

MCAD - Carson and Brynn: A Family's Story

We live in Phoenix, AZ and have two kids with Medium-chain acyl-CoA dehydrogenase deficiency (MCAD), Carson (age 9) and Brynn (age 4). Carson was born in 2004 and at that time MCAD was not part of the newborn screening panel. He was healthy until his first crisis when he was 16 months old. He had been sick and was only eating small amounts, but was still drinking fluids. I let him sleep in the next morning and noticed something wasn't right when I woke him up. He was opening his eyes but not responding to me. I rushed him to the ER, which was only a few miles from our house. They took one look at him and rushed him back; it was the scariest day of my life. His blood sugar was 6 (normal blood sugar levels in children are usually between 60 and 100). After a week in the hospital and many tests, the doctor called to give us his diagnosis – MCAD. We had never heard of it before. His blood sugar was so low during his crisis that they said he could have had brain damage. We are very thankful that he survived that day and had no long-lasting effects. Carson had surgery this year and once again we were reminded how easily his body can decompensate. He went in for a tonsillectomy and was released from the hospital two days later. Once we got home, he stopped eating and drinking all together because it was so painful. He started decompensating and his blood sugar began to drop, so we called Dr. Aleck and headed to Phoenix Children's Hospital. He had to be on an IV for two days, and they kept him an extra day to ensure that he could eat and drink enough to keep his body fueled.

We knew with our second child that there was a 25% chance she would have MCAD. She was tested at birth and we received the results 4 days later. Brynn has been hospitalized several times each year because of her MCAD.

MCAD has changed our lives, but knowing how to treat it makes it manageable. We know how to handle illness at home and we know when it's time to head to the hospital. We know that we have to be proactive with our children's schools and caregivers in teaching them about MCAD and how our kids are different than other kids. Thankfully, it has been added to the disorders newborn screening tests for, and parents will know right away about the disorder and how to treat it.

We have a great metabolic team at Phoenix Children's Hospital and our pediatrician, Dr. Curran at East Valley Children's Center, is wonderful and helps us with whatever we need.

