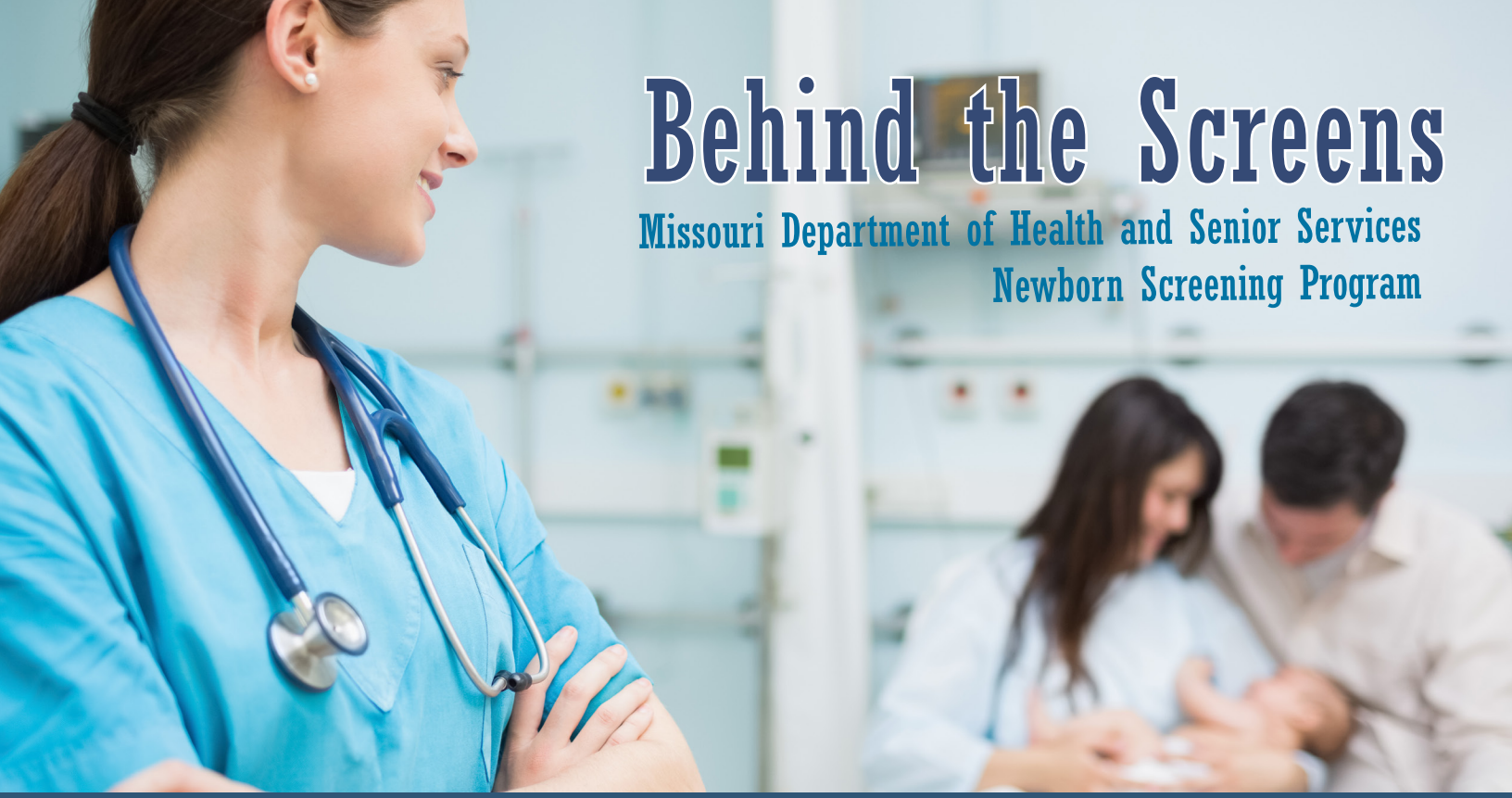

Behind the Screens

Missouri Department of Health and Senior Services
Newborn Screening Program

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Featured Disorder

Citrullinemia (CIT) is an inherited amino acid disorder that causes ammonia and other toxic substances to accumulate at high levels in the blood. While having a child with CIT is rare, when both parents are carriers, more than one child in the family can be affected with the condition.

The presentation of CIT falls on a spectrum that ranges from severe, to mild, or no symptoms. CIT type I, also known as classic CIT, is the most severe form, which presents in newborns during the first week of life. Patients with non-classic CIT type II can also have elevated citrulline but different clinical symptoms. People with CIT type II may have milder symptoms that appear later, presenting from childhood to adulthood. Sometimes, adult women with CIT may only present during pregnancy or right after delivery. There are also patients with CIT who may not develop high ammonia levels or any symptoms. CIT type I is the most common form of the disorder affecting about 1 in 250,000 newborns.

Affected infants typically appear normal at birth, but as ammonia builds up, they experience a progressive lack of energy (lethargy), poor feeding, vomiting, seizures, and loss of consciousness. Many of these signs may occur when the baby eats foods, or takes breast milk or formula, that their body cannot break down. The signs can be triggered by long periods of time without eating, illnesses, and infections.

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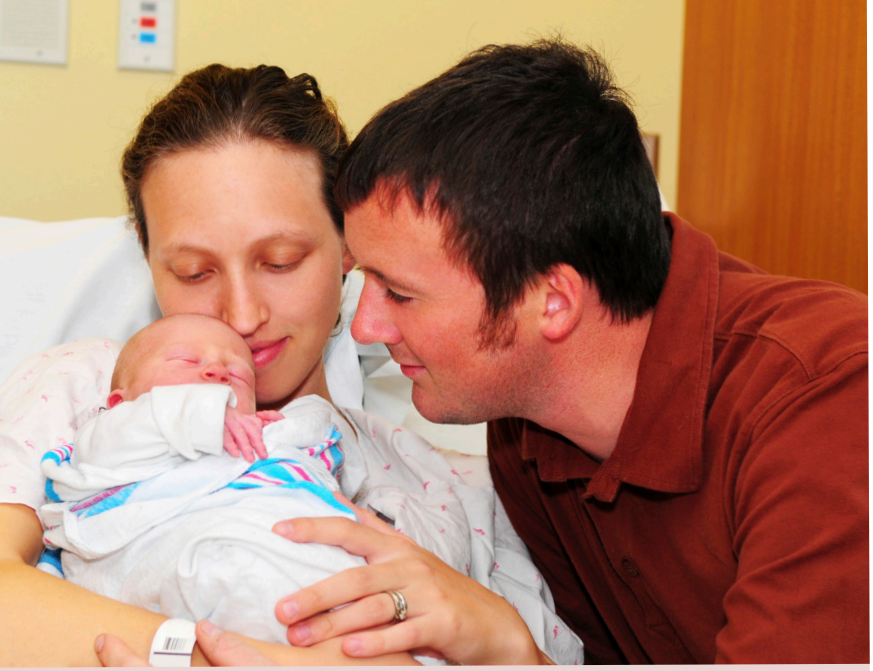
What's New?

The Newborn Screening Program on behalf of the Department of Health and Senior Services (DHSS) is excited to bring you "Discover DHSS." These are short videos on YouTube. The videos have been created to provide education and bring awareness to the various programs and services that are offered at DHSS.

Three videos have been created for the newborn screening program. The links to the videos are:

1. Discover DHSS: What we do at the Newborn Screening Program: https://www.youtube.com/watch?v=o25S0PeZXbQ
2. Discover DHSS: Meet the Scientist at the Newborn Screening Lab: https://www.youtube.com/watch?v=hgyja6vJ78c
3. Discover DHSS: Meet the Nurses at the Newborn Screening Program: https://www.youtube.com/watch?v=Ts67ra3RCvo



Did You KNOW?

Did you know there is a recommended list of conditions for which every baby should be screened? The Advisory Committee on Heritable Disorders in Newborns and Children works to set national guidelines to guide and support states in the development of their newborn screening programs. In order to keep screening up to date, they utilize parent advocates, organizations, and experts. The Committee's recommendations are reviewed by the Secretary of the U.S. Department of Health and Human Services.

The Committee and the Secretary work together to create the Recommended Uniform Screening Panel, or RUSP. The RUSP is not law, but simply a guide for states. Each state is able to choose which conditions it will screen for with guidance from the RUSP. The RUSP currently includes 35 core conditions and 26 secondary conditions. For more information about the RUSP go to https://www.hrsa.gov/advisory-committees/heritable-disorders/rusp/index.html.

Featured Disorder continued from page 1

When a baby screens positive for CIT on a newborn screen, the diagnoses needs to be confirmed through specific medical testing. Genetic testing can help identify the gene mutation in the family. Due to the rapidly increasing amount of ammonia and other toxins building up in the baby's system, it is critical the newborn screen be collected at the optimal time of collection, between 24 to 48 hours of age, to minimize the chances of brain and nerve damage caused by the high levels of ammonia.

Treatment for CIT includes medications to help remove excess ammonia from the blood. Dialysis can also be used to clear ammonia from the blood. Babies are given special low-protein formula instead of breast milk or regular formula. CIT can also be treated with a liver transplant that is typically done before one year of age.

Babies who receive early treatment for CIT can have healthy growth and development. This is why newborn screening for CIT is so important.

Missouri began screening all newborns born in Missouri in 2005. From 2005 to 2019, 11 babies have been confirmed to have CIT type I.

Tech Tips

- A pulse oximetry reading of 90-94% in BOTH the right hand and foot OR a difference of 4% or more between the right hand and foot when performing a critical congenital heart disease (CCHD) screening, should be repeated within one hour.
- Report hearing screening results no later than seven days from the date of screening. Timely submission of hearing screening results allow the Newborn Screening Program to quickly contact parents of babies who have either missed or failed the hearing screening.
- Instead of discarding a "ruined" or poor quality specimen, write VOID across the front of the screening form and return it to the Newborn Screening Laboratory along with the "good" newborn screening sample. You will be credited for the VOIDED specimen form.

PATIENT SPOTLIGHT

Meet Isabelle

"Our daughter was born May 9, 2017 - 6lb 15 oz, 19.5 inches long. She was perfect, and absolutely adorable as can be. Such a strong little lady. We took her to her five-day appointment and she had already surpassed her birth weight, which is a rare thing for breastfed-only babies. I was shocked to hear this because I had noticed she slept for really long periods of time. She was already sleeping through the night and would sleep 8 hours at a time during the day and she would fall asleep when it was time to eat. We went home after our appointment so proud to hear about our perfect baby. Later in the evening, I get a phone call from our pediatrician. He is a good friend of ours, so his call wasn't too odd. He proceeded to tell me that they received her newborn screen results (after her appointment) and that they had come back abnormal. Her results for her TSH - thyroid stimulating hormone - were pretty high. I am a nurse and I knew what that meant, medication for the rest of her life. Everything made sense in my head at that moment. The long napping and sleepiness, it fell into place. He requested we go get her labs rechecked ASAP, that night if possible. Tears streaming down my face I told her dad that we needed to take our daughter to the hospital to go get her labs drawn because of what the doctor had just told me. Her labs came back conclusive for elevated TSH again, off the charts elevated. You go through a short phase of denial, hoping it was just a fluke, but reality sets in (especially after tons of research) and medicine is the best choice. Rotating back to the newborn screen, had we not had her checked, our perfect child would not have been hitting milestones as she is now. As parents, you don't want to have anything wrong with your child, but I cannot even begin to describe how lucky we are to have something happen to our child that one: we caught early enough to treat, and two: is easily treated with medicine. The newborn screen provided us the way to catch and treat early enough. Had we waited two weeks or more after birth it would have been too late to treat effectively. We are very grateful for the newborn screen."

> "The newborn screen provided us the way to catch and treat early enough."

Thank you for your contribution to ensuring the best possible start for Missouri newborns.

MISSOURI DEPARTMENT OF HEALTH AND SENIOR SERVICES

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Newborn Blood Spot, Hearing, and CCHD Programs
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