CHU Sainte-Justine
Le centre hospitalier universitaire mère-enfant

Université de Montréal

MOLECULAR DIAGNOSTICS LABORATORY - Molecular Genetics

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3175, Côte Sainte-Catherine, Montréal, Q C, H3T 1C5, 514-345-4931

ATTENTION: Shaded bloc required

F-583A # INV.:30004403
BM Génétique ang. (RÉV.2014-05)

REFERRING PHYSICIAN: _____

Licence No:_____

Date:_____ Time:_____

Outside client: Please indicate for results and billing.

Name: _____

Department: _____

Address: _____

_____

Phone:_____ Fax:_____

Identification of patient

Name: _____ First name: _____

Sex: F ☐ M ☐

File or # Health care no.: _____

Date of birth: _____

The signature of the referring physician is obligatory.
I certify that I have explained the following to the patient: the nature of the requested test, its benefits, limitations and potential risk for the patient and his or her family. I certify that I have obtained signed informed consent for the test from the patient or his or her legal guardian.

_____, _____, _____, _____
NAME | No de pratique | Signature | Date

TYPE OF SAMPLE

☐ Blood: 5 ml (EDTA, purple topped tube) _____tubes
☐ Amniocytes
☐ Chorionic biopsy ☐ DNA
☐ Filter paper blood specimen (ARSACS, CGD, COX-SLSJ and HHH)
☐ Other: _____

TEST REQUESTED

☐ Diagnosis patient ☐ symptomatic or ☐ at risk
☐ Carrier detection
☐ Foeto-maternal contamination testing
☐ Pregnacy
Specify diagnosis:_____
☐ LMP: _____Week of pregnancy: _____
☐ Other: _____

TEST (EDTA, purple topped tube)

☐ Spinal Muscular Atrophy (SMA, SMN1)
☐ Friedreich's Ataxia (FXN)
☐ Craniosynostoses
  ☐ Muenke Synd. Mutation P250R (FGFR3)
  ☐ Apert Synd. P253R & S252W (FGFR2)
☐ Duchenne/Becker Muscular Distrophy in DMD, BMD
  ☐ Detection of a known deletion in males
  ☐ Family Linkage Study
☐ Familial Hypercholesterolemia (LDLR)
Clinical state: ☐ Homozygous ☐ Heterozygous
Tests 1, 2 and 3 will be performed, in that order, until 2 (homozygote) or 1 (heterozygote) mutant allele(s) is/are identified. Only those tests performed will be billed.
  ☐ 1. Deletions of 15 kb and 5 kb
  ☐ 2. W66G
  ☐ 3. Group of rare mutations: C152W, 681ins7, E207K, R329X, Y354C, C347R, C646Y and Y468X.
  ☐ 4. A specific mutation among the group of rare mutations
  Specify: _____
☐ Out of province testing Specify:_____

MUTATIONS PREVALENT IN QUEBEC (EDTA, purple topped tube)

☐ Autosomal Recessive Spastic Ataxia of the Charlevoix-Saguenay (ARSACS): mutations 6594delT and 5254 C>T in SACS
☐ North American Indian Childhood Cirrhosis (NAIC): mutation R565W in CIRH1A
☐ Congenital Disorders of Glycosylation (CGD-Ib): mutation R295H in MPI
☐ Cytochrome C Oxidase Deficiency, Saguenay-Lac St-Jean (COX-SLSJ)
  ☐ Mutations A354V and C1277Xdel18 in (LRPPRC)
  ☐ Mutation A354V ONLY
☐ Cree Leukoencephalopathy (CLE): mutation R195H in (eIF2B5)
☐ Cree Encephalitis (CL): mutation R164X (TREX1)
☐ Syndrome HHH: mutation F188del (SLC 25A15)
☐ Lipoprotein Lipase Deficiency (LPL): mutations Pro207Leu and Gly188Glu

FAMILY INFORMATION

Relation to affected individual _____
Name of affected individual _____
Were other samples sent from this family?_____
Family name_____

Samples should be sent to

Laboratoire central
CHU Sainte-Justine
2nd floor, bloc 9
3175, Côte Sainte-Catherine
Montréal (Québec) H3T 1C5
Phone: 514 345-4931 extension 6229
Fax: 514 345-2339

PEDIGREE

Sampling performed by: Date : 20 ____ - ____ - ____ Time :