Behind the Screens

Missouri Department of Health and Senior Services
Newborn Screening Program

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Featured Disorder

Malonic Acidemia (MAL) is a condition in which the body cannot break down certain proteins, leading to harmful levels of organic acids and toxins. Because of this, MAL is considered an organic acid disorder. This disorder is extremely rare, with fewer than 30 reported cases in the literature.

Symptom onset age varies, but it typically occurs in early childhood. Symptoms include developmental delays, weak muscle tone, diarrhea, vomiting and seizures. Specialists may prescribe L-carnitine supplements to help break down fats. Additionally, dieticians and nutritionists can help create a diet that avoids high-fat foods. Avoiding illness, infection and long periods without eating can help prevent the symptoms of the disease.

A genetic counselor can help families understand the causes of MAL, discuss genetic testing for MAL, and explain what this diagnosis means for other family members and future pregnancies. Because MAL is so rare, the outcomes are not well known; however, without early intervention and treatment, children with MAL will die in infancy.

The Missouri Newborn Screening Program screens for many organic acid conditions, including MAL, and collaborates with specialists across the state to ensure children receive timely intervention and treatment.

If you or someone you know has been affected by MAL, support and additional information can be found at the Organic Acidemia Association and Baby's First Test.

Patient Spotlight

Charlee

"Charlee, short for Charlotte, entered the world three weeks earlier than expected, weighing just 4 pounds, 1 ounce. Even before she was born, Charlee faced challenges, struggling to grow due to intrauterine growth restriction. Her arrival was fraught with concerns, as doctors worked to stabilize her dangerously low blood sugar levels. She was quickly placed in a special care nursery, where an IV provided the support her tiny body needed.

Those first days were a whirlwind of medical tests, assessments, and anxious moments. Amid the chaos, her newborn screening—a routine procedure—didn't initially stand out to us. But Charlee's condition continued to deteriorate. She wasn't eating well, she was losing weight, and we were desperate for answers.

Then came the call that changed everything. Charlee's neonatal doctor received an urgent phone call from the state health department, urging immediate testing of her thyroid levels. It's exceedingly rare, the doctor told us, for the state to bypass its usual process of notifying parents or pediatricians via letter or email. This level of urgency was unprecedented.

After additional tests, Charlee was diagnosed with congenital hypothyroidism. Her tiny body wasn't producing thyroid hormones, and further examination revealed why: Charlee had been born without a thyroid gland. The thyroid is a critical organ, responsible for regulating body temperature, metabolism, growth, and development, among other vital functions. Without it, life as we know it isn't possible.

But hope came in the form of a simple, daily medication: levothyroxine. Just days after beginning treatment, Charlee's appetite improved, and she started gaining weight. Slowly but surely, our beautiful baby was thriving.

Today, Charlee is a bright, loving, and spirited almost-two-year-old. Her smile lights up our days, and her unique personality shines through. Each morning, she cheerfully takes her thyroid medication—an act so ordinary now that it's hard to fathom how extraordinary her journey has been.

We are profoundly grateful for newborn screening, for the doctors who acted quickly, and for the medication that allows Charlee to live a full and happy life. Without these interventions, Charlee's future could have been tragically different. Instead, we have the incredible gift of watching her grow, thrive, and simply be—just as she was always meant to."

Stacy, Charlee's Mom

DID YOU KNOW?

When a high-risk result is identified, the Missouri Department of Health and Senior Services collaborates with specialists with expertise in the disorder. These specialists also collaborate with the primary provider listed on the newborn screen to create a plan of care for the baby. Missouri has contractual agreements with MU Health Care in Columbia, Children's Mercy Hospital in Kansas City, St. Louis Children's Hospital in St. Louis, and SSM Cardinal Glennon in St. Louis. These agreements allow laboratory staff, screening staff, providers and specialists to work together as a team.

MU Health Care

Children's Mercy KANSAS CITY

SSM Health Cardinal Glennon Children's Hospital

Children's HOSPITAL • ST. LOUIS BJC HealthCare

Provider Tip

Timely follow-up and clear communication are key to early hearing detection and intervention. When a newborn fails the initial hearing screening, it is essential to clearly explain the next steps to the family, assist in scheduling follow-up testing, and document and report screening results promptly to the Missouri Department of Health and Senior Services.

MISSOURI DEPARTMENT OF HEALTH & SENIOR SERVICES

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