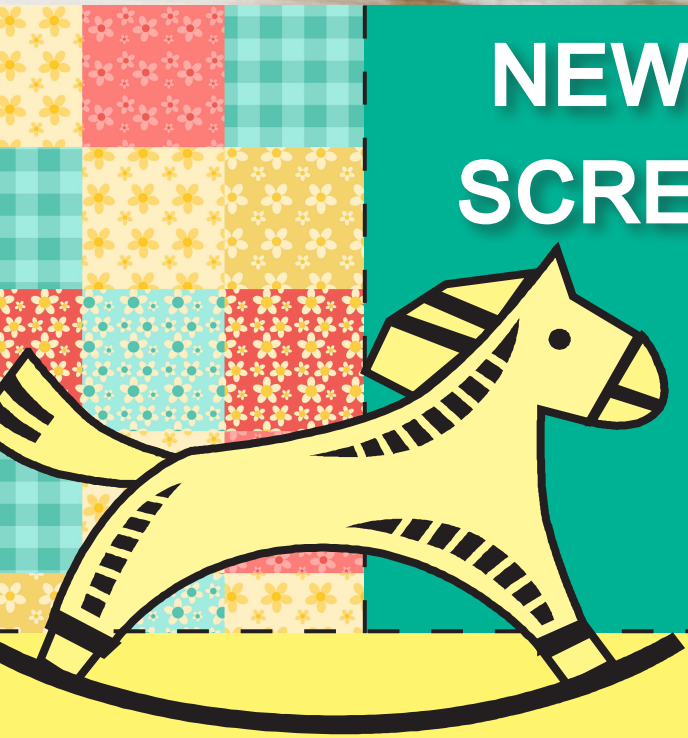
NEWBORN SCREENING



health.mo.gov/newbornscreening

MISSOURI DEPARTMENT OF HEALTH AND SENIOR SERVICES

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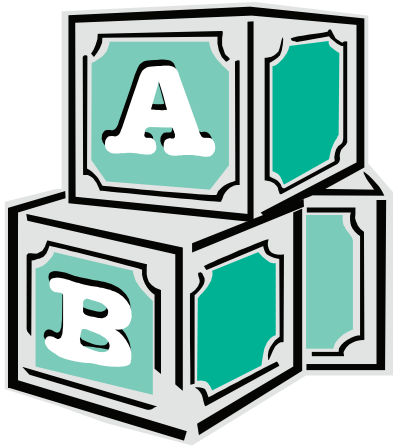


Newborn screening refers to screenings performed on newborns shortly after birth to protect them from the dangerous effects of disorders that otherwise may not be detected for several days, months or even years. Missouri law requires all babies born in the state to be screened for certain rare, but serious conditions. All newborns are screened for more than 70 disorders, including hearing loss and critical congenital heart disease. A small sample of blood is collected from your baby's heel shortly after birth and is then sent to the Missouri Department of Health and Senior Services State Public Health Laboratory for testing.

Whether your baby is born in the hospital or at home, the newborn screen should be collected between 24 and 48 hours of age. The results of your baby's blood screen will be given to your baby's health care provider and the birthing hospital or midwife. Sometimes, more than one newborn screen is needed. If an additional newborn screen is needed you will be notified either by your baby's health care provider, the hospital, midwife, or staff from the Department of Health and Senior Services. If so, it is very important that you bring your baby back for a repeat newborn screen as soon as possible.

Frequently Asked Questions

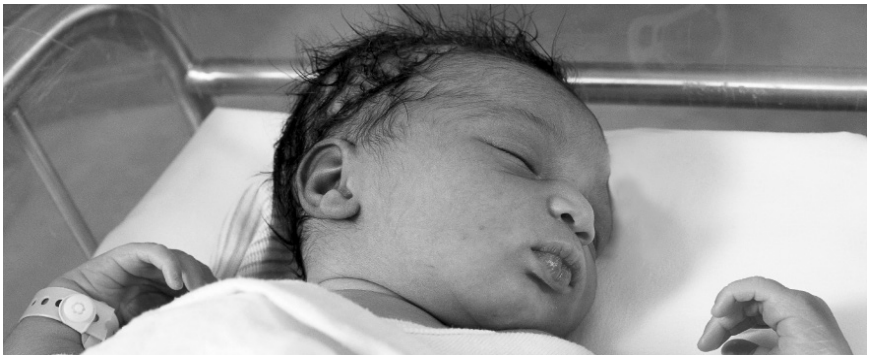
Why is newborn screening important?

The disorders your baby will be screened for are rare. However, these disorders can result in severe injury to the brain, organs or nervous system, and/or may result in death if not treated. Newborn screening helps to identify babies needing diagnosis and treatment, such as a special diet or medication. Since symptoms are not generally noticeable at birth, the only way to find these disorders before permanent damage occurs is by newborn screening. Early treatment will help your baby grow up as healthy as possible.

What if the screening results are abnormal?

A "positive" or abnormal screening result only means that a baby might have a disorder. Sometimes positive screening results are found in babies that do not have the disorder. A diagnosis of a disorder is not made with the first lab test. Further testing will be necessary to determine if your baby actually has the disorder.

If you are asked to repeat the newborn screen or to take your baby to have additional testing, please act quickly so tests can be completed and final results obtained. If needed, treatment must be started as soon as possible to prevent the onset of developmental delays or other damaging results.



When is rescreening needed?

Some things can cause problems with the screening tests. When these things happen, the results of the tests are not reliable. Some of the reasons parents are asked to bring their babies back for rescreening are:

- The specimen was collected too early (less than 24 hours of age).
- The specimen was not collected before a blood transfusion or too soon after a blood transfusion.
- A problem occurred with the quality of the specimen (e.g., problem with collection and/or handling of the dried blood spots).

Is there a cure for the disorders?

There are no cures for most of these disorders. If these disorders are found and a baby gets treatment early, serious problems can be prevented or reduced. If babies with these disorders get early and continuous treatment, most can grow and develop normally and live healthy lives.

What can parents do?

The goal of the Newborn Screening Program is to prevent serious health problems through early screening. You can help!

- Leave two phone numbers (your number and that of a relative, a neighbor or a friend) with the hospital or midwife and health care provider, to assure you can be contacted regarding the screening results.
- Let the hospital or midwife know the name of your baby's health care provider.
- Make sure your baby is screened before you leave the hospital or 24 to 48 hours after birth if your baby is born at home.
- Ask your baby's health care provider or midwife about your baby's newborn screening results.
- Listen to your baby's health care provider and follow directions if more tests or medical appointments are needed.

Do not rely on others to make sure your baby gets tested!

Disorders Included in Newborn Screening

Amino Acid Disorders

This group of disorders prevents a baby's system from breaking down certain waste products in their blood such as phenylalanine, ammonia or other amino acids. Buildup of amino acids and/or bi-products of amino acid metabolism in the blood causes severe medical complications. In each of these disorders, the lack of early identification and treatment may result in serious medical consequences, including developmental delays, failure to thrive and/or death. Early identification and treatment with a special diet or medications can help a baby to grow and develop as healthy as possible.

Amino Acid Disorders screened for include:

- Arginemia (ARG, arginase deficiency)
- Argininosuccinate acidemia (ASA, argininosuccinase)
- Citrullinemia type I (CIT-I, argininosuccinate synthetase)
- Citrullinemia type II (CIT-II, citrin deficiency)
- Defects of biopterin cofactor biosynthesis (BIOPT-BS)
- Defects of biopterin cofactor regeneration (BIOPT-RG)
- Homocystinuria (HCY, cystathionine beta synthase)
- Hyperphenylalaninemia (H-PHE)
- Hypermethioninemia (MET)
- Maple syrup urine disease (MSUD, branched-chain ketoacid dehydrogenase)
- Phenylketonuria (PKU, phenylalanine hydroxylase)
- Tyrosinemia type I (TYR-1, fumarylacetoacetate hydrolase)*
- Tyrosinemia type II (TYR-II, tyrosine aminotransferase)
- Tyrosinemia type III (TYR-III, hydroxyphenylpyruvate dioxygenase)

*There is a lower probability of detection of this disorder during the immediate newborn period.

Biotinidase Deficiency

A disorder found in babies who are missing the enzyme biotinidase. This can lead to seizures, developmental delay, eczema and hearing loss. Problems can be prevented with biotin (a vitamin) treatment started in the first few weeks of life.

Congenital Adrenal Hyperplasia

A disorder caused by an enzyme deficiency that results in the adrenal gland producing too little of one hormone and too much of another. Some babies with this disorder are at risk of sudden death. Problems can be prevented with hormone treatment started early.

Congenital Hypothyroidism

A disorder caused by not having enough thyroid hormone. The most common effects of congenital hypothyroidism are developmental delays and poor growth. If treatment with thyroid medication starts in the first few weeks of life, these children usually develop normally.

Cystic Fibrosis

A disorder that causes thick, sticky mucus to build up in the lungs, digestive system and other organs of the body. This can lead to respiratory and digestive problems, which can be very serious. Early detection and treatment are important for lung health, growth and development. Newborn screening may detect some, but not all, carriers of cystic fibrosis.

Fatty Acid Oxidation Disorders

This is a group of disorders that causes a baby to have trouble using fat for energy. This leads to a buildup of toxic fatty acids, which may cause metabolic crisis. A metabolic crisis can lead to seizures, failure to breathe, cardiac arrest and death and/or result in serious brain damage. However, screening can provide diagnosis before symptoms occur. Early diagnosis and treatment of these disorders allows for proactive treatment and management to prevent or control metabolic crisis effectively.

Fatty Acid Oxidation Disorders screened for include:

- Carnitine acylcarnitine deficiency (CACT)
- Carnitine uptake defect (CUD, carnitine transport defect)*
- Carnitine palmitoyl transferase deficiency I (CPT-1a)
- Carnitine palmitoyl transferase deficiency II (CPT-II)
- Dienoyl-CoA reductase deficiency (DE-RED)
- Glutaric acidemia type II (GA-II, multiple acyl-CoA dehydrogenase deficiency)
- Long-chain hydroxyacyl-CoA dehydrogenase deficiency (LCHAD)
- Medium-chain acyl-CoA dehydrogenase deficiency (MCAD)
- Medium-chain ketoacyl-CoA thiolase deficiency (MCKAT)
- Medium/Short chain L-3-hydroxy acyl-CoA dehydrogenase deficiency (M/SCHAD)
- Short-chain acyl-CoA dehydrogenase deficiency (SCAD)
- Trifunctional protein deficiency (TFP)
- Very long-chain acyl-CoA dehydrogenase deficiency (VLCAD)

Galactosemia

A disorder in which a simple sugar called “galactose” cannot be broken down in the body. Galactose is found in breast milk, many formulas and milk products. If it remains at high levels in the body, galactose will harm the baby’s eyes, liver and brain. If left untreated, classical galactosemia results in death. When started early, a special diet can prevent these problems.

Sickle Cell Disease

Sickle cell disease is mainly found in African-Americans and those of Mediterranean background. In sickle cell disease, the red blood cells change from the normal round shape to an abnormal sickle shape. These sickled cells can clog blood vessels so not enough oxygen can be carried to the body. Babies with sickle cell disease are more likely to have anemia, episodes of pain, strokes and life-threatening infections. Early treatment with antibiotics, immunizations and parent education can help. These measures can prevent serious infections in childhood, and reduce health problems. It is important that affected children be under the care of a health care provider early in life for medication and other treatment.

Disorders that may be detected include:

- Sickle cell anemia (Hb S/S)
- Sickle hemoglobin-C disease (Hb S/C)
- Sickle beta zero thalassemia disease
- Sickle beta plus thalassemia disease
- Sickle hemoglobin-D disease
- Sickle hemoglobin-E disease
- Sickle hemoglobin-O-Arab disease
- Sickle hemoglobin Lepore Boston disease
- Sickle Hereditary Persistence of Fetal Hemoglobin disease (S/HPFH)
- Sickle "Unidentified"
- Hemoglobin-C beta zero thalassemia disease
- Hemoglobin-C beta plus thalassemia disease
- Hemoglobin-E beta zero thalassemia disease
- Hemoglobin-E beta plus thalassemia disease
- Hemoglobin-H disease
- Homozygous beta zero thalassemia disease
- Homozygous-C disease
- Homozygous-E disorder
- Double heterozygous beta thalassemia disease

Newborn screening may also identify carrier or trait conditions associated with sickle cell disease and other hemoglobin variants.

Lysosomal Storage Disorders

Lysosomal Storage Disorders (LSDs) are a group of genetic disorders that result in enzyme deficiencies within the lysosomes of the body's cells. Lysosomes are the recycling bins of the cells and they use very specific enzymes in order to break down and recycle large unwanted waste products. In the LSDs, one of these enzymes is absent or not working. This causes a buildup of waste products in the lysosomes that results in damage to muscles, nerves and/or certain organs in the body. If left untreated, the severe infantile forms of these disorders lead to critical and irreversible disabilities, requiring continuous and extraordinary care, and ultimately lead to death. Treatments may be available for these disorders if caught early, before symptoms begin to appear in the infant.

Lysosomal Storage Disorders screened for include:

- Fabry disease
- Gaucher disease
- Hurler syndrome
- Krabbe disease
- Pompe disease

Organic Acid Disorders

This group of disorders prevents a baby's system from removing certain waste products of proteins and other substances from their blood. These disorders can have a variety of effects on babies from mild to severe including metabolic crisis and problems with the heart, muscles and some organs. Babies with these disorders may become rapidly sick, have seizures, go into a coma and could die without treatment.

Organic Acid Disorders screened for include:

- 2-Methyl-3-hydroxybutyric aciduria (2M3HBA)
- 2-Methylbutyryl-CoA dehydrogenase deficiency (2MBG, SBCAD)
- 3-Hydroxy 3-methylglutaric aciduria (HMG, 3-Hydrox 3-methylglutaryl-CoA lyase)
- 3-Methylcrotonyl-CoA carboxylase deficiency (3-MCC)

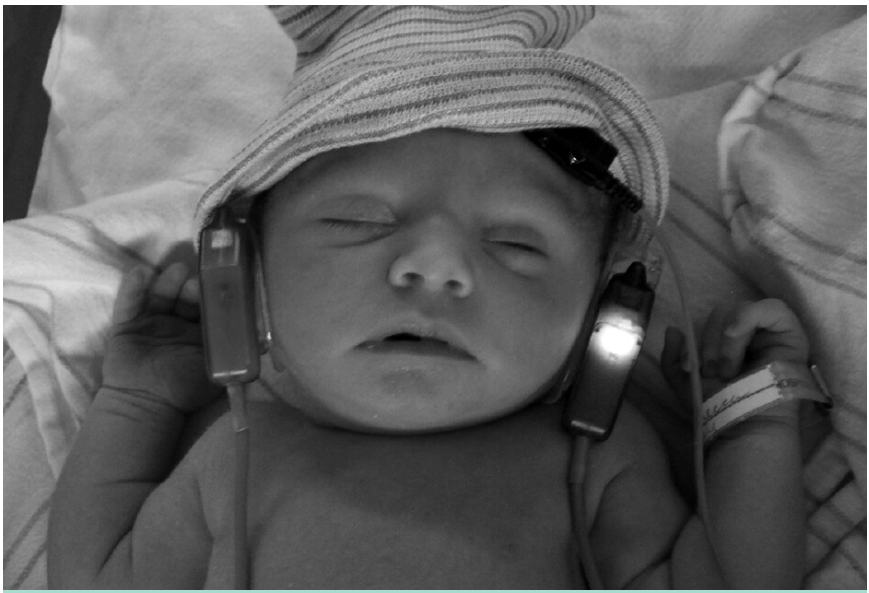
- 3-Methylglutaconic aciduria (3MGA, Type I hydratase deficiency)
- Beta ketothiolase (BKT, mitochondrial acetoacetyl-CoA thiolase, short-chain ketoacyl thiolase)
- Glutaric acidemia type I (GA-1, glutaryl-CoA dehydrogenase)
- Isobutyryl-CoA dehydrogenase deficiency (IBG)
- Isovaleric acidemia (IVA, Isovaleryl-CoA dehydrogenase)
- Malonic acidemia (MAL, malonyl-CoA decarboxylase)
- Methylmalonic acidemia (CBL A,B; vitamin B12 disorders)
- Methylmalonic acidemia (CBL C,D)
- Methylmalonic acidemia (MUT, methylmalonyl-CoA mutase)
- Multiple carboxylase deficiency (MCD, holocarboxylase synthetase)
- Propionic acidemia (PROP, propionyl-CoA carboxylase)

Severe Combined Immunodeficiency

Severe Combined Immunodeficiency (SCID), sometimes in the past referred to as "Bubble Boy Disease," is a rare, but very serious genetic defect characterized by extremely low counts of T-cells, which are a specific type of white blood cell. White blood cells are responsible for your immune system's ability to fight off infection. Babies affected by SCID may appear normal and healthy for the first few months of life until the immunity given to them by their mother begins to disappear. Children with a normal immune system can fight off everyday germs, but children with SCID will not be able to recover from something as simple as the common cold. Without early detection and treatment, children affected by SCID will die of infections before their first or second birthday. Treatment with bone marrow transplant within the first few months of life, before infection has occurred, can cure SCID in most cases.

Newborn Hearing Screening

All babies receive a safe and painless hearing screening shortly after birth to identify possible hearing loss. Hearing loss is one of the most common birth defects, occurring in 1 to 3 infants per 1,000. If hearing loss is not detected and managed early, it can impede speech, language and cognitive development. If the final screening results indicate your baby may have hearing loss, it is important to work with your health care provider to make an appointment with an audiologist to perform a more thorough hearing test before your baby is 3 months old.



Do you know the results of your baby's hearing screening?

Critical Congenital Heart Disease

Critical congenital heart disease (CCHD) is the name given to specific congenital heart defects. These defects in the heart occur before birth, cause blood to flow in an abnormal pattern and may lead to blockage of blood flow throughout the body. If left untreated, these defects can lead to death or can cause serious developmental delay.

CCHD screening is a simple bedside test to determine the amount of oxygen in the baby's blood. Low oxygen levels can be a sign of CCHD. The test is done using a machine called a pulse oximeter. The pulse oximeter is an infrared light sensor that is gently wrapped around the baby's hand or foot. Light passes through the skin and tissue and is read by the sensor to estimate the blood oxygen level. The test is painless and takes just a few minutes.

If the results are "negative," it means that the baby's test results did not show signs of a CCHD. This type of screening test does not detect all CCHDs, so it is possible to still have a CCHD or other congenital heart defect with a negative screening result. If the results are "positive" ("fail" or out-of-range result), it means that the baby's test results showed low levels of oxygen in the blood, which can be a sign of a CCHD. This does not always mean that the baby has a CCHD. It just means that more testing is needed.

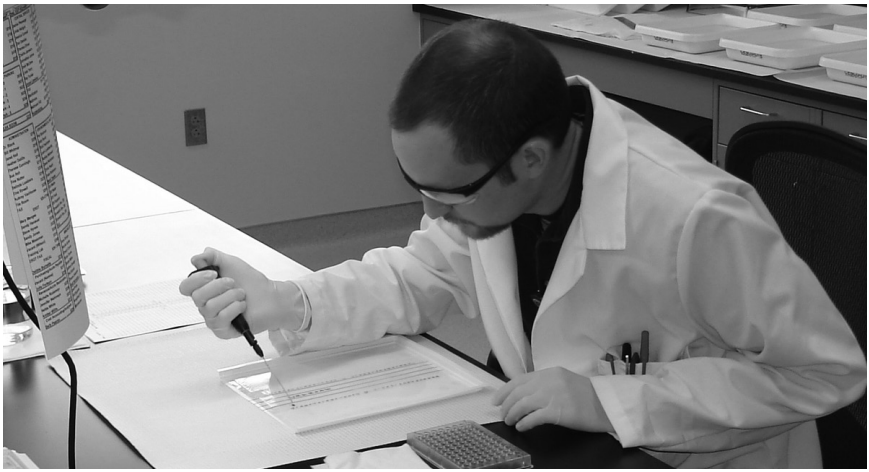
Pulse oximetry screening is most likely to detect seven specific CCHDs. These include:

- Pulmonary atresia
- Tetralogy of fallot
- Tricuspid atresia
- Truncus arteriosus
- Hypoplastic left heart syndrome
- Total anomalous pulmonary venous return
- Transposition of the great arteries

Newborn Screening Samples

Missouri Newborn Screen Sample Storage and Release Policy

Once the newborn screening test is completed, the Missouri State Public Health Laboratory will store the remaining newborn screening sample for five 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 detecting 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.



Scientist at the Missouri State Public Health Laboratory, Jefferson City, Missouri, testing newborn screening samples.

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 extra newborn screening sample back to you.
- Destroy the newborn screening sample after the newborn tests are completed.
- Store the extra newborn screening sample for five 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 three options from above and write to the laboratory director at:

Missouri State Public Health Laboratory
Newborn Screening Laboratory
P.O. Box 570
Jefferson City, MO 65102

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

If you have questions or need assistance, please contact the Newborn Screening Laboratory at 573-751-2662.

health.mo.gov/lab/newborn

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 newborn 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.

Special Notes

The newborn screening tests are not diagnostic. They are a "screen" designed to detect newborns who need further testing to determine if they have certain disorders. The screening tests are very efficient and provide newborns with the best opportunity for having the disorders identified early. However, like most laboratory tests, the tests used for newborn screening cannot guarantee that every affected newborn will be identified, or that only infants at higher risk of being affected will be identified. Therefore, it is important to recognize that there will be some "false positives" (newborns with a positive or abnormal screen result who are later found to have normal results), and the possibility of "false negatives" (newborns with normal screening results who are later found to have one of the conditions).

Medical Homes

The Missouri Department of Health and Senior Services supports and encourages access to a medical home for all children, with and without special health care needs. A medical home is defined by the Health Resources and Services Administration as, "a home base for any (person's) medical and non-medical care. The medical home is a cultivated partnership between the patient, family, and primary provider in cooperation with specialists and support from the community."

For more information on medical homes, please visit:
http://www.hrsa.gov/healthit/toolbox/Childrenstoolbox/BuildingMedicalHome/whyimportant.html

For more information on newborn screening, please call the Missouri Department of Health and Senior Services at 800-877-6246 or visit the newborn screening website at health.mo.gov/newbornscreening.

Additional Resources

www.babysfirsttest.org
www.marchofdimes.org
www.aap.org

Questions for My Doctor

Missouri Department of Health and Senior Services
Division of Community and Public Health
P.O. Box 570
Jefferson City, MO 65102
Phone: 800-877-6246 Fax: 573-751-6185
www.health.mo.gov
For information regarding other Maternal and
Child Health Services call: 800-TEL-LINK (800-835-5465)

Alternate forms of this publication for persons with disabilities may be obtained by contacting the Missouri Department of Health and Senior Services at the number listed above.

Citizens who are hearing or speech impaired can dial 711.

AN EQUAL OPPORTUNITY/AFFIRMATIVE ACTION EMPLOYER
Services provided on a nondiscriminatory basis.

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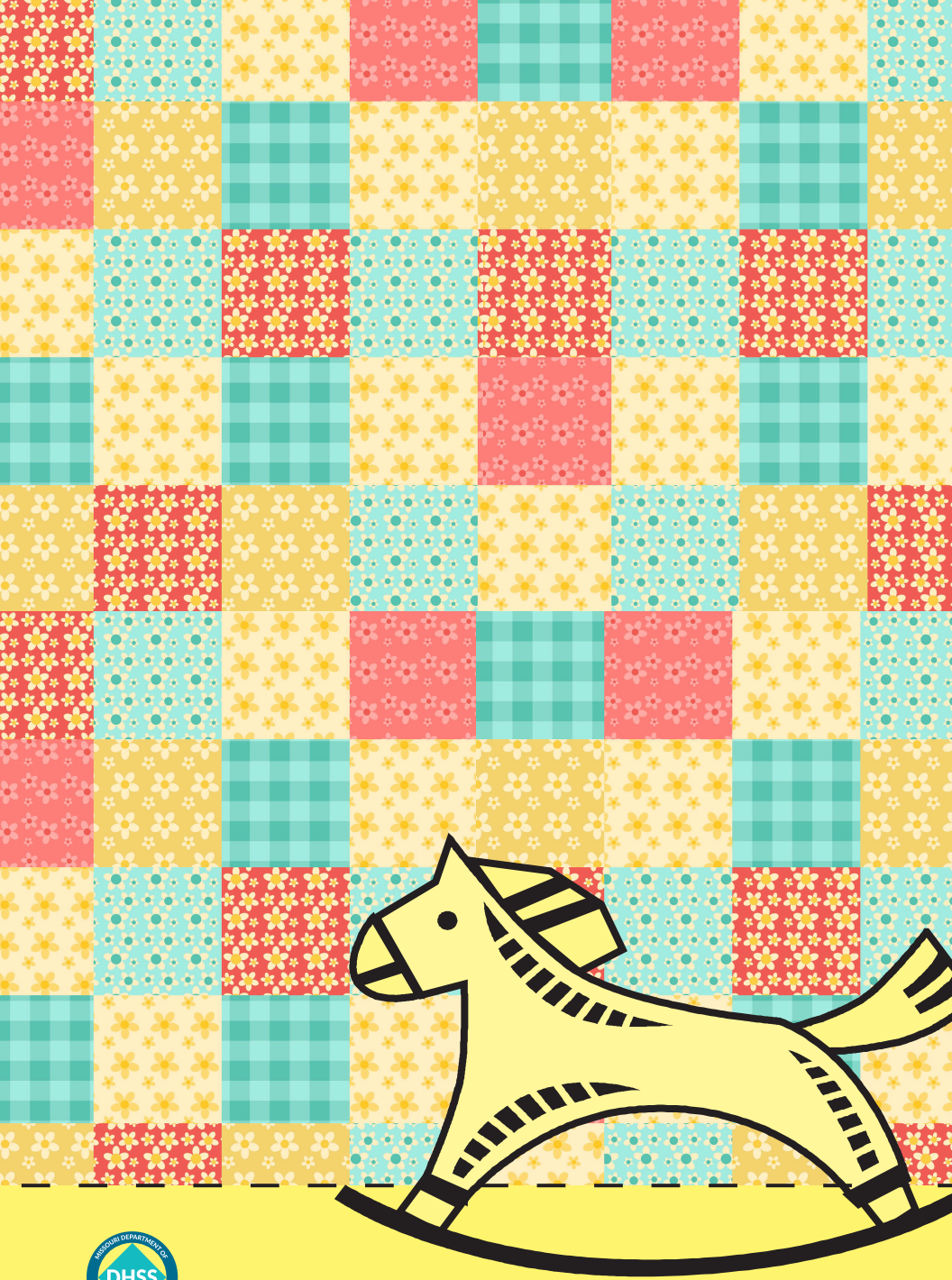

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