



# Behind the Screens

Missouri Department of Health and Senior Services  
Newborn Screening Program

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## Featured Disorder

**Cystic Fibrosis (CF)** is a progressive, autosomal recessive genetic disorder. Mutations in the cystic fibrosis transmembrane conductance regulator (CFTR) gene cause the body to produce thick, sticky mucous that obstructs passageways in both the lungs and pancreas leading to severe lung damage and nutritional deficiencies. Babies with CF may exhibit no bowel movement in the first 24-48 hours of life, salty-tasting skin, delayed growth, or failure to thrive. Other symptoms people may experience later in life are chronic lung infections, severe digestive and bowels problems, persistent phlegmy cough, and infertility problems.

The Cystic Fibrosis Foundation and the Centers for Disease Control and Prevention recommend that all newborns be screened for CF. In 2007, Missouri added CF to the list of disorders screened on the blood spot filter form. Each year 25-30 Missouri babies are identified as having CF via newborn screening. Missouri uses a two-step process for testing which includes measuring the level of Immunoreactive Trypsinogen (IRT) in the baby's blood from the dried blood spot sample. If the IRT level is elevated, then a secondary test is performed on the same dried blood spot sample to test for some of the most common mutations in the CF gene.

Early detection and treatment through newborn screening improves quality of life as well as life expectancy for those living with CF. Advances in research and treatments such as airway clearance techniques, inhaled medications, and pancreatic enzyme supplements are continuing to make CF a more manageable disorder. Today the current life expectancy for people with CF is 40 years, whereas in the 1950's, children with CF rarely survived to school age. While newborn screening is not a definitive diagnostic test for CF, it can lead to early referral to genetic specialists where confirmatory sweat testing can be done and life-saving intervention can begin.

## What's New?

The Missouri Department of Health and Senior Services' Newborn Screening Program is excited to inform you that as of January 3, 2017, all newborn screening samples are being tested for Severe Combined Immunodeficiency (SCID).

SCID is a rare, genetic condition in which a baby is born without a functioning immune system. These babies are unable to fight off infections as simple as the common cold. Children born with SCID appear normal and healthy at birth, but if left untreated, will die before their first or second birthday. Regular immunoglobulin replacement therapy can help children fight infections, however, the most effective treatment for SCID is bone marrow transplant. Treatment with bone marrow transplant has a 95% successful cure rate if given within the first three months of life.

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# Did You? KNOW!



Did you know sickle cell disease (SCD) is the most common inherited blood disorder in the United States? The disease occurs predominantly among people of African descent, but is also found in individuals with Mediterranean, Middle Eastern, Indian, and Latin American ancestry. In Missouri, approximately 1 in 400 black or African American newborns are affected by SCD. The symptoms of SCD may not develop until a few months after birth or later childhood, therefore, early detection is vital for a better quality of life for these children.

The Missouri Department of Health and Senior Services' Newborn Screening Program works to ensure that all infants identified with SCD receive timely follow-up with confirmatory testing and early diagnosis of the disease. If detected early, life-prolonging care can begin as soon as possible. When left undetected or untreated, SCD can lead to severe health complications and even death in early childhood.

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## Tech Tips



- When collecting a newborn blood spot screen, including repeat specimens, always document the gestational age and weight at time of birth on the collection form.
- Paper Hearing Only forms may be submitted for babies who: are transferred from another hospital, return for repeat screening, or had a delayed screening due to NICU stay. Paper Hearing Only forms are available free of charge from the DHSS warehouse at <http://health.mo.gov/warehouse> under catalog number BGD-3, or they can be printed at [http://health.mo.gov/living/families/genetics/newbornhearing/pdf/HearScreen\\_Only\\_Form.pdf](http://health.mo.gov/living/families/genetics/newbornhearing/pdf/HearScreen_Only_Form.pdf).
- A pulse oximetry reading of 90-94% in BOTH the right hand and foot OR a difference of 4 percent or more between the right hand and foot when performing a critical congenital heart disease (CCHD) screening should be repeated in one hour.

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*...always document the gestational age and weight at time of birth on the collection form.*

# PATIENT

# SPOTLIGHT

# Feb

Jeb is a Missouri baby who was diagnosed through the accurate and timely collection and transport of his newborn screening. Jeb is now thriving as a normal, happy, and healthy baby. This is what his mother, Kristen, had to say about their experience:



"My husband John and I welcomed our sweet baby boy, Jeb, on December 8, 2016. As new parents, we were filled with excitement as we transitioned home. At a week old, we were informed Jeb had a positive result on his newborn screening test. Preliminary results showed that Jeb had a metabolic condition known as Very Long Chain Acyl-CoA Dehydrogenase Deficiency (VLCAD). The next few weeks to months were challenging as we learned about VLCAD and possible complications. We were referred to a great team of specialists who have created a lifelong plan for Jeb's health that will enable him to thrive and be as active as he would like to be. We are so thankful for early detection through newborn screening that allowed for early intervention in what can be a life-threatening condition, not only for Jeb, but for our potential future children as well."

Kristen, Jeb's mother

*"We are so thankful for early detection through newborn screening that allowed for early intervention in what can be a life-threatening condition..."*

**Thank you  
for your  
contribution  
to ensuring  
the best  
possible start  
for Missouri  
newborns.**



MISSOURI DEPARTMENT OF  
HEALTH AND SENIOR SERVICES

**Bureau of Genetics and Healthy Childhood**  
Newborn Blood Spot, Hearing, and CCHD Programs  
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