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# 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 and those from certain **regions of Quebec** can be found here
- Canadian carrier screening recommendations for **cystic fibrosis, fragile X syndrome** and **spinal muscular atrophy** are on page 2.
- 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.

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# Reproductive Genetic Carrier Screening in Canada: Condition specific

**Table 1. Canadian recommendations for condition specific reproductive genetic carrier screening.**

| Condition | Screening Recommendations |
|---|---|
| Cystic fibrosis (CF) | General population carrier screening for CF **is not** recommended.<br><br>DNA-based analysis of the *CFTR* gene is recommended for:<br>✓ Individuals with a personal or family history of CF<br>✓ Both padiorents of a fetus with an ultrasound finding of echogenic bowel<br>✓ All couples from the Saguenay Lac-St-Jean and Charlevoix regions in Québec where the CF carrier frequencies are 1/15 and 1/20, respectively<br><br>Newborn screening for CF is offered as part of most all Canadian provincial screening programs with the exception of Quebec and Nunavut (Kivilliq). |
| Fragile X syndrome (FXS) and related conditions | General population carrier screening for FXS **is not** recommended.<br><br>Offering FXS carrier screening is **recommended for** any female (46, XX) with a personal or family history of:<br>✓ Fragile X syndrome or fragile X-related conditions<br>✓ Unexplained intellectual disability or developmental delay<br>✓ Autism<br>✓ Premature menopause i.e. ovarian insufficiency with elevated follicle stimulating hormone at age<40 years of unknown etiology<br>✓ History of male (46,XY) relatives with isolated cerebellar ataxia and tremor<br><br>Due to the complexity of interpreting FXS carrier screening results, pre- and post-test genetic counselling is strongly recommended. |
| Spinal Muscular Atrophy (SMA) | General population carrier screening for SMA **is not** recommended.<br><br>Any couple where one member has a family history of SMA **should be offered referral** for reproductive genetic counselling for consideration of carrier screening. |

**Reference 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


— Click on each condition in the table to link to a patient/family resource


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