

Behind the Screens

Missouri Department of Health and Senior Services
Newborn Screening Program

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Featured Disorder

Tyrosinemia, Type 1, or TYR I, is a rare genetic condition affecting about one in every 100,000 individuals worldwide. It is caused by the body's inability to break down amino acids in the body. Amino acids are considered the building blocks of proteins. There are three types of tyrosinemia; however, type 1 is considered the most severe. Possible signs and symptoms include diarrhea, vomiting, tiredness, irritability, yellowing of the skin, easy bruising or bleeding, swelling of the legs or abdomen, and developmental delays. Symptoms are often triggered by fasting, illness and infection.

Treatment for TYR I includes dietary restrictions, specialized formulas and foods, vitamin D supplementation and a medication called nitisinone. Early detection and treatment for TYR I can prevent the most severe effects of the disorder, including liver, kidney and brain damage. If you or someone you know has been affected by TYR I, support is available from the Tyrosinemia Society and the Network of Tyrosinemia Advocates. If you would like to learn more about Tyrosinemia, or any other disorder Missouri screens for, visit Baby's First Test.



Provider Tip

Calibrate hearing screening devices (OAE/ABR) regularly to ensure they are properly maintained in accordance with manufacturer guidelines. This helps avoid false results and reduces the need for repeat screenings. If you require assistance, contact the manufacturer of your equipment or the Newborn Hearing Screening program at 573-751-6266.

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Patient Spotlight

Grady



"Grady was diagnosed at 6 months with Infantile Onset Pompe Disease. Pompe disease is caused by mutations in a gene that makes the enzyme acid alpha-glucosidase (GAA). This enzyme is needed to break down glycogen. Glycogen is a type of sugar stored in muscle. Without this enzyme, people experience progressive weakening of their muscles. Grady was diagnosed when he failed to meet milestones. At his 6-month appointment, Grady's doctor became concerned about his weight and his low muscle tone. Blood work showed extremely high CK levels. We were immediately admitted to the University of Missouri Children's Hospital. There, we met Dr. Hillman and our genetic counselor, Dawn Peck. They immediately got to work trying to diagnose Grady. It was discovered that day that Grady's heart was also enlarged and had been missed on an X-ray that he had back at our hometown hospital at 4 months. Knowing this was a sign of Pompe disease and being pretty confident in his diagnosis based on his symptoms, they sent Grady's blood sample to the state newborn screening (NBS) lab. Missouri NBS was in the stages of validating the testing of Pompe Disease, but it was not yet available for full population screening. Dr. Hillman and Dawn figured they could help Grady the quickest way possible by running that sample quickly and also help the NBS lab by giving them a potentially positive result to help validate their test and go on to help screen newborn babies being born in Missouri.

Pompe disease was added to Missouri newborn screening in January 2013, about nine months after Grady's diagnosis. Missouri was one of the first states to add Pompe to newborn screening. Without diagnosis and treatment, most babies with Pompe will not live to their first birthday. Early diagnosis is lifesaving with this disease.

Grady started receiving enzyme replacement therapy (ERT) when he was 6 months old and has had hundreds and hundreds of infusions. He receives biweekly IV infusions of Nexviazyme. These infusions replace the missing enzyme and last about 4-6 hours.

Grady has overcome major obstacles and accomplished things that doctors said he would never do, from walking to talking. He has always proved everyone wrong. Grady is now a normal teenage boy who loves football, video games, his dogs, hanging out on the farm, and just relaxing at home.

While our lives may look a bit different than we ever imagined, we are so thankful for the life we have been blessed with. We know it could have had a much grimmer outcome, without the help of our amazing medical team."

Meghan, Grady's Mom

Share Your Story!

If you or someone you know would like to share your newborn screening story, please contact the program via the patient spotlight form.

DID YOU KNOW?



The “Detach This Copy and Give to Mother of Newborn!” sheet is on the front of every blood spot collection form. This sheet provides parents with vital information about Missouri’s blood spot screening program. The front and back efficiently outline the screening, why it is crucial, Missouri’s newborn screening sample and release policy, parents’ rights, benefits of storing leftover samples, and essential contact information. Prior to collection, it is imperative that parent education includes a review of this sheet by a nurse or provider to ensure parents make informed decisions. Then, at the time of blood spot sample collection, the sheet must be detached and handed to the newborn’s parent/guardian. This rule is outlined in [19 CSR 25-36.010 Testing for Metabolic and Genetic Disorders](#).

INITIAL NEWBORN SCREENING

MO 580-1377

EXP 2031-09-30 LOT 7277826 V8251 REF 10534735 Rev.AI

Detach This Copy and Give to Mother of Newborn!

Dear Parents of Newborn Baby:

To help give your baby a good start, Missouri law requires your baby’s blood to be tested for more than 70 disorders. A few small drops of blood taken from the baby’s heel are put on special paper and then sent to the State Public Health Laboratory for testing.

Many disorders are not noticed at birth. Yet they can put the baby at risk for disability or death if not found early. If results from this screening test are not normal, your healthcare provider will tell you about more testing that needs to be done. Please make sure the hospital staff gives you a Newborn Screening pamphlet.

Once the newborn screening test is done, the Missouri State Public Health Laboratory will store the remaining newborn screening sample for five (5) years. The storage is secure. Missouri state law allows for the stored sample to be used for research. The research may help improve methods for spotting illnesses. The research may also find better ways to test, treat and cure major childhood diseases. Your baby is not identified to the researcher in any way. After five years, the rest of the newborn screening sample will be destroyed.

The law allows the parent or legal guardian the option of not having their baby’s leftover newborn screening sample stored or studied. You may ask the State Laboratory to:

- Give the leftover newborn screening sample back to you
- Destroy the leftover newborn screening sample after the newborn tests are done
- Store the leftover newborn screening sample for 5 years but do not release it for study

If you choose NOT to allow your baby’s leftover newborn screening sample to be studied, select one of the above three options. Then write to the Laboratory Director at:

Missouri State Public Health Laboratory
Newborn Screening Laboratory
101 N. Chestnut Street
Jefferson City, MO 65101

Give this information:
Baby’s name
Baby’s date of birth
Mother’s first and last name
Place where baby was born
The option you selected from above
State if you are the parent and legal guardian
Your current address
Your signature and current date (See other side)

If you have questions or need assistance, please contact the Newborn Screening Laboratory at 573-751-3334.

Benefits of storing newborn screening samples:

There are many reasons why newborn screening samples are kept, many of which benefit your family and other Missouri families.

- In some cases, samples are requested by the family or the baby’s health care team.
- The baby’s sample is available to you for other health-related testing within five years of storage.
- The baby’s sample is available to help identify a missing or deceased child within five years of storage.
- If your child has an illness and is enrolled in a research study, parents may request that their baby’s newborn screening sample be returned to them in order that they may send it to the researcher within five years of storage.

For research purposes, all identifying information is removed from the samples (baby’s name, parent’s name, parent’s address, hospital of birth, etc.). The researcher does not know who the baby is. These samples may be used to:

- Provide quality assurance in the screening.
- Do public health studies and research to help develop newborn screening tests and better understand diseases for the benefit of the general public.
- Search for new markers for chronic diseases such as childhood leukemia, sickle cell disease, autism and diabetes.

Only those research projects that undergo careful scientific and ethical review will be given approval to use newborn screening samples.

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