

Perinatal Services BC Neonatal Guideline 9: NEWBORN SCREENING

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While every attempt has been made to ensure that the information contained herein is clinically accurate and current, Perinatal Services BC and BC Newborn Screening Program acknowledge that many issues remain controversial, and therefore may be subject to practice interpretation.

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Executive Summary

BC Newborn Screening Program

BC's Newborn Screening Program screens for 22 disorders.^{1,2}

- About 40 babies per year will be identified with one of these disorders. The most common disorders are congenital hypothyroidism (CH), cystic fibrosis (CF), medium-chain acyl-CoA dehydrogenase deficiency (MCAD), phenylketonuria (PKU), and congenital adrenal hyperplasia (CAH).

Consent for Newborn Screening

- It is best to give the parent(s)/guardian information about newborn screening prenatally and reinforce after the baby's birth (pamphlet on newborn screening is available in multiple languages at www.newbornscreeningbc.ca).
- Newborn screening is standard of care and is performed on all babies unless declined by the parent(s)/guardian. An "informed refusal" process is outlined in this guideline.

Blood Spot Card Collection

- Fill all four areas on the blood spot card completely with the blood sample. For babies where blood is difficult to draw (e.g. premature babies), a minimum of two spots is required.
- Collect blood spot cards for all babies, including babies who are breast/bottle fed, on Total Parenteral Nutrition (TPN) or intravenous fluids or fasting.
- Take blood from arterial lines, when available.
- Do not use cord blood. Maternal blood contamination may interfere with screening result interpretation (CLSI Guideline, 2007).
- Utilize appropriate measures to enhance the comfort of the baby during blood spot collection (e.g., breastfeeding and skin-to-skin contact).

Timing of Collection

1. Babies discharged after 24 hours of age:

Collect the blood spot card prior to discharge and between 24 and 48 hours of age (pre-term and term babies). If collection is not completed during this timeframe, collection should be done no later than 7 days of age.

2. Babies discharged before 24 hours of age:

Recommended Practice:

Collect the blood spot card prior to discharge. The Newborn Screening (NBS) Laboratory at BC Children's Hospital (BCCH) will request, through the baby's physician/midwife, that a repeat sample be collected by 2 weeks (14 days) of age. Rationale for collecting 2 samples on babies discharged before 24 hours of age:

- The first blood screen will identify over 80% of disorders and will help to prevent life threatening events such as severe or potentially fatal bacterial infections in babies with galactosemia or significant metabolic crises in

¹ Test panel includes:

Metabolic disorders	Phenylketonuria (PKU), Maple Syrup Urine Disease (MSUD), Citrullinemia (CIT), Argininosuccinic Acidemia (ASA), Homocystinuria (Hcy), Tyrosinemia I (Tyr), Medium-chain Acyl-CoA Dehydrogenase Deficiency (MCAD), Long-chain Hydroxyacyl-CoA Dehydrogenase Deficiency (LCHAD), Trifunctional Protein Deficiency (TFP), Very-long chain Acyl-CoA Dehydrogenase Deficiency (VLCAD), Propionic Acidemia (PROP), Methylmalonic Acidemia (MUT), Cobalamin Disorders (Cbl A,B), Glutaric Aciduria Type 1 (GA I), Isovaleric Acidemia (IVA), and Galactosemia (GALT).
Endocrine disorders	Congenital Hypothyroidism (CH) and Congenital Adrenal Hyperplasia (CAH).
Hemoglobinopathies	Sickle Cell Disease (HbSS), Sickle Cell/Hemoglobin C (HbSC) and Sickle Cell/ β -thalassemia (HbS/ β -thal).
Cystic fibrosis (CF)	Cystic fibrosis (CF).

² Nine secondary disorders that are not primary targets of the screening program will be identified as "byproducts" of the screening process (see Appendix 6, question 7 for a listing of these disorders).

babies with medium-chain acyl-CoA dehydrogenase deficiency (MCAD), very-long chain acylCoA dehydrogenase deficiency (VLCAD), maple syrup urine disease (MSUD), or congenital adrenal hyperplasia (CAH).

- The second screen optimizes detection of phenylketonuria (PKU), cystic fibrosis (CF) and homocystinuria (Hcy) which are time sensitive and cannot be reliably detected until 24 hours or more after birth.
- Most health authorities (HAs) or hospitals do not have processes in place to track and follow-up babies in the community who did not have a blood spot card collected prior to discharge.
- A pilot study at BC Women's Hospital to trial deferral of testing revealed that up to 8 percent of parents did not return with their baby to the hospital for blood collection. Many parents find it difficult to return to the lab shortly after discharge with their baby, despite their best intentions. If a second sample is never collected, at least the baby will have received most of the benefits of screening.

Note: If an initial blood spot card is collected, the NBS Laboratory will track that a follow-up card(s) is collected (if required). If an initial card is never collected, the NBS Laboratory is unable to identify or track babies for screening.

An "informed deferral" process is outlined in the guideline for babies discharged before 24 hours of age where the parent(s)/guardian wishes to defer blood spot card collection.

Exceptions to Recommended Practice:

Collection of two blood spot cards may not apply if a health authority/hospital has a standard process in place to follow-up after discharge to ensure a blood spot card is collected or if the baby is under the care of a registered midwife.

3. Babies who receive a blood transfusion:

If a blood transfusion is anticipated, whenever possible, collect a blood spot card prior to the transfusion regardless of the baby's age. If not collected prior to the transfusion, collect at 24 – 48 hours of age. The NBS Laboratory will request, through the baby's physician/midwife, repeat cards as required.

4. Very low birth weight babies (1,500 grams or less):

Collect an initial blood spot card between 24 and 48 hours of age. The NBS Laboratory will request, through the baby's physician/midwife, a repeat card at three weeks (21 days) of age or on discharge from hospital to home, whichever is sooner. Rationale: Very low birth weight babies who have congenital hypothyroidism (CH) may have a delayed rise in thyroid stimulating hormone (TSH). The first screen (done at 24 – 48 hours) may miss the detection of CH.

Follow-up of Positive Screens

- The NBS Laboratory will contact the baby's physician/midwife by phone to discuss a positive screen and coordinate next steps.

Storage, Use, Retention, and Disposal of Blood Spot Cards

- Blood spot cards are stored for 10 years. This is consistent with the period of time in which the card has any potential clinical use.
- The primary use of the blood spot card is to conduct blood tests for treatable disorders in order to achieve early diagnosis and improve health outcomes in children. All other uses of the blood spot card are secondary. Secondary uses include clinical, research and other purposes.
- Information about the storage, use, and retention of blood spot cards is provided to the parent(s)/legal guardian via the NBS Parent Information Sheet (A Simple Blood Test Could Save Your Baby's Life) and the NBS Program website.
- Parents/legal guardians have the option to request their baby's blood spot sample be destroyed after newborn screening testing is complete. They may also request the card be returned to them. Details of the process are outlined in this guideline and on the NBS Program website.

Further Information

- BC Newborn Screening Program website: www.newbornscreeningbc.ca or www.perinatalervicesbc.ca.

Neonatal Guideline 9: **NEWBORN SCREENING**

The goal of BC's Newborn Screening (NBS) Program is to identify babies who have a treatable disorder detectable through a blood test. These babies appear normal at birth and, unless they are screened, might otherwise not be diagnosed with one of these disorders before irreversible damage has occurred. If not treated, these conditions are associated with recurrent illnesses and/or developmental disabilities and/or death. Early detection of these disorders allows treatment that may prevent severe mental handicap, growth problems, health problems and sudden infant death (Dietzen, 2009).

Babies born in British Columbia and the Yukon are screened for the following 22 disorders:

Metabolic Disorders

Amino Acid Disorders:

- Phenylketonuria (PKU)
- Maple Syrup Urine Disease (MSUD)
- Citrullinemia (CIT)
- Argininosuccinic Acidemia (ASA)
- Homocystinuria (Hcy)
- Tyrosinemia I (Tyr I)

Fatty Acid Oxidation Disorders:

- Medium-chain Acyl-CoA Dehydrogenase Deficiency (MCAD)
- Long-chain Hydroxyacyl-CoA Dehydrogenase Deficiency (LCHAD)
- Trifunctional Protein Deficiency (TFP)
- Very-long chain AcylCoA Dehydrogenase Deficiency (VLCAD)

Organic Acid Disorders:

- Propionic Acidemia (PROP)
- Methylmalonic Acidemia (MUT)
- Cobalamin Disorders (Cbl A,B)
- Glutaric Aciduria Type 1 (GA I)
- Isovaleric Acidemia (IVA)

Galactosemia (GALT)

Endocrine Disorders

- Congenital Hypothyroidism (CH)
- Congenital Adrenal Hyperplasia (CAH)

Hemoglobinopathies

- Sickle Cell Disease (HbSS)
- Sickle Cell/Hemoglobin C (HbSC)
- Sickle Cell/ β -thalassemia (HbS/ β -thal)

Cystic Fibrosis (CF)

Nine secondary disorders that are not primary targets of the screening program will be identified as "byproducts" of the screening process. See Appendix 6, question 7 for a listing of these disorders.

Additional information on newborn screening for health care providers and families is available on: www.newbornscreeningbc.ca or www.perinatalservicesbc.ca.

2.0 Consent for Blood Spot Card Collection

A. World Health Organization (WHO) Position Statement & Current Practices

The World Health Organization (1998) states that “newborn screening should be mandatory if early diagnosis and treatment will benefit the newborn.”

BC, other Canadian provinces and territories and most other jurisdictions in the world do not require explicit verbal or written consent for newborn screening. It is considered standard of care and is performed on all babies unless declined by the parent(s)/guardian. The rationale is that:

1. There is minimal risk associated with the collection of the blood versus significant consequences of a missed case.
2. The screening test is in the best interests of the baby/child.

B. Process of Informing Parent(s)/Guardian about Newborn Screening

1. Provide the parent(s)/guardian with information about newborn screening. This information is:
 - Best given to parent(s)/guardian prenatally and reinforced after the baby's birth.
 - Available in several languages on the website: www.newbornscreeningbc.ca (see Appendix 1 for an English version of the pamphlet).
2. If the parent(s)/guardian initially declines newborn screening:
 - i. Notify the attending physician/midwife.
 - ii. Review the newborn screening material with the parent(s)/guardian, answer any questions and recommend newborn screening a second time.
 - iii. If the parent(s)/guardian still wishes to decline newborn screening, complete the “Informed Refusal: Newborn Screening” Form (see Appendix 2) and have the physician/midwife and parent(s)/guardian sign.
 - Place the original of the signed form on the baby's health record.
 - Send a copy of the signed form to the baby's physician.
 - iv. Document that testing has been declined (a) in the baby's health record and (b) on the Newborn Record Part 2.

3.0 Blood Spot Card Collection

A. General Notes

- a. Fill all four areas on the blood spot card completely with the blood sample. For babies where blood is difficult to draw (e.g. premature babies), a minimum of two spots is required.
- b. Collect blood spot cards for all babies, including babies who are breast/bottle fed, on Total Parenteral Nutrition (TPN) or intravenous fluids or fasting.
- c. Take blood from arterial lines, when available. Clear line by withdrawing 2ml of blood. After clearing line, collect blood in syringe, remove needle and immediately apply 100µl (0.1 ml) to each printed circle.
- d. Do not use cord blood. Maternal blood contamination may interfere with screening result interpretation (CLSI Guideline, 2007).
- e. Although not the method of choice, specimens may be obtained by applying blood collected in sterile heparinized capillary tubes to the blood spot card. As heparin is known to interfere with molecular testing, samples containing heparin that are tested for the cystic fibrosis gene (approximately 3%) will be invalid and will require recollection. If this type of collection is necessary (e.g., babies in isolation), please indicate the collection method on the requisition.
- f. Complete the blood spot card (see Appendix 3 for a sample card):
 - Check the “Y” box under “Newborn on TPN” if the baby is on TPN at the time the card is collected. If not, check the “N” box.
 - Check the “Y” box under “RBC Transfusion” if the baby has had a red blood cell transfusion prior to collection of the card (also note the date of the transfusion). If the baby has not had a transfusion, check the “N” box.
 - List the physician/midwife that will be caring for the baby in the community under “Physician/Midwife (to be notified of results).” This will usually be a family physician, midwife or pediatrician.

B. Timing of Collection

i. Babies Discharged After 24 Hours of Age

Collect the blood spot card prior to discharge and between 24 and 48 hours of age (pre-term and term babies). If collection is not completed during this timeframe, collection should be done no later than 7 days of age to ensure early detection of these treatable disorders.

Document the collection of the blood spot card on the Newborn Record Part 2.

ii. Babies Transferred Between Hospitals

If a baby is transferred to another facility:

1. Before 24 hours of age: the receiving hospital collects the blood spot card.
2. After 24 hours of age: the sending hospital collects the blood spot card. The receiving hospital confirms that the card was collected and, if it was not, collects the card.

Document the collection of the blood spot card on the Newborn Record Part 2.

iii. Babies Discharged Before 24 Hours of Age

Recommended Practice:

1. Collect the blood spot card prior to discharge and document on the Newborn Record Part 2.
2. The Newborn Screening Laboratory (NBS) will request, through the baby’s physician/midwife, that a repeat sample be collected by two weeks (14 days) of age.

Rationale for collecting 2 samples on babies discharged before 24 hours of age:

- The first blood screen will identify over 80% of disorders and will help to prevent life threatening events such as severe or potentially fatal bacterial infections in babies with galactosemia or significant metabolic crises in babies with medium-chain acyl-CoA dehydrogenase deficiency (MCAD), very-long chain acylCoA dehydrogenase deficiency (VLCAD), maple syrup urine disease (MSUD), or congenital adrenal hyperplasia (CAH).
- The second screen optimizes detection of phenylketonuria (PKU), cystic fibrosis (CF) and homocystinuria (Hcy) which are time sensitive and cannot be reliably detected until 24 hours or more after birth.
- Most HAs or hospitals do not have processes in place to track and follow-up babies in the community who did not have a blood spot card collected prior to discharge.
- A pilot study at BC Women's Hospital to trial deferral of testing revealed that up to 8 percent of parents did not return with their baby to the hospital for blood collection. Many parents find it difficult to return to the lab shortly after discharge with their baby, despite their best intentions. If a second sample is never collected, at least the baby will have received most of the benefits of screening.

Note: If an initial blood spot card is collected, the NBS Laboratory will track that a follow-up card(s) is collected (if required). If an initial card is never collected, the NBS Laboratory is unable to identify or track babies for screening.

Note: Irrespective of best practices, occasionally parents refuse 2 blood collections for their baby. Follow the "Informed Deferral" process (below) if the parent(s)/guardian declines collection of the sample prior to discharge. The Informed Deferral: Newborn Screening Form acknowledges the parents'/guardian's responsibility for taking their baby to a laboratory to have a blood card collected within the recommended timeframe (preferably between 24 and 48 hours of age, but no later than 7 days).

- a. Complete the "Informed Deferral: Newborn Screening" Form (see Appendix 4) and have the physician and parent(s)/guardian sign.
 - Place the original of the signed form on the baby's health record.
 - Send a copy of the signed form to the baby's physician.
- b. Document that testing has been deferred on the Newborn Record Part 2.

Exceptions to the Recommended Practice:

There are two circumstances where a baby may be discharged from hospital without having a blood spot card collected prior to discharge. In both circumstances a standard process is in place to track and follow-up babies to ensure a card is collected within the recommended timeframe (preferably between 24 and 48 hours of age, but no later than 7 days).

Exception #1: HA/hospital has a process in place to track and follow-up babies (see Appendix 5 for key components of such a process).

- HA/hospital designate arrangements for the collection of the blood spot card within the recommended timeframe.
- HA/hospital tracks and follows-up to ensure a blood spot card is collected.
- Parent(s)/guardian signs an "Informed Deferral: Newborn Screening" form (sample in Appendix 4).

Exception #2: Baby is under the care of a Registered Midwife (RM).

- Registered Midwives (RMs), as per the College of Registered Midwives of British Columbia (CRMBC) policy, are responsible for ensuring a blood spot card is collected within the recommended timeframe. The RM collects the card during a home visit and/or makes alternative arrangements for babies born at home or discharged from hospital at less than 24 hours old.
- Parent(s)/guardian sign a "Newborn Screening Agreement for Midwifery Clients" form. This form is provided by the Registered Midwife (RM) and acknowledges the RMs responsibility for collecting the blood spot card

and/or making alternative arrangements.

- See www.cmbc.bc.ca: midwives only section for the College of Midwives of BC (CMBC) policy on collection of blood spot cards.

iv. Babies Who Receive a Blood Transfusion

- Blood transfusions are known to affect the results of hemoglobinopathy and galactosemia screens and may affect other screens as well (Reed, 2000; Korson, 1990).
- If a blood transfusion is anticipated, whenever possible, collect a blood spot card prior to the transfusion regardless of the baby's age.
- If the baby is less than 24 hours old when the card is collected, the NBS Laboratory will request, through the baby's physician/midwife, that a repeat card be collected by two weeks (14 days) of age.
- If a blood spot card is not collected prior to the transfusion, collect at 24 – 48 hours of age. The NBS Laboratory will request, through the baby's physician/midwife, that two repeat cards be collected, one at three weeks (21 days) and one at four months (120 days) after the date of the transfusion.

v. Very Low Birth Weight Babies (1,500 Grams or Less)

- Very low birth weight babies who have congenital hypothyroidism (CH) may have a delayed rise in thyroid stimulating hormone (TSH). The first screen (done at 24 – 48 hours) may miss the identification of CH in some of these babies (Tylek-Lemariska D, 2005; Grufeiro-Papendieck L., 2005).
- For this reason, the NBS Laboratory will request, through the baby's physician/midwife, that a repeat card be collected at day 21 or on discharge from hospital to home, whichever is sooner. If the baby is transferred to another hospital:
 - Before 21 days of age: the receiving hospital collects the blood spot card.
 - After 21 days of age: the sending hospital collects the blood spot card. The receiving hospital confirms that the card was collected and if it was not, collects the card.
- For babies where blood is difficult to draw (e.g., premature babies), it is acceptable to fill 2 out of the 4 available spots on the card.

C. Procedure for Blood Spot Card Collection

The procedure for collection of blood spot cards is outlined below and is found on the back of the newborn screening blood spot card. Refer to references 2, 3, 13 and 14 for further details. Questions about collection of blood in specific circumstances may be directed to the Supervisor of the Blood Collection Department at BC Children's Hospital (ph: 604 875-2139).

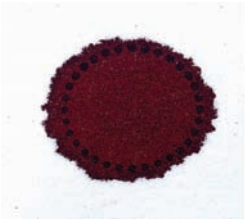
1. Utilize appropriate measures to enhance comfort for the baby during blood spot collection (e.g., breastfeeding, skin-to-skin contact).
2. Wash hands vigorously. Wear powder-free gloves and change gloves between babies.
3. Select puncture site.



Shaded areas indicate safe areas for puncture site (medial or lateral portion of the plantar surface of the baby's heel). Avoid previous puncture sites or the curvature of the heel.

4. If the baby's heel is cool or the first puncture was unsuccessful, warm the heel for 3 minutes (may help increase blood flow).
 - Use a soft cloth moistened with warm (not hot) tap water. Tap water that is too hot may cause burns.

- If a commercial heel warmer is used, follow the manufacturer's guidelines. Remember there is a higher likelihood of burning the skin of babies if the skin is fragile (e.g., premature babies).
5. Place the baby's leg lower than the heart (to increase venous pressure).
 6. Cleanse puncture site with 70% isopropyl alcohol.³
 7. Allow heel to air dry.
 8. Make puncture using sterile safety lancet or heel incision device with tip not longer than 2.0 mm (shorter for babies less than 1,500 grams in weight).
 - Incision devices are recommended as they make a standardized incision (better blood flow, less damage to the heel and fewer punctures required).
 9. Wipe away the first drop of blood with a dry sterilized gauze pad (initial drop contains tissue fluids which might dilute the sample).
 10. Allow a second, large drop of blood to form.
 11. Touch the underside of the filter paper gently against the large drop of blood and, in one step, allow a sufficient quantity of blood to soak through and completely fill a preprinted circle.



Do not press the filter paper against the puncture site on the heel.

Do not layer successive drops of blood or apply blood more than once in the same collection circle.


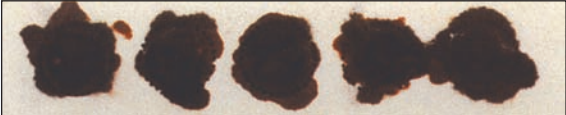
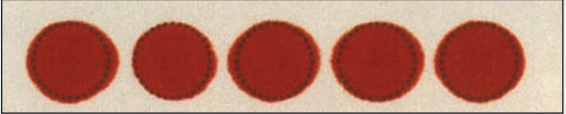

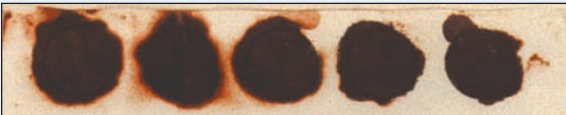
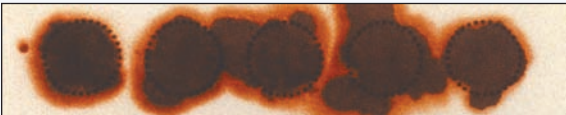


Avoid touching or smearing spots.

Valid Blood Spot (see Table 1 for examples of invalid blood spots)

12. Fill remaining circles with blood in the same manner as step 11, with successive drops of blood. If blood flow is diminished, repeat steps 3 to 8.
13. After blood has been collected, elevate the baby's foot and place a sterile gauze pad against the puncture site until the bleeding stops.
14. Allow blood spots to thoroughly air dry for at least three hours at ambient temperature (18⁰ – 25⁰) on a horizontal, nonabsorptive surface, away from direct heat and sunlight. Do not refrigerate.
15. Once dried, place blood spot cards into the sleeves provided. Place sleeves into an envelope and send to the Newborn Screening Laboratory, BC Children's Hospital, 4480 Oak Street, Room 2F27, Vancouver, BC, V6H 3V4 within 24 hours of collection.

³ 70% isopropyl alcohol is recommended as the antiseptic for the collection of blood spot specimens by the Clinical and Laboratory Standards Institute (CLSI). Refer to the BC Newborn Screening Program website (www.newbornscreeningbc.ca) or contact the NBS Laboratory (p: 604-875-2148) if considering an alternative antiseptic to ensure the solution does not interfere with the results of the test.

Table 1: Examples of Invalid Blood Spot Cards and Possible Causes

Example	Possible Causes
<p>1 Specimen quantity insufficient for testing.</p> 	<ul style="list-style-type: none"> • Removing filter paper before blood has completely filled the circle or before blood has soaked through to second side. • Applying blood to filter paper with a capillary tube. • Allowing filter paper to come into contact with gloved or ungloved hands or substances such as hand lotion or powder, either before or after blood specimen collection.
<p>2 Specimen appears scratched or abraded.</p> 	<ul style="list-style-type: none"> • Applying blood with a capillary tube or other device.
<p>3 Specimen not drying before mailing.</p> 	<ul style="list-style-type: none"> • Mailing specimen before drying for a minimum of three hours.
<p>4 Specimen appears supersaturated.</p> 	<ul style="list-style-type: none"> • Applying excess blood to filter paper, usually with a device. • Applying blood to both sides of filter paper.
<p>5 Specimen appears diluted, discoloured or contaminated.</p> 	<ul style="list-style-type: none"> • Squeezing or “milking” of area surrounding the puncture site. • Allowing filter paper to come into contact with gloved or ungloved hands or substances such as alcohol, formula, antiseptic solutions, water, hand lotions or powder, etc, either before or after blood specimen collection. • Exposing blood spots to direct heat.
<p>6 Specimen exhibits serum rings.</p> 	<ul style="list-style-type: none"> • Not wiping alcohol from puncture site before making skin puncture. • Allowing filter paper to come into contact with alcohol, hand lotion, etc • Squeezing area around puncture site excessively. • Drying specimen improperly. • Applying blood to filter paper with capillary tube.
<p>7 Specimen appears dotted or layered.</p> 	<ul style="list-style-type: none"> • Touching the same circle on filter paper to blood drop several times. • Filling circle on both sides of filter paper.
<p>8 No blood.</p> 	<ul style="list-style-type: none"> • Failure to obtain blood specimen.

Source: Simple Spot Check, modified with permission from Whatman. Information provided by the New York State Department of Health.

4.0 Storage, Use, Retention, and Disposal of NBS Blood Spot Cards

A. Storage, Retention, and Disposal

1. The storage, use, retention and disposal policy aligns with relevant provincial legislation and policies.
2. Blood spot cards are stored for 10 years. This is consistent with the period of time in which the card has any potential clinical use. After 10 years and upon direction provided by the Director of the NBS Program, the cards are disposed of by incineration according to standard operating procedure for biohazardous materials.
3. Blood spot cards are stored in one of two secured locations: NBS Laboratory storage or off-site storage. Access to stored cards may only be requested by the Director of the NBS Program and senior staff designated by the Director. All staff that access stored cards have received training in data privacy.

B. Uses

The primary use of the blood spot card is to conduct blood tests for treatable disorders in order to achieve early diagnosis and improve health outcomes in children. All other uses of the blood spot card are secondary. Secondary uses are divided into clinical, research and other purposes.

1. Clinical Uses

- a. Stored blood spot cards may be used for clinical uses such as:
 - Re-running a test in the event the first test result was not clear.
 - Re-testing in the event of an unexplained illness or death of a baby/child.
 - Laboratory quality control: reference range adjustments, investigation of possible false negative and/or false positive cases.
 - Laboratory method improvements, test protocol refinement and development of new tests.
- b. Release of the card for the follow up of an unexplained baby/child illness or death requires the consent of the parent(s) or legal guardian(s).

2. Research Uses

There is great potential to advance science and clinical care for newborns and children utilizing stored blood spots for health research. Public health research contributes to the public good through increased scientific knowledge. Therefore, residual blood spot cards may be used for health research.

The parent(s)/legal guardian/public is informed that blood spot cards may be used to conduct anonymous NBS research via the Parent Information Pamphlet (see Appendix 1) and the BC Newborn Screening website. All research must be approved by a Clinical Research Ethics Board.

3. Other Uses

Newborn screening blood spot cards are occasionally required to be released if requested by Court order or other legislated authority for purposes such as a coroner or forensic investigation.

C. Duty to Inform

The statements around informing parents/legal guardians and the public are based on the principles of transparency, accountability and respect of privacy.

The parent(s)/legal guardian is provided with information about the storage, retention and uses of newborn screening blood spot cards via:

- NBS Program Information Pamphlet: A Simple Blood Test Could Save Your Baby's Life (see Appendix 1)
- NBS Program Website: www.newbornscreeningbc.ca

D. Opt-out Options

Parent(s)/legal guardian(s) have the option to request their baby's blood spot sample be destroyed after newborn screening testing is complete. They may also request the card be returned to them.

Parent(s)/legal guardians who wish to have their baby's blood card destroyed must:

- a. Sign and send in a directive authorizing destruction of the card (see Appendix 7) to the NBS Laboratory.
 - Both parent(s)/legal guardian(s) must sign the consent.
 - If there is only one parent/legal guardian, the parent/legal guardian must attest to that fact.
- b. Provide proof of identity (photocopy of driver's license or other government issued photo ID).

Parent(s)/legal guardian(s) who wish to have their baby's blood spot card (after being autoclaved*) returned must:

- a. Sign and send in a consent authorizing the release of the card (see Appendix 8).
 - Both parents/legal guardians must sign the release.
 - If there is only one parent/legal guardian, the parent/legal guardian must attest to that fact.
- b. Provide proof of identity (photocopy of driver's license or other government issued photo ID).
- c. Pick up the blood card in person from the NBS Laboratory.
 - Card must be picked up by at least one parent/legal guardian.

*Note: Autoclaving the card is required to remove any potential biohazard but will make the card unusable for further biological testing. If the card is being requested for a medical investigation, the parent(s)/legal guardian(s) should discuss this with the Director of the NBS Program.

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For more information on Newborn Screening, please contact:

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BC Children's Hospital
4480 Oak Street, Room 2F27, Vancouver, BC, V6H 3V4
Telephone: (604) 875-2148; Fax: (604) 875-3836
www.newbornscreeningbc.ca

College of Midwives of British Columbia
Suite 210, 1682 West 7th Avenue,
Vancouver, BC, V6J 4S6
Telephone: (604) 742-2230
www.cmbc.bc.ca (midwives only section)

BC Newborn Screening Program Information Sheet

A Simple Blood Test Could Save Your Baby's Life



Why is my baby screened?

A small spot of your baby's blood can be used to get important information about his or her health. A newborn baby can look healthy but have a rare and serious disorder that you and your doctor or

midwife may not know about. Newborn screening finds babies who may have one of a number of these rare disorders. When these disorders are found and treated early, the chances of serious health problems are prevented or reduced later in life. If not treated, these disorders can cause severe mental handicap, growth problems, health problems and sudden infant death.

In British Columbia there are about 40 babies born each year (1 out of every 1,000) who are found to have one of these rare disorders.

How is my baby screened?

Your baby's heel is pricked and a few drops of blood are taken and put onto a special card. Your baby may cry, but taking the blood sample does not harm your baby. You can help your baby by holding and breastfeeding her or him while the blood is being taken. The blood sample is sent to the laboratory at BC Children's Hospital for testing. The same blood sample is used to screen for all disorders.

How soon after birth will my baby be screened?

The blood sample is usually taken between 24 and 48 hours after birth. This will be done before your baby leaves the hospital or, if a home birth, by your midwife at home.

What if my baby goes home before 24 hours old?

A blood sample will be taken in the hospital before leaving. Over 80% of disorders can be screened using this blood sample. You will be given instructions on how to have the sample repeated within 2 weeks. The purpose of the second sample is to double check the few disorders that can be missed on the first (early) screen.

Can I wait and have my baby tested later?

The earlier these treatable disorders are found, the better the outcome for babies with these disorders. It is strongly advised that your baby not leave the hospital without a blood sample being taken. If you decide you do not want your baby to have a blood sample taken before he or she leaves the hospital, you will be asked to sign a form to show you understand the reasons for the test and the possible outcome for your baby if your baby is not tested and has one of these disorders.

If your baby is under the care of a Registered Midwife, the midwife may review options with you to have the blood sample drawn at home.

How do I find out the results of the screening?

Your baby's screening results are reported to the hospital where your baby was born and your baby's doctor or midwife.

What does it mean if the screen is negative?

A negative screen means that the chance that your baby has one of these disorders is very low. Very rarely, the test may miss a baby with one of these disorders.

What does it mean if the screen is positive and what happens next?

A positive screen tells that there might be a problem. It does not mean that your baby has one of these disorders, but it is possible. More tests are needed.

Will screening for these disorders find anything else?

Screening for sickle cell disease and cystic fibrosis may also tell if your baby is a carrier for one of these disorders. Babies who are carriers are healthy and no more likely to get sick than any other baby. If your baby is a carrier, you will be provided with more information to find out what this means for your baby, yourself and your family.

Which disorders are included in the Newborn Screening?

In British Columbia, babies are screened for 22 rare but treatable disorders. These include:

Metabolic disorders. These occur when the body is not able to break down (metabolize) certain substances in food like fats, proteins or sugars. These substances can build up in the body and cause serious health problems. Serious health problems can usually be prevented with early treatment.

Endocrine disorders. Babies with endocrine disorders of either the thyroid or adrenal glands make too little of certain hormones. Babies with these disorders can receive hormones to replace the ones their bodies cannot make. Replacement of thyroid hormone prevents growth problems and mental handicap. Replacement of adrenal gland hormones can prevent serious health problems such as shock or unexpected death.

Blood disorders. Blood disorders happen when the part of the red blood cell that carries oxygen (hemoglobin) throughout the body is changed. Hemoglobin is important because it picks up oxygen in the lungs and carries it to the other parts of the body. Serious health problems can be prevented through medicines and special treatments.

Cystic Fibrosis. Cystic fibrosis is an inherited life-limiting disorder. It causes thick mucus to build up in the lungs, digestive system (and pancreas) and other organs. Most people with CF get chest infections. They also have problems digesting their food and, as a result, they may not gain weight as well as they should. Early treatment can be started with medicines and physical therapy that help babies with cystic fibrosis digest food and keep their lungs clear of mucus. CF affects about 1 in every 3,600 babies in BC.

What if the results show that my baby has one of the disorders after all the tests are done?

Your baby will need treatment from a doctor who specializes in the disorder. You will be referred to a specialist right away. Treatment can start in a few days.

What happens to my baby's blood spot card when the testing is complete?

Your baby's card with the leftover blood will be kept for 10 years in secure storage by the BC Newborn Screening Program. Occasionally, the dried blood spot samples may be used for other purposes after the testing is finished. These include 1) re-running a test if the first test result was not clear; (2) trying to find the reason for a health problem that has developed later in a child's life or trying to find the cause of an unexplained illness or death of a child; (3) checking the quality of testing done by the laboratory to make sure that results are accurate; and (4) developing better tests for screening of disorders. Samples may also be used for health research if the research has been approved by a Clinical Research Ethics Board. In these cases, all information that may identify the baby is removed.

If you do not wish your baby's stored blood spot card to be used for these purposes, you may fill out a form called a Directive to Destroy Leftover Newborn Screening Blood Samples and send to the BC Newborn Screening Program. See website for details.

If you need more information:

Talk to your doctor or midwife. Visit the Newborn Screening website at www.newbornscreeningbc.ca

We are committed to protecting the privacy of personal information:

The BC Newborn Screening Program collects, uses and discloses personal information only as authorized under section 26 (c) of the BC Freedom of Information and Protection of Privacy Act and other legislation. We take all reasonable steps to make sure that personal information is treated confidentially, is only used for the intended purpose and is securely stored. Should you have any questions regarding the collection, use or disclosure of your baby's personal information, please contact the BC Newborn Screening Program at (604) 875-2148.

Revised November 2010

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The BC Newborn Screening Program is a collaboration of BC Women's Hospital & Health Centre, BC Children's Hospital and Perinatal Services BC, all part of the Provincial Health Services Authority



NEWBORN SCREENING LABORATORY
 BC Children's Hospital
 4480 Oak Street, Room 2F27
 Vancouver, BC V6H 3V4
 Ph: (604) 875-2148
 Fax: (604) 875-3836

Put HA logo here.

Put addressograph label here.

INFORMED REFUSAL: NEWBORN SCREENING

I, _____
 Parent/legal guardian first and last name

the parent/guardian of baby _____
 Circle one Baby's name

born _____ at _____
 Date of birth Name of hospital

DO NOT CONSENT TO HAVE MY BABY'S BLOOD TAKEN FOR NEWBORN SCREENING TESTS
 (tests are listed below).

I make this choice knowing that:

- The screening needs only a few drops of blood from my baby's heel.
- The screening is for 22 treatable disorders (listed below).
- My baby can look perfectly normal at birth and still have one of these disorders.
- These disorders can cause severe mental handicap, growth problems, health problems and sudden infant death. When found and treated early, these problems may be prevented or reduced.

 Signature of parent/guardian

 Date


 Signature of physician/midwife

 Date

 Signature of witness

 Date

Newborn Screening Tests			
Metabolic Disorders <i>Amino Acid Disorders:</i> <ul style="list-style-type: none"> • Phenylketonuria (PKU) • Maple Syrup Urine Disease (MSUD) • Citrullinemia (CIT) • Argininosuccinic Acidemia (ASA) • Homocystinuria (Hcy) • Tyrosinemia I (Tyr I) 	<i>Fatty Acid Oxidation Disorders:</i> <ul style="list-style-type: none"> • Medium-chain Acyl-CoA Dehydrogenase Deficiency (MCAD) • Long-chain Hydroxyacyl-CoA Dehydrogenase Deficiency (LCHAD) • Trifunctional Protein Deficiency (TFP) • Very-long chain AcylCoA Dehydrogenase Deficiency (VLCAD) 	<i>Organic Acid Disorders:</i> <ul style="list-style-type: none"> • Propionic Acidemia (PROP) • Methylmalonic Acidemia (MUT) • Cobalamin Disorders (Cbl A,B) • Glutaric Aciduria Type 1 (GA I) • Isovaleric Acidemia (IVA) 	<i>Galactosemia</i> (GALT)
Endocrine Disorders <ul style="list-style-type: none"> • Congenital Hypothyroidism (CH) • Congenital Adrenal Hyperplasia (CAH) 	Hemoglobinopathies <ul style="list-style-type: none"> • Sickle Cell Disease (HbSS) • Sickle Cell/Hemoglobin C (HbSC) • Sickle Cell/β-thalassemia (HbS/β-thal) 	Cystic Fibrosis (CF)	

 <p>COMPLETELY FILL ALL CIRCLES WITH BLOOD. SOAK THRU</p> <p>Whatman 903® [LOT] W0XX XXXXXXXX</p>			
YYY-MM [REF] 10534567 Rev.3 Whatman 903®		[LOT] W0XX XXXXXXXX	
1) Do not touch sample area 2) Do not use if damaged		NBS lab use only	
<p>BC Children's Hospital NEWBORN SCREENING LABORATORY, Dept. of Pathology 4480 Oak Street, Room 2F27, Vancouver, BC V6H 3V4 Ph: (604) 875-2148, Fax: (604) 875-3836</p>			
PRINT CLEARLY WITH BALLPOINT PEN DO NOT USE ADDRESSOGRAPH			
NEWBORN			
Name		<input type="checkbox"/> M <input type="checkbox"/> F	
Surname	First Name		
PHN	Date of Birth	Time of Birth	
	dd/Month/yy	__:__:__	
Hospital or Place of Birth		Unit/Hospital #	
Birth Weight (g)	Multiple Birth	Gestational Age:	
	<input type="checkbox"/> Y <input type="checkbox"/> N	<input type="checkbox"/> Term <input type="checkbox"/> Prem ____ (wks)	
Newborn on TPN?	RBC Transfusion?	Last Date:	
<input type="checkbox"/> Y <input type="checkbox"/> N	<input type="checkbox"/> Y <input type="checkbox"/> N	dd/Month/yy	
MOTHER			
Name			
Surname	First Name		
PHN			
PHYSICIAN/MIDWIFE (to be notified of results)			
Name		Billing #	
Surname	First Name		
COLLECTION			
Collected By	Collection Date	Collection Time	
	dd/Month/yy	__:__:__	
Collection Location (if NOT hospital of birth)			
If not an initial Newborn Screen, check below: <input type="checkbox"/> Repeat Screen <input type="checkbox"/> Amino Acid Monitor <input type="checkbox"/> Acylcarnitine Profile <input type="checkbox"/> _____			
Collection instructions on reverse. For information on disorders screened, visit: www.newbornscreeningbc.ca			

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Put HA logo here.

Put addressograph label here.

INFORMED DEFERRAL: NEWBORN SCREENING

I, _____
Parent/legal guardian first and last name

the parent/guardian of baby _____
Circle one Baby's name

born _____ at _____
Date of birth Name of hospital

DO NOT WISH TO HAVE MY BABY'S BLOOD TAKEN FOR NEWBORN SCREENING TESTS PRIOR TO DISCHARGE FROM HOSPITAL (tests are listed below).

- ✓ I will take my baby to a hospital laboratory or follow other plans to have blood taken when my baby is between 24 and 48 hours of age.
- ✓ I am aware that the BC Newborn Screening Laboratory cannot track or follow up on testing if a first blood sample is never collected.

If my baby does not have newborn screening testing, I make this choice knowing that:

- The screening needs only a few drops of blood from my baby's heel.
- The screening is for 22 treatable disorders (listed below).
- My baby can look perfectly normal at birth and still have one of these disorders.
- These disorders can cause severe mental handicap, growth problems, health problems and sudden infant death. When found and treated early, these problems may be prevented or reduced.

 Signature of parent/guardian

 Date

 Signature of physician/midwife

 Date

 Signature of witness

 Date

Newborn Screening Tests			
Metabolic Disorders <i>Amino Acid Disorders:</i> <ul style="list-style-type: none"> • Phenylketonuria (PKU) • Maple Syrup Urine Disease (MSUD) • Citrullinemia (CIT) • Argininosuccinic Acidemia (ASA) • Homocystinuria (Hcy) • Tyrosinemia I (Tyr I) 	<i>Fatty Acid Oxidation Disorders:</i> <ul style="list-style-type: none"> • Medium-chain Acyl-CoA Dehydrogenase Deficiency (MCAD) • Long-chain Hydroxyacyl-CoA Dehydrogenase Deficiency (LCHAD) • Trifunctional Protein Deficiency (TFP) • Very-long chain AcylCoA Dehydrogenase Deficiency (VLCAD) 	<i>Organic Acid Disorders:</i> <ul style="list-style-type: none"> • Propionic Acidemia (PROP) • Methylmalonic Acidemia (MUT) • Cobalamin Disorders (Cbl A,B) • Glutaric Aciduria Type 1 (GA I) • Isovaleric Acidemia (IVA) 	<i>Galactosemia (GALT)</i>
Endocrine Disorders <ul style="list-style-type: none"> • Congenital Hypothyroidism (CH) • Congenital Adrenal Hyperplasia (CAH) 	Hemoglobinopathies <ul style="list-style-type: none"> • Sickle Cell Disease (HbSS) • Sickle Cell/Hemoglobin C (HbSC) • Sickle Cell/β-thalassemia (HbS/β-thal) 	Cystic Fibrosis (CF)	

Appendix 5: Blood Spot Card Collection for Babies Discharged Before 24 Hours of Age

Guidelines for Health Authorities/Hospitals

Note: If an initial blood spot card is collected, the NBS Laboratory will track that a follow-up card(s) is collected (if required). If an initial card is never collected, the NBS Laboratory is unable to identify or track babies for screening.

The recommended timeframe for a blood spot card to be collected is between 24 and 48 hours of age (pre-term and term babies). If collection is not completed during this timeframe, collection should be done no later than 7 days of age to ensure early detection of these treatable disorders.

For babies discharged from hospital before 24 hours of age, collection of a blood spot card prior to discharge is “recommended practice.” The NBS Laboratory will request, through the baby’s physician/midwife, that a repeat card be collected by two weeks (14 days) of age.

Health Authorities (HAs)/hospitals may develop their own processes for tracking and follow-up of babies who have not had a blood spot card collected prior to discharge. Such processes would allow for a single blood spot card to be collected within the recommended timeframe. Recommended components of such a deferral process include:

1. Individual designated to oversee the deferral process.
2. Process to identify babies who have not had a blood spot card collected.
3. Process to collect blood spot cards post-discharge (e.g., hospital laboratory, public health nurse in the home or in a clinic, or physician in the office).
4. Process to track that a blood spot card was collected.
5. Process to follow-up cases in which a blood spot card was not collected.
6. Consideration for tracking and follow-up of babies born in an HA hospital but living outside the HA/hospital catchment area.

Birth hospitals/Health Authorities considering setting up a process to allow deferral of testing are advised to contact the Risk Management department within their HA to review the process.

If standard processes are not in place, it is recommended that a blood spot card be collected prior to hospital discharge and again by two weeks of age (as per the “recommended practice” in the guideline).

Appendix 6: Questions & Answers about Newborn Screening in BC

Disorders Screened and Test Accuracy

1. What disorders does the newborn screen detect?
 - BC's Newborn Screening Program screens for 22 disorders.

Table 1: Disorders Screened

Grouping	Metabolites Measured	Disorder	Abbrev.
Metabolic Disorders			
Amino Acid Disorders	Amino Acids	Phenylketonuria	PKU
		Maple Syrup Urine Disease	MSUD
		Citrullinemia	CIT
		Argininosuccinic Acidemia	ASA
		Homocystinuria	Hcy
		Tyrosinemia, Type I	Tyr I
Fatty Acid Oxidation Disorders	Acylcarnitines	Medium-chain Acyl-CoA Dehydrogenase Deficiency	MCAD
		Long-chain Hydroxyacyl-CoA Dehydrogenase Deficiency	LCHAD
		Trifunctional Protein Deficiency	TFP
		Very-long Chain AcylCoA Dehydrogenase Deficiency	VLCAD
Organic Acid Disorders	Acylcarnitines	Propionic Acidemia	PROP
		Methylmalonic Acidemia	MUT
		Cobalamin Disorders	Cbl A, B
		Glutaric Aciduria, Type I	GA I
		Isovaleric Acidemia	IVA
Galactosemia	GALT enzyme activity	Galactosemia	GALT
Endocrine Disorders	Thyroid stimulating hormone (TSH)	Congenital Hypothyroidism	CH
	17OH-progesterone (1 st tier) Steroid panel (2 nd tier)	Congenital Adrenal Hyperplasia	CAH
Hemoglobinopathies	Hemoglobin HPLC	Sickle Cell Disease	HbSS
		Sickle Cell/Hemoglobin C	HSC
		Sickle Cell/ β -thalassemia	HbS/ β -thal
Cystic Fibrosis	Immunoreactive trypsinogen (IRT) (1 st tier) CFTR mutation panel (2 nd tier)	Cystic Fibrosis	CF

2. Why screen for these disorders and not others?

- The disorders on the screening panel were selected by the Newborn Screening Advisory Committee of BC following an evidence-based review process.
- Factors considered in the decision process included: incidence and seriousness of the disorder, evidence of improved health outcomes with early detection, test performance (accuracy and reliability), availability of confirmatory testing and follow-up treatment.
- The Newborn Screening Advisory Committee will review the panel of screening tests periodically and recommend changes as new tests and/or information about disorders becomes available.

3. What are the possible results of a newborn screen?

- "Negative screen" for all disorders. The NBS Laboratory will send a "negative screen" report to the physician/midwife listed on the blood spot card and to the baby's birth hospital.
- "Repeat sample requested". The NBS Laboratory will send a report to the physician/midwife listed on the blood spot card to request a repeat sample. A copy of the report will be sent to the baby's birth hospital. Usual reasons are:
 - Baby was less than 24 hours old at the time of collection;
 - Baby was less than 1,500 grams at birth;
 - Baby had a blood transfusion prior to collection of the sample; or
 - Sample was unsatisfactory.
- "Positive screen" for one of the disorders. A positive screen does not mean that the baby has a disorder, but only that further testing is required. See question #4.
- Baby is a "CF carrier" or has a hemoglobinopathy "trait." See questions #5 and #6.

4. What is the process if a baby has a "positive screen" for one of these disorders?

- The NBS Laboratory will contact the baby's physician/midwife by phone to discuss the positive screen and coordinate the next steps.
- Generally, the physician/midwife will be asked to contact the family to assess the clinical state of the baby and organize the repeat testing. Contact information for an appropriate specialist on-call will also be provided in cases in which there are immediate clinical concerns.
- The NBS Laboratory will contact the baby's physician/midwife with the repeat testing results as soon as they are available and will coordinate referral to the appropriate clinical specialty if the repeat results are positive.

5. What is the process if a baby is identified as a cystic fibrosis (CF) carrier or as having a hemoglobinopathy trait?

- Screening for cystic fibrosis and hemoglobinopathies may identify a baby that is a CF carrier or has a hemoglobinopathy trait.
- It is important for the parents to know if the baby is a CF carrier or has a hemoglobinopathy trait so they can:
 - tell their child later in life. His or her future partner can choose to have testing to identify the couple's chances of having a baby with CF, or a clinically significant hemoglobinopathy.
 - decide whether they wish to be tested. If the baby is a CF carrier or has a hemoglobinopathy trait, one parent is almost certainly a carrier. There is a small risk that both parents are carriers which would have implications for future pregnancies.
- Resources are available to assist in counseling families with regards to these issues. Referral to the BC Medical Genetics program (see question #6) for genetic counseling may also be appropriate under some circumstances.

6. **If parents wish to have carrier testing for CF or hemoglobinopathy, what is the process?**
- Pre-test counselling and testing can be done or arranged by the physician.
 - If parents wish additional genetic counselling, the physician may refer them to the Provincial Medical Genetics Program in Vancouver (604-875-2157) or Vancouver Island Medical Genetics in Victoria (250-727-4461).
 - Questions from physicians and nurses about carrier testing and the interpretation of results may be directed to a genetic counsellor in one of the Medical Genetics clinics listed above.
7. **Will the newborn screening test identify disorders other than the 22 targeted disorders?**
- Nine secondary disorders that are not primary targets of the screening program may be identified as “by-products” of the screening process:
 - a. Amino Acid Disorders
 - i. Hypermethioninemia (MET)
 - ii. Citrin Deficiency (CIT II)
 - iii. Mild Hyperphenylalaninemia (H-Phe)
 - iv. Biopterin Biosynthesis Defects (BIOPT BS)
 - v. Biopterin Recycling Defects (BIOPT REC)
 - b. Organic Acid Disorders
 - i. Cobalamin C/D (Cbl C/D)
 - ii. 2-methylbutyrylglycinuria (2MBG)
 - c. Fatty Acid Oxidation Disorders
 - i. Multiple Acyl-CoA Dehydrogenase Deficiency (MAD)
 - d. Hemoglobinopathies
 - i. Variant Hemoglobinopathies (Var Hb)
8. **How accurate are the newborn screening tests?**
- The testing accuracy varies between disorders but the testing protocols have been designed to maximize sensitivity (proportion of cases detected) and positive predictive value (PPV: percentage of babies with a positive screen that truly have the disorder.)
 - Some tests have multiple "tiers" of testing where a rapid first tier is completed on all samples to identify those at greatest risk for a disorder, and a more complicated second tier test is then performed to identify those with a false positive result on the first test.

Table 2: Sensitivity and Positive Predictive Value of Newborn Screens for Each Disorder¹

Disorder	Abbrev	# Tiers	Sensitivity	Positive Predictive Value (PPV)
Metabolic Disorders				
Amino Acid Disorders				
Phenylketonuria	PKU	1	99%	~100%
Maple Syrup Urine Disease	MSUD	2	95%	95%
Citrullinemia	CIT	1	99%	~50%
Argininosuccinic Acidemia	ASA	1	99%	~50%
Homocystinuria	Hcy	2	99%	95%
Tyrosinemia, Type I	Tyr I	1	99%	~100%
Fatty Acid Oxidation Disorders				
Medium-chain Acyl-CoA Dehydrogenase Deficiency	MCAD	1	99%	100%
Long-chain Hydroxyacyl-CoA Dehydrogenase	LCHAD	1	~95%	30%
Trifunctional Protein Deficiency	TFP	1	~95%	30%
Very-long Chain AcylCoA Dehydrogenase Deficiency	VLCAD	1	98%	~80%
Organic Acid Disorders				
Propionic Acidemia	PROP	2	97%	30%
Methylmalonic Acidemia	MUT	2	95%	30%
Cobalamin Disorders	Cbl A, B	2	95%	30%
Glutaric Aciduria, Type I	GA I	1	99%	39%
Isovaleric Acidemia	IVA	1	98%	50%
Galactosemia	GALT	1	99%	20%
Endocrine Disorders				
Congenital Hypothyroidism	CH	1	~90%	26%
Congenital Adrenal Hyperplasia	CAH	2	99%	~90%
Hemoglobinopathies				
Sickle Cell Disease	HbSS	1	~99%	~85%
Sickle Cell/Hemoglobin C	HbSC	1	~99%	~85%
Sickle Cell/ β -thalassemia	HbS/ β -thal	1	~99%	~85%
Cystic Fibrosis	CF	1	95%	15%

¹ Sensitivity and positive predictive values estimated from BC empirical data (where available) or literature values for equivalent methods.

Consent for Newborn Screening

9. Is newborn screening mandatory?

- Newborn screening is considered “standard of care” and is recommended for all babies born in BC.
- Newborn screening is not mandatory. Parents or the legal guardian may decline screening after having received information on the purpose and benefits of the screening (see “Informed Refusal” process outlined in this guideline).
- Note: A surrogate mother will not likely have completed the papers to be the legal guardian by the time blood is collected for the newborn screening tests. Consult your health authority policy and/or risk management department as to the acceptability of a surrogate mother signing the refusal form if legal documentation is not in place.
- Information about newborn screening is available in several languages on the website: www.newbornscreeningbc.ca.

Timing of Blood Spot Card Collection

10. What is the best time to collect blood spot cards for newborn screening?

- Blood spot cards are best collected prior to discharge and between 24 and 48 hours of age (pre-term and term babies). If collection is not completed during this time, collection should be done no later than 7 days of age in order to ensure early detection of these treatable disorders.
- If a baby is discharged from hospital prior to 24 hours of age, an initial card is collected before discharge. A second card is collected by 2 weeks (14 days) of age.

11. Is it too late to collect a specimen if the baby is older than 7 days old?

- No, newborn screening can still be done, but the sensitivity of some of the screening tests will be reduced if the sample is collected at greater than 4 weeks of age.

12. Must the baby have started feeding before collecting the specimen?

- No, as long as the specimen was collected after 24 hours, the screen will detect the conditions regardless of whether the baby has started feeding (breast/bottle) or is on Total Parenteral Nutrition (TPN) or intravenous fluids.

13. What if the baby has had a blood transfusion?

- Blood transfusions are known to affect the results of hemoglobinopathy and galactosemia screens and may affect other screens as well (Reed, 2000; Korson, 1990).
- If a blood transfusion is anticipated, whenever possible, collect a blood spot card prior to the transfusion regardless of the baby’s age.
- If the baby is less than 24 hours old when the card is collected, the NBS Laboratory will request, through the baby’s physician/midwife, that a repeat card be collected by two weeks (14 days) of age.
- If a blood spot card is not collected prior to the transfusion, collect at 24 – 48 hours of age. The NBS Laboratory will request, through the baby’s physician/midwife, that two repeat cards be collected, one at three weeks (21 days) and one at four months (120 days) after the date of the transfusion.

14. What if the baby is less than 1,500 grams at birth?

- Very low birth weight babies who have congenital hypothyroidism (CH) may have a delayed rise in thyroid stimulating hormone (TSH). The first screen (done at 24 – 48 hours) may miss the identification of CH in some of these babies (Tylek-Lemariska D, 2005; Grufeiro-Papendieck L., 2005).
- For this reason, the NBS Laboratory will request, through the baby’s physician/midwife, that a repeat card be collected at day 21 or on discharge from hospital, whichever is sooner.
- For babies where blood is difficult to draw (e.g., premature infants), it is acceptable to fill 2 out of the 4 available spots on the card.

15. Does the baby have to return to the hospital of birth to have a repeat specimen collected?
- No, the baby can go to any birthing hospital laboratory in British Columbia.
16. For babies discharged from hospital at less than 24 hours of age, what is the benefit of collecting the blood spot card in hospital if a second card is required anyway?
- The first blood screen will identify over 80% of disorders and will help to prevent life threatening events such as severe or potentially fatal bacterial infections in babies with galactosemia or significant metabolic crises in babies with medium-chain acyl-CoA dehydrogenase deficiency (MCAD), very-long chain acylCoA dehydrogenase deficiency (VLCAD) or maple syrup urine disease (MSUD).
 - The 2nd screen optimizes detection of phenylketonuria (PKU), cystic fibrosis (CF) and homocystinuria (Hcy) which are time sensitive and cannot be reliably detected until 24 hours or more after birth.
17. Why not defer testing and have the parent(s)/guardian return to the hospital lab to have the blood spot card collected later?
- A pilot study at BC Women’s Hospital to trial deferral of testing revealed that up to 8 percent of parents did not return with their baby to the hospital for blood collection. Many parents find it difficult to return to the lab shortly after discharge with their baby, despite their best intentions. If a second sample is never collected, at least the baby will have received most of the benefits of screening.
18. What if parents do not wish to have their baby tested prior to discharge from hospital?
- Provide the parent(s)/guardian information about the rationale for collecting blood prior to discharge and the risks if newborn screening is not done.
 - If they still decline collection of the card prior to discharge from hospital, have them sign the “Informed Deferral: Newborn Screening” Form (see Appendix 4) and make arrangements for them to have a blood card collected after discharge.
 - Exceptions to collection of two blood spot cards may apply if a health authority/hospital has a standard process in place to follow-up after discharge to ensure a blood spot card is collected or if the baby is under the care of a registered midwife. See Appendix 5 in this guideline for information about setting up a deferral process.
- Note: If an initial blood spot card is collected, the NBS Laboratory will track that a follow-up card(s) is collected (if required). If an initial card is never collected, the NBS Laboratory is unable to identify or track babies for screening.**

Refusal of Newborn Screening

19. What if parents do not wish to have their baby screened at all?
- Ask the parent(s)/guardian to read the brochure “A simple blood test could save your baby’s life” (available in multiple languages at www.newbornscreeningbc.ca).
 - Discuss the benefits of newborn screening and answer questions/address concerns. Often, their reservations are due to lack of understanding and can be easily resolved.
 - If they still decline screening, have them sign the “Informed Refusal: Newborn Screening” form.
 - Place a copy of the signed form in the baby’s health record and send a copy to the baby’s physician/midwife.

Storage, Use, Retention, and Disposal of Blood Spot Cards

20. What happens to the blood spot card after the newborn screening tests are done?
- After the tests are done, a very small amount of dried blood is left on the card. The amount left depends on how much blood was collected and whether more tests were needed to make sure the results were accurate. The amount left is usually smaller than a dime.
 - The BC Newborn Screening Program stores the cards with the leftover blood sample for 10 years in one of two secure locations: BC Children’s Hospital NBS laboratory storage or off-site storage. Blood spot cards may only be requested from storage by the Director of the NBS Program and senior staff designated by the Director. All staff that access stored cards have received training in data privacy. After 10 years and upon direction provided by the Director of the BC NBS Program, the cards are disposed of by incineration according to standard operating procedure for biohazardous materials.

21. Why keep blood spot cards after the newborn screening tests are done?

Blood spot cards are kept for clinical purposes such as:

1. Re-running a test in the event the first test result was not clear. This means the test can be repeated without having to get another blood sample from the baby.
2. Trying to find the reason for a health problem that has developed later in a child's life or trying to find the cause of an unexplained illness or death of a baby/child. Sometimes testing the leftover blood spot sample will help to find the cause.
3. Checking the quality of testing done by the laboratory to make sure that results are accurate.
4. Developing better tests for the disorders currently screened or for developing new tests to screen for other treatable disorders.

22. Are blood spot cards ever used for health research?

- Yes, a few studies have been conducted on stored blood spots. In all cases, stored blood spot samples were anonymized, meaning that all the information that identifies the baby (e.g., name, PHN and date of birth) was removed to protect privacy in accordance with the Freedom of Information and Protection of Privacy Act (FOIPPA). All research projects have been approved by a Clinical Research Ethics Board (CREB) to ensure high ethical standards.
- There is great potential to advance science and clinical care for newborns and children utilizing stored blood spots for health research. Public health research contributes to the public good through increased scientific knowledge.

23. Are baby's blood spot cards ever released to third parties?

- Blood spot cards are treated the same as hospital records or medical files held by doctors and hospitals in relation to the powers of the Court. On rare occasions the Court or other legislative authority (e.g., Coroner's Act) may order access to a blood spot card or related information. Without legislated authority, blood spot cards are not released to third parties.

24. Can a baby have the newborn screening tests done but not have the leftover blood card stored with the BC Newborn Screening Program?

- Yes. Parents/legal guardians who do not wish their baby's blood spot card to be stored with the BC Newborn Screening Program may request to have the card destroyed.
- Parents/legal guardians who wish to have their baby's blood spot card destroyed must sign a form called a Directive to Destroy Leftover Newborn Screening Blood Samples. The signed form is sent to the BC Children's Laboratory with proof of identity.
- Once a directive to destroy a card is received, the NBS laboratory separates the filter paper containing the blood spots from the blood spot card. The filter paper and blood spots are destroyed. A letter is sent to the parents/legal guardians to tell them the blood spots have been destroyed. Destroying the card means that the card will no longer be available for any purpose, including further tests if the baby or child develops an illness later in life.
- Both parents/legal guardians must sign the request to destroy their baby's card. If the baby has only one parent/legal guardian, that parent/legal guardian must sign the form to say they are the only parent/legal guardian.
- Occasionally, parents/legal guardians may request to have their baby's blood spot card returned. This may be possible in exceptional circumstances but would require the parents/legal guardians to come to the BC Children's Laboratory with proof of identity to pick up the blood spot card. The card itself will be heat treated (autoclaved) before returning in order to remove any potential biohazard associated with the dried blood. This means the card will no longer be usable for any type of biological testing.

NEWBORN SCREENING LABORATORY
 BC Children’s Hospital
 4480 Oak Street, Room 2F27
 Vancouver, BC V6H 3V4
 Ph:(604) 875-2148 ; Fax: (604) 875-3836



PLEASE READ CAREFULLY AS THIS DOCUMENT AFFECTS YOUR LEGAL RIGHTS.

Directive to Destroy Leftover Newborn Screening Blood Samples

Details of blood sample (so correct card is identified):

Baby’s name: _____ Mother’s name: _____
 Baby’s date of birth: _____ Baby’s hospital/place of birth: _____
 Baby’s Personal Health Number (PHN): _____ Mother’s PHN: _____

Details of requestor:

Both parents must sign to have the card released unless they attest to being the only parent/legal guardian.

We/I, _____ and _____, parents or legal guardians of the baby described above, hereby request the BC Newborn Screening Program to destroy our/my baby’s newborn screening blood spot card. We understand that newborn screening blood spot cards are typically stored by the BC Newborn Screening Program and would be available in the future for any further health-related testing that might be deemed necessary for our child. For example, the ability to diagnose if a disease is congenital or acquired after birth may be limited.

We/I also hereby release the BC Newborn Screening Program, the Provincial Health Services Authority and any of its employees, officers, directors and physicians from any liability whatsoever for destroying this card and for the consequences of not having it available for my/our child’s care in the future.

Date: _____

Name (mother/legal guardian): _____

Witness name: _____

Signature (mother/legal guardian): _____

Witness signature: _____

Date: _____

Name (father/legal guardian): _____

Witness name: _____

Signature (father/legal guardian): _____

Witness signature: _____

If one parent/legal guardian:

I am the only parent/legal guardian of the baby described above.

Parent/Legal Guardian’s signature: _____

Proof of identify MUST be supplied (photocopies only):

1. Baby’s birth certificate; **AND**
2. Parent(s) passport photo page or drivers license; **AND**
3. If legal guardian, provide proof of guardianship.

For each blood spot card, we will:

1. Separate the filter paper containing the blood spots from the blood spot card.
2. Destroy the filter paper and blood spots.
3. Notify you after the blood spots and filter paper have been destroyed, and the date destroyed.
4. File the remaining requisition, your original written request and, a copy of our letter to you.

Please return form, with photocopies of proof of identity to:

NEWBORN SCREENING LABORATORY
 BC Children’s Hospital, Department of Pathology
 4480 Oak Street, Room 2F27, Vancouver, BC V6H 3V4

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 4480 Oak Street, Room 2F27
 Vancouver, BC V6H 3V4
 Ph:(604) 875-2148; Fax: (604) 875-3836



PLEASE READ CAREFULLY AS THIS DOCUMENT AFFECTS YOUR LEGAL RIGHTS.

Release of Leftover Newborn Screening Card

Details of blood sample (so correct card is identified):

Baby’s name: _____ Mother’s name: _____
 Baby’s date of birth: _____ Baby’s hospital/place of birth: _____
 Baby’s Personal Health Number (PHN): _____ Mother’s PHN: _____

Details of requestor:

Both parents must sign to have the card released unless they attest to being the only parent/legal guardian.

We/I, _____ and _____, parents or legal guardians of the baby described above, hereby request the BC Newborn Screening Program to return our/my baby’s newborn screening blood spot card. We understand that newborn screening blood spot cards are typically stored by the BC Newborn Screening Program and would be available in the future for any further health-related testing that might be deemed necessary.

We/I also hereby release the BC Newborn Screening Program, the Provincial Health Services Authority, and any of its employees, officers, directors and physicians from any liability whatsoever for the consequences of not having this card available. (Note the card will be autoclaved as blood is considered a biohazard)

Date: _____

Name (mother/legal guardian): _____ Witness name: _____

Signature (mother/legal guardian): _____ Witness signature: _____

Date: _____

Name (father/legal guardian): _____ Witness name: _____

Signature (father/legal guardian): _____ Witness signature: _____

If one parent/legal guardian:

I am the only parent/legal guardian of the baby described above.

Parent/Legal Guardian’s signature: _____

Proof of identify MUST be supplied (photocopies only):

1. Baby’s birth certificate; AND
2. Parent(s) passport photo page or drivers license; AND
3. If legal guardian, provide proof of guardianship.

Please return form, with photocopies of proof of identity to:

NEWBORN SCREENING LABORATORY
 BC Children’s Hospital, Department of Pathology
 4480 Oak Street, Room 2F27, Vancouver, BC V6H 3V4

Please also bring your proof of identity with you to the BC Children’s Hospital Newborn Screening Laboratory when you come to retrieve the card (the lab will contact you when it is ready for pickup).

Please note the card will be heat treated (autoclaved) prior to returning to remove any potential biohazard from the dried blood spots. As a result the card will no longer be useable for biological testing.