Behind the Screens

Missouri Department of Health and Senior Services
Newborn Screening Program

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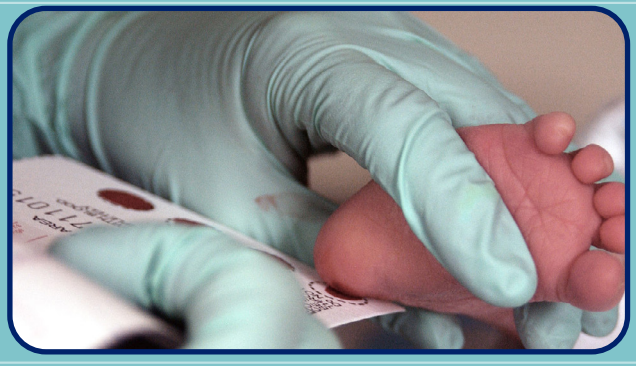
Featured Disorder

Mucopolysaccharidosis type I (MPS I) is an inherited disorder in the lysosomal storage disorder category. Lysosomes, the cell's recycling center, can't break down certain sugars and substances in MPS I patients. This leads to harmful buildup and various symptoms. Symptoms vary from severe to mild.

Early detection and treatment can delay severe health problems. Severe MPS I affects 1 in 100,000 newborns. The milder form impacts 1 in 500,000. In severe cases, babies show symptoms within their first year, while mild cases might not show signs until later in childhood. Symptoms include a large head, hearing loss, distinct facial features, hernias, and enlarged liver and spleen. Developmental delays and learning disabilities are also common.

There is no cure for MPS I, but treatments exist. These include enzyme replacement through IV infusions or stem cell transplants. Other options are therapy, diet changes, and medications or surgeries. Early detection through newborn screening is crucial for effective treatment and a healthy life.

Click here to learn more about MPS I Baby's First Test.



What's New?

The National Academies of Sciences, Engineering and Medicine are studying how newborn screening works in the United States. They will look at new technologies, suggest improvements, and offer a future plan. This work is funded by the Department of Health and Human Services Office on Women's Health and the Chan Zuckerberg Initiative.

Click here to learn more about the committee.

Patient Spotlight

Ezra

"Ezra is our first and only child. We were soaking up all the firsts of new parents during his first few days of life before we got the call that he had a a rare and fatal genetic disorder called Krabbe leukodystrophy. The first thing we did was research online, which was a mistake, as almost everything said these children typically passed away by age two. But then, we found a glimmer of hope. We found an organization that connected us with one of two specialists in this disorder. She informed us that because Ezra was screened as a newborn, he was eligible for treatment in the form of a transplant if he wasn't symptomatic. Time was extremely critical in this disease. We were terrified, but more than that, we finally felt a semblance of relief. Ezra was one month old when he was transplanted. Although he is regularly followed by his incredible team of physicians and participates in weekly therapies—he's a silly, social and adventurous seven-year-old boy. He is an extrovert who makes friends wherever he goes (likely due to his smile that feels like sunshine). Ezra loves going to school, cooking in the kitchen and dancing to Taylor Swift. If you ask him his favorite color, he will quickly tell you "Blue". Our family and friends cannot imagine our world without Ezra in it. To think that our lives would have a dramatically different outcome had it not been for the Missouri Newborn Screening is jarring. Without it, we wouldn't have been able to be put in touch with a specialist so quickly, and he certainly wouldn't be here with us today. We can never express the depths of appreciation that we have for those who completed his newborn screening panels."

Ryan and Karlita. Ezra's parents

Ezra's mom, Karlita, is a true champion in the Krabbe community. She has spoken on numerous local and national platforms, emphasizing the critical importance of newborn blood spot screening, early detection, and early intervention. Her advocacy for newborn screening and early intervention and treatment inspires hope for other families, showing that it is possible to raise a healthy child despite their diagnosis.

Tech Tips

- The CDC reports, 90% of infants born with hearing loss have two hearing parents. Babies who are deaf or hard of hearing can make the same sounds as babies without hearing loss. This is why early hearing detection and intervention refers to the practice of screening every newborn for hearing loss prior to hospital discharge.
- All babies born in Missouri should be screened for critical congenital heart defects (CCHDs) 24 to 48 hours after birth. CCHDs happen when a baby's heart or major blood vessels do not form correctly, causing a defect. There are many different types of heart defects that range from mild to severe. Babies with "critical" heart defects need urgent treatment, which may include medicine or surgery. Without treatment, these defects can lead to death or can cause serious health issues.
- Pulse oximetry screening is most likely to detect seven specific CCHDs. These include:
  - Hypoplastic left heart syndrome
  - Pulmonary atresia
  - Tetralogy of fallot
  - Total anomalous pulmonary venous return
  - Transposition of the great arteries
  - Tricuspid atresia
  - Truncus arteriosus

Click here to learn more about CCHDs.

Did You KNOW?

We have partnered with ***Navigate Newborn Screening*** to help families learn more about newborn screening and develop leadership skills with a **free** online learning module. This learning module will teach you more about the newborn screening system and will help you develop skills and confidence to support your personal leadership journey.

To access the online module, click on one of the links below. You will be taken to an enrollment page where you will be asked to enroll. Once you have enrolled, you can access all Navigate Newborn Screening courses. The module includes five base courses and two optional learning courses. Most courses take five minutes or less and can be easily viewed from your computer or phone.

- Navigate Newborn Screening English

For more information visit:
https://health.mo.gov/newbornscreening

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