Behind the Screens

Missouri Department of Health and Senior Services
Newborn Screening Program

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Featured Disorder

Early detection and treatment can help children with 3-MCC lead healthy lives. 3-methylcrotonyl-CoA carboxylase deficiency (3-MCC deficiency) is an inherited type of organic acid disorder. 3-MCC is an enzyme that helps break down the amino acid, leucine (a building block of protein). Infants with 3-MCC deficiency appear normal at birth, but can develop signs and symptoms in infancy or early childhood--although some may never show any signs of the disorder. 3-MCC affects one out of every 36,000 babies worldwide and is inherited as an autosomal recessive pattern. This means that a child must inherit an abnormal gene from each parent to have the disorder.

Mutations in the methylcrotonyl-CoA carboxylase 1 (MCCC1) gene or methylcrotonyl-CoA carboxylase 2 (MCCC2) gene can cause 3-MCC deficiency. These two important genes provide instructions for making different parts of the enzyme 3-MCC known as “subunits”. Mutations in the MCCC1 or MCCC2 gene reduce or completely eliminate the activity of 3-MCC. This prevents the body from processing or breaking down leucine properly and, as a result, toxic byproducts of leucine accumulate to harmful levels. This can result in brain damage or even death.

The signs and symptoms of this condition are different with each child and can range from mild to life-threatening. These include feeding difficulties such as poor appetite, recurrent episodes of vomiting and diarrhea, sleeping longer or more often with excessive tiredness (lethargy), weak muscle tone (hypotonia), muscle tightness, and behavioral changes. If untreated, this disorder can lead to breathing problems, liver failure, delayed development, seizures, coma, brain damage, and death. Many of these complications can be prevented with early detection and lifelong monitoring.

Treatment options for 3-MCC include supplements, medication and dietary restrictions. A very specific diet low in protein will need to be followed due to the inability to break down leucine. A supplement prescribed by a physician called L-carnitine is a natural substance that helps remove damaging waste products from the body.

If 3-MCC is detected and treated early, children often exhibit typical growth and development. Most children diagnosed with 3-MCC never need any special treatment and can live healthy lives.

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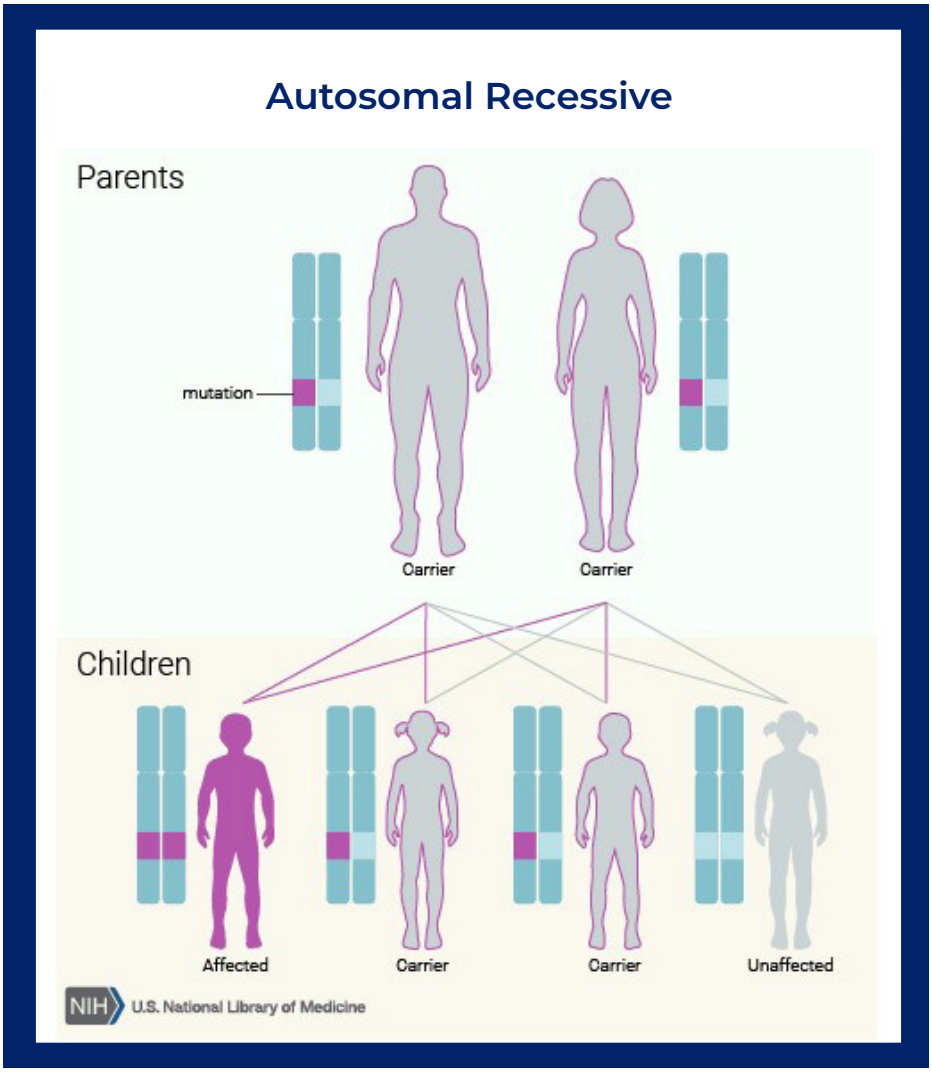

Featured Disorder References

babysfirsttest.org/newborn-screening/conditions/3-methylcrotonyl-coa-carboxylase-deficiency

medlineplus.gov/genetics/condition/3-methylcrotonyl-coa-carboxylase-deficiency/

TECH TIPS

- Report hearing screening results no later than seven days from the date of screening.
- Accurate date and time of blood spot collection is crucial. This data cannot be amended once the specimen is received by the state public health laboratory.
- NEVER throw away a newborn screening (NBS) collection form. If you make a mistake or if you suspect the specimen will be a poor quality sample, allow the specimen to dry completely, write VOID across the front of the form and return it to the State NBS Laboratory along with the correct form. The voided form will be replaced free of charge.
- It is important to identify the health care provider who will care for the newborn after discharge for all newborn screenings. Provide the health care provider's first and last name. A clinic name or office phone number will also suffice.

Patient Spotlight EVERETT

Everett is our second child and was born March 6, 2021. Our oldest was born healthy so, we expected Everett to be the same. Although my husband and I are in the medical field and have a great deal of combined knowledge, I was not prepared when we received a phone call about a week after Everett was born saying, "Hi, this is the genetics team from Cardinal Glennon and we are calling in regards to Everett's newborn screening." My heart sank, I instantly knew something was wrong. The genetics team knew I worked at Cardinal Glennon Children's Hospital as a nurse already, so they went into great detail of everything we needed to do. We went in for a follow up and blood work the next day for both Everett and I. They explained that sometimes the newborn screens for 3-methylcrotonyl-CoA carboxylase deficiency (3-MCC) can come back positive for the infant, when in reality, the mom will actually have the disease. So they worked both of us up. I was still in shock that something could be so wrong, yet my baby looks so healthy. Everett's liver enzymes were elevated, which is an indicator of 3-MCC. Everything on my lab work came back normal, which meant that it was indeed Everett that we had to keep an eye on. We were instructed to never let him fast and to feed him every 2-3 hours until we could confirm the diagnosis at around 3-4 months of age. As Everett got older, we were able to increase the amount of time between feeds, but if he was to ever fast longer than the recommended times, it could increase chance for a metabolic crisis. This could include seizures, coma, or even death. Along with never fasting, we also were instructed to limit protein intake, because the enzyme that was missing breaks down leucine, an amino acid present in proteins.

A helpful resource tool we encountered while on this journey was a Facebook page that had people all ages living with 3-MCC, they talked about their experiences and the diets that they follow. This disorder is very rare, so there were only a couple hundred people on it. I am just surprised at how welcoming and available people are in tough situations like these. My husband and I also spent a lot of time researching treatments, signs and symptoms, and more information about 3-MCC just to give a perspective of what we may be encountering.

After this appointment, we went on as normal until we were able to get DNA testing around 3-4 months old. It was 3 months of sleepless nights breastfeeding every 2-3 hours. When we were finally able to get the testing done, it only took about 2 weeks to get the results back.

We were so relieved to find out that he was only a carrier for this disorder. Both my husband and I were offered DNA testing to find out who was the carrier or if we both may be carriers, but we declined. We already have 1 healthy son, so we are taking our chances that only one of us has the gene. We are so very thankful that we know more about this disorder, in case we have grandchildren one day with 3-MCC.

This experience has also helped in my career as a postpartum/pediatric nurse who performs newborn screenings on the daily. I am very intentional about educating my patients on the importance of newborn screening and when they go home on what to expect if they receive that same phone call.

We are so very thankful for everyone who helped us through this journey as well. It was unexpected, but very insightful.

Kristin, Everett's mom

Did You KNOW?

Blood spot screens are reported with results that fall into one of the following categories: normal, low-risk or high-risk. Normal results do not require any further action unless clinically indicated. Low-risk results are those that are almost normal, but not quite, and it is recommended that providers repeat the screen. A repeat screen often will normalize, but occasionally the results will remain abnormal and require further action. High-risk results indicate an increased risk that a baby may be affected by a particular disorder and require additional testing to confirm or rule out disease. When a screen is high-risk, the Missouri Newborn Screening Program refers the baby to specialty centers within the state for follow-up. The Program contracts with Children's Mercy Hospital in Kansas City, University of Missouri Women & Children's Hospital in Columbia, Cardinal Glennon Children's Hospital in St. Louis, and Saint Louis Children's Hospital in St. Louis. The centers consist of a variety of multidisciplinary teams, some of which are geneticists, pulmonologists, hematologists, genetic counselors, dieticians, endocrinologists, neuromuscular specialists and immunologists, among others. Families in Missouri are in great hands with the local experts who partner with the Program to ensure the best care for our youngest citizens!

thank you

SSMHealth Cardinal Glennon Children's Hospital

Women's and Children's Hospital
University of Missouri Health Care

Children's Mercy KANSAS CITY

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Bureau of Genetics and Healthy Childhood
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Congenital Heart Disease Programs
573.751.6266 or 800.877.6246

Missouri State Newborn Screening Laboratory
573.751.2662
www.health.mo.gov/newbornscreening