GEC KO
Genetics Education Canada - Knowledge Organization
Centre d'éducation en génétique canadien - Connaissances organisées

POINT OF CARE TOOL

Reproductive Genetic Carrier Screening in Canada

All couples planning their families should have a three-generation family history taken, ideally in the preconception period. Attention should be paid to the red flags in Box 1 to assess risk to future offspring.

A personal or family history of:

- congenital anomaly e.g. congenital heart defect, neural tube defect
- intellectual disability or developmental delay
- genetic syndrome e.g. neurofibromatosis, Noonan syndrome
- chromosomal disorder e.g. Down syndrome (trisomy 21), familial translocation
- muscular disorder e.g. X-linked Duchenne and Becker muscular dystrophies
- bleeding disorder e.g. X-linked hemophilia A or B
- stillbirth
- sudden unexplained death
- other major health concerns such as cardiomyopathy, neurological disease, epilepsy, hearing loss, autism, and psychiatric disorders
- consanguinity

Box 1. Personal and family history red flags that should prompt a referral for genetic consultation, ideally when individuals are planning a family (preconception).

A history of any of these red flags should prompt referral for genetic consultation. Individuals and their partners should be encouraged to make their best efforts to obtain confirmatory information such as medical records, genetic test results, even family photos.

One's ethnicity is an important piece of risk assessment as some populations are known to have a higher incidence of certain genetic conditions due to a **founder effect**. Founder effect confers reduced genetic diversity in a population descended from a small number of ancestors. A founder mutation refer to a specific pathogenic genomic variation in a specific population due to the presence of that genomic variation in a single or small number of ancestors.

Other considerations:

— There is a higher incidence of **hemoglobinopathies** in certain populations, screening recommendations can be found here

— Canadian recommendation for reproductive carrier screening in individuals of **Ashkenazi Jewish** ethnicity can be found here, and those from certain **regions of Quebec** are on page 2

— Canadian carrier screening recommendations for **cystic fibrosis, fragile X syndrome** and **spinal muscular atrophy** can be found here

— Individuals who are of Cree ancestry have a higher carrier frequency of **Cree encephalitis** (1/30-1/17) and **Cree Leukoencephalopathy** (~1/10). Screening programs have been developed in some regional communities. The CE-CLE Screening Program is offered to adults in the Awash clinics and to high school students.

— **Aboriginal Manitoba** populations have a higher incidence of cerebro-oculo-facio-skeletal syndrome

— **Newfoundland** populations have a higher incidence of Bardet Biedl syndrome and neuronal ceroid lipofuscinosis.

— A maternal family history of bleeding disorders in a woman's male relatives (father, brother, and/or maternal uncles) should prompt referral for consideration of carrier screening of **X-linked hemophilia.**

— Families **Amish, Mennonite, or Hutterite** background based on family history and/or geographic or religious settlement locality, in addition to a three-generation family history, should be offered referral for genetic consultation.

Expanded carrier testing is privately available genetic testing which screens an individual for more than just guideline/ethnicity-based conditions. See our Education Module for more information on this type of testing.

Table 1. Canadian recommendations for reproductive carrier screening of French Canadians originating from specific geographic regions. See this map for Quebec health regions. Screening should be offered when both members of a couple have at least one grandparent originating from the specified region.

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CHEO
Mount Sinai Hospital
Sinai Health System
Joseph & Wolf Lebovic Health Complex
Family & Community Medicine
UNIVERSITY OF TORONTO
uOttawa

Reproductive Genetic Carrier Screening in Canada: French Canadian

References and Resources:

— Wilson RD, De Bie I, Armour CM, et al. Joint SOGC-CCMG Opinion for Reproductive Genetic Carrier

Saguenay Lac-St-Jean (SLSJ) and Charlevoix regions		
Condition	**Carrier frequency**	**Screening recommendations**
Cystic fibrosis	SLSJ, 1/15 Charlevoix, 1/20	DNA-based analysis is recommended for: ✓ all at-risk couples For couples in Quebec, see the provincial screening program site for steps to obtaining testing. For couples outside of Quebec, consider contacting your local genetics centre for referral criteria or assistance ordering testing.
Tyrosinemia type I	1/19	
Leigh syndrome (French Canadian type)	1/23	
Autosomal Recessive Spastic Ataxia, Charlevoix-Saguenay (ARSACS)	1/23	
Agenesis of the Corpus Callosum with peripheral neuropathy	1/23	
For individuals from the SLSJ region, attention to family histories of myotonic dystrophy type I, congenital disorder of glycosylation type 1B, Tay-Sachs disease, and mucolipidosis II should also be considered. Turnaround time for this testing can be lengthy and out-of-province approval may be necessary prior to ordering testing which can increase wait time for results. To maximize a couple's family planning options, these processes should be initiated as soon as possible.		

Quebec Bas-St-Laurent (Rimouski) and Gaspésie, adjoining New Brunswick		
Condition	**Carrier frequency**	**Screening recommendations**
Tay-Sachs disease	1/14	If there is a positive family history, consider screening by targeted DNA-based analysis for the familial pathogenic variant. Contact your local genetics centre for how to order testing.
When only one member of a couple is of French Canadian ethnicity, the decision to screen the couple should take into consideration the frequency of the condition and the availability of reliable screening in non-French Canadian individuals.		

Screening: An Update for All Canadian Providers of Maternity and Reproductive Healthcare in the Era of Direct-to-Consumer Testing. J Obstet Gynaecol Can 2016;38(8):742-762.e3

— Offre de tests de porteur pour quatre maladies héréditaires récessives chez les personnes originaires des régions du Saguenay–Lac-Saint-Jean, de Charlevoix et de la Haute-Côte-Nord

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