Behind the Screens

Missouri Department of Health and Senior Services
Newborn Screening Program

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Featured Disorder

Maple Syrup Urine Disease (MSUD) is a type of amino acid condition. The body is unable to break down certain amino acids, the building blocks of proteins. MSUD is named for the sweet smell of the urine of untreated babies. Early detection and treatment are key in preventing severe outcomes of the condition.

MSUD is an autosomal recessive condition. A baby must inherit two copies of the non-working gene for MSUD, one from each parent, in order to have the condition. MSUD affects one out of every 185,000 babies worldwide. It is found in specific ethnic groups. Approximately one out of every 380 babies in the Old Order Mennonite population is affected by the condition. MSUD is also common in people of French-Canadian ancestry and Ashkenazi Jewish ancestry. Missouri began screening for MSUD in 2005.

Classic MSUD is the most common form. Symptoms develop shortly after birth and can include poor appetite, feeding difficulties, weight loss, lethargy, high-pitched cry, irritability, and sweet-smelling urine. These can be triggered when baby consumes food that their body cannot breakdown, illnesses and infection, or long periods of not eating.

Treatment options for MSUD include dietary treatments, supplements, and medications. A child diagnosed with MSUD may need to be on a protein-restricted diet. This may include special formula and foods. These will likely continue into adulthood. Thiamine supplements are helpful for those babies determined to have a type of MSUD that is thiamine-responsive. The primary care provider will be able to help determine this.

If MSUD is treated early, children can have healthy growth and development. Some may still experience signs of MSUD even with treatment. Babies who do not receive treatment for MSUD are at risk for brain damage, coma, or death. Learn more by visiting https://www.babysfirsttest.org/newborn-screening/conditions/maple-syrup-urine-disease-msud.

What's New?

New and Improved Education Materials

The Bureau of Genetics and Healthy Childhood is working on updating some of our education materials. Be on the lookout for these new items to be arriving in your mailboxes soon!

Please utilize our Newborn Screening webpages:

Newborn Screening Laboratory
health.mo.gov/lab/newborn

Newborn Blood Spot Screening
health.mo.gov/newbornscreening

Newborn Hearing Screening
health.mo.gov/newbornhearingscreening

Newborn CCHD Screening
health.mo.gov/cchd

Did You KNOW?

Did you know many preterm, sick, or low birth weight infants require more than one newborn blood spot screening to ensure accurate testing? Sick babies, babies born less than 34 weeks gestation, or babies with a birth weight less than 2000 grams often have certain medical problems that require special treatments. Special treatments such as total parenteral nutrition (TPN) and red blood cell (RBC) transfusions affect the newborn blood spot screening results. Prematurity, even without other complications, can cause false positive or even false negative blood spot screening results.

The optimum time to collect a blood spot sample for a healthy, term baby is 24-48 hours of life. For sick or preterm babies, the recommended collection times differ to ensure accurate and complete results are obtained taking into consideration treatments they may receive and their gestational age.

To learn more about the Missouri Guidelines for premature, low birth weight, sick or NICU infants, visit https://health.mo.gov/lab/newborn/pdf/nicuguidelinesandkeyfornbs.pdf. For all rules regarding newborn blood spot collection in Missouri, please visit https://s1.sos.mo.gov/cmsimages/adrules/csr/current/19csr/19c25-36.pdf.

Tech Tips

- The NBS test is not run "stat," however you should contact the MO State Public Health Laboratory by phone at 573.751.2662 if results from a particular test are needed as soon as possible.
- Each hospital should designate a person responsible for carrying out the newborn hearing screening program at their facility. This person is known as the program manager. The Department of Health and Senior Services (DHSS) Newborn Hearing Screening Program keeps a list of program managers and their contact information in order to clarify screening results and to send program-related notifications. Please notify the DHSS Newborn Hearing Screening Program of changes to the program manager or contact information within 30 days of the change. You may reach the DHSS Newborn Hearing Screening Program by phone (573.751.6266) or email Catherine.Harbison@health.mo.gov.
- Make sure the correct primary care provider (PCP), who will be seeing the child, is completely and accurately named. First and last name of the PCP must be written on the form. If both the first and last names are not included, the PCP area will be reported as unknown. This will delay reporting of results and can cause significant delays in babies receiving follow-up care.

PATIENT Spotlight

Lindsey

"My name is Lindsey Dahler, and I was born in 1984. Growing up with a genetic disorder like Phenylketonuria (PKU) was challenging. I remember being hungry all the time as a child and being jealous of my other siblings, because they could eat whatever they wanted. Luckily, I had a sister that had PKU as well. We leaned on each other a lot. We always stuck together, because being the only kids at school with PKU was hard. We always felt like outcasts, because we had to bring our own lunch and snacks. Our lunch normally consisted of vegetable soup, low protein bread, and some kind of fruit. For snacks we would bring 4 vanilla wafers or marshmallows.

PKU can create problems with comprehension, ADHD, emotional issues, etc. if the low-phe (low protein) diet is not followed. I had trouble in school when it came to comprehending what I was reading in text books. I was diagnosed with a learning disability in the sixth grade. High school was really hard for me. After high school, I went to cosmetology school and earned my degree. I worked in a salon for several years. Currently, I work in the Department of Social Services and have been with the State of Missouri for about five years.

I married in 2011 and decided to have children. I consulted with my genetic specialist and my dietician. From there they taught me how to effectively follow my diet, so I could start planning my pregnancies. I was required to reach "normal" phenylalanine levels (which is 4-6) for three months before starting my family. I worried constantly about the health of my baby, so being pregnant was nerve-racking.

I saw my genetic specialist, a high-risk specialist, and my regular OB. I had weekly blood draws to ensure my levels were at the normal range all throughout my pregnancy. I had ultrasounds twice a month to make sure the baby was developing correctly. My dietician provided me a meal plan that made sure I had proper nutrition and my medical formula three times a day. I now have two children Elliana and Anniston. Both of them are happy and healthy and do not have PKU (but are carriers).

Growing up, we were told that we should not have children, because the risks were too high. If the protein levels are too high throughout the pregnancy, it can cause damage to the baby such as cognitive and physical disabilities. Now research has proven, and I am living proof, that you can have beautiful, healthy babies if you follow your strict low-phe diet.

I would like to stress here that the newborn screening tests saved my life because if left undetected I would have become mentally disabled due to high phe levels (sending toxic gas to the brain killing brain cells). I would not have been able to accomplish all of this if it wasn't for the newborn screening that identified this genetic disorder. Even though it has been a hard road, and still is sometimes, it's well worth it, because I was able to have my dream children. Thank you to my genetic specialist, dietician, obstetrician, my high-risk obstetrician, Missouri State Newborn Screening Program, and the Missouri State Metabolic Formula Program."

LINDSEY

thank you

for your contribution in ensuring the best possible start for Missouri newborns!

Missouri DHSS Department of Health and Senior Services

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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