TEST

Newborn screening tests

Newborn screening uses blood, hearing, and oxygen tests to find certain serious conditions soon after birth.

WHAT IT MEASURES
Blood markers, oxygen level, and hearing responses

SAMPLE
Heel-prick blood; none for oxygen and hearing tests

FASTING NEEDED?
No

HOW LONG IT TAKES
Several minutes per test, usually during the birth stay

RESULTS IN
Immediate to several days, varying by laboratory and program

USED FOR
Screening

MAIN LIMITATION
Positive screens need confirmation; some conditions can be missed

Overview

Newborn screening is a group of tests offered shortly after birth. The aim is to find certain health conditions before they cause noticeable problems, when early assessment or treatment may help.

Screening usually has three parts:

The blood sample is checked for signs of selected genetic, hormone, blood, immune, and metabolic conditions. Metabolic conditions affect how the body processes nutrients or produces energy. The exact conditions included differ between countries, states, and screening programs.

Pulse oximetry helps identify some critical congenital heart defects. These are serious structural heart conditions present from birth. The hearing screen checks how the ears or hearing nerves respond to sound.

These are screening tests, not diagnostic tests. An out-of-range result means your baby needs further assessment. It does not confirm that your baby has a condition.

- A heel-prick blood sample, also called a dried blood spot test
- A hearing screen
- A pulse oximetry test, which measures oxygen in the blood

Why the test is ordered

Many screened conditions do not cause clear signs immediately after birth. Without screening, they may not be found until a baby becomes unwell or misses an important stage of development.

Finding a possible condition early can allow diagnostic testing to begin promptly. Depending on the condition, early care may involve changes in feeding, medicines, surgery, hearing support, or specialist monitoring.

Newborn screening does not test for every health condition. It also does not replace newborn examinations, pregnancy testing, family-history assessment, or medical care when a baby has symptoms.

Who should have the test

Newborn screening is routinely offered to newborn babies. The exact process, timing, consent requirements, and conditions tested for depend on where your baby is born.

Babies born at home or discharged early still need screening arrangements. Ask the midwife, hospital, or newborn screening program how and when testing will happen.

Premature or unwell babies are also screened, but they may need repeat samples. Treatments used in neonatal care can affect results. Babies transferred between hospitals need clear records showing which tests have been completed.

Parents may sometimes decline screening, depending on local law and policy. Before deciding, ask what conditions are included, what testing involves, and what could happen if a condition is not identified early.

How to prepare

Your baby does not need to fast. Usual breast milk or formula feeds can continue unless the clinical team gives different instructions for another reason.

Tell the screening team if your baby:

If your baby has gone home, bring any screening paperwork to the appointment. Feeding or comforting your baby during the heel prick may reduce distress.

- Was born prematurely or has a low birth weight
- Has received a blood transfusion
- Is receiving intravenous nutrition, oxygen, or medicines
- Has a family history of an inherited condition or childhood hearing loss
- Had prenatal genetic testing or a known pregnancy finding

What happens during the test

For the blood spot test, a healthcare professional warms and cleans your baby's heel, then makes a small prick. Several drops of blood are placed onto special filter paper and allowed to dry. The card is sent to a screening laboratory.

Blood collection is usually performed after your baby has had time to begin feeding. The recommended timing varies by screening program. A sample taken very early may need to be repeated.

For pulse oximetry, a soft sensor is placed around a hand and foot. It shines light through the skin to estimate blood oxygen. The test is painless and usually takes a few minutes when the baby is calm.

Hearing screening uses small sensors or a soft earpiece. One method measures echoes produced by the inner ear. Another measures the hearing nerve and brain's response to quiet clicking sounds. The test is painless and works best while your baby is settled or asleep.

Understanding your results

A result may be reported as within the expected range, out of range, incomplete, or unsuitable for analysis. The wording differs between programs.

An expected result means the screen did not find evidence of the conditions tested at that time. It cannot rule out every condition, and it does not guarantee that a condition will never develop.

An out-of-range or positive screen means more testing is needed. Many babies with positive screens do not have the condition. Screening thresholds are designed to identify babies who

may benefit from prompt follow-up, so some false alarms are expected.

A repeat result may be requested because the first sample was collected too early, contained too little blood, was damaged, or was affected by the baby's age or treatment. Being asked for another test does not by itself mean that a condition is likely.

Reference ranges and interpretation vary by laboratory, age, sex, pregnancy status, medicines, and clinical context. Pregnancy status does not apply to the newborn, but pregnancy history, maternal medicines, and the baby's treatment can influence some results.

What affects the results

Several factors can change a newborn screening result even when the laboratory has performed the test correctly.

The baby's age at collection is especially important. Some blood markers take time to rise, while others change rapidly after birth. Samples collected earlier or later than the program's recommended window can be harder to interpret.

Prematurity, low birth weight, serious illness, and immature liver or kidney function can alter hormone and metabolic markers. This can produce an out-of-range result without confirming an inherited condition.

A recent blood transfusion can replace or dilute the baby's own red blood cells. This may mask some blood disorders or affect enzyme testing. Screening may be needed before transfusion and again later, according to the local program.

Intravenous nutrition can raise certain amino acids and fats measured by blood spot screening. Medicines and treatments, including antibiotics like amoxicillin or doxycycline , steroids like prednisone or hydrocortisone , oxygen, and dialysis, may also affect selected tests. Do not stop treatment for screening.

For pulse oximetry, crying, movement, cold skin, poor circulation, supplemental oxygen, and an incorrectly placed sensor can affect the reading. Altitude may also influence oxygen levels.

Fluid or birth material in the ear canal, background noise, movement, and testing soon after birth can affect hearing screening. These commonly lead to a repeat screen.

Poor blood collection can also make a sample unusable. Too little blood, overlapping drops, contamination, moisture, heat, or delayed transport may prevent reliable analysis.

Accuracy and limitations

Newborn screening is designed to find most babies with the included conditions, but no screening method finds every affected baby. A condition may be missed if its marker has not changed yet, the baby has a milder form, or transfusion or treatment has altered the result.

Screening can also suggest a condition when the baby does not have it. Prematurity, illness, early collection, and normal biological differences are common reasons for these false-positive results.

The blood spot does not diagnose a specific disorder. Depending on the finding, better diagnostic tests may include a fresh blood or urine sample, enzyme testing, genetic testing, or assessment by a metabolic, hormone, blood, or immune specialist.

Pulse oximetry does not find every serious heart defect. Some defects do not lower oxygen levels during the screening period. An echocardiogram, which uses ultrasound to show the heart's structure, is the main diagnostic test when a heart defect is suspected.

A hearing screen can miss mild, progressive, or later-onset hearing loss. If concerns continue, a full assessment by a pediatric hearing specialist is more informative than repeating routine screening indefinitely.

Risks and complications

A heel prick causes brief pain and may leave a small bruise. Infection or continued bleeding is uncommon.

Pulse oximetry and hearing screening are painless. Sensors or adhesive pads may occasionally cause temporary skin redness.

An out-of-range result can cause worry and lead to additional appointments. Prompt confirmatory testing helps resolve whether the screening result represents a condition or a false alarm.

What happens next

Results for oxygen and hearing screening are often available immediately. Blood spot results usually take several days, although timing varies by laboratory and screening program. Results needing urgent follow-up may be communicated sooner.

If results are within the expected range, routine newborn and child health appointments continue. Keep a record of completed screening, particularly if your baby moves between hospitals or regions.

If a screen is incomplete or unclear, another screen may be arranged. If it is positive, the screening team will explain which diagnostic tests are needed and how quickly they should happen. Attend this follow-up even if your baby appears well.

A pending or normal screening result should not delay care for symptoms. Call emergency services if your baby has blue or grey lips or skin, severe difficulty breathing, a seizure, or cannot be woken. Seek urgent medical advice for poor feeding with unusual sleepiness, repeated vomiting, worsening jaundice, fever, or any sudden change that concerns you.

Common questions

Can my baby feed before the heel-prick test?

Yes. Fasting is not usually needed, and normal feeding can continue. Feeding or skin-to-skin contact may help comfort your baby during collection.

Does the time of day matter?

Usually not. Your baby's age in hours or days matters more than the clock time. Follow the timing arranged by the local screening program.

Can my baby exercise or move beforehand?

No preparation is needed. Movement and crying can temporarily interfere with pulse oximetry, so staff may wait until your baby is calm before taking a reading.

Why does my baby need another blood spot?

The first sample may have been early, too small, damaged, or affected by prematurity, transfusion, illness, or treatment. A repeat request does not confirm a health condition.

Does a positive result mean my baby has the condition?

No. It means the screening result crossed a threshold and diagnostic testing is needed. False-positive results occur because screening is designed to avoid missing babies who may need early care.

Can a normal screen miss something?

Yes. Screening covers selected conditions and can occasionally miss one. It also cannot rule out disorders that were not included or hearing and heart problems that develop later.

Sources

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