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Newborn Screening Advisory Committee for British Columbia and the Yukon

Terms of Reference

Updated Sept 20, 2010

Background

Newborn screening is a population-based screening program that provides early diagnosis for a range of treatable disorders. Without newborn screening, affected infants may not otherwise be diagnosed soon enough to prevent serious health problems, including global developmental delay, blindness, liver problems, or even death. Early diagnosis and treatment can make the difference between lifelong impairment and healthy development. See Appendix 1 for a listing of disorders screened.

Responsibilities of the Newborn Screening Advisory Committee

General

1. Establish newborn screening standards, policies and guidelines.

2. Regularly review disorders for which infants could potentially be screened according to an agreed upon framework. Assumption: the committee will have appropriate representation and use established screening principles and evaluation tools.

3. Monitor, review and respond in a timely manner to *emerging* clinical evidence and research in newborn screening.

4. Advise the program on the implementation, modification, and where necessary, the cessation of newborn screening tests.

5. Advise on screening algorithms, referral, and care pathways for each of the disorders.

6. Maintain linkages and communication with newborn screening programs across Canada.

Education and communication:

7. Advise on educational materials for health care providers and parents on newborn screening.

Quality improvement:

8. Establish standards and review outcomes with respect to appropriate blood spot card collection and the transportation system.

9. Establish and maintain surveillance standards and processes (to ensure screening is available for all newborns in BC and the Yukon).

10. Review quality of procedures for testing and interpretation of test results.

11. Track false positive and false negative rates.

12. Review communication protocols.

13. Identify outcome parameters to assess the efficiency and effectiveness of diagnostic testing in follow-up to presumptive positive test results.

14. Track the outcome of treatment of infants and children with disorders detected by the program.

Reporting Relationships:

The Newborn Screening Program and the Newborn Screening Advisory Committee (NSAC) report to the PHSA Executive through the Provincial Executive Director, Perinatal Services BC.

The NSAC will collaborate with regional health authorities and other stakeholders on specific topic areas.

See Appendix 2 for a diagrammatic representation of the reporting relationships relevant to the Newborn Screening Program and Advisory Committee.

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Membership:

Committee members will rotate on a 3 year term (assuming an alternative is available).

Representation	Participants
• Expert in lab medicine related to NBS	Dr Hilary Vallance Dr Graham Sinclair
• Expert in metabolic diseases	Dr Sandra Sirrs Dr Sylvia Stockler
• Expert in one of the following: pediatric endocrinology, CF, hematology	Dr Mark Chilvers
• Expert in genetics	Dr Lorne Clarke Dr Laura Arbour David Koehn
• Neonatologist/pediatrician/FP physician	Dr John Smyth
• Representative from Perinatal Services BC / PHSA	KimWilliams

Corresponding Members (receive agenda + minutes; attend as able):

Representation	Proposed participants
• Representative from Ministry of Health Services (MOHS)	Richele Shorter
• Representative from Ministry of Healthy Living and Sport (MHLS)	Carla Springinotic

The role of the representatives from the MOHS and MHLS is to:

- 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. Ombudsman and Patient Safety and Quality Council).
- Receive information from and liaise with PHSA Executive representative on significant recommendations/emerging issues.

Meeting Frequency:

Meetings will be held 2 – 3 times per year and at the call of the chair.

Much of the detailed work will be done through working groups which will report to the Newborn Screening Advisory Committee.

Other

Terms of reference will be reviewed annually by the committee.

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Appendix 1
Summary of Newborn Screening Tests

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	CbI A, B
		Glutaric Aciduria, Type I	GA I
		Isovaleric Acidemia	IVA
Galactosemia	GALT enzyme activity		GALT
Endocrine Disorders	Thyroid stimulating hormone (TSH)	Congenital Hypothyroidism	CH
	17OH-progesterone (1st tier) Steroid panel (2nd tier)	Congenital Adrenal Hyperplasia	CAH
Hemoglob-inopathies	Hemoglobin HPLC	Sickle Cell Disease	HBSS
		Sickle Cell/Hemoglobin C	HSc
		Sickle Cell/β-thalassemia	Hb S/β-thal
Cystic Fibrosis	Immunoreactive trypsinogen (IRT) (1st tier) CFTR mutation panel (2nd tier)		CF

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Appendix 2
Reporting Relationships of the Newborn Screening Program and Advisory Committee