

Information on Factors that Can Cause a False Positive or False Negative Newborn Screen Result



MARITIME
Newborn Screening
Dépistage néonatal
DES MARITIMES

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There are factors that can affect the results of newborn screening. Many of these variables cannot be avoided. Some of the variables can be addressed (i.e., proper collection of the blood spot sample) and help to minimize the chance of a false positive or false negative result. If you have questions about any of the information below, please contact Maritime Newborn Screening (MNBS) by phone at 902-470-2783 or email us at MNBSinfo@iwk.nshealth.ca.

Age at Time of Collection for Full Term Infant

Recommended age for collection of a newborn screening sample is **24-48 hours of life**. Infants should be more than 24 hours of age to ensure they have ingested enough nutrition to show accurate levels of amino acids in the blood. If collected before 24 hours, amino acid levels could be falsely low. If collected after 72 hours the infant may no longer be in a catabolic state, which could cause a false negative result for other metabolic conditions on the newborn screen.

Unsatisfactory Blood Spot Samples

Poor quality samples can cause false positive or false negative results. Examples of poor-quality samples are as follows:

- Quantity of blood insufficient
- Supersaturated blood spots
- Clotted or layered blood spot
- Scratched filter paper
- Wet/discoloured
- Serum rings contain too much tissue fluid in the specimen
- Contamination of blood spot with oils from hands touching it, spilling of substances on the sample, or piling samples on one another
- Use of anticoagulant in collection tube

Prematurity

If the infant is born prematurely their liver and immune systems are less mature and this can affect some of the results for the newborn screen. Samples are collected between 24-48 hours and then again at time of discharge. *A sample should still be collected at 24-48 hours as some of the screening is not affected by prematurity.*

Transfusions

Specifically, packed red blood cell transfusions (PRBC). If a PRBC transfusion is administered prior to a sample draw, it can affect hemoglobinopathy, biotinidase, and galactosemia results. *If at all possible, the newborn screen should be taken before a PRBC transfusion is done, even if this is before the ideal collection time of 24-48 hours of life.*

Conditions of the Newborn

The wellbeing of a newborn can affect the newborn screen result. An infant who is unwell is more likely to have a false positive result. In some cases, a condition may cause a false negative result (see below for action required). Conditions of the newborn that can affect the newborn screen result are as follows:

- Liver immaturity or disease
- Meconium ileus (MI) → ****contact MNBS (phone 902-470-2783) as soon as possible if a newborn has this condition, as MI affects the screening process for cystic fibrosis****
- Hypoxia
- Hyperbilirubinemia
- Acute illness

Total Parental Nutrition (TPN)

TPN can alter the amino acid levels in the blood of an infant and could cause a false positive result for the metabolic portion of the screen.

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Nothing by Mouth (NPO)

If a baby has not eaten anything, this can cause a false negative result. This can happen because the baby has not yet ingested anything with protein or fat, so it is not possible to determine how they process it, which is part of the screen.

Infant Medications

- Antibiotics
- Dopamine
- Steroids
- Iodine
- MCT (medium chain triglycerides) oil
- Dextrose
- Carnitine Supplementation

Expired Blood Collection Cards

The date on each blood spot card needs to be checked prior to collection. An expired collection card could cause false positive or false negative results.

Family History

Unexplained infant death and other family history may indicate an underlying genetic condition in the family. Contact MNBS (902-470-2783) if there is a family history of one of the conditions on the newborn screening panel.

Twin/Multiple Pregnancy

Results can be affected if the pregnancy has more than one baby that shares the same placenta/blood supply. Pregnancies with twin-to-twin transfusion syndrome can lead to false positive or false negative results. All same sex twins/multiples are retested at 2 weeks of age to ensure that the results are accurate for each baby.

Cord Blood Sample

Cord blood can be contaminated by blood from the gestational carrier so it is not suitable for newborn screening, as it can cause a false positive or false negative result.

Medications in Pregnancy

If the gestational carrier is taking one of the following medications, it may affect the results of newborn screening:

- Trikafta® (Elexacaftor/tezacaftor/ivacaftor) → ****contact MNBS (phone 902-470-2783) as Trikafta taken during pregnancy affects the screening process for cystic fibrosis****
- Steroids
- Iodine Supplementation

Pregnancy Conditions

If the gestational carrier has one of the following conditions, it may affect the results of newborn screening:

- Thyroid disease
- Congenital adrenal hyperplasia (CAH)
- Phenylketonuria (PKU) or hyperphenylalaninemia
- Vitamin B12 deficiency
- Carnitine deficiency
- Glutaric acidemia Type 1 (GA1)
- 3-MCC deficiency



IWK Health

Maritime Newborn Screening

IWK Health, Halifax, N.S.

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