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THIS IS A PERSONAL STORY SHARED BY A FAMILY WHOSE CHILD HAS
MEDIUM CHAIN ACYL-COA DEHYDROGENASE DEFICIENCY (MCADD).

My son Isaac was born with a rare genetic disorder called Russell Silver Syndrome (RSS) and congenital hypothyroidism. RSS is a form of primordial dwarfism and even though he was born full term he weighed only 4lbs 12 oz and was 15" long. One issue with RSS is hypoglycemia so my husband and I have made sure that Isaac eats every 3-4 hours since birth. His hypothyroidism was detected by the state mandated screen and I was unaware that supplemental screening even existed.



Around 12 am Friday June 17, 2005 I heard Isaac crying and when I went to his room he had thrown up everywhere. My in-laws had a stomach bug the previous week and Isaac had spent some time over there so I figured he had gotten it. He threw up repeatedly all night long. I called the after hours nurse at my pediatrician's office and she told me to give him Pedialyte about 1 tablespoon every hour and to call the doctor in the morning. Isaac continued throwing up all day and I gave him 2-3 oz of Pedialyte every hour. I continued giving it to him through the night. At 4 am I had given him 3 oz of Pedialyte and I woke up around 5:15 am and he was gasping and shaking. I called 911 and the paramedics rushed us to a children's hospital in the area. He was lifeless and pale. They checked his blood sugar and it was a 4! Normal is at least 60. We were admitted to the hospital and the doctors thought that Isaac had a seizure due to the low blood sugar so they scheduled an MRI to check for brain damage. The MRI came back clean and through various tests they performed that week they found out that Isaac has MCADD. The doctors told me the startling news that it is a condition that is fatal if undiagnosed. They set up an appointment with a metabolic geneticist so we can learn how to handle this disorder.

Isaac was discharged from the hospital on Friday June 24th, 2005 and his doctor admitted that he had his "it's not your fault" speech prepared because he sincerely believed Isaac would have brain damage because of the seizure. He said that "someone up there was looking out for your little boy". I thank God that we found out before it was too late. I am 27 weeks pregnant with Isaac's little sister and she will have the extended newborn screen since she is at risk for MCADD.

As I look at Isaac laughing and playing with his toys I get choked up thinking about how we almost lost him and how a simple blood test told us about this deadly condition. One day I will tell him how the awful events of last week caused us to test his little sister... he may have saved her life.

Written June 27, 2005 by Mimi Garza
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