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

Reviewed March 2023

Canadian recommendations for reproductive carrier screening of individuals who are of Ashkenazi Jewish (AJ) ethnicity

Ashkenazi Jews are descendants of the Jewish communities of Germany, Poland, Austria and Eastern Europe. For genetics purposes, an individual with one biological grandparent of Ashkenazi Jewish heritage is considered a candidate for carrier screening.

Condition	Carrier frequency	Screening recommendations
Tay-Sachs disease	1/30	DNA-based analysis is recommended for all at-risk couples
Canavan disease	1/37 - 1/53	
Familial dysautonomia	1/32	

When only one member of a couple is of AJ ethnicity, the non-AJ partner should be screened for Tay-Sachs disease, using biochemical hexosaminidase enzyme activity since available DNA testing targets specific pathogenic variants found in AJ populations (*founder mutations*).

No screening for Canavan Disease and Familial Dysautonomia is recommended in non-AJ individuals, as both conditions are very rare in the general population.

Some provinces offer carrier screening panels that include more than the three conditions above. See page 2 for more on testing in your province.

Screening for other genetic conditions where common pathogenic variants are known can also be considered, particularly when there is a positive family history, for example:

Condition	Carrier frequency	Screening recommendations
Gaucher disease	1/18	If there is a positive family history, consider screening by DNA-based targeted analysis (ideally for the familial pathogenic variant, although gene panels with common AJ pathogenic variants may be considered).
Cystic fibrosis (CF)	1/24	
Fanconi anemia group C	1/89	
Niemann-Pick type A	1/90	
Mucolipodosis type IV	1/100	
Bloom syndrome	1/104	

When only one member of a couple is of AJ ethnicity, the decision to screen the couple should consider the frequency of the condition and the availability of reliable screening in the non-AJ partner.

E.g. Where there is a positive family history of CF and one member of a couple is of AJ ethnicity and the other member of the couple is of Northern European ethnicity; offering carrier screening for CF to this couple is reasonable. However, offering screening for a family history of the rare Bloom syndrome would not be recommended as the carrier frequency and utility of screening in non-AJ individuals is unknown.

References and resources

- Wilson et al 2016 J Obstet Gynaecol PMID: 27638987
- The National Council of Jewish Women of Canada
- Jewish Genetic Disease Consortium

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Ashkenazi Jewish Genetic Carrier Screening across Canada

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Province	How to order testing	Conditions screened
Alberta	• Testing is not available in province • Approval for out of province funding must first be obtained through the Genetic Resource Centre • Applications are reviewed on a case-by-case basis	o Tay-Sachs disease o Canavan disease o Familial dysautonomia
British Columbia & Yukon	• Instructions, requisitions and counselling information found here ◦ Molecular Genetics Laboratory at BC Children's and BC Women's Hospitals	o Tay-Sachs disease o Canavan disease o Familial dysautonomia o Fanconi anemia type C
Manitoba	• Requisition is available here through the Diagnostic Services of Manitoba ◦ Diagnostic Services of Manitoba	o Tay-Sachs disease o Canavan disease o Familial dysautonomia o Fanconi anemia type C
Maritimes (New Brunswick, Nova Scotia & Prince Edward Island)	Contact local genomics centre	o Tay-Sachs disease o Canavan disease o Familial dysautonomia o Fanconi anemia type C
Newfoundland and Labrador	Contact local genomics centre	
Ontario	Hospital for Sick Kids Department of Pediatric and Laboratory Medicine • Full panel requisition here • Tay Sachs biochemical requisition here	o Tay-Sachs disease o Canavan disease o Familial dysautonomia o Fanconi anemia group C o Bloom syndrome o Mucolipidosis type IV o Niemann Pick disease
Québec	McGill University Health Centre Jewish Genetic Condition Screening Program Additional Québec resources	o Tay-Sachs disease o Canavan disease o Familial dysautonomia
Saskatchewan	Saskatchewan Health Authority Ashkenazi Jewish Genetic Panel Refer to Saskatoon Medical Genetics Centre	o Tay-Sachs disease o Canavan disease o Familial dysautonomia o Fanconi anemia group C o Bloom syndrome o Mucolipidosis type IV o Niemann Pick disease

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

Both members of a couple are of AJ ethnicity

And
currently pregnant

Yes

- **Offer** AJ panel based carrier screening to both members concurrently
- Indicate ongoing pregnancy to expedite results

Both members are carriers of the **same** condition

Yes

Refer to your local genetics center with results

No

STOP

No, preconception

Offer AJ panel based carrier screening to **female** member first

Positive result

Yes

Offer the same AJ panel based carrier screening to male member

Both members are carriers of the **same** condition

Yes

Refer to your local genetics center with results

No

STOP

No

STOP

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One member of a couple is of AJ ancestry

And
currently pregnant

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Concurrently
rather than sequentially for prompt receipt of the couple's results

Offer AJ panel based carrier screening to AJ member only

Consider offering Tay-Sachs disease screening to the non-AJ member with biochemical analysis (vs genetic testing)

Indicate ongoing pregnancy to expedite results

Both members are carriers of Tay Sachs disease

Yes

Refer to your local genetics center with results

No

STOP

AJ member is positive for another condition

Pause

Consider the frequency of the condition and the availability of reliable screening in the non-AJ partner e.g. screening for familial dysautonomia or Canavan disease is not recommended in non-AJ individuals as the carrier frequency is low and testing/interpretation of genetic results is limited.

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

One member of a couple is of AJ ancestry

And not currently pregnant (preconception)

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Offer AJ panel based carrier testing to AJ member **only**

Positive result

Yes

Positive result for Tay Sachs disease

Yes

Offer Tay Sachs disease screening to partner with biochemical analysis (vs genetic testing)

Positive result

Yes

Refer to your local genetics center with results

No

STOP

No

STOP

No, positive for another condition

Pause

Consider the frequency of the condition and the availability of reliable screening in the non-AJ partner e.g. screening for familial dysautonomia or Canavan disease is not recommended in non-AJ individuals as the carrier frequency is low and testing/interpretation of genetic results is limited.

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

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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.