INSTITUT DE CARDIOLOGIE DE MONTRÉAL

Test requisition form - CARDIOVASCULAR GENETICS

Molecular Diagnostic Laboratory

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

Please attach :

☐ **Consent Form for Genetic Testing F-G-CONS-3860 AND/OR**
☐ **Toxicology results and autopsy report for all molecular autopsy requests**

Last name, First name :

Date of birth:
Gender:
File Number:
RAMQ Number:

Addressograph

Provide at least two (2) unique identifiers.

A- Sampling/Sample Type — MANDATORY

Date: AAAA - MM - JJ **By:** _____

☐ **Whole blood - EDTA lavender tube (at least 1 mL)**
☐ **Extracted DNA - One tube (minimum 3-5 µg)** **Source*:** _____

**Avoid liver sample. If the patient has a hematologic malignancy, DNA extracted from a non-hematologic source is strongly recommended.*

☐ **Specimen already sent to MDL**

For MDL use

Shipping instructions to the above address

Whole blood: Tubes are shipped at room temperature, according to current biomedical specimen transport standards. Reception of specimens must be within ten (10) days after collection.

DNA: Samples can be sent at room temperature or in a cooler if they have been previously frozen.

B- Reason for this request — MANDATORY

☐ **Request for cardiovascular genetic analysis** Family number (if known) : _____
☐ **Adding tests to a request** Deceased patient, date of death: AAAA - MM - JJ
☐ **Specimen banking**

Priority request: ☐ *Pregnancy* ☐ *ICD implantation* ☐ *Surgical decision* ☐ *Other (justify request by email to the MDL)*

C- Analysis request for specific variant(s) — FAMILY MEMBER only

Known or suspected familial diagnosis: _____

Clinical Indication (required): Co-segregation study: ☐ Affected patient ☐ Unaffected patient ☐ Patient with unclear phenotype
☐ Predictive testing following genetic counseling ☐ Other : _____

Family variant(s) targeted:	**Gene**	**Transcript**	**Nucleic change**	**Protein change**
For variant(s) not reported by our laboratory, attach copy of a family member's report				

D- Analysis request for a complete profile (*check Panel AND Criteria*) : — PROBAND only

Panel test and sequenced genes	Clinical indications for test eligibility
Idiopathic or genetic/hereditary cardiomyopathy (CM) Medical genetics consultation is recommended if diagnosis < 14 years, dysmorphia, or extracardiac involvement. Specialty clinic opinion is required for all conditions except TTR amyloidosis.	
☐ Hypertrophic CM	
ACTC1, ACTN2, ALPK3, CACNA1C, CSRP3, DES, FHL1, FHOD3, FLNC, GLA, JPH2, LAMP2, MYBPC3, MYH7, MYL2, MYL3, PLN, PRKAG2, PTPN11, RAF1, RIT1, TNNC1, TNNI3, TNNT2, TPM1, TRIM63, TTR	☐ Left ventricular (LV) wall ≥ 15 mm, OR ☐ LV wall ≥ 13 mm with family history, OR ☐ LV wall with Z score > 2 (in children) **AND** ☐ LV thickening not fully explained by overload condition **AND** ☐ Evaluation in a specialized medical genetics or cardiogenetics clinic
☐ Dilated CM or left ventricular noncompaction	
AARS2, ACTC1, ACTN2, BAG3, DES, DMD, DOLK, DSC2, DSG2, DSP, EMD, FKRP, FKTN, FLNC, HCN4, JPH2, JUP, LAMP2, LMNA, MYBPC3, MYH7, NEXN, NKX2-5, NRAP, PKP2, PLN, PPCS, PPP1R13L, RAF1, RBM20, RYR2, SCN5A, TAFAZZIN, TMEM43, TNNC1, TNNI3, TNNI3K, TNNT2, TPM1, TTN, VCL	☐ Dilated CMP with conduction disorder (bifasicular block or atrioventricular block) OR ☐ CMP with family history of non-ischemic heart disease and/or malignant arrhythmia and/or conduction disorder OR ☐ CMP with myocardial fibrosis and/or ventricular arrhythmia OR ☐ Severe CMP requiring invasive treatment (defibrillator, mechanical support, heart transplant) OR ☐ Dilated CMP with diagnosis at young age (e.g. < 40 years) **AND** ☐ Evaluation in a specialized medical genetics or cardiogenetics clinic

Isolated left ventricular noncompaction in adults without myocardial fibrosis, ventricular arrhythmia, ventricular dysfunction, or family history is not an indication for genetic analysis.

☐ Arrhythmogenic right ventricular CM	
ANK2, CDH2, DES, DSC2, DSG2, DSP, FLNC, JUP, LMNA, PKP2, PLN, TMEM43	☐ Definitive or borderline diagnosis according to TF2010 criteria (Pubmed ID: 20172911) **AND** ☐ No left ventricular involvement (if associated left ventricular involvement, the dilated CMP panel is recommended) **AND** ☐ Evaluation in a specialized medical genetics or cardiogenetics clinic
☐ TTR-related cardiac amyloidosis	
TTR	☐ Positive histological evaluation for amyloidosis and confirmation of TTR subtype OR ☐ Positive histological evaluation for amyloidosis and absence of circulating monoclonal protein OR ☐ Bone scan (e.g. PYP) positive for cardiac amyloidosis and absence of circulating monoclonal protein
Hereditary arrhythmias	
☐ Andersen-Tawil syndrome	
KCNJ2	☐ Clinical diagnosis according to the diagnostic criteria (Pubmed ID: 29125635) **AND** ☐ Evaluation in a specialized medical genetics, cardiogenetics or neurogenetics clinic
☐ Brugada syndrome	
SCN5A	☐ ECG showing a spontaneous type I Brugada pattern OR ☐ ECG showing a sodium blocker-induced type I Brugada pattern in a clinical setting suggesting Brugada syndrome **AND** ☐ Evaluation in a specialized medical genetics or cardiogenetics clinic
☐ Long QT syndrome	
CACNA1C, CALM1, CALM2, CALM3, KCNE1, KCNE2, KCNH2, KCNJ2, KCNQ1, SCN5A, TECRL, TRDN	☐ Survivor of cardiac arrest with QT prolongation on resting ECG OR ☐ Prolonged QTc (> 480 ms for ♀ and > 460 ms for ♂) OR ☐ Borderline QTc (460-480 ms for ♀ and 450-460 ms for ♂) in the presence of syncope AND Abnormal T-wave AND/OR Suspicious family history **AND** ☐ Evaluation in a specialized medical genetics or cardiogenetics clinic
☐ Short QT syndrome	
CACNA1C, KCNH2, KCNJ2, KCNQ1, SCN5A, SLC4A3	☐ QTc < 330 ms, OR ☐ QTc < 360 ms in the presence of clinical suspicion of short QT syndrome based on personal history (cardiac arrest, suspected syncope) or family history **AND** ☐ Evaluation in a specialized medical genetics or cardiogenetics clinic
☐ Catecholaminergic polymorphic ventricular tachycardia	
CALM1, CALM2, CALM3, CASQ2, KCNJ2, RYR2, TECRL, TRDN	☐ Clinical suspicion of catecholaminergic polymorphic ventricular tachycardia **AND** ☐ Evaluation in a specialized medical genetics or cardiogenetics clinic
☐ Familial progressive cardiac conduction defect	
DES, EMD, GLA, GNB5, HCN4, LAMP2, LMNA, NKX2-5, PRKAG2, SCN5A, SGO1, TBX5, TNNI3K, TRPM4, TTR	☐ Conduction disorder requiring pacemaker implantation <50 years OR ☐ Conduction disorder requiring pacemaker implantation <60 years of age in the presence of a family history of conduction disorder, sudden death, or cardiomyopathy. **AND** ☐ Evaluation in a specialized medical genetics or cardiogenetics clinic
☐ Unexplained cardiac arrest	
AARS2, ACTC1, ACTN2, ALPK3, ANK2, BAG3, CACNA1C, CALM1, CALM2, CALM3, CASQ2, CDH2, CSRP3, DES, DMD, DOLK, DPP6 (NM_130797.4:c.244-141059C>T), *DSC2, DSG2, DSP, EMD, FHL1, FHOD3, FKRP, FKTN, FLNC, GLA, GNB5, HCN4, JPH2, JUP, KCNE1, KCNE2, KCNH2, KCNJ2, KCNQ1, LAMP2, LMNA, MYBPC3, MYH7, MYL2, MYL3, NXN, NKX2-5, NRAP, PKP2, PLN, PPA2, PPCS, PPP1R13L, PRKAG2, PTPN11, RAF1, RBM20, RIT1, RYR2, SCN5A, SGO1, SLC4A3, TAFAZZIN, TBX5, TECRL, TMEM43, TNNC1, TNNI3, TNNI3K, TNNT2, TPM1, TRDN, TRIM63, TRPM4, TTN, TTR, VCL*	☐ Survivor of cardiac arrest with documented ventricular fibrillation, when the etiology of the cardiac arrest remains unclear after clinical evaluation **OR** ☐ Sudden death that remains unexplained after evaluation by the coroner and/or pathologist including toxicology, autopsy and cardiac autopsy **AND** ☐ Evaluation of the patient or case by a specialized medical genetics or cardiogenetics clinic

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Aortopathies (thoracic aortic aneurysm/dissection)

Medical genetics advice recommended if diagnosis < 18 years, dysmorphia and/or extracardiac damage

☐ Non-syndromic familial aortic aneurysm

ACTA2, COL1A1, COL3A1, FBN1, LOX, MYH11, MYLK, PRKG1, SMAD2, SMAD3, SMAD4, TGFB2, TGFB3, TGFBR1, TGFBR2	☐ Dilation of the thoracic aorta in the absence of systemic signs **AND** ☐ Family history of aortopathy in at least one 1er degree relative OR Age < 65 years in the absence of risk factors (hypertension, aortic atherosclerosis, etc.) **AND** ☐ Evaluation in a specialized medical genetics or cardiogenetics clinic

☐ Syndromic aortic aneurysm

ACTA2, CBS, COL1A1, COL3A1, COL5A1, COL5A2, EFEMP2, ELN, FBLN5, FBN1, FKBP14, FLNA, LOX, MYH11, MYLK, PLOD1, PRKG1, SKI, SLC2A10, SMAD2, SMAD3, SMAD4, TGFB2, TGFB3, TGFBR1	☐ Aortopathy with non-specific syndromic clinical picture OR ☐ Presence of clinical signs of connective tissue disease that do not meet the clinical criteria for Marfan syndrome OR ☐ Thoracic aortic dissection < 65 years in the absence of atherosclerosis at autopsy **AND** ☐ Evaluation in a specialized medical genetics or cardiogenetics clinic

☐ Classical Ehlers-Danlos syndrome

COL1A1, COL5A1, COL5A2	☐ Clinical diagnosis of classic Ehlers-Danlos syndrome **AND** ☐ Evaluation in a specialized medical genetics or cardiogenetics clinic

☐ Vascular Ehlers-Danlos syndrome

COL1A1, COL3A1	☐ Clinical diagnosis of vascular Ehlers-Danlos syndrome **AND** ☐ Evaluation in a specialized medical genetics or cardiogenetics clinic

☐ Loeys-Dietz syndrome

SMAD2, SMAD3, TGFB2, TGFB3, TGFBR1, TGFBR2	☐ Clinical signs suggestive of Loeys-Dietz syndrome OR ☐ Aortopathy with syndromic clinical picture suggesting Loeys-Dietz syndrome **AND** ☐ Evaluation in a specialized medical genetics or cardiogenetics clinic

☐ Marfan syndrome

FBN1	☐ Clinical diagnosis of Marfan syndrome OR ☐ Aortopathy with syndromic clinical picture suggesting Marfan syndrome **AND** ☐ Evaluation in a specialized medical genetics or cardiogenetics clinic

Additional gene(s) (valid for a gene present on the requisition form only) : _____

E- Pedigree (provide family tree and indicate known mutations) *OPTIONAL*

☐ Family tree will be provided as an attachment to this application

F- Health Care Professional Information: *MANDATORY*

By signing, 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 patient's representative/legal tutor the indicated genetic test and its purpose, limitations, risks and benefits and have satisfactorily answered all related questions, as indicated on the consent form.

Last name, first name: _____ License Number: _____

Hospital Center: _____ Fax : * _____ **for sending results*

_____ Signature: _____ Date: _____

☐ c.c. to referring physician, coroner, genetic counselor, laboratory or other :

Last name, first name: _____ License Number: _____

Hospital Center: _____ Fax : _____

_____

Warning: Any incomplete or non-compliant request may be refused