Behind the Screens

Missouri Department of Health and Senior Services
Newborn Screening Program

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Featured Disorder

Propionic acidemia (PROP) is a genetic condition in which the body is unable to break down certain proteins and fats. Normally, when food is ingested, enzymes help break the food down into amino acids. One enzyme that breaks down those amino acids is propionyl-CoA carboxylase (PCC). Individuals with PROP either do not make enough or make non-working PCC enzymes. When PCC is not working correctly, harmful levels of toxins build up in the body.

PROP affects one in every 35,000 to 75,000 babies born in the United States. It is most commonly found in people from the Inuit population of Greenland, some Amish communities, and Saudi Arabia. Each year, in Missouri, there are typically one to two cases of PROP identified through newborn screening. The harmful effects of PROP can occur just days after birth, which is why early detection through newborn screening is key. Some early signs of PROP include: poor feeding, vomiting, tiredness, poor muscle tone, and sleeping longer or more often.

Children with PROP will be put on a carefully planned diet from birth. Special formulas are available for those with PROP and are often used throughout adulthood. L-carnitine and biotin supplements may be prescribed. These natural compounds, which help to break down fats and proteins, may not be found in a child with PROP. If left untreated, babies with PROP can die within the first year of life. Treating PROP early can help reduce some of the early complications of the condition and children can experience normal growth and development. However, some children receiving treatment can still experience learning disabilities or developmental delays. Early detection, intervention, and treatment accomplished through newborn screening gives these children the greatest chance at having a positive outcome.

What's New?

On January 1, 2018, the Missouri State Public Health Laboratory (MSPHL) began formally reporting Krabbe disease on laboratory reports. Although every baby born in Missouri has been screened for Krabbe since 2012, those results have not previously been available on the newborn screening laboratory reports. The newborn screening laboratory reports are available to submitters and primary care providers after the newborn screening tests have been completed. These reports not only outline the newborn screening results, but they also provide health care providers with comments regarding any recommended action such as a repeat screen or consultation with a specialist.

Now that the MSPHL is formally reporting Krabbe disease screening results, you may see some new comments on the newborn screening laboratory reports unique to Krabbe disease.

Continued on page 3

Did You KNOW?

Did you know cytomegalovirus (CMV) infection is the leading non-genetic cause of hearing loss at birth? A woman who develops an active CMV infection during pregnancy can pass the virus to her baby. While CMV is a common virus that harmlessly affects people of all ages, CMV during pregnancy may cause severe health complications to an unborn baby including cognitive impairment, cerebral palsy, developmental delay, and vision and hearing loss.

Every year, more than 40,000 women in the U.S. experience CMV infection during pregnancy. One in 150 babies is born with congenital CMV. One in every five children born with CMV suffers lifelong disabilities.

A woman who is pregnant or planning to become pregnant can protect her baby from CMV by protecting herself.

The best way to avoid CMV is hand washing after changing a diaper and after wiping a child's nose, mouth, or tears. DHSS provides a no-cost brochure titled "Cytomegalovirus (CMV) Infection: What women who are pregnant or plan to become pregnant need to know." (http://health.mo.gov/living/families/genetics/newbornhearing/pdf/CMVBrochure-Bifold-071317.pdf). Consider making this essential information available to the mothers of the newborns you screen.

The brochure can be ordered here: http://health.mo.gov/living/families/genetics/newbornhealth/orderform.php.

Tech Tips

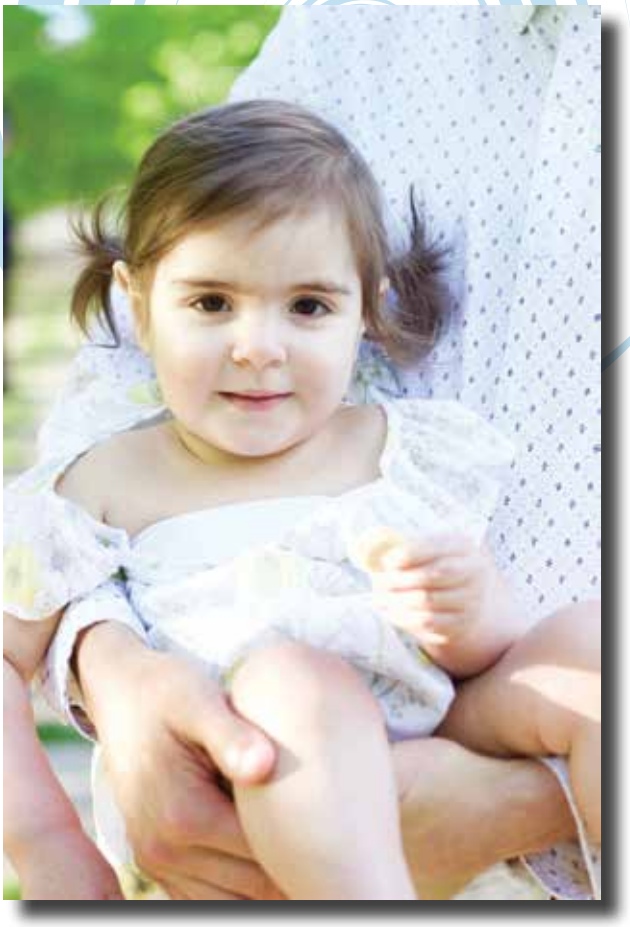
- It is important to ensure that the mother's address and phone number are current and correct when filling out the blood spot collection form. This information is vital when a family needs to be contacted to seek intervention for a high risk result on the newborn screening.
- Adequate blood flow is needed to obtain an accurate pulse ox reading. When performing a pulse ox, make sure the infant's extremities are warm and never attempt to obtain a pulse ox reading on the same extremity that you have an automatic blood pressure cuff on.
- The initial hearing screening results entered into MoEVR should be from the last hearing screening performed prior to discharge from the hospital. For example, if a "refer" result is obtained twelve hours after birth and a "pass" result is obtained forty hours after birth, enter the "pass" result into MoEVR.

THANK YOU

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

PATIENT — SPOTLIGHT

Arrow



Arrow is a Missouri child who was diagnosed with a congenital disorder as a result of newborn screening. Without the timely collection, transport, and testing of Arrow's newborn blood spot specimen, she may not have had the chance to grow into the happy, healthy child she is today. Arrow's father has graciously shared her story with us:

"In early 2015, my wife and I welcomed our daughter, Arrow, into our family. She was our third child that we had birthed at home and like our first two children, everything had gone really smooth. Around day ten, we received a phone call from our midwife that the Missouri newborn screening test had detected an extreme deficiency in her thyroid levels. We were instructed to immediately take Arrow to Children's Mercy in downtown Kansas City, where she was diagnosed and treated for congenital hypothyroidism. If this condition had not been caught and treated in the first two weeks of Arrow's life, she could have suffered severe intellectual disabilities and growth problems, with irreversible damage. We thank God for the Missouri newborn screening program that surfaced an illness that was invisible to us – now Arrow will live to her full potential."

> **"We wanted to share Arrow's (story) because we believe the newborn screening saved her life."**

Continued from page 1

These comments and recommendations might look a little different from the ones you are used to seeing due to the complexity of this disorder. If you have any questions or concerns regarding Krabbe newborn screening results, please feel free to contact the Newborn Screening Program or the MSPHL.

For additional information, phone numbers for Missouri's genetic tertiary referral centers can be found at http://health.mo.gov/living/families/genetics/referralcenters.php.

MISSOURI DEPARTMENT OF HEALTH AND SENIOR SERVICES

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Newborn Blood Spot, Hearing, and CCHD Programs
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