

<sup>1</sup>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

For questions or comments about this report, contact John Adams at [john.adams@canpku.org](mailto:john.adams@canpku.org)

Province/Territory (east to west/ west to east)	Core <sup>1</sup> 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									regional 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	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); CF universal pilot since May, 2005 in Calgary Region; C = 13 to be universally offered by April 2007
British Columbia	C	A						A			Expansion under review; universal hearing screening by end 2008
Yukon	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

### Deficiency/Disorder Abbreviations and Names

<b>BIO</b>	Biotinidase	<b>CF</b>	Cystic fibrosis	<b>GALT</b>	Transferase deficient galactosemia (Classical)	<b>HB S/C</b>	Sickle – C disease	<b>HEAR</b>	Hearing screening
<b>CAH</b>	Congenital adrenal hyperplasia	<b>CH</b>	Congenital hyperthyroidism	<b>HB S/S</b>	Sickle cell disease	<b>HB S/A</b>	S-βeta thalassemia		

### Other Disorders

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

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Province/Territory (east to west/ west to east)	Core <sup>1</sup> 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
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	
Yukon			A																A	
Northwest Territories - western																			A	
Northwest Territories - eastern																			A	
Nunavut - western																			A	
Nunavut - eastern						A	A	A	A		A			A	A				A	A

<sup>1</sup>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)**

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

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Province/ Territory (east to west; west to east)	Secondary Target <sup>1</sup> 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-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	B	B	A	A	A	A		B	B	B
Alberta																									
British Columbia																									
Yukon																									
Northwest T. - western																									
Northwest T. - eastern																									
Nunavut - western																									
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**Deficiency/Disorder Abbreviations and Names (optional names)**

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