



Newborn Screening in Canada Status Report – Page 1 of 3 - updated Jan. 13, 2012

¹Follows format used by the U.S. National Newborn Screening & Genetics Resource Center

A dot "●" indicates that universal screening for the condition is required by law

A = universally offered but not yet required, **B** = offered to select populations or by request, **C** = testing required or offered universally but not yet implemented, **D** = likely to be detected (and reported) as a by-product of MRM screening (MS/MS) targeted by law

For questions or comments about this report, contact John Adams at john.adams@canpku.org

Province/Territory (east to west/ west to east)	Core ¹ Conditions:											Additional Conditions Included (universally offered unless otherwise indicated)	
	Endocrine			Hemoglobin			Others						
	HEAR	CH	CAH	S/S/S/S	S/C	S/A	BIO	GALT	CF	CCHD	SCID		
Newfoundland & Labrador	B	A											
Prince Edward Isl.	A	A											Covered by NBS Lab in Nova Scotia
Nova Scotia	A	A											CF and HB pilot under consideration
New Brunswick	A	A											collaborate with Nova Scotia; PKU in NB
Quebec	B	A											Universal hearing announced 2010 but not implemented; Bloodspot expansion under review; 2nd screen by urine collected by parent at newborn Day 21: 12 conditions of amino, urea cycle & organic acids & 11 transport disorders of amino acids (Fanconi syndrome, Cystinurias, Hartnup syndrome, Cystathioninemia, Prolidase deficiency, etc.)
Ontario	A	A	A	A	A	A	A	A	A				SCID under consideration
Manitoba	B	A	A				A	A	A				targeted screening GA1 (Oji-Cree), CPT1 (Hutterites) Plan to add MS/MS in 2007
Saskatchewan	B	●	C				C	B/C	C				Plan to add BIO, GALT, CAH & CF in 2007 Q2
Alberta	B	A	A				A		A				Pilot hearing screening
British Columbia	A	A	A	A	A	A		A	A				
Yukon	B												Covered by NBS lab British Columbia; hearing screen only available in Whitehorse
Northwest Territories	B	A					A						Covered by NBS lab in Alberta, hearing screen available only in Yellowknife
Nunavut - Kitimeot region		A					A		A				Covered by NBS lab in Alberta
Nunavut - Kivilliq region	B	A	A				A	A					Covered by NBS lab in Manitoba
Nunavut - Baffin region	B	A											Covered by NBS labs in Quebec; Second screen by urine - 23 conditions

¹Terminology consistent with American College of Medical Genetics report - Newborn Screening: Toward a Uniform Screening Panel and System 2005, p. 63.

Deficiency/Disorder Abbreviations and Names

BIO	Biotinidase	CF	Cystic fibrosis	GALT	Transferase deficient galactosemia (Classical)	HB S/C	Sickle – C disease	HEAR	Hearing screening
CAH	Congenital adrenal hyperplasia	CH	Congenital hypothyroidism	HB S/S	Sickle cell anemia	HB S/A	S-beta thalassemia	SCID	Severe Combined Immunodeficiency
CCHD	Critical Congenital Heart Disease								

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Province/Territory (east to west/ west to east)	Core ¹ Conditions: Metabolic																			
	Fatty Acid Disorders					Organic Acid Disorders									Amino Acid Disorders					
	CUD	LCHAD	MCAD	TFP	VLCAD	GA-I	HMG	IVA	3-MCC	Cbl-A,B	BKT	MUT	PROP	MCD	ASA	CIT	HCY	MSUD	PKU	TYR-I
Newfoundland & Labrador			A														A		A	A
Prince Edward Island	A	A	A	A	A	A		A										A	A	
Nova Scotia	A	A	A	A	A	A		A										A	A	
New Brunswick:	A	A	A	A	A	A		A										A	A	
Quebec						A	A	A	A	A		A		A	A				A	A
Ontario	A	A	A	A	A	A	A	A	A	A	A	A	A	A	A	A	A	A	A	A
Manitoba						B													A	
Saskatchewan		A	A	A	A	A	A	A	A	A	A	A	A	A	A	A	A	A	●	A
Alberta	A	A	A	A	A	A	A	A		A		A			A			A	A	A
British Columbia		A	A	A	A	A		A		A		A		A	A			A	A	A
Yukon		A	A	A	A	A		A		A		A		A	A			A	A	A
Northwest Territories																			A	
Nunavut - Kitimeot region																			A	
Nunavut - Kivilliq region																			A	
Nunavut – Baffin region						A	A	A	A	A		A		A	A				A	A

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Deficiency/Disorder Abbreviations and Names (optional nomenclature)

3-MCC	3-Methylcrotonyl-CoA carboxylase	CUD	Carnitine uptake defect (Carnitine transport defect)	LCHAD	Long-chain hydroxyacyl-CoA dehydrogenase	PKU	Phenylketonuria/hyperphenylalaninemia
ASA	Argininosuccinate acidemia	GA-I	Glutaric acidemia type 1	MCAD	Medium-chain acyl-CoA dehydrogenase	PROP	Propionic acidemia (Propionyl-CoA carboxylase)
BKT	Beta ketothiolase (mitochondrial acetoacetyl-CoA thiolase ; short-chain ketoacyl thiolase; T2)	HCY	Homocystinuria (cystathionine beta synthase)	MCD	Multiple carboxylase (Holocarboxylase synthetase)	TFP	Trifunctional protein
CBL A,B	Methylmalonic acidemia (Vitamin B12 Disorders)	HMG	3-Hydroxy 3 - methylglutaric aciduria (3-Hydrox 3- methylglutaryl-CoA lyase)	MSUD	Maple syrup urine disease (branched-chain ketoacid dehydrogenase)	TYR-I	Tyrosinemia Type 1
CIT I	Citrullinemia type I (Argininosuccinate synthetase)	IVA	Isovaleric acidemia (Isovaleryl-CoA dehydrogenase)	MUT	Methylmalonic Acidemia (methylmalonyl-CoA mutase)	VLCAD	Very long-chain acyl-CoA dehydrogenase

