SCREENING TEST VS. DIAGNOSTIC TEST FOR NEWBORN SCREENING

LEGEND Baby without Inherited Disorder Baby with an Inherited Disorder

All babies born in the United States receive newborn screening and the tests included in these screening panels vary based on each state's requirements. These screening tests are designed to catch everyone who has the screened condition, whether they show symptoms or not. A NEGATIVE screening result is very trustworthy. In other words, these tests are very sensitive.



DIAGNOSTIC TESTS

Because Screening tests are designed to detect the condition, they sometimes incorrectly identify an individual as positive (false positives). In this example, four of the babies from the group above have a positive screening result. A diagnostic test administered by laboratory professionals is necessary to confirm any positive screening results. Diagnostic tests are designed differently from screening tests and are used to test small groups of people who are already suspected to have the condition, such as a positive screening test. Positive results from diagnostic tests are much less likely to be false. In other words, diagnostic tests are more specific than screening tests. In this example, only one of the four babies has a positive diagnostic test, and therefore only this baby has the condition.

A baby is diagnosed with a specific disorder or condition after a positive (newborn) screening test result AND confirmation with a positive diagnostic test result.





NOTE: The combination of screening followed by diagnostic testing helps make sure every child with the condition is identified and gets appropriate treatment. Medical intervention may prevent neonatal death and/or improve the quality of life for a baby with positive test results on newborn screening and the confirmatory diagnostic test.

outreach.amp.org

Disclaimer: The number of circles and squares are not an actual representation of statistically significant occurrences of inherited disorders present in the population and are simply used for educational purposes only.