

Guidance on Heel-stick Specimen Collection for PCP Offices

CONNECTICUT NEWBORN SCREENING PROGRAM • December 2019

The Connecticut Newborn Screening (CT NBS) Program at the State Laboratory will ask your office to collect a heel-stick specimen for the following reasons:

- A NBS heel-stick specimen was never collected because of
 - early discharge from the birthing center
 - home birth
 - parent refusal
- The 1st specimen was collected at less than 24 hours of age and is invalid for amino acid and endocrine disorder screening
- The 1st specimen was unsatisfactory for screening
- The 1st specimen was collected following a transfusion of a blood product
- A borderline out of range result necessitates re-screening

NBS heel-stick specimen collection in the PCP office:

The heel-stick specimen must be collected within 2 business days of receipt of the request from the CT NBS Program (unless otherwise instructed) and sent to the state laboratory within 24 hours of collection. Your office will be asked to expedite collection of the specimen and shipping of the specimen to the state lab if the infant never had an initial newborn screening completed for any reason.

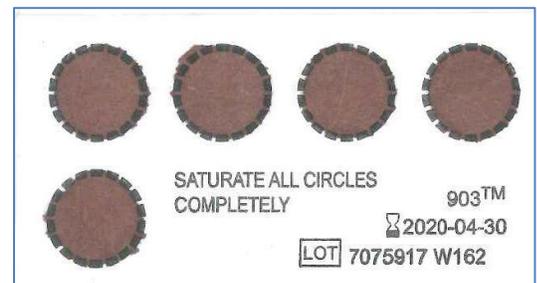
If there are extenuating circumstances that will delay heel-stick specimen collection or there is a family history of the disorder being screened for, please contact the CT NBS Program at the State Laboratory immediately.

The CT NBS Program has heard from multiple practices that they are not equipped to collect NBS specimens in the office. CT NBS Program follow-up staff are available to answer your questions and can provide support and training (860.920.6628 or dph.nbstracking@ct.gov). Please see the next page for answers to frequently asked questions and rationale behind collecting NBS specimens in the PCP office.

Importance of timeliness in newborn screening:

CT's NBS panel has expanded tremendously since its inception in 1964 with testing for PKU. CT currently screens newborns for 64 disorders and anticipates adding 3 additional disorders to the blood-spot panel by mid-2020. A number of these disorders are time critical/sensitive and even small delays in identification and treatment can cause permanent damage to or the death of an infant. More information on national newborn screening timeliness recommendations is available at: <https://www.hrsa.gov/advisory-committees/heritable-disorders/newborn-screening-timeliness.html>.

It is highly recommended all practices caring for newborns are prepared to collect NBS heel-stick specimens. Sending the family elsewhere for specimen collection or ordering bloodwork as opposed to collecting a repeat specimen has repeatedly led to delays in screening and follow-up.



Frequently Asked Questions (FAQs)

Q: Can I send a newborn to the laboratory for a repeat NBS rather than collect the specimen in the office?

A: It is highly recommended that the PCP office collect the heel-stick specimen and submit it to the state laboratory directly, rather than sending the infant to a laboratory for specimen collection.

- Most commercial laboratories in Connecticut **will not** collect a heel-stick specimen. If your office has an arrangement with a commercial laboratory to collect heel-stick specimens, it is your office's responsibility to assure that the specimen is collected within two business days of receiving the request for the specimen from the CT NBS Program and that the specimen is shipped to the State Laboratory within 24 hours of collection

Q: Can I send the newborn back to the Birth Hospital for heel-stick specimen collection?

A: In most cases, no. The majority of CT Birth Hospitals will not collect heel-stick specimens from an infant that has been discharged from their care.

- If you contact the birth hospital about collecting a heel-stick specimen, speak with the administrator who oversees newborn screening for the birthing center. Do not speak with the hospital laboratory staff; in most cases the hospital laboratory is not able to collect a heel-stick specimen. If the birth hospital agrees to collect a heel-stick specimen, it remains your office's responsibility to assure that the specimen is collected within two business days of receiving the request for the specimen from the CT NBS Program and that the specimen is shipped to the State Laboratory within 24 hours of collection

Q: Can the CT NBS Program tell me which laboratories or birth hospitals are able to collect a heel-stick specimen?

A: No. There are dozens of birth hospitals and hundreds of lab collection sites in CT. The CT NBS Program does not maintain this information.

Q: Can I order bloodwork instead of collecting a repeat heel-stick specimen?

A: Collection of a heel-stick specimen is strongly recommended over ordering venous blood work for the following reasons:

- The CT NBS Program tests over 70 analytes using age related cut-offs and specialized algorithms and tools. Most other laboratories do not have age related cut-offs for newborns which can affect the interpretation of test results
- Collection of venous blood is more invasive than heel-stick specimen collection and requires a specialized skillset. Most phlebotomists are not skilled at collecting blood from a newborn. This can create significant stress for the infant and family. Also, travel to a location other than the PCP office can be a barrier for a family and lead to delays in specimen collection.
- The PCP office has more control over the timing of collection and submission of a heel-stick specimen to the state laboratory vs. timing of collection of venous blood collection. When a specimen arrives at the state laboratory for repeat screening, the screening process is expedited. Preliminary results are generally available within one business day of receipt of the specimen at the state laboratory.
- Repeat screening conducted through the state laboratory allows for the most accurate comparison between the initial and repeat test results. Different laboratories may use different testing platforms, measure different analytes and use different units of measurement which can affect the interpretation of results (continued on next page)

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- There is no cost to the parent or PCP office for screening conducted by the Connecticut Newborn Screening Program when a heel-stick specimen is collected and submitted to the state laboratory by the PCP office. Insurance may not cover newborn screening or other tests ordered through a commercial laboratory, when ordered in lieu of recommended repeat bloodspot screening conducted by the state laboratory.
- The term “repeat NBS” means different things to different people. CT screens for 64 different disorders by analyzing over 70 different analytes. This can be confusing for office and laboratory staff not experienced with ordering lab work following an out of range newborn screening result and can result in errors that lead to delays repeat screening.



Example 1: The CT NBS Program requested that a practice collect a heel-stick sample following an out of range Adrenoleukodystrophy (X-ALD) newborn screening panel. It is not uncommon for healthcare workers to refer to newborn screening as the “PKU test”. In this case the practice ordered a “repeat PKU test¹”. Upon receipt of the “PKU” order, the laboratory ran a serum phenylalanine test which is of no use in X-ALD screening.

Example 2: The CT NBS Program requested that a practice collect a heel stick specimen following an out of range Amino Acid Panel on the initial newborn screen. PCP office staff ordered a Thyroid Stimulating Hormone level (TSH) level after asking a commercial laboratory what to order for a “repeat NBS”. Repeat Congenital Hypothyroidism screening (TSH level) is a fairly common request. In this case the laboratory staff incorrectly equated the request for a repeat NBS as a need for a serum TSH level.

Q: If I choose to order bloodwork can the CT NBS Program advise me what to order?

A. No, if you choose to order bloodwork over collecting a heel-stick specimen, you will need to consult with the specialist in the area of genetics, endocrinology, hematology, immunology or neurology, depending on the reason for the request for repeat NBS. Please send the results of bloodwork ordered in lieu of a repeat NBS to the CT NBS Program Follow-up Unit at the State Laboratory at 860.730.8385 (fax) or dph.nbstracking@ct.gov.

Q: Where do I obtain specimen collection supplies and instructions?

A. Contact the DPH Outfit Room at the state laboratory (860.920.6674 or dph.outfitroom@ct.gov).

Q: Who do I call if I have a question about specimen collection or need results of repeat screening?

A. Contact the CT Newborn Screening Program Follow-up Unit at the State Laboratory (860.920.6674 or dph.nbstracking@ct.gov).

¹ See [Newborn Screening: More Than a PKU Screen](#) attached