Newborn Screening in Canada Status Report – Page 1 of 3 - updated July 3, 2008

[1]For consistency this report follows 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

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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; second screen by urine collected by parent at newborn Day 21 – 90% participation: 12 conditions of amino, urea cycle & organic acids plus 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	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 Biotindase (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	C	C	C		A	C	expansion announced July 2008 – fully implemented by 2010; universal hearing screening by end 2008
Yukon	A	A	C	C	C	C		A	C	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

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-βeta 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-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	A
British Columbia		A	A	C	C	A		C		C		C	C		C	C	C	C	A	C
Yukon		A	A	C	C	A		C		C		C	C		C	C	C	C	A	C
Northwest Territories - western																			A	
Northwest Territories - eastern																			A	
Nunavut - western																			A	
Nunavut – eastern						A	A	A	A	A		A			A	A			A	A

[1]Terminology 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 (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		Hbg
	CACT	CPT-Ia	CPT- II	DE-RED.	GA-II	MCKAT	M/SCHAD	SC AD	2M3HBA	2MBG	3MGA	Cbl-C,D	IBG	MAL	ARG	BIOPT-BS	BIOPT-RG	CIT-II	H-PHE	MET	TYR- II	TYR- III	GALE	GALK	Variant hemoglobins
Nfld. & Labrador																					A				
Prince Edward Island	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					C					C		C				A	A	C	A						
Yukon					C					C		C				A	A	C	A						
Northwest T. - western																									
Northwest T. - eastern																									
Nunavut - western																									
Nunavut - eastern												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-Methylbutyrly-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 biopterin cofactor biosynthesis	**CPT-II**	Carnitine palmitoyltransferase II	**IBG**	Isobutyryl-CoA dehydrogenase	**TYR-II**	Tyrosinemia type II
BIOPT-REG	Defects of biopterin cofactor regeneration	**De-Red**	Dienoly-CoA reductase	**M/SCHAD**	Medium/Short chain L-3-hydroxy acyl-CoA dehydrogenase	**TYR-III**	Tyrosinemia type III