

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. [Founder mutations](#) refer to specific gene mutations observed at high frequency in a specific population due to the presence of that gene mutation 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 regions or 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 with **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.

Saguenay Lac-St-Jean (SLSJ) and Charlevoix regions		
Condition	Carrier frequency	Screening recommendations
Cystic fibrosis	SLSJ, 1/15 Charlevoix, 1/20	DNA-based mutation analysis is recommended for: ✓ all at-risk couples Contact your local genetics centre for how to arrange testing.
Tyrosinemia type I	1/19	DNA-based mutation analysis is recommended for: ✓ all at-risk couples
Leigh syndrome (French Canadian type)	1/23	DNA-based mutation analysis is recommended for: ✓ all at-risk couples
Autosomal Recessive Spastic Ataxia, Charlevoix-Saguenay (ARSACS)	1/23	DNA-based mutation analysis is recommended for: ✓ all at-risk couples
Agenesis of the Corpus Callosum with peripheral neuropathy	1/23	DNA-based mutation analysis is recommended for: ✓ all at-risk couples
<p>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 mucopolipidosis II should also be considered.</p> <p>Turnaround time for this testing can 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.</p>		
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 DNA-based targeted mutation analysis (ideally for the familial gene mutation). Contact your local genetics centre for how to order testing.
<p>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.</p>		

References and Resources:

[Wilson RD, De Bie I, Armour CM, et al. 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. J Obstet Gynaecol Can 2016;38\(8\):742-762.e3](#)

Offre de test génétique au SLSJ www.genetique.santesaglac.com