



Connecticut Newborn Screening Timing of Blood-Spot Collection Guidelines



CONNECTICUT NEWBORN SCREENING PROGRAM • April 2019

Timeliness in Newborn Screening

The Connecticut Newborn Screening Program at the State Laboratory screens for over 60 disorders through bloodspot screening. The timely collection of the Newborn Screening (NBS) bloodspot specimen is crucial. Failure to submit the NBS specimen in a timely fashion can lead to delays in the identification and treatment of a disorder. Bloodspot specimens collected after 48 hours of age may not allow enough time for results to be interpreted and reported before serious symptoms and in some cases, death, occur.

Some of the most recent recommendations (March 2015) from the US Department of Health and Human Services (USDHHS) [Advisory Committee for Heritable Disorders in Newborns and Children](#) (ACHDNC) for timeliness in newborn screening¹ are as follows

1. Presumptive positive results for time-critical conditions detected by newborn screening should immediately be reported to the child's healthcare provider and no later than 5 days of life.
2. All presumptive positive results for time sensitive conditions should be reported to the healthcare provider as soon as possible but no later than 7 days of life.
3. In order to achieve these goals (and reduce delays in newborn screening):
 - Initial NBS specimens should be collected in the appropriate time frame for the baby's condition but **no later than 48 hours after birth**.
 - NBS specimens **should be received at the Laboratory as soon as possible; ideally within 24 hours of collection**.

Timing of blood spot collection is also important for the accurate interpretation of results. Specimens collected prior to 24 hours of age¹ are invalid for congenital hypothyroidism (CH) and Congenital Adrenal Hyperplasia (CAH) screening and amino acid (AA) screening results are less reliable.

Specimens collected too soon following a blood transfusion invalidate Biotinidase Deficiency (Bio), Galactosemia (Galt), Severe Combined Immunodeficiency (SCID) and Hemoglobinopathy (Hgb) screenings.

In addition, specimens collected too early or too late may increase the chance of false positive or false negative results.

Please see the next page for the specifics of CT NBS Program Blood-Spot Collection Guidelines.

¹ <https://www.newsteps.org/news-and-education/news/update-suggested-recommendations-timeliness-newborn-screening>

Connecticut Newborn Screening Bloodspot Collection Guidelines

Timing of Bloodspot Specimen Collection

Previous CT NBS Program specimen collection guidelines recommended that specimen collection take place between 24 and 48 hours of life, preferably as soon as possible after 24 hours of life. However, there is evidence that demonstrates when the first NBS specimen is collected at greater than 28 hours of life the number of false positive Congenital Hypothyroidism screens decreases.

Effective April 1, 2019, the CT NBS Program recommends that the first specimen be collected within 24 to 48 hours after birth, preferably as close as possible to 30 hours of life. Parenteral nutrition should be held for at least 4 hours prior to specimen collection, unless medically contraindicated.

Special circumstances

- **Specimen collection at less than 24 hours of age**

There are some circumstances where it is appropriate to collect a bloodspot specimen prior to 24 hours of life. These are:

- Prior to the transfusion of blood products
- When death is imminent
- Prior to early discharge
- Prior to transfer to another facility

When a specimen is collected at less than 24 hours of age, a second specimen must be collected at greater than 24 of age for endocrine screening and repeat amino acid and acylcarnitine screening.

- **Transfusion**

Every effort should be made to collect a NBS specimen prior to transfusion of blood products, however if the first specimen is collected after a transfusion:

- a second NBS specimen must be collected at greater than 48 hours post transfusion for Severe Combined Immunodeficiency (SCID) screening
- a third NBS specimen must be collected at 120-days post transfusion for Biotinidase Deficiency, Galactosemia and Hemoglobinopathy screenings.

- **The medically fragile newborn**

If the infant's condition precludes collection of the first newborn screening specimen before 48 hours of life, this must be documented before 48 hours of life in the Maven laboratory package by selecting "baby medically fragile, specimen collection pending" and entering the date of the entry was made.

- **The dying infant:**

If an infant is likely to die, it is appropriate to collect a newborn screening specimen prior to death, or if not possible to collect prior to death, as soon as possible after death.

- While dying infants may have abnormal results as a response to organ failure, the specimen may also provide a diagnosis of an early onset disorder.
- If it is not possible to obtain a NBS specimen prior to or immediately following death, or the parent of a newborn expected to die declines screening, this must be documented in the notes section of Maven and in the Maven laboratory package by selecting “baby medically fragile, unable to collect specimen prior to death”.

- **Parent refusal:**

[Connecticut General Statutes, Section 19a-55](#) allows a parent or guardian of an infant to refuse permission for newborn screening blood-spot testing, because such a test is in conflict with the parent’s or guardian’s religious beliefs and practice.

- It is essential that the parent understand the consequences of refusing newborn screening for their infant. Have the parent review the information found at one or more of these sites
 - <https://portal.ct.gov/DPH/Laboratory/Newborn-Screening/Newborn-Screening-Program>
 - <http://www.savebabies.org/index.html>
 - <https://kidshealth.org/en/parents/newborn-screening-tests.html?ref=search>
- If after learning more about newborn screening, the parent still wants to decline screening, complete the [CT NBS Program Waiver form](#) and fax to the CT NBS Program at 860-730-8385.

[Please also see the Connecticut Newborn Screening Blood-Spot Specimen Collection Guidelines](#)