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## Newborn Screening

Your newborn infant has screening tests before leaving the hospital. There may be different tests depending on the state where you live. They include:

- Tests on a few drops of blood from pricking the baby's heel. The tests look for inherited disorders. All states test for at least 30 of these conditions.
- A hearing test that measures the baby's response to sound
- A skin test that measures the level of oxygen in the blood. This can tell if the baby has a congenital heart defect.

These tests look for serious medical conditions. If not treated, some of these conditions can cause lifelong health problems. Others can cause early death. With early diagnosis, treatment can begin right away, before serious problems can occur or become permanent.

If a screening shows that your baby might have a condition, the health care provider or the state health department will call you. It is important to follow up quickly. Further testing can verify whether your baby has the condition. If so, treatment should start right away.

*NIH: National Institute of Child Health and Human Development*

## Start Here

- How Is Newborn Screening Done? [<https://www.nichd.nih.gov/health/topics/newborn/conditioninfo/how-done>]  (Eunice Kennedy Shriver National Institute of Child Health and Human Development)  
Also in Spanish [<https://espanol.nichd.nih.gov/salud/temas/newborn/informacion/realiza>]
- Newborn Screening [<https://www.cdc.gov/newborn-screening/>] (Centers for Disease Control and Prevention)
- Newborn Screening Tests [<https://kidshealth.org/en/parents/newborn-screening-tests.html>] (Nemours Foundation)  
Also in Spanish [<https://kidshealth.org/es/parents/newborn-screening-tests.html>]
- What Disorders Are Newborns Screened for in the United States?  
[<https://www.nichd.nih.gov/health/topics/newborn/conditioninfo/disorders>]  (Eunice Kennedy Shriver National Institute of Child Health and Human Development)  
Also in Spanish [<https://espanol.nichd.nih.gov/salud/temas/newborn/informacion/trastornos>]

## Related Issues

- How Are My Newborn's Screening Results Used? [<https://www.nichd.nih.gov/health/topics/newborn/conditioninfo/how-used>]  (Eunice Kennedy Shriver National Institute of Child Health and Human Development)  
Also in Spanish [<https://espanol.nichd.nih.gov/salud/temas/newborn/informacion/utilizan>]

## Specifics

- Galactosemia Tests [<https://medlineplus.gov/lab-tests/galactosemia-tests/>]  (National Library of Medicine)  
Also in Spanish [<https://medlineplus.gov/spanish/pruebas-de-laboratorio/prueba-de-galactosemia/>]
- Hearing Tests for Children [<https://medlineplus.gov/lab-tests/hearing-tests-for-children/>]  (National Library of Medicine)

Also in Spanish [<https://medlineplus.gov/spanish/pruebas-de-laboratorio/pruebas-de-audicion-para-ninos/>]

- Newborn Screening Tests for Your Baby [<https://www.marchofdimes.org/find-support/topics/parenthood/newborn-screening-tests-your-baby>] (March of Dimes Foundation)  
Also in Spanish [<https://nacersano.marchofdimes.org/bebe/pruebas-de-deteccion-para-recien-nacidos.aspx>]
- Phenylketonuria (PKU) Screening [<https://medlineplus.gov/lab-tests/phenylketonuria-pku-screening/>]  
 (National Library of Medicine)  
Also in Spanish [<https://medlineplus.gov/spanish/pruebas-de-laboratorio/prueba-de-deteccion-de-fenilcetonuria-fcu/>]
- Screening for Critical Congenital Heart Disease in Newborns  
[<https://www.ahajournals.org/doi/10.1161/CIRCULATIONAHA.113.008522>] (American Heart Association)

## Genetics

- 21-hydroxylase deficiency: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/21-hydroxylase-deficiency>]  
 (National Library of Medicine)
- 3-hydroxy-3-methylglutaryl-CoA lyase deficiency: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/3-hydroxy-3-methylglutaryl-coa-lyase-deficiency>]  
 (National Library of Medicine)
- 3-hydroxyacyl-CoA dehydrogenase deficiency: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/3-hydroxyacyl-coa-dehydrogenase-deficiency>]  
 (National Library of Medicine)
- 3-methylcrotonyl-CoA carboxylase deficiency: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/3-methylcrotonyl-coa-carboxylase-deficiency>]  
 (National Library of Medicine)
- 3-methylglutaconyl-CoA hydratase deficiency: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/3-methylglutaconyl-coa-hydratase-deficiency>]  
 (National Library of Medicine)
- Adenosine deaminase deficiency: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/adenosine-deaminase-deficiency>]  
 (National Library of Medicine)
- Alpha thalassemia: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/alpha-thalassemia>]  
 (National Library of Medicine)
- Arginase deficiency: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/arginase-deficiency>]  
 (National Library of Medicine)
- Argininosuccinic aciduria: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/argininosuccinic-aciduria>]  
 (National Library of Medicine)
- Barth syndrome: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/barth-syndrome>]  
 (National Library of Medicine)
- Beta thalassemia: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/beta-thalassemia>]  
 (National Library of Medicine)
- Beta-ketothiolase deficiency: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/beta-ketothiolase-deficiency>]  
 (National Library of Medicine)
- Biotinidase deficiency: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/biotinidase-deficiency>]  
 (National Library of Medicine)
- Carnitine palmitoyltransferase I deficiency: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/carnitine-palmitoyltransferase-i-deficiency>]  
 (National Library of Medicine)
- Carnitine palmitoyltransferase II deficiency: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/carnitine-palmitoyltransferase-ii-deficiency>]  
 (National Library of Medicine)
- Carnitine-acylcarnitine translocase deficiency: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/carnitine-acylcarnitine-translocase-deficiency>]  
 (National Library of Medicine)
- Citrullinemia: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/citrullinemia>]  
 (National Library of Medicine)
- Congenital adrenal hyperplasia due to 11-beta-hydroxylase deficiency: MedlinePlus Genetics  
[<https://medlineplus.gov/genetics/condition/congenital-adrenal-hyperplasia-due-to-11-beta-hydroxylase-deficiency>]  
 (National Library of Medicine)

- Congenital hypothyroidism: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/congenital-hypothyroidism>]  (National Library of Medicine)
- Costeff syndrome: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/costeoff-syndrome/>]  (National Library of Medicine)
- Critical congenital heart disease: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/critical-congenital-heart-disease>]  (National Library of Medicine)
- Cystic fibrosis: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/cystic-fibrosis>]  (National Library of Medicine)
- Dilated cardiomyopathy with ataxia syndrome: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/dilated-cardiomyopathy-with-ataxia-syndrome>]  (National Library of Medicine)
- Ethylmalonic encephalopathy: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/ethylmalonic-encephalopathy>]  (National Library of Medicine)
- Galactosemia: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/galactosemia>]  (National Library of Medicine)
- Glucose-6-phosphate dehydrogenase deficiency: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/glucose-6-phosphate-dehydrogenase-deficiency>]  (National Library of Medicine)
- Glutamate formiminotransferase deficiency: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/glutamate-formiminotransferase-deficiency>]  (National Library of Medicine)
- Glutaric acidemia type I: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/glutaric-acidemia-type-i>]  (National Library of Medicine)
- Glutaric acidemia type II: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/glutaric-acidemia-type-ii>]  (National Library of Medicine)
- Glutathione synthetase deficiency: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/glutathione-synthetase-deficiency>]  (National Library of Medicine)
- Histidinemia: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/histidinemia>]  (National Library of Medicine)
- Homocystinuria: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/homocystinuria>]  (National Library of Medicine)
- HSD10 disease: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/hsd10-disease>]  (National Library of Medicine)
- Hyperlysineemia: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/hyperlysineemia>]  (National Library of Medicine)
- Hypermethioninemia: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/hypermethioninemia>]  (National Library of Medicine)
- Hyperprolinemia: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/hyperprolinemia>]  (National Library of Medicine)
- Inherited thyroxine-binding globulin deficiency: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/inherited-thyroxine-binding-globulin-deficiency>]  (National Library of Medicine)
- Isobutyryl-CoA dehydrogenase deficiency: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/isobutyryl-coa-dehydrogenase-deficiency>]  (National Library of Medicine)
- Isovaleric acidemia: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/isovaleric-acidemia>]  (National Library of Medicine)
- Krabbe disease: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/krabbe-disease>]  (National Library of Medicine)
- Long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/long-chain-3-hydroxyacyl-coa-dehydrogenase-deficiency>]  (National Library of Medicine)

- Malonyl-CoA decarboxylase deficiency: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/malonyl-coa-decarboxylase-deficiency>]  (National Library of Medicine)
- Maple syrup urine disease: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/maple-syrup-urine-disease>]  (National Library of Medicine)
- Medium-chain acyl-CoA dehydrogenase deficiency: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/medium-chain-acyl-coa-dehydrogenase-deficiency>]  (National Library of Medicine)
- Methylmalonic acidemia: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/methylmalonic-acidemia>]  (National Library of Medicine)
- Mitochondrial trifunctional protein deficiency: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/mitochondrial-trifunctional-protein-deficiency>]  (National Library of Medicine)
- Nonketotic hyperglycinemia: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/nonketotic-hyperglycinemia>]  (National Library of Medicine)
- Ornithine transcarbamylase deficiency: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/ornithine-transcarbamylase-deficiency>]  (National Library of Medicine)
- Ornithine translocase deficiency: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/ornithine-translocase-deficiency>]  (National Library of Medicine)
- Phenylketonuria: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/phenylketonuria>]  (National Library of Medicine)
- Primary carnitine deficiency: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/primary-carnitine-deficiency>]  (National Library of Medicine)
- Short-chain acyl-CoA dehydrogenase deficiency: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/short-chain-acyl-coa-dehydrogenase-deficiency>]  (National Library of Medicine)
- Short/branched chain acyl-CoA dehydrogenase deficiency: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/short-branched-chain-acyl-coa-dehydrogenase-deficiency>]  (National Library of Medicine)
- Sickle cell disease: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/sickle-cell-disease>]  (National Library of Medicine)
- Succinate-CoA ligase deficiency: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/sucl2-related-mitochondrial-dna-depletion-syndrome>]  (National Library of Medicine)
- Tetrahydrobiopterin deficiency: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/tetrahydrobiopterin-deficiency>]  (National Library of Medicine)
- Tyrosinemia: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/tyrosinemia>]  (National Library of Medicine)
- X-linked severe combined immunodeficiency: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/x-linked-severe-combined-immunodeficiency>]  (National Library of Medicine)
- ZAP70-related severe combined immunodeficiency: MedlinePlus Genetics [<https://medlineplus.gov/genetics/condition/zap70-related-severe-combined-immunodeficiency>]  (National Library of Medicine)

## Clinical Trials

- ClinicalTrials.gov: Neonatal Screening [<https://clinicaltrials.gov/search?intr=%22Neonatal+Screening%22&aggFilters=status:not%20rec>]  (National Institutes of Health)

## Journal Articles

References and abstracts from MEDLINE/PubMed (National Library of Medicine)

- Article: Speculum-free retinopathy of prematurity screening with a proparacaine-soaked cotton-swab: A randomized... [<https://www.ncbi.nlm.nih.gov/pubmed/40725872>]

- Article: Assessment of the Children's Hospital of Philadelphia Score Algorithm for Screening... [<https://www.ncbi.nlm.nih.gov/pubmed/40717047>]
- Article: Japanese experience of newborn screening for lysosomal storage diseases and adrenoleukodystrophy. [<https://www.ncbi.nlm.nih.gov/pubmed/40708026>]
- Newborn Screening -- see more articles [<https://pubmed.ncbi.nlm.nih.gov/?term=%22Neonatal+Screening%22%5Bmajr%3Aexp%5D+AND+humans%5Bmh%5D+AND+english%5Bla%5D+AND+%22last+1+Year%22+%5Bdat%5D+NOT+%28letter%5Bpt%5D+OR+case+reports%5Bpt%5D+OR+editorial%5Bpt%5D+OR+comment%5Bpt%5D%29+AND+free+full+text%5Bsb%5D+>]

## Reference Desk

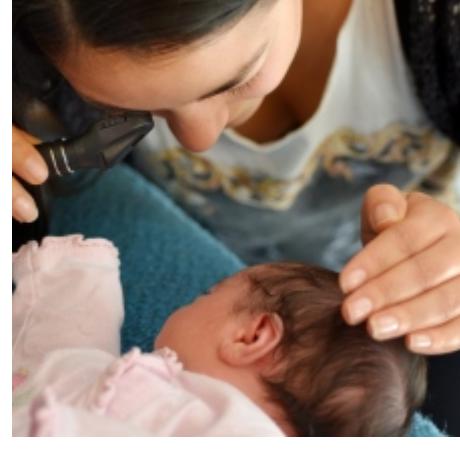
- GeneReviews Glossary [<https://www.ncbi.nlm.nih.gov/books/NBK5191/>]  (National Center for Biotechnology Information)

## Find an Expert

- Eunice Kennedy Shriver National Institute of Child Health and Human Development [<https://www.nichd.nih.gov/>]   
Also in Spanish [<https://espanol.nichd.nih.gov/>]
- March of Dimes Foundation [<https://www.marchofdimes.org/>]  
Also in Spanish [<https://nacersano.marchofdimes.org/>]
- National Human Genome Research Institute [<https://www.genome.gov/>] 

## Patient Handouts

- Newborn screening tests [<https://medlineplus.gov/ency/article/007257.htm>] (Medical Encyclopedia)  
Also in Spanish [<https://medlineplus.gov/spanish/ency/article/007257.htm>]



## MEDICAL ENCYCLOPEDIA

- Aminoaciduria [<https://medlineplus.gov/ency/article/003366.htm>]
- Apgar score [<https://medlineplus.gov/ency/article/003402.htm>]
- Hemoglobin electrophoresis [<https://medlineplus.gov/ency/article/003639.htm>]
- Methylmalonic acid blood test [<https://medlineplus.gov/ency/article/003565.htm>]
- Neonatal cystic fibrosis screening test [<https://medlineplus.gov/ency/article/003409.htm>]
- Newborn screening tests [<https://medlineplus.gov/ency/article/007257.htm>]
- Plasma amino acids [<https://medlineplus.gov/ency/article/003361.htm>]
- Serum phenylalanine screening [<https://medlineplus.gov/ency/article/003362.htm>]
- TORCH Screen [<https://medlineplus.gov/ency/article/003350.htm>]

## Related Health Topics

Genetic Testing [<https://medlineplus.gov/genetictesting.html>]

## National Institutes of Health

The primary NIH organization for research on *Newborn Screening* is the Eunice Kennedy Shriver National Institute of Child Health and Human Development [<https://www.nichd.nih.gov/Pages/index.aspx>]

## Other Languages

Find health information in languages other than English [<https://medlineplus.gov/languages/newbornscreening.html>] on *Newborn Screening*

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