TEST ID: NBSE
NEWBORN SCREENING EXPANDED PANEL, BLOOD SPOT

USEFUL FOR
Presymptomatic identification of disorders for which we can screen using an expanded panel to allow for early initiation of treatment and consequent improvement in the long-term prognosis of affected patients.

CLINICAL INFORMATION
Newborn screening as a public health measure was initiated in the early 1960s for the identification of infants affected with phenylketonuria (PKU). Since then, additional genetic and nongenetic conditions were included in screening programs. The goal of newborn screening is to detect diagnostic markers of selected disorders in blood spots collected from presymptomatic newborns. Early identification of affected newborns allows for early initiation of treatment to avoid mortality, morbidity, and disabilities due to these disorders.

The US Secretary of Health and Human Services (HHS) recommends all programs screen for 34 core disorders (www.hrsa.gov/advisorycommittees/mchbadvisory/heritabledisorders/recommendedpanel/). These conditions are considered to fulfill 3 basic principles:

- Condition is identifiable at a period of time (12-48 hours after birth) at which it would not ordinarily be clinically detected.
- Test with appropriate sensitivity and specificity is available.
- Demonstrated benefits of early detection, timely intervention, and efficacious treatment.

The 34 core disorders comprise the Recommended Uniform Screening Panel (RUSP).

Screening tests do not conclusively determine disease status, but measure analytes, which in most cases are not specific for a particular disease. This is the reason why the HHS Secretary also recognizes more than 25 additional conditions as secondary targets that do not meet all inclusion criteria, but are identified nevertheless because most of them are components of the differential diagnosis of screening results observed in core conditions. Even for the secondary conditions, the possibility of making a diagnosis early in life not only helps avoid unnecessary diagnostic testing, but is also beneficial to the patient’s families because genetic counseling and prenatal diagnosis can be offered.

This test includes 32 of 34 core conditions* included in the RUSP and all secondary conditions listed with the RUSP. In addition, it is expanded to include screening for an additional 4 lysosomal storage disorders (Krabbe, Fabry, Gaucher, and Niemann-Pick types A and B diseases), guanidinoacetate methyltransferase (GAMT) deficiency, and glucose-6-phosphate dehydrogenase (G6PD) deficiency. Our screening approach is designed to identify all newborns affected with at least the classic variants of the diseases, but is not expected to detect milder forms of these conditions.

*This test does not screen for critical congenital heart disease and congenital hearing loss, both of which are tested in the nursery using methods other than blood spots (audiometry, pulse oximetry).

REFERENCE VALUES
Negative

ANALYTIC TIME
3 days

CONTENT AND VALUES SUBJECT TO CHANGE. SEE THE MAYO MEDICAL LABORATORIES TEST CATALOG FOR CURRENT INFORMATION.
INTERPRETATION

An interpretive report is provided.

The quantitative measurements of informative metabolites and related ratios and their bioinformatic evaluation using the Collaborative Laboratory Integrated Reports (CLIR) system support the initial interpretation of the complete profile and may suggest the need to perform the measurement of more specific biomarkers using the original newborn screen specimen (second-tier test). Nevertheless, abnormal results are not sufficient to conclusively establish a diagnosis of a particular disease. To verify a preliminary diagnosis, independent biochemical (ie, in vitro enzyme assay) or molecular genetic analyses are required, many of which are offered within Mayo Clinic’s Division of Laboratory Genetics and Genomics.

The reports are in text form only. In a case with a completely normal profile, where the interpretation is reported as negative for all of the listed groups of conditions, no values are provided. A report for an abnormal screening result includes a quantitative result for the relevant abnormal biomarkers including those of a second-tier test when applicable, the CLIR score indicating the similarity of the newborn’s results to those derived from known patients with the relevant disease, a detailed interpretation of the results, and recommendations for additional biochemical testing and confirmatory studies (enzyme assay, molecular analysis).

CLINICAL REFERENCE


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