ATTENTION: Filling of shaded zones is required

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

☐ Blood : 2-5 ml EDTA _____ (number of vials)

☐ Biological fluid (source/mL): _____

☐ Muscular tissur (30-50 mg) : _____

☐ Other tissu (source / mg) : _____

☐ Cultured fibroblasts (1-2 x T23, 80% confluency)

☐ DNA (≥3 µg) : _____
(quantity, source, lab no)

PRENATAL (# of weeks: _____)

☐ DNA (≥1 µg) : _____
(quantity, source, lab no)

☐ Amniotic fluid (minimum 10 mL):

☐ Amniocytes (2 X T25 - 80% confluency)

☐ Cultured chorionic villi (CVS) (2 X T25 - 80% confluency)

☐ Direct chorionic villi (CVS, minimum 10 mg)

* Exclusion of maternal cell contamination is strongly advised for every prenatal test.

INDICATION FOR TESTING

☐ Diagnostic (symptomatic patient)

☐ Neonatal screening confirmation (PQDNS)

☐ Carrier status determination

☐ Population screening

☐ Predictive testing

☐ Prenatal diagnosis (please advise laboratory ahead of time). Note that prenatal diagnosis and exclusion of maternal cell contamination of the fetal specimen requires a sample from the mother.

☐ For future analysis

FAMILIAL INFORMATIONS

☐ Index case name : _____

☐ Relationship with index case : _____

☐ Additional sample sent for this family : _____

☐ Family no : _____

PEDIGREE

Include a pedigree of the family and relevant clinical information

SAMPLE RECEPTION

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

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Last name: _____ First name: _____

Gender: F ☐ M ☐

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Date of birth: _____

TESTS

☐ Achondroplasia-Hypochondroplasia (FGFR3) ACHH
(Panel of common mutations) [1]

☐ Mitochondrial genome (SNG): ADNM1
☐ Sequencing and deletion detection
☐ Known variation : _____
(HGVS nomenclature)

☐ Spinal muscular atrophy (dél/dup exon7-SMN1) [2] ASQT
☐ Copy number of SMN2 [3]

☐ Friedreich ataxia (FXN intron 1 GAA expansion) FRIE2

☐ North American Indian Childhood Cirrhosis (UTP4) RMCNA
(CIRH1A): p.R565W)

☐ Craniosynostosis:
☐ Apert Syndrome (FGFR2:p.P253R et p.S252W) RMFG2
☐ Muenke Syndrome (FGFR3:p.P250R) RMFG3

☐ Thanatophoric Dysplasia type I and II (FGFR3) TD12
(Panel of common mutations) [1]

☐ Duchenne/Becker Muscular Dystrophies (DMD) [2] DMD
(deletion/duplication)

☐ FMR1 CGG Expansion FRAGI
☐ Fragile X Syndrome (FRAXA)
☐ Fragile X-associated premature ovarian insufficiency (FXPOI)
☐ Fragile X-associated Tremor/Ataxia(FXTAS)

☐ Familial Hypercholesterolemia, LDLR :
☐ 15Kb and 5Kb Deletions HFDEL
☐ Panel of common mutations [1] HFMUT
☐ Specific LDLR variation : _____ BMSSA
(HGVS nomenclature)

☐ Lipoprotein Lipase Deficiency (LPL: p.P234L et p.G215E) RMLPL
(Historically known : p.P207L et p.G188E)

☐ Mucolipidosis II (GNPTAB: c.3503_3504delTC) GNPTA

☐ Analysis of familial variations BMSSA
Please attach the test report for an affected family member/carrier for any variation to be analysed in case it was not previously tested in the laboratory, ans include a sample from a family member in whom the variation was identified (a family positive control)

Name of the index case : _____
Familial relationship with index case : _____
Gene (HGVS nomenclature) : _____
Variation (HGVS nomenclature) : _____

☐ Maternal Cell Contamination : _____ VCFM5
(specify indication)

☐ Other analysis : _____

☐ HHH Syndrome (SLC25A15: p.Phe188del) RMHHH

☐ Rett Syndrome (MECP2) : RETT
☐ Sequencing (coding exons + splice junctions)
☐ Deletion/Duplication [2]
☐ Specific variation : _____
(HGVS nomenclature)

☐ Tyrosinemia type I : FAH
☐ FAH Sequencing (coding exons + splice junctions)
☐ Specific variation : _____
(HGVS nomenclature)

Population Screening

☐ Cree Population Frequent Disorders :
☐ Cree Encephalitis (TREX1: p.R164X) CREE2
☐ Cree Leucoencephalopathy (ELF2B5: p.R195H) CREL2

☐ Congenital disorder of glycosylation CDG1b (MPI:p.R295H) RMMPI

☐ Four recessive Diseases of Saguenay-Lac-Saint-Jean
☐ COX-SLSJ (LRPPRC): c.1061C>A (p.A345V)
☐ Tyrosinemia 1 (FAH): c.1062+5G>A (IVS12+5G>A)
☐ NSM/ACC (SLS12A6): c.2436delG (p.T813Pfs)
☐ ARSACS (SACS):
☐ c.8844delT (p.I2949fs) (also known : 6594delT)
☐ c.7504C>T (p.R2502Ter) (also known : 5254C>T)

Next-Generation Sequencing (NGS) Panels - RQDM*

☐ Noonan/Rasopathies Panel NOONA
☐ Mitochondrial Disorder Panel (nuclear) MINUC
☐ Muscular Diseases (myopathies)
☐ Myopathy Panel (global muscular disease) MANUS
☐ Muscular Dystrophy Panel DYMUS
☐ Malignant Hyperthermia Panel HYPMA
☐ Congenital Myasthenia Panel MYAST
☐ Rhabdomyolysis Panel BMRHA

☐ Intellectual Disability (DI)/GDD DINTE
Trio (probant/mother/father)
Comment(s) : _____

☐ Other NGS Analysis : _____

Checklist for health professionals and Blood Drawing Centers :

☐ Are included the details of the testing indication and clinical information.

☐ Form of complementary clinical information for NGS sequencing.

☐ Consent for NGS sequencing

- Incomplete requisition from unauthorised health professionals and inadequate samples (ex. hemolysed, inappropriately labelled) will be deemed not conformed and will be rejected.
- For shipping conditions, NGS clinical and consent forms please consult the website for each individual test : //www.chusj.org/fr/Labotest

* It is the responsability of the prescriber to check beforehand the availability of the tests.
1. Please refer to the website for details of the mutations tested : https://www.chusj.org/fr/Labotest.
2. This test requires a fresh blood sample.
3. SMN2 exon-7 copy number is provided for SMA affected individuals only.