

NEWBORN SCREENING

TESTING TO DETECT GENETIC, ENDOCRINE, AND METABOLIC DISORDERS IN NEWBORNS



WHY IS NEWBORN SCREENING IMPORTANT?

Newborn screening is intended to detect serious health conditions for which early intervention can greatly improve healthy development and positive outcomes. Each year, more than four million newborns are screened for congenital disorders in the U.S. Of these newborns, 12,500 per year are diagnosed with one of the 34 core conditions detected through screening.¹ These conditions, when left untreated, can result in severe physical and neurological impairment or death.

In the United States, recommendations for newborn screening are developed by the Secretary of Health and Human Services. These recommendations are known as the Recommended Uniform Screening Panel (RUSP). The RUSP is constantly in flux as newly discovered treatments or emerging laboratory technologies enable additional recommendations with two new conditions recommended in 2016.

FEATURED TESTS

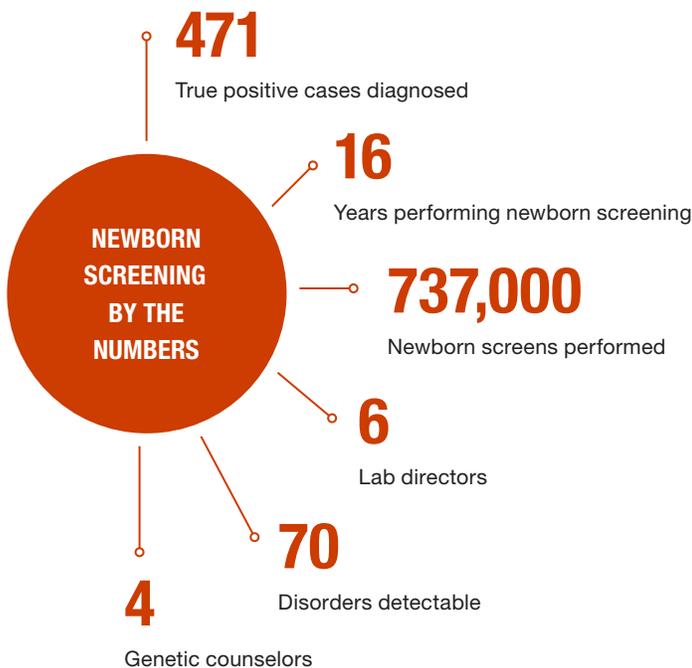
- ▶ **Newborn Screen Recommended Panel, Blood Spot (Mayo ID: NBSR)**
Panel that includes all recommended conditions on the RUSP.
- ▶ **Newborn Screen Expanded Panel, Blood Spot (Mayo ID: NBSE)**
Expanded panel that includes additional disorders beyond the recommended panel.

CONSULT WITH MAYO CLINIC GENETIC COUNSELORS ABOUT TEST OPTIONS

Genetic counselors bring real value to proper test utilization by making sure the most appropriate test has been ordered. Genetic counselors are available to discuss diagnostic testing options and strategies.

Mayo Clinic genetic counselors are available to:

- ▶ Support the ordering process.
- ▶ Provide additional information about testing options.
- ▶ Offer results interpretation.
- ▶ Assist with case review and coordination.



MAYO CLINIC'S SOLUTION TO THE FALSE POSITIVE PROBLEM

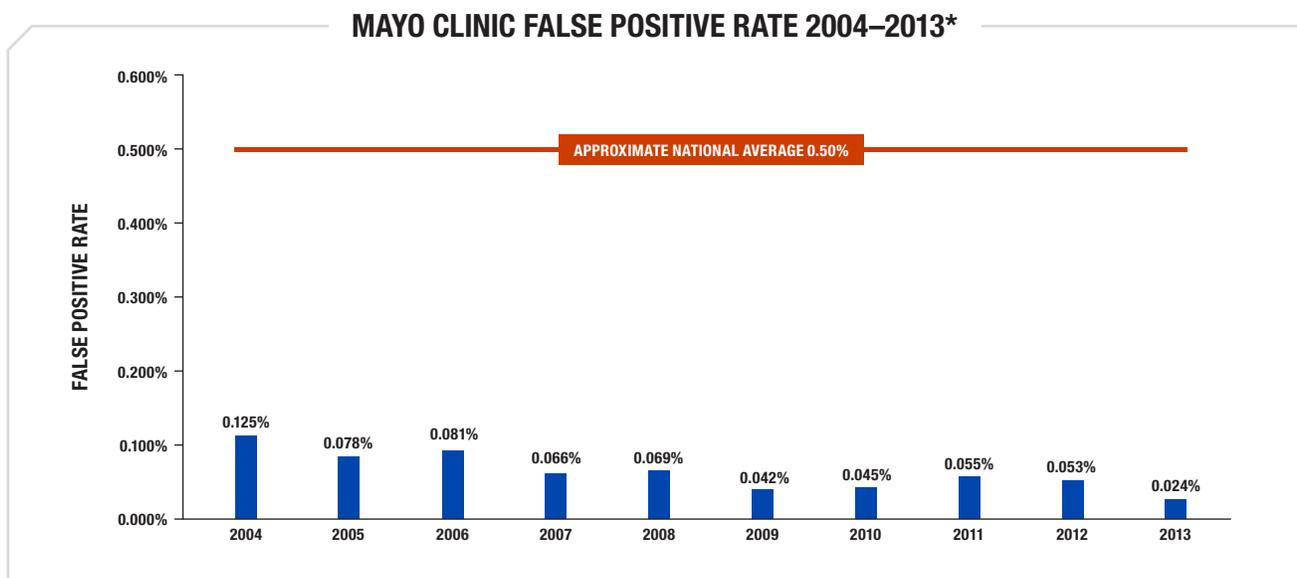
In the U.S., the national false-positive rate is approximately 0.50 percent. False-positives require additional patient contact and unnecessary stress during a critical bonding period. Newborn screening assays at Mayo Medical Laboratories are designed to keep the false positive rate as low as possible. Our strategy has two main post-analytical components: data analysis using an internally-developed multivariate pattern recognition software, Collaborative Laboratory Integrated Reports (CLIR), and an extensive menu of second tier tests.

CLIR SOFTWARE

CLIR software creates and maintains an integrated database of clinical and laboratory data which is used to produce on-demand, post-analytical tools. Our CLIR database consists of laboratory data from thousands of true positive cases submitted by participating collaborators world-wide. Rather than relying on a standard cut-off value, these tools merge the analysis of multiple analytes, each ranked according to clinical significance, into a single score which measures the likelihood of disease. The CLIR score is ranked against the data of true positive cases stored in the database. This analysis is more informative than a simple cut off value traditionally found in newborn screening because it scores how similar is an individual result profile is to those observed in patients known to have a disease.

SECOND TIER TESTING

Second tier tests employ more sensitive methodologies that may not be suitable as a primary, high-throughput screening test. These tests are performed on the original newborn screening specimen when the primary screening assay yields abnormal results. This approach requires no additional patient contact and significantly increases the positive predictive value of the screening. At Mayo Medical Laboratories, all of our newborn screening panels include the performance of second tier testing when appropriate.



* Testing performed via tandem mass spectrometry



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GENETIC COUNSELORS

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CLINICAL REFERENCES

- CDC. (2012). CDC Grand Rounds: newborn screening and improved outcomes. Morbidity and Mortality Weekly Report 61(21), 390-393. Retrieved March 22, 2017 from <http://www.cdc.gov/mmwr/preview/mmwrhtml/mm6121a2.htm>

TAP INTO THE EXPERTISE OF MAYO CLINIC

The Mayo Clinic Biochemical Genetics Laboratory is an interdisciplinary group of physicians, scientists, genetic counselors, and laboratory professionals that includes six laboratory directors and four genetic counselors specializing in newborn screening and diagnostic testing for inborn errors of metabolism. This team provides testing and result interpretation of the highest quality for the diagnosis and clinical care of patients with inborn errors of metabolism and has a long track record of innovative test improvement and test development to achieve newborn screening with the highest sensitivity and specificity, paying particular attention to reducing false positive rates. Offering one of the most comprehensive test menus for inborn errors of metabolism, more than 150 qualitative and quantitative genetic assays are available for newborn screening, diagnosis, and treatment monitoring.

FOR MORE INFORMATION ABOUT NEWBORN SCREENING, VISIT

MayoMedicalLaboratories.com/NBS