Behind the Screens

Missouri Department of Health and Senior Services
Newborn Screening Program

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Featured Disorder

Congenital hypothyroidism (CH) is an endocrine condition that affects the body's thyroid gland. Individuals with CH are unable to produce enough thyroid hormone, which is essential for healthy growth and development. When there's not enough thyroid hormone production, the body produces more of a chemical called thyroid stimulating hormone (TSH) in an attempt to get the body to produce more thyroid hormone. As a result, the newborn screening will show a high amount of TSH.

About one in every 3,000 to 4,000 babies in the United States is born with CH. Twice as many females as males are affected, and it occurs in all ethnic groups. In Missouri, in 2016, there were 45 diagnoses of CH as a result of newborn screening. If CH is not detected right away, most babies will begin to show signs at three to four weeks after birth. Some early signs of CH include: jaundice of the skin or eyes, weak muscle tone, swelling around the eyes, swollen tongue, a hoarse-sounding cry, and delayed growth and weight gain.

Treatment for CH most commonly consists of hormone replacement therapy. The baby will need to be started on a synthetic thyroid hormone to replace the natural hormone that his or her body doesn't produce in adequate amounts. There may also be dietary restrictions such as restricting the amount of soy or iron in the baby's diet, as both soy and iron affect how the body absorbs the synthetic hormone. If treatment for CH starts soon after birth, affected children can have healthy growth and development. Delayed treatment can put children at risk for intellectual disabilities, learning disabilities, developmental delays, and delayed growth. This is another reason why the timely and accurate collection of newborn screenings is so important.

What's New?

In October 2017 the American Academy of Pediatrics published an updated policy statement regarding cord blood banking. The policy statement is intended to assist pediatricians, obstetricians and other health care providers in their discussions with parents about the life-saving potential of banked cord blood. The statement addresses all issues of collection and storage including the regulation, cost, need, and benefits of private versus public banking. In addition, care providers are advised of the various ethnic specific disparities in current banking practices. Overall, the AAP recommends the routine banking of cord blood for its proven potential in the treatment of certain fatal diseases.

Cord blood is an excellent source for stem cells needed for hematopoietic stem cell transplantation for treatment of certain hematologic malignancies, hemoglobinopathies, severe forms of

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Did You KNOW?

The white, detachable paper on the front of the newborn screening (NBS) collection card is information regarding the Missouri Newborn Screening Sample Storage Policy that was implemented on July 1, 2011. Missouri State Law (Section 191.317) requires the Missouri State Public Health Laboratory to retain the leftover NBS samples for five years after the testing has been completed and then to destroy them after the five years of storage has ended.

There are numerous benefits to public health in retaining residual NBS samples. Some of these benefits include: the law allows the department to release the samples for the purpose of anonymous research, quality assurance and improvement for the NBS laboratory, research for new treatments and cures for major childhood illnesses, and sometimes the NBS sample is the only remaining evidence available to a family from their child if their child becomes missing.

It is important to provide and review this information sheet with parents, as it also provides three opt-out options if they do not wish the department to release their child's leftover NBS sample for anonymous research. For more information about the Sample Storage Policy, please visit http://health.mo.gov/lab/newborn/pdf/nbsstoragereleasepolicy.pdf.

Tech Tips

- When placing a pulse ox sensor on a baby's skin for a critical congenital heart disease screening, there should not be gaps between the sensor and the baby's skin. The sides of the sensor should be directly opposite of each other.
- Do not confuse meconium ileus with meconium aspiration on the newborn screening collection form. Check the box for meconium ileus only if an ileus is present, as this is often a sign of cystic fibrosis.
- Report hearing screening results no later than seven days from the date of screening. Timely submission of hearing screening results allows the Newborn Screening Program to quickly contact parents of babies who have either missed or failed the hearing screening.

THANK YOU

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

PATIENT — SPOTLIGHT

DOMINIC

Dominic is a happy, spunky, little boy with cystic fibrosis (CF). Newborn screening aided in the timely diagnosis and intervention of his condition. His mother, Christina, said this:

"Dominic was born on May 29, 2015 and before leaving to go home and enjoy our newborn, his newborn screening was collected. At our two day check-up with his pediatrician, we mentioned that we had noticed that Dominic had been vomiting green bile. After many hours in the E.R. between our local hospital and St. Louis Children's Hospital, we discovered that Dominic was suffering from meconium ileus, even though he had a normal bowel movement shortly after delivery. This was our first sign that he may have CF. After emergency surgery to remove the obstruction, within just 24 hours of our visit with the pediatrician the day before, we were told that there was a good chance that Dominic's newborn screening would come back positive for CF. While Dominic was in the NICU recovering from surgery, we were told that the screening came back only showing one CF mutation after checking for the first 80 percent of mutations. Soon his blood was drawn and sent back to check for the other 20 percrent of CF mutations. A week after we were home, we received 'the call.' They had found his other CF mutation, which meant that we would soon have to meet with the CF team at Children's Hospital and complete a sweat test to verify his diagnosis. We would also start him on enzymes to help him gain weight and grow to a strong, young boy. Since his diagnosis, he has been admitted to the hospital four times for CF related complications, but he has been growing at an acceptable rate and continues to thrive regardless of his diagnosis."

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T-lymphocyte and other immunodeficiencies, and metabolic diseases. Universal newborn screening for SCID has exponentially increased the estimated need for conveniently accessed stem cells. Continuous addition to the newborn screening panel of disorders that can be treated with stem cell transplantation has increased the anticipation of the need for a larger volume of publicly stored cord blood.

Education of the public, parents, and health care providers is paramount in expanding the availability of cord blood stem cells. To learn more about this topic, please visit http://pediatrics.aappublications.org/content/early/2017/10/26/peds.2017-2695.

Missouri DHSS Department of Health and Senior Services

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