Behind the Screens

Missouri Department of Health and Senior Services

Newborn Screening Program

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Featured Disorder

Gaucher (go-shay) disease is a rare genetic disorder in which the glucocerebrosidase (GBA) enzyme is either missing or does not function properly. It is classified as a lysosomal storage disorder because the missing or malfunctioning enzyme prevents lysosomes from breaking down certain fats, increasing harmful molecules in the body. Gaucher disease occurs in about one in every 20,000 births. There are three main types of Gaucher disease, each with its own age of onset and signs or symptoms.

The most common form is Type 1, which occurs more frequently in individuals of Eastern and Central European Jewish descent. Type 1, or non-neuronopathic form, of Gaucher disease does not affect the brain or spinal cord. Symptoms can begin at any time. They include a swollen stomach, easy bruising and broken bones, exhaustion, bone pain, nosebleeds, and yellowing of the eyes. Types 2 and 3, or neuronopathic forms, affect the brain and spinal cord. Type 2 usually occurs by 3 months of age and progresses rapidly, while Type 3 usually occurs by 2 years of age and progresses more slowly. Symptoms of Types 2 and 3 include the ones listed above, but they can also consist of abnormal eye movements, epilepsy and seizures, developmental delays, muscle tightening, and difficulty breathing and swallowing. Additionally, there are rarer types of Gaucher disease, one that presents with life-threatening complications in the first few days of life, and a cardiovascular form that can include heart symptoms in addition to the other features noted above.

There is no cure for Gaucher disease. Treatments aim to delay symptoms and improve quality of life. Early identification and treatment often allow children with Gaucher disease types 1 and 3 to live healthier lives with fewer complications; this is why newborn screening for Gaucher disease is essential. Treatments for Gaucher disease may include enzyme replacement therapy and/or medications like miglustat, pain relievers, and vitamin supplements. Blood transfusions, joint replacement surgery, and bone marrow transplants may also be considered. More information about Gaucher disease can be found at Baby's First Test, Children's Gaucher Research Fund and the National Gaucher Foundation.

Patient Spotlight

Vernin Theo

"Early in my pregnancy, I had the Non-Invasive Prenatal Testing (NIPT) done at 11 weeks. Everything came back negative, and we found out we were having a BOY! I was relieved to know everything had come back negative since my husband had been adopted at birth, and we didn't have a full family history. I had a normal pregnancy, labor, and delivery! We were over the moon with our perfect baby boy, Vernin Theo.

At about eight days old, I received a call from our pediatrician. She told me that he had an abnormal result from his newborn screen and that he had an irregular result for a Cystic Fibrosis (CF) gene. I was immediately terrified. My mind went to a movie I had seen about a teen with CF whose life was greatly impacted by the disease, which ultimately took her at an early age. As I sat and cried trying to explain to my husband what our pediatrician had said I just couldn't stop thinking that I would soon lose this perfect boy of mine. We were quickly speaking with a specialist's office in Columbia.

Within a few days, we were on our way to Columbia for a sweat test. This is the gold standard test for CF. The results of the initial sweat test were indeterminate. My husband and I both completed a full genetic test. It was finally discovered that my husband and I were both carriers of a mutated gene that is responsible for CF. A full genetic panel on our son showed he did have both gene mutations that did in fact give him the diagnosis of Cystic Fibrosis. Fortunately, in the couple of months it took for his diagnosis, he didn't exhibit any signs of the disease.

After his diagnosis, he was started on twice-daily nebulizer treatments and manual chest percussion as a way of preventing and protecting his lungs. Vernin is now 16 months old, and he continues not to show any signs of Cystic Fibrosis. We feel very fortunate that this was caught so early with the newborn screen. If he hadn't been diagnosed at birth, my husband and I wouldn't know what to look for or what precautions to take. Without that knowledge, his life and future could have had a negative impact. Now that the initial stress of learning all this information about our son is behind us, we feel more comfortable believing that our son has a bright future ahead of him! And, of course, he is still our perfect baby boy! I am a labor and delivery nurse. When patients say they don't want to receive the newborn screening on their new baby, my coworkers and I have first-hand knowledge of how much of an impact it really can make!"

Katie, Vernin's Mom

IN the NEWS

At the end of 2024, the American Academy of Pediatrics (AAP) updated its Critical Congenital Heart Disease (CCHD) screening recommendations and algorithm. Please visit the AAP website for more information on these CCHD screening changes. Additionally, The Missouri State Public Health Laboratory (MSPHL) and the Newborn Screening Program have updated the newborn blood spot collection form to include Critical Congenital Heart Disease (CCHD) screening results. Once you receive an order including the new forms, use them to report CCHD results. Until then, continue using the Missouri Electronic Vital Records (MoEVR) reporting system. More information on this will be outlined in our next newsletter and with the new collection form orders.

Provider Tip

An inpatient screening should be limited to two hearing screening sessions using the same technology, separated by several hours. A hearing screening session should be limited to two screening attempts. The newborn should always receive a bilateral screen, regardless of previous screening results.

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Newborn Blood Spot, Hearing and
Critical Congenital Heart Defects Programs
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