Behind the Screens

Missouri Department of Health and Senior Services
Newborn Screening Program

Summer 2023 ★ Missouri Department of Health and Senior Services ★ Health.Mo.Gov ★ Volume 6 Number 2

Featured Disorder

Isovaleric acidemia (IVA) is considered an organic acid disorder. This is an inherited disorder that occurs when the body cannot breakdown certain parts of the proteins found in food. This condition consists of the buildup of toxins that can lead to serious complications known as a metabolic crisis. Untreated crises can lead to brain damage and even death. Early intervention and treatment is vital for children diagnosed with IVA to lead healthy lives. IVA affects one out of every 230,000 babies born in the United States.

There are two forms of IVA, acute and chronic/intermittent IVA which differ by age of onset. In the acute form, signs appear one to 14 days after birth. Acute attacks can be seen within the first days of life and include nausea/vomiting, refusal to eat, listlessness, and a sweaty foot odor. With chronic/intermittent IVA, the signs of IVA do not appear until later in infancy or childhood and lead to failure to thrive and developmental delays. These two forms share similar signs, but the signs of chronic/intermittent IVA are less severe than acute IVA in babies. There is no cure for IVA, management of symptoms includes a low protein diet with leucine restrictions. With these modifications and avoiding triggers of attacks, babies diagnosed with IVA have the greatest chance at normal growth and development.

Tech Tips

3 C's

Always remember the 3 C's when filling out the newborn screening blood spot form. To complete the form CLEARLY, COMPLETELY and CORRECTLY, including the mother, baby, and health care provider's full name and contact information.

Remember to list the pediatrician or primary health care provider who will care for the baby after discharge.

Patient Spotlight
AnnaRae

AnnaRae is our youngest daughter and was born on August 29, 2022. On Friday, September 2, we received a phone call from Children's Mercy Hospital in Kansas City that Anna's newborn screening had been flagged "high risk" for an Organic Acid Disorder, Isovaleric Acidemia. Both my husband and I were blindsided by this as our oldest daughter, MaryGrace, is a healthy toddler and we had never heard of the disorder. The genetics team insisted we make the drive to begin treatment immediately while waiting for the final confirmation of diagnosis. At this point in time, AnnaRae had very few signs of IVA, but we headed straight to the hospital. Upon arrival at the hospital, the nurses immediately drew blood for lab work and the genetics team began a lengthy discussion of what Isovaleric Acidemia is and how we will manage it for her entire life. When her lab work came back we were immediately transferred to the NICU as her ammonia and acidosis levels were too high and her body was spiraling into a metabolic crisis very quickly. We spent a short amount of time in the NICU before being transferred to the PICU for our Anna to be set up on dialysis to remove the harmful substances as fast as possible. When we woke up that Friday morning, we never imagined that by midnight our daughter would be in a coma in the PICU receiving life-saving medical attention. At that time we knew and understood next to nothing concerning Isovaleric Acidemia and were watching our newborn daughter fight for her life. Thanks to the incredible team at Children's Mercy, AnnaRae pulled through her first metabolic crisis. Now, Anna has a g-tube to help us safely administer her metabolic care and now at 6 months old she is growing and thriving despite her metabolic crisis and Isovaleric Acidemia.

AnnaRae still receives exceptional care from her metabolic team at Children's Mercy Hospital. Up until recently, we traveled there weekly for lab work and meetings with the genetic team, now we have spaced out her appointments to biweekly and hopefully as she grows, we can space them even more. The members of her team have ensured we are educated on Isovaleric Acidemia and have all the necessary tools/resources to care for our daughter.

The Missouri newborn screening truly helped save her life. The screening results came in early enough for us to be aware of the disorder and at a hospital with a team equipped to save her life. Without the results from her newborn screening we would not have had the knowledge of her disorder and we may not have been able to save her. I cannot stress the importance of this screening nor express the depths of our gratitude to the nurses who completed Anna's screening properly.

Ashley,
AnnaRae's mom

Did You KNOW?

The Missouri Department of Health and Senior Services, Bureau of Genetics and Healthy Childhood and the Office of Epidemiology launched an interactive dashboard displaying data about early hearing detection and intervention (EHDI) in Missouri.

The dashboard recaps data from 2015-2020 for any infants who have missed any step of the EHDI process and are considered lost to follow-up. To view the newborn hearing screening dashboard, please visit https://health.mo.gov/living/families/genetics/newbornhearing/dashboard-ehdi.php

For more information on topics in this edition, please visit:

- https://www.babysfirsttest.org/newborn-screening/conditions/isovaleric-acidemia
- https://rarediseases.org/rare-diseases/acidemia-isovaleric

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