



Molecular Diagnostic Laboratory

Montreal Heart Institute
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Phone : 514 376-3330 ext 3712
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Last Name, First Name : _____
Date of Birth : _____
Sex : _____
Record number : _____ *Addressograph* _____
RAMQ number (if available): _____
Contact (Phone number) : _____

A- Sample Information

Required

Date : - -
Collected by (Name) : _____
Laboratory reference number : _____
 Patient's consent was obtained (A copy of the signed consent form must be provided)
Sample type (check) :
 Lavender tube (EDTA) – One 4 ml tube required
 DNA Quantity required : 5 µg Concentration (must be indicated on tubes) : _____

For laboratory use

N.B.: DNA samples extracted from Formalin-fixed, paraffin-embedded (FFPE) tissues will not be accepted.

B- Reason for this request (Please check all that apply) :

Required

Genetics test request (Please complete section D or E) Family number (if known) : _____
Patient ethnicity (ex : french canadian) : _____
 DNA banking
 Additional testing (Please complete section D or E)
 Deceased patient (date of death): _____

C- Known or suspected diagnostic (Please check all that apply) :
Required

D- Patient's Phenotype **Required for patients non-treated at the MHI**

Familial condition (known or suspected)	Clinical data :	Family History :
<input type="checkbox"/> Familial hypercholesterolemia <input type="checkbox"/> Familial hyperalphalipoproteinemia <input type="checkbox"/> Familial hyperchylomicronemia <input type="checkbox"/> Sitosterolemia <input type="checkbox"/> Other : _____	Pharmacotherapy : _____ LDL-C (mmol/L) : _____ Before treatment : _____ After treatment initiation : _____ Tg (mmol/L) : _____	
Please also complete the HF Dutch Lipid Clinic Network Diagnostic Criteria grid (back).		

E- Targeted variant testing only

Required for family member

Specific Variant:	Gene	Transcript	Nucleic Change	Protein Change
For variant(s) not reported by our laboratory, please attach a copy of a family member's report if available.	_____	_____	_____	_____
	_____	_____	_____	_____

F- Dyslipidemia Panel Test (Please check all that apply)

Required for proband

Please note that sequential analyzes are performed. If no clinically significant variant is identified within the first-line analysis, the second and third-line panels will automatically be completed.

Familial dyslipidemia	First-Line	Second-Line	Third-Line
<input type="checkbox"/> Familial hypercholesterolemia (FH)	LDLR (Copy Number Variations - CNV) Only whole blood samples are accepted for this test	APOB, LDLR, LDLRAP1, PCSK9	-
<input type="checkbox"/> Familial hyperalphalipoproteinemia	CETP	-	-
<input type="checkbox"/> Familial hyperchylomicronemia	LPL	APOA5, APOC2, GPIIIBP1, LMF1	-
<input type="checkbox"/> Sitosterolemia	ABCG5, ABCG8	-	-

Additional gene(s) :

Cardiovascular Genetics Test Requisition Form - Familial dyslipidemia

G- Health Care Professional Information :

Required

I confirm that this test is medically indicated for the stated clinical condition and that the results will be used for clinical purposes for the patient. I have explained to the patient or his / her representative / legal guardian the indicated genetic analysis and its objectives, limitations, risks and benefits and have satisfactorily answered all related questions.

Last Name & First Name: _____
 Full address: _____

License number : _____
 Phone : _____
 Fax : _____
 Date : _____
 Signature : _____

c.c. Additional reporting provider, coroner, genetic counselor or other:

Last Name & First Name: _____
 Full address : _____

License number : _____
 Phone : _____
 Fax : _____

H- Laboratory Contact Information :

Institution : _____
 Full address : _____

Contact person : _____
 Phone : _____
 Fax : _____

Dutch Lipid Clinic Network diagnostic criteria for familial hypercholesterolemia

Please circle the criteria that apply

Criteria	Points
Family history	
i. First-degree relative with premature coronary heart disease (M < 55 years; F < 60 years) OR	1
ii. First-degree relative with known LDL-C level > 95th percentile	
iii. First-degree relative with tendinous xanthoma and / or arcus cornealis OR	2
iv. Children aged < 18 y/o with LDL-C level > 95th percentile	
Clinical history	
i. Patient with premature coronary heart disease (M < 55 years; F < 60 years)	2
ii. Patient with premature cerebral or peripheral vascular disease	1
Physical examination	
i. Tendinous xanthoma	6
ii. Arcus cornealis < 45 years	4
LDL cholesterol level	
i. LDL-C level > 8,5 mmol/L	8
ii. LDL-C level 6,5 - 8,4 mmol/L	5
iii. LDL-C level 5,0 - 6,4 mmol/L	3
iv. LDL-C level 4,0 - 4,9 mmol/L	1
DNA analysis	
Causative variant in <i>LDLR</i> , <i>APOB</i> or <i>PCSK9</i> genes	8
TOTAL SCORE :	

Diagnosis (based on the total number of points obtained) :

Definite familial hypercholesterolemia: > 8 points
 Probable familial hypercholesterolemia: 6-8 points
 Possible familial hypercholesterolemia: 3-5 points
 Unlikely familial hypercholesterolemia: 0-2 points

Indication for genetic testing :

Heterozygous FH

Patient with a score > 5 points

Patient with xanthoma, high cholesterol and family history of coronary heart disease

First-, second-, and third-degree relative of a proband known with a causative variant in one of the genes associated with familial hypercholesterolemia (Section E-Targeted variant testing only)

Homozygous FH

Patient in whom both parents are known with a causative variant in one of the genes associated with familial hypercholesterolemia

OR

Untreated patient with LDL-C level > 12,0 mmol/L and treated patient with LDL-C level > 8,0 mmol/L

AND

Cutaneous or tendonous xanthoma < 10 years

OR

Both parents with elevated LDL-C levels reflecting heterozygous HF