



Newborn Screening in Canada Status Report – Page 1 of 3 - updated Dec. 15, 2009

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)	
	Hearing	Endocrine		Hemoglobin (HB)			Other				
	HEAR	CH	CAH	S/S	S/C	S/A	BIO	GALT	CF		
Newfoundland & Labrador		A									
Prince Edward Island	A	A									Covered by NBS Lab in Nova Scotia
Nova Scotia		A									HB pilot under consideration; discussing CF
New Brunswick	A	A									collaboration with Nova Scotia; maintain PKU in NB
Quebec	B	A									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	A	ACMG core panel of 29 disorders
Manitoba	B	A	A				A	A			targeted screening for GA1 (Oji-Cree), CPT1 (Hutterites) and Duchenne muscular dystrophy (males) Plan to add MS/MS in 2007
Saskatchewan	B	●	C				C	B / C	C		Plan to add Biotinidase (BIO), Galactosemia (GALT), Congenital Adrenal Hyperplasia (CAH) and Cystic Fibrosis in 2007 Q2
Alberta	B	A	A				A			A	Pilot for universal hearing screening in 4 regions (Calgary, Chinook, Mistahia & Palliser)
British Columbia	C	A	C	A	A	A		A	A		Expansion 11/ 2009 including 2 nd tier test for HCY, MSUD, PA, MMA, CBL A/B,, add CAH 10/2010; universal hearing screening by end 2008
Yukon	A	A	C	A	A	A		A	A		Covered by NBS lab in British Columbia
Northwest Territories	B	A					A				Covered by NBS lab in Alberta
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 sample at newborn Day 21 – 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 hyperthyroidism	HB S/S	Sickle cell disease	HB S/A	S-beta thalassemia		

Other Disorders

5-OXO	5-oxoprolinuria (pyroglutamic aciduria)	G6PD	Glucose 6 phosphate dehydrogenase	NKH	Nonketotic hyperglycinemia
CPS	Carbamoylphosphate synthetase	HHH	Hyperammonemia/ornithinemia/ citrullinemia (Ornithine transporter defect)	OTC	Ornithine transcarbamylse
EMA	Ethylmalonic encephalopathy	HIV	Human immunodeficiency virus	TOXO	Toxoplasmosis

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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-1	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	A	A
British Columbia		A	A	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	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-1	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-1	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

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Province/ Territory (east to west; west to east)	Secondary Target ¹ Conditions																								
	Fatty Acid Disorders							Organic Acid Disorders					Amino Acid Disorders							Other Metabolic		Hgb			
	CACT	CPT-Ia	CPT-II	DE-RED.	GA-II	MCKAT	M/SCHAD	SCAD	2M3HBA	2MBG	3MGA	Cbl-C,D	IBG	MAL	ARG	BIOPT-BS	BIOPT-REG	CIT-II	H-PHE	MET	TYR-II	TYR-III	GALE	GALK	Variant hemoglobins
Nfld. & Labrador																									
Prince Edward Island	A	A	A		A																A				
Nova Scotia	A	A	A		A																				
New Brunswick	A	A	A		A																				
Quebec											A			A			A								
Ontario																									
Manitoba		B																							
Sask.	A	A	A	B	A		A		A	A	A		A	A	B	B	A	A	A	A		B	B	B	
Alberta																									
British Columbia					A				A		A				A	A	A	A	A						
Yukon					A				A		A				A	A	A	A	A						
Northwest T.																									
Nunavut - Kitimeot																									
Nunavut - Kivilliq																									
Nunavut - Baffin												A		A			A								

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Deficiency/Disorder Abbreviations and Names (optional names)

2M3HBA	2-Methyl-3-hydroxy butyric aciduria	CACT	Carnitine acylcarnitine translocase	GA-II	Glutaric acidemia Type II	MAL	Malonic acidemia (Malonyl-CoA decarboxylase)
2MBG	2-Methylbutyryl-CoA dehydrogenase	CBL-C,D	Methylmalonic acidemia (Cbl C,D)	GALE	Galactose epimerase	MCKAT	Medium-chain ketoacyl-CoA thiolase
3MGA	3-Methylglutaconic aciduria	CIT-II	Citrullinemia type II	GALK	Galactokinase	MET	Hypermethioninemia
ARG	Arginemia (Arginase deficiency)	CPT-Ia	Carnitine palmitoyltransferase I	H-PHE	Benign hyperphenylalaninemia	SCAD	Short-chain acyl-CoA dehydrogenase
BIOPT-BS	Defects of biotin cofactor biosynthesis	CPT-II	Carnitine palmitoyltransferase II	IBG	Isobutyryl-CoA dehydrogenase	TYR-II	Tyrosinemia type II
BIOPT-REG	Defects of biotin cofactor regeneration	De-Red	Dienoly-CoA reductase	M/SCHAD	Medium/Short chain L-3-hydroxy acyl-CoA dehydrogenase	TYR-III	Tyrosinemia type III