
TRIFUNCTIONAL PROTEIN DEFICIENCY (TFP)

After enduring one year of surgeries for endometriosis, I went through in vitro-fertilization to get pregnant. I was so excited when I found out I was pregnant. Unfortunately, I started to have complications very early on. At 4 months I went through a spinal tap and almost lost the baby. My migraine headaches were getting severe and the doctors wanted to rule out meningitis. At 5 months a C-Line was put in me so I could eat because I was unable to keep anything down. I was hospitalized almost every month for dehydration. In my 7th month I fainted in a grocery store and was whisked away in an ambulance. At 30 weeks I was in labor. The doctors kept things under control until 35 weeks, when Sean was born on September 16, 1998.



I will never forget that day. I was so tired and it hurt to move. I stayed in bed and told my doctor that I stopped taking my medicine to stop the labor because it was making me ill. At 5 pm she called me to tell me to go to the hospital. I told her I couldn't. I could not move. I was in so much pain and very tired. I just wanted to sleep. At 8 pm she convinced me to get to the hospital. When I got there, blood was immediately drawn. About 20 minutes later, my doctor came in and told me that I'm having the baby tonight. I told my husband to go home and get the labor bag. She said "No, you don't understand. We have to do an emergency c-section. You and the baby are in distress." I was then told that my liver levels were at 863 and they are not supposed to be above 40. My body was full of toxic poisoning caused by my baby. We were both dying.

At 9:50 I was in the OR and by 9:55pm my beautiful blue-eyed baby boy was born - all 5lbs3oz of him. I didn't see him until the next day. I was almost in a coma. I was told that I lost a lot of blood and almost didn't make it.

We came home from the hospital 4 days later. His pediatrician wanted to keep an eye on his weight. We went in for weight checks every couple of days the first weeks of his life. The next few months were almost uneventful. I questioned his doctor about his head being so flat on one side and he told me that happens when the baby sleeps favorably on one side. I questioned the doctor about his constant spitting up. I was told all babies spit up. And I questioned the doctor about Sean's breathing, I was told some babies breathe funny. Since this was my first baby, I put all my trust in the doctor.

While I was still pregnant I interviewed every pediatrician in my area and I liked this particular doctor right from the start. He had the right credentials, and was kind. From the time Sean was born he had a severe snoring problem. He would breathe very loudly in a flat lying down position and was quiet when he would be put in a 30 degree angle position. Besides his heavy breathing Sean took forever to finish a bottle and then would

spit up or vomit most of it up. His head started to get flat on one side. I asked his pediatrician about his breathing, his head going flat, and the spitting up. I got for a response "you are a first time mom, some babies breathe funny, some spit up and some take longer to feed". Since I was a first time mom, I believed him. Sean was doing pretty well until January 1999. He was hospitalized for the croup and sent home after 3 days. The following week he was hospitalized with bronchitis and sent home again after 3 days.

The night we got home from the hospital, Sean was having problems breathing. His regular doctor was away so the doctor covering for him told me to take him to a Children's Hospital in Jacksonville. So that is where I took him. He was subsequently diagnosed with Craniosynostosis-Lamidodial Suture, Laryngomalacia, Hypotonia, Gastro-Esophageal Reflux. He was discharged 4 days later with surgery the following month. My trust in doctors now gave way to fear, disbelief, and misgivings. Everything that I questioned Sean's doctor about and had gotten an 'everything is ok' reply, were now definitely not 'ok.'

Early in February I had a hysterectomy after my doctor told me that if I ever got pregnant again I would be dead by my 4th month. Three weeks later I took Sean to the Children's hospital to have his surgery. The doctor opened his skull in the form of a horseshoe to repair his head. The suture had to be surgically opened or his brain would continue to push his forehead out causing him to look deformed. After surgery his head started to look round again. We were home a week later.

That's when the ear infections started, one right after the other. I kept taking Sean to his new Doctor to see what was wrong. I was told that some children are just prone to ear infections. In April Sean's doctor took a six-week vacation and had a different doctor covering for him. During this time Sean came down with a case of the runs. The doctor that was covering ordered a series of tests that came back negative, thankfully.

On June 4 1999, I got a knock on my door. Unassuming me answers the door to find a representative from the Department of Children and Families standing there. I was being investigated for Abuse. I strongly believe, for various reasons, that a family member, whom had never gone with me to Sean's Dr's visits, had a nurse whom had NEVER seen Sean before, call in a report because I was taking Sean to so many doctors. The authorities believed that I had to have Munchausen Syndrome by Proxy. This family member also inaccurately told the Department that I was not taking my medication for Borderline Personality Disorder - a disorder I have NEVER been diagnosed with.

The next day I had to appear in court with my son. When I told the Judge that my son had skull surgery and was diagnosed with other ailments, he took my son from me and ordered him to be put in Foster Care. I was unable to get any money from my husband for a lawyer, so I used my engagement ring. I was able to find a lawyer that would take it for payment. After 36 psychological tests with the Department's psychologist, 2 sessions with a psychiatrist, 2 sessions with a therapist, I was finally awarded my son after no doctor could find anything wrong with my mental state. Five weeks went by before I could put my son to sleep in his own crib. That was the 2nd happiest day of my life.

The Department still wouldn't give up the hold they had on me. In August I was told that I had to sign a six-month voluntary contract. It included marriage counseling, putting Sean in day care and only taking Sean to one pediatrician. I signed it. My husband and I went to marriage counseling. Our marriage counselor thought it best to do divorce counseling instead. In November, I had to get a restraining order against my husband after he was arrested for battery on me. After all of that, I went back to work and put Sean in day care.

Sean was getting sicker and sicker. I was always taking days off to take care of him. Then the Department of Children & Families came back in my life. Sean was tested for developmental delays and was found to be at approximately 9 months to 14 months. At the time he was 17 months old. My caseworker and I had several arguments. He wanted me to put Sean in a special day care for physically challenged children. I wanted Sean

to stay at the school he was in. He liked his teachers, got along with the other children and I thought that such a change would be very disruptive to him. Instead of "patching up" Sean's delays, I wanted to find out the cause of the delays. The Department of course did not agree with me. I took Sean to his doctor, another pediatrician; only this time I chose a group of doctors, to get a referral to see a specialist.

Sean saw Dr. Hammond, a neurologist at Nemours Children Clinic in Jacksonville in March 2000. Dr. Hammond ran a Creatinine Kinase (CK) blood test. The results came back at 10,350. I was told that figure was high and to get another lab drawn. I took Sean to my local hospital for the blood work. This time the results came in at 150. At our next visit with Dr. Hammond, I was told that Sean could have Muscular Dystrophy with the high numbers in his first CK labs. Dr. Hammond referred us to another Doctor at Nemours who could order the right tests; the only problem was we had to wait six weeks before we could be seen.

In May I was served with divorce papers at work, and it was also the month that my restraining order ended. My parents graciously decided to hire an attorney for me, and thus the hunt began. My father accompanied me to our first interview and we decided that he would be the best "man" for the job.

June 8, 2000 was the day Sean and I met our Guardian Angel face to face. I took Sean to the Genetics Division at Nemours Children Clinic in Jacksonville. Sean and I were playing on the floor of the examination room when our angel walked in. He thought that Sean might have a genetic defect and wanted to start to do studies right away. On June 16th Sean was hospitalized for further studies that proved that Sean had a breakdown in his Long-chain 3 Hydroxy acyl CoA Dehydrogenase Deficiency, or LCHAD for short. I was also informed that I did not need the hysterectomy. The complications from pregnancy arose due to my baby's genetic problem.

Dr. Perszyk told me that many of his mothers have been accused of Munchausen Syndrome. He told me that I was not crazy, that Sean was very sick, and he gave me my dignity and pride back. He gave Sean life. Without this diagnosis Sean would have died when he had an episode two weeks later and wound up in the Pediatric Intensive Care with CK levels at 68,000.

In July a nasal feeding was placed in Sean's nose. I had to learn how to change the tube every week. He was put on a feeding pump and special formula. I had to quit my job to take care of him. In September Sean underwent G-Tube surgery and received the stomach "button". During that hospital stay, Sean was seen by a Pulmonary Doctor who assessed him as having underlying Asthma. His nebulizer treatment medication was changed and now he gets his breathing treatments three times daily. So far there has not been any breakdown of the major muscles, the heart, lungs, kidneys or liver. His eyes were checked as well and were unaffected.

As of today he still has his "bad" days when he just seems to wobble, or chooses to not walk. He crawls with his head down and he sleeps a lot. Those are Sean's warning signs that his CK levels are rising. It is this period that is critical that his "Angel" Doctor become involved. Children with this disease can die with little advance warning. Sean could have easily died but thankfully he did not. Genetic Disorders can be detected at birth if testing is done. The HMO's decided to not offer the testing to save money. If newborn testing was done a 3-day-old baby would not have died in her mother's arms, a one year old would not have died in Intensive Care from Liver damage, and a 17-year-old boy would not be crippled. The list goes on and on.

The one thing that I do not understand is how the Department of Children and Families has so much power that they can legally 'kidnap' a child only on hearsay without proof. The Police force has Internal Affairs. Even the Health Insurance Companies have a watchful eye. Who is looking out for the children? I was told that if parents change doctors more than twice in one year, the Department of Children & Families could take legal action. If you move more than once in a three-year period you could have your child taken away. How do parents get back their God-given right to raise their children, to pick their doctor, and to move if they feel it neces-

sary?

The divorce trial ended May 2001. The Judge ordered 50/50 custody. I take care of Sean while his father is at work. He doesn't have to pay child support or reimburse me for any medical equipment. In November 2001 He evicted me and Sean out of the marital home. We have now relocated to a new city and made new friends. The judge ordered that Sean get dropped off to me by 6:45 am and be picked up by 3:30 pm so that I can work. His father drops him off at 6:15 am (waking him out of a sound sleep) and picks him around 4:30 pm.

In May 2000 Sean entered into the research protocol in Dallas TX with Dr. Charles Roe. Further testing proved that Sean has Tri Functional Protein Deficiency TFP not LCHAD. He was started on a new formula with new oil and it has proved to be more effective than the MCT oil he was on. In spite of the benefits from the new formula, Sean needs special molded boots to support his ankles. His therapist and I are working together in acquiring weight shirts for him to wear daily to help with balance. Sean overall is doing better but there are more and more obstacles that we face on a day to day basis. I am just very very thankful that he is still alive.

The attorneys that I have talked to all said that I have a great case, unfortunately I do not have the funds to hire them.

If Sean was diagnosed at birth, there is substantial proof that he would not have a g-tube or have the developmental delays he has now.

Sean and I are living solely on what SSI gives us, \$455 a month. We have turned to every organization for financial aid which proved useless. Out of desperation I started Metabolic Disorders National Affiliation, a 501C non profit foundation geared to help with financial aid to those who have children suffering from metabolic disorders. You can find us on the web at [www.metdna.org](http://www.metdna.org). God Bless.

Sincerely,

Keri Calisto

Written August 2002 by Keri Calisto

Mom to Sean

Palm Beach, Florida