Behind the Screens

Missouri Department of Health and Senior Services

Newborn Screening Program

Summer 2025 ★ Missouri Department of Health and Senior Services ★ Health.Mo.Gov ★ Volume 8 Number 2

Featured Disorder

Severe combined immunodeficiency (SCID), also known as Bubble Boy Syndrome, is a group of disorders characterized by the absence or dysfunction of T and B lymphocytes. Newborns with SCID are at risk for severe, life-threatening infections because they do not have a working immune system. Without timely diagnosis and adequate treatment, patients with SCID do not survive past the age of two. SCID is a rare genetic disorder, with an estimated prevalence of 1 in 58,000 births. SCID is not contagious – both parents must be carriers of the genetic mutation for their child to be affected. Early symptoms of SCID include frequent and resistant infections, diarrhea, failure to thrive and thrush.

Most infants with SCID are treated with stem cell transplantation (SCT) and gene therapy to restore normal immune function. Patients who undergo SCT or gene therapy should be isolated, and supportive care with antibiotics and antifungals may be given to prevent infections. Patients who are diagnosed early and are infection-free before treatment begins have a survival rate of about 95%. Early diagnosis and intervention are crucial for newborns with SCID, which is why accurate and timely newborn screening is so vital.

The Newborn Screening Follow-up Program works with the State Public Health Laboratory to quickly identify newborns at high risk for having SCID through accurate and timely blood spot screenings. The Program connects primary care providers and families with immunology specialists throughout the state to ensure these babies receive treatment as quickly as possible. If you, someone you know, or one of your patients is diagnosed with SCID, there are support services available through the Immune Deficiency Foundation (IDF) and SCID Compass, an educational program of IDF.

Resources:

IDF and SCID Compass program: primaryimmune.org

SCID: babysfirsttest.org

Featured Disorder 1

Patient Spotlight 2

Did You Know 2

Patient Spotlight

Oliver

"Approximately three days after our son Oliver was born, we received a phone call from his pediatrician stating that he had screened positive for Spinal Muscular Atrophy (SMA) on the Missouri state newborn screen. With both of us being in the health care field, we knew how devastating this diagnosis was. We were immediately contacted by Children's Mercy Hospital in Kansas City, MO and were in for an appointment within a few days. During this time, we learned much more about SMA, and the genetic testing done in the newborn screening process. There have been recent groundbreaking advances in the treatment of SMA, but due to the degenerative nature of the disease, time is crucial to getting treated prior to the onset and progression of symptoms. With the type of SMA that Oliver has, he would likely have started to develop symptoms between 6 months and 2 years of life. The newborn screen and genetic testing allowed Oliver to be treated while he was asymptomatic, and his life has been drastically changed for the better.

He recently turned two years old and continues to be symptom-free while meeting all his milestones. He is running, jumping and climbing all over the place! Screening for SMA has only recently been added to the newborn state screen across the country and we cannot imagine how different our lives would be if it had not been. Without the newborn screen, Oliver would not have received treatment prior to the start of the degenerative process occurring which would have been too late. We are forever grateful for the advocacy of the SMA foundation and the advancements in testing and treatment of SMA. It has forever changed our son's life!"

Josh and Bailey, Oliver's parents

Did You Know?

120 BABIES SAVED PER YEAR

Screening for critical congenital heart defects (CCHD) was added to Missouri's required newborn screening tests in January 2014. According to the Centers for Disease Control and Prevention, these screenings have decreased early infant deaths from CCHD by 33% (that is 120 babies saved per year)!

For more information about CCHD, visit cdc.gov/heart-defects/hcp/screening/index.html.

MISSOURI DEPARTMENT OF HEALTH & SENIOR SERVICES

Bureau of Genetics and Healthy Childhood
Newborn Blood Spot, Hearing and Critical Congenital Heart Defects Programs
573.751.6266 or 800.877.6246

Missouri State Newborn Screening Laboratory
573.751.2662
Health.Mo.Gov/newbornscreening