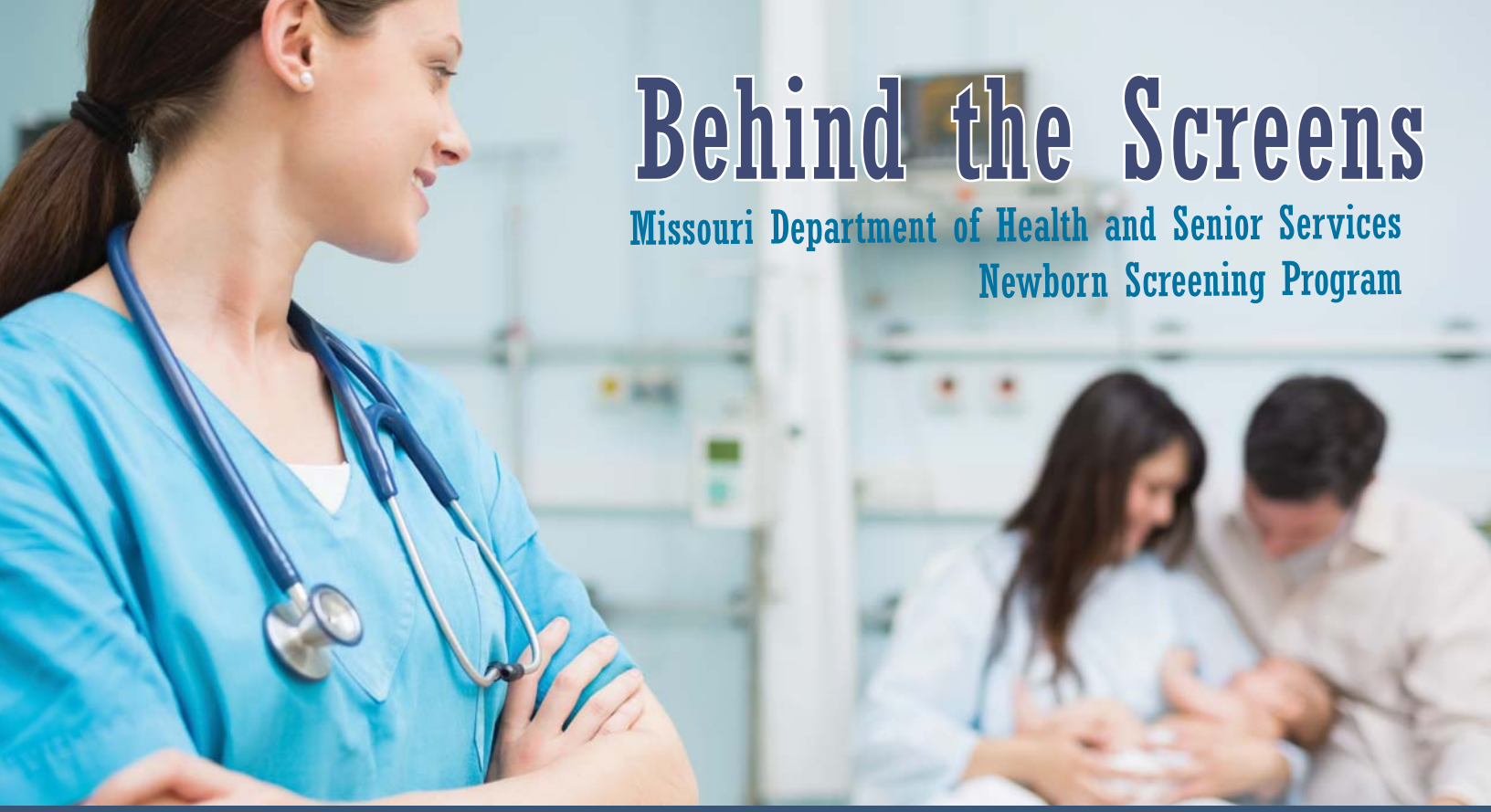

Behind the Screens

Missouri Department of Health and Senior Services
Newborn Screening Program

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Featured Disorder

Spinal muscular atrophy (SMA) is a progressive, autosomal recessive genetic disorder that affects the part of the nervous system that controls voluntary muscle movement. It is the number one genetic cause of death for infants. SMA involves the loss of nerve cells called motor neurons in the spinal cord and is classified as a motor neuron disease. The incidence of SMA is between 1:6,000 and 1:11,000 births. Based on this incidence, Missouri's Newborn Screening Program would expect to identify 7 to 12 babies in Missouri with SMA each year.

There are four types of SMA – I, II, III, and IV. The type of SMA is determined by age of onset and highest physical milestone achieved. SMA type I is the most common and severe form with symptoms becoming apparent within the first six months of life including progressive muscle weakness and poor muscle tone which leads to significant developmental delay. Individuals with SMA have difficulty performing the basic functions of life, like breathing and swallowing. However, SMA does not affect a person's ability to think and learn. Some children develop scoliosis or other skeletal abnormalities. Without treatment, most children affected with SMA type I are not able to sit up or stand, and the vast majority do not survive past two years of age due to respiratory failure.

SMA cannot be cured, however treatments are available to manage symptoms and improve quality of life. The first Federal Drug Administration approved treatment for SMA is nusinersen (Spinraza) and more treatments continue to be developed. Continued treatment with nusinersen allows many babies with SMA to reach and maintain age appropriate developmental milestones, including sitting, crawling, and walking. On average, breathing problems, nutrition problems, and hospital admissions have also decreased. However, response to treatment varies.

What's New?

The Newborn Screening Program is excited to inform you that all newborn screening samples are now being tested for Hunter syndrome (MPS II) and spinal muscular atrophy (SMA).

Hunter syndrome, also known as mucopolysaccharidosis II or MPS II, is a rare disease with an incidence of 1:162,000 births. Hunter syndrome is caused by a deficient or absent enzyme which results in the body's inability to break down certain types of complex sugars. Those sugars build up in cells throughout the body resulting in a variety of symptoms. Individuals with the most severe form of Hunter syndrome usually begin to exhibit symptoms between the ages of two and four. While Hunter syndrome is progressive and life-limiting, treatments are available to help manage symptoms and improve quality of life.

SMA is a progressive, genetic disorder that affects the part of the nervous system that controls voluntary muscle movement. To learn more about SMA, please see this issue's Featured Disorder article.

Did You KNOW?

Did you know that meconium ileus (MI) is NOT the same as meconium present at birth or during labor?

Meconium ileus is a bowel obstruction that occurs when the meconium in a baby's intestine is even thicker and stickier than normal meconium, creating a blockage in the ileum. The earliest signs of MI are abdominal distention (a swollen belly), bilious (green) vomit and no passage of meconium.

Most infants with MI have a disease called cystic fibrosis which is one of the disorders detected through newborn screening. Newborns with MI are at risk for false negative newborn screens for cystic fibrosis, therefore if a baby has a meconium ileus, it is important to mark the box indicating so, in the Altered Health Status portion of the blood spot form. This will flag the specimen and allow the Newborn Screening Laboratory to initiate second tier DNA testing to provide the most accurate screening results possible.

Tech Tips

Blood Spot

- Blood spot collection forms ordered from the Missouri State Public Health Laboratory after October 1, 2018, will have a new category in the Altered Health Status box. If a baby exhibits signs or symptoms of drug withdrawal, at the time of blood spot collection, please mark the box "Drug Withdrawal Signs".

Hearing

- Screening babies after they are 24 hours old will help ensure that debris has cleared from the ear canal. Since babies born by C-Section tend to have more debris in the ear canal, wait to do their hearing screening until as close to discharge as possible.

Critical Congenital Heart Disease (CCHD)

- If you complete a critical congenital heart disease screening on a baby transferred into your birthing facility from another hospital, you will not be able to document the screening in MoEVR. Please complete a paper form at https://health.mo.gov/living/families/genetics/birthdefects/pdf/MO580-31254-16CCHD.pdf and submit to the Missouri Department of Health and Senior Services according to the directions on the form.

PATIENT — SPOTLIGHT

LUCCA

Meet Lucca! She was born in Missouri in 2017, and was diagnosed with a rare disorder as a result of newborn blood spot screening. Thankfully, she is now a thriving toddler. Her mother, Katie, shares their story:

"My fiancé and I were overjoyed when our sweet girl Lucca Rose came into this world on June 20, 2017. As first time parents we were not quite sure what to expect but we were excited, sleep deprived, and madly in love. Not even 24 hours after we returned home from the hospital I received a call from genetics at St Louis Children's Hospital. I was confused and in shock that my little girl's heel prick test had detected an inherited fatty acid oxidation disorder known as Short Chain Acyl-CoA Dehydrogenase Deficiency (SCADD). Sure enough, after numerous tests it was confirmed our little girl had SCADD. SCADD is a rare condition so there is not a lot of available information online and our pediatrician was not very familiar. The not knowing made us extremely worried about our little girl's health and what her future might be like. However, genetics at St Louis Children's Hospital was our strongest advocate and a true blessing to our family. They put our daughter on a treatment plan immediately. If I ever had a question or concern they were always there to answer it. We are forever grateful for the prompt attention and support we received during this very scary time. Today our little girl is happy, healthy, and growing up fast!"

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THANK YOU for your contribution to ensuring the best possible start for Missouri newborns.

Missouri DHSS Department of Health and Senior Services

MISSOURI DEPARTMENT OF
HEALTH AND SENIOR SERVICES

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