INSTITUT DE CARDIOLOGIE DE MONTRÉAL

Test requisition form – Familial Dyslipidemia

Molecular Diagnostic Laboratory

Montreal Heart Institute
5000, Belanger Street, C-1760
Montreal (Quebec) H1T 1C8
Phone : 514 376-3330 ext. 3712 Fax : 514 593-2577
Email : ldm@icm-mhi.org

Last Name, First Name :
Date of Birth :
Sex :
Record Number :
RAMQ Number (if available):
Contact (phone number) :

Adressograph

Please provide at least two (2) unique identifiers

For any test request, you must provide:
☐ **Test requisition form – Familial Dyslipidemia**
☐ **Consent form for genetic testing F-G-CONSEN-9297**

For any molecular autopsy request, you must provide:
☐ **Test requisition form – Familial Dyslipidemia**
☐ **Toxicological analysis result and autopsy report**

A- Sample information — *REQUIRED*

Date : YYYY - MM - DD
Collected by (Initials) : _____
Laboratory reference number : _____
Sample type (check) :
☐ **Lavender tube (EDTA) – One tube (at least 1 mL)** ☐ **Specimen already sent to MDL**
☐ **DNA – One tube (at least 3-5 µg) :**
Requirements: 25 µL minimal volume with a 260/280 ratio between 1.8 and 2.2. Following buffers accepted: Tris-EDTA and elution buffer provided in a commercial DNA extraction kit
N.B.: DNA samples extracted from formalin fixed and paraffin-embedded tissues (FFPE) are not accepted. In addition, if the patient has a hematologic malignancy, DNA extracted from a non-hematologic source is strongly recommended. Please contact the laboratory as needed.

For laboratory use

B- Reason for this request (please check all that apply) : — *REQUIRED*

☐ **Genetic test request** (*please complete section D or E and F*)
Patient ethnicity (ex : french canadian) : _____
Family Number (if known) : _____
Deceased patient, Date of death : YYYY - MM - DD

☐ **Specimen banking**
☐ **Additionnal testing**
(*please complete section D or E and F*)

C- Patient's phenotype — *REQUIRED*

Clinical data :
Pharmacotherapy : _____
LDL-C level before treatment (mmol/L): _____
LDL-C level after treatment initiation (mmol/L): _____
Tg level before treatment (mmol/L): _____
Tg level after treatment initiation (mmol/L): _____

Familial history :

The HF Dutch Lipid Clinic Network Diagnostic Criteria grid must be completed (page 3).

D- Dyslipidemia Panel Test (Please check all that apply): — *REQUIRED FOR PROBAND*

Panel Test	Sequenced Genes Analysis includes detection of single nucleotide genetic variations (SNV), small genetic variations (InDel) and copy number variations (CNV), as well as certain intronic variants associated with hereditary cardiovascular diseases A list of intronic variants is available at https://www.icm-mhi.org/en/health-care-and-services/clinics-and-services/cardiovascular-genetics-center.
Familial dyslipidemia	
☐ Familial Hypercholesterolemia	*APOB, LDLR, LDLRAP1, PCSK9*
☐ Familial hyperalphalipoproteinemia	*CETP*
☐ Familial hyperchylomicronemia	*APOA5, APOC2, GPIHBP1, LMF1, LPL*
☐ Sitosterolemia	*ABCG5, ABCG8*
Additional gene(s) (only for gene present on this request):	_____

E- Targeted variant testing only : *REQUIRED FOR FAMILY MEMBER*

Known or suspected diagnosis : _____

Specific variant(s) :

For variant(s) not reported by our laboratory, please attach a copy of a family member's report.

Gene	Transcript	Nucleic change	Protein change

F- 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 : _____

G- Laboratory Contact Information : *OPTIONAL*

Institution : _____
Full Address : _____
_____

Contact person : _____
Phone : _____
Fax : _____

Warning: Any incomplete or non-compliant analysis request will be refused

H- Shipping instruction :

Specimen Dispatching :

Whole blood : The whole blood tubes are sent at room temperature as per the current transportation standards for biomedical specimens. The specimens should be received at the latest ten (10) days after the sampling date.

DNA : DNA samples can be sent at room temperature or in an icebox if samples were previously frozen.

Shipping Address :

Molecular Diagnostic Laboratory (C-1760)
Montreal Heart Institute
5000, Belanger Street
Montreal (Quebec) H1T 1C8

The HF Dutch Lipid Clinic Network Diagnostic Criteria grid must be completed (page 3).

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** ii. First-degree relative with known LDL-C level > 95th percentile	1
iii. First-degree relative with tendinous xanthoma and / or arcus cornealis **OR** iv. Children aged < 18 y/o with LDL-C level > 95th percentile	2
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
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 :
Heterozygote 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)
Homozygote 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 FH