

IMPORTANT NEONATAL METABOLIC SCREENING CONSIDERATIONS

Newborn metabolic screening tests are not diagnostic. Due to biologic variability, some affected infants may have normal neonatal metabolic screening results. The possibility of a false negative or a false positive result must always be considered, especially when screening newborns for metabolic disorders. A newborn metabolic screen may detect specific mutations only (e.g., CAH due to 21-hydroxylase or 11-beta-hydroxylase deficiency).

Collection Form Information:

IT IS IMPERATIVE TO PROVIDE COMPLETE INFORMATION ON THE COLLECTION FORM. The lab needs the information to correctly assign test result interpretations. Complete information also makes it possible to positively identify the infant in the event of similar names and to contact the submitting facility or attending healthcare practitioner if necessary.

Collection Times:

Early collections compromise some of the test results. Recommended time for screening is between 24 hours and 5 days of age. If an infant is less than 24 hours old at the time of collection, ANOTHER SPECIMEN SHOULD BE COLLECTED AND SENT FOR TESTING AS SOON AS POSSIBLE. However, a newborn metabolic screen should always be collected before an infant leaves the hospital and before transfusion, even if the infant is less than 24 hours of age.

Specimen Collection:

Blood should be applied DIRECTLY from the infant's heel onto the filter paper. Blood collections into containers with preservatives (e.g., EDTA) may cause false positive and/or false negative test results, depending on the testing technology. Blood should never be applied to both sides of the filter paper or "drawn" on with a capillary tube.

Premature or Ill Infants:

Infants in the neonatal intensive care unit have so many critical needs that their newborn metabolic screening may be overlooked. All infants transferred to another medical facility must be screened by the receiving facility unless the infant has already been screened. It is also advisable to draw the neonatal metabolic screen on an infant before transfusion.

Transferred Infants:

Hospitals transferring an infant to another facility are responsible for notifying the receiving facility of the status of the neonatal metabolic screen.

Transfusions:

Red blood cell (RBC) transfusions interfere with the interpretation of some newborn metabolic screening results. Whenever possible, the newborn screen should be collected prior to a RBC transfusion, even if less than 24 hours of age. IF an 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.

Treatment:

Under no circumstances should treatment be initiated without consultation with program medical consultants. Treatment prior to diagnostic confirmation may interfere with confirmatory testing. Moreover, it may cause irreversible harm to the infant.

Total Parenteral Nutrition (TPN):

Infants on some types of TPN may show elevated levels of amino acids (e.g., phenylalanine). Indication of TPN status on the collection form is necessary for clarifying some test results.

Hormone (Steroid) Therapy:

Steroids administered to the mother during pregnancy, or to the infant immediately after birth, can interfere with congenital adrenal hyperplasia test results. Contact the endocrine consultants regarding management for these situations, (319) 356-2838.

Thyroid Medications:

When thyroid medications are administered to the mother during pregnancy, Congenital Hypothyroidism screening results are not reliable. Contact the Pediatric Endocrinology Consultants regarding management for these situations, (319) 356-2838.