

# 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  
Biotpterin 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  
Galactokinase deficiency  
Galactoepimerase deficiency  
Galactosemia  
Glutaric acidemia (2 types)  
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-beta 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](http://www.health.state.mn.us/newbornscreening)

### MN Early Hearing Detection & Intervention Program

[www.improveehdi.org/mn](http://www.improveehdi.org/mn)

### U.S. Health Resources & Services Administration

<https://newbornscreening.hrsa.gov>

### Baby's First Test

[www.babysfirsttest.org](http://www.babysfirsttest.org)



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