Canadian recommendations for reproductive carrier screening of French Canadian individuals

Click here to see a map of Québec regions.

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Recommendations for those who are from the 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 Québec, see the provincial screening program site for steps to obtaining testing. For couples outside of Québec, 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	
Agenesis of the corpus callosum with peripheral neuropathy	1/23	

For individuals from the SLSJ region, pay attention to family histories of myotonic dystrophy type I, congenital disorder of glycosylation type 1B, Tay-Sachs disease, and mucolipidosis II as testing for these conditions may 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.

Recommendations for those who are from Québec 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 DNA-based targeted analysis for the familial pathogenic variant

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.

References and resources

- Wilson et al 2016 J Obstet Gynaecol PMID: 27638987
- Dépistage et offre de tests de porteur en Québec:
  - 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

GECKO
Genetics Education Canada: Knowledge Organization
Centre d'éducation en génétique canadien: Connaissances organisées
POINT OF CARE TOOL

Reproductive genetic carrier screening in Canada

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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 the box below to assess risk to future offspring.

NOTE: Canadian guidelines (Wilson et al 2016 J Obstet Gynaecol PMID: 27638987) are currently under review by the Canadian College of Medical Geneticists, and a new publication is not yet available. We will update our content when new recommendations are released.

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 spectrum disorder, and psychiatric disorders
- consanguinity

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 refers 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 are here, and those from **certain regions of Québec** can be found here
- 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 male relatives (father, brother, and/or maternal uncles) should prompt referral for consideration of carrier screening of X-linked hemophilia.
- Families of **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.