Newborn Screening Family Story:

Jennifer and Madison

"During my pregnancy with Madison, I consistently measured behind during my checkups. My 20 -week anatomy scan had shown no concerning issues. When I was roughly 30 weeks or so, I began to have thoughts that something was wrong and expressed that to my OBGYN. She assured me that I am a petite person, and that is the reason why I was measuring behind.

When I was 36 weeks 5 days pregnant, I went into labor. Madison had not turned around and she was breech. Due to this, I had an unplanned c section. She was born 5 pounds, 3 ounces and 17 inches. She was tiny.

A few hours after she was born, I was being wheeled away to my new room in the hospital when a nurse began saying "she's blue". I was tired and just had major surgery and this did not register in my mind. Madison was then taken for some tests, including newborn heart screenings.

The next morning, a doctor came in and sat on the edge of the bed. He informed me that Madison had a hole in her heart and that she needed a pediatric cardiologist immediately.

We got sent home a few days later and I began the search for a cardiologist. In the meantime, Madison's feedings at home were not going well. Her pediatrician at the time had me bring her in for daily weight checks. At 2 weeks old, she dropped down to 4 pounds 8 oz and her skin color was ashy. I will never forget this appointment- her doctor said I needed to take her to the children's hospital because they suspected failure to thrive. Stunned, trying to recover from major surgery and trying to figure out motherhood, I drove Madison to a children's hospital in Chicago. She was immediately admitted and diagnosed as "failure to thrive". While waiting for a room to open, a doctor came in and informed me that they suspected she had a genetic disorder called Di George. I had never heard of this and began to research feverishly.

"She had open heart surgery when she was 11 months old, and this is when she suddenly began to make strides!"

Due to her low weight, taking blood for tests was only limited to once a day. One of the tests ran was a genetic sequence called FISH that would be able to detect missing chromosomes. She was placed on a high calorie formula, and we were sent home.

Roughly 6 weeks later, I had gotten a phone call from the genetic counselor. I could not make an in-person appointment and asked for the results over the phone. I was floored when I was informed that she had two very rare genetic mutations called 3q29 deletion and 1q21.1 deletion. I was given a laundry list of roughly 30-40 different health and developmental issues to look out for. I was told that Madison is the only known case at the moment with both of those deletions together.

Shortly after this, she began to see her cardiologist. She was diagnosed with two hearts defects: ASD (the hole in her heart) and Pulmonary Stenosis. It was deemed mild at the time, and she required monthly checkups. Her first year was a whirlwind. Any time she got sick, she ended up needing to be hospitalized in the PICU. She was a "frequent flyer" and I got to know the doctors and nurses. At 2 months old, she underwent her first surgery for 2 hernias. At 4 months old, she got her first feeding tube. It was an NG tube, which is inserted through the nose. She began feeding therapy at this point as well. At 8 months old, she still was not making progress in feeding therapy and gaining the amount of weight her doctors wanted. She had a G tube placed; a feeding tube directly inserted into her stomach. It was at this point as well that her heart defects appeared to be worsening. At 10 months old, she had an appointment with cardiology and her doctor informed me that he needed to consult with the team on how to proceed, as she was still very tiny. I got a phone call a few days later that she needed to have open heart surgery to repair the two heart defects because she was facing the possibility of heart failure or a stroke. She had open heart surgery when she was 11 months old, and this is when she suddenly began to make strides! Since the open heart, she hasn't needed to be hospitalized when she gets sick, her hair began to grow, and she had a burst of growth in her development in speech and physical therapy!

The original genetic counselor said that she may not walk- she defied that odd and she began to walk at 18 months old, the same day she had tubes put in her ears for reoccurring ear infections!

"She is tiny...However, she is mighty."

At 2 years old, I applied for Madison to receive a grant for a feeding tube weaning program and she was accepted. I began the process of working with a team to wean her off the feeding tube. She had made a considerable amount of progress in feeding therapy at this point and we had the blessing from all doctors and specialists. She successfully weaned off the feeding tube by the age of 3 and had it removed at age 4! There's been a small list of diagnoses over the last few years, including ADHD and Microcephaly.

Today, she is 5 years old. She is tiny, she's only 27 pounds and roughly 31 inches tall. However, she is mighty. She begins Kindergarten in the Carmel Clay school district this fall. She has an IEP and still requires speech therapy. However, this little girl has overcome big obstacles and I am so proud to be her mom!"

Families are at the heart of all newborn screening efforts. Hearing from families is essential in spreading awareness about newborn screening and the conditions detected.

Do you have a story to share about how newborn screening has impacted you and your family's lives? Reach out to ISDHNBS@isdh.in.gov to share your story!

NEWBORN SCREENING
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To promote, protect, and improve the health and safety of all Hoosiers

Indiana Department of Health
2 North Meridian Street • Indianapolis, Indiana 46204 • 317-233-1325 • health.in.gov