

Common medical tests for babies



Why babies have medical tests

Medical tests in babies serve several purposes. Screening tests look for selected conditions in infants who appear well, because some metabolic, endocrine, genetic, hearing, or cardiac disorders may not cause immediate visible symptoms. Early identification can allow timely treatment, monitoring, or referral and may reduce the risk of serious complications.

Other tests are performed because a clinician notices a finding during an examination, such as poor weight gain, persistent jaundice, unusual breathing, a heart murmur, feeding difficulty, or reduced responsiveness. These are diagnostic evaluations rather than routine screening. The clinician selects them based on the baby's age, examination, symptoms, and relevant history.

It is useful to distinguish a screening result from a diagnosis. Screening is designed to identify babies who may need additional assessment. Results can be affected by timing, prematurity, illness, transfusions, medications, or technical factors. A healthcare professional should interpret the result and explain whether repeat screening, confirmatory laboratory testing, imaging, or specialist review is appropriate.

Newborn blood-spot screening

The newborn blood-spot test, often called the heel-prick test, is usually collected during the first days after birth. A clinician briefly punctures the baby's heel and places several drops of blood on a special card. The card is sent to a public health or laboratory service, where the sample is tested for a panel of conditions. The exact panel varies by jurisdiction; in the United States, state programs determine which conditions are included.

Depending on the local program, screening may identify conditions involving amino-acid metabolism, fatty-acid oxidation, organic-acid metabolism, hormones, hemoglobin, or other inherited pathways. Examples commonly included in screening programs are phenylketonuria, congenital hypothyroidism, medium-chain acyl-CoA dehydrogenase deficiency, and sickle cell disease. The purpose is not to test for every possible illness, but to detect selected disorders for which early action can be particularly valuable.

Collection is quick, although a baby may cry because of the heel puncture. Holding, feeding, or comforting the baby afterward may help. Some babies need a repeat sample because the first specimen was collected too early, was inadequate, or requires confirmation. Parents should ask how results are delivered and provide reliable contact information before leaving the birth facility. A request for repeat testing should be addressed promptly, while remembering that it does not automatically mean the baby has a disorder.

Hearing screening in the newborn period

Newborn hearing screening checks whether a baby responds normally to certain sounds. It is generally painless and can often be performed while the baby is asleep. Two commonly used technologies are otoacoustic emissions testing, which measures sound generated by the inner ear in response to a stimulus, and automated auditory brainstem response testing, which measures electrical activity associated with the auditory pathway.

A result described as "refer," "did not pass," or requiring rescreening is not the same as confirmed hearing loss. Temporary fluid in the ear, movement, crying, background noise, or an imperfect fit of the probe can affect the first test. The recommended next step may be a repeat screen or a comprehensive

audiologic assessment, depending on the result and the baby's risk factors.

Follow-up matters because hearing supports language, communication, and social development. Even when a newborn passes screening, caregivers and clinicians continue to observe responses to sound during routine visits. Concerns such as inconsistent startle responses, lack of reaction to voices, or limited vocal development should be discussed rather than waiting for a future appointment.

Oxygen screening for critical heart conditions

Pulse oximetry screening estimates the percentage of hemoglobin carrying oxygen in the blood. A small sensor, usually placed on a baby's hand or foot, uses light to obtain an oxygen saturation reading. The test is noninvasive and typically takes only a few minutes. It is commonly performed after the baby is at least 24 hours old, or as close to that timing as practical before discharge.

The screening is intended to help detect certain critical congenital heart defects, especially conditions that may produce low oxygen levels before obvious symptoms develop. A low or borderline result may lead to repeat measurements, assessment by a clinician, or further testing such as echocardiography. Oxygen saturation can also be influenced by lung disease, infection, circulation changes, or other conditions, so the screening result requires clinical interpretation.

Pulse oximetry does not detect every heart defect. A baby can have a normal screening result and still develop a cardiac problem or have an abnormality that the screen is not designed to identify. New breathing difficulty, blue or gray coloration, marked sleepiness, sweating or tiring during feeds, or poor feeding requires prompt medical attention.

What happens at routine well-baby visits

Routine checkups combine observation, physical examination, preventive care, and developmental surveillance. The clinician measures weight, length, and head circumference and compares the pattern over time with standardized growth references. One measurement is less informative than the trajectory, so bringing prior records helps the clinician identify meaningful changes.

The physical examination may include assessment of skin color, hydration, head and fontanelles, eyes, mouth, abdomen, hips, genitalia, muscle tone, and neurologic reflexes. The clinician listens to the heart and lungs, evaluates pulses, and observes breathing. Depending on age and risk factors, the visit may include vision or hearing assessment, review of immunizations, and discussion of feeding, sleep, elimination, and safety.

Developmental screening uses structured questions or standardized tools to examine communication, motor skills, problem-solving, and social-emotional behavior. Screening is different from informal reassurance: it helps identify babies who may benefit from closer observation or early intervention. Behavioral and developmental concerns should be discussed even if the baby appears physically well.

Some examinations are repeated at several visits because infant abilities change rapidly. A clinician may ask whether the baby smiles responsively, makes sounds, tracks objects, reaches, rolls, sits, or uses both sides of the body in expected ways. Milestones have a range, but a loss of previously acquired skills, marked asymmetry, or persistent concern merits timely evaluation.

Additional tests when a concern arises

A baby may need testing outside the standard schedule when symptoms, examination findings, or risk factors warrant it. A blood test can evaluate anemia, infection, inflammation, glucose, electrolytes, bilirubin, or organ function. The specific tests depend on the clinical question. Blood collection may use a heel stick, finger stick, or venous sample; the clinician should explain the reason and expected timing of results.

Urinalysis and urine culture may be used when a urinary infection is suspected, although obtaining a reliable sample in an infant requires an appropriate collection method. Stool testing may occasionally be considered for persistent diarrhea, blood, or unusual gastrointestinal findings. These tests should be ordered and interpreted by a healthcare professional because normal infant variation can resemble disease and because contamination can produce misleading results.

Imaging is selected according to the suspected problem. Ultrasound uses sound

waves and is often used to examine the brain through an open fontanelle, kidneys, abdomen, or hips. An echocardiogram uses ultrasound to assess heart structure and function. X-rays provide information about bones and the chest but involve ionizing radiation, so clinicians use them when the expected benefit justifies the exposure. MRI can provide detailed images without ionizing radiation, but an infant may need specialized preparation or sedation, which requires careful risk assessment.

Genetic testing, metabolic testing, or referral to audiology, cardiology, neurology, ophthalmology, or another specialty may be recommended when screening or examination findings suggest a specific concern. Ask what question the test is intended to answer, whether preparation is needed, when results will be available, and who will contact you.

Preparing for testing and follow-up

Before an appointment, write down feeding patterns, wet and soiled diapers, sleep changes, temperatures, medications or supplements, and any episodes that worried you. Bring discharge papers, newborn screening information, immunization records, and the names of relevant family medical conditions. If the baby has seen another clinician, ask whether records have been transferred.

For many routine tests, no special preparation is needed. Do not withhold feeds, water, or medication unless the clinician specifically gives instructions. A familiar caregiver, feeding, swaddling when appropriate, and a calm environment can make a brief procedure easier. Ask about pain-relief or comfort measures that are suitable for the baby's age and situation.

Results may be available immediately, after several days, or later depending on the test. Confirm the communication plan rather than assuming that no call means a normal result. Keep copies of reports and ask whether a repeat test or follow-up visit is required. If instructions are unclear, contact the testing facility or the baby's healthcare professional.

Seek urgent care for severe breathing difficulty, blue or gray lips or skin, unresponsiveness, a seizure, rapidly worsening illness, or signs of serious dehydration. For babies younger than three months, a measured fever should receive prompt professional guidance. These concerns should not wait for a

scheduled screening appointment.