National Newborn Screening and Genetics Resource Center

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	

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

[1]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)

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	SC AD	2M3HBA	2MBG	3MGA	CbI-C,D	IBG	MAL	ARG	BIOPT-BS	BIOPT-RG	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 - Kivallig		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 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**	Dienoyl-CoA reductase	**M/SCHAD**	Medium/Short chain L-3-hydroxy acyl-CoA dehydrogenase	**TYR-III**	Tyrosinemia type III