

Caden

THIS IS A PERSONAL STORY SHARED BY A FAMILY WHOSE CHILD
HAS GLUTARIC ACIDEMIA TYPE II (GA-II).

On July 13, 1998, Michael Caden Richards was lucky enough to have been born to a couple of the most persistent parents there are. Immediately after he was born, we recognized an array of abnormal distinctions about him. He was a full term baby and weighed in at 5lbs 12oz. His skin was golden-yellow and he had very little fat under his skin. His skin was very loose and wrinkly. He seemed to be unusually hungry all of the time and his left foot was turned in very badly. Nothing was mentioned by the staff so, we didn't think too much more of his appearance.



I had also just endured a brush with death as a result of pre eclampsia and felt like we were just lucky to be alive. Three years later, we found out that he had been treated for hypoglycemia while in the nursery. We were never made aware of this! The nightmare began when we came home with our little, now 5 lb 9 oz package.

Caden was an inconsolable baby, to say the very least. Many times we resorted to giving him pain medication unsure what was wrong. We thought it might have been belly pain from the extreme amount of bottles he was drinking. Naturally, we complained to his Nurse Practitioner about this and was assured we were making too much of his crying. Finally, we got some lab work done. His labs were showing that he was in complete kidney failure! A few days later they were redone and his kidneys showed NO sign of failure! Prayer? He did have a left sided hydronephrosis, in which they attributed his crying to possible pain from the kidney holding urine. Weeks turned into months and his symptoms were increasing. It was ear infection after another, which resulted in surgery for bilateral tubes in ears. His system was in constant chaos. Some of his unexplained symptoms were: loss of weight, projectile vomiting, gastroenteritis, hypotonia, upper respiratory infections, periodic loss of appetite, pale with dark circles under his eyes, thrush, polydipsia & polyuria, malaise, extreme irritability, insomnia, dysuria, dehydration and oily/clay colored stools.

When he turned 9 months old, it was obvious that the left side of his face was different. The left side of his face from under his eye down had fallen. His eye still looked fine but the left side of his mouth and neck, appeared to have no muscle function. He was immediately sent for an MRI, which was normal. It was racked up to be some sort of facial paralysis, and then dismissed. As time has gone by, it is barely visible now. When he cries, you can still see the asymmetry in his mouth and his less defined jaw line on the left side, but other than that, he has made a remarkable recovery from "something". In February of 2000, when Caden was 19 months old, he started showing signs of un-coordination. We took him to the ER and they did a finger stick for blood

glucose and got a reading of 206.

Later, they drew blood and naturally, it was normal! They explained how inaccurate blood glucose machines were and so on. Nevertheless, we were still going to keep an eye on his blood sugar. We probably checked him a few times here and there and stopped because we didn't get anything abnormal.

In October of 2000, at 27 months, he woke up one morning staggering and shaking very bad. He had ran a fever the night before. I had obtained a blood sugar monitor through his insurance by this time. His blood sugar was 31! I thought there must have been an error. I used 3 glucose strips to confirm the number. I treated him with Popsicles and soda. It was now 292 when we left for the clinic. It stayed high for some time. According to their meter it was still at 245 and confirmed by laboratory.

There after, he had so many high and low blood sugars that I was afraid to go to sleep at night. He had a 26 and 260 in one day, documented at the Clinic. I don't know if it was out of frustration or pure disbelief, but his Nurse Practitioner who had taken care of him since birth, flat out told me that he was just fine. Nothing was wrong and it was very normal for his blood sugar to be 31 or 245! She suggested we stop checking his blood sugar and for me to get some psychiatric counseling! I left, never to go back. How could I? He was going to die, unless I kept looking for answers! There was only one other doctor's office in town. We made our first visit and presented our case with no problem. His blood sugar issues were of great concern as well as his severe leg pain. His new pediatrician had a bone scan ordered, after I asked him to. It was normal. The leg pains had originated from very early on, even before he could talk because his body language told us so. You guessed it, Growing Pains! Yes, I knew better but what can you do?

He had also started having some "spells". That's all we knew to call them. He would become delirious and unresponsive. In January 2001, when Caden was 30 months old, I found an endocrinologist on my own. After our first visit, we received a diagnosis of early Type I Diabetes. We were told to keep an eye on his blood sugar as he would probably just wake up diabetic one morning and we needed to prevent ketoacidosis. This endocrinologist basically took the easy way out. He based Caden's diagnosis on a hand full of facts: sister w/diabetes and occasional hyperglycemia and even though, his HBA1C was an average of 65! HBA1C gives you the average blood sugar for a 3-month period. He said that his pancreas was failing and this was all part of how type I diabetes evolved. His explanations did sound really good.

After a month or so of blood sugar monitoring, and a really bad illness related high blood sugar of 496, he was started on ½ unit of humalog in the mornings. This did not go over too well. He would drop too low after the shot. He wouldn't get hypoglycemic but hit the 60 mark and it made me very nervous. I opted to stop the insulin injections myself, even though the endocrinologist did not agree. Insulin could have killed our son!

Following several ER visits with low blood sugars in the 30's and 40's, we were told that we were welcome to get a second opinion. We decided to do so. All the while, his new pediatrician was forming his own opinions without my knowledge.

In May 2001, when Caden was 34 months old, he was admitted to a Children's Hospital in a near by state in the endocrinology dept. for a work-up to rule out diabetes. We stayed for 5 days and Caden done quite well. He was not sick (virus, cold etc.) nothing to influence his numbers. He had limited activity and very structured meals. Just the day before he was admitted, he was jerking due to a blood sugar level of 64. A blood sugar level of 64 is not medically considered hypoglycemic. Our main argument was Numbers. We were told that 60 is not low and there's no way his symptoms could be blood sugar related, when his numbers are this high. Our son felt terrible at 75. Why? Our conclusion was, that 9 times out of 10, during his "hypoglycemic" symptoms, his blood sugar will be anywhere from 60-75. The other 1 out of 10 was usually much lower.

These were very consistent readings documented by home glucometer and laboratory testing. Before we left, we met with the endocrinologist and his colleague. They basically took us behind closed doors and laughed in our faces! We were sent home even more disgusted than before. Our child had just endured almost a week of torture: 5 IV's, numerous finger sticks and starved him for nearly a whole day, for NOTHING!

According to medical records, prior to Caden's evaluation, they had already interviewed his former NP and current pediatrician, both of which helped entertain the diagnosis of Munchausen Syndrome by Proxy! Needless to say, we were very upset and sick to our stomachs that we were being accused of child abuse, for trying to help our sick child! It is the most sickening feeling I have ever had in my life and will never get over the hurt and betray. I had lost faith in the medical system. I knew there were good physicians out there. I just found it hard to believe that we would ever find one.

By the grace of God, a desperate mother looking for her daughter's diagnosis, found Caden's old web page on the Children with Diabetes website. She emailed me to let me know about the similarities in our children and that they were heading to Mayo. She promised to let me know any information she learned. She wound up sending me a lot of great information along with the FOD website address. After reading the FOD information on the site, immediately, I signed up on the Family Support Group email list. I knew I had found my son's diagnosis!

We went back to the Children's hospital for our follow up a few weeks later for the remaining test results and to discuss the MSBP suspicion. I was armed with information about FOD's. I had proof that there were other things (besides Diabetes) that could cause my son's symptoms. The endocrinologist even told me that there was NOTHING else that could be causing these symptoms, which were "waxing and waning"! Nothing! For every accusation of MSBP, I had an explanation. Symptoms of a FOD can and do "wax and wane." They are associated with high and low blood sugars and more importantly, could account for each and every one of Caden's unexplained symptoms. He did not deny the possibility of a Fatty Acid Oxidation Disorder but strongly discouraged further workup. He finally agreed to send Caden to the Genetics dept.

We met with the Head Biochemical Geneticist. We were brought in and talked to like mere idiots once again. This person met with us approximately 15 minutes and wound up making us sound more unstable than ever. She read over his prior reports and again formed an opinion based on other's opinions. She, very reluctantly, agreed to do an Acyl Carnitine Profile. In reading her summary of our visit, she said that there were NO real low blood sugars documented (There were); NO way that High blood sugars were associated with FOD's, (They are) and his "whining" for food was NOT associated with blood sugar levels! (It was and is). She also stated in the summary that she only did the Acyl Carnitine Profile test to "put the issue to rest!" She said that she expected it to be perfectly normal. I guess you could say we were looking for trouble when we decided to go on to the neurology dept. per the endocrinologist suggestions. At least he kept referring us. Maybe he was trying to get all of the proof he could get; I don't know. Whatever the reasons, I am glad he did.

It was now October 2001 and we had moved 40 minutes away to my husband's hometown, where he was opening his new business. We met with a new pediatrician who seemed to be kind of laid back but.. OK. Nothing outstanding; he would do. We filled him in on all of the happenings and he agreed to continue with his care. Following our move, we suspect that Caden's old pediatrician, called Social Services, because he told us he would if we switched Dr.'s! . Long story short, Social Services had their medical team review Caden's labs and found no evidence of any abuse but overwhelming evidence of a sick child! It had been 3 months since we had saw the geneticist and our neurology appointment was coming up. We wanted to know the results of the Acyl Carnitine Profile. I put in a call to the geneticist who was "putting this issue to rest". She returned my phone call with a scorn in her voice like no other I have ever heard. She kept insisting that he only had a "Red Flag" and that it meant nothing.

When we went in for Caden's neurology appointment, the neurologist ordered the results from the lab and lo and behold, there it was. A significantly abnormal Acyl Carnitine Profile that suggested MADD or GAIL. The neurologist noted several abnormalities, from hypotonia to hyporeflexia, and hyper mobile joints with loose ligaments, along with other distinctive abnormalities, including labs from his endocrinology work up. This was the very first doctor who took an interest in our son. She listened to us carefully and did NOT form an opinion based on others. From then on, the work up began, as she too felt he could have a FOD. He had a muscle and skin biopsy per her request.

After waiting, for what seemed like forever, we learned that his skin cells also suggested MADD/GAIL. We decided to go on to seek one other opinion, since we had proven there was an abnormality and we had at least one Dr. on our side. It was also a huge hassle to even go to this hospital where our son's wonderful neurologist was, due to the heat the other two doctors caused us. Before we could get to our second opinion, Caden got very sick. He had eaten peanuts and a hot dog. We had no idea that he was eating a deadly combination of foods. He woke up at 3 Am vomiting, on Jan. 27, 2002. He continued to vomit nonstop. By the time we got him to the hospital, he was very dehydrated. His heart rate was 190. He was so lethargic and here we were trying to tell the ER staff what we "thought" was wrong with him. His neurologist coordinated that hospital visit. She made sure he got the D10 and so on.

After he was discharged, he started having some stomach pains that were getting much worse by the hour. It was January 29, 2002 when we met with a wonderful geneticist. He was very compassionate and understanding. He laid it out to us plain and simple in terms we could understand. We learned a lot about what it could be, but still no definite answers. He had a thorough physical exam and blood work. We learned about his carnitine deficiency, and he had an abnormal urine acylglycine test, which also suggested GAIL. Caden was still not recovering from the stomach pains. They became much more severe. We were taking him to his new pediatrician and he was telling us to give him Maalox. He had even received copies of his reports indicating a metabolic disease. Still, he assured us that kids would typically have belly pain for at least a week after throwing up. We did give him Maalox for a week.

Caden had endured all he could by the second week and laid in our kitchen floor in pain. We immediately picked him up and took him to the largest, nearby hospital. It was determined by X ray that his intestines were swollen. They admitted him at once. He was on IV D10 and L-Carnitine for days. Caden wound up having a seizure like episode and hypoglycemic spell while there from taking him off of his D10 for an MRI. After a week of no answers, we asked to be discharged. We could give him Tylenol at home. That's all they were doing. We continued his oral Carnitor at home from this point on. We were told by the Pediatrician in charge that we may just have to live with his stomach pain! This was not acceptable! I still knew in my heart that my son had a FOD. It is now nearing the end of February 2002 and Caden is 3 yrs and 7 months old.

Caden was still sick with his stomach pain. He had now developed a high fever and vomiting. He was throwing up food that resembled an ESOPHAGUS! It was compacted food, 4 or 5 inches long and undigested. We took him to Children's, where his neurologist was, the next morning. He was admitted under an "on call Dr." that read his chart and made me out to be over anxious Mother, who was "inflicting anxiety on to my son and causing stomach pain!" Is this all anyone could come up with?! He has no idea what anxiety is! Anxiety is having a sick child that no one will help!

By now, Caden was sleeping 15 hours a day and sweating profusely. His liver enzymes were also elevated. He would sweat so severely we would have to keep switching his pillows. The GI doctor actually became a bit concerned at this point and mentioned some very atypical lymphocytes in his blood. They are often associated with the Epstein Bar virus or Leukemia. Luckily test proved he had neither. By now, we were in the very last days of March 2002 and Caden's new pediatrician had become aggravated with treating him. He asked us to pick up our medical records on our last visit. He had no pediatrician for 2 months before we could find an-

other.

April 2, 2002 we met with a new neurologist, one who specializing in metabolics. He agreed with Caden's first neurologist. He agreed that it was most likely GAI. He was started on Riboflavin and increased his dose of Carnitor. He had a thorough metabolic workup and another skin biopsy. The skin biopsy failed to grow. Luckily, his first neurologist still had some frozen skin cells and she sent it out for a specific GAI enzyme analysis. May 19, 2002, we received a phone call from Caden's original neurologist's nurse. She told me about his defective enzyme called the ETF-QO, that they located via his skin cells. She said that this defect causes the disease Multiple Acyl CoA Dehydrogenase Deficiency or aka. Glutaric Aciduria type II. The enzyme is responsible for the breakdown of fat and protein. His enzyme is only functioning at around 20 to 25%. The Multiple in MADD means that Caden has problems with multiple dehydrogenases, (i.e. short, medium and long chain fatty acids.). It also involves other dehydrogenases and all branched chain amino acids. I am unsure of whatever else is involved. When the endocrinologist, who accused me of MSBP, heard of Caden's diagnosis through his neurologist, I received a phone call from him, apologizing! I did not accept, but talked with him and urged him to never wipe a child under the plate again because of "waxing and waning" symptoms.

On our return neurology appointment, June 18, 2002, we officially had a diagnosis of GAI/MADD. We also learned that Caden's last urine analysis (prior his DX) was not good, but hopefully, after starting his low fat, low protein, high carbohydrate diet, things will improve with him. He will be having a blood protein check often to ensure he is receiving enough protein for proper growth and development. Too much protein is very toxic to his system and can lead to a metabolic crisis or death. The over accumulated fats can build up in the brain, heart and liver quickly, so diet is very important and used as the main treatment. He may also have to have G-tube surgery for low blood sugar and low body weight. We could not obtain a prognosis, which is a bit unnerving.

However, it is understandable, as so many complications can arise in the blink of an eye. It is very hard to predict the life span of someone who has such an unpredictable disease. Caden was only 25 days shy of his 4th birthday before he got a diagnosis! How could it have taken so long to uncover? It was not that this disease was so terribly hard to diagnose, it was a matter of finding the right doctor to do the right test.

The day he was diagnosed was the most bittersweet day of our lives. We were so busy thinking of all of the people we were going to say "I told you so, to," that we forgot to be sad. It was a very low moment but the stress and hopelessness had consumed our lives so, that we were lost for emotion. All we could think about was revenge! Now, a few months later, reality has set in we realize what we are dealing with (I think). We are also relieved. How could we be relieved? Knowing is better than Not knowing. At least now, we could begin a treatment plan to help our son.

Every day is a struggle. There are new issues to deal with every day and just when you think things are settling down, something new will come up or something old will resurface. We do have pity parties occasionally, but have to stop and remind ourselves that if we feel this tired and ran down, how must our son feel? We push ourselves that much harder, hopefully, encouraging Caden to do so.

Once we get things under control (if that is a possibility), I plan to work hard in advocating for New Born Screening. A simple heel stick that costs \$25.00 could have saved so much heartache. Caden is very lucky to have survived 4 years undiagnosed. Our job now, is to prevent further damage by staying on top of his diet, watching for early signs metabolic decompensation and learning everything we can about this disease. As you can tell, we are not a family that gives up. We will continue to battle this disease with the help of wonderful Specialists' like we have now. In addition, I urge anyone in a similar situation to find and join a support group. A support group will help you keep your sanity! You will find that there are other families out there dealing with similar situations. They may be on the other side of the world, but you will never feel the distance. The

information and support that we have received from the FOD Family Support Group is priceless.

Thanks for taking the time to read our son's story.

Mike & Krystena Richards

Parents to:

Caden Richards DOB: 7/13/98 GAI/MADD- Diagnosed: 6/18/02

Warren Prater - half sibling/7 yrs unaffected

Jaime Richards - half sibling/10 yrs unaffected

Carsen Richards- DOB 6/9/03 GAI/MADD