

Newborn Screening (NBS) Tests - General Information

Use

Screening of newborn infants for certain metabolic, endocrine, and genetic disorders that can adversely affect a child's long-term health or survival.

Clinical Significance

The OSDH PHL NBS Panel includes measurement of analytes of newborns that are associated with the following conditions:

- Biotinidase deficiency
- Congenital adrenal hyperplasia
- Congenital hypothyroidism
- Cystic fibrosis
- Galactosemia
- Hemoglobinopathy
- Fatty acid oxidation disorders
- Organic acid disorders
- Amino acid disorders
- X-Linked adrenoleukodystrophy
- Lysosomal storage disorders
- Severe combined immunodeficiency
- Spinal muscular atrophy

Methodology

Various methodologies are used to measure the different analytes that form part of the NBS panel, including flow injection-tandem mass spectrometry; fluorescence immunoassay analysis; isoelectric focusing gel electrophoresis; polymerase chain reaction (PCR) amplification.

Specimen Type

Dried blood spots (DBS) prepared by applying fresh whole blood on Whatman 903, Ahlstrom PerkinElmer 226, or equivalent filter paper from infants less than 6 months of age, in accordance with the current CLSI Section NBS01-A6 standards.

Note: *Blood must not be applied to the filter paper from EDTA, heparin, or citrate anticoagulated blood.*

Minimum Volume/Size

5 filled circles

Collection

See [Guidance for Collection of NBS Dried Blood Spots](#)

The following NBS collection kits (filter paper/demographics form) are available to birthing facilities, midwives and private physicians free of charge:

- Newborn Screening Kit (ODH #450) – used for NBS and PKU monitoring
- Child/Adult Sick Cell Screening Kit (ODH #485) – used for screening children and adults for hemoglobinopathies (sickle cell)

These forms can be ordered from the OSDH PHL Client Services Department at (405) 444-6494 during regular business hours, email PHLSupplyOrders@health.ok.gov, or submit a Laboratory [Supply Order Request](#) to arrange shipment.

Turn-around Time

Within 5 working days of receipt (7 working days if reflex testing is required)

Reports

NBS test results can be accessed using the [Newborn Screening Results Web Portal](#). Healthcare providers are encouraged to use the portal for accessing current and past NBS test results (results are available from 2002). Written reports are also sent to the infant's designated pediatrician by NBS Follow-up Program, as indicated on the hospital report, and to the sample collection site.

To request access to the NBS Results Web Portal or to ask questions regarding the portal, please call (405) 564-7753 or email PublicHealthlab@health.ok.gov the OSDH PHL.

For healthcare providers located outside of Oklahoma but caring for infants born in Oklahoma:

- For infants under one year old, please fax request on clinic letterhead with infant's name, date of birth, birthing hospital, and mother's name to 405-900-7556.
- For infants older than one year, please fax a signed HIPAA release with the above information on the clinic letterhead to 405-900-7556.

Interpretation

See specific NBS tests for details.

Limitations/Interferences

NBS is performed to assess risk in clinically asymptomatic newborns for a host of disorders; it is not intended to establish diagnosis. Abnormal biomarker levels, identified through screening and evaluated using cutoffs, only indicate that a newborn may be at increased risk for a screened disorder. NBS Follow-up may reach out to healthcare providers to recommend additional testing or provide consultation based on initial test results. The healthcare provider may request additional diagnostic testing to determine if the newborn has the disorder in question.

Healthcare providers should understand that in instances where a newborn has a family history of a disease or is symptomatic, additional diagnostic testing is necessary regardless of the NBS result.

Even though algorithms are used to determine infants at highest risk, NBS may not detect all affected newborns. A positive (i.e., abnormal) screening result is not a confirmed diagnosis of a disorder, and a negative (i.e., normal) screening result is not a confirmed exclusion of a disorder.

CPT Code

Variable depending on specific tests.

Additional Resources

- [OSDH Newborn Screening Follow-up Program](#)
- [American College of Medical Genetics and Genomics](#) - ACT Sheets and Algorithms for specific disorders, provider next steps
- [Baby's First Test](#) - For parents or staff wanting more information