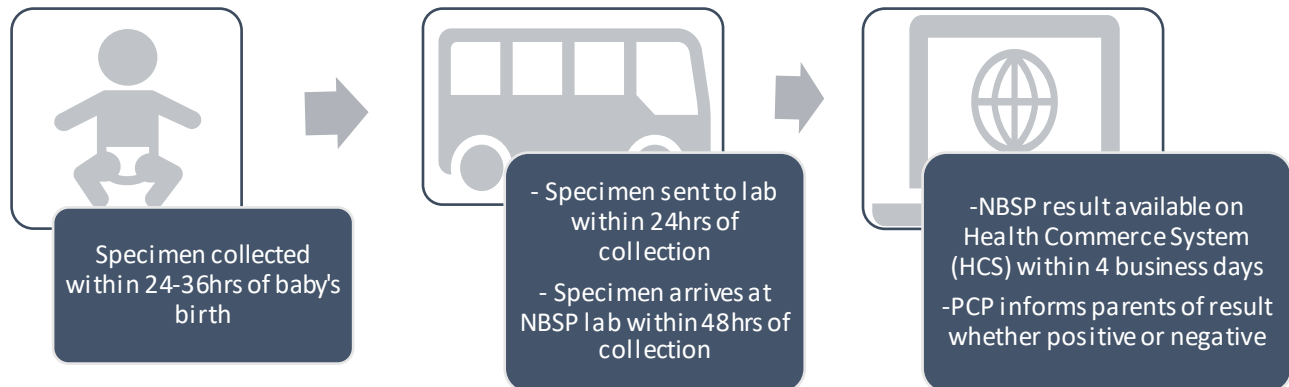


Guide to Newborn Screening for Primary Care Providers in New York State

- Introduction:** The New York State Newborn Screening Program (NYS NBSP) is a public health service provided to all infants born in NYS for early detection of treatable disorders that can affect newborns or children. The NBSP screens for more than 50 disorders that, without treatment, may permanently impact the health and overall quality of life of newborns. Newborn screening is mandated in New York State by Public Health Law, sections 2500-a and 2500-f and written permission from parents is not required to conduct this screening, though they may refuse for religious reasons. There is no charge for the newborn screen in New York State. Approximately 215,00 babies are screened per year and approximately 3% of these are abnormal (borderline or referrals). Timely action taken by providers to obtain repeat specimens or diagnostic testing will improve health outcomes for presumed positive babies. Results are usually reported within 3-4 business days, although abnormal results may be communicated out sooner. The newborn screen is a screen, not a diagnostic test; if a baby shows any signs or symptoms of any of the disorders on the panel, they should be evaluated accordingly.
- “The PKU”:** Although some people still refer to the newborn screen as the “PKU” or the “PKU test”, Phenylketonuria (PKU) was simply the first disorder on the newborn screening panel. The Newborn screening panel is now made up of more than 50 disorders. Please refer to the testing as the “*newborn screen*” and not the “PKU test” to avoid confusion.

Newborn Screening Process:



Newborn Screening Communication:

- Primary Care Physicians (PCP) may communicate with NBSP staff via phone calls, secure emails, faxes, mail, Secure File Transfer on the NYSDOH Health Commerce System website, and/or the internet Case Management System (iCMS). For more information about any of these electronic communication options please contact the NBS Program.
- If you do not have access to look up NBSP reports, reach out to nbsinfo@health.ny.gov with your Health Commerce System (HCS) username to request access.

Types of Newborn Screening Results:

| NBS Report Type | Description | Action Needed | Necessary Follow-up Actions |
|--------------------------------------------------------------|----------------------------------------------------------------------------------------------------------------|----------------------------------------------------------------------------------------|------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| Screen Negative | Results are within acceptable limits for all disorders | None | <ul style="list-style-type: none"> • Provider communicates NBSP result to parents. |
| Sub-optimal or unsuitable or collected at < 24 hours of life | Specimen is poor quality or collected too early and results could not be interpreted for one or more disorders | Repeat specimen required, arranged by HOB and primary care provider (PCP) | <ul style="list-style-type: none"> • NBSP staff sends electronic communication to hospital of birth (HOB) with result. • HOB and NBSP contacts PCP and parents about the need for a repeat specimen via mail. • PCP and/or HOB arranges for repeat specimen collection and sends sample to NBSP. |
| Borderline | Specimen has an abnormal result that is below the referral cut-off for one or more disorders | Repeat specimen or appropriate independent testing required, arranged by HOB and PCP | <ul style="list-style-type: none"> • NBSP staff sends electronic communication to hospital of birth (HOB) with result. • HOB and NBSP contacts PCP and parents about the need for a repeat specimen via mail. • PCP and/or HOB arranges for repeat specimen collection and sends sample to NBSP lab. • If independent testing is done, PCP/HOB should complete diagnosis form and forward independent results to NBSP. |
| Referral | Specimen has an abnormal result above the referral cut-off for one or more disorders | Prompt consultation with Specialty Care Center (SCC) and appropriate follow-up testing | <ul style="list-style-type: none"> • NBSP notifies PCP and SCC of result. • PCP notifies the parents of the result. • PCP and/or SCC orders appropriate confirmatory testing, discuss whether treatment is required. |

Recommendations for Discussing Newborn Screen Results with Families:

- Share specific newborn screening results with parents helping them to understand that a positive screen is serious while assuring them of your help, support and guidance.
- Engage with the family, providing information at their desired pace and level, avoid using complex medical terms.
- Explore the family's emotions and available support systems.
- Discuss with the family a shared concrete and specific plan including what, where, when and how evaluation and if needed, treatment will occur. Provide appropriate resources.
- Looking for more information about a specific disorder? Visit the NBSP website: www.wadsworth.org/programs/newborn/screening/screened-disorders



Primary Care Providers (PCPs) are important stakeholders in the newborn screening system and have regulatory responsibilities. The regulation that dictates how Public Health Law, sections 2500-a and 2500-f are carried out is called "Newborn Screening for Phenylketonuria and Other Diseases," Subpart 69-1 of Title 10 (Health) of the Official Compilation of Codes, Rules and Regulations. *It lists the following responsibilities for PCPs:*



- When notified by the testing laboratory that a repeat specimen is required, the responsible provider shall notify the parent(s) within one (1) business day that a repeat specimen is required from the newborn. The responsible provider, or his or her designee, shall collect the repeat specimen, pursuant to guidance issued by the testing laboratory, and submit it to the testing laboratory as soon as practicable.
- If a satisfactory specimen or repeat specimen is not collected due to newborn mortality, the responsible provider shall submit a written and/or electronic notification to the testing laboratory within five (5) days after death.
- The responsible provider shall document on the newborn's health record the LAB I.D., as it appears on the specimen collection form, the date and time of specimen collection, and all screening results. The responsible provider shall inform parents of the newborn screening result.
- Except when a newborn is under the care of a specialty care center, the responsible provider shall report diagnostic evaluation and test results, case management information, and follow-up reviews to the testing laboratory no later than ninety (90) days following receipt of testing laboratory test results. The responsible provider shall arrange for a diagnostic evaluation and case management with an approved specialty care center as necessary.
- Before performing any tasks relating to collection of specimens, the responsible provider, and any staff under his/her supervision performing specimen collection, shall complete comprehensive specimen collection training, and shall complete such training annually thereafter. The responsible provider shall retain documentation of all such training.
- The responsible provider shall establish written policies and procedures, which shall be available for the Department's review, for:
 - the collection, storage, and shipping of specimens
 - the tracking and disposition of test results.

Common Misconceptions:

- If you do not hear back about your patients' newborn screen results, do not assume they are normal. It is the responsibility of PCPs to look up their patients' NBSP results. The NBS Program and HOB will notify you of abnormal results, but no news may mean that we do not know you are the baby's PCP or have the wrong contact information for your office.
- We do not currently have a mechanism to receive updates about your practice's contact info if you updated it with other DOH entities. Please consider notifying us too if there is a change!

Have any questions or concerns? Please reach out to us, we are here to help.



Newborn Screening Program Contact Information:

Phone: 518-473-7552

Fax 1: 518-474-0405

Fax 2: 518-473-8627

Email: nbsinfo@health.ny.gov

NBSP Website: <https://www.wadsworth.org/programs/newborn/screening>