Behind the Screens

Missouri Department of Health and Senior Services

Newborn Screening Program

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Featured Disorder

Short-Chain Acyl-CoA Dehydrogenase Deficiency, or SCAD, is a fatty acid oxidation condition in which the body cannot break down certain fats. Individuals with SCAD are unable to convert fats into the energy the body needs to function. This autosomal recessive genetic condition affects approximately 1 in every 40,000 to 100,000 newborns.

Symptoms of SCAD are often triggered by long periods of time without eating or from recent illness or infection. Symptoms include sleepiness, irritability, poor appetite, muscle weakness, learning delays, fever, diarrhea, vomiting, seizures, low blood sugar, and trouble breathing. Treatment focuses on dietary adjustments and the use of supplements and medications. Treatment is collaborative, including the primary health care provider and genetic specialist. Nutritionists can also help identify the foods those with SCAD need to avoid. In addition to eating often and taking Vitamin B2 supplements (Riboflavin), some with SCAD take prescription L-carnitine supplements. L-carnitine supplements can help the body break down fats and eliminate harmful substances. Newborn screening of SCAD can help identify and treat the disease early, preventing the serious health outcomes of SCAD and allowing children to grow up healthy and with typical development. Resources for those affected by SCAD can be found at fodsupport.org.

Patient Spotlight

Phoenix

Pictured are Phoenix and her parents, Stephanie and Jeremy.

"My husband, Jeremy, and I would like to share our journey that we have experienced with our daughter Phoenix. She was closely monitored during the last four months of my pregnancy due to intrauterine growth restriction (IUGR). I was induced on March 29, 2024, and I had Phoenix via C-section at 8:23 in the morning on April 2, 2024. She weighed 5 pounds 0.8 ounces at birth and was 18 inches long; she was very tiny. She spent 13 days in the NICU for various reasons, during which she failed her newborn hearing test multiple times in her right ear. It was also discovered that she had elevated bilirubin levels and a heart murmur that were consistent with a family history (Phoenix's older sister, my husband's oldest) of Alagille syndrome (a rare genetic disorder that affects the bile ducts in the liver, as well as the heart, ear and more).

After we were discharged, we returned to the hospital a couple weeks later to attempt the hearing screening again in hopes that her ear canals had grown, and she would pass the hearing test. She failed it again, and they gave us the option of coming in again or being referred to an audiologist. We took the referral. At seven weeks old, Phoenix had her first Auditory Brainstem Response test. It was discovered that she has severe/profound hearing loss in her right ear. She has no hearing loss in her left ear. She had her second Auditory Brainstem Response test at 2 months old to confirm the results, after which we were referred to Missouri First Steps.

Through First Steps we were introduced to The Moog Center for Deaf Education, where we transferred her audiology services to. We have been trialing the Ponto System as her first hearing aid. The Ponto is a bone conduction hearing aid attached to a band that she wears on her head on the right side. The Ponto works by hearing the sound with a microphone on the side with hearing loss and sending it to the hearing ear. This provides all the sounds, but not necessarily where the sound is coming from. We are getting ready to switch to the more traditional style (behind-the-ear) hearing aid in February 2025. Phoenix has enough of a loss to one day possibly have a cochlear implant. We are just taking it a hearing aid system at a time to find what works best for her and our family. If not for newborn screening tests, we wouldn't have caught her hearing loss so early. We are very lucky to have a great team of doctors and audiologists to help us navigate through this to make sure that Phoenix gets everything she needs to thrive."

Stephanie, Phoenix's Mom

IN the NEWS

As mentioned in our previous newsletter, the Missouri State Public Health Laboratory and the Newborn Screening Program have updated the newborn blood spot collection form to include critical congenital heart disease (CCHD) screening results. Once you receive an order including the new forms, use them to report CCHD results. Until then, continue using the Missouri Electronic Vital Records reporting system.

This change does not affect newborn hearing screening reporting or birth certificate submission. If you do not have access to MoEVR and instead use the Department of Health and Senior Services CCHD Reporting Form, please use the new blood spot collection form to report results as a replacement for the paper form. If you are not submitting a blood spot sample but need to report CCHD results, use the electronic CCHD reporting form available at Health.Mo.Gov/cchd.

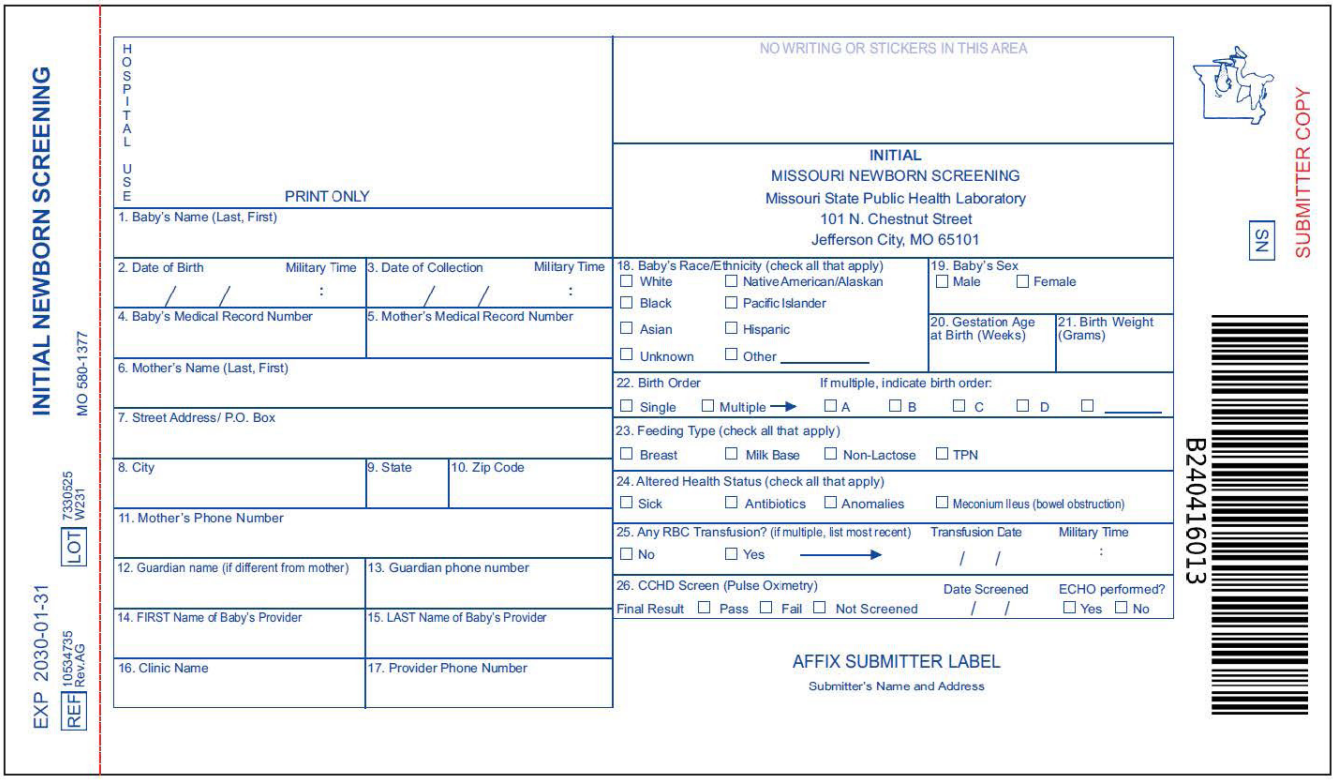
The new blood spot collection form will include a box to record the date of the CCHD screening, the pass or fail result, and whether an echocardiogram was performed. Pulse oximeter readings will no longer be reported to the Newborn Screening Program. CCHD screening should be conducted according to the most current American Academy of Pediatrics recommendations, which were recently updated at the end of 2024 and are detailed in the journal article "Newborn Screening for Critical Congenital Heart Disease: A New Algorithm and Other Updated Recommendations: Clinical Report," published in Pediatrics, January 2025, Volume 128, No.1.

More information about Missouri's Newborn Screening Program visit Health.Mo.Gov/newbornscreening, For questions, please contact Lori Swartz at 573-751-6266 or lori.swartz@health.mo.gov.

Provider Tip

In 2024, the Missouri State Public Health Laboratory (MSPHL) received 1,811 poor-quality newborn screening samples, each delaying lifesaving care a baby may need if it is found to have a disease screened for by Missouri. Poor-quality specimen examples include an insufficient quantity of blood, incomplete saturation of the filter paper, and overfilled, supersaturated, layered, or diluted pre-printed circles. Poor quality specimens may also occur due to issues in the drying or transportation processes. A reliable resource for collecting acceptable blood spot specimens is the Clinical and Laboratory Standards Institute (CLSI). They offer a Dried Blood Spot Specimen Collection for Newborn Screening manual for purchase. They also offer free learning resources on their YouTube channel. For more information on the Missouri State Public Health Laboratory, visit Health.Mo.Gov/lab/newborn. If you require additional assistance with troubleshooting poor-quality samples, please contact the Newborn Screening team at 573-751-6266.

Completing A New Blood Spot Collection Form



1. Baby's Name: As it appears on the medical record.

2-3. Date of Birth and Date of Collection: Both are critical for results. The date of collection box cannot be amended if is incomplete or inaccurate.

6. Mother's Name: Birth mother's legal name.

7. Street Address: Address where baby will live.

12-13. Guardianship Information: Only use if someone other than birth mother will have legal guardianship of baby.

14-17. Provider Information: First and last name, clinic name and phone number for provider who will care for baby after discharge. If the baby is in the NICU, use the neonatologist.

18. Baby's Race/Ethnicity: Race as mother would identify and list "unknown" if you aren't sure.

20. Gestational Age at Birth: Length of pregnancy, listed in weeks.

21. Birth Weight: Birth weight, not adjusted weight or current weight.

24. Altered Health Status: Mark "Sick" if baby is in NICU. Mark "Antibiotics" if given to baby, not the mother, in the last 48 hours. Mark "Meconium Ileus" if baby has a bowel obstruction. This is different than meconium-stained fluid.

25. RBC Transfusion: Report date and ending time of most recent transfusion.

26. CCHD Screen: Critical congenital heart disease (CCHD) is now being reported on this form. Pulse oximetry numbers no longer need to be reported.

MISSOURI DEPARTMENT OF HEALTH & SENIOR SERVICES

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Newborn Blood Spot, Hearing and
Critical Congenital Heart Defects Programs
573.751.6266 or 800.877.6246
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