ATTENTION: Zone grise obligatoire

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, QC, H3T 1C5, 514-345-4642

Requesting Institution/Unit : _____

Address : Civic number _____ Street _____

City _____ Province/Country _____

Postal code _____

Phone : _____ FAX: _____

Referring physician : _____ Licence No : _____

Sampling Date : _____ **Time** _____

Sampled by : _____

Patient information

Last name: _____ First name: _____

Gender: F ☐ M ☐

Medical records number / Provincial health number: _____

Date of birth: _____

Clinical information: _____

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.

SAMPLE TYPE

POSTNATAL

- ☐ EDTA Blood : 5ml (EDTA vial) _____ # of tubes
- ☐ Tissue : _____
- ☐ DNA : _____ (specify origin aside from blood)
- ☐ Filter (Guthrie) card filled with EDTA blood (accepted only for LRPPRC and SACS analysis

PRENATAL (# of weeks :_____)

- ☐ DNA : _____(specify origin)
- ☐ Amniocytes
- ☐ Chorionic villi
- ☐ Cultured chorionic villi (T25 flask(s))
- ☐ Maternal cell contamination analysis : _____ (specify indication)

INDICATION FOR TESTING

- ☐ Diagnostic (symptomatic patient)
- ☐ Carrier status determination
- ☐ Prenatal diagnosis

FAMILIAL INFORMATIONS

Index case name : _____

Relationship with index case : _____

Additional sample sent for this family : _____

Family # : _____

TESTS

- ☐ Achondroplasia-Hypochondroplasia (FGFR3: sequencing of exons 10 and 13)
- ☐ Congenital Disorder of Glycosylation, Type Ib (MPI:p.R295H)
- ☐ Craniosynostoses:
  - ☐ Muenke Syndrome (FGFR3:p.P250R)
  - ☐ Apert Syndrome (FGFR2:p.P253R and FGFR2:p.S252W)
- ☐ Cree Population Frequent Disorders :
  - ☐ Cree Encephalitis TREX1: p.R164X
  - ☐ Cree Leukoencephalopathy: eIF2B5: p.R195H
- ☐ DNA extraction (indication): _____
- ☐ Duchenne/Becker Muscular Dystrophies, DMD[1] : Deletion/duplication
- ☐ Familial Hypercholesterolemia, LDLR :
  Phenotype (mandatory) ☐ Homozygote ☐ Heterozygote
  - ☐ 15Kb and 5Kb Deletions
  - ☐ Frequent Mutations Panel[3]
- ☐ Friedreich Ataxia (FXN intron 1 GAA expansion)[1]
- ☐ HHH Syndrome (SLC25A15 : p.F188del)
- ☐ Leigh Syndrome, French Canadian Variant:
  - ☐ LRPPRC: p.A354V
  - ☐ LRPPRC: p.C1277Xdel8
- ☐ Lipoprotein lipase deficiency (LPL:p.P207L and LPL:p.G188E)
- ☐ Mitochondrial DNA[1]:
  - ☐ Sequencing
  - ☐ Deletion/duplication
  - ☐ Known mutation[2]
- ☐ North American Indian Childhood Cirrhosis (UTP4 (CIRH1A):p.R565W)
- ☐ Rett Syndrome, MECP2[1] :
  - ☐ Sequencing (coding exons)
  - ☐ Deletion/Duplication
  - ☐ Known Mutation[2]
- ☐ Spastic Ataxia, Charlevoix-Saguenay Type (ARSACS)
  - ☐ SACS: c. 6594delT
  - ☐ SACS: c. 5254 C>T
- ☐ Spinal Muscular Atrophy (SMN1 exon 7 copy number)[1] :
- ☐ Thanatophoric Dysplasia type I and II (FGFR3 : sequencing of exons 7, 10, 15 and 19)
- ☐ Other analysis :_____

1 : This test requires an EDTA blood sample. Extracted DNA will not be accepted.
2 : Please include the report from an affected/carrier relative when requesting known mutation testing
3 : Please refer to our website for additional information regarding specific mutations tested : https://www.chusj.org/fr/Labotest

SAMPLE RECEPTION

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PEDIGREE