Behind the Screens

Missouri Department of Health and Senior Services
Newborn Screening Program

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Featured Disorder

Krabbe is a lysosomal storage disorder (LSD). LSDs cause a buildup of toxins in the body resulting in enzyme deficiencies. Krabbe disease occurs when there is a mutation in the gene that encodes for an enzyme called galactocerebroside beta-galactosidase (GALC). GALC is necessary for myelin production. Myelin surrounds and protects the nerves in the body. When the body does not produce enough GALC to create the myelin properly, a toxin is formed instead. This toxin causes death of brain cells and nerves.

Krabbe occurs in approximately 1 out of every 100,000 individuals in the United States. In August 2012, Missouri began screening all babies for Krabbe through the newborn screen.

Krabbe is classified two ways; early onset and later onset. The early onset form of Krabbe begins in infancy and has three stages that progress from general irritability, to deterioration of mental capabilities and muscle stiffness, and finally to complete blindness, deafness and eventually death. The later onset type of Krabbe begins in childhood or early adulthood. These symptoms have a slower progression and include vision problems, hearing loss and difficulty conducting basic movements. Muscle rigidity may also be present. The late onset form has variable conditions and there is no definitive set of stages as there are with the early onset form.

Unfortunately, there is no cure for Krabbe. Bone marrow transplant and umbilical cord blood stem cell transplant have been found to be the most effective treatment options when conducted before the onset of symptoms. Early identification and treatment may prevent some of the serious health issues caused by the condition and improve quality of life.

Visit https://www.babysfirsttest.org/newborn-screening/conditions/krabbe for more information about Krabbe.

What's New?

Submitter Recognition Certificates: Certificates of recognition were sent out in September to recognize those facilities that have collected a newborn blood spot screen resulting in a confirmed diagnosis. These certificates will be mailed out on a quarterly basis. If you receive one of these certificates, we hope you will proudly display them to highlight your hard work and commitment to newborn screening.

Coming soon! Hospital newborn hearing screening programs that conduct outpatient rescreenings will soon be able to submit rescreening results electronically through the Missouri Electronic Vital Record (MoEVR) system. Gone are the days of paper "Hearing Screen Only" forms! Bulky mailings and endless faxes will be a thing of the past! The change to electronic reporting will save time and make it easy to enter and submit your rescreening results to

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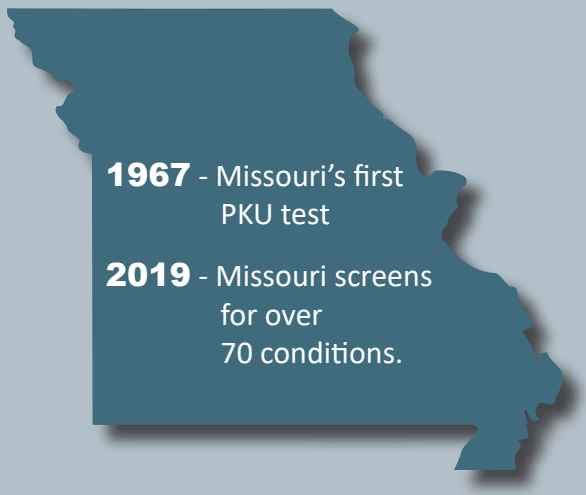
Did You KNOW?



Did you know, newborn blood spot screening (NBS) is more than a phenylketonuria (PKU) screen? NBS began in the 1960s with a screen for PKU (Missouri 1967). Today, Missouri screens for over 70 conditions. Through the years as conditions continue to be added to the screening panel, many healthcare professionals still use the term "PKU test." This term is no longer accurate. Using the term "PKU test" creates confusion for both parents and clinicians. Newborn blood spot screen has become the accepted term used to describe the collective group of conditions screened for at birth.

You may ask, "Why do the words matter?" Here is an example from Baby's First Test of just how important word choices are. "A family received an urgent call informing them that their child needed follow-up for an abnormal newborn screen. Arriving at the lab, a staffer came up to them and said 'You must be the people here for the PKU test.' Returning home, this family spent all weekend researching PKU. Arriving at the metabolic clinic, they were surprised to find their newborn was not suspected of having PKU at all. Instead, their child was actually at risk for an entirely different condition. Clinic staff had to spend a lot of time helping the family 'unlearn' all the information they had gathered, a time-consuming task which could have been avoided had the correct terminology for newborn screening been used from the beginning."

Learn more about what conditions Missouri screens for at https://www.babysfirsttest.org/newborn-screening/states/missouri.

Tech Tips

- Bright or infrared light, including bilirubin lamps and surgical lights, can affect the accuracy of a pulse ox reading. Ensure that the infant is not placed in bright or infrared light while critical congenital heart disease (CCHD) screening is being performed. You may cover the pulse ox sensor with a blanket to ensure that extraneous light does not affect the accuracy of your reading.

- It is imperative to completely and accurately document dates and times on the NBS collection form. The date and time of birth as well as the date and time of sample collection is crucial for accurate screening results. Many of the screening tests are dependent on the baby's age at collection. If these data elements are not provided on the NBS collection form, the results cannot be accurately determined and the baby will have to have another screen collected. This decreases timely identification of disorders that could threaten a baby's health. Always double check the completion of the information on the blood spot form.

- Remember to submit your hearing screening and hearing rescreening results to the Missouri Department of Health and Senior Services Newborn Hearing Screening Program (MNHSP) within seven days of the screening. Timely submission of results means parents don't receive unnecessary reminders in the mail from the MNHSP. To review how to report hearing screening results, visit https://health.mo.gov/living/families/genetics/newbornhearing/guidelines.php.

PATIENT Spotlight

"Our Hallie Jo was diagnosed with Isovaleric Acidemia (IVA) at 4-days old. It was found through the newborn screening that they did at the hospital. Our pediatrician called and had us go to the hospital for additional lab work. They said she had a mild case, but that she would need a few extra supplements to keep her levels balanced and to make sure that her body was breaking down protein correctly. It was extremely scary in the beginning, but she continued to do well and didn't have any metabolic crisis that many babies with IVA do.

Almost 12-months later, her geneticist and dietician gave us the fabulous news that she no longer needed any supplements! They believe she is just a carrier of the IVA gene and unless we notice any symptoms, she doesn't need to have any diet restrictions. Although our case is very different than most since she no longer has to be treated, we are very happy that our doctors were very cautious and that the newborn screening flagged possible health concerns."

Katie Royal (mom)

> "...we are very happy that our doctors were very cautious and that the newborn screening flagged possible health concerns."

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the Missouri Department of Health and Senior Services Newborn Hearing Screening Program (MNHSP). The new electronic rescreening form will be found on a tab next to the initial hearing screening form in MoEVR. Three new fields will be added to the rescreening form: 1) Was a referral made to an audiologist? (yes/no); 2) If yes, a text field will appear to enter the name of the audiology clinic; and 3) If yes, a date picker will appear to enter the date of the appointment. This information will serve to improve MNHSP's follow-up efforts. The MNHSP will send an email to all hospital hearing screening program managers when the electronic rescreening form is ready for use. Until then, if you need to update your contact email with the MNHSP, send your new email address to Marie Duggan at Marie.Duggan@health.mo.gov.

MISSOURI DEPARTMENT OF HEALTH AND SENIOR SERVICES

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Newborn Blood Spot, Hearing, and CCHD Programs
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Missouri State Newborn Screening Laboratory
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