

Sean: A Family's Story



On November 23, 2011, our baby boy, Sean, was born. He is our second child and we have been filled with joy ever since. We were able to go home four days after Sean was born. After being home for two days and just getting settled in with our little guy, we received a phone call from our pediatrician. He said that something had shown up on the first newborn screening test and that we needed to take Sean in for the second test right away. I called my husband to come home from work early and we took him in for the second test that day. I couldn't believe what the pediatrician told me. He couldn't explain everything, but I just knew that it couldn't be good. I was so heartbroken. No parent wants their child to be hurting or have to suffer through anything. The very next day, a geneticist called us from Phoenix Children's Hospital and explained that we needed to come to Phoenix Children's Hospital to get more testing to see whether Sean had a metabolic disorder. I remember being so confused, worried and unsure of all that was happening.

When we met with the geneticist, he explained to us that the newborn screening test showed that Sean might have a metabolic disorder called Glutaric Acidemia Type 1 (GA-1), where his protein intake needed to be limited. He explained that the test has high false positives to catch kids who might have it. They said that there are high false positives because to have a child slip through without knowing if they have GA-1 could cause great consequences, such as brain injury, coma and even death. So Sean had to get another test that specifically tested for GA-1. Sean was put on a restrictive diet immediately as a precautionary measure while we waited for the results of his test.

The genetics office was so wonderful about educating us on what to do while we waited for the results. We took every precaution with Sean since simple infections like an earache or a cold could cause brain injury, coma or death. The genetics team had protocols in place to help prevent these things from happening if he became sick.

A few months later, we received the phone call that confirmed Sean did have GA-1. This year our son will be celebrating his second birthday. He is a beautiful and joyful little guy.

We learned that the Arizona newborn screening test did not specifically test for GA-1 until 2006. That means they had only been testing for it for five years when Sean was born. We wouldn't have been able to take all the precautions if we hadn't known. We are very big advocates of newborn screening because it saved our son's life and we are so thankful to the test and those who made it possible for Sean to get it at birth.

