



Behind the Screens

Missouri Department of Health and Senior Services
Newborn Screening Program

October 2018 ★ Missouri Dept. of Health and Senior Services ★ www.health.mo.gov ★ Volume 2 Number 4

Featured Disorder

Medium-chain acyl-CoA dehydrogenase deficiency (MCAD) is an autosomal recessive genetic condition in which the body is unable to convert some fats into energy. Fatty acids are chain-like acids that come in varying lengths – short, medium, long, or very long. An enzyme called medium-chain acyl-CoA dehydrogenase is responsible for breaking down medium-length fatty acids. In babies with MCAD, there is either not enough of this enzyme or it does not work properly. This becomes dangerous because a baby’s body and heart needs fat for energy when sugars are gone, such as between meals.

MCAD affects approximately one in every 15,000 babies born in the United States and is more commonly seen in those of northern European ancestry. MCAD symptoms usually begin in infancy or early childhood, but a matter of hours can make all the difference for these babies. Many symptoms occur when an affected baby eats food that his or her body cannot break down. They can also be triggered by long periods without eating, illnesses, and infections. The first signs of MCAD include sleeping longer or more often, irritable mood, poor appetite, fever, diarrhea, low blood sugar, and vomiting.

Babies with MCAD will need a restricted diet to avoid the types of fats their bodies cannot break down. Specialists can assist families in planning a high-carbohydrate, low-fat diet that still provides affected babies with the nutrients they will need for healthy growth and development. Eating often and regularly can also help prevent babies with MCAD from experiencing symptoms. L-carnitine supplements might be prescribed to help break down fats and remove harmful substances. If left untreated, children with MCAD may experience breathing problems, seizures, brain damage, coma, and possibly death. Early screening and treatment can lead to normal, healthy lives for babies with MCAD.

What’s New?

Thanks to newborn hearing screening, identification of infants who are deaf/hard-of-hearing (DHH) happens quickly. Parents are thrust into a new reality involving many professionals, complex decisions about medical interventions and conversations about communication options. Studies indicate that being engaged with another parent of a child who is DHH positively influences the emotional well-being, confidence and competence of a parent new to parenting a child who is DHH.

The Missouri Department of Health and Senior Services’ (DHSS) Newborn Hearing Screening Program recently joined with the DHSS’ Family Partnership to provide parent-to-parent support to all families with children who are DHH. The Family Partnership Program provides no-cost information and support to children and youth

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Did You KNOW?



The goal of the Missouri Newborn Screening Program is to ensure all babies are screened shortly after birth. However, Missouri law does allow parents the right to refuse screening. As a front-line team member of newborn screening, your role is paramount in providing accurate and up to date information to ensure families understand the value of early detection and intervention. It is vital that parents have an understanding that newborn screening can detect over 70 different, serious disorders, including hearing loss and critical congenital heart disease (CCHD). Providing education will help parents make informed decisions.

Free educational materials are available at health.mo.gov/newbornscreening and the sample storage policy can

be found on the top, white, detachable sheet of the newborn blood spot collection card. If parents choose to refuse newborn screening after being fully informed, it is your responsibility to document the refusal per your facility's policy. Then fax a copy of the refusal stating which screenings are being refused with the parent's signature, baby's full name, date of birth, mother's name, and birthing facility to 573-751-6185, attention: Newborn Screening Follow Up.

Tech Tips

- When submitting CCHD paper reports on babies that have been transferred into your facility, please remember that reports are only required on those babies born in Missouri. If a baby was born in another state and then transferred into your care, their CCHD results do not need to be reported to Missouri's Newborn Screening Program.
- All ill and premature newborns require a repeat blood spot screen between 7 and 14 days of life. All newborns less than 34 weeks gestation or less than 2000 grams at birth require a third screen at 28 days of life. Please review Missouri's guidelines at <https://health.mo.gov/lab/newborn/pdf/nicuguidelinesandkeyfornbs.pdf>.
- In MoEVR, Discharge Disposition must be entered on the hearing screening page. This field assists the Newborn Hearing Screening Program in locating babies who may have failed or missed the newborn hearing screening or have risk factors for late-onset hearing loss. If an infant is still hospitalized after the hearing screening is completed, enter "Home" if he/she will likely go home or "Unknown" if he/she is unlikely to be discharged to home.

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with special health care needs and their families. The Family Partnership's new Family Partner for Families with Children who are DHH is the parent of a child with hearing loss. She contacts families with newly diagnosed infants to provide non-biased information about communication opportunities, explain the benefits of early intervention, and share her parent perspective. The Family Partner is available to provide emotional support and explore options and solutions with any Missouri family that has a child who is DHH. To reach the Family Partner for DHH, call the Family Partnership at 1-800-451-0669.



PATIENT — SPOTLIGHT

Julianna and her family's lives were forever changed when she was diagnosed with PKU through newborn screening. Now, nine years later, Julianna is living a life like any other girl her age. Julianna is a perfect example of how early detection and intervention can make all the difference in the life of a child. Her mother, Jaci, said this:

“Early in my pregnancy I faced the age old question – do you want a boy or girl? My answer, like so many other expectant mothers, was that all I wanted was a healthy baby. My husband and I took our new baby girl, Julianna, home four days after a scheduled cesarean section. She was healthy, just slightly jaundiced. At five days old, we took her to see the pediatrician for the first checkup.

Everything checked out fine at that appointment. Two hours after returning home, I received the phone call that changed everything. Julianna's pediatrician apologized for giving me information over the phone since we had just been in his office, but Julianna's heel stick test came back positive for Phenylketonuria (PKU). I believe I asked him at least four times how to spell Phenylketonuria as I jotted down the phone numbers to her new genetic specialist.

I was scared, confused, and sad all at once. I didn't know at that time she had had a newborn screening completed when she was in the hospital after being born. I certainly had never even heard of Phenylketonuria in my life.

The following week I felt like I took a crash course on PKU – learning from Julianna's genetic doctor, counselor and nutritionist how to properly care for a child with PKU. That first year I was an emotional wreck. I had a beautiful baby girl who looked healthy, but made weekly trips to the hospital for blood tests. I learned how to weigh and prepare her medical formula, as well as how to figure out her daily Phenylalanine intake once she started eating baby food. Any blood test result that wasn't within the correct range would send me into a frantic state of worry and confusion. I was looking for any signs of abnormal child development.

Gradually, I learned what a great gift the newborn screening has been to us all. I was able to get Julianna on a low protein diet her first week of life. She continues to enjoy drinking her medical formula and complies very well



Julianna

with her low protein diet. She's learned how to make her own formula, how to cook and bake low protein foods, and how to weigh her food and record them on her food diary.

Julianna is 9 years old and has a friendly, sweet and caring personality who shows no signs of cognitive delay or intellectual deficits. She enjoys school and has been a member of the local YMCA swim team for five years. I've watched her grow and develop into a happy, healthy young child.

Although she was born with a rare genetic metabolic disorder, PKU does not define her. I am forever grateful she had that newborn screening test. Without it, I wouldn't have been able to say I have a healthy daughter, which is all I ever wished for to begin with.”

“**Although she was born with a rare genetic metabolic disorder, PKU does not define her.**”



MISSOURI DEPARTMENT OF HEALTH AND SENIOR SERVICES

Bureau of Genetics and Healthy Childhood
Newborn Blood Spot, Hearing, and CCHD Programs
573.751.6266 or 800.877.6246

Missouri State Newborn Screening Laboratory
573.751.2662

www.health.mo.gov/newbornscreening

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An EO/AA employer: Services provided on a nondiscriminatory basis.
Hearing- and speech-impaired citizens can dial 711.