MARITIME
Newborn Screening
PROGRAM

PROGRAMME DE
dépistage néonatal
DES MARITIMES

Newborn Screening Manual

A Guide for Health Care Providers

Edition 1.0 | OCTOBER 2022

Newborn Screening Manual:

A guide for Health Care Providers

This manual was created by the Maritime Newborn Screening Program (MNSP) to provide health care providers (HCPs), including submitting facilities, with a comprehensive guide to Newborn Screening. The goals are to ensure that all infants born in the Maritimes receive high quality newborn screening.

This manual reviews recommended practices for newborn screening. It can be found, free of charge online at www.maritimenewbornscreening.ca. This manual is intended to be used by hospitals and health care providers that submit, or receive results from, the Maritime Newborn Screening Program. Revised or additional pages may be updated periodically.

If you have any questions about the information contained in this manual, please contact MNBSinfo@iwk.nshealth.ca.

Table of Contents

SECTION ONE: DEFINITIONS

Words that are defined will be in blue text throughout the document for referral back to this section for its definition/explanation.

The Maritimes
Nova Scotia, New Brunswick and Prince Edward Island

MNSP
This acronym stands for Maritime Newborn Screening Program.

MNSP Advisory Committee
The MNSP Advisory Committee is an advisory body of health and other professionals from each province with expertise in newborn and childhood screening. The mandate of the Advisory Committee is to provide the MNSP with advice regarding the policies and practices related to newborn screening.

MNSP Clinical Working Groups
The group of specialists involved with the various categories of disorders screened, that meet to discuss positive case results and efficiency of the clinical follow-up process.

Maritime Newborn Screening Program Newborn Screening card:
The MNSP newborn screening card consists of four main parts:

1) The filter paper at the top where the blood will be applied
2) The parent information sheet at the front
3) The requisition with required fields (and 2 carbon copies)
4) The decline/defer form

Screen Negative Results
These results mean that the infant has a low chance of having the disease. A negative result, however, does not completely rule out a disease; an infant showing symptoms of the disease should have prompt diagnostic evaluation.

Screen Follow-Up Results
These results mean that another sample is required because either i) the initial sample was unsatisfactory, ii) the result of a specific screen was unclear.

Screen Positive Results
These results mean that there is an increased chance for the disease. A screen positive result, however, does NOT mean a baby has a disease; the MNSP will refer the infant to specialists for diagnostic testing to confirm or rule out the disease.

Analytes
The newborn blood biomarkers that are used to screen for specific conditions.

Newborn Screening Sample
A newborn screening blood spot collection card that is collected between one day (24 hours) and two days (48 hours) after birth.

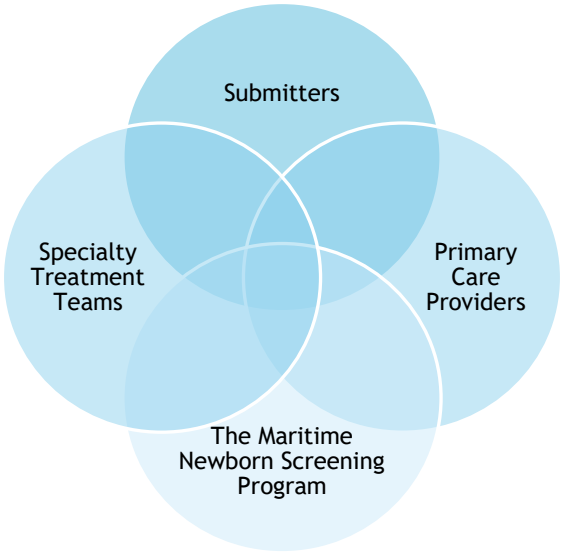
SECTION TWO: BACKGROUND INFORMATION

2.1 Introduction

The primary goal of newborn screening is early identification of infants with serious disorders so that treatment can be initiated before damaging health concerns arise. While all of the diseases screened for are relatively rare and not usually apparent at birth, in 2016, 84 infants in the Maritimes were found to have one of these conditions (1 in 359 infants screened). Timely newborn screening, early diagnosis, and treatment can help these children have the best start in life. The cost of missing one of these conditions can be significant to the life of that newborn. Untreated infants can develop mental disability, serious health problems, or even die, sometimes without a diagnosis being made.

Prior to the creation of the specific Maritime Newborn Screening Program in April 2014, newborn screening was performed at several independent sites throughout the Maritimes. Nova Scotia began testing for new analytes and at that time New Brunswick and Prince Edward Island contracted the IWK Health Centre to provide testing and follow up services for them. All testing is currently located in the IWK Children's Health Centre in Halifax, NS.

Newborn Screening requires coordinated interaction between 4 main groups of Health Care Providers.



The main responsibilities of each group are as follows:

1) Submitters
   This group, which includes hospitals, birthing centers, and midwifery practices, is responsible for providing information to parents, for providing the MNSP with birth records, for recording accurate and complete information on the collection card, for collecting a satisfactory blood spot, for timely and efficient transport of samples to the IWK testing facility, and for prompt follow-up in the event that an initial screen was missed, a sample was unsatisfactory, or a follow-up sample is required for analysis.

2) Primary Care Providers
   This group includes health care providers, physicians, nurse practitioners and midwives who are involved in the care of the baby, both prenatally and postnatally. Clinicians

are responsible to educate their patients about newborn screening and to ensure an order is placed. For patient information materials please visit www.maritimenewbornscreening.ca. Clinicians involved in postnatal care are responsible to check that a newborn screen sample was taken, to assist in coordinating any follow-up samples that are needed, and to educate parents about the results of the newborn screening.

3) The Maritime Newborn Screening Program
This group includes the staff from the program's administration and laboratory. The Maritime Newborn Screening Program is responsible for efficient and accurate screening of all samples, for distributing materials and educational resources needed for newborn screening, for keeping accurate records, for communication with clinicians and collecting facilities regarding missing, unsatisfactory or follow-up samples, for coordinating and tracking clinical involvement required for infants with a positive newborn screen, and for educating parents, health care providers, and the public about newborn screening.

4) Specialty Treatment Teams
This group includes any clinic or facility to which infants with positive newborn screening results are referred. They are responsible for ensuring swift confirmatory testing for infants with positive newborn screening results, for management of confirmed cases, for providing the MNSP with final diagnosis/follow-up information, and for providing education to primary health care clinicians.

2.2 MNSP History

Date	Newborn Screening Events
1972	The IWK Health Centre started screening for Phenylketonuria (PKU)
1977	Congenital Hypothyroidism (CH)
October 2000	Medium-Chain Acyl-CoA Dehydrogenase Deficiency (MCAD)
September 2004	Very Long-Chain Acyl-CoA Dehydrogenase Deficiency (VLCAD) and Glutaric Acidemia Type I (GAI)
April 2005	Carnitine Palmitoyl Transferase I Deficiency (CPTI), Carnitine Palmitoyl Transferase II Deficiency (CPTII), Carnitine Uptake Disorder (CUD), and Carnitine-Acylcarnitine Translocase Deficiency (CACT)
2005	Long-Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency (LCHAD), Trifunctional Protein Deficiency (TFP), Isovaleric Acidemia (IVA), and Maple Syrup Urine Disease (MSUD)
April 2014	Screening for Cystic Fibrosis (CF) and hemoglobinopathies begins
October 2014	New Brunswick contracts with the Nova Scotia newborn screening program to provide services. Name changes to the Maritime Newborn Screening Program
November 2015	Prince Edward Island contracts services from the Maritime Newborn Screening Program
March 2016	Severe Combined Immunodeficiency (SCID)
February 2018	Argininosuccinic Acidemia (ASA), Citrullinemia Type I, Methylmalonic Acidemia Mutase Deficiency (MMA/MUT), Methylmalonic Acidemia Cobalamin A or B Defect (MMA/CblA or MMA/CblB), and Propionic Acidemia (PA)
May 2019	Biotinidase Deficiency (BIOT) and Classic Galactosemia (GALT)

2.3 MNSP Contact Information

The MNSP can be contacted in several ways:

Phone	Local: 902-470-7998 Toll Free: 902-470-5888 extension 7998
Fax	902-470-6974
Email	MNBSinfo@iwk.nshealth.ca
Mail	Maritime Newborn Screening Program IWK Health Centre 5850/5980 University Avenue, PO Box 9700 Halifax, Nova Scotia, Canada B3K 6R8
Website	www.maritimenewbornscreening.ca

Office hours are Monday to Friday (excluding holidays), 8:00am – 16:00pm

2.4 Newborn Screening Basics

1) **Please use the term "Newborn Screening" not "PKU test".**
The term "PKU test" can be confusing to parents and other healthcare providers. PKU (Phenylketonuria) is only one of the diseases screened for in the newborn screening. For a complete list of the diseases screened, please visit our website www.maritimenewbornscreening.ca.

2) **Every infant is at risk.**
Newborn screening detects very rare diseases that are not apparent at birth. Most infants will not have a family history of the disease; therefore ***every*** infant is at risk.

3) **Screen every infant PRIOR to discharge from the hospital.**
Infants discharged prior to 24 hours of age should have a newborn screen taken prior to discharge, even though this is outside of the recommended range of 24-48 hours. This is due to the fact that there is a higher rate of missed newborn screens if the babies are discharged prior to 24 hours of age, and some (but not all) disorders can be picked up before 24 hours of age. Inform parents of the need for a repeat screen during the recommended time of 24-48 hours after birth. If parents refuse to have this early sample collected, please have them complete the Defer/Decline Form located at the back of the newborn screening card.

4) **The goal of newborn screening is to detect and treat disease EARLY.**
If left undiagnosed, the diseases screened for will likely cause intellectual disability, serious health concerns, or even death. Early detection and treatment of affected infants can greatly improve the outcome, and sometimes even save their life.

5) **Newborn Screening is strongly recommended for all infants, but is not mandatory.**
Ensure that newborn screening has been thoroughly explained to the parents. If the parents do not consent to testing, it is extremely important to document this by having them sign the Decline Form located at the back of the newborns screening card.

6) **Unsatisfactory samples require a repeat sample taken immediately.**
Delays in obtaining a repeat sample can lead to delays in diagnosis and serious health concerns for affected infants.

7) **The newborn screening cards MUST be filled out accurately and completely.** The information asked for on the newborn screening card is essential for accurate interpretation of the results. Incorrect or missing information can lead to false positive and false negative results as well as delays in contacting the family/healthcare provider in the event of a screen positive result or a result indicating follow up testing is required.

2.5 Conditions Screened for through the MNSP

The disorders screened for by the Maritime Newborn Screening Program (MNSP) are based on advice from the MNSP Advisory Committee comprised of healthcare providers from the three Maritime Provinces and from a variety of specialty areas. The advisory committee reviews disorders and provides advice about testing that would be most beneficial to the population it serves.

METABOLIC SCREEN

Maple Syrup Urine Disease (MSUD)
Phenylketonuria (PKU)
Glutaric Acidemia Type I (GA1)
Isovaleric Acidemia (IVA)
Carnitine Palmitoyl Transferase I Deficiency (CPTI)
Carnitine Palmitoyl Transferase II Deficiency (CPTII)
Carnitine Uptake Disorder (CUD)
Carnitine-Acylcarnitine Translocase Deficiency (CACT)
Long-Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency (LCHAD)
Medium-Chain Acyl-CoA Dehydrogenase Deficiency (MCAD)
Very Long-Chain Acyl-CoA Dehydrogenase Deficiency (VLCAD)
Trifunctional Protein Deficiency (TFP, a variety of LCHAD)
Argininosuccinic Acidemia (ASA)
Methylmalonic Acidemia Mutase Deficiency (MMA/MUT)
Methylmalonic Acidemia Cobalamin A or B Defects (MMA/CblA or MMA/CblB)
Citrullinemia Type I
Propionic Acidemia (PA)
Biotinidase Deficiency (BIOT)
Classic Galactosemia (GALT)

OTHER DISORDERS

Congenital Hypothyroidism (CH)
Cystic Fibrosis (CF)
Hemoglobinopathies (Sickle Cell Disease, Hemoglobin Variants, Hemoglobin H Disease)
Severe Combined Immunodeficiency (SCID)

A more detailed review of these disorders, including primary marker, incidence, symptoms, and treatment can be found in section 5.1.

2.6 Maritime Newborn Screening Program Reports

Each infant will receive an OVERALL newborn screening result.

All Newborn screening reports will be sent to:

- The ordering healthcare provider indicated on the card
- The family physician indicated on the card
- The birth hospital indicated on the card

Potential newborn screening results are:

Screen Negative Results

- These results mean that the infant has a low chance of having the disease. A screen negative result, however, does not completely rule out a disease; an infant showing symptoms of the disease should have prompt diagnostic evaluation.

Screen Follow-Up Results

- These results mean that another sample is required because either i) the initial sample was unsatisfactory, ii) the result of a specific screen was unclear.

Screen Positive Results

- These results mean that there is an increased chance for the disease. A screen positive result, however, does NOT mean a baby has a disease; the MNSP will refer the infant to specialists for diagnostic testing to confirm or rule out the disease.

Please see the appendix for examples of reports with these results.

2.7 Newborn Screening Limitations

Newborn Screening Limitations

As with any type of screening, false negatives and false positives can occur in newborn screening. False positives may increase parental anxiety and expose the infant to unnecessary testing.

False negative results, although rare, may result in potential harmful outcomes for the infant. If a newborn screening result is negative, it does not mean with certainty that the infant does not have any of the diseases on the panel. **If an infant or child in your care shows symptoms of a particular disease, diagnostic investigation should be initiated regardless of the newborn screening result.** The relevant specialist should be contacted for further advice. False negative results may provide a misleading sense of reassurance to Primary Care Providers, which is why clinical symptoms must always be evaluated independently of newborn screening results.

SECTION THREE: SAMPLE COLLECTION

3.1 Responsibility for Sample Collection

It is the responsibility of all submitting facilities (birth hospitals, midwifery practices, or birthing centres) and all prenatal and postnatal primary health care providers to ensure that all infants born in the Maritimes are offered newborn screening.

For infants born in hospitals, ensuring that the screen has been offered should be part of the pre-discharge checklist.

For infants born at home/under the care of midwife, ensuring that the screen has been offered should be part of the first or second postpartum visit.

*It is extremely important to collect quality newborn screening samples the **first** time.* Unsatisfactory newborn screen specimens can cause unreliable, misleading, and clinically inaccurate levels for one or more of the analytes used for newborn screening. Unsatisfactory newborn screen specimens place burden on the whole newborn screening system. They can cause unnecessary trauma to the infant and anxiety for the parents, they cause submitting facilities to spend time to re-collect the sample, and they can delay the identification and treatment of an affected infant. The turnaround time for results and treatment is imperative in preventing irreversible organ damage or death for affected infants.

3.2 Informed Consent for Parents/Guardians

The MNSP strives to make sure that education is available for parents to make informed decisions for their infant. It is important that parents know that newborn screening could save their infant's life and/or prevent serious health problems. Brochures on newborn screening are available on the MNSP website: www.maritimenewbornscreening.ca and hardcopies can also be ordered through the MNSP. See Section 8 for instructions.

The MNSP newborn screening card includes a sheet full of information about newborn screening. This information sheet should be given to parents before the newborn screening sample is drawn. A copy of this information sheet is provided in the Appendix. For the most up to date version, please refer to our website at www.maritimenewbornscreening.ca.

3.3 Parental/Guardian Decline Procedures

Newborn screening is considered standard of care. The vast majority of parents wish to have their child screened. However, it is currently not mandated by law and parents can decline the screening if they wish.

The decision to decline newborn screening must be discussed with a health care provider to ensure parents are making an informed decision. The health care provider should document the discussion and the decision that they have refused this screening for their infant in the infant's medical record and have the parents sign the Newborn Screening Decline form.

For your convenience, the MNSP has provided a Newborn Screening Decline or Defer Form. It is included as the last page of the MNSP newborn screening card. It documents the information that the parents should be given and allows them to sign to say that they understand and are choosing to decline the screening. A health care provider should also sign the form.

IMPORTANT: Should parents decline newborn screening, please still fill out the demographics portion of the newborn screen and send this, along with the signed decline form, to the MNSP. This will prevent us from contacting the health care provider or patient when we do not receive a newborn screen for the patient (as explained in more detail in section 4.2, we have tracking systems in place to check for newborn screens on all babies born within health care facilities in the Maritimes).

3.4 Parental/Guardian Deferral Procedures

As will be discussed later in this section, the ideal time for a newborn screening sample to be collected is 24-48 hours of age. If a healthy infant is discharged prior to 24 hours of age, it is advisable to still take a newborn screen before discharge, AND then another in the recommended time frame of 24-48 hours of age. This is because some of the disorders can be picked up before 24 hours of age and if the child is indeed affected, any delay in diagnosis could prove harmful.

However, the parent may choose to defer the screening for a later date. To do this, the parents and a healthcare provider (can be a nurse) must sign the Newborn Screening Decline or Defer Form (included as the last page of the MNSP newborn screening card), which states that they understand the potential risks of delayed screening. This signed form must be mailed to the MNSP program.

It is up to the collecting facility to ensure the parents return for a newborn screen.

3.5 Accurate Documentation on the MNSP Newborn Screening Card

The MNSP newborn screening card consists of four main parts: 1) the filter paper at the top for specimen collection, 2) the parent information sheet at the front, 3) the requisition with required fields (and 2 carbon copies), 4) and the decline/defer form.

There are many, many things that can affect the results and interpretation of a newborn screen. It is vital to the accuracy of the newborn screening that all fields on the requisition are filled out accurately and completely. Accurate analysis of the analytes and a complete report cannot be issued if certain critical fields are not completed (see the section 3.10 for more information on the rejection policy).

The MNSP newborn screening card requisition should be filled out with a black or blue ball point pen (no gel pens). If a hospital sticker or stamp is used, all copies of the screening card must be stickered/stamped.

Please ensure that stickers are printed properly and that information asked for is complete and legible.

The MNSP newborn screening card requisition must be filled out PRIOR to sample collection to reduce the risks for sample mix-up. When the sample is collected, it is the submitter's responsibility to accurately and completely record the date and time of collection.

To complete the newborn screening accurately, the MNSP newborn screening card must be filled out with health information regarding the following:

The Infant – we need to know:

- The main identifications for the infant (Name, healthcare number (Hospital ID number for babies born in New Brunswick), DOB, and sex)
- The time of birth to verify the age of collection.
- Whether the infant is part of a multiple birth, and if their twin is the same sex. This is important because same sex multiples require a repeat newborn screen.
- The birth weight. This is important because low birth weight babies require a repeat newborn screen.
- The gestational age. This is important because premature babies require a repeat newborn screen.

The Clinical Status – we need to know:

- Whether the baby was admitted to the NICU **at the time of** collection
- How the baby is feeding **at the time of collection** (particularly if they were receiving TPN)
- If the baby received a Packed Red Blood Cell (PRBC) transfusion either after birth or in utero, then please check off YES. Please include the date of the first transfusion. This is important because if the transfusion was before the newborn screening sample was collected, a repeat newborn screen will be required. You may indicate "no" for transfusion status on the newborn screening card requisition if an infant has only received fresh frozen plasma (FFP) and/or platelets, or has not received any kind of red blood cell transfusion.
- Whether the mother had complications, specifically, whether she had acute fatty liver of pregnancy (AFLP), Gestational Diabetes, HELLP syndrome (hemolysis, elevated liver enzymes, low platelet count), or a previous sudden infant death syndrome (SIDS). This is important because the clinical status of the mother can influence the infant's newborn screen results.

The Mother – we need to know:

- The main identifications for the mother (Name, DOB, HCN)
- The contact information. This is extremely important should we need to reach the family urgently because of positive screening results.
- If the baby is adopted or the biological mother is a surrogate, we also need the information for the adoptive/legal parents.

The Healthcare Provider – we need to know:

- The birth hospital, midwifery practice, or birthing center. This is important as it allows us to send results to the appropriate facility. If the baby was born at one hospital and the blotter is collected after a transfer to another hospital, please provide information for both health centres.
- The ordering health care provider; this individual must be a physician, midwife, or nurse practitioner.
- The baby's primary health care provider. This is critical information because most babies have already been discharged and we will need to contact the primary healthcare provider if the results need to be followed up on.

The Collection - we need to know:

- Whether this is a first full newborn screen or a subsequent follow-up. For non-newborn screening samples please specify if it is for an acylcarnitine profile or a monitoring sample for a patient with known PKU.
- The date and time of collection. This is important because a repeat sample is needed if the newborn screening sample is collected outside of optimal timelines (24-48 hours of age).
- The name of the collector, printed legibly

Additional Information:

- If the birth is via a woman who is a surrogate: please include the information about the woman who was the surrogate as well as the guardian who will have custody of the child.
- If the baby will be adopted at birth: please include the information about the birth mother as well as the adoptive guardian.
- If the child is not in the custody of the birth mother: please include this information and who has custody of the child.
- Other information: if there is other information that may be relevant to the child's care and reporting of medical results, please include this information.
- Additional information may not fit on the newborn screening card. Please label an extra piece of paper with the baby's information and identifiers, and include this additional information. You can attach this extra piece of paper on the bottom of the blotter card (the other side than the blood spots). Please do not staple through or damage the blood spots in any way.

It is important to have demographic information on the biological and/or birth parents for clinical follow up. The newborn screen may be Screen Positive because a biological and/or birth parent is affected with a certain condition, not the infant. Therefore, we need to have demographic information about the biological and/or birth parents so we can contact them, if the results are important to their health.

Information about who is the Legal Guardian must be included when a newborn screening sample is sent to the IWK Health Centre. This is to ensure information is passed on to the appropriate care giver when necessary.

Please see the Appendix for a copy of the MNSP newborn screening card which shows the demographic/medical history information you are required to fill out. For the most up to date version of these documents, please visit our website at www.maritimenewbornscreening.ca.

Submitters are responsible for ensuring that all of the fields on the MNSP newborn screening card are filled out accurately and completely BEFORE it is sent to the MNSP.

It can be helpful to have a "quality control check" procedure, where all MNSP newborn screening cards are checked by a charge nurse, unit clerk, manager or laboratory staff before they are sent out to ensure it has been fully and accurately completed and to determine if the specimen was collected at or after 24 hours of age. This quality control check can also serve to make sure the specimen is satisfactory, as discussed in a later section.

3.6 Recommendations for Timing and Frequency of Sample Collection

Full term healthy infants

A newborn screening sample should be taken between 24 hours and 48 hours after the birth of the baby. It is during this timeframe that the levels of analytes are optimized for accurate detection of the diseases screened.

Infants that are discharged early (<24 hours)

The optimal time for collection of the newborn screen is 24-48 hours of age. Samples collected at less than 24 hours of age are considered unreliable because of the risk for false negative results. However, if an infant is discharged prior to 24 hours of age, it is advisable to still take a newborn screen before discharge, AND THEN another newborn screen must also be collected during the optimal time of 24-48 hours of age. This is because some of the disorders can be picked up before 24 hours of age and if the child is indeed affected, even a day's delay in diagnosis could prove harmful. Samples collected before 24 hours are still run by MNSP because of this chance that the screen could show screen positive result. If this happens, the infant will be referred for diagnostic evaluation for that disease in order that diagnosis can happen ASAP. A repeat newborn screen will still need to be collected in order for screening to be completed for other diseases that cannot be picked up before 24 hours.

Parents may defer the newborn screen prior to 24 hours of age, but they must sign the deferral form and be counselled on the potential risks. The deferral form should be sent to the MNSP.

The date and time of specimen collection should be clearly indicated on the newborn screening card. Based on the information received, the MNSP tracking system flags screens taken before 24 hours of age. The Maritime Newborn Screening Program will follow-up with submitting facilities and/or the primary health care provider to make sure this repeat is done.

Premature (<34 weeks gestational age) and Low Birth Weight (<2000g) Infants

The physiological prematurity of these infants can affect the levels of some of the analytes screened, creating the potential for false negative or false positive results. Many of these babies will be in the hospital/NICU for some period of time.

These babies should have two newborn screening samples taken:

1) The first newborn screening sample should be collected at 24-48 hours after birth.
2) The second newborn screening sample should be collected at 14 days of age OR prior to discharge from the hospital/NICU, whichever comes first.

The gestational age and birth weight of every baby should be clearly indicated on the MNSP newborn screening card. Based on this information, the MNSP tracking system flags premature and low birth weight babies. The MNSP will contact the birth/NICU facility by phone and/or fax to help make sure the second sample is collected.

Same Sex Twins

There is a chance that twin to twin transfusion occurred in sets of identical multiples. To make sure we are measuring the levels correctly for each individual infant, same sex multiples should receive two newborn screening samples:

1) The first sample should be taken between 24 - 48 hours of age.
2) The second sample should be taken at 14 days of age.

Based on the information we receive on the newborn screening blotter card, the MNSP tracking system flags same sex twins. The MNSP will contact the primary health care provider

(or the hospital should the infants still be admitted) by phone and/or fax to help make sure the second sample is collected.

Infants that are transferred to another facility
If an infant requires a transfer to another facility, a specimen should be collected prior to transfer regardless of the patient's age (for the same reasons as those for infants discharged early). If the sample was not collected or was collected too early, the transferring facility is responsible for informing the receiving facility of the need for sample collection. The receiving facility is responsible for ensuring follow-up.

Infants receiving Packed Red Blood Cell (PRBC) transfusions
If an infant has received donor packed red blood cells before the initial newborn screen was taken, we cannot be sure that the newborn screening result for hemoglobinopathies, Biotinidase Deficiency or Classic Galactosemia are accurate.

The initial newborn screen should be collected prior to transfusion, no matter the age.
If an infant does receive a PRBC transfusion before the first newborn screening sample was taken, they should have a second newborn screening done three months after the date of the last transfusion.

Based on the information we receive on the newborn screening blotter card, the MNSP tracking system flags infants who have been transfused. The MNSP will contact the primary health care provider by phone and/or fax to help make sure the second sample is collected.

Samples that are received by the lab >10 days after collection
The stability of the analytes in the dried blood specimen can be significantly affected if there is a delay in transportation and receipt of the sample by the MNSP. The samples will still be run, but due to the decreased reliability, another specimen must be collected.

Based on the information we receive on the newborn screening blotter card, the MNSP tracking system flags infants whose samples are received by the lab >10 days after collection. The MNSP will contact the submitting facility or the primary health care provider by phone and/or fax to help make sure the second sample is collected.

Initial samples collected after 2 weeks of age
The levels of many of the screening markers naturally drop or rise over the first two weeks of life. Initial samples taken from infants older than two weeks of age will be analyzed, however the accuracy is decreased. It is possible that different algorithms will have to be used to minimize the risk of false negative results.

3.7 Pain Reduction Strategies

Parents may be hesitant to participate in newborn screening because of the pain associated with the heel poke/stick. Pain reduction strategies can be used that decrease or even eliminate the discomfort the infant feels.

Nonpharmacologic approaches are generally more effective when used in combination than when used alone. The use of pharmacologic pain management is not recommended for a newborn screening heel poke/stick.

Breastfeeding
Breastfeeding or oral administration of breast milk has been shown to be as effective in pain management as analgesic measures. It elevates endorphin and oxytocin levels in newborns, which reduce pain and divert the infant's attention away from the painful stimulus. Breastfeeding newborns have been shown to cry less and have smaller increases in heart rate during painful procedures. Oral glucose or sucrose administration has also been shown to have a similar effect.

Non-nutritive sucking
It has been shown that for both preterm and term infants, pacifier use results in decreased crying and lower increases in heart rate in response to painful stimuli.

Swaddling or facilitated tucking
Gently maintaining the arms and legs in a flexed position facilitates self-soothing behaviors (e.g. hand-to-mouth movement, non-nutritive sucking) and is developmentally supportive. It has been shown to decrease the pain an infant experiences during the newborn screening heel poke/stick.

Skin-to-skin contact
Skin-to-skin contact has long been established as an effective and safe way to reduce neonatal pain. The most effective skin-to-skin contact is known as kangaroo care which consists of the infant resting between the mother's (or other family members) breasts. This position stimulates a number of different tactile systems in the infant and reduces the neonatal pain response.

Sensorial saturation
The use of touch, massage, voice, and/or smell has been used to decrease the pain an infant experiences by the heel poke/stick. Combinations of sensorial experiences are more effective at reducing pain than one modality alone. Keep in mind that too much stimulation causes distress and may sensitize the infant to pain. Consider talking or singing softly to the infant while gently rubbing the cheeks or arms. A calming or familiar smell could also be used to help minimize neonatal pain.

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Campbell-Yeo M, Johnston C, Benoit B, Latimer M, Vincer M, Walker CD, Streiner D, Inglis D, Caddell K. Trial of repeated analgesia with Kangaroo Mother Care (TRAKC Trial). BMC Pediatr 2013: 13:182

3.8 Recommended Procedure for Sample Collection

The MNSP newborn screening card consists of four main parts:

1. The filter paper at the top for specimen collection,
2. The parent information sheet at the front,
3. The requisition with required fields (and 2 carbon copies),
4. The decline/defer form.

Avoid touching the filter paper portion of the MNSP newborn screening card before, during, and after collection of the sample. Do not allow water, feeding formulas, antiseptic solutions, glove powder, hand lotion, or other materials to come into contact with the MNSP newborn screening card, especially the filter paper, before or after its use.

Step One

Ensure that the expiry date on the MNSP newborn screening card has not passed. If it has, use another non-expired card for the collection. If you ONLY have expired cards, order new cards immediately (see section 8.2 for instructions). In the meantime, collect the specimen on the expired cards, but note that the sample will be considered unsatisfactory and a repeat newborn screen will be required.

Step Two

Complete all of the required information on the requisition portion of the MNSP newborn screening card as per the detailed instructions in section 11.1. This must be done prior to specimen collection to reduce the risk for sample mix-up. A blue or black ballpoint pen (not gel pen) should be used so that the carbon copies will be clear.

Step Three

Collect the required number of blood spots in the circles drawn on the filter paper. Currently, we require 5 spots. This may seem like a lot, but should we find any discrepant or abnormal results, we take several different punches from the same card to verify the results. Failure to collect the appropriate number of blood spots can cause the sample to be unsatisfactory due to an insufficiency of blood. Each of the 5 circles on the MNSP newborn screening card filter paper requires approximately 75-100 µL of blood to fill it completely and soak all the way through to the back of the filter paper.

Heel poke/stick (method of choice)

The method of choice for collection of the newborn screening sample is the heel poke/stick method.

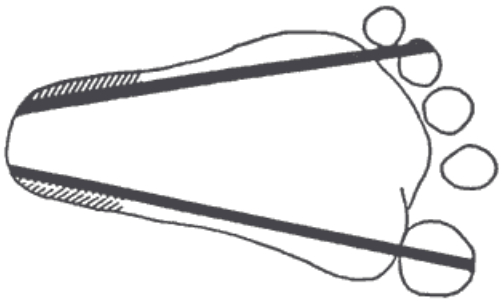
Precautions

Confirm the identity of the infant and ensure accuracy of the information on the newborn screening requisition.

Wash hands vigorously before proceeding. Use appropriate personal protection precautions such as wearing powder-free gloves and changing gloves between infants. Dispose of sharps in the appropriate sharps container.

Site Preparation
The medial or lateral portion of the infant's heel is the preferred site of collection.

Warming the infant's heel can help increase blood flow; use a warm moist towel for three minutes. Positioning the infant's heel lower than the heart can also increase bloodflow.



CAUTION: topical anesthetics should not be used as they may impair blood flow and could also contaminate the sample leading to unreliable results.

Disinfect the area with 70% alcohol and allow the skin to air dry.

Heel Poke/stick
Puncture the appropriate site with a disposable sterile lancet or retractable incision device. Any device used should not puncture beyond 2mm. Scalpel blades or needles must not be used.

Application of Blood
Wipe away the first drop of blood with a sterile gauze pad or cotton ball. Allow a second, large blood drop to form. You can apply gently, intermittent pressure as the blood drop is forming.

Excessive milking or squeezing should be avoided as it can cause hemolysis of the blood or result in a mixture of tissue fluids, both of which can cause inaccurate results.

Carefully touch the filter paper against the large blood drop and, in one step, allow the blood to soak through and completely fill one preprinted circle. Do not touch the filter paper directly to the skin.

The blood circle should be uniform and have saturated through to fill the back side of the filter paper as well. Do NOT apply blood to both sides separately. Do not layer another blood drop on top of a partially filled circle, as this causes layering, which results in non-uniform analyte concentrations that can lead to inaccurate results.

Repeat this procedure, using large drops of blood to completely fill each of the five preprinted circles on the filter paper.

After the blood has been collected, the foot should be elevated above the heart and a sterile gauze pad or cotton ball pressed against the puncture site until bleeding stops.

Drying and preparation for shipping
After application to the blood spot collection card, avoid touching or smearing the blood spots.

Place the freshly collected bloodspot card on a horizontal, level, non-absorbent, open surface and allow it to air dry at room temperature (15-22ºC) for at least 3 hours.

Keep the specimen away from direct sunlight (indirect room light is not usually detrimental unless accompanied by heat).

Blood spots should not be touched, stacked, or allowed to touch other surfaces during the drying process. Even once the blood spots are dry, they should not be allowed to touch other specimens due to the risk of cross-contamination. Please use the fold over paper attachment included with the MNSP newborn screening card filter paper; this will help protect the specimens. Do not use stables or tape to secure the flap.

Shipping - See section 3.12

3.9 Collection by Means Other Than the Heel Poke

Capillary Tube

This method should only be used if the heel poke method is not an option. A specimens can be obtained by applying blood to the blood spot collection card from a sterile, anticoagulant-free capillary tube. The capillary tube collection method may also apply to blood collected from other sources and transferred onto filter paper.

The use of anticoagulants should be avoided during the collection of the newborn screening sample. Ethylenediaminetetraacetic acid (EDTA) may cause interference with some laboratory tests. Since heparin is a known inhibitor of the Polymerase Chain Reaction (PCR) is should be avoided as it may result in test failure in some circumstances. **This method should only be used if the heel poke method is not an option.**

Collection
Using a fresh sterile, plain (additive-free) capillary tube for each circle to be filled on the blood spot collection card, collect the appropriate volume of blood (each of the five 11 mm circles requires approximately 75-100 uL) required for the newborn screen.

Allow blood to flow into the capillary tube by capillary action from the blood drop formed at the heel puncture site. This can be done by touching the end of the capillary tube directly to the blood drop formed on the babies' foot. Fill rates might be improved by holding the tube in a near-horizontal position when touching to the blood drop. Collect enough blood to fill all the circles.

Application
After filling the capillary tube, immediately apply the contents of that tube to the centre of a single, preprinted circle on the filter paper, completely filling the circle. Waiting too long before application will allow cells and plasma to separate or the blood to clot. Both of these will result in a sample that is not able to be used for newborn screening analysis.

To avoid damaging the filter paper fibers, **do not allow the capillary tube to touch the filter paper**. Actions such as "colouring in" the circle, repeated dabbing around the circle, or any

technique that might scratch, abrade, compress, or indent the paper should not be used. These actions may lead to compression of the filter paper and inaccurate blood volume collection.

Do not reuse capillary tubes.

Apply blood to only one side of the filter paper. Do not apply multiple capillary specimens to the same circle, since caking or heterogeneous spreading will occur and might adversely affect test results.

Collect the required number of uniform blood spots. Failure to collect the appropriate number of blood spots might result in the sample being unsatisfactory for analysis due to insufficient blood.

After blood has been collected from the heel of the newborn, the foot should be elevated above the body, and a sterile gauze pad or cotton swab pressed against the puncture site until the bleeding stops.

Venous/Arterial Sample

Although not the method of choice, blood collection from needle puncture of a vein or artery and its application directly onto the preprinted circles of the filter paper is possible. Blood should not be drawn from an extremity into which IV fluids (including blood) are being or have been infused.

Babies receiving Total Parental Nutrition (TPN) should not have a sample collected with this method.

The routine practice of dorsal hand vein collection is discouraged. Problematic issues include:

- Test results might be affected by blood from different vessel sources
- Hand veins might be needed for IV fluids
- Venous sampling is more invasive than a heel stick

Collection and Application
For sample collection from a dorsal hand vein, select the appropriate sized winged blood collection set (butterfly). Remove or shorten catheter length so blood can flow freely onto the circle on the filter paper. Use standard pediatric venous collection procedures.

For all other venous or arterial sample collections, utilize collection and application procedures described in the heel poke method.

Collect the required number of uniform blood spots. Failure to collect the appropriate number of blood spots might result in the sample being unsatisfactory for analysis due to insufficient blood.

Syringe collection of blood for application onto a blood spot collection card is not recommended because time delays may allow for clot formation and settling of cells producing heterogeneous specimens since anticoagulants are not used.

Special Circumstances

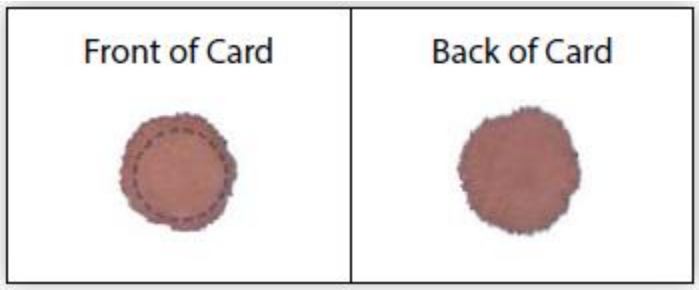
Unique situations may require samples to be collected at different times or in different ways. If you have questions, or need guidance in a certain situation, please call the Maritime Newborn Screening Office at 902-470-7998.

3.10 Satisfactory and Unsatisfactory Newborn Screening Samples

The reference ranges and calculations used for newborn screening analytes are based on the receipt of a satisfactory sample. Testing may not be able to be performed, or if it can, it may not be accurate. This is why specimen quality is of utmost importance. When samples are received in the MNSP, each is reviewed for quantity and quality.

Satisfactory Newborn Screening Samples

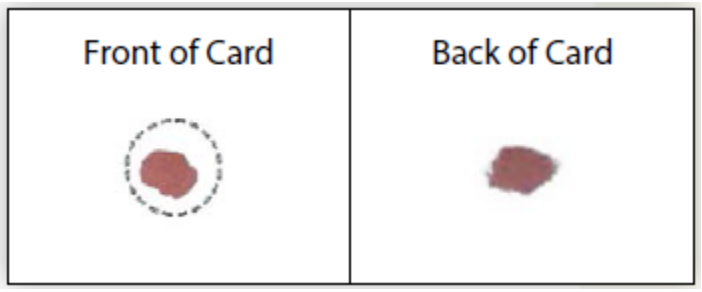
1) Specimen Satisfactory
    a. The blood fills the entire circle.
    b. The blood has saturated both sides of the filter paper (front and back).
    c. The blood is evenly distributed.
2) Data Satisfactory
    a. The entire requisition is filled out accurately and completely.



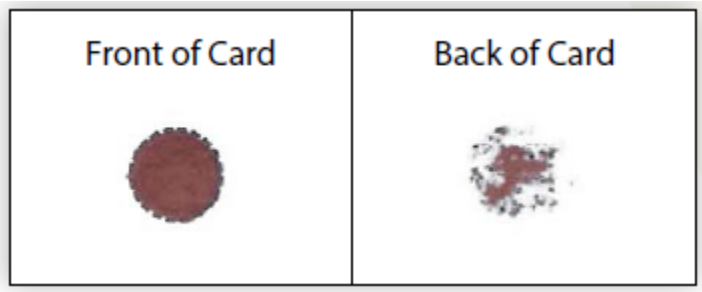
Here is an example of a satisfactory specimen. The blood is fully soaked through to the back of the filter paper. There are no areas of white visible on the front or back of the blotter paper and the circle is completely filled in with blood. It is estimated that 75 – 100 uL of blood is required to fill one circle on the filter paper. The newborn screening test calculations assume that the blood is evenly distributed within the circle and completely saturates both sides of the filter paper.

Unsatisfactory Newborn Screening Samples

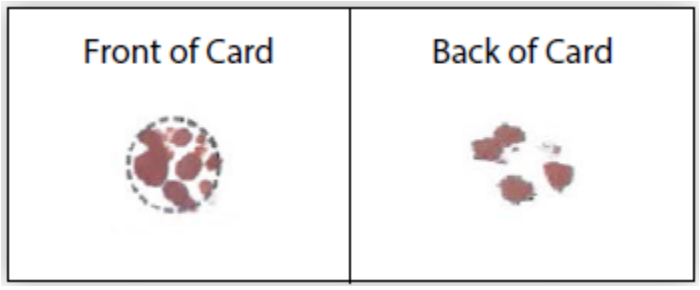
1. Quantity of blood is insufficient



This is an example of a sample that does not have sufficient blood to perform the newborn screen. Although the blood has soaked through to the back of the card, the volume is not sufficient for testing.



Even if the entire circle is filled in with blood, if the blood does not soak through to the back of the filter paper, it will also not be enough sample to run the newborn screen. Sometimes specimens will appear to be collected properly from the front, but will be insufficient when viewed from the back.



Both sides of the filter paper should be examined to assure that the blood has uniformly penetrated and saturated the paper.

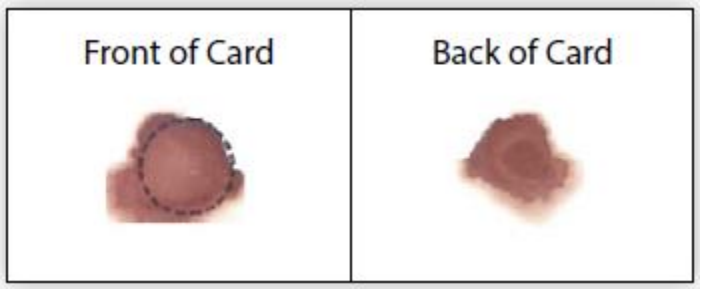
Please do NOT apply blood to both sides of the card.

Failure to collect the appropriate number of blood spots may result in the specimen being unsatisfactory for analysis due to insufficient blood.

2. Blood spots appear scratched or damaged

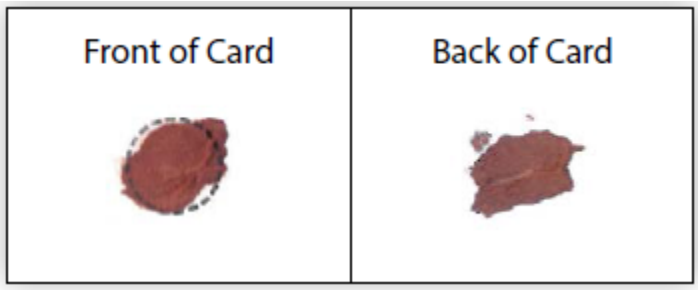
Sometimes the filter paper fibers can be damaged if a capillary tube or butterfly was used to collect a blood specimen and accidentally touches the filter paper. Repeatedly dabbing or "colouring in" the circle may also damage the filter paper. Any technique that might scratch, abrade, compress, or indent the paper should not be used. Do not use the infant's heel to attempt to force the blood through to the back side of the blood spot collection card. This may damage the fibres of the filter paper. These actions may lead to compression of the filter paper and inaccurate blood volume collection.

3. Blood spots are wet and/or discoloured



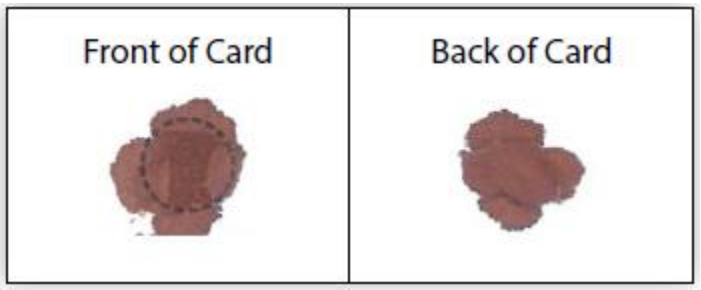
Do not allow water, feeding formulas, antiseptic solutions, glove powder, hand lotion, or other materials to come into contact with the specimen card before or after use. Ensure that the infant's heel is dry and free of alcohol prior to performing the heel stick.

4. Blood spots are supersaturated



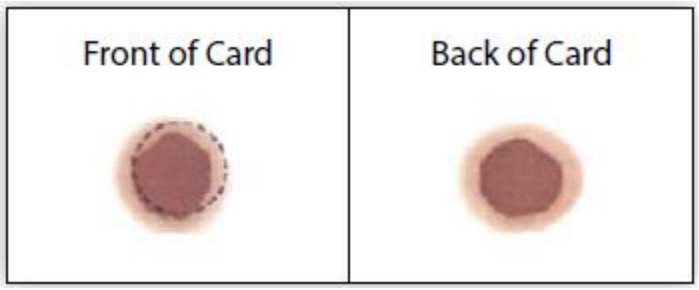
Repeated application of blood in the same area or supersaturation of the filter paper may lead to an excess volume of blood being analyzed during testing, potentially resulting in false negative or false positive screening results.

5. Blood spots appear clotted or layered



Applying successive drops of blood to already partially dried spots causes "layering" and inaccurate blood volume collection, which results in non-uniform analyte concentrations.

6. Blood spots show serum rings



Excessive squeezing or milking the puncture may cause hemolysis of the specimen or result in a mixture of tissue fluids with the specimen which can adversely affect the screening results.

7. Blood spots were damaged during delivery

The blood spot collection cards arrived in a wet or damaged envelope.

ADDITIONAL RESOURCES

1. Simple Spot Check Poster:

   Available at no charge from Whatman:
   www.whatman.com/NeonatalScreeningProducts.aspx

2. Clinical and Laboratory Standards Institute: www.clsi.org
   - Making a Difference Through Newborn Screening: Blood Collection on Filter Paper (DVD)
   - Blood Collection on Filter Paper for Newborn Screening Programs
   - Newborn Screening Quick Guides

8. Information provided on sample collection card was unsatisfactory

The newborn screening requisition portion of the blood spot collection card must be completed to ensure proper specimen labeling for positive identification of the patient. All fields on the blood spot collection card should be filled in as completely as possible. A complete newborn screening report cannot be issued if certain critical fields are not completed.

Failure to provide all information on the card may result in delays in notification of relevant care providers for an infant who screens positive or may make interpretation of the results more difficult.

Common mistakes that cause the requisition to not be satisfactory:

a. Missing Critical Fields (please see below for list)
b. Incorrect date of collection if the sample is collected close to midnight
c. Using the mother's health card number in the infant's health card number field. For babies born in New Brunswick who do not yet have a health card number, please fill in that section with the Hospital ID Number.

Critical Fields

Please ensure that the following fields on the newborn screening requisition are completed:

- The infant's last name
- The infant's date and time of birth
- The date and time of specimen collection
- The date of transfusion (if applicable)
- Birth weight
- Mother/Guardian information
- Submitting Health Care Provider and Family Doctor
- Gestational age

3.11 Rejection Policy

If a specimen has been found to unsatisfactory due to specimen quality, a representative from the Maritime Newborn Screening Program Office will contact the submitter. It is the submitter's responsibility to arrange for a repeat specimen to be collected. The submitter should attempt to communicate the need for a repeat specimen directly to the infant's family or health care provider.

If the data on the specimen card is unsatisfactory, a representative from the Maritime Newborn Screening Program Office will contact the birth hospital to obtain the missing information. Every effort should be made to correctly fill out the information on the screening card. The potential adverse impact of missing and/or inaccurate demographic information on the interpretation of an infant's specimen can have very serious

consequences. Inaccurate information can lead to an infant screening negative for conditions on the panel when truly affected (false negatives), or screening positive when they are not truly affected (false positives). Both scenarios place unnecessary burden on the infant, their family and the health care system. Failure to provide complete information for the mother/guardian and/or primary health care provider can lead to delays in locating an infant who has screened positive for a disorder on the panel – and subsequent delays in the initiation of treatment.

3.12 Specimen Shipping and Transport

Samples should be sent to the MNSP every business day, including Friday's, from all provinces. ***It is NOT acceptable to wait several days and batch samples for send out***. Delays could have serious and irreversible consequences for affected infants. See section 8 for information on shipping supplies.

Specimen Tracking System

Submitters are encouraged to develop an internal system to track that each infant born at their facility has a newborn screen performed. This system should also be used to confirm that a result is received for each sample sent to MNSP.

3.13 Recommendations for Quality Assurance

- Implement a process for ensuring that a newborn screening test is collected/offered for every newborn.
- If the newborn screen is collected, ensure a "spot check" to make sure that the specimen and all required information is satisfactory **before** it is sent.
- Designate individuals responsible for:
    a. Filling in the newborn screening card
    b. Specimen collection
    c. Recording the collection in the infant's chart
    d. Sending the specimen to Maritime Newborn Screening Program
    e. Ensuring test results are received and entered into the infant's chart
- Establish procedure for:
    a. Ensuring specimen collection prior to discharge
    b. Informing parent or guardian for need of repeat testing if the initial specimen was collected prior to 24 hours, after a blood transfusion, or other special circumstances (premature infant, transfer, etc)
    d. Documentation should a parent or guardian decline testing
- Implement a process for ensuring that HCPs and staff are informed of their responsibilities in the newborn screening process.
- Implement the guidelines for specimen collection in this handbook along with those in the Clinical and Laboratory Standards Institute's "Blood Collection on Filter Paper for Neonatal Screening Programs."
- Ensure any new staff are trained and familiar with the processes of newborn screening.

SECTION FOUR: MISSED NEWBORN SCREENS

4.1 Maritime Newborn Screening Program Procedure for Tracking Newborn Screens

It is the responsibility of all submitting facilities (birth hospitals, midwifery practices, or birthing centres) to send complete and correct lists of infants born (Birth Lists). These lists should be sent daily or, at the very least, weekly. Many centres have set up an automatic, electronic fax system to do this.

The MNSP compares these birth lists to completed newborn screens to ensure that all babies born in the Maritimes have been offered Newborn Screening. Should a baby on a birth list not have a newborn screen in our records, the MNSP will contact the submitting facility to determine why the infant has not had a newborn screen. This is to reduce the number of missed newborn screens and improve care for babies born in the Maritimes.

4.2 Common Reasons for a Missed Newborn Screen

The MNSP has identified several common reasons for identification of a missed newborn screen. These include:

- The sample was taken but not sent OR sample collection delayed OR batched sample sending.
- Newborn screen declined by family.
- Infant born in hospital and discharged to midwifery care or home without a newborn screen being collected prior to discharge or after discharge.
- Sample not collected as a result of an error by collecting facility.
- Facility transfer (both hospitals failed to collect the newborn screen).

4.3 Responsibility for Notification and Collection of a Missed Newborn Screen

It is the responsibility of the facility where the infant was born or the midwifery program caring for the baby, to investigate why the newborn screen has not been taken and to help, as needed, in obtaining a sample that was missed.

If you receive a call from the MNSP regarding a potentially missed Newborn Screen:

1) Investigate why it appears that the infant has not had a newborn screen completed. This can be done by looking in hospital records, checking the drying or send out area for Newborn Screens in your facility, and potentially calling the courier services.
2) If it is revealed that a family declined the newborn screening, you must send the MNSP documentation, preferably the signed decline form from the MNSP newborn screening card.
3) For true missed newborn screens, either the submitting facility or the HCP will need to contact the family and have them return as soon as possible to have the newborn screen done. Follow the instructions provided from the MNSP.

4.4 Homebirths

Babies that are born at home with the assistance of a midwife should be captured by the Maritime Newborn Screening Program. They will be listed on the birth lists that all midwife practices send to the program on a daily or weekly basis. Information about newborn screening should be provided to families at the time of the home birth, as well as before the birth during prenatal care. The families should be advised to bring the neonate into their local hospital between 24 to 48 hours after birth to have the newborn screening sample collected.

Babies that are born at home without the assistance of a care provider may not be captured by the Maritime Newborn Screening Program. If a baby is born at home and does not come to the attention of medical providers, then it will not be captured on a birth list, a newborn screening specimen will not be collected, and the baby will be missed. Unfortunately, there is no way, currently, for the Maritime Newborn Screening Program to capture infants in these situations. If a neonate is seen by a health care provider, and the neonate was born at home, please ensure the family is aware of newborn screening and has had this sample collected.

SECTION FIVE: DISEASE INFORMATION

The disorders screened for by the Maritime Newborn Screening Program (MNSP) are based on advice from the Maritime Newborn Screening Advisory Committee comprised of healthcare providers from the three Maritime Provinces and from a variety of specialty areas. The advisory committee reviews disorders and makes recommendations about testing that would be most beneficial to the population it serves. This committee reviews and approves all information created by the MNSP.

Clinical follow-up and case management for patients with a positive newborn screen is conducted by the MNSP Clinical Working Groups. These groups are composed of specialists involved with the various categories of disorders screened.

It must be emphasized that newborn screening is NOT diagnostic, and there is a possibility of false negative and false screen positive results. Therefore, if an infant is experiencing clinical symptoms of any of the diseases included on the newborn screening panel, diagnostic testing should be initiated immediately.

5.1 Summary of Disorders Screened

Disease	Incidence	Primary Analyte Measured	Screening can prevent these symptoms	Treatment
Metabolic Diseases: Amino Acid Disorders				
Maple Syrup Urine Disease (MSUD)	1/200,000 babies born	Leucine/Isoleucine	Failure to thrive, seizures, developmental delay, coma, death	Low protein diet
Phenylketonuria (PKU)	1/12,000	Phenylalanine	Severe and irreversible developmental delay	Phenylalanine restricted diet, supplementation
Metabolic Diseases: Organic Acid Disorders				
Glutaric Acidemia Type I (GA1)	Unknown	C5DC	Developmental delay, spacticity, encephalopathy, coma, death	Avoidance of fasting, low protein diet, medications
Isovaleric Acidemia (IVA)	1/100,000-1/200,000 babies born	C5	Encephalopathy, neurologic damage, coma, death	Avoidance of fasting, low protein diet, medications
Methylmalonic Acidemia (Cobalamin A or B Defects)	1/100,000 babies born	C3	Failure to thrive, encephalopathy, neurologic damage, coma, death	Avoidance of fasting, low protein diet, supplements and/or medications
Methylmalonic Acidemia (Mutase Deficiency)	1/50,000-1/100,000 babies born	C3	Metabolic decompensation, failure to thrive, cardiomyopathy, coma, death	Avoidance of fasting, low protein diet, supplements and/or medications, liver and/or kidney transplantation
Propionic Acidemia (PA)	1/100,000 babies born	C3	Growth impairment, intellectual disability, seizures, pancreatitis, cardiomyopathy, death	Avoidance of fasting, low protein diet, supplements and/or medications, liver and/or kidney transplantation
Metabolic Diseases: Urea Cycle Disorders				
Argininosuccinic Acidemia (ASA)	1/70,000 babies born	Citrulline	Failure to thrive, seizures, developmental delay, coma, death	Dietary management, supplements and/or

				medications and liver transplantation.
Ctrullinemia Type I	1/60,000 babies born	Citrulline	Failure to thrive, seizures, developmental delay, coma, death	Dietary management, supplements and/or medications and liver transplantation.
Metabolic Diseases: Fatty Acid Disorders				
Carnitine Palmitoyl Transferase I (CPTI)	Unknown	C0/C16	Lethargy, hypoketotic hypoglycemia, hepatomegaly, vomiting, myopathy, seizures, death	Avoidance of fasting, high carbohydrate, low fat diet, supplementation
Carnitine Palmitoyl Transferase II (CPTII)	Unknown	C16	Hypoglycemia, seizures, hepatomegaly, cardiomyopathy, arrhythmia, coma, death	Avoidance of fasting, high carbohydrate, low fat diet, supplementation
Carnitine Uptake Disorder (CUD)	Unknown	C0	Cardiomyopathy, hypotonia, hepatomegaly, coma, death	Carnitine supplementation, avoidance of fasting
Long-Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency (LCHAD)	Unknown	C16OH	Cardiomyopathy, seizures, developmental delay, coma, death	Avoidance of fasting, diet low in long-chain fats
Medium-Chain Acyl-CoA Dehydrogenase Deficiency (MCAD)	1/10,000	C8	Seizures, coma, death	Avoidance of fasting, aggressive treatment of illness
Very Long-Chain Acyl-CoA Dehydrogenase Deficiency (VLCAD)	Unknown	C14:1	Developmental delay, failure to thrive, hepatomegaly, cardiomyopathy, coma, death	Avoidance of fasting, special diet
Metabolic Disease: Carbohydrate Disorder				
Classic Galactosemia (GALT)	1/60,000 babies born	Enzyme activity assay	Failure to thrive, liver damage, sepsis, death	Galactose restricted diet
Metabolic Disease: Biotin Disorder				
Biotinidase Deficiency (BIOT)	1/60,000 babies born	Enzyme activity assay	Developmental delay, hypotonia, seizures, skin rash, hair loss, death	Biotin (vitamin) supplementation
Endocrine Diseases:				
Congenital Hypothyroidism	1/3,000 babies born	TSH	Severe and irreversible developmental delay, failure to thrive	Hormone Replacement
Hemoglobinopathies:				
Hemoglobinopathies	1/400 in some ethnic populations	Hemoglobin (Hgb) patterns	Painful sickle crises, tissue ischemia, infection, sepsis, organ damage, anemia	Prophylactic antibiotics, pain management, blood transfusions, bone marrow transplant
Immune Deficiency Disorders:				
Severe Combined Immunodeficiency (SCID)	1/50,000-1/100,000 babies born	TREC (Tcell receptor excision circles)	Severe, life-threatening infections	Prophylactic antibiotics, enzyme therapy, bone marrow transplant
Other Genetic Disorders:				
Cystic Fibrosis (CF)	1/3,600 babies born	Immunotrypsinogen (IRT), CFTR gene mutation analysis	Severe growth failure, chronic lung disease, early death	Enzyme therapy, Pulmonary Therapy

SECTION SIX: POSITIVE NEWBORN SCREEN PROCEDURES

6.1 Metabolic Disorders:

Metabolic disorders occur when the body either cannot metabolize or produce certain amino acids, organic acids, fatty acids, carbohydrates or vitamins, resulting in toxic accumulation of some substances and/or the deficiency of others. Amino acids are derived from protein, thus treatment often involves a low-protein diet and/or a diet low in specific amino acids. Organic acidemias are a class of inherited metabolic disorders that result in the accumulation of organic acids in the blood and urine. Treatment often involves a special diet and may include dietary supplements, medical foods or medications. Fatty acid oxidation defects result in the accumulation of fatty acids in the body and treatment may involve special dietary supplementation and the avoidance of fasting. Urea cycle disorders are caused by a deficiency in one of six enzymes in the urea cycle, which is responsible for removing ammonia from the blood stream through the urine. If this does not happen, then nitrogen builds up in the blood in the form of ammonia and has a toxic effect. Carbohydrate disorders are caused by deficiencies of enzymes involved in glycogen, galactose and fructose metabolism. Children with disorders of carbohydrate metabolism may present with lethargy, encephalopathy, hypoclycemia and hepatomegaly. Biotin disorders are caused by a deficiency in the enzyme biotinidase, leading to a decrease in the vitamin biotin. Treatment involves supplementation with biotin. For a complete list of metabolic conditions included in this screen please refer to Section 5.1. Updated information about all conditions screening for by the MNSP can be found on our website at: www.maritimenewbornscreening.ca

Once a baby has screened positive for a metabolic disorder, the MNSP will contact the family's Health Care Provider to disclose this information. The family will be contacted and told about the screen positive result by the MNSP and an appointment will be arranged for the next metabolic clinic at the IWK. The multidisciplinary metabolic clinic will assess the infant and initiate follow-up investigations to properly diagnose the infant, or rule-out an inborn error of metabolism (false positive newborn screen result). Further medical care will be based on the diagnosis.

6.2 Endocrine Disease Congenital Hypothyroidism (CH):

CH is an endocrine disease which can cause the thyroid gland to make too little thyroid hormone. Babies with this disorder can receive hormones to replace the ones their bodies cannot make. Replacement of thyroid hormone prevents growth problems and mental handicaps.

Once a baby has screened positive for congenital hypothyroidism, the MNSP will contact the IWK Endocrinology Department and communicate the results for patients born in Nova Scotia and Prince Edward Island. The MNSP will then contact the family's Primary Health Care Provider and the family to disclose the screen positive results.

For babies born in New Brunswick, the MNSP will contact the family's Primary Health Care Provider and disclose the screen positive results. The MNSP will then contact a local New Brunswick Endocrinology Department who will inform the family of the screen positive results and coordinate appropriate follow-up.

For all babies who have a screen positive result for CH, an appointment will be scheduled as soon as possible within specific hospitals, depending on which Maritime province the family lives in. Follow-up testing will be used to properly diagnose the infant, or rule-out congenital hypothyroidism (false positive newborn screen result). Further medical care will be based on the diagnosis.

6.3 Hemoglobinopathies:

Hemoglobinopathies constitute a number of inherited conditions that affect the number and/or shape of the red blood cells. Some conditions can cause life-threatening symptoms, while others may not result in any clinical manifestation. When severe cases are left untreated, they can cause a shortage of red blood cells, organ damage, or death. Treatment may include medications and/or blood transfusions.

Once a baby has screened positive for a possible hemoglobinopathy disease or trait, the MNSP will contact the family's Health Care Provider to disclose this information. The family will be contacted and told about the screen positive result. If the screening results indicate a hemoglobinopathy trait, then genetic counselling will be offered to the family through a referral to the Maritime Medical Genetics Service. If the screening results indicate a hemoglobinopathy disease, then a referral will be made to the Hematology Clinic as well as the Maritime Medical Genetics Service. The multidisciplinary team will assess the family and initiate follow-up investigations to properly diagnose the infant and family. Further medical care will be based on the diagnosis.

6.4 Severe Combined Immunodeficiency (SCID):

SCID is an inherited condition in which the baby's body is unable to fight off serious and life-threatening infections. This is due to a low functioning immune system, which puts the baby at risk for getting infections which could lead to death. Treatment for SCID may include isolation from other young children, immunoglobulin replacement therapy, and/or bone marrow transplant.

Once a baby has screened positive for SCID, the MNSP will contact the family's Health Care Provider to disclose this information. The family will be contacted and told about the screen positive result and asked to submit another blood sample at their earliest convenience. Specific precautionary measures will be communicated to the family. Once the secondary blood work results have been interpreted by the IWK Immunologist, the family will be contacted and may be scheduled for an appointment with Immunology based on the results. Further medical care will be based on findings from both the blood work and appointment with the Immunology clinic.

6.5 Cystic Fibrosis (CF):

CF is an inherited disorder of the mucus glands. Mucus is secreted in a baby's body to cover and protect the lungs, digestive system, reproductive system, and other internal organs and tissues. CF causes the baby to produce excess mucus that is abnormally thick and sticky, which can lead to a variety of health problems. If left untreated, CF can cause serious lifelong health problems and a shortened lifespan. However, if the condition is identified early, and proper treatment is initiated, many symptoms of CF can be controlled and the child's lifespan can be extended significantly. Treatment of CF includes dietary changes, supplements, medications, airway clearance therapy, and sometimes lung transplant.

Once a baby has screened positive for CF, the MNSP will contact the family's Health Care Provider to disclose this information. The family will be contacted and told about the screen positive result and asked to come in to the next available Cystic Fibrosis Clinic appointment. At this appointment, the family will meet with members of an interdisciplinary team and secondary diagnostic testing will be initiated. Further medical care and results will come from the Cystic Fibrosis Clinic clinicians.

SECTION SEVEN: PRIVACY AND CONFIDENTIALITY

The MNSP is committed to maintaining the highest level of privacy and confidentiality of the information and dried blood spots it receives. This section outlines the MNSP's policies and procedures surrounding the use and storage of an infant's:

1) Health information found on the newborn screening card, and
2) Dried blood spot specimens.

7.1 Privacy, Confidentiality, and Disclosure of Health Information

Heath Information refers to the information written on the requisition portion of the Newborn Screening Card. As described in detail in section 3.5, this includes health information about the infant, the clinical information, the mother, the healthcare provider, and the collection.

The MNSP limits the collection, use and disclosure of personal health information to the minimum amount necessary to achieve the purpose for which it is collected. As permitted by law, the IWK only collects personal health information for the following purposes:

1) To provide healthcare and medical treatment
2) For the administration and management of the IWK
3) To plan, administer, and manage the quality of the care provided by the IWK
4) To meet any legislative and regulatory requirements (e.g. Vital Statistics Act requirements)
5) To support and promote ethics-board approved research and education at the IWK

The MNSP may also use the health information provided on the cards to share information with healthcare providers who are involved in the care of the infant, or in order to optimize the care we provide.

The MNSP will not use any information from the blotter card that can be traced back to the baby unless it is needed for direct care.

The newborn screening reports for each infant are sent to:

1) The ordering healthcare provider indicated on the card
2) The family physician indicated on the card
3) The birth hospital indicated on the card

An example of the Newborn Screening report issued by the MNSP is found in the Appendix.

If the screen shows a screen positive result, the report will also be provided to the specialist team who will need to provide diagnostic testing for the infant.

7.2 Storage of the Dried Blood Spot Specimens

Newborn Screening Cards (including health information found on the requisition and the dried blood spot samples) are kept in a secure storage facility for ten years. Then they are destroyed, as per IWK policy.

7.3 Use of the Dried Blood Spot Specimens

The dried blood spots are primarily used to provide direct health care to the baby, most importantly for the initial newborn screen.

In the very rare case that an infant with a negative initial newborn screen is subsequently diagnosed with one of the conditions that is screened for (a false negative result), the infant's stored newborn screening sample is re-tested to try and determine why it was missed. The dried blood spot can also be released to another laboratory for other testing requested by a health care provider as per the parent's/guardian's request.

7.4 Additional Uses of the Dried Blood Spot Specimens

When the initial newborn screen is complete and retained dried blood spots are no longer required for direct health care for the patient, the dried blood spots may occasionally be used for other purposes.

1) Dried blood spots may be used as quality control samples, for validation of equipment and methods, or for educational purposes provided patient information is removed or all validation data is maintained in confidential storage.
2) Dried blood spots may be requested by legal warrant or court order.
3) Dried blood spots may be used for research purposes as outlined in Section 7.5.

7.5 Use of the Dried Blood Spot Samples for Research

Research that needs an infant's sample to be linked with his/her identity can only be done after obtaining written consent from the child (if they are old enough to give consent) or from their surrogate decision maker (a parent or guardian). The study would have to be approved by a research ethics board. The child or parent/guardian would be fully informed of the purpose of the research as well as the pros and cons of participating in the research and would have the ability to choose to participate or decline to participate in the research study.

Research that requires an infant's sample may be allowed without obtaining the child's (or their surrogate decision maker's) consent ONLY IF the research is approved by a research ethics board AND all identifying information has been removed so it is impossible to link an individual with the research results.

7.6 Destruction and Release of the Dried Blood Spot Samples

After ten years, the stored Newborn Screening Card (including the health information on the requisition and the dried blood spot) will be securely and confidentially destroyed. Screen positive Newborn Screening Cards are kept for longer periods of time.

7.7 Parental Rights

Parents have the right to:

1) Request access to the personal health information of their child
2) Request a correction to personal health information if it is incorrect
3) Withdraw their consent for any uses as described above.

To decline, a parent should contact the Maritime Newborn Screening Program by phone or email (contact information included in section 2.3).

SECTION EIGHT: ORDERING NEWBORN SCREENING SUPPLIES

8.1 Ordering MNSP Newborn Screening Cards

To order Maritime Newborn Screening Program brochures or collection cards, contact MNBSinfo@iwk.nshealth.ca or call 902-470-7998.

You will need to provide:

- Your healthcare facility
- The shipping address
- How many Newborn Screening Cards/brochures you would like

The MNSP keeps records of all orders of Newborn Screening Cards and brochures.

8.2 Ordering Shipping Supplies

Newborn screening samples are transported to the MNSP using the courier services of Canada Post or a facilities contracted courier service.

For facilities serviced by Canada Post as part of their Newborn Screening contract, order supplies by contacting the MNSP at 902-470-7998 or email MNBSinfo@iwk.nshealth.ca

8.3 Interruptions in Canada Post Services

In the event of Canada Post downtime, the MNSP will send a bulletin with alternate instructions to ensure that sample transport is not disrupted. This is for facilities that have Canada Post shipping as part of their contract with the MNSP.

You can also always feel free to contact the MNSP should you have any concerns.

SECTION NINE: EDUCATIONAL RESOURCES

9.1 Educational Materials Available

Education about newborn screening is vital to the success of the program. It is recommended that facilities provide an orientation about newborn screening to all new employees, including review of this manual.

In addition, the staff at MNSP will work with you to meet the educational needs of your facility and address specific screening practice questions.

The MNSP has created various resources for parents, healthcare providers, and the public.

The majority of these resources are available free of charge on our website: www.maritimenewbornscreening.ca

Hardcopies of the Newborn Screening Brochure can be obtained by emailing MNBSinfo@iwk.nshealth.ca , calling 902-470-7998 or visiting the website.

General Resources

Maritime Newborn Screening Program Brochure: A comprehensive brochure outlining the process and benefits of Newborn Screening. This brochure is designed to be given out prenatally and/or postnatally PRIOR to Newborn Screening. This brochure is available in several languages on the MNSP website.

Resources for Health Care Providers

We have a variety of materials for health care providers to learn about what the newborn screening program does as well as the disorders that we screen for. Clinical information can also be found in these resources, which are all located on our website.

- **Newborn Screening Manual:** For healthcare providers and facilities that collect newborn screening samples.
- **Collecting a Blood Spot for Newborn Screening:** To assist healthcare professional in collecting and submitting satisfactory newborn screening samples.
- **Disorder Fact Sheets for Health Providers:** These fact sheets are designed for healthcare providers and outline what will happen should their patient have a positive newborn screen for a specific disorder. They also include information on the clinical features, etiology, and treatment.
- **Things that can affect Newborn Screening results:** Information on several conditions and/or situations that may create false positive or negative results.

- **Recommendations for the Timing and Frequency of Specimen Collection:** Outline of the policies for timing of specimen collection for different populations of infants. These are also explained in detail in the Newborn Screening Manual.

- **Newborn Screening Decline or Defer Form:** A copy of the Decline/Defer form which is also included in the Newborn Screening Card.

Resources for Parents

Parent education is essential to successful newborn screening. Informed parents are better able to understand screen positive results and the next steps in the process. In addition, informed parents may experience less anxiety associated with a repeat test request for an unsatisfactory sample. Informed parents are also better able to make decisions about their child's health.

- **NBS Information Sheet for Parents/Guardians:** General information regarding newborn screening: the process, the potential results, and the retention policies. This sheet is also included in the Newborn Screening Card and should be provided to parents at the time the Newborn Screening Sample is collected.

- **Newborn Screening Decline or Defer Form:** A copy of the decline/Defer form, which is also included in the Newborn Screening Card.

- **My baby needs a Repeat Newborn Screen:** A resource for parents whose infants require a repeat newborn screen.

- **My Baby Screened Positive for…:** These fact sheets are designed for parents and outline what will happen should their baby have a screen positive result for a specific disorder.

- **Sweat Test Information Sheet:** Information for parents whose infant must undergo a sweat test after a screen positive result for cystic fibrosis.

SECTION TEN: MNSP SCREENING PROCESSES

10.1 Specimen Processing

Every newborn screening specimen the MNSP receives is considered urgent. Analysis and reporting is done on an Urgent basis.

1) Specimen Accessioning

Specimens collected outside the IWK Health Centre are delivered by courier, date & time stamped, and sent to the Newborn Screening accessioning area. Specimens collected within the IWK Health Centre are retrieved from drying racks the following morning and sent to the Newborn Screening accessioning area. Specimens delivered to the accessioning area after

10:30 am (Monday to Friday) may not be accessioned or tested until the following business day.

Specimens are first sorted by screen level (initial vs follow-up vs monitoring) and then by province. At this time, a quality check is performed that identifies issues with specimen quality, missing/incorrect demographic information and/or positive patient identification; these specimens are pulled from the batch for review by a senior technologist to determine if specimen can be analyzed or if a repeat collection is required. For information on Unsatisfactory samples, please see Section 3.10.

Every specimen then receives a special accessioning barcode to facilitate punching, demographic entry, analysis, reporting and follow up (if required). A "punch" is an excised circle of blood which is subsequently used for sample analysis. Each and every sample received is tracked at all times.

2) Data Entry
Specimens are registered and tests ordered in the laboratory information system and then specimen demographics are entered into the newborn screening software, a robust software that facilitates all aspects of our screening program. If any critical information is missing, a data entry clerk contacts the submitter to obtain the missing information. In the event that an infant screens positive, this information allows our newborn screening program to contact the family immediately to arrange further testing.

3) Punching
Each specimen is scanned and a small portion of the blood spot is punched into a series of microtiter plates, facilitated by a specialized instrument that is equipped with blood detection technology that informs the user of the best punching location on the blood portion of the blotter card. The instrument and software also tracks the location of the punches and takes a high resolution image of each punch that can be used for troubleshooting and/or educational purposes. All samples are punched into barcoded microplates that are designated for a specific method/procedure.

10.2 Screening Methodology

I. Procedure for Metabolic Disease screening by amino acid and acylcarnitine analysis by Tandem Mass Spectrometry (MS/MS)

The mass spectrometer is an instrument that separates and quantifies ions based on their mass/charge ratios. TMS permits rapid, sensitive, and accurate measurements of many different types of metabolites with minimal sample preparation. This system is able to detect amino acidemias, organic acidemias, and disorders of fatty acid oxidation in a high-throughput configuration. The TMS platform detects certain errors of inborn metabolism and are separated into two categories of analytes: amino acids and carnitines (free and acyl).

To detect these potential disease states, our lab employs a series of tandem mass spectrometers - instruments widely used for this type of assay because of their specificity and sensitivity. A mass spectrometer is capable of separating components in a matrix and comparing the profile to a library of profiles for positive identification. In the simplest terms, the mass spectrometer is a filter.

The specimen blood spots are punched into a 96 well microtiter plate in which an internal standard mixture was previously added. The dots are then treated with a series of chemicals to extract the analytes from the filter paper and to derivitize the compounds, a step required to enhance analysis in the mass spectrometers.

Once in liquid form, the specimen must become a charged gas to be suitable for measurement in the mass spectrometers. This is achieved by the use of super-heaters and an ion source. Under vacuum, the charged compound enters the first of two filters – only the charged particles (called parent ions) that we are searching for are identified and allowed to proceed to the next stage. In the next section, the ions are fragmented with nitrogen gas to form smaller ions (called daughter ions). The parent and daughter ions then proceed to the final stage where they are filtered once again. All the filtered ions then strike a detector which quantitates or counts the amount of each parent/daughter ion pair. This detector count is then converted into a reportable concentration using sophisticated software.

The process is driven by external hardware, such as pumps, a degasser unit and an auto sampler.

II. Procedure for Congenital Hypothyroidism screening by Thyroid Stimulating Hormone (TSH) analysis

The TSH analysis is performed by a fluorescent immune-assay analyzer that can measure fluorescence, or the emission of light. The specimen blood spot is punched into a 96 well mictrotitre plate that contains antibodies to the TSH molecule, creating an antibody complex. A fluorescent antibody is then attached to the complex and incubated to enhance the process. After an incubation period, a solution is added to the specimen that boosts the formation of highly fluorescent chelates which are in turn measured by the analyzer. The fluorescence of each specimen is proportional to the concentration of TSH in the specimen.

III. Procedure for Cystic Fibrosis screening by Immunoreactive Trypsinogen (IRT) and CFTR gene mutation analysis

The first-tier phenotypic test for CF is the IRT measurement. IRT is made in the pancreas and for infants with CF, pancreatic ducts are partially blocked, leading to an increase of IRT levels in the blood. This can occur for infants with CF that is pancreatic sufficient or insufficient and can also be elevated for heterozygous carriers of CF. The IRT analysis is performed by a fluorescent immune-assay analyzer that can measure fluorescence or the emission of light. The specimen blood dot is punched into a mictrotitre plate that contains antibodies to the IRT molecule, creating an antibody complex. A fluorescent antibody is then attached to the complex and incubated to enhance the process. After an incubation period, a solution is added to the specimen that boosts the formation of highly fluorescent chelates which are in turn measured by the analyzer. The fluorescence of each specimen is proportional to the concentration of IRT in the specimen. Infants who have an elevated IRT measurement are then flagged for second-tier testing via gene mutation analysis. The *CFTR* gene is analyzed for over 30 of common gene mutations, which account for over 90% of CF cases. These genetic

results, in combination with the IRT measurements provide evidence for the newborn screening result.

IV. Procedure for Hemoglobinopathy analysis by High Performance Liquid Chromatography (HPLC) and Capillary Electrophoresis

Newborn screening for hemoglobinopathies is a two-part testing process which includes initial testing performed by HPLC and confirmatory testing performed by electrophoresis.

The initial instrument for hemoglobinopathy newborn screening is based on the chromatographic separation of hemoglobins by ion-exchange High Performance Liquid Chromatography (HPLC). The sample blood spots are punched into 96 well microtiter plates with DI water to allow for the elution of the dried blood for testing. The instrument pumps buffer solutions of increasing ionic strength through an analytical cartridge to a detector. The hemoglobins are eluted out at different retention times, depending on their ionic strengths within the cartridge. When hemoglobin is present, it produces a change in absorbance. A chromatogram is plotted using the change in absorbance vs. time in order to identify specific hemoglobin's present. They are identified by comparing the retention times obtained, to the retention times of known hemoglobins.

If the initial instrument identifies a potential abnormal hemoglobin, the sample is then repunched for confirmation. The confirmatory instrument has eight capillaries that function in parallel and are able to detect abnormal and normal hemoglobins using capillary electrophoresis in free solution. The sample blood dots are punched into 8 well segments with DI water to allow for the elution of the dried blood for testing. The anodic end of the capillary injects the diluted sample and voltage is applied. The hemoglobin fractions are separated by their mobility in an alkaline buffer. After separation, each hemoglobin is detected and quantified at the cathodic end of the capillary using a specific absorbance. The analyzer separates the hemoglobin fractions into one of 13 zones. Each zone has a list of possible normal hemoglobins or hemoglobin variants known to be found in that zone.

V. Procedure for Severe Combined Immunodeficiency by T-Cell Receptor Excision Circles (TREC) analysis

T-cell Receptor Excision circles (TRECs) are circular DNA fragments generated during T-cell receptor rearrangement. In healthy neonates, TRECs are made in large numbers, while in infants with SCID they are barely detectable.

Blood spots are punched into 96 well microtiter plates and go through a PCR amplification process. TREC copy number in blood can be used to distinguish T-cell lymphopenic SCID infants from healthy babies. However, low TRECs copy numbers can also be the results of other immunodeficiency, such as 22q11.2 deletion syndrome (DiGeorge Syndrome or velocardiofacial syndrome), and sometimes as a result of immunosuppression drugs. Confirmatory tests are needed for the diagnosis of SCID and for the determination of the form of SCID.

I. Procedure for Classic Galactosemia (GALT) and Biotinidase Deficiency (BIOT) Screening using Enzyme Activity Assays

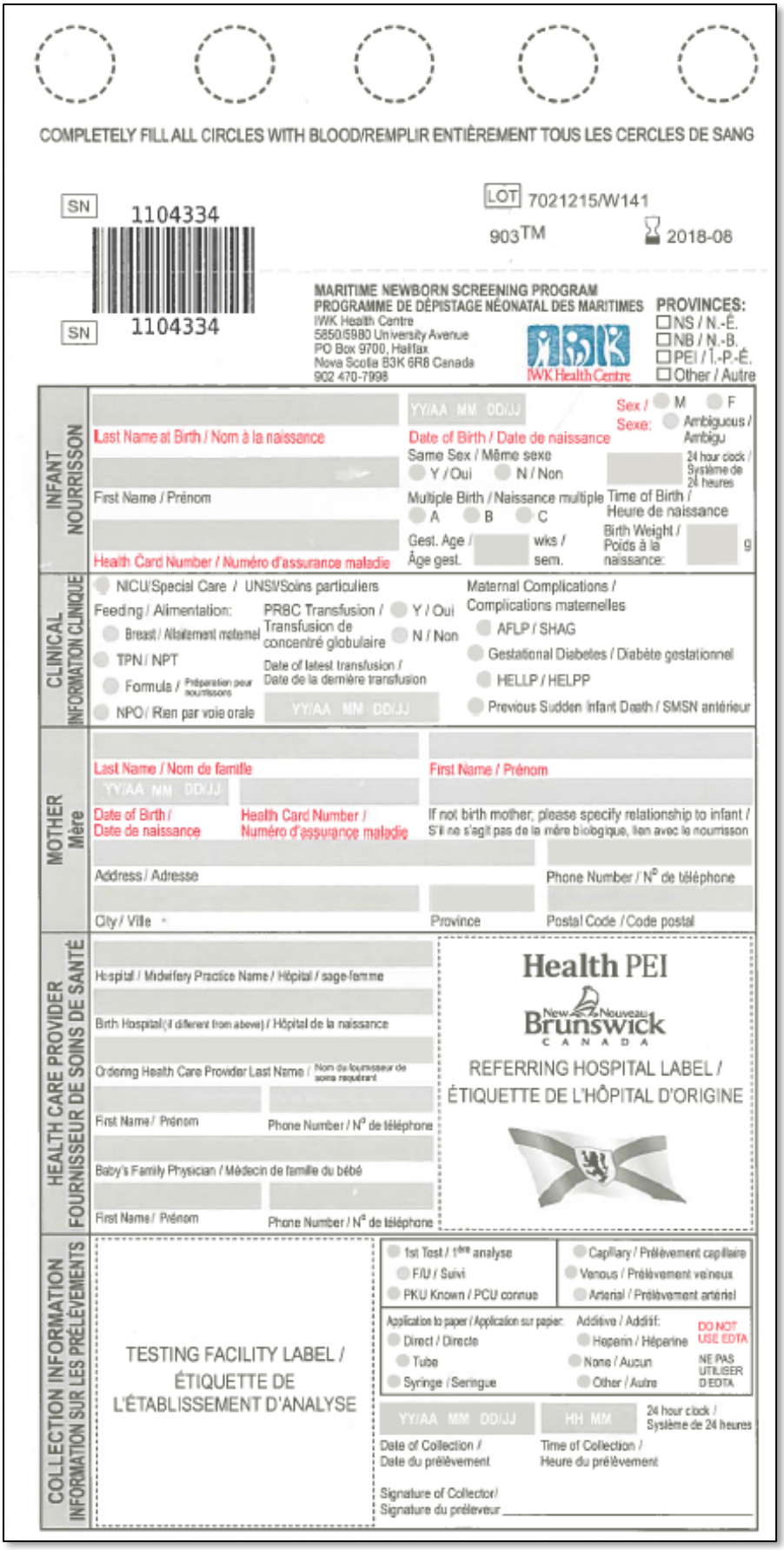
The GALT and BIOT analysis is performed by a fluorescent immune-assay analyzer that can measure fluorescence, or the emission of light. The specimen blood spot is punched into a 96 well mictrotitre plate that contains antibodies to specific enzymes. To screen for GALT, the enzyme being evaluated is galactose-1-phsphate uridylyltransferase. To screen for BIOT, the enzyme being evaluated is biotinidase. The antibodies to these enzymes will bind to form an antibody complex. A fluorescent antibody is then attached to the complex and incubated to enhance the fluorescent signal. After an incubation period, a solution is added to the specimen that boosts the formation of highly fluorescent chelates which are in turn measured by the analyzer. The fluorescence of each specimen is proportional to the amount of enzyme present in the specimen. For patient samples affected by GALT and/or BIOT, low levels of fluorescence will indicate low levels of enzyme.

SECTION ELEVEN: REFERENCES and APPENDICES

11.1 - Newborn screening blood collection card
11.2 - Parent Info sheet
11.3 - Decline/defer form

11.1 Newborn Screening Blood Collection Card

The Newborn Screening blood collection card (blotter card) consists of a form for specific demographic information and patient information, and a blotter area for 5 blood spots. It is imperative that the patient information and blood spots are filled in correctly. For further reference, please see Section 3.5 and 3.8 respectively. An example of the blood collection card can be seen on the next page. For the most up-to-date version, please refer to our website at www.maritimenewbornscreening.ca



COMPLETELY FILL ALL CIRCLES WITH BLOOD/REMPLIR ENTIÈREMENT TOUS LES CERCLES DE SANG

SN 1104334

1104334 SN

LOT 7021215/W141

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2018-08

MARITIME NEWBORN SCREENING PROGRAM
PROGRAMME DE DÉPISTAGE NÉONATAL DES MARITIMES
IWK Health Centre
5850/5980 University Avenue
PO Box 9700, Halifax
Nova Scotia B3K 6R8 Canada
902 470-7998

IWK Health Centre

PROVINCES:
☐ NS / N.-É.
☐ NB / N.-B.
☐ PEI / Î.-P.-É.
☐ Other / Autre

INFANT NOURRISSON

Last Name at Birth / Nom à la naissance

First Name / Prénom

Health Card Number / Numéro d'assurance maladie

YY/AA MM DD/JJ
Date of Birth / Date de naissance

Same Sex / Même sexe ○ Y / Oui ○ N / Non

Multiple Birth / Naissance multiple ○ A ○ B ○ C

Gest. Age / Âge gest. ____ wks / sem.

Sex / Sexe: ○ M ○ F ○ Ambiguous / Ambigu

24 hour clock / Système de 24 heures
Time of Birth / Heure de naissance

Birth Weight / Poids à la naissance: ____ g

CLINICAL INFORMATION CLINIQUE

○ NICU/Special Care / UNSI/Soins particuliers

Feeding / Alimentation:
○ Breast / Allaitement maternel
○ TPN / NPT
○ Formula / Préparation pour nourrissons
○ NPO / Rien par voie orale

PRBC Transfusion / Transfusion de concentré globulaire ○ Y / Oui ○ N / Non

Date of latest transfusion / Date de la dernière transfusion
YY/AA MM DD/JJ

Maternal Complications / Complications maternelles
○ AFLP / SHAG
○ Gestational Diabetes / Diabète gestationnel
○ HELLP / HELPP
○ Previous Sudden Infant Death / SMSN antérieur

MOTHER Mère

Last Name / Nom de famille

First Name / Prénom

YY/AA MM DD/JJ
Date of Birth / Date de naissance

Health Card Number / Numéro d'assurance maladie

If not birth mother, please specify relationship to infant / S'il ne s'agit pas de la mère biologique, lien avec le nourrisson

Address / Adresse

Phone Number / N° de téléphone

City / Ville

Province

Postal Code / Code postal

HEALTH CARE PROVIDER FOURNISSEUR DE SOINS DE SANTÉ

Hospital / Midwifery Practice Name / Hôpital / sage-femme

Birth Hospital (if different from above) / Hôpital de la naissance

Ordering Health Care Provider Last Name / Nom du fournisseur de soins requérant

First Name / Prénom

Phone Number / N° de téléphone

Baby's Family Physician / Médecin de famille du bébé

First Name / Prénom

Phone Number / N° de téléphone

Health PEI

New Nouveau Brunswick CANADA

REFERRING HOSPITAL LABEL / ÉTIQUETTE DE L'HÔPITAL D'ORIGINE

COLLECTION INFORMATION INFORMATION SUR LES PRÉLÈVEMENTS

TESTING FACILITY LABEL / ÉTIQUETTE DE L'ÉTABLISSEMENT D'ANALYSE

○ 1st Test / 1ère analyse
○ F/U / Suivi
○ PKU Known / PCU connue

○ Capillary / Prélèvement capillaire
○ Venous / Prélèvement veineux
○ Arterial / Prélèvement artériel

Application to paper / Application sur papier:
○ Direct / Directe
○ Tube
○ Syringe / Seringue

Additive / Additif:
○ Heparin / Héparine
○ None / Aucun
○ Other / Autre

DO NOT USE EDTA
NE PAS UTILISER D'EDTA

YY/AA MM DD/JJ
Date of Collection / Date du prélèvement

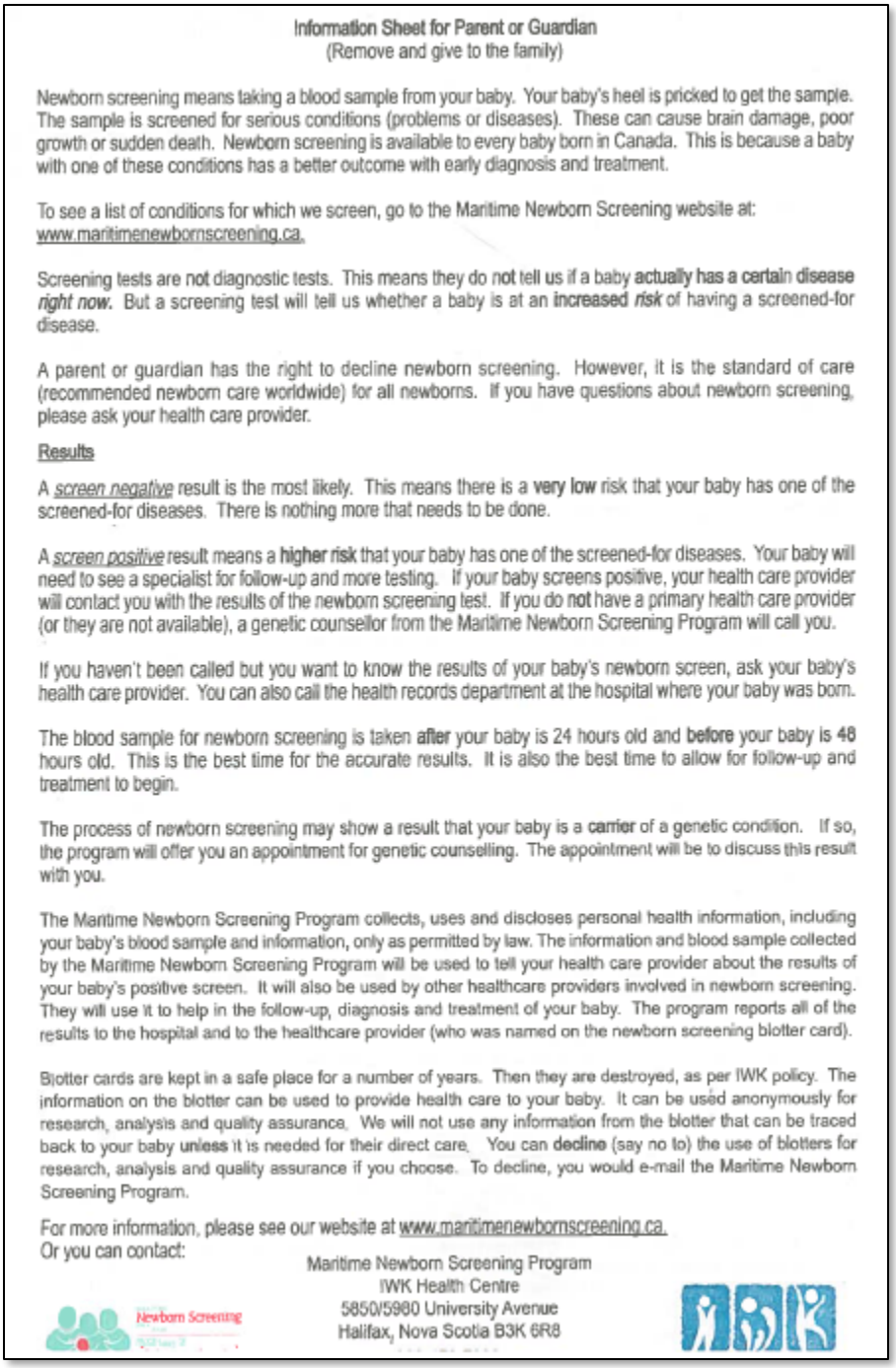
HH MM
Time of Collection / Heure du prélèvement

24 hour clock / Système de 24 heures

Signature of Collector / Signature du préleveur _____

11.2 Parent Information Sheet

The parent information sheet is the top page of the blood collection card. This information sheet can be removed from the blood collection card and given to the family. It contains useful information about the newborn screen, screening results, the screening process, communication of results, and where to find more information. An example of the parent information sheet can be seen below. For the most up-to-date version, please refer to our website at www.maritimenewbornscreening.ca



Information Sheet for Parent or Guardian
(Remove and give to the family)

Newborn screening means taking a blood sample from your baby. Your baby's heel is pricked to get the sample. The sample is screened for serious conditions (problems or diseases). These can cause brain damage, poor growth or sudden death. Newborn screening is available to every baby born in Canada. This is because a baby with one of these conditions has a better outcome with early diagnosis and treatment.

To see a list of conditions for which we screen, go to the Maritime Newborn Screening website at: www.maritimenewbornscreening.ca.

Screening tests are not diagnostic tests. This means they do not tell us if a baby **actually has a certain disease** ***right now.*** But a screening test will tell us whether a baby is at an **increased** ***risk*** of having a screened-for disease.

A parent or guardian has the right to decline newborn screening. However, it is the standard of care (recommended newborn care worldwide) for all newborns. If you have questions about newborn screening, please ask your health care provider.

Results

A *screen negative* result is the most likely. This means there is a **very low** risk that your baby has one of the screened-for diseases. There is nothing more that needs to be done.

A *screen positive* result means a **higher risk** that your baby has one of the screened-for diseases. Your baby will need to see a specialist for follow-up and more testing. If your baby screens positive, your health care provider will contact you with the results of the newborn screening test. If you do **not** have a primary health care provider (or they are not available), a genetic counsellor from the Maritime Newborn Screening Program will call you.

If you haven't been called but you want to know the results of your baby's newborn screen, ask your baby's health care provider. You can also call the health records department at the hospital where your baby was born.

The blood sample for newborn screening is taken **after** your baby is 24 hours old and **before** your baby is 48 hours old. This is the best time for the accurate results. It is also the best time to allow for follow-up and treatment to begin.

The process of newborn screening may show a result that your baby is a **carrier** of a genetic condition. If so, the program will offer you an appointment for genetic counselling. The appointment will be to discuss this result with you.

The Maritime Newborn Screening Program collects, uses and discloses personal health information, including your baby's blood sample and information, only as permitted by law. The information and blood sample collected by the Maritime Newborn Screening Program will be used to tell your health care provider about the results of your baby's positive screen. It will also be used by other healthcare providers involved in newborn screening. They will use it to help in the follow-up, diagnosis and treatment of your baby. The program reports all of the results to the hospital and to the healthcare provider (who was named on the newborn screening blotter card).

Blotter cards are kept in a safe place for a number of years. Then they are destroyed, as per IWK policy. The information on the blotter can be used to provide health care to your baby. It can be used anonymously for research, analysis and quality assurance. We will not use any information from the blotter that can be traced back to your baby **unless** it is needed for their direct care. You can **decline** (say no to) the use of blotters for research, analysis and quality assurance if you choose. To decline, you would e-mail the Maritime Newborn Screening Program.

For more information, please see our website at www.maritimenewbornscreening.ca.
Or you can contact:

Maritime Newborn Screening Program
IWK Health Centre
5850/5980 University Avenue
Halifax, Nova Scotia B3K 6R8

11.3 Decline/Defer Form

This form comes attached to the newborn screening blood collection card. It is located at the back behind all other forms. It contains information about declining or deferring the screen as well as a parental signature space indicating they have read and understand their decision. If this form is filled out, the baby's demographic information must also be filled out and sent to the IWK Health Centre like any other blotter card. An example of this form can be seen on the next page. For the most up-to-date version, please refer to our website at www.maritimenewbornscreening.ca

Newborn Screening Decline or Defer Form

Please have the form completed and signed by the parent or guardian who declines or defers the newborn screening. Return the entire requisition to the Maritime Newborn Screening Program.

I have been informed and understand that:

1. Newborn screening is considered standard of care for all newborns born in the Maritimes.
2. Newborn screening is done to detect treatable disorders that may not cause symptoms for several weeks or months after birth.
3. My baby can look perfectly normal at birth and can still have one of the diseases that are screened for.
4. If treatment is delayed for these conditions, it may cause permanent damage to my child, including severe brain disability, growth and health problems and/or sudden infant death.
5. The goal of newborn screening is early detection so that treatment can begin immediately. This provides the potential for the child to have the best health outcome possible through early screening.
6. Newborn screening samples should be obtained between 24-48 hours of age. Some of the diseases on the screening panel can cause serious health problems within the first few weeks of life so it is recommended that a sample be collected at hospital discharge even if this occurs before 24 hours of age.

Please complete one section below:

☐ I choose NOT to have my baby's blood taken for newborn screening tests: either for the original screen or as required for repeat testing that has been requested.
I understand the above information and I am making an informed decision as my child's guardian to refuse and/or discontinue screening.
I am aware screening is recommended by my child's health care provider and the potential for serious health impact in refusing should I not return to have my child screened or return for screening as requested.

I have chosen to not have my child screened because _____

OR

☐ My baby is/will be discharged before 24 hours of age and I plan to have my baby screened on _____.

Formulaire de refus ou de report du dépistage néonatal

Faire signer le formulaire rempli par le parent ou tuteur qui refuse ou reporte le dépistage néonatal. Retourner la requête au Programme de dépistage néonatal des Maritimes.

On m'a fourni l'information suivante et je comprends ce qui suit :

1. Le dépistage néonatal est considéré comme une norme de soins pour tous les nouveau-nés des Maritimes.
2. Le dépistage néonatal est fait pour dépister des troubles traitables qui peuvent ne pas causer de symptômes durant plusieurs semaines ou mois après la naissance.
3. Mon bébé peut sembler tout à fait normal à la naissance, mais tout de même avoir une des maladies pour lesquelles on fait un dépistage.
4. Un retard dans le traitement de ces affections peut causer des dommages permanents à mon enfant, dont une déficience au niveau du cerveau, des problèmes de croissance et de santé et la mort subite du nourrisson.
5. Le but du dépistage néonatal est de détecter rapidement les affections de façon à débuter immédiatement le traitement. Cela donne la possibilité à l'enfant d'obtenir les meilleurs résultats de santé possibles grâce au dépistage précoce.
6. Les échantillons du dépistage néonatal devraient être prélevés entre 24 et 48 heures après la naissance. Comme certaines des maladies vérifiées dans le cadre du dépistage peuvent causer de graves problèmes de santé dans les premières semaines de vie, il est recommandé qu'un échantillon de sang soit prélevé au moment du congé de l'hôpital, même si le congé a lieu avant les 24 premières heures du bébé.

Veuillez remplir la section ci-dessous.

☐ Je choisis de ne PAS faire subir de prélèvement d'un échantillon de sang à mon bébé pour les tests de dépistage et ce, pour le dépistage initial ou les nouvelles épreuves qui sont demandées.
Je comprends l'information présentée ci-dessus et je prends, à titre de parent ou de tuteur de l'enfant, la décision éclairée de refuser ou de cesser le dépistage.
Je sais que le dépistage est recommandé par le fournisseur de soins de santé de mon enfant et je reconnais la possibilité que mon refus puisse entraîner de graves répercussions pour la santé si je ne reviens pas pour le dépistage de mon enfant ou si je ne reviens pas pour un dépistage demandé.

J'ai choisis de ne pas faire subir le dépistage à mon enfant parce que _____.

OU

☐ Mon bébé a reçu ou recevra son congé avant d'avoir 24 heures et je prévois revenir pour le dépistage le _____.

Baby's name / Nom du bébé	Date of Birth / Date de naissance

Parent/Guardian signature Signature du parent ou tuteur	Parent/Guardian name please print Nom du parent ou tuteur (lettres moulées svp)	Date
Physician/Midwife signature Signature du médecin ou de la sage-femme	Physician/Midwife name please print Nom du médecin ou de la sage-femme (lettres moulées svp)	Date

REF 10550001 Rev.AA