

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

Province/Territory	Core ¹ 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	
Province										
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 ²
Saskatchewan	B	●	C				C	B/C	C	
Territory										
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	

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

²Including Fanconi syndrome, Cystinurias, Hartnup syndrome, Prolidase deficiency, etc.

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		

Additional Disorders

DMD	Duchenne Muscular Dystrophy

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Province/Territory	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
Province																				
Alberta	A	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
Territory																				
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

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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	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-REG	CIT-II	H-PHE	MET	TYR-II	TYR-III	GALE	GALK	Variant hemoglobins
Province																									
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
Territory																									
Northwest Territories																									
Nunavut - Kitikmeot																									
Nunavut - Kivalliq		B																							
Nunavut - Baffin											A			A			A								
Yukon					C				C		C				A	A	C	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	Dienoyl-CoA reductase	M/SCHAD	Medium/Short chain L-3-hydroxy acyl-CoA dehydrogenase	TYR-III	Tyrosinemia type III