NEWBORN SCREENING BC

Terms of Reference

Newborn Screening Advisory Committee (NSAC)

APPROVAL

This section contains the signatures of the working group chair(s) or other sponsor(s), indicating approval of the terms of reference. The project manager is the team member in charge of collecting approval for the terms of reference.

SIGNATURES			
Robert Finch	Project sponsor	Dated:	
Ellen Giesbrecht	Project sponsor	Dated:	
Hilary Vallance	Chair	Dated:	

VERSION CONTROL

This section is used to document any changes and serves to control the development and distribution of revisions to the terms of reference. Attach any supporting documentation related to changes in an appendix.

VERSION	STATUS	DATE UPDATED	UPDATED BY	REVISED BY	COMMENTS
V0.1	Under review	09/15/2022	Navi Atwal	Graham Sinclair, Hilary Vallance	Incorporated feedback and proposed changes
V0.2	Draft, under review	11/03/2022	Nazanin Fatemi	Graham Sinclair, Hilary Vallance, Julie MacFarlane	Updated scope, responsibilities, principles and other changes as discussed with the project sponsors
V0.3	Draft, under review	12/02/2022	Nazanin Fatemi	Rob Finch	Incorporated edits from project sponsor
V0.4	Draft	12/12/2022	Nazanin Fatemi	Julie Macfarlane	Incorporated edits
V0.5	Draft final	03/16/2023	Carolyn Ma	Graham Sinclair, Hilary Vallance, Julie MacFarlane	Incorporated last feedback, finalized membership

Newborn Screening Advisory Committee (NSAC)

Terms of Reference

BACKGROUND

Newborn Screening BC (NSBC) facilitates screening to all newborns in British Columbia (B.C.), and Yukon for a group of treatable conditions in order to allow for early intervention and improved health outcomes in children. It is a population-based screening program that provides early diagnosis for a range of treatable conditions. Without newborn screening, affected infants may not otherwise be diagnosed soon enough to prevent serious health problems. Early diagnosis and treatment can make the difference between lifelong impairment and healthy development.

Currently, the Newborn Screening BC (NSBC) program consists of blood spot card screening and point of care biliary atresia screening:

- An updated list of disorders screened by the blood spot card are available at: **Disorders Screened (perinatalservicesbc.ca)**
- Information about biliary atresia screening can be found at: **Biliary Atresia (perinatalservicesbc.ca)**

PURPOSE

The Newborn Screening Advisory Committee (NSAC or Committee) is responsible for providing evidence-based advice and guidance to NSBC for consideration of novel screening tests and strategies, and assessment of ongoing program quality.

SCOPE

In-scope:

- Newborn blood spot screening program in B.C. and Yukon
- Newborn biliary atresia screening program in B.C.
- Consideration of other provincial newborn screening activities

Out of scope:

- Consideration of screening tests that are not for newborns
- Screening tests for newborns born outside of B.C. and Yukon
- Prenatal genetic screening

- Maternal screening

RESPONSIBILITIES

General

- Monitor, evaluate and provide recommendations on key performance indicators for Newborn Screening BC
- Review and approve new standards, policies and guidelines, as applicable
- Establish working groups to regularly review disorders in response to changes in the rare disease landscape, including but not limited to new therapies, biomarkers/genomic technologies and information systems/technologies. *Assumption: the Committee will have appropriate representation and use established screening principles, evaluation tools and evidence-based knowledge.*
- Review and provide recommendations from working groups to NSBC with respect to addition (or removal) of conditions to NSBC
- Advise NSBC on the implementation, modification, and where necessary, the cessation of newborn screening tests
- Provide policy and knowledge translation advice for NSBC programs including but not limited to changes to the testing panel, technologies, screening protocols, consent, storage and access to residual samples or alignment with other relevant screening systems
- Advise on research necessary to enhance understanding of matters related to program effectiveness

Education and Communication

- Advise on educational materials for health care providers and parents on newborn screening
- Advise on the response to questions referred to the Committee by NSBC, the Ministry of Health, or other stakeholders
- Review communication protocols
- Collaborate with regional health authorities and other stakeholder on specific topic areas, as needed. See Appendix A for a diagrammatic representation of the reporting relationships relevant to NSBC and the Committee.

Quality Improvement

- Monitor quality metrics with respect to appropriate blood spot card collection
- Review key performance indicators of the blood spot specimen transportation system
- Review outcomes to ensure screening is available for all newborns in B.C.
- Review quality of procedures for testing and interpretation of test results
- Monitor and review key program performance metrics such as positive predictive value, false positive and false negative rates, participation and timeliness

- Monitor the efficiency and effectiveness of diagnostic testing in follow-up to presumptive positive test results
- In collaboration with clinical programs, evaluate outcomes of treatment in infants and children with disorders detected by the program
- Review and advise on all aspects of the NSBC program effectiveness and quality, including evaluation of performance indicators and related data and the use of audits, as needed
- Identify potential collaborative partners and initiatives to enhance screening and rare disease care for newborns and children in B.C. and Yukon. The Committee may advise NSBC to work with such bodies.

WORKING PRINCIPLES

Consolidated principles for screening based on a systematic review and consensus process (Mark J Dobrow et al)[1] has been chosen as this committee's working principles:

PRINCIPLE	DESCRIPTION
Disease/condition	**1. Epidemiology of the disease or condition** is adequately understood and the disease or condition is an important health problem **2. Natural history of disease or condition** is adequately understood, the disease or condition is well-defined, and there is a detectable preclinical phase **3. Target population for screening** is clearly defined, identifiable and able to be reached
Test/intervention	**4. Screening test performance characteristics** are appropriate for the purpose, with all key component specific to the test being accurate and reliable or reproducible; test should be acceptable to the target population and possible to perform to administer it safely, affordably and efficiently **5. Interpretation of screening test results** such that screening test results are clearly interpretable and determinate to allow identification of the screening participants who should (and should not) be offered diagnostic testing and post-screening care

---

[1] Dobrow MJ et al, Consolidated principles for screening based on a systematic review and consensus process, CMAJ 2018 April 9; 190:E422-9. doi: 10.1503/cmaj.171154

	6. Post-screening test options such that the burden of testing on all participants should be understood and acceptable, and the effect of false-positive and false-negative tests should be minimal
Program/system	**7. Screening program infrastructure** or a clear plan to develop adequate infrastructure is required to allow timely access to all components of the screening program **8. Screening program coordination and integration** with the broader health care system is needed to optimize care continuity and ensure no screening participate is neglected **9. Screening program acceptability and ethics** such that all components of screening program should be clinically, socially and ethically acceptable to screening participants, and there are effective methods for providing screening participants with informed choice, promoting their autonomy and protecting their rights **10. Screening program benefits and harms** and their expected range and magnitude are clearly defined, acceptable and supported by existing high-quality scientific evidence that indicates that the overall benefit of screening program outweighs its potential harms **11. Economic evaluation of screening program** is conducted to assess the full costs and effects of implementing, operating and sustaining the screening program while clearly considering the opportunity costs and effect of allocating resources to other potential non-screening alternatives for managing the disease or condition **12. Screening program quality and performance management** such that the screening program has clear goals and objectives that are explicitly linked to program planning, monitoring, evaluating and reporting activities, with dedicated information systems and funding, to ensure ongoing quality control and achievement of performance targets

MEMBERSHIP

The membership of the Committee should reflect a range of backgrounds and disciplines relevant to the mandate of NSBC. It will be composed of representatives from a variety of health authorities. In order to remain effective and efficient, the number of members will not exceed 15 persons.

1.1. Standing Membership

- Executive Director, Perinatal Services BC
- Provincial Medical Director, Perinatal Services BC
- Screening Director, Perinatal Services BC
- Medical Director, Newborn Screening BC (Chair)

- Director, Newborn blood spot screening laboratory

Representation from:

- Operations, Provincial Laboratory Medicine Services
- Newborn Screening BC Data and Surveillance, Perinatal Services BC
- Ministry of Health, Provincial and Specialized Services division *(see below)*

The role of the representative(s) from the Ministry of Health will be to:

- Receive meeting agendas and minutes, and be invited to attend the annual meeting
- Monitor and ensure appropriate linkages within the government for resolution of significant program recommendations/emerging issues
- Ensure appropriate linkages and liaison with oversight bodies (e.g., Ombudsperson and BC Patient Safety and Quality Council)
- Receive information from and liaise with PHSA Executive representative on significant recommendations/emerging issues

1.2. Rotating Membership and other members as required

- Expert in one of the following: Pediatric endocrinology, cystic fibrosis, hematology, severe combined immunodeficiency disorder (SCID) pediatrician, pediatric neurology
- Biochemical diseases expert
- Medical geneticist
- Neonatologist/pediatrician
- Genetic counsellor/nurse practitioner
- Family physician, midwife

See Appendix B for a list of Committee members.

DISEASE SPECIFIC WORKING GROUPS

There will be Disease Specific Working Groups reflecting the NSBC target diseases that will be formed on an ad-hoc basis. These working groups will report to the Committee and have separate Terms of Reference. A representative of these working groups can attend the Committee meeting(s) on an as-needed basis.

MEETING FREQUENCY, LOCATION, AND ATTENDANCE

- As needed with a minimum of two times per year
- Members are expected to attend all meetings or provide a delegate

DECISION-MAKING AND QUORUM

Decisions will be evidence-based and always be made with the best interests of children and families in mind served by NSBC. The Committee will strive to base its decisions on consensus. If consensus is not possible, the chairperson may call a vote. A simple majority of members present can pass a vote. Decisions are binding and all members will support the decisions and work of NSAC after decisions have been made.

Quorum is set at 50 per cent plus one of membership for decisions.

TERM LIMITS

All Committee members (except Standing Members) will serve an initial term of three years which may be renewed for two renewal terms of three years.

ACCOUNTABILITY AND REPORTING

The committee, via the chair, is responsible for reporting to Perinatal Services BC Steering Committee as required. Agenda will be circulated to all members at least one week before the meeting. Draft minutes will be distributed to the membership for review within two weeks of the meeting.

CONFIDENTIALITY

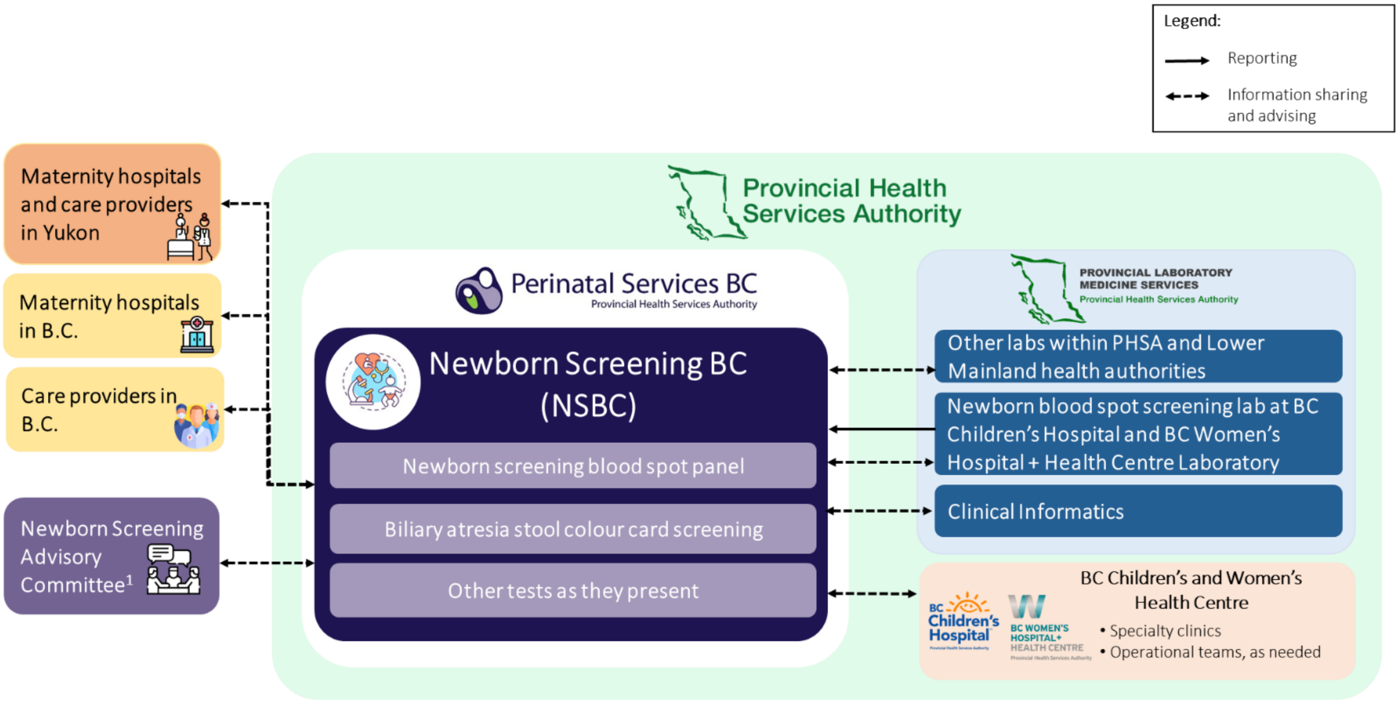
All members will treat information shared in meetings with confidentiality unless the information is to be shared as noted in the meetings.

REVIEW

The Terms of Reference will be reviewed annually by the Committee

Appendices

APPENDIX A: NEWBORN SCREENING BC PROGRAM MODEL



Legend:

⟶ Reporting

⇠⇢ Information sharing and advising

Maternity hospitals and care providers in Yukon

Maternity hospitals in B.C.

Care providers in B.C.

Newborn Screening Advisory Committee[1]

Provincial Health Services Authority

Perinatal Services BC
Provincial Health Services Authority

Newborn Screening BC (NSBC)

- Newborn screening blood spot panel
- Biliary atresia stool colour card screening
- Other tests as they present

PROVINCIAL LABORATORY MEDICINE SERVICES
Provincial Health Services Authority

- Other labs within PHSA and Lower Mainland health authorities
- Newborn blood spot screening lab at BC Children's Hospital and BC Women's Hospital + Health Centre Laboratory
- Clinical Informatics

BC Children's and Women's Health Centre

- Specialty clinics
- Operational teams, as needed

---

[1] Comprised of subject matter experts, operational leads, disease specific ad-hoc working groups, when applicable; Chair: Newborn Screening BC Medical Director

APPENDIX B: MEMBERSHIP OF NSAC

Standing Membership

Representative of	Member name	Title
Perinatal Services BC	Robert Finch	Executive Director, Perinatal Services BC
	Ellen Giesbrecht	Provincial Medical Director, Perinatal Services BC
	Julie Macfarlane	Screening Director, Perinatal Services BC
	Hilary Vallance	Medical Director, Newborn Screening BC (Chair)
Provincial Laboratory Medicine Services	Graham Sinclair	Director, Newborn blood spot screening laboratory
Operations, Provincial Laboratory Medicine Services	Brenda Jackson	Senior Executive Director, Clinical Laboratory Operations
Newborn Screening BC Data and Surveillance, Perinatal Services BC	Kenny Der, or Kathryn Berry-Einarson	Director, Informatics Director, Quality and Research, Perinatal Services BC
Ministry of Health		Provincial and Specialized Services division

Rotating Membership

Representative of	Member name	Title
Expert in Cystic Fibrosis Pediatrician	Mark Chilvers	Medical Director of CF Care BC Medical Director, Child Health BC CF Clinic Director Pediatric Respirologist, BC Children's & BC Women's Hospital
Biochemical Diseases	Ramona Salvarinova	Division Head, Biochemical Diseases, Department of Pediatrics, BC Children's & Women's Hospital
Medical Geneticist	Laura Arbour	Professor, UBC Department of Medical Genetics Clinical Geneticist, Victoria, Island Health
Genetic Counsellor	Gurdip Hulait	Genetic Counsellor, Newborn Screening Laboratory, BC Children's & BC Women's Hospital

Family Physician	Melissa Gillis	Family Physician, Nanaimo, Island Health
Midwife	Dina Davidson	Registered Midwife and Lactation Consultant, Port Moody, Fraser Health
Pediatrician	Anamaria Richardson	Pediatrician, Vancouver, Vancouver Coastal Health