

WHEN TO COLLECT A BLOOD SPECIMEN

Full-term Infant/Hospital Birth setting

A filter paper specimen must be collected from each newborn infant as close as possible to the time of discharge and no later than 7 days of age by the institution or hospital where initial or subsequent newborn care was provided, regardless of prematurity, illness, feeding history or antibiotics given. The preferred time of collection is 24-48 hours of age.

Specimen Collected Early (< 24 hours)

If the initial specimen is collected before 24 hours of age, a second specimen must be collected within 2 weeks of age.

Premature/Sick Infants

A specimen should be collected as close as possible to discharge and no later than 7 days of life, unless a transfusion is imminent. The appropriate strategy is to ***always*** collect a newborn screening sample immediately **before any transfusions, regardless of the infant's age.**

Transfused Infant

Red blood cell (RBC) transfusions interfere with the interpretation of some newborn screening results. The appropriate strategy is to ***always*** collect a newborn screening sample immediately **before any transfusions, regardless of the infant's age.** Since red blood cells and plasma transfusions can cause false negative results, post-transfusion follow-up at the appropriate time is essential.

- Whenever possible, the newborn screen specimen should be collected prior to a transfusion of blood products, even if less than 24 hours of age.
- If the infant was transfused at the time of collection, a follow-up filter paper specimen must be collected at least 8 weeks after the last transfusion.

IMPORTANT: Always indicate on the specimen collection card if the infant has been transfused and document the date of the most recent transfusion.

Total Parenteral Nutrition (TPN Therapy - Hyperalimentation)

Infants on some types of total parenteral nutrition (TPN) may show elevated levels of amino acids (e.g. phenylalanine). Indications of TPN status on the collection form is necessary for clarifying some test results. The infant does not have to be off TPN before collecting the specimen.

The newborn screening test, like any laboratory test, may have false positives and false negatives. If signs and symptoms of one of the disorders are clinically evident, the physician should proceed to diagnostic testing, pending the results of the screening test or in spite of the results of the screening test.

Clinical Signs or Family History

There are a number of clinical situations that will modify the usual approach of obtaining a newborn screening specimen and waiting for the result. The following are brief suggested guidelines for particular situations that may arise in clinical practice.

Regardless of any diagnostic or therapeutic interventions, a newborn screen should be obtained on all infants to test for the other conditions included in the panel. When in doubt about the course of management for any of the conditions on the screening test, consultation with a specialist is advised.

- **If the results of the newborn screen are pending:**
For any of the screened conditions, but especially those in which the metabolite accumulation is dangerous, such as galactosemia, **treat as if the infant has the condition.** For other conditions, contact a metabolic center or metabolic physician for assistance with rapid diagnosis and institution of dietary treatment; for galactosemia, begin a soy-based formula until the screening results are known.
- **If the newborn screening test result was "normal":**
If clinical symptoms suggest one of the screened conditions despite a "normal" screening test, the physician should **proceed as if the patient has the condition** and immediately contact a consultant specialist for instructions on further evaluation of the patient.

Newborn Screening of Infants with an Affected Close Relative

As many of the conditions tested for by newborn screening are genetic, it is possible that multiple members of a family may be affected. Prenatal diagnosis is possible for many of these conditions. If prenatal diagnosis determines that the infant is affected, any appropriate treatment (e.g., special diet) should be initiated immediately after birth.

If prenatal diagnosis predicts an unaffected baby, practitioners should bear in mind that no prenatal diagnostic test is 100% accurate. Neonates who are siblings or close relatives of an affected individual are not part of the "general population" for whom newborn screening is designed. For any infant with a positive family history, providers should contact appropriate consultant specialists, ideally prenatally or immediately at birth, to determine the proper diagnostic tests and proper timing of those tests.

Transferred Infants

When possible, the originating hospital should draw a newborn screening specimen **before** transferring the infant to another hospital. Transfer information should be included in the Certifier's Worksheet for Birth Certificate filing. The information will assist with follow-up if the newborn screening is not done prior to transfer.

Infants born in South Dakota must have testing performed by the designated laboratory. If an infant is transferred to another hospital before 48 hours of age, the receiving hospital must collect a specimen at an appropriate time within the first 48 hours of life. If an infant is transferred to another hospital after 48 hours of age, the transferring hospital must collect a specimen before the transfer and within the first 48 hours of age. When an infant born in South Dakota is transferred out of state, **the specimen needs to be submitted to the SD designated laboratory for testing.**

Home/Out-of Hospital Births

The parents, guardian, or custodian of each infant are responsible for having blood tests for metabolic disorders performed.

If a birth attendant was not present for the birth, the local registrar shall inform the parent or guardian of the need for a blood test for metabolic disorders when inquiring about or filing a certificate of birth.