Behind the Screens

Missouri Department of Health and Senior Services
Newborn Screening Program

Spring 2026 ★ Missouri Department of Health and Senior Services ★ Health.Mo.Gov ★ Volume 9 Number 1

Featured Disorder

Critical congenital heart disease (CCHD) refers to specific congenital heart defects that happen when a baby's heart or major blood vessels inside the heart do not develop properly. Most of the time, the cause of these abnormalities remains unknown; however, some defects may be caused by changes in genes or chromosomes, or environmental factors, such as drugs, illnesses or chemicals, that a pregnant woman is exposed to during pregnancy, which can affect how the baby's heart develops.

About two out of every 1,000 babies are born with CCHD. These heart defects occur before birth and can range from mild to severe. Heart defects require prompt treatment; if left untreated, they can cause cyanosis (a bluish tint to the skin, lips, and fingernails), rapid breathing, swelling in the face, hands, feet, legs, or areas around the eyes, shortness of breath, sweating around the head, poor weight gain, developmental delays, or even death. In Missouri, all babies should be screened for CCHD around 24 hours old. A failed CCHD screen may require referral to a pediatric cardiologist, who may order an echocardiogram and further testing. Most babies who pass the CCHD screening will not have CCHD; however, this test does not detect all defects, so it is still possible to have a heart defect even with a negative screening result.

Without screening shortly after birth, babies with CCHD might go home from the hospital or birthing center without proper care because they look healthy. Sometimes, even babies who had a normal prenatal ultrasound can have hidden CCHD. At home, these babies can develop serious health problems and often need emergency care. Treatment for CCHD depends on the type of heart defect present. Most heart defects can be corrected or improved with surgery, catheter procedures, and/or medications. CCHD screening may save a baby's life by helping to find a severe heart defect quickly so that the baby can be treated and lead a healthier life. Support services and more information are available at babysfirsttest.org.

Patient Spotlight

Charles

After a chaotic labor and delivery, we welcomed our son, Charles, into our lives. He was beautiful and perfect. I remember, in between holding him and admiring my new baby, the hospital did so many little tests just to make sure he was healthy. A nurse gave us a paper with information about the newborn screening and told us, "Don't worry, no news is good news." Well, a few weeks later, we were contacted with news. I immediately felt scared, remembering what the nurse said. In the midst of the wonderful and exhausting days of taking care of a newborn, we had completely forgotten about the newborn screening. Our baby seemed so healthy, could something really be wrong? His results showed lower than normal levels of the GAA enzyme, indicating a possibility of having a lysosomal storage disorder called Pompe disease. A quick search on Google let us know this could be very serious, causing a shortened lifespan, with some affected babies not making it to their very first birthday.

First things first, they wanted to retest to rule out a false positive. We immediately went to get a second blood draw done. While waiting on the results, we educated ourselves on what life could look like for us if our son, in fact, had Pompe disease. During our research, we learned that the quicker the treatment starts, the better the outcome we could have. We also learned that not all states test for this specific disease, meaning that despite this being unpleasant news, we knew we were physically in the best possible place for the future of our baby if the second test indicated Pompe disease. Because the state of Missouri screens newborns for Pompe disease, we would be able to get a quick diagnosis and a fast track to treatment, which would affect the longevity of my son's life. Although we were terrified, stuck in the middle of the unknown results, we truly felt so thankful towards the state of Missouri for including this test. Our second results came back indicating even more of a decrease in the GAA enzyme with a present gene mutation. We were then referred to the Children's Mercy Genetic Clinic. During this time, we felt so alone and anxious about the future, but we were also in disbelief because our son didn't seem sick at all. To our surprise, someone who worked with the newborn screening called us to ask about how things were, and it made us feel seen and cared for. It was so unexpected that a standardized test that we previously knew nothing about could have interjected itself so heavily into our lives. By the absolute grace of God, our son turned out to be only a carrier of Pompe disease and isn't actually affected by the disease. Because our son is a carrier of Pompe disease, they told us it's likely that either I or Charles's dad was also a carrier, possibly even both of us. This was very important information to have when deciding to have more children. We both did genetic testing and learned that neither one of us carries Pompe disease. The genetics specialists told us that the chance of our son being a carrier without a parent carrier was extremely rare, about a 0.5% chance.

What a wild journey we were on for the first couple of months of Charles's life! The newborn screening allowed us to learn very important information for Charles to know when he decides to have kids of his own. What a wonderful thing for something as simple as a blood draw for a newborn screening to allow you to be proactive instead of reactive to whatever results you get. It's such a blessing. Charles is now two and a half and thriving! Praise the Lord!"

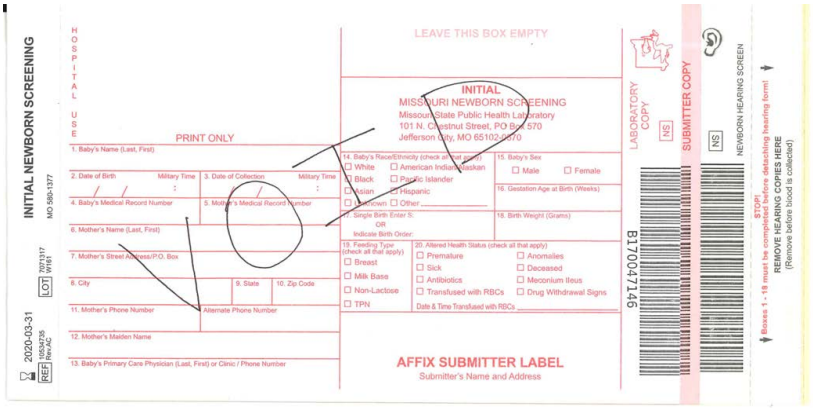
Rachel, Charles' Mom

IN the NEWS

Missouri continues to advocate for language equity and foundational development with the LEAD-K Act - legislation ensuring children who are Deaf or Hard of Hearing build a strong language foundation before kindergarten. Passed in August 2023, the initiative provides milestone checklists and parent-focused resources through the LEAD-K Missouri Early Connections platform. More information can be found at Missouri's Early Care & Education Connections website at earlyconnections.mo.gov.

Provider Tip

A newborn blood spot screen form costs $130. While a birthing facility or health care provider cannot charge a patient more than the cost of the form, they may have additional charges associated with the blood spot screening. This may include a collection charge, processing fees, supply charge, etc. If the suitability of a collected specimen is questionable, it is best to start over and recollect. The old specimen does not need to be discarded; instead, write "VOID" across the front of the form and return it to the Missouri State Public Health Laboratory with the new, satisfactory sample. The lab will then replace the old form with a new form to the submitter, free of charge.



MISSOURI DEPARTMENT OF HEALTH & SENIOR SERVICES

Bureau of Genetics and Healthy Childhood
Newborn Blood Spot, Hearing and
Critical Congenital Heart Disease Programs
573.751.6266 or 800.877.6246
Missouri State Newborn Screening Laboratory
573.751.3334, Option 6
Health.Mo.Gov