

For consistency this report follows the format used by the U.S. National Newborn Screening & Genetics Resource Center

Dot "●" indicates that screening for the condition is universally 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

Questions regarding the Canada Status Report should be directed to John Adams at [john.adams@canpku.org](mailto:john.adams@canpku.org)

Province/Territory	Core <sup>1</sup> Conditions:									Additional Conditions Included (universally offered unless otherwise indicated)
	Hearing	Endocrine		Hemoglobin (HB)			Other			
	HEAR	CH	CAH	S/S	S/A	S/C	BIO	GALT	CF	
<b>Province</b>										
Alberta	B	A	A				A		A	
British Columbia	C	C		C	C	C		A	C	
Manitoba	B	A	A				A	A		DMD (B) ( males)
New Brunswick	A	A								
Newfoundland & Labrador		A								
Nova Scotia		A								
Ontario	A	A	A	A	A	A	A	A	A	
Prince Edward Island	A	A								
Quebec	B	A								Amino acid transport disorders <sup>2</sup>
Saskatchewan	B	●	C				C	B/C	C	
<b>Territory</b>										
Northwest Territories	B	A	A				A		A	
Nunavut - Kitikmeot region		A	A				A		A	
Nunavut - Kivalliq region	B	A	A					A		
Nunavut - Baffin region	B	A								
Yukon	A	A	C	C	C	C		A	C	

<sup>1</sup>Terminology consistent with ACMG report - Newborn Screening: Towards a Uniform Screening Panel and System. Genetics in Medicine. 2006; 8( Supp 1): S32-S33.

<sup>2</sup>Including Fanconi syndrome, Cystinurias, Hartnup syndrome, Prolidase deficiency, etc.

**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-beta thalassemia		

**Additional Disorders**

<b>DMD</b>	Duchenne Muscular Dystrophy

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Province/Territory	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
<b>Province</b>																				
Alberta	A	A	A	A	A	A	A	A	A				A			A	A	A	A	A
British Columbia		A	A	C	C			C		C		C	C		C	C	C	C	A	C
Manitoba						B													A	
New Brunswick	A	A	A	A	A	A		A										A	A	
Newfoundland & Labrador			A														A		A	A
Nova Scotia	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
Prince Edward Island	A	A	A	A	A	A		A										A	A	
Quebec						A	A	A	A	A		A			A	A			A	A
Saskatchewan	B	A	A	A	A	A	A	A	A	A	A	A	A	A	A	A	A	A	●	A
<b>Territory</b>																				
Northwest Territories	A	A	A	A	A	A	A	A		A			A			A	A	A	A	A
Nunavut - Kitikmeot region	A	A	A	A	A	A	A	A		A			A			A	A	A	A	A
Nunavut - Kivalliq region						B													A	
Nunavut - Baffin region						A	A	A	A	A		A			A	A			A	A
Yukon		A	A	C	C			C		C		C	C		C	C	C	C	A	C

<sup>1</sup>Terminology consistent with ACMG report - Newborn Screening: Towards a Uniform Screening Panel and System. Genetics in Medicine. 2006; 8( Supp 1): S32-S33.

**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	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	SC AD	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
<b>Province</b>																									
Alberta																									
British Columbia					C				C		C				A	A	C	A							
Manitoba		B																							
New Brunswick	A	A	A		A																				
Newfoundland & Labrador																				A					
Nova Scotia	A	A	A		A																				
Ontario																									
Prince Edward Island	A	A	A		A																				
Quebec											A			A			A								
Saskatchewan	A	A	A	B	A		A	A	A	A	A		A	A	B	B	A	A	A	A			B	B	B
<b>Territory</b>																									
Northwest Territories																									
Nunavut - Kitikmeot																									
Nunavut - Kivalliq		B																							
Nunavut - Baffin											A			A			A								
Yukon					C				C		C				A	A	C	A							

<sup>1</sup>Terminology consistent with ACMG report - Newborn Screening: Towards a Uniform Screening Panel and System. Genetics in Medicine. 2006; 8( Supp 1): S32-S33.

**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 biotin 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 biotin cofactor regeneration	<b>De-Red</b>	Dienoyl-CoA reductase	<b>M/SCHAD</b>	Medium/Short chain L-3-hydroxy acyl-CoA dehydrogenase	<b>TYR-III</b>	Tyrosinemia type III