Genetic Carrier Screening in Canada: Condition specific

Reviewed March 2023

Current Canadian guidelines recommend against general population carrier screening for cystic fibrosis, Fragile X syndrome and spinal muscular atrophy. See the table below for current screening recommendations.

Condition	Screening recommendations
Cystic fibrosis (CF)	General population carrier screening for CF is not recommended. DNA-based analysis of the *CFTR* gene is recommended for: • Individuals with a personal or family history of CF • Both parents of a fetus with an ultrasound finding of echogenic bowel • All couples from the Saguenay Lac-St-Jean and Charlevoix regions in Québec, as the CF carrier frequencies are 1/15 and 1/20, respectively Newborn screening for CF is offered as part of most Canadian provincial screening programs.
Fragile X syndrome (FXS) and related conditions	General population carrier screening for FXS is not recommended. Offering FXS carrier screening is recommended for any person (46, XX) with a personal or family history of: • Fragile X syndrome or fragile X-related conditions • Unexplained intellectual disability or developmental delay • Autism spectrum disorder • Premature menopause i.e. ovarian insufficiency with elevated follicle stimulating hormone of unknown etiology under 40 years of age • History of male (46,XY) relatives with isolated cerebellar ataxia and tremor 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. Any couple where one member has a family history of SMA should be offered referral for reproductive genetic counselling for consideration of carrier screening.

Remember to also take a family history targeting red flags that may suggest additional testing.

References and resources

- Wilson et al 2016 J Obstet Gynaecol PMID: 27638987
- Cystic Fibrosis Canada
- Fragile X Research Foundation of Canada
- Cure SMA Canada
- Muscular Dystrophy Canada

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

Reproductive genetic carrier screening in Canada

Reviewed March 2023

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.