

TITLE

NEWBORN BLOOD SPOT SCREENING FOLLOW-UP

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Provincial

DOCUMENT

HCS-32-01

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Newborn Blood Spot Screening Policy (#HCS-32)

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NOTE: The first appearance of terms in bold in the body of this document (except titles) are defined terms – please refer to the Definitions section.

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OBJECTIVES

- To outline the procedure to be followed when follow-up is required within the **Alberta Newborn Screening Program (ANSP) (formerly the Newborn Metabolic Screening [NMS] Program)**.
- To ensure follow-up communication and actions occur in a timely manner and are monitored for completion.
- To ensure consistent accommodation of special circumstances related to follow-up within the ANSP.

APPLICABILITY

Compliance with this document is required by all Alberta Health Services employees, members of the medical and midwifery staffs, students, volunteers, and other persons acting on behalf of Alberta Health Services (including contracted service providers as necessary).

ELEMENTS**1. Accommodating Special Circumstances**

- 1.1 If an infant is admitted to a neonatal intensive care unit (NICU) or special care nursery, refer to the Alberta Health Services (AHS) *Newborn Blood Spot Screening Neonatal Intensive Care Unit* Guideline.

- 1.2 If any of the following special circumstances occur, refer to Section 14 for additional information:
- a) infant moved out of Alberta;
 - b) **parent** or **guardian** does not present infant for **sample collection**; or
 - c) repeat sample collection for multiple births.

2. Types of Follow-up

- 2.1 Follow-up shall be completed for:
- a) **abnormal results**;
 - b) **borderline results**;
 - c) **inadequate results**;
 - d) sickle cell trait notifications;
 - e) invalid demographic data (e.g., incorrect, missing or illegible demographic data);
 - f) missing initial screens;
 - g) low birth weight infants (see Section 9 below);
 - h) preterm infants (see Section 10 below); and
 - i) flagged registrations (e.g., out of province registrations, transcription errors or temporary identifier used).
- 2.2 For more information about the types of follow-up, see the ANSP staff education resource *Following Up Summary*.

3. Follow-up Communication

- 3.1 The ANSP's application and/or alternative sources (e.g., **laboratory information system**) as required shall be accessed every business day (Monday to Friday, except for statutory holidays) by:
- a) the **Alberta Newborn Screening Laboratory (formerly the NMS Laboratory)** to receive information about the need for follow-up of abnormal results, sickle cell trait notifications and flagged registrations.
 - b) the **ANSP coordination team (formerly the NMS coordination team)** to receive information about the need for follow-up of borderline results, inadequate results, invalid demographic data, missing initial screens, low birth weight and preterm infants.

- 3.2 The need for follow-up of abnormal results shall be communicated by the Alberta Newborn Screening Laboratory to the infant's Physician or Midwife and the appropriate Alberta Children's Hospital and Stollery Children's Hospital specialty clinics (e.g., metabolic, endocrine, immunology, hematology, cystic fibrosis) every business day.
- 3.3 The need for follow-up of borderline results, inadequate results, invalid demographic data, missing initial screens, low birth weight and preterm infants shall be communicated by the ANSP coordination team to **zone public health nursing services** every business day.
- a) For infants under the care of a Midwife, zone public health nursing services shall **transfer** the need for follow-up to the appropriate midwifery service.
- b) For infants within a neonatal intensive care unit (NICU), special care nursery or pediatric inpatient unit, zone public health nursing services shall transfer the need for follow-up to the appropriate service area.
- 3.4 The need for follow-up of borderline results, inadequate results, invalid demographic data, missing initial screens, low birth weight or preterm infants for infants residing in an Indigenous community shall be communicated by the ANSP coordination team to the appropriate public health team in the Indigenous community every business day.

4. Follow-up Actions: Abnormal Results

- 4.1 The Alberta Newborn Screening Laboratory, in accordance with their local service area resources (e.g., laboratory standard operating procedures, job aids, worksheets), shall refer infants with abnormal results to their Physician or Midwife and the appropriate speciality clinic for clinical assessment and diagnostic testing.
- a) As soon as an abnormal result is available, all attempts shall be made to telephone the infant's Physician or Midwife and the appropriate speciality clinic.
- (i) If the infant's Physician or Midwife is unknown or unavailable, all attempts shall be made to telephone an alternate Physician or Midwife until a **most responsible health practitioner (MRHP)** is established for follow-up.
- b) The *Alberta Newborn Screening Laboratory Preliminary Results Sheet* and *ANSP Condition Fact Sheet* shall be sent to the infant's Physician or Midwife and the appropriate speciality clinic.
- c) A formal laboratory report shall be sent to the infant's Physician or Midwife and made available to the appropriate speciality clinic.
- d) Actions shall be documented in the infant's **health record**.

- 4.2 The Physician or Midwife and/or appropriate speciality clinic shall work with parents or guardians of infants with abnormal results to arrange clinical assessment and diagnostic testing to confirm or exclude a diagnosis of a **screened condition**.
- 4.3 If parents or guardians of infants with abnormal results refuse clinical assessment and diagnostic testing, the MRHP shall consider communicating the refusal to Children's Services based on the infant's medical history in accordance with the *Child, Youth & Family Enhancement Act*.
- a) Actions shall be documented by the MRHP in the infant's health record and communicated to the ANSP coordination team and the Alberta Newborn Screening Laboratory.
- 4.4 The appropriate speciality clinic shall provide diagnostic outcomes for infants with abnormal results to the Alberta Newborn Screening Laboratory in a timely manner.
- a) In the rare circumstance that an infant with a normal screen result is diagnosed with a screened condition, the appropriate speciality clinic shall report this to the Alberta Newborn Screening Laboratory and the ANSP coordination team. The specialty clinic, Alberta Newborn Screening Laboratory and the ANSP coordination team shall follow the AHS *Reporting of Clinical Adverse Events, Close Calls, and Hazards* Procedure. The ANSP coordination team shall report to Alberta Health.
- 4.5 The Alberta Newborn Screening Laboratory shall provide diagnostic outcomes to the ANSP coordination team in a timely manner for quality management purposes.
- 5. Follow-up Actions: Borderline Results or Inadequate Results**
- 5.1 Infants with borderline results or inadequate results shall have a repeat **sample** collected within 96 hours of the need for repeat sample collection being identified (except in the case of increased tyrosine).
- a) In the case of increased tyrosine, the repeat sample shall be collected when the infant is between 28 and 42 days of age.
- b) In case of multiple birth sets (e.g., twins, triplets) the repeat sample shall be collected in accordance with Section 14.3 below.
- 5.2 Zone public health nursing services shall coordinate and discuss repeat sample collection with:
- a) the parent or guardian in accordance with the AHS *Newborn Blood Spot Screening: Contacting Parents or Guardians* Guideline; or
- b) a NICU, special care nursery or Midwife as appropriate in accordance with zone-specific practices.

- 5.3 The repeat sample shall be collected in accordance with the AHS *Newborn Blood Spot Screening Sample Collection Procedure*.
- 5.4 Actions shall be documented by the MRHP in the infant's health record and communicated to the ANSP coordination team.
- 5.5 For additional information, see the ANSP staff education resource *Following Up on Infants Essentials*.
- 6. Follow-up Actions: Sickle Cell Trait Notification**
- 6.1 The Alberta Newborn Screening Laboratory, in accordance with their local service area resources (e.g., laboratory standard operating procedures, job aids, worksheets), shall notify the health care provider (as indicated on the requisition) of infants with sickle cell trait of their carrier status including a notification letter and education materials within a week.
- a) If the infant's health care provider is unknown, all attempts shall be made to inform an alternate Physician or Midwife until a MRHP is established.
- b) Actions shall be documented in accordance with Alberta Newborn Screening Laboratory local services area resources.
- 7. Follow-up Actions: Invalid Demographic Data**
- 7.1 Zone public health nursing services shall investigate invalid demographic data in accordance with their local service area resources (e.g., decision-making trees, operational practices, investigation tools) in order to validate and ensure the infant's demographic data is corrected (e.g., date of birth, time of birth, gender).
- 7.2 Invalid demographic data shall be validated by:
- a) cross-checking the infant's demographic data in the **notice of birth**, Netcare and/or infant health record with the infant's demographic data in the **Person Directory**.
- 7.3 Invalid demographic data shall be corrected by:
- a) completing the *Alberta Newborn Screening Laboratory Correction Form* and submitting it to the Alberta Newborn Screening Laboratory who will then correct the infant's demographic data in the laboratory information system; and
- b) requesting corrections and/or making corrections to the infant's demographic data and/or the record that is determined to be incorrect.
- 7.4 Health Information Management may be contacted for assistance with either validation or correction of the infant's demographic data.

- 7.5 If the identity of the infant is in doubt after the correction of invalid demographic data, the Alberta Newborn Screening Laboratory shall re-report the sample to ensure repeat sample collection is arranged in accordance with Sections 5.2 to 5.5 above.
- 7.6 Actions shall be documented in the infant's health record and communicated to the ANSP coordination team.
- 7.7 For additional information, see the ANSP staff education resource *Following Up on Infants Essentials*.
- 8. Follow-up Actions: Missing Initial Screens**
- 8.1 Zone public health nursing services shall investigate missing initial screens in accordance with their local service area resources (e.g., decision making trees, operational practices, investigation tools) in order to determine if an initial sample was collected or if sample collection is required.
- 8.2 If it is determined by documented evidence that an initial sample was never collected or an initial sample was collected but was not received by the Alberta Newborn Screening Laboratory, arrangements shall be made for sample collection as soon as possible in accordance with the AHS *Newborn Blood Spot Screening: Contacting Parents or Guardians* Guideline, unless there is one of the following documented reasons for not collecting the sample:
- a) neonatal death;
 - b) parent or guardian refusal;
 - c) parent or guardian does not present infant for sample collection;
 - d) **Physician refusal;**
 - e) infant moved out of province or unable to locate family; or
 - f) sample collected under another identity.
- 8.3 Sample collection shall occur in accordance with the AHS *Newborn Blood Spot Screening Sample Collection* Procedure.
- 8.4 Actions shall be documented in the infant's health record and communicated to the ANSP coordination team.
- 8.5 For additional information, see the ANSP staff education resource *Following Up on Infants Essentials*.

9. Follow-up Actions: Low Birth Weight Infants

- 9.1 For infants less than 2000 grams at birth a repeat sample shall be collected between 21-28 days after birth in order to support the detection of congenital hypothyroidism or congenital adrenal hyperplasia in low birth weight infants.
- 9.2 Zone public health nursing services shall coordinate and discuss repeat sample collection with:
- a) the parent or guardian in accordance with the AHS *Newborn Blood Spot Screening: Contacting Parents or Guardians* Guideline; or
 - b) a NICU, special care nurse or midwife as appropriate in accordance with zone-specific practices.
- 9.3 The repeat sample shall be collected in accordance with the AHS *Newborn Blood Spot Screening Sample Collection* Procedure.
- 9.4 Actions shall be documented by the MRHP in the infant's health record and communicated to the ANSP coordination team.
- 9.5 For additional information, see the ANSP staff education resource *Following Up on Infants Essentials*.

10. Follow-up Actions: Preterm infants

- 10.1 For infants less than 37 weeks gestational age, a repeat sample may be required to be collected between 21-28 days after birth in order to support the detection of severe combined immunodeficiency and sickle cell disease in preterm infants.
- a) The ANSP coordination team shall send out notifications when these repeat samples are required.
- 10.2 Zone public health nursing services shall coordinate and discuss repeat sample collection with:
- a) the parent or guardian in accordance with the AHS *Newborn Blood Spot Screening: Contacting Parents or Guardians* Guideline; or
 - b) a NICU, special care nurse or Midwife as appropriate in accordance with zone-specific practices.
- 10.3 The repeat sample shall be collected in accordance with the AHS *Newborn Blood Spot Screening Sample Collection* Procedure.
- 10.4 Actions shall be documented by the MRHP in the infant's health record and communicated to the ANSP coordination team.
- 10.5 For additional information, see the ANSP staff education resource *Following Up on Infants Essentials*.

11. Follow-up Actions: Flagged Registrations

11.1 The Alberta Newborn Screening Laboratory shall investigate flagged registrations in accordance with their local service area resources (e.g., laboratory standards operating procedures, job aids, worksheets) in order to validate and ensure flagged registrations are resolved and when necessary corrected.

- a) Registrations may be flagged when a **unique lifetime identifier** requires validation or a temporary identifier is used for an infant within the Alberta Newborn Screening Laboratory.

11.2 Flagged registrations shall be validated by:

- a) cross-checking the infant's registration information in the Person Directory with the infant's demographic information in the laboratory information system.

11.3 Flagged registrations shall be resolved by:

- a) making corrections in the laboratory information system;
- b) contacting Health Information Management for assistance with obtaining information necessary to update temporary identifiers to a Unique Lifetime Identifier;
- c) continuing to use a temporary identifier for the infant; and/or
- d) acknowledging registration as valid for the infant's circumstances.

11.4 Actions shall be documented in accordance with Alberta Newborn Screening Laboratory local service area resources.

12. Monitoring

12.1 The need for follow-up shall be monitored by the ANSP coordination team every business day to ensure appropriate communication and actions are completed.

- a) If follow-up communication (as outlined in Section 3 above) and follow-up actions (as outlined in Sections 4 to 11 above) are not completed or are not completed in a timely manner, a rationale shall be documented in the infant's health record.
- b) Documentation on other sources may be required in accordance with:
 - (i) ANSP coordination team local service area resources (e.g., operational practices); and
 - (ii) Alberta Newborn Screening Laboratory local area resources (e.g., laboratory standard operation procedures, job aids, worksheets).

13. Completion

- 13.1 Follow-up shall be considered complete when one of the following requirements has been met:
- a) a normal, adequate screen result is reported;
 - b) a confirmation of diagnostic outcome is received by the Alberta Newborn Screening Laboratory; or
 - c) a valid reason for not undertaking initial or repeat sample collection is documented in the infant's health record and communicated to the infant's Physician or Midwife.

14. Accommodating Special Circumstances: Additional Information

- 14.1 Infant moved out of Alberta:
- a) If an infant has moved out of Alberta, the new location of residence is determined through attempts to contact the parents or guardians in accordance with the *AHS Newborn Blood Spot Screening: Contacting Parents or Guardians* Guideline.
 - b) When an infant has moved out of Alberta, the need for follow-up of abnormal results shall be communicated by the Alberta Newborn Screening Laboratory to the newborn screening program in the infant's new location of residence.
 - c) When an infant has moved out of Alberta, the need for follow-up of borderline results or inadequate results shall be communicated by zone public health nursing services to:
 - (i) the agency responsible for newborn screening follow-up in the infant's new location of residence; and
 - (ii) the Alberta Newborn Screening Laboratory who shall provide borderline results to the newborn screening program in the infant's new location of residence, if requested.
 - d) When an infant has moved out of Alberta, the need for follow-up of a missing initial screen shall be communicated by zone public health nursing services to the agency responsible for newborn screening follow-up in the infant's new location of residence.
 - e) Actions shall be documented in the infant's health record and communicated to the ANSP coordination team.
 - f) For additional information, see the ANSP staff education resource *Special Situations when Following Up Essentials*.

- 14.2 Parent or guardian does not present infant for sample collection:
- a) Parents or guardians who have not refused sample collection but have not booked and/or not presented for sample collection or repeat sample collection on three (3) separate occasions are considered non-compliant.
 - b) Zone public health nursing services shall notify the infant's Physician or Midwife that the parent or guardian has not booked or did not present for sample collection after three (3) times.
 - c) Actions shall be documented in the infant's health record and communicated to the ANSP coordination team.
 - d) For additional information, see the ANSP staff education resource *Special Situations when Following Up Essentials*.
- 14.3 Repeat sample collection for multiple births:
- a) All infants within multiple birth sets (e.g., twins, triplets) shall have a repeat sample collected if any infant in the multiple birth set has borderline results reported, given that the genetics of one infant in the multiple birth set may affect the screen results of others in the birth set.
 - b) Zone public health nursing services shall coordinate and discuss repeat sample collection with:
 - (i) the parent or guardian in accordance with the AHS *Newborn Blood Spot Screening: Contacting Parents or Guardians* Guideline; or
 - (ii) a NICU, special care nursery or Midwife as appropriate in accordance with zone-specific practices.
 - c) The repeat sample shall be collected in accordance with the AHS *Newborn Blood Spot Screening Sample Collection* Procedure.
 - d) Actions shall be documented in the infants' health records and communicated to the ANSP coordination team.
 - e) For additional information, see the ANSP staff education resource *Special Situations when Following Up Essentials*.

DEFINITIONS

Abnormal result means a screen result that is positive or has been reported as borderline twice for the same condition.

Alberta Newborn Screening Laboratory (formerly the Newborn Metabolic Screening Laboratory) means the Alberta Health Services designated laboratory for newborn blood spot screening located at the University of Alberta Hospital.

Alberta Newborn Screening Program (ANSP) (formerly the Newborn Metabolic Screening Program) means an organized population-based newborn blood spot screening program delivered by Alberta Health Services.

Alberta Newborn Screening Program (formerly the Newborn Metabolic Screening Program) coordination team means the Alberta Health Services designated team within Population, Public and Indigenous Health that integrates, organizes and manages the ANSP and its operations.

Borderline result means a screen result that is neither positive nor negative and requires follow-up through repeat sample collection.

Guardian means, where applicable:

For a Minor:

- a) A guardian as defined by the *Family Law Act*, a divorced parent with custody of the minor, or a person appointed pursuant to a will, personal directive, court order, agreement or by authorization of legislation (e.g. *Child, Youth and Family Enhancement Act*).

For an Adult:

- a) an individual appointed by the Court in accordance with the *Adult Guardianship and Trusteeship Act* to make to make decisions on behalf of the adult patient when the adult patient lacks capacity.

Health record means the Alberta Health Services legal record of the patient's diagnostic, treatment and care information.

Inadequate result means the screen results showed that the sample was not adequate for adequate analysis.

Laboratory information system means a class of software that receives, processes and stores information generated by medical laboratory processes.

Most responsible health practitioner means the health practitioner who has responsibility and accountability for the specific treatment/procedure(s) provided to a patient and who is authorized by Alberta Health Services to perform the duties required to fulfill the delivery of such a treatment/procedure(s) within the scope of their practice.

Notice of birth means the document used to collect personally identifiable information about the mother and her infant or about a stillbirth.

Parent means the guardian of a child with the legal authority to make decisions of behalf of the minor in accordance with the Alberta *Family Law Act*.

Physician refusal within the Alberta Newborn Screening Program means the physician does not allow a sample collection on an infant for clinical reasons (i.e., the infant is medically unstable).

Person Directory means a secure, web-enabled application that delivers person-identifiable demographic and eligibility information to authorized health service providers.

Sample means the blood spots and health information collected on the requisition card for the purpose of newborn blood spot screening.

Sample collection means the process of completing the requisition card and poking the infant's heel to obtain the blood spots for the purpose of newborn blood spot screening.

Screened condition means one or more of the treatable conditions currently screened for within the Alberta Newborn Screening Program.

Transfer means re-assignment or physical re-location to/from a health care setting, service, health care provider or level of care.

Unique Lifetime Identifier (ULI) means a unique and permanent number assigned to all persons who receive health services in Alberta. Unique Lifetime Identifiers are assigned to all Alberta residents, residents of other provinces/territories or other countries.

Zone public health nursing services means the public health nursing services responsible for providing newborn blood spot screening services at the zone level.

REFERENCES

- Alberta Health Services Governance Documents:
 - *Disclosure of Harm Procedure* (#PS-95-01)
 - *Immediate Management of Clinical Adverse Events Procedure* (#PS-95-02)
 - *Newborn Blood Spot Screening Policy* (#HCS-32)
 - *Newborn Blood Spot Screening Contacting Parents or Guardians Guideline* (#HCS-32-03)
 - *Newborn Blood Spot Screening Neonatal Intensive Care Unit Guideline* (#HCS-32-04)
 - *Newborn Blood Spot Screening Sample Collection Procedure* (#HCS-32-02)
 - *Ongoing Management of Clinical Adverse Events Procedure* (#PS-95-03)
 - *Patient Safety Alerts and Safer Practices Notices Procedure* (#PS-95-05)
 - *Patient Safety Learning Summary Procedure* (#PS-95-06)
 - *Recognizing and Responding to Hazards, Close Calls and Clinical Adverse Events Policy* (#PS-95)
- Alberta Health Services Forms:
 - *Alberta Newborn Screening Laboratory Correction Form* (#21409)

- Alberta Health Services Resources:
 - *Following Up on Infant's Essentials*
 - *Following Up Summary*
 - *Alberta Newborn Screening Laboratory Preliminary Results Sheet*
 - *ANSP Condition Fact Sheet*
 - *Special Situations when Following Up Essentials*
- Non-Alberta Health Services Documents:
 - Child, Youth & Family Enhancement Act (Alberta)

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