

Newborn Screening in Canada Status Report – Page 1 of 3 – updated September 3, 2015

¹Follows 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

U - screened by urine

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Province/Territory						Additional Conditions Included						
(east to west/		End	ocrine	Hemog			Condi		Others		(universally offered unless otherwise	
west to east)	HEAR	СН	САН	S/S S/S	S/C	S/A	BIO	GALT	CF	CCHD	SCID	indicated)
Newfoundland &												
Labrador	В	Α							A			
Prince Edward												Part of Maritime NBS program;
Island	A	Α		A	A	A	В	В	A		В	SCID, BIO & GALT active planning
Nova Scotia												Part of Maritime NBS program;
	A	Α		A	A	A	В	В	A		В	SCID, BIO & GALT active planning
New Brunswick												Part of Maritime NBS program;
	A	A		A	A	A	В	В	Α		В	SCID, BIO & GALT active planning
Quebec												Hearing announced 2010, HB
Q												announced 2012; PKU, CH, TYR &
												MCAD by bloodspot; 2nd screen by
	_				_	_						urine collected by parent at newborn
	В	Α		В	В	В						Day of Life 21 – see note 1
Ontario	A	A	A	A	A	A	A	A	Α		A	
Manitoba												targeted molecular screening GA1
	В	A	A	В	В	В	A	A	A			variant (Oji-Cree)
Saskatchewan	В	•	C				A	A	A			
Alberta												Pilot hearing screening; evaluating
	В	A	A				A		A			HGbs, SCID< GALT & TYR
British Columbia												In 2015 completing pilot for GAMT
												(guanidinoacetate methyltransferase)
	A	A	A	A	A	A		A	A			Funding review: SCID, CUD & BIO
Yukon												Covered by NBS lab in BC; hearing
	В	A	C	A	A	A		A	A			screen available in Whitehorse only
Northwest												Covered by NBS lab in Alberta,
Territories												hearing screen available only in
	В	Α					Α		A			Yellowknife
Nunavut												Covered by NBS lab in Alberta
- Kitimeot region		Α					A		A			Covered by 11DS lab in Alberta
Nunavut												Covered by NBS lab in Manitoba
- Kivilliq region	В	A	A				A	A				Covered by 14DS lab in Maintoba
Nunavut												
- Baffin region												Covered by NBS lab in ON
	В	A	A	A	A	A	A	A	A		A	

¹Terminology 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

BIO	Biotinidase	CF	Cystic fibrosis	GALT	Transferase deficient galactosemia (Classical)	HB S/C	Sickle – C disease	HEAR	Hearing screening
САН	Congenital adrenal hyperplasia	СН	Congenital hypothyroidism	HB S/S	Sickle cell anemia	HB S/A	S-βeta thalassemia	SCID	Severe Combined Immunodeficiency
CCHD	Critical Congenital He	eart Dis	ease						

Note 1: Quebec Mass Urine Screening covers additional conditions: 12 conditions of amino acids, urea cycle & organic acids & transport disorders of amino acids (Cystinuria, Dicarboxylic aminoaciduria, Fanconi syndrome, Hartnup syndrome, Hyperoxaluri, Prolidase deficiency, and Pyroglutamic aciduria. Other conditions reported on pages 2 and 3 of this report.



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							Co	re ¹ (Cond	ition	s: N	Ietabo	lic							
Province/Territory		Fatty Acid Disorders					0	rgan	ic Ac	cid D	isoro	lers			Amino Acid Disorders					ers
(east to west/ west to east)	CUD	LCHAD	MCAD	TFP	VLCAD	GA-I	HMG	IVA	3-MCC	Cbl-A,B	BKT	MUT	PROP	MCD	ASA	CIT	нсу	MSUD	PKU	TYR-1
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			U	U	U	U	U		U	U		U	U			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	Α
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
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	A	A	A	A	A	A	A	A	A	A

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Deficiency/Disorder Abbreviations and Names (optional nomenclature)

3-МСС	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)
вкт	Beta ketothiolase (mitochondrial acetoacetyl-CoA thiolase; short-chain ketoacyl thiolase; T2)	НСҮ	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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										S	Secon	dary	Tar	get 1	Con	ditio	18								
Province/	Fatty Acid Disorders								Oı	Organic Acid Disorders					Amino Acid Disorders							Other Metabolic		Hbg	
Territory (east to west; west to east)	CACT	CPT-Ia	CPT-II	DE-RED.	GA-II	MCKAT	M/SCHA D	SCAD	2М3НВА	2MBG	3MGA	Cbl-C,D	IBG	MAL	ARG	BIOPT- BS	BIOPT- RG	CIT-II	н-рне	MET	TYR- II	TYR- III	GALE	GALK	Variant hemoglob ins
Nfld. & Labrador																					A				
Prince Edward Is.	A	A	A		A																				
Nova Scotia	A	A	A		A																				
New																									
Brunswick	A	A	A		A																				
Quebec												U			U			U							
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	В
Sask.	A	A	A	В	A			A		A	A	A		A	A	В	В	A	A	A	A		В	В	В
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																									

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Deficiency/Disorder Abbreviations and Names (optional names)

2М3НВА	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	Н-РНЕ	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