

MOLECULAR DIAGNOSTICS LABORATORY - Molecular Genetics

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3175, Côte Sainte-Catherine, Montréal, QC, H3T 1C5, 514-345-4642

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 <input type="checkbox"/> M <input type="checkbox"/> 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	PRENATAL (# of weeks : _____)
<input type="checkbox"/> EDTA Blood : 5ml (EDTA vial) _____ # of tubes <input type="checkbox"/> Tissue : _____ <input type="checkbox"/> DNA : _____ (specify origin aside from blood) <input type="checkbox"/> Filter (Guthrie) card filled with EDTA blood (accepted only for LRPPRC and SACS analysis)	<input type="checkbox"/> DNA : _____ (specify origin) <input type="checkbox"/> Amniocytes <input type="checkbox"/> Chorionic villi <input type="checkbox"/> Cultured chorionic villi (T25 flask(s)) <input type="checkbox"/> Maternal cell contamination analysis : _____ (specify indication)
INDICATION FOR TESTING	FAMILIAL INFORMATIONS
<input type="checkbox"/> Diagnostic (symptomatic patient) <input type="checkbox"/> Carrier status determination <input type="checkbox"/> Prenatal diagnosis	Index case name : _____ Relationship with index case : _____ Additional sample sent for this family : _____ Family # : _____
TESTS	
<input type="checkbox"/> Achondroplasia-Hypochondroplasia (FGFR3: sequencing of exons 10 and 13) <input type="checkbox"/> Congenital Disorder of Glycosylation, Type Ib (MPI:p.R295H) <input type="checkbox"/> Craniosynostoses: <input type="checkbox"/> Muenke Syndrome (FGFR3:p.P250R) <input type="checkbox"/> Apert Syndrome (FGFR2:p.P253R and FGFR2:p.S252W) <input type="checkbox"/> Cree Population Frequent Disorders : <input type="checkbox"/> Cree Encephalitis TREX1: p.R164X <input type="checkbox"/> Cree Leukoencephalopathy: eIF2B5: p.R195H <input type="checkbox"/> DNA extraction (indication): _____ <input type="checkbox"/> Duchenne/Becker Muscular Dystrophies, DMD ¹ : Deletion/duplication <input type="checkbox"/> Familial Hypercholesterolemia, LDLR : Phenotype (mandatory) <input type="checkbox"/> Homozygote <input type="checkbox"/> Heterozygote <input type="checkbox"/> 15Kb and 5Kb Deletions <input type="checkbox"/> Frequent Mutations Panel ³ <input type="checkbox"/> Friedreich Ataxia (FXN intron 1 GAA expansion) ¹ <input type="checkbox"/> HHH Syndrome (SLC25A15 : p.F188del) <input type="checkbox"/> Leigh Syndrome, French Canadian Variant: <input type="checkbox"/> LRPPRC: p.A354V <input type="checkbox"/> LRPPRC: p.C1277Xdel8 <input type="checkbox"/> Lipoprotein lipase deficiency (LPL:p.P207L and LPL:p.G188E)	<input type="checkbox"/> Mitochondrial DNA ¹ : <input type="checkbox"/> Sequencing <input type="checkbox"/> Deletion/duplication <input type="checkbox"/> Known mutation ² <input type="checkbox"/> North American Indian Childhood Cirrhosis (UTP4 (CIRH1A):p.R565W) <input type="checkbox"/> Rett Syndrome, MECP2 ¹ : <input type="checkbox"/> Sequencing (coding exons) <input type="checkbox"/> Deletion/Duplication <input type="checkbox"/> Known Mutation ² <input type="checkbox"/> Spastic Ataxia, Charlevoix-Saguenay Type (ARSACS) <input type="checkbox"/> SACS: c. 6594delT <input type="checkbox"/> SACS: c. 5254 C>T <input type="checkbox"/> Spinal Muscular Atrophy (SMN1 exon 7 copy number) ¹ : <input type="checkbox"/> Thanatophoric Dysplasia type I and II (FGFR3 : sequencing of exons 7, 10, 15 and 19) <input type="checkbox"/> 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	PEDIGREE
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