Newborn Screening in Canada Status Report – Page 1 of 3 – updated May 5, 2012

[1]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)
		Endocrine		Hemoglobin (HB)			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 NS; 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 2011
Manitoba	B	A	A	B	B	B	A	A	A			targeted molecular screening GA1 variant (Oji-Cree), CPT1 (Hutterites)
Saskatchewan	B	●	C				A	A	A			
Alberta	B	A	A				A		A			Pilot hearing screening
British Columbia	A	A	A	A	A	A		A	A			
Yukon	B	A	C	A	A	A		A	A			Covered by NBS lab in BC; hearing screen available in Whitehorse only
Northwest Territories	B	A					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

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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-βeta 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	A
Ontario	A	A	A	A	A	A	A	A	A	A	A	A	A	A	A	A	A	A	A	A
Manitoba	A	A	A	A	A	A&B	A	A	A	A	A	A	A	A	A	A	A	A	A	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	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	A	A	A	A	A	A	A		A			A			A	A	A	A	A
Nunavut - Kitimeot region	A	A	A	A	A	A	A	A		A			A			A	A	A	A	A
Nunavut - Kivilliq region	A	A	A	A	A	A	A	A	A	A	A	A	A	A	A	A	A	A	A	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		Hbg
	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-RG	CIT-II	H-PHE	MET	TYR-II	TYR-III	GALE	GALK	Variant hemoglobins
Nfld. & Labrador																					A				
Prince Edward Is.	A	A	A		A																				
Nova Scotia	A	A	A		A																				
New Brunswick	A	A	A		A																				
Quebec												A			A			A							
Ontario																									
Manitoba	A	A	A	A	A	A	A	A	A	A	A	A	A	A	A	A	A	A	A	A	A	A		A	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																									
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-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