OPTILAB Montréal-CUSM

Centre universitaire de santé McGill — McGill University Health Centre

PATIENT STAMP OR LABEL HERE

Molecular Genetics Requisition - CMDL

Core Molecular Diagnostic Laboratory (CLIA #99D1042152)
1001 Decarie boul., E05.5051
Montreal, QC, H4A 3J1 Canada
Tel: 514-934-1934 x23383 / x23298 Fax: 514-843-1661

Patient Information:

Name (Last, First):*

Birth date (YYYY-MM-DD):* / /

Gender:* ☐ Male ☐ Female ☐ Unknown

Father's name:

Mother's name:

For Canada only:

Provincial Health Card #:*

Issuing Province:*

Referring Physician:

Name (Last, First):*

License #:*

Institution:*

e-mail address:*

Address:*

Tel:* Fax:*

(Fax # to send results)

Genetic counsellor:

Tel:* Fax:*

Signature:* Date:* / /

I acknowledge that the patient/guardian is aware of the benefits, limitations and risks associated with the requested test(s) and that I have obtained informed consent to perform genetic testing for this patient.
I authorize the laboratory to fax results to the number provided above.

Sample Information:

Collection date (YYYY-MM-DD):* / /

☒ Blood in EDTA (purple top tube): min 5 mL (2 mL for newborns)

☐ DNA: min 5 ug – Source:

☐ Amniotic fluid: min 10 mL

☐ Cultured amniocytes: 2 confluent T25 flasks

☐ Direct CVS: min 10 mg direct villi

☐ Cultured CVS: 2 confluent T25 flasks

☐ Tissue – Specify:

☐ Other – Specify:

CMDL - Laboratory use only:

Date - Time received:

/ /

h min

SAMPLE LABEL HERE

Sample type and # of tubes:

Patient #:

Test Requested *(write below and check box(es) on page 2)*:*

FH panel

Reason for Testing:*

☐ Confirm diagnosis (symptomatic)

☐ Carrier testing (for recessive conditions)

☐ Predictive testing (for dominant conditions)

☐ Prenatal testing (maternal sample required)

☐ Other – Specify:

Reason for expedited testing *(if applicable)*:

☐ Pregnancy (Gestational age: weeks on / /)

☐ Other reason – Specify:

Familial Variant Analysis:

For cases where a familial variant is known, please complete below and attach a copy of the proband's report. If the familial variant was not previously tested at the CMDL, please provide a sample from a family member known to be positive for this variant (i.e. positive control).

Gene (HGNC symbol):

Variant(s) (HGVS nomenclature):

CMDL Family number:

Name of proband:

Relationship to proband:

Pedigree/Clinical Information:

Please draw or attached pedigree and provide all relevant information.

Ethnicity:*

Ordering Checklist:

☐ Specimen tube labelled with at least two identifiers

☐ Completed test requisition (this form)

☐ Completed testing eligibility criteria form (if applicable)

☐ Consent form (or signature that consent form was obtained)

Required information. Samples will not be processed if information is missing.

PATIENT STAMP OR LABEL HERE

Name (Last, First):

Birthdate (YYYY-MM-DD): / /

CMDL - Laboratory use only:

Date - Time received:

/ / h

SAMPLE LABEL HERE

min

Sample type and # of tubes:

Patient #:

List of Tests:

For prenatal testing and analysis of familial variants, please include information on page 1.

Cystic fibrosis and *CFTR*-related conditions

- ☐ *CFTR* gene sequencing
- ☐ Known familial variant(s)

Angelman / Prader-Willi syndrome

Phenotype: ☐ Angelman ☐ Prader-Willi

- ☐ Methylation and deletion/duplication analysis
- ☐ UPD15 analysis *(please submit parental samples)*

Ashkenazi Jewish Carrier Screening

- ☐ *HEXA*, *ASPA*, *IKBKAP* recurrent variant analysis (Tay-Sachs disease, Canavan disease, familial dysautonomia)
- ☐ Known familial variant(s)

Hearing loss

- ☐ *GJB2* gene sequencing and *GJB6* recurrent deletions analysis
- ☐ Known familial variant(s)

Hidrotic ectodermal dysplasia (Clouston syndrome)

- ☐ *GJB6* gene sequencing
- ☐ Known familial variant(s)

MCAD deficiency

- ☐ *ACADM* gene sequecing
- ☐ Known familial variant(s)

***PAH* deficiency**

Phenotype: ☐ PKU ☐ Hyperphenylalaninemia

- ☐ *PAH* gene sequencing
- ☐ Known familial variant(s)

Hexosaminidase A deficiency (Tay-Sachs disease)

- ☐ *HEXA* gene sequencing (*please provide enzymatic testing results*)
- ☐ Known familial variant(s)

Male infertility

- ☐ Y-chromosome microdeletion analysis

***HBB*-related hemoglobinopathies**

Phenotype: ☐ HbS ☐ HbC ☐ HbE ☐ Other

β-thalassemia: ☐ Major ☐ Intermedia ☐ Trait/Minor

- ☐ *HBB* gene sequencing *(please provide haematological studies)*
- ☐ Known familial variant(s)

Huntington disease

- ☐ *HTT* trinucleotide repeat expansion analysis *(please provide clinical findings and family history)*

Hereditary Breast and Ovarian Cancer

- ☐ *BRCA1*, *BRCA2* French Canadian recurrent variant analysis
- ☐ *BRCA1*, *BRCA2, PALB2* French Canadian recurrent variant analysis
- ☐ *PALB2* recurrent variant analysis (c.2323C>T, p.Gln775Ter)
- ☐ *BRCA1*, *BRCA2* Ashkenazi Jewish recurrent variant analysis
- ☐ *BRCA1* Eastern European recurrent variant analysis
- ☐ *BRCA1* Greek recurrent variant analysis
- ☐ Known familial variant(s)

Methylmalonic acidemia

- ☐ *MMACHC* gene sequencing
- ☐ Known familial variant(s) (*MMACHC*, *LMBRD1*, *MMAA*, *MMAB*, *MCEE*, *CD320*, and *MUT* genes)

Familial hypercholesterolemia

- ☐ *LDLR*, *APOB*, *PCSK9* panel
- ☐ Known familial variant(s)

Microsatellite analysis

- ☐ Maternal cell contamination analysis *(please submit maternal sample)*
- ☐ Zygosity analysis
- ☐ Tissue matching analysis
- ☐ Postnatal QF-PCR for aneuploidy detection

Other test *(Please call first for information)*

- ☐ Specify: