Newborn Screening FACT SHEET

What is newborn screening?

Newborn screening identifies babies at risk for conditions that can affect their long-term health or survival. Most of these conditions cannot be seen at birth but can be treated or helped if found early. Newborn screening includes blood spot, hearing, and pulse oximetry screening.

Blood spot screening checks for over 60 conditions, a list of which can be found in the box to the right. Early detection can help prevent serious health problems, disability, and even death.

Hearing screening checks for hearing loss in the range where speech is heard. Identifying hearing loss early helps babies stay on track with speech, language, and communication skills.

Pulse oximetry screening checks for a set of serious, life-threatening heart defects known as critical congenital heart disease (CCHD). If detected early, babies with CCHD can often be treated with surgery or other medical interventions.

Blood spot screening checks babies for:

Arginemia
Argininosuccinate acidemia
Beta ketothiolase deficiency
Biopterin cofactor defects (2 types)
Biotinidase deficiency
Carnitine acylcarnitine translocase deficiency
Carnitine palmitoyltransferase deficiency (2 types)
Carnitine uptake defect
Citrullinemia (2 types)
Congenital adrenal hyperplasia
Congenital cytomegalovirus (cCMV)
Congenital hypothyroidism
Cystic fibrosis
Dienoyl-CoA reductase deficiency
Duchenne muscular dystrophy (DMD)
Galactokinase deficiency
Galactoepimerase deficiency
Galactosemia
Glutaric acidemia (2 types)
Guanidinoacetate methyltransferase (GAMT) deficiency
Hemoglobinopathy variants
Homocystinuria
Hypermethioninemia
Hyperphenylalaninemia
Isobutyryl-CoA dehydrogenase deficiency
Isovaleric acidemia
Krabbe disease
Long-chain hydroxyacyl-CoA dehydrogenase deficiency
Malonic acidemia
Maple syrup urine disease
Medium-chain acyl-CoA dehydrogenase deficiency
Medium/short-chain hydroxy acyl-CoA dehydrogenase deficiency
Medium-chain keto acyl-CoA thiolase deficiency
Methylmalonic acidemia (3 types)
Mucopolysaccharidosis type I
Multiple CoA carboxylase deficiency
Phenylketonuria
Pompe disease
Primary T-cell lymphopenias
Propionic acidemia
Severe combined immunodeficiency
Short-chain acyl-CoA dehydrogenase deficiency
Sickle cell disease
Sickle-C disease
Spinal muscular atrophy (SMA)
S-βeta thalassemia
Trifunctional protein deficiency
Tyrosinemia (3 types)
Very long-chain acyl-CoA dehydrogenase deficiency
X-linked adrenoleukodystrophy
2-Methyl-3-hydroxybutyric acidemia
2-Methylbutyryl-CoA dehydrogenase deficiency
3-Hydroxy-3-methylglutaryl-CoA lyase deficiency
3-Methylcrotonyl-CoA carboxylase deficiency
3-Methylglutaconyl-CoA hydratase deficiency

What happens to the remaining blood spots and results after screening?

Following newborn screening, test results and any leftover blood spots are stored to allow for follow-up testing, if needed. Stored blood spots and test results are also used for making sure screening is accurate, improving tests, and developing new newborn screening tests. They are not used for research without the parent's written informed consent.

Parents have options for the storage of their child's blood spots and test results. You may request that your child's blood spots and results be destroyed, or you may request to obtain the blood spots through your child's primary care provider at any time. You may also choose to allow your child's blood spots and results to be used for research. Ask your provider or visit the Newborn Screening Program website for forms and instructions on how to request these options.

When will I get my baby's results?

Your baby's hearing and pulse oximetry screen results will be available on the same day of screening. Discuss the results with your provider.

The blood spot screening process takes a few days, but your baby's primary care provider will contact you as soon as possible if the results suggest a problem. The first well-child visit is also a good time to talk to your baby's primary care provider about results, as results are typically available when your baby is about a week old.

What personal information is provided to the Minnesota Department of Health (MDH)?

The newborn screening card that is sent to MDH for testing contains only the information about the birthing parent and baby that will help staff interpret test results and contact your baby's primary care provider if more testing or follow-up is needed. This includes, but is not limited to, baby's name, date of birth, time of birth, birthing parent's name, and the name of baby's primary care provider or clinic.

Can I refuse screening for my baby?

Yes. If you do not want your baby screened, you must complete the *Parental Refusal or Delay of Newborn Screening* form. You can ask your birth provider for a copy of the form or download it from the Newborn Screening Program website.

For more information on newborn screening:

Minnesota Newborn Screening
www.health.state.mn.us/newbornscreening

MN Early Hearing Detection & Intervention Program
www.health.state.mn.us/people/childrenyouth/improveehdi/state.html

U.S. Health Resources & Services Administration
https://newbornscreening.hrsa.gov

Baby's First Test
www.babysfirsttest.org

To obtain this information in a different format, call (800) 664-7772

Printed on recycled paper

DEPARTMENT OF HEALTH

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