

Requesting Institution/Unit : _____ Address : <u>Civic number</u> _____ <u>Street</u> _____ <u>City</u> _____ <u>Province/Country</u> _____ <u>Postal code</u> _____ 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: _____
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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

PRENATAL (# of weeks:)

- ☐ 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 μ g) : _____
(quantity, source, lab no)

- ☐ DNA ($\geq 1 \mu\text{g}$) : _____
(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

FAMILIAL INFORMATION

- ☐ 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

- ☐ 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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TESTS

- ☐ Achondroplasia-Hypochondroplasia (FGFR3) ACHH
(Panel of common mutations) ¹
- ☐ Mitochondrial genome (SNG): ADN1
☐ 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 FRAX1
☐ 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 : _____
 (HGVS nomenclature) BMSSA
- ☐ Lipoprotein Lipase Deficiency (LPL: p.P234L et p.G215E) RMLPL
(Historically known : p.P207L et p.G188E)
- ☐ Mucopolipidosis 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 (TRES1: 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
 Trio (proband/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 responsibility 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.