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 : 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

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PEDIGREE