Carrier Screening in Canada: Counselling Aid

In 2016, the Society of Obstetrics and Gynecology Canada (SOGC) and the Canadian College of Medical Geneticists (CCMG) published a joint opinion titled, *Joint SOGC-CCMG Opinion for Reproductive Genetic Carrier Screening: An Update for All Canadian Providers of Maternity and Reproductive Healthcare in the Era of Direct-to-Consumer Testing*. Two key recommendations were: (1) "A primary discussion about the value and risk of reproductive carrier screening should be offered to all women/families considering a pregnancy (pre-conception) and to all pregnant women at their first prenatal visit [...]." (2) "The option of [Expanded Carrier Screening] should be part of the informed consent process." In addition, this table summarizes the SOGC-CCMG recommendations for ethnicity-based carrier screening:

Condition Name	Available with LifeLabs	Associated Gene(s) †	Any Ethnicity	Ashkenazi Jewish	African	Mediterranian	Southeast Asian	French Canadian (SLSJ+C)‡	French Canadian (BSL, Gasp, NB)‡	Aboriginal Manitoba	Newfoundland	Cree
Cystic Fibrosis	X	CFTR	F	F				X				
Spinal Muscular Atrophy	X	SMN1	F									
Tay-Sachs Disease	X	HEXA		X				F	X			
Canavan Disease	X	ASPA		X								
Familial Dysautonomia	X	IKBKAP		X								
Bloom Syndrome	X	BLM		F								
Gaucher Disease	X	GBA		F								
Fanconi Anemia, Type C	X	FANCC		F								
Mucolipidosis IV	X	MCOLN1		F								
Niemann Pick Disease	X	NPC2		F								
Glycogen Storage Disease, Type 1a	X	G6PC		F								
Familial Hyperinsulinism	X	KCNJ11		F								
Maple Syrup Urine Disease, Type 1B	X	BCKDHB		F								
Dihydrolipoamide Dehydrogenase Deficiency	X	DLD		F								
Usher Syndrome, Type 3	X	CLRN1		F								
Nemaline Myopathy, Type 2	X	NEB		F								
Joubert Syndrome, Type II	X	TMEM216		F								
Walker-Warburg	X	FKTN		F								
Sickle Cell Disease	X	HBB			X	X	X					
Alpha Thalassemia	X	HBA1/HBA2			X	X	X					
Fragile X Syndrome	X	FMR1	F									
X-linked Hemophilia (Hemophilia B)	X	F9	F									
Tyrosinemia, Type 1	X	FAH						X				
Leigh Syndrome, French Canadian Type	X	LRPPRC						X				
Spastic Ataxia, Charlevoix Saquenay Type (ARSACS)	X	SACS						X				
Andermann Syndrome	X	SC12A6						X				
COFS Syndrome (Cockayne Syndrome Type II)	X	ERCC6								F		
Congenital Disorder of Glycosylation, Type 1B	X	MPI						F				
Mucolipidosis II	X	GNPTAB						F				
Neuronal Ceroid-Lipofuscinosis	X	CLN3/CLN5/CLN6									F	
Bardet-Biedl	X	BBS1/BBS2/BBS10/BBS12									F	
Cree Leukoencephalopathy												X
Cree Encephalitis												X

X: Recommended by the SOGC as a population screen; **F**: Recommended by SOGC for patients with a family history.
†Additional genes may be associated with some conditions, which are not part of the Invitae* screen.
‡(SLSJ+C) = French Canadian from Saguenay Lac-St-Jean and Charlevoix Regions;
(BSL, GASP, NB) = French Canadian from Bas-St-Laurent, Gaspesie, and nearby New Brunswick regions.
#Note about table: Amish, Mennonite, and Hutterite religious groups are at an increased risk for greater than 150 genetic conditions, but are not included in the table above. An in-depth three generation family history assessment is recommended for these groups.

ORDER EXPANDED CARRIER SCREENING THROUGH LIFELABS GENETICS

LifeLabs Genetics offers Expanded Carrier Screening through our partner, Invitae. Our team of certified genetic counsellors is available to support healthcare providers throughout the ordering process.

1. Download the following documents from www.LifeLabsGenetics.com:
   (1) LifeLabs Genetics Requisition Form; (2) Patient Consent Form; (3) Ministry of Health funding application, if applicable
2. Your patient can visit one of the many LifeLabs Patient Service Centres to have their blood drawn.
   • Self-collection saliva kits are also available. Please contact us for more details.
3. Results will be sent to the healthcare provider within 2-3 weeks from receipt of the sample at Invitae in California.
4. All patients are eligible for post-test counselling with a certified genetic counsellor through Invitae.
5. Patients are encouraged to discuss their results and the outcome of the genetic counselling session with their healthcare provider.

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