ATTENTION: Zone grise obligatoire

CHU Sainte-Justine
Le centre hospitalier universitaire mère-enfant
Université de Montréal

MOLECULAR DIAGNOSTICS LABORATORY - Molecular Genetics

MOLECULAR DIAGNOSTICS LABORATORY - Molecular Genetics

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 : 2-5ml (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)
- ☐ Cultured amniocytes ☐ Amniotic fluid
- ☐ 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 (inform the lab in advance)
- ☐ For future analysis

FAMILIAL INFORMATIONS

Index case name : _____

Relationship with index case : _____

Additional sample sent for this family : _____

Family # : _____

TESTS

- ☐ Achondropalsia-Hypochondroplasia (FGFR3 sequencing: NM_000142.4, exons 9 & 12)
- ☐ Mitochondrial DNA[1]:
  - ☐ Sequencing and deletion detection
  - ☐ Known mutation[2]: _____
- ☐ Spinal Muscular Atrophy[1] :
  - ☐ Deletion/duplication exon 7 (SMN1)[3]
  - ☐ Family linkage analysis
- ☐ Friedreich's ataxia (FXN intron 1 GAA expansion)[1]
- ☐ Spastic Ataxia, Charlevoix-Saguenay Type (ARSACS) :
  - ☐ SACS: c. 6594delT
  - ☐ SACS: c. 5254 C>T
- ☐ North American Indian Childhood Cirrhosis (UTP4 (CIRH1A):p.R565W)
- ☐ Craniosynostosis:
  - ☐ Muenke Syndrome (FGFR3: p.P250R)
  - ☐ Apert Syndrome (FGFR2: p.P253R et p.S252W)
- ☐ Lipoprotein lipase deficiency (LPL: p.P207L and LPL: p.G188E)
- ☐ Thanatophoric Dysplasia type I and II (FGFR3 sequencing (NM_000142.4) exons 7, 9, 14 et 18)
- ☐ Duchenne/Becker Muscular Dystrophies, DMD[1] (deletion/duplication)
- ☐ Tyrosinemia type I : ☐ FAH sequencing
  - ☐ Known FAH mutation[2] : _____
- ☐ Mucolipidosis type II : GNPTAB c.3503_3504delTC
- ☐ Familial hypercholesterolemia, LDLR:
  Phenotype (mandatory): ☐ Homozygote ☐ Heterozygote
  - ☐ 15Kb and 5Kb deletions
  - ☐ Frequent Mutations Panel[4]
  - ☐ Specific LDLR mutation[2]: _____
- ☐ Cree Population Frequent Disorders :
  - ☐ Cree Encephalitis: TREX1: p.R164X
  - ☐ Cree Leukoencephalopathy: eIF2B5: p.R195H
- ☐ Leigh Syndrome, French Canadian Variant :
  - ☐ LRPPRC: p.A354V
  - ☐ LRPPRC: p.C1277Xdel8
- ☐ HHH Syndrome (SLC25A15: p.F188del)
- ☐ Rett Syndrome, MECP2 :
  - ☐ Sequencing (coding exons)
  - ☐ Deletion/duplication
  - ☐ Known mutation[2]: _____
- ☐ Congenital Disorder of Glycosylation, type 1b: MPI:p.R295H
- ☐ DNA extraction: (mandatory indication): _____
- ☐ Other analysis: _____

1 : This test requires an EDTA blood sample. Extracted DNA will not be accepted.
2 : Please include the lab report of an affected/carrier relative when requesting known mutation testing; the mutations should be described according to the standard HGVS nomenclature.
3 : The copy number of SMN2 exon 7 will be also included in the result report only for the affected cases with zero SMN1 copies.
4 : Please refer to our website for additional information regarding specific mutations tested : https://www.chusj.org/en/Labotest

SAMPLE RECEPTION

Laboratoire Central
CHU Sainte-Justine
2nd floor, bloc 9
3175, Côte Sainte-Catherine
Montréal (Québec) H3T 1C5
Phone : 514-345-4642 Fax : 514-345-2339

PEDIGREE