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

FAMILIAL HYPERCHOLESTEROLEMIA (FH)

Updated Oct 2024

GENETIC TESTING FOR FH

Genetic testing is clinically available in all provinces with the exception of British Colombia. It is possible for any physician to order genetic testing in Ontario and Québec. In other provinces, testing can currently only be ordered by a specialist (e.g. medical genetics, endocrinology, cardiology). Enlarge the table below to find more details about testing in your province.

GENETIC TESTING FOR FH ACROSS CANADA

Province	Clinical testing available (publicly funded)	Research testing available	Private pay testing available	Specialist required to order testing	Links to laboratory requisitions
BC	N	Y	Y	N/A	Refer to endocrinology, cardiology, or internal medicine
AB	Y	N	Y	Y	Refer to any specialist
SK	Y	N	Y	Y	Refer to medical genetics, endocrinology, cardiology or internal medicine
MB	Y	N	Y	Y	Refer to [illegible]
ON	Y	Y	Y	N	• London Health Sciences Centre molecular lab • Trillium Health Partners Credit Valley Site • Hamilton Regional Laboratory Medicine Program
QC	Y	Y	Y	N	• The Core Molecular Genetics Laboratory at the McGill University Health Centre • CHU Sainte Justine Molecular Laboratory
Maritimes	Y	N	Y	Y	Refer to medical genetics, endocrinology or cardiology
NFLD	Y	N	Y	Y	Refer to medical genetics

Genetic testing for FH across Canada

WHAT DO I DO WITH MY PATIENT'S GENETIC TEST RESULTS?

Positive results

One or more pathogenic/likely pathogenic (P/LP) variants in an FH gene have been identified and are known to be associated with FH.

This confirms a diagnosis of FH.

These results can be used to guide management and refine risk stratification.

For those with two copies of a P/LP variant in FH genes, homozygous FH, consider referral to specialized lipid clinic.

Cascade Screening

This patient now becomes the index case and their first-and second-degree relatives can be offered genetic testing for the familial gene variant(s).

Facilitate family communication and testing.

Cascade screening is the most cost-effective approach for identification of new FH cases.

Cascade screening reduces the average age at which an individual is diagnosed and results in an increased number of individuals who are treated with statins and have subsequent lowered lipid levels.

Genetic counselling

Genetics clinics vary in their referral criteria and may or may not accept referrals for familial testing of FH. Check your local genetics centre for more.

Negative results (uninformative)

No pathogenic/likely pathogenic (P/LP) variants in the FH genes tested, were identified.

This **does not rule out a diagnosis of** FH

Utility of the results depend upon the extent of analysis (e.g. ancestry based targeted testing vs. full gene sequencing) as well as the genes included on the panel.

While P/LP variants in the L*DLR, APOB* and *PCSK9* genes account for the majority (~80%) of FH, many other genes are known to affect hereditary risk of hypercholesterolemia.

Genetic testing cannot be offered to unaffected relatives. Lipid screening can be used for at-risk relatives.

Genetic testing may improve and re-referral may be considered.

True Negative results

The familial P/LP variant in the FH gene tested is not present.

This person is not at risk for FH.

This testing *only* looked for the presence or absence of the familial variant.

Variant of Uncertain Significance (VUS)

A variant in FH-related gene is detected, but there is insufficient evidence to determine if it is truly associated with disease.

This **does not rule out a diagnosis of** FH

Cascade screening of relatives is rarely offered in the context of a VUS.

Genetic counselling may be available. Check your local genetics centre for referral criteria.

Lipid screening can be used for at-risk relatives.

Over time a VUS may be reclassified, re-referral to genetics can be considered.

Patient resources

https://www.fhcanada.net/patients

HF Canada Hypercholestérolémie Familiale
FH Canada Familial Hypercholesterolemia

More FH resources

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