Newborn Screening Tests

What Is Newborn Screening?

Newborn screening is a public health service done in each U.S. state. Every newborn is tested for a group of health disorders that aren't otherwise found at birth.

With a simple blood test, doctors can check for rare genetic, hormone-related, and metabolic conditions that can cause serious health problems. Newborn screening lets doctors diagnose babies quickly and start treatment as soon as possible.

Which Screening Tests Are Offered?

Screening varies by state. Tests offered can change as technology advances and treatments improve. Although there are national recommendations for newborn screening, it is up to each state to decide which tests to include.

Newborn screening includes tests for:

Metabolic problems. Metabolism is the process that converts food into energy the body can use to move, think, and grow. Enzymes are special proteins that help with metabolism by speeding up the chemical reactions in cells. Most metabolic problems happen when certain enzymes are missing or not working as they should. Metabolic disorders in newborn screening include:

- phenylketonuria (PKU)
- methylmalonic acidemia
- maple syrup urine disease (MSUD)
- tyrosinemia
- citrullinema
- medium chain acyl CoA dehydrogenase (MCAD) deficiency

Hormone problems. Hormones are chemical messengers made by glands. Hormone problems happen when glands make too much or not enough hormones. Hormone problems in newborn screening include:

- congenital hypothyroidism
- congenital adrenal hyperplasia

Hemoglobin problems: Hemoglobin is a protein in red blood cells that carries oxygen throughout the body. Some of the hemoglobin problems included in newborn screening are:

- sickle cell disease and sickle cell trait
- hemoglobin SC disease
- beta thalassemia

Other problems. Other rare but serious medical problems included in newborn screening are:

- galactosemia
- biotidinase deficiency
- cystic fibrosis
- severe combined immunodeficiency (SCID)
- Pompe disease (glycogen storage disease type II)
- mucopolysaccharidosis type 1
- X-linked adrenoleukodystropy
- spinal muscle atrophy (SMA)

Most states also screen for hearing loss and critical congenital heart disease, which are not done by testing the blood.

Talk to your doctor if you think your baby may need other newborn screening tests not offered through your state program.

How Is Newborn Screening Done?

A small blood sample taken by pricking the baby's heel is tested. This happens before the baby leaves the hospital, usually at 1 or 2 days of age. Talk to your doctor about newborn screening if your baby was not born in a hospital.

The blood sample should be taken after the first 24 hours of life. Some babies are tested within the first 24 hours, though, because sometimes moms and newborns are discharged within 1 day. If this happens, experts recommend taking a repeat sample after the baby is more than 24 hours old. Some states routinely do two tests on all infants.

When Are the Results Ready?

Results of newborn screening for hearing loss and heart disease are available as soon as the test is done.

Blood test results usually are ready by the time a baby is 5–7 days old. Often, parents won't hear about results if screening tests were normal. They are contacted if a test was positive for a condition. A positive newborn screening test does not mean a child definitely has the medical condition. Doctors order more tests to confirm or rule out the diagnosis. Parents can talk to their child's doctor about the newborn screening results.

If a diagnosis is confirmed, doctors might refer the child to a specialist for more testing and treatment. When treatment is needed, it's important to start it as soon as possible. Treatment may include special formula, diet restrictions, supplements, medicines, and close monitoring.

Visit Baby's First Test for more information on newborn screening and to find out which conditions your state checks for.

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