INSTITUT DE CARDIOLOGIE DE MONTRÉAL

Cardiovascular Genetics Test Requisition Form
Familial dyslipidemia

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 : yyyy - mm - dd
Collected by (Name) :
Laboratory reference number :
☐ **Patient's consent was obtained *(A copy of the signed consent form must be provided)***

For laboratory use

Sample type (check) :
☐ Lavender tube (EDTA) – One 4 ml tube required
☐ DNA Quantity required : 5 µg Concentration (must be indicated on tubes) :

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 E **or** F*)
Family number (if known) :
Patient ethnicity (ex : french canadian) :
Deceased patient Date of death :
☐ DNA banking
☐ Additional testing (Please *complete section E **or** F*)

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

Familial condition (known or suspected)
☐ Familial hypercholesterolemia
☐ Familial hyperalphalipoproteinemia
☐ Familial hyperchylomicronemia
☐ Sitosterolemia
☐ Other :

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

Clinical data :
Pharmacotherapy :
LDL-C (mmol/L) :
Before treatment :
After treatment initiation :
Tg (mmol/L)

Family History :

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.				

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-line panels will automatically be completed.

Familial dyslipidemia	**First-Line**	**Second-Line**
☐ Familial hypercholesterolemia (FH)	*LDLR* (Copy Number Variations - CNV) **Only whole blood samples are accepted for this test**	*APOB, LDLR, LDLRAP1, PCSK9*
☐ Familial hyperalphalipoproteinemia	*CETP*	-
☐ Familial hyperchylomicronemia	*LPL*	*APOA5, APOC2, GPIHBP1, LMF1*
☐ Sitosterolemia	*ABCG5, ABCG8*	-
Additionnal gene(s) :		

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

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

Last Name & First Name: _____ License number : _____
Full address : _____ Phone : _____
Fax :

H- Laboratory Contact Information :

Institution : _____ Contact person : _____
Full address : _____ 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 *LDLR, APOB* or *PCSK9* 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