OPTILAB Montréal-CUSM

LISTE DES ANALYSES HORS-QUEBEC
List for testing done out of Québec
LABORATOIRE CLINIQUES CUSM
MUHC CLINICAL LABS

Centre universitaire de santé McGill — McGill University Health Centre

Appelation approuvée de l'analyse ou maladie associée [avec numéro de phénotype MIM] / Approved test name or Associated Disease [with MIM Phenotype Number]	**Code Test** / Test Code **(Dictionnaire MSSS)**	**SYMBOL HGNC DU GÈNE et NOM OMIM DU GÈNE** / HGNC Gene Symbol and OMIM Gene Number	**RÈGLES DE MANUTENTION** / Handling Conditions	**LABORATOIRE DÉSIGNÉ** / Designated laboratory	**RENSEIGNEMENTS SUR L'ANALYSE** / Test details	**MÉTHODE** / Methode	**NOTE PARTICULIÈRE** / Special comment	**Date de la révision** / Date of revision
1,5-anhydroglucitol (GlycoMark) [500115] [GLYC]	1465		Gold Top or Red Top (Serum) or Lavender (plasma). Centrifuge and aliquot serum to transport tube within one hour of collection. Store and ship forzen. Stable at -20°C for 14 days.	LabCorp/Dynacare (https://www.labcorp.com/)		Enzymatic, colorimetric assay.		10-Jan-21
15 Mold Panel	1465			In-Common Laboratories	Done at Alletess			12-Mar-20
2,3-Dinor-11Beta-Prostaglandin F2 Alpha, Urine [23BPG]	1		Urine. No preservative. Collect for 24-h or random sample. Refrigeate 4°C during collection. Store frozen.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	systemic mastocytosis; mast cell activation disorders including systemic mastocytosis; Replaces 11 BETA-PROSTAGLANDIN F2 ALPHA	LC-MS/MS	May also order Leukotriene E4	4-Mar-21
5-Methyltetrahydrofolate (CSF) [NC01]			Collect 1 milliliter of CSF. Spin sample if contaminated with red blood cells and freeze the clear CSF at -80°Celsius. Store frozen at -80°Celsius.	MNG Laboratories (www.medicalneurogenetics.com)/Dynacare	Cerebral folate deficiency			14-May-20
5'Nucleotidase	4		Serum. Stable 4h at RT, 7 days at 4°C or 14 days at -20°C.	Dynacare		enzymatic	Requires preappova as tets is of limited utility	22-Oct-20
a1-Acid glycoprotein [A1AGP]			Serum. Freeze if > 72 hours	In-Common Laboratories	Other names: Orosomucoid, Acid Glycoprotein, Alpha 1 Acid Glycoprotein	Nephelometry		6-Sep-18
ABCC6 Gene Analysis in Pseudoxanthoma Elasticum [2642]	1323	ABCC6 (603234)	Lavender top (EDTA)	GeneDx (www.genedx.com)	Pseudoxanthoma Elasticum (264800)	Tier 1: sequencing for commom mutations [2641]; Tier 2: full gene sequencing [2642]; del/dupl (MLPA) [906]		9-Dec-19
Acetaminophen [ACETA]			Serum	In-Common Laboratories	quantitation; to determine clearance			6-Sep-18
Acetylcholine Receptor Antibodies by RIPA (AChR Ab, 91020/91021 only)	18		Serum (Gold SST) or CSF. Store at -20°C.	BC Neuroimmunology (bcneuro.ca)	myasthenia gravis	RIPA		31-Oct-20
Acetylcholine Receptor Antibodies with reflex to Muscle Specific Tyrosine Kinase Antibodies (MuSK Ab)	18		Serum (Gold SST) or CSF. Store at -20°C.	BC Neuroimmunology (bcneuro.ca)	myasthenia gravis	RIPA		31-Oct-20
Acyclovir, Serum/Plasma [FACY]			Red Top or Lavender Top (NOT SST)	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)		LC-MS		
ADAR Full Gene Sequencing Analysis [MOL309]		ADAR (146920)	Lavender Top (EDTA)	MNG Laboratories (www.medicalneurogenetics.com)/Dynacare	Aicardi-goutieres syndrome 6 (615010); Dyschromatosis symmetrica hereditaria	Sanger sequencing	NGS Panel available	14-May-20
Adenosine Deaminase, Pleural Fluid [FADPF]	11		Must be frozen within 24 hours of collection. No freeze/thaw cycles.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)		Ultraviolet Spectrophotometry		11-Feb-21
Adiponectin [FADIO]			Draw blood in a plain red-top or Gold SST, serum gel tube(s) is acceptable. Spin down and send 1 mL of serum refrigerated in a plastic vial. Store frozen. Overnight fasting is required.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)		ELSIA	HIC: RUO	31-Oct-20
ADmark® Phospho-Tau/Total-Tau/Ab42 CSF Analysis & Interpretation (Symptomatic) [177]	596		Cerebrospinal Fluid (CSF), 2 mL. POLYPROPYLENE TUBES ONLY.Store at -20°C.	Athena Diagnostics (www.athenadiagnostics.com)	Alzheimer Disease (104300)	ELISA	Alernate site : MML	3-Aug-18
Adrenal hyperplasia due to 21-hydroyxlase deficiency (201910)		CYP21A2 (613815)	Lavender Top (EDTA)	Alberta Mol Dx Laboratory (Calgary)		sequencing & MLPA		26-Sep-18
Afirma GEC	1680		Fine needle aspirate	Groupe TMTC (https://grouptmtc.com)				21-Jan-21
Agammaglobulinemia Panel, Sequencing (9 Genes) and Deletion/Duplication (6 Genes) [2011151]	999	BLNK, BTK, CD79A, CD79B, IGHM, IGLL1, LRRC8A, PIK3R1, SH2D1A	Lavender Top (EDTA)	ARUP Laboratories (www.aruplab.com)	Agammaglobulinemia 1, Autosomal Recessive ◦ X-Linked Agammaglobulinemia ◦ SH2D1A-Related Lymphoproliferative Disease, X-Linked ◦ Agammaglobulinemia 2, Autosomal Recessive ◦ Agammaglobulinemia 3, Autosomal Recessive ◦ Agammaglobulinemia 4, Autosomal Recessive ◦ Agammaglobulinemia 5, Autosomal Dominant ◦ Agammaglobulinemia 6, Autosomal Recessive ◦ Agammaglobulinemia 7, Autosomal Recessive	Massively Parallel Sequencing/Exonic Oligonucleotide-based CGH Microarray		
AGXT Gene, Full Gene Analysis [AGXMS]		AGXT (604285)	Lavender Top (EDTA)	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	Hyperoxaluria, primary, type 1 (259900)	Sanger sequencing and MLPA		
Aicardi-Goutieres Syndrome (NextGen Sequencing Panel and Copy Number Analysis; 6 Genes) [NGS344]		ADAR (601059); ALDH7A1 (107323); RNASEH2A (606034); RNASEH2B (610326); RNASEH2C (610330); SAMHD1 (606754); TREX1 (606609)	Lavender Top (EDTA)	MNG Laboratories (www.medicalneurogenetics.com)/Dynacare	Aicardi-Goutieres Syndrome	NextGen Sequencing		14-May-20

Appelation approuvée de l'analyse ou maladie associée [avec numéro de phénotype MIM] / Approved test name or Associated Disease [with MIM Phenotype Number]	**Code Test** / Test Code **(Dictionnaire MSSS)**	**SYMBOL HGNC DU GÈNE et NOM OMIM DU GÈNE** / HGNC Gene Symbol and OMIM Gene Number	**RÈGLES DE MANUTENTION /** Handling Conditions	**LABORATOIRE DÉSIGNÉ** / Designated laboratory	**RENSEIGNEMENTS SUR L'ANALYSE** / Test details	**MÉTHODE /** Methode	**NOTE PARTICULIÈRE /** Special comment	**Date de la révision /** Date of revision
ALA Dehydratase [ALAD]			Green Top (Na heparin), 4°C only.Do not freeze. Send M-W only.	In-Common Laboratories	Aminolevulinic Acid Dehydratase Deficiency Porphyria (612740)		Sent to USA	6-Sep-18
Albendazole	1917		RED TOP ONLY. Serum without gel, centrifuge immediately after collection, freeze and ship with dry ice	Inselspital Bern Freiburgstrasse 10 Zentrum für Labormedizin Zentrale Annahme INOF / z. Hd. Y. Aebi CH-3010 Bern, Switzerland	Measures albendazole sulfoxide due to 1st pass effcets			6-Apr-21
Albright's hereditary Osteodystrophy (103580)	440	GNAS (139320)	Lavender Top (EDTA) 2 x 4 mL	Fulgent Genetics (fulgentdiagnostics.com)	Albright's hereditary Osteodystrophy (103580)	NextGen Sequencing		28-Feb-20
Alexander Disease via the GFAP Gene [3775]		GFAP (137780)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Alexander disease (203450)	Sanger sequencing		17-Mar-17
Alpha aminoadipic semialdehyde (Urine) [MET20]	12	Alpha-aminoadipic semialdehyde	Collect 1 milliliter urine (random) and freeze at -20°Celsius. Store frozen at –20°Celsius and ship frozen.	MNG Laboratories (www.medicalneurogenetics.com)/Dynacare	Pyridoxine-dependent epilepsy (266100)	MS/MS		14-May-20
Alpha aminoadipic semialdehyde (Whole Blood) [AASA]	12	Alpha-aminoadipic semialdehyde	Specimen: Whole Blood Container(s): Dark Green/Sodium Heparin or Lt. Green/Lithium Heparin Tube. **Reject due to**: If sample is not spun and frozen within 1 hour of collection. 48 hr storage at -20 C is acceptable. Store **plasma** at -70°C.	Seattle Children's Hospital. http://seattlechildrenslab.testcatalog.org/	Pyridoxine-dependent epilepsy (266100)			27-Jan-20
ALPHA-1-ANTITRYPSIN DEFICIENCY (613490)	1395	SERPINA1 (104400)	Lavender Top (EDTA)	Attn: Norine Freedman or Lynn Coleman Special Chemistry Laboratory St Paul's Hospital 1081 Burrard Street Vancouver, B.C. V6Z 1Y6. Contact Dr. A. Mattman BEFORE sending sample.	ALPHA-1-ANTITRYPSIN DEFICIENCY (613490)	Sanger sequencing		
alpha-actin (skeletal muscle form)-related myopathy via the ACTA1 gene [358]		ACTA1 (102610)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	nemaline myopathy (NEM3; OMIM #161800) and congenital fiber-type disproportion (CFTD1; OMIM #255310)			
Alpha-aminoadipic semialdehyde (CSF) [NC08]	12	Alpha-aminoadipic semialdehyde	Collect 1 milliliter of CSF. Spin sample if contaminated with red blood cells and freeze the clear CSF at -80°Celsius. Store frozen at -80°Celsius.	MNG Laboratories (www.medicalneurogenetics.com)/Dyncare	Pyridoxine-dependent epilepsy (266100)	MS/MS	Seattle for Heparin plasma	14-May-20
Alpha-Subunit Pituitary Tumor Marker, Serum [APGH]	55		Red Top	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	Alternate name: Alpha Glycoprotein Subunit			26-Sep-18
Alpha-Thalassemia (604131)	826	HBA1 (141800); HBA2 (141850)	Lavender Top (EDTA)	Molecular Genetics Laboratory - McMaster University Medical Centre	targeted mutations at HSJ	Sanger sequencing & del/dupl		
Alport Syndrome NGS Panel	785	COL4A3, COL4A4, COL4A5 COL4A6	Lavender Top (EDTA)	Fulgent Genetics (fulgentdiagnostics.com)		NextGen Sequencing		3-Feb-20
ALZHEIMER'S DISEASE, FAMILIAL VIA THE APP GENE (604)		APP (104760)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)		Sanger sequencing	For known mutations only. Otherwise use Fulgent panel.	
ALZHEIMER'S DISEASE, FAMILIAL VIA THE PSEN1 GENE [1414]		PSEN1 (104311)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Alzheimer Disease, Type 3 (607882); Frontotemporal Dementia (600274)	Sanger sequencing	For known mutations only. Otherwise use Fulgent panel.	
ALZHEIMER'S DISEASE, FAMILIAL VIA THE PSEN2 GENE (1415)		PSEN2 (600759)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Alzheimer Disease, Type 4 (606889)	Sanger sequencing	For known mutations only. Otherwise use Fulgent panel.	
AMELOGENESIS IMPERFECTA VIA THE DLX3 GENE [1603]		DLX (600525)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Tricho-Dento-Osseous Syndrome (190320); Amelogenesis Imperfecta, Type IV (104510)	Sanger sequencing		
Aminolevulinic Acid Dehydratase (ALAD), Whole Blood [ALAD]			Green Top, 4°C only	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	Aminolevulinic Acid Dehydratase Deficiency Porphyria (612740)			31-Oct-20
AMYLOID PROTEIN ID, PAR, LC MS/MS [AMPIP]	1123		Paraffin section	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)		LC-MS/MS	ADD CHARGE FOR MICRODISSECTION	6-Jul-18
Amyotrophic Lateral Sclerosis and Frontotemporal Dementia Panel [10359]	853	ANG ANXA1APP ARHGEF28 ATXN2 C9orf72 CFAP410 CHCHD10 CHMP2B DAO DCTN1 ERBB4 FIG4 FUS GRN HNRNPA1 HNRNPA2B1 ITM2B KIF5A MAPT MATR3 MOBP NEFH 9 NEK1 OPTN PFN1 PSEN1 PSEN2 SETX SOD1 SQSTM1 TAF15 TARDBP TBK1 TREM2 TUBA4UBQLN2 UNC13A VAPB VCP	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Frontotemporal Dementia And/Or Amyotrophic Lateral Sclerosis (105550), Amyotrophic Lateral Sclerosis Type 6 (608030); Amyotrophic Lateral Sclerosis Type 9 (611895); Amyotrophic Lateral Sclerosis Type 1 (105400); Amyotrophic Lateral Sclerosis Type 10 (612069); Amyotrophic Lateral Sclerosis Type 12 (613435)	NextGen Sequencing		2-Nov-20
Amyotrophic Lateral Sclerosis NGS Panel	853	ABCD1, ABHD12, ALS2, ANG, ARHGEF28, ATXN1, ATXN2, C9orf72, CHCHD10, CHGB, CHMP2B, CRYM, DAO, DCTN1, ERBB4, FIG4, FUS, GRN, HNRNPA1, HNRNPA2B1, LUM, MAPT, MATR3, NEFH, NEK1, OPTN, PFN1, PRPH, PSEN1, SETX, SIGMAR1, SOD1, SPG11, SPG20, SQSTM1, TAF15, TARDBP, TBK1, TREM2, TRPM7, TUBA4A, UBQLN2, UNC13A, VAPB, VCP, VEGFA (46 genes)	Lavender Top (EDTA) 2 x 4 mL	Fulgent Genetics (fulgentdiagnostics.com)		NextGen Sequencing	Always add del/dupl	22-Aug-17

Appelation approuvée de l'analyse ou maladie associée [avec numéro de phénotype MIM] / Approved test name or Associated Disease [with MIM Phenotype Number]	**Code Test** / Test Code **(Dictionnaire MSSS)**	**SYMBOL HGNC DU GÈNE et NOM OMIM DU GÈNE** / HGNC Gene Symbol and OMIM Gene Number	**RÈGLES DE MANUTENTION** / Handling Conditions	**LABORATOIRE DÉSIGNÉ** / Designated laboratory	**RENSEIGNEMENTS SUR L'ANALYSE** / Test details	**MÉTHODE** / Methode	**NOTE PARTICULIÈRE** / Special comment	**Date de la révision** / Date of revision
Andersen-Tawil syndrome (170390)		KCNJ2 (600681)	Lavender Top (EDTA)	Invitae (www.invitae.com)	ANDERSEN CARDIODYSRHYTHMIC PERIODIC PARALYSIS (proper name); Andersen syndrome (170390), long QT syndrome 7, PERIODIC PARALYSIS, POTASSIUM-SENSITIVE CARDIODYSRHYTHMIC TYPE			7-Oct-18
Angelman Syndrome: Methylation and Copy Number Analysis	311	SNRPN	Lavender Top (EDTA)	Genome Diagnostics The Department of Paediatric Laboratory Medicine The Hospital for Sick Children		Methylation-Specific-MLPA of SNPRN		19-Feb-20
Angelman Syndrome: UPD 15 Analysis	311		Lavender Top (EDTA)	Genome Diagnostics The Department of Paediatric Laboratory Medicine The Hospital for Sick Children		STR analysis		19-Feb-20
Angiotensin Converting Enzyme (ACE), CSF	1919		CSF. Store Frozen	In-Common Laboratories				
Anti-Aquaporin 4 (anti-AQP4 AB) + Anti-Myelin Oligodendrocyte Glycoproteins Antibodies (Anti-MOG AB)	157		Serum only (Red Top OR Gold SST). Store at -20°C. (CSF is not an appropriate sample as Ab is made by plasma cells peripheraly.)	BC Neuroimmunology (bcneuro.ca)	Anti-Aquaporin 4 (Anti-AQP4 Ab) $150; Anti-Myelin Oligodendrocyte Glycoproteins antibody (Anti-MOG) $100	CBA live	NMO avaialble at CHUM for CSF and Serum as "NMO-Ig"	31-Oct-20
Antibodies to clustered acetylcholine receptors (AChR Ab CBA)	1465		Serum (Gold SST) or CSF. Store at -20°C.	BC Neuroimmunology (bcneuro.ca)	myasthenia gravis	CBA		31-Oct-20
Anti-C1Q Ab, IgG (RDL) [520147]	1402		Serum. Store and send frozen. Stable for 60 days.	LabCorp/Dynacare (https://www.labcorp.com/)		ELISA		7-May-21
Anti-DNAse B Titer, Serum [ADNAS]	1465		Serum (Gold or Red Top). Stable for 28 days at 4°C (preferred) or -20°C.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)		Nephelometry		21-May-21
Anti-DPPX (dipeptidyl aminopeptidase-like 6)	1735		Gold SST	Mitogen Advanced Diagnostics	encephalitis	CBA		2-Jun-21
Anti-dsDNA (Double-stranded) Ab by Farr method (RDL) [520059]	1465		Serum (Red Top or Gold SST). Store at -20°C.	LabCorp/Dynacare (https://www.labcorp.com/)		RIA		23-Nov-20
Anti-**Elaprase** antibody	1406		Draw blood before infusion for patients on treatment. If a specimen is being drawn due to an infusion reaction, please wait between 4 and 24 h to allow ERT to clear from blood. Collect 6 ml of blood (3 ml serum to send frozen the day of collection ; if not possible to send it the same day of collection keep frozen at - 80C until shipping)	LabCorp (https://www.labcorp.com/)	Antibody produced in patients on Enzyme Replacement Therapy for Hunter syndrome (MPSII)			
Anti-Enterocyte Antibody	1404		Serum. Store frozen.	The Children's Hospital of Philadelphia (https://www.testmenu.com/chop)				23-Mar-20
Anti-GMCSF Autoantibodies [GMCSFA]	1279		Serum gel (Gold top); Freeze at -70°C.	National Jewish Health Laboratories		ELISA		23-May-19
Anti-IFNg Autoantibody [IFNGE]	1147		Gold SST or Red Top. 2-8°C for 48 hours, >1 month at -70°C	National Jewish Health Laboratories				18-Mar-21
Anti-IgA [FIGA]	908		Gold SST	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)				4-Mar-21
Anti-IgLON5	1465		Gold SST	Mitogen Advanced Diagnostics		CBA		2-Jun-21
Anti-MeCP2	1465		Gold SST	Mitogen Advanced Diagnostics			Part of Arthitis Panel?	29-Nov-17
Anti-myelin associated glycoproteins (MAG)	40		Gold SST	Mitogen Advanced Diagnostics		ELISA		2-Jun-21
Anti-RNA pol I/III			Gold SST	Mitogen Advanced Diagnostics				
Anti-TB Drug	58	Azithromycin; Ethambutol; Rifabutin; Isonazid; Pyrazinamide; Rifampin	Serum	National Jewish Health Laboratories	TDM	LC/GCMS/LCMS	Same price for all anti-TB drugs per determination	2020-20-29
Anti-THSD7A (thrombospodin)	1465		Gold SST; store serum at -20°C	Mitogen Advanced Diagnostics	primary membranous nephropathy	CBA		2-Jun-21
anti-Tr/DNER (Delta/Notch-like epidermal growth factor-related receptor)			Gold SST	Mitogen Advanced Diagnostics	anti-Purkinje cell antibody		Availble as part of panel	21-Apr-17
Apert/Pfeiffer Syndrome (recurrent mutation in FGFR1 gene)		FGFR1 (136350); FGFR2 (176943)	Lavender Top (EDTA)	Molecular Genetics Laboratory, Hospital for Sick Children (www.sickkids.ca/PaediatricLaboratoryMedicine/Laboratories-Services/Molecular-Genetics-Laboratory/index.html)	Craniosynostosis: Apert syndrome (101200), Pfeiffer syndrome (101600)	Sanger sequencing: FGFR2 (exon 7); FGFR1 (p.Pro252Arg)		
Aripiprazole (Abilify) [FARI]	1142		Red top tube (Gold top SST tube not accepted)	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)		LC-MS/MS		5-May-21
Arrhythmia Panel [CA1601]	289	ABCC9 AKAP9 ANK2 CACNA1C* CACNB2 CALM1* CALM2 CALM3 CASQ2 CAV3 CDH2 CTNNA3 DBH DSC2 DSG2 DSP C FLNC* GATA6 HADHA HCN4 KCNA5 KCNE1 KCNH2 KCNJ2 KCNJ5 KCNQ1 LDB3 LEMD2 LMNA MYH6 MYH7 MYL4 NKX2-5 NOS1AP PKP2#* PLN PPA2 RYR2 SALL4 SCN10A SCN1B SCN3B SCN5A TBX5 TECRL TGFB3 TMEM43 TNNI3 TNNI3K TNNT2 TRDN TRPM4 TTN*	Lavender Top (EDTA) [2 tubes]	Blueprint Genetics http://blueprintgenetics.com/		NextGen Sequencing		20-Feb-20
Ataxia with Oculomoter Apraxia Type 1/2 NGS Panel		APTX (606350); SETX (608465)	Lavender Top (2 x 4 mL)	Fulgent Genetics (fulgentdiagnostics.com)	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia (208920); Amyotrophic lateral sclerosis 4, juvenile (602433); Ataxia-ocular apraxia-2 (606002)	NextGen Sequencing		5-Apr-19

Appelation approuvée de l'analyse ou maladie associée [avec numéro de phénotype MIM] / Approved test name or Associated Disease [with MIM Phenotype Number]	**Code Test /** Test Code **(Dictionnaire MSSS)**	**SYMBOL HGNC DU GÈNE et NOM OMIM DU GÈNE /** HGNC Gene Symbol and OMIM Gene Number	**RÈGLES DE MANUTENTION /** Handling Conditions	**LABORATOIRE DÉSIGNÉ /** Designated laboratory	**RENSEIGNEMENTS SUR L'ANALYSE /** Test details	**MÉTHODE /** Methode	**NOTE PARTICULIÈRE /** Special comment	**Date de la révision /** Date of revision
Ataxia/Episodic Ataxia Disorders (NextGen Sequencing Panel and Copy Number Analysis; 330 Genes + mtDNA + FRDA Repeat Expansion Analysis) [NGS419]	583	AAAS (605378), AARS2 (612035), ABCB7 (300135), ABCC8 (600509), ABCD1 (300371), ABHD5 (604780), ACO2 (100850), ADCK3 (606980), ADSL (608222), AFG3L2 (604581), AHI1 (608894), ALDH5A1 (610045), ALG6 (604566), AMACR (NA), ANO10 (613728), APOPT1 (616003), APTX (606350), ARL13B (608922), ARL6 (608845), ARSA (607574), ASL (608310), ASS1 (603470), ATM (607585), ATN1 (607462), ATP1A2 (182340), ATP1A3 (182350), ATP8A2 (605870), ATPAF2 (608918), ATXN1 (601556), ATXN10 (603516), ATXN2 (601517), ATXN3 (607047), ATXN7 (607640), AUH (600529), B9D1 (614144), BBS1 (209901), BBS10 (610148), BBS12 (610683), BBS2 (606151), BBS4 (600374), BBS5 (603650), BBS7 (607590), BBS9 (607968), BCKDHA (608348), BCKDHB (248611), BCS1L (603647), BEAN1 (612051), BOLA3 (613183), BSCL2 (606158), BTD (609019), C10ORF2 (606075), C12orf65 (613541), C19orf12 (614298), C5ORF42 (614571), CA8 (114815), CACNA1A (601011), CACNA1G (604065), CACNB4 (601949), CAMTA1 (611501), CASK (300172), CC2D2A (612013), CCDC28B (610162), CCDC88C (611204), CECR1 (607575), CEP290 (610142), TSGA14 (610523), CHCHD10 (615903), CLCN2 (600570), CLN5 (608102), CLN6 (606725), CLN8 (607837), CLPP (601119), COG4 (606976), COQ2 (609825), COX10 (602125), C12ORF62 (614478), COX15 (603646), FAM36A (614698), COX6B1 (124089), COX8A (123870), CP (117700), CPS1 (608307), CSPP1 (611654), CSTB (601145), CTDP1 (604927), CTSD (116840), CTSF (603539), CYP27A1 (606530), DARS2 (610956), DBT (248610), DCX (300121), DHFR (126060), DKC1 (300126), DLAT (608770), DLD (238331), DNAJC19 (608977), DNAJC5 (611203), DNMT1 (126375), DPM1 (603503), DYRK1A (600855), EEF2 (130610), EGR2 (129010), ELOVL4 (605512), ELOVL5 (611805), ERCC2 (126340), ERCC5 (133530), ERCC6 (133540), ETHE1 (608451), FA2H (611026), FASTKD2 (612322), FBXL4 (605654), FGF14 (601515), FLVCR1 (609144), FMR1 (309550), FOXRED1 (613622), FTL (134790), FXN (606829), GABRB3 (137192), GAMT (601240), GBA (606463), GBA2 (609471), GCH1 (600225), GFAP (137780), GJA1 (600309), GJB1 (304040), GJC2 (608803), GLB1 (611458), GMPPB (615320), GOSR2 (604027), GPI (172400), GPR56 (604110), GRM1 (614831), GRN (138945), GSS (601002), HARS (142810), HARS2 (600783), HCN1 (602780), HEPACAM (611642), HEXB (606873), HTR1A (109760), HTRA1 (602194), HTT (613004), INPP5E (613037), ITM2B (603904), ITPR1 (147265), KCNA1 (176260), KCNC1 (176258), KCNC3 (176264), KCND3 (605411), KCTD7 (611725), KIF1A (601255), KIF1B (605995), KIF5A (602821), KIF7 (611254), LARS2 (604544), LMNB1 (150340), LRPPRC (607544), LYRM7 (615831), MAN2B1 (609458), MARS2 (609728), MBD5 (611472), MECP2 (300005), MED13L (608771), MESD8 (611124)	Lavender Top (EDTA)	MNG Laboratories (www.medicalneurogenetics.com)		NextGen Sequencing		14-May-20
Ataxia/Episodic Ataxia Disorders (NextGen Sequencing Panel and Copy Number Analysis; 534 Genes + mtDNA) [NGS324]	583	AAAS (605378), AARS2 (612035), ABCB7 (300135), ABCC8 (600509), ABCD1 (300371), ABHD5 (604780), ACO2 (100850), ADCK3 (606980), ADSL (608222), AFG3L2 (604581), AHI1 (608894), ALDH5A1 (610045), ALG6 (604566), AMACR (NA), ANO10 (613728), APOPT1 (616003), APTX (606350), ARL13B (608922), ARL6 (608845), ARSA (607574), ASL (608310), ASS1 (603470), ATM (607585), ATN1 (607462), ATP1A2 (182340), ATP1A3 (182350), ATP8A2 (605870), ATPAF2 (608918), ATXN1 (601556), ATXN10 (603516), ATXN2 (601517), ATXN3 (607047), ATXN7 (607640), AUH (600529), B9D1 (614144), BBS1 (209901), BBS10 (610148), BBS12 (610683), BBS2 (606151), BBS4 (600374), BBS5 (603650), BBS7 (607590), BBS9 (607968), BCKDHA (608348), BCKDHB (248611), BCS1L (603647), BEAN1 (612051), BOLA3 (613183), BSCL2 (606158), BTD (609019), C10ORF2 (606075), C12orf65 (613541), C19orf12 (614298), C5ORF42 (614571), CA8 (114815), CACNA1A (601011), CACNA1G (604065), CACNB4 (601949), CAMTA1 (611501), CASK (300172), CC2D2A (612013), CCDC28B (610162), CCDC88C (611204), CECR1 (607575), CEP290 (610142), TSGA14 (610523), CHCHD10 (615903), CLCN2 (600570), CLN5 (608102), CLN6 (606725), CLN8 (607837), CLPP (601119), COG4 (606976), COQ2 (609825), COX10 (602125), C12ORF62 (614478), COX15 (603646), FAM36A (614698), COX6B1 (124089), COX8A (123870), CP (117700), CPS1 (608307), CSPP1 (611654), CSTB (601145), CTDP1 (604927), CTSD (116840), CTSF (603539), CYP27A1 (606530), DARS2 (610956), DBT (248610), DCX (300121), DHFR (126060), DKC1 (300126), DLAT (608770), DLD (238331), DNAJC19 (608977), DNAJC5 (611203), DNMT1 (126375), DPM1 (603503), DYRK1A (600855), EEF2 (130610), EGR2 (129010), ELOVL4 (605512), ELOVL5 (611805), ERCC2 (126340), ERCC5 (133530), ERCC6 (133540), ETHE1 (608451), FA2H (611026), FASTKD2 (612322), FBXL4 (605654), FGF14 (601515), FLVCR1 (609144), FMR1 (309550), FOXRED1 (613622), FTL (134790), FXN (606829), GABRB3 (137192), GAMT (601240), GBA (606463), GBA2 (609471), GCH1 (600225), GFAP (137780), GJA1 (600309), GJB1 (304040), GJC2 (608803), GLB1 (611458), GMPPB (615320), GOSR2 (604027), GPI (172400), GPR56 (604110), GRM1 (614831), GRN (138945), GSS (601002), HARS (142810), HARS2 (600783), HCN1 (602780), HEPACAM (611642), HEXB (606873), HTR1A (109760), HTRA1 (602194), HTT (613004), INPP5E (613037), ITM2B (603904), ITPR1 (147265), KCNA1 (176260), KCNC1 (176258), KCNC3 (176264), KCND3 (605411), KCTD7 (611725), KIF1A (601255), KIF1B (605995), KIF5A (602821), KIF7 (611254), LARS2 (604544), LMNB1 (150340), LRPPRC (607544), LYRM7 (615831), MAN2B1 (609458), MARS2 (609728), MBD5 (611472), MECP2 (300005), MED13L (608771), MESD8 (611124)	Lavender Top (EDTA)	MNG Laboratories (www.medicalneurogenetics.com)/Dynacare		NextGen Sequencing		14-May-20
Atypical Hemolytic Uremic Syndrome and Membranoproliferative Glomerulonephritis Panel: Sequencing		CD46 CFB CFH CFHR5 CFI C3 THBD APLN	Lavender Top (EDTA) 2 x 4 mL	The Hospital for Sick Children Rapid Response Laboratory 170 Elizabeth Street, Room 3642 Toronto, ON M5G 2G3 Canada Phone: 416-813-7200 Phone: 1-855-381-3212	aHUS Familial Hemolytic-Uremic syndrome Hereditary Hemolytic-Uremic syndrome MPGN; Mesangiocapillary glomerulonephritis	Sanger sequencing		

Appelation approuvée de l'analyse ou maladie associée [avec numéro de phénotype MIM] / Approved test name or Associated Disease [with MIM Phenotype Number]	**Code Test** / Test Code **(Dictionnaire MSSS)**	**SYMBOL HGNC DU GÈNE et NOM OMIM DU GÈNE** / HGNC Gene Symbol and OMIM Gene Number	**RÈGLES DE MANUTENTION** / Handling Conditions	**LABORATOIRE DÉSIGNÉ** / Designated laboratory	**RENSEIGNEMENTS SUR L'ANALYSE** / Test details	**MÉTHODE** / Methode	**NOTE PARTICULIÈRE** / Special comment	**Date de la révision** / Date of revision
Atypical Hemolytic Uremic Syndrome and Membranoproliferative Glomerulonephritis Panel: Sequencing	672	CFH (134370); CD46 (120920); CFI (217030); CFB (138470); CFHR5 (608593); C3 (120700); THB (187940); APLN (300297)	Lavender Top (EDTA)	Molecular Genetics Laboratory, Hospital for Sick Children (www.sickkids.ca/PaediatricLaboratoryMedicine/Laboratories-Services/Molecular-Genetics-Laboratory/index.html)	HUS, Familial Hemolytic-Uremic syndrome, Hereditary Hemolytic-Uremic syndrome, MPGN; Mesangiocapillary glomerulonephritis	Sanger sequencing	CD46 $850; CFB $850; CFH $1000; CFHR5 $850; CFI ($850; C3 $2000; THBD $600; APLN $600	
Autism Spectrum Disorders and Intellectual Disability (ASD-ID) Comprehensive Sequencing Panel with CNV Detection [5045]	717	complete gene list available at https://www.preventiongenetics.com/documents/ASD_IDGeneList.pdf?07c00668	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Replaces Fulgent ID Panel when proband available only (adopted child…). If 'Trio' (proband + parents) send for Autism/ID Xpanded Panel at GeneDx (3500 USD for 3 samples)	NextGen Sequencing		2-Feb-18
Autism/ID Xpanded Panel [952]	717		Lavender Top (EDTA)	GeneDx (www.genedx.com)		NextGen Sequencing		9-Dec-19
Autoimmune Dysautonomia Evaluation, Serum (ADE)			Red Top	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)		IFA, RIA, EIA, WB, CBA		
Autoimmune Liver Diseases Profile PLUS	1330		Gold SST	Mitogen Advanced Diagnostics	primary biliary cirrhosis, autoimmune hepatitis: Mitochondrial oxaloacid dehydrogenase M2/M3 Soluble liver antigen (SLA) Liver-Kidney-Microsome (LKM) SP100 gp210 PML 3EBP0 LC-1 Ro52/TRIM21	LIA		2-Jun-21
Autoimmune Myopathy /Myositis Profile PLUS (Includes Mup44 & Immune Mediated Necrotizing Profile)	153		Gold SST	Mitogen Advanced Diagnostics	Jo-1, Mi2, Mi2-α, Mi2β, MDA5, NXP2, TIF1γ PL7, PL12, PM/Scl75, PM/Scl100, Ku, SRP, EJ, OJ, Ro52, HMGCR, anti-NT5C1 A/Mup44	LIA, ELISA, ALBIA (LDT)		2-Jun-21
Autoimmune Polyendocrinopathy Syndrome Type 1 via the AIRE Gene [1224]		AIRE (607358)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)		Sanger sequencing		15-Mar-17
Autoimmune Retinopathy Panel by Immunoblot [ARP]	720		Serum. Store at 4°C. DO NOT FREEZE.	Ocular Immunology Laboratory, OHSU Biomedical Research Building, Room 253 3181 SW Sam Jackson Park Road Portland, OR 97239, USA	CAII (carbonic anhydrase II), HSP27 (heat shock protein 27), GAPDH (glyceraldehyde 3-phosphate dehydrogenase), Aldolase, Enolase, Arrestin, Tubulin, PKM2 (pyruvate kinase M2)	Immunoblot		11-Apr-20
Autoinflammatory Syndromes Panel [08120]	838		Lavender Top (EDTA)	Invitae (www.invitae.com)		NextGen Sequencing		3-Oct-19
Autosomal dominant lateral temporal lobe epilepsy (Epilepsy, familial temporal lobe, 1) (600512)		LGI-1 (604619)	Lavender Top (EDTA)	Centogene AG (www.centogene.com)		1. single exon testing 2. full gene sequencing 3. deletion/duplication testing		
Avian IgG Antibodies Panel, Serum (Budgie and Pigeon)	1419		Red Top; Gold SST	In-Common Laboratories	budgie = parakeet	FEIA		10-Mar-20
Avian precipitins: Pigeon IgG Antibodies	1419		Red Top; Gold SST	In-Common Laboratories		FEIA		8-Nov-18
Axenfeld-Rieger Syndrome (FOXC1) [TB18]	1069	FOXC1 (601090)	Lavender Top (EDTA)	GeneDx (www.genedx.com)	Axenfeld-Rieger syndrome, type 3 and Rieger or Axenfeld anomalies (602482)	Sanger sequencing		9-Dec-19
Baller-Gerold syndrome (218600)		RECQL4 (603780)	Lavender Top (EDTA)	Centogene AG (www.centogene.com)		1. single exon testing 2. full gene sequencing 3. deletion/duplication testing		
Bartter Syndrome NGS Panel	804	ATP6V1B1, BSND, CA2, CASR, CLCNKA, CLCNKB, CLDN16, CLDN19, FXYD2, GNA11, HNF1B, HSD11B2, KCNJ1, KCNJ10, KLHL3, MAGED2, NR3C2, SCNN1A, SCNN1B, SCNN1G, SLC12A1, SLC12A2, SLC12A3, SLC4A1, SLC4A4, WNK1, WNK4	Lavender Top (EDTA) 2 x 4 mL	Fulgent Genetics (fulgentdiagnostics.com)				28-Feb-20
Basal Ganglia Calcification NGS Panel	1192	CA2; MYORG; PDGFB (190040); PDGFRB; SLC20A2 (158378); XPR1	Lavender Top (EDTA) 2 x 4 mL	Fulgent Genetics (fulgentdiagnostics.com)	Basal ganglia calcification, idiopathic, 5 (615483); Basal ganglia cancification, idiopathic, 1 (213600)	NextGen Sequencing		13-Feb-20
Bb	792		0.5 mL EDTA plasma (serum also accepted) –spun, separated, frozen within 2 hrs of collection, separate aliquot each test; ship on dry ice	Cincinnati Children's Hospital Medical Center 3333 Burnet Avenue NRB 1042 Cincinnati, OH 45229 (www.cincinnatichildrens.org)				15-Nov-17
Beckwith-Wiedemann syndrome: CDKN1C Sequencing (Step 3)	571	CDKN1C (600856)	Lavender Top (EDTA) [2 tubes]	Genome Diagnostics The Department of Paediatric Laboratory Medicine The Hospital for Sick Children	Wiedemann-Beckwith Syndrome (130650)			19-Feb-20
Beckwith-Wiedemann syndrome: Methylation & Copy Number (Step1)	571		Lavender Top (EDTA) [2 tubes]	Genome Diagnostics The Department of Paediatric Laboratory Medicine The Hospital for Sick Children	Wiedemann-Beckwith Syndrome (130650)	Methylation-Specific-MLPA of imprinting centers 1 and 2		19-Feb-20
Beckwith-Wiedemann syndrome: UPD11 Analysis (Step 2)	571	H19 (103280); KCNQ1OT1 (604115); CDKN1C (600856)	Lavender Top (EDTA) [2 tubes]	Genome Diagnostics The Department of Paediatric Laboratory Medicine The Hospital for Sick Children	Wiedemann-Beckwith Syndrome (130650)	UPD11 studies via STR (short tandem repeat) analysis.		19-Feb-20
Beta-Thalassemia (613985)	823	HBB (141900)	Lavender Top (EDTA)	Molecular Genetics Laboratory - McMaster University Medical Centre		Sanger sequencing		

Appelation approuvée de l'analyse ou maladie associée [avec numéro de phénotype MIM] / Approved test name or Associated Disease [with MIM Phenotype Number]	**Code Test** / Test Code **(Dictionnaire MSSS)**	**SYMBOL HGNC DU GÈNE et NOM OMIM DU GÈNE** / HGNC Gene Symbol and OMIM Gene Number	**RÈGLES DE MANUTENTION** / Handling Conditions	**LABORATOIRE DÉSIGNÉ** / Designated laboratory	**RENSEIGNEMENTS SUR L'ANALYSE** / Test details	**MÉTHODE** / Methode	**NOTE PARTICULIÈRE** / Special comment	**Date de la révision** / Date of revision
Bile Acid, Serum (Bile Acid Profile)	9		Urine 5-30mL Serum 0.5-2mL; Bile Fluid 1-2mL. FREEZE URINE and SERUM ASAP.Note: If possible send Urine & Serum. Urine is analyzed for all patients – if Urine shows evidence of a metabolic abnormality, Serum will be tested. URSO can mask detection of bile acid synthetic defects it is preferable for patients to be off URSO or ACTIGAL for 5 DAYS before SAMPLE Collection	Mass Spectrometry Lab – MLC 7019; Cincinnati Children's Hospital Medical Center	Note: This is refelex test from Bile Acids, Urine. Both sample should be sent if possible.	FAB-MS	Available at MCL ($153.70) [BAFS]	8-Aug-17
Bile Acid, Urine (Bile Acid Profile)	9		Urine 5-30mL Serum 0.5-2mL; Bile Fluid 1-2mL. FREEZE URINE and SERUM ASAP. Note: If possible send Urine & Serum. Urine is analyzed for all patients – if Urine shows evidence of a metabolic abnormality, Serum will be tested. URSO can mask detection of bile acid synthetic defects it is preferable for patients to be off URSO or ACTIGAL for 5 DAYS before SAMPLE Collection	Mass Spectrometry Lab – MLC 7019; Cincinnati Children's Hospital Medical Center		FAB-MS	NOT AVAILABLE AT MCL	8-Aug-17
Birt-Hogg-Dubé Syndrome (135150)	740	FLCN (607273)	Lavender Top (EDTA)	Alberta Mol Dx Laboratory (Calgary)		NGS		16-Jun-20
BMPR1A, SMAD4 Gene Sequencing & Del/Dup [717]		SMAD4 (600993); BMPR1A (601299)	Lavender Top (EDTA)	GeneDx (www.genedx.com)	174900 (juvenile polyposis syndrome); 175050 (juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome)	Sanger Sequencing, Exon Array CGH		9-Dec-19
Bone Marrow Failure Syndrome Panel [HE0801]	1427	135 genes	Lavender Top (EDTA) [2 tubes]	Blueprint Genetics http://blueprintgenetics.com/		NextGen Sequencing		28-Feb-20
BP 180 and BP230			Serum	Immunodermatology Laboratory Department of Dermatology 30 North 1900 East, 4A330 SOM Salt Lake City, Utah 84132 Email: immunoderm@hsc.utah.edu		ELISA	$132.32 US	14-Sep-20
Brain-Iron Accumulation NGS Panel	11213	ATP13A2, C19orf12, COASY, CP, DCAF17, FA2H, FTL, GTPBP2, PANK2, PLA2G6, SCP2, WDR45	Lavender Top (EDTA) 2 x 4 mL	Fulgent Genetics (fulgentdiagnostics.com)		NextGen Sequencing		28-Feb-20
Breast and Gynaecological Cancer Specific Panel	227		Lavender Top (EDTA)	Invitae (www.invitae.com)	This is a special panel from the Cedars cancer Centre.	Next-Generation sequencing		6-Feb-20
Breast Cancer Panel (Invitae) [01202]	227	ATM (), BARD1 (), BRCA1 (113705), BRCA2 (600185); BRIP1(), CDH1 (192090); CHEK2 (), NBN (), NF1 (), PALB2 (601728) PTEN (601728), RAD50 () STK11 (602216), TP53 (191170)	Lavender Top (EDTA)	Invitae (www.invitae.com)	Breast-Ovarian Cancer, Familial, type 1 (604370); Breast-Ovarian Cancer, Familial, type 2 (612555); Ovarian carcinoma, somatic (167000); Fanconi anemia, complementation group N (610832); PTEN hamartoma tumor syndrome (_); Pancreatic cancer (260350); Peutz-Jeghers syndrome (175200); Breast cancer (114480)	Next-Generation sequencing		7-Oct-18
Bromine - Total Blood [FBROM]	1465		Royal Blue (EDTA) top tube	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)		ICP/MS	INSPQ?? TO BE VERIFIED - NO	30-Apr-21
Bullous Autoimmune Skin Disease Profile	750		Serum gel (Gold top)	Mitogen Advanced Diagnostics	BP180, BP230, Desmoglein 1, Desmoglein 3	CBA		2-Jun-21
C3 Nephritic Factor	792		1 ml frozen serum	Molecular Otolaryngology & Renal Research Laboratory, U. of Iowa Carver College of Medicine (www.medicine.uiowa.edu/morl)	Dense Deposit Disease (DDD, aka Membranoproliferative Glomerulonephritis Type II, MPGNII)	1. Immunofixation electrophoresis (IFE) combines the techniques of electrophoresis with immunofixation to detect C3 degradation products, an indirect measure of dysregulation of C3 convertase. Conversion of C3 to C3c is quantitated (Koch, et al., 1981). 2. C3 Convertase Stabilizing Assay (C3CSA) measures the ability of C3Nefs to stabilize C3 convertase on sheep erythrocytes. 3. C3 Convertase Stabilizing Assay with Properdin (C3CSAP) measures the ability of C3Nefs to stabilize C3 convertase built with properdin on sheep erythrocytes.	Available as Facteur C3 nephritique (activité) at CHUQ (HEJ)	7-Feb-19
C3 Nephritic Factor	792		1 ml frozen serum	Cincinnati Children's Hospital Medical Center 3333 Burnet Avenue NRB 1042 Cincinnati, OH 45229 (www.cincinnatichildrens.org)	Dense Deposit Disease (DDD, aka Membranoproliferative Glomerulonephritis Type II, MPGNII)		Available as Facteur C3 nephritique (activité) at CHUQ (HEJ)	15-Nov-17
C3G Functional Panel	792		2 mL serum + 2 mL frozen EDTA plasma. Store frozen.	Molecular Otolaryngology & Renal Research Laboratory, U. of Iowa Carver College of Medicine (www.medicine.uiowa.edu/morl)	DDD & C3GN	serology		18-Sep-19

Appelation approuvée de l'analyse ou maladie associée [avec numéro de phénotype MIM] / Approved test name or Associated Disease [with MIM Phenotype Number]	**Code Test** / Test Code **(Dictionnaire MSSS)**	**SYMBOL HGNC DU GÈNE et NOM OMIM DU GÈNE** / HGNC Gene Symbol and OMIM Gene Number	**RÈGLES DE MANUTENTION /** Handling Conditions	**LABORATOIRE DÉSIGNÉ** / Designated laboratory	**RENSEIGNEMENTS SUR L'ANALYSE** / Test details	**MÉTHODE /** Methode	**NOTE PARTICULIÈRE /** Special comment	**Date de la révision /** Date of revision
C5a			1 ml frozen serum	Cincinnati Children's Hospital Medical Center 3333 Burnet Avenue NRB 1042 Cincinnati, OH 45229 (www.cincinnatichildrens.org)				21-Nov-18
C9orf72 with reflex to SOD1 and ATXN2 [13039]	853	C9orf72 (614260); SOD1 (147450)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Amyotrphic lateral sclerosis		If C9orf72 is only performed, then the price is only $250 USD	2-Nov-20
CACNA1A Full Gene Sequencing Analysis [MOL033]		CACNA1A (601011)	Lavender Top (EDTA)	MNG Laboratories (www.medicalneurogenetics.com)/Dynacare	Familial hemiplegic migraine 1 (141500); episodic ataxia type 2 (108500)	Sanger sequencing		14-May-20
CACNB4 Full Gene Sequencing Analysis [MOL227]		CACNB4 (601949)	Lavender Top (EDTA)	MNG Laboratories (www.medicalneurogenetics.com)/Dynacare	Episodic ataxia, type 5 (613855)	Sanger sequencing		14-May-20
Calpain 3 DNA Sequencing Test [563]		CAPN3 (114240)	Lavender Top (EDTA)	Athena Diagnostics (www.athenadiagnostics.com)	LGMD2A (253600), Calpainopathy			
CAMURATI-ENGELMANN DISEASE (CED) VIA THE TGFB1 GENE [787]		TGFB1 (190180)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Camurati-Engelmann Disease (131300)	Sanger sequencing		
Carbohydrate Deficient Transferrin for Congenital Disorders of Glycosylation, Serum [CDG]	406		Red Top. Store serum frozen.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)		Affinity Chromatography-Mass Spectrometry	This is a confirmatory test following the screening test for CDT testing at the MUHC.	16-Dec-20
Carbohydrate, Urine [CHOU]	1572		Random urine. Store frozen. Stable 21 days.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	Replaces "reducing substances"	TLC		31-Oct-20
Cardio-Facio-Cutaneous Syndrome NGS Panel (5 genes)		BRAF (164757), KRAS (190070), MAP2K1 (176872), MAP2K2 (601263), SOS1 (182530)	Lavender Top (EDTA) 2 x 4 mL	Fulgent Genetics (fulgentdiagnostics.com)	Cardiofaciocutaneous syndrome (115150); Cardiofaciocutaneous syndrome 2 (615278); Cardiofaciocutaneous syndrome 3(615279); Cardiofaciocutaneous syndrome 4 (615280); Fibromatosis, gingival (135300)	NextGen Sequencing		
Cathartic Laxatives Profile, Stool [FCLPS]			Stool	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	Mg, Pi			20-Oct-20
CAVEOLINOPATHY TESTING VIA THE CAV3 GENE [467]		CAV3 (601253)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Muscular Dystrophy, Limb-Girdle, Type 1C (607801)	Sanger sequencing		
CDKN2A (p16) & CDK4 (Exon2) Sequencing [2021]		CDKN2A (600160); CDK4 (123829)	Lavender Top (EDTA)	GeneDx (www.genedx.com)	Melanoma, cutaneous malignant, 3			9-Dec-19
Central Hypoventilation And Apnea Panel [PU0401]	775	CHAT, CHRNA1,CHRNB1, CHRND, CHRNE, COLQ, EDN3, GLRA1, MEP2, PHOX2B, PAPSN, RET, SCN4A, SLC6A5, ZEB2	Lavender Top (EDTA)	Blueprint Genetics http://blueprintgenetics.com/		Next-Generation sequencing		26-Sep-18
CEREBRAL CAVERNOUS MALFORMATIONS PANEL [1943]	823	KRIT (604214), CCM2 (607292) , PCCD10 (609118)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Cerebral Cavernous Malformations 1(116860), Cerebral Cavernous Malformations 2 (603284), Cerebral Cavernous Malformations 3 (603285)	Sequencing and CNV Detection via NextGen Sequencing using PG-Select Capture Probes		2-Feb-20
CEREBRAL CAVERNOUS MALFORMATIONS VIA THE CCM2 GENE (603284) [122]		CCM2 (607929)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Cerebral calvernous malformations-2	Sanger sequencing		
CEREBRAL CAVERNOUS MALFORMATIONS VIA THE KRIT1/CCM1 GENE [121]		KRIT1 (604241)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Cerebral calvernous malformations-1 (116860)	Sanger sequencing		
CEREBRAL CAVERNOUS MALFORMATIONS VIA THE PDCD10/CCM3 (603285) [123]		PDCD10 (609118)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Cerebral calvernous malformation-3	Sanger sequencing		
CerebroTendinous Xanthomatosis (CTX) via the CYP27A1 Gene [1670]		CYP27A1 (606530)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	CerebroTendinous Xanthomatosis (213700)	Sanger sequencing		31-Mar-17

Appelation approuvée de l'analyse ou maladie associée [avec numéro de phénotype MIM] / Approved test name or Associated Disease [with MIM Phenotype Number]	**Code Test** / Test Code **(Dictionnaire MSSS)**	**SYMBOL HGNC DU GÈNE et NOM OMIM DU GÈNE** / HGNC Gene Symbol and OMIM Gene Number	**RÈGLES DE MANUTENTION** / Handling Conditions	**LABORATOIRE DÉSIGNÉ** / Designated laboratory	**RENSEIGNEMENTS SUR L'ANALYSE** / Test details	**MÉTHODE** / Methode	**NOTE PARTICULIÈRE** / Special comment	**Date de la révision** / Date of revision
Charcot Marie Tooth Disease (NextGen Sequencing Panel and Copy Number Analysis; 189 Genes + mtDNA) [NGS345]	643	AAAS (605378), AARS (601065), ABCD1 (300371), ACO2 (100850), ACOX1 (609751), AFG3L2 (604581), AIFM1 (300169), ALDH3A2 (609523), ALDH6A1 (603178), ALG2 (607905), AMPD2 (102771), AP1S2 (300629), AP3D1 (607246), AP4M1 (602296), APTX (606350), ARCN1 (600820), ARHGEF10 (608136), ARNT2 (606036), ARSA (607574), ASNS (108370), ASPA (608034), ATAD3A (612316), ATP13A2 (610513), ATP7B (606882), B4GALNT1 (601873), BAG3 (603883), BRPF1 (602410), BSCL2 (606158), C10ORF2 (606075), C12orf65 (613541), C19orf12 (614297), CLP1 (608757), COQ7 (601683), COX6A1 (602072), CSF1R (164770), CTDP1 (604927), CTSD (116840), CYP2U1 (610670), DARS2 (610956), DDHD1 (614603), DDOST (602202), DGUOK (601465), DHFR (126060), DHTKD1 (614984), DNAJB2 (604139), DNAJC3 (601184), DNM1L (603850), DNM2 (602378), DPM1 (603503), DYNC1H1 (600112), EGR2 (129010), ELOVL4 (605512), ELOVL5 (611805), ERCC3 (133510), ERCC6 (609413), ERCC8 (609412), EXOSC3 (606489), FBLN5 (604580), FGD4 (611104), FGF14 (601515), FIG4 (609390), FLVCR1 (609144), FOLR1 (136430), FOXG1 (164874), FOXP1 (605515), FTL (134790), GAA (606800), GABRB3 (137192), GALC (606890), GAN (605379), GARS (600287), GBE1 (607839), GDAP1 (606598), GIF (609342), GJB1 (304040), GJC2 (608803), GLUL (138290), GNB4 (610863), GPR56 (604110), HADHB (143450), HARS (142810), HINT1 (601314), HK1 (142600), HSD17B4 (601860), HSPB1 (602195), HSPB8 (608014), HTRA1 (602194), IARS2 (612801), IBA57 (615316), IER3IP1 (609382), IGHMBP2 (600502), INF2 (610982), ITPA (147520), KARS (601421), KIDINS220 (615759), KIF1A (601255), KIF1B (605995), KIF5A (602821), LITAF (603795), LMNA (150330), LRSAM1 (610933), MANBA (609489), MARS (156560), MAT1A (610550), MED25 (610197), MFN2 (608507), MICU1 (605084), MME (120520), MOCS1 (603707), MOCS2 (603708), MPZ (159440), MRPS22 (605810), MTMR2 (603557), MTTP (157147), NAGA (104170), NAGLU (609701), NARS2 (612803), NDRG1 (605262), NEFH (162230), NEFL (162280), OPA1 (605290), PC (608786), PDK3 (300906), PDYN (131340), PEX10 (602859), PEX11B (603867), PEX16 (603360), PEX19 (600279), PEX2 (170993), PEX5 (600414), PEX7 (601757), PGBD3 (609413), PGM3 (172100), PHGDH (606879), PIGV (610274), PLA2G6 (603604), PLEKHG5 (611101), PMP22 (601097), PNPLA6 (603197), SGK196 (615247), PRDM12 (616458), PRICKLE1 (608500), PRMT7 (610087), PRPS1 (311850), PRX (605725), PSAP (176801), PURA (600473), PYROXD1 (617220), RAB7A (602298), RMND1 (614917), RNASEH1 (604123), SBF1 (603560), SBF2 (607697), SEPT9 (604061), SH3TC2 (608206), SLC12A6 (604878), SLC16A2 (300095), SLC1A4 (600229), SLC25A1 (190315), SLC25A12 (603667), SLC25A19 (606521), SLC25A22 (609302), SLC25A46	Lavender Top (EDTA)	MNG Laboratories (www.medicalneurogenetics.com)/Dynacare	Charcot-Marie tooth Disease (302800)	Next-Generation sequencing	Alt: CTGT 42 genes & del/dupl	14-May-20
Childhood-Onset Epilepsy Panel [542]	356	ADSL, ALDH5A1, ATP1A3, ATRX, CACNA1A, CASK, CDKL5, CHD2, CHRNA2, CHRNA4, CHRNA7, CHRNB2, CLCN4, CLN3, CLN5, CLN6, CLN8, CNTNAP2, CSTB, CTSD, DDX3X, DEPDC5, DYRK1A, EEF1A2, EHMT1, EPM2A, FOLR1, FOXG1, GABRA1, GABRB2, GABRB3, GABRG2, GAMT, GATM, GNAO1, GOSR2, GRIN1, GRIN2A, IQSEC2, KANSL1, KCNA2, KCNC1, KCNMA1, KCNT1, KCTD7, KDM6A, KIAA2022, LGI1, MAGI2, MBD5, MECP2, MEF2C, MFSD8, NALCN, NGLY1, NHLRC1, NPRL3, NRXN1, PACS1, PCDH19, PIGN, PNKP, POLG, PPP2R5D, PPT1, PURA, SCN1A, SCN1B, SCN2A, SLC19A3, SLC2A1, SLC6A1, SLC6A8, SLC9A6, SMC1A, SPATA5, STX1B, SYNGAP1, TBC1D24, TCF4, TPP1, UBE3A, WDR45, ZEB2	Lavender Top (EDTA) [2 tubes]	GeneDx (www.genedx.com)		NextGen Sequencing		28-Feb-20
Cholestanol [FCHO]	400		Lavender top (EDTA)	Mayo Clinical Laboatories (Kennedy Kreiger Institute (Biochemical Genetics))	Cerebrotendinous xanthomatosis	GC-MS		18-Sep-17
Cholestasis Panel	400	ABCB11 MYO5B* NOTCH2* NR 2I 27 NPHP1 PEX1 PEX10 PEX12 PEX2 PEX26 PEX5 PEX6 SERPINA1 SLC25A13 SLC26A3 SMPD1 SPINT2 TJP2 TMEM216 TRMU L TTC37 UGT1A1 VIPAS39 VPS33B ABCB4 ABCC2 AKR1D1 ATP8B1 BAAT CFTR CREB3L3 CYP7B1 DCDC2 DGUOK EPCAM 80 FAH HSD3B7 JAG1	Lavender Top (EDTA)	Blueprint Genetics http://blueprintgenetics.com/		NextGen Sequencing		28-May-19
Choreoacanthocytosis (200150)		VPS13A (605978)	Lavender Top (EDTA)	North York General		2 mutations		
Christianson Type X-Linked Mental Retardation via the SLC9A6 Gene [562]		SLC9A6 (300231)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Mental Retardation, X-Linked, Syndromic, Christianson Type (300243)	Sanger sequencing		

Appelation approuvée de l'analyse ou maladie associée [avec numéro de phénotype MIM] / Approved test name or Associated Disease [with MIM Phenotype Number]	**Code Test** / Test Code **(Dictionnaire MSSS)**	**SYMBOL HGNC DU GÈNE et NOM OMIM DU GÈNE** / HGNC Gene Symbol and OMIM Gene Number	**RÈGLES DE MANUTENTION** / Handling Conditions	**LABORATOIRE DÉSIGNÉ** / Designated laboratory	**RENSEIGNEMENTS SUR L'ANALYSE** / Test details	**MÉTHODE** / Methode	**NOTE PARTICULIÈRE** / Special comment	**Date de la révision** / Date of revision
Chronic Granulomatous Disease NextGen Sequencing (NGS) Panel [1971]		CYBA (608508); CYBB (300481); NCF2 (608515); NCF4 (601488)	Lavender Top (EDTA)	Fulgent Genetics (fulgentdiagnostics.com)		NextGen Sequencing		
Chronic Inflammatory Demyelinating Polyneuropathy (CIDP) Test Panel - Nodal and Paranodal Antibodies	336	Neurofascin 140 (NF140), Neurofascin 186 (NF186); Contactin-1 (CNTN1), Contactin-associated protein 1 (CASPR1), and Neurofascin 155 (NF155)	Serum or CSF. Store at -20°C.	BC Neuroimmunology (bcneuro.ca)		CBA fixed	Replaces contactin and neurofascin tesing at Washington U.	31-Oct-20
Chronic Pancreatitis NGS Panel	402	CASR, CFTR, CTRC, PRSS1, SPINK1	Lavender Top (EDTA) 2 x 4 mL	Fulgent Genetics (fulgentdiagnostics.com)		NextGen Sequencing		8-Feb-19
CLCN1 Full Gene sequencing Analysis [MOL355]; Paramyotonia Congenita (168300)	607	CLCN1 (118425)	Lavender Top (EDTA)	MNG Laboratories (www.medicalneurogenetics.com)/Dynacare	Paramyotonia Congenita (168300)	Sanger sequencing		14-May-20
COCKAYNE SYNDROME VIA THE ERCC6 GENE [1008]		ERCC6 (609413)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Cockayne Syndrome, Type B (133540)	Sanger sequencing		
Collagen Type II Antibodies [FFTYC]	1399		Red Top; Gold SST. Stable for 7 d at 4°C or longer at -20°C.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)		Enzyme Linked Immunosorbent Assay (ELISA)		15-May-19
Collagen VII IgG Antibody Level	1399		Red Top or Gold SST. Stability Ambient: 7 days Refrigerated: 14 days Frozen: Indefinitely	Immunodermatology Laboratory Department of Dermatology 30 North 1900 East, 4A330 SOM Salt Lake City, Utah 84132 Email: immunoderm@hsc.utah.edu		Enzyme Linked Immunosorbent Assay (ELISA)		6-Jul-20
Colon Cancer NGS Panel	231	APC, AXIN2, BMPR1A, BUB1B, CDH1, CDKN2A, CHEK2, EPCAM, EXO1, FLCN, GALNT12, MLH1, MSH2, MSH6, MUTYH, PMS1, PMS2, PTEN, SMAD4, STK11, TP53	Lavender Top (EDTA) 2 x 4 mL	Fulgent Genetics (fulgentdiagnostics.com)		NextGen Sequencing		Decmber 12, 2018
Colorectal cancer (Li-Fraumeni syndrome)	231	TP53 (191170)	Lavender Top (EDTA)	Molecular Genetics Laboratory, Hospital for Sick Children (www.sickkids.ca/PaediatricLaboratoryMedicine/Laboratories-Services/Molecular-Genetics-Laboratory/index.html)	Colorectal cancer (114500); Li-Fraumeni syndrome (151623)	1. Sanger sequencing 2. gene dosage		
ColoVantage (Methylated Septin 9) [16983]	1395	SEPT9	Plasma (EDTA); handle at 4°C; freeze plasma in plastic container. Minimum 10 mL.	Quest Diagnostics Chantilly 14225 Newbrook Dr. Chantilly, VA 20153-0841				11-Apr-18
Combined Mito Genome Plus Mito Nuclear Gene Panel [615]	518	AARS, AARS2, ABCB11, ABCB4, ABCB7, ABCD4, ACAD9, ACADM, ACADVL, ACO2, ACSF3, ADCK3 (CABC1; COQ8), ADCK4, AFG3L2, AGK, AGL, AIFM1, ALAS2, ALDOA, ALDOB, ALG1, ALG11, ALG13, ALG2, ALG3, ALG6, ALG9, AMACR, APOPT1, APTX, ARG1, ASL, ASS1, ATP5A1, ATP5E, ATP7B, ATP8B1, ATPAF2 (ATP12), AUH, B4GALT1, BCKDHA, BCKDHB, BCS1L, BOLA3, C10ORF2, C12ORF65, C19orf12, CA5A, CARS2, CHKB, CISD2, CLPB, COA5 (C2ORF64), COA6, COASY, COG4, COG5, COG6, COG7, COG8, COQ2, COQ4, COQ6, COQ9, COX10, COX14 (C12ORF62), COX15, COX20 (FAM36A), COX42, COX6A1, COX6B1, COX7B, CPS1, CPT1A, CPT2, CYC1, DARS, DARS2, DBT, DDHD1, DDHD2, DDOST, DGUOK, DLAT, DLD, DMGDH, DNA2, DNAJC19, DNM1L, DNM2, DOLK, DPAGT1, DPM1, DPM3, EARS2, ECHS1, ELAC2, ENO3, ETFA, ETFB, ETFDH, ETHE1, FAH, FARS2, FASTKD2, FBP1, FBXL4, FDX1L, FH, FLAD1, FOXRED1, G6PC, GAA, GAMT, GARS, GATM, GBE1, GCDH, GFER, GFM1 (EFG1), GFM2, GLRX5, GMPPA, GSS, GTPBP3, GYG1, GYG2, GYS1, GYS2, HADHA, HADHB, HARS2, HCFC1, HIBCH, HLCS, HMGCL, HMGCS2, HSD17B10, HSPD1, IARS2, IBA57, ISCA2, ISCU, IVD,	Lavender Top (EDTA)	GeneDx (www.genedx.com)	Cerebral Creatine Deficiency Syndrome-1 (CCDS1), Combined D-2- and L-2-Hydroxyglutaric Aciduria, Oxidative Phosphorylation (OXPHOS) Deficiency, Congenital Disorder of Glycosylation, Congenital Sideroblastic Anemia with B-cell Immunodeficiency, Periodic fevers, and Developmental delay (SIFD), Fanconi-Bickel Syndrome, Glutathione Synthetase Deficiency (GSSD), Glycogen Storage Disease IIIa; Glycogen Storage Disease IIIb, Glycogen Storage Disease IV, Glycogen Storage Disease IXc (GSD9C), Glycogen Storage Disease IXd (GDS9D), Glycogen Storage Disease 0, Muscle (GSD0B), Glycogen Storage Disease Type IXa, Glycogen Storage Disease Type IXb (GSD9B), Glycogen Storage Disease VII (GSD7), Glycogen Storage Disease X (GSD10), Glycogen Storage Disease XI (GSD11), Glycogen Storage Disease XII (GSD12), Glycogen Storage Disease XIII (GSD13), Glycogen Storage Disease XV (GSD15), Hereditary Fructose Intolerance, Alacrima, Achalasia, and Mental Retardation Syndrome (AAMR), 2,4-Dienoyl-CoA Reductase Deficiency (DECRD),			9-Dec-19
Common Variable Immunodeficiency Panel (Inviate) [08112]	1042	CD27, CR2, CTLA4, ICOS, IL21, IL21R, LRBA, NFKB2, PIK3CD, PIK3R1, PLCG2, PRKCD, RAC2, STAT3, TNFRSF13B, TNFRSF13C, TNFSF12	Lavender Top (EDTA)	Invitae (www.invitae.com)		Next-Generation sequencing		2-Dec-07
Complement Profile (C2, C3, C4, C5, C7, C8, C9, C1Q, Factor B, Factor H, Factor I. Properdin, C1 inhibitor, C4HP)			0.5 mL red top serum- spun, separated, frozen within 2 hrs of collection; ship on dry ice	Cincinnati Children's Hospital Medical Center 3333 Burnet Avenue NRB 1042 Cincinnati, OH 45229 (www.cincinnatichildrens.org)				15-Nov-17

Appelation approuvée de l'analyse ou maladie associée [avec numéro de phénotype MIM] / Approved test name or Associated Disease [with MIM Phenotype Number]	**Code Test /** Test Code **(Dictionnaire MSSS)**	**SYMBOL HGNC DU GÈNE et NOM OMIM DU GÈNE /** HGNC Gene Symbol and OMIM Gene Number	**RÈGLES DE MANUTENTION /** Handling Conditions	**LABORATOIRE DÉSIGNÉ /** Designated laboratory	**RENSEIGNEMENTS SUR L'ANALYSE /** Test details	**MÉTHODE /** Methode	**NOTE PARTICULIÈRE /** Special comment	**Date de la révision /** Date of revision
Complement System Disorder Panel Plus Anlaysis [IM0701]	803	ADIPOQ Complement system AD/AR 2 8 ADIPOR1* Complement system AD/AR 4 ADIPOR2 Complement system AD/AR ARMC4* Ciliary dyskinesia AR 13 15 C1QA C1q deficiency AR 2 7 C1QB C1q deficiency AR 4 7 C1QBP Primary immunodeficiency AD/AR 6 C1QC C1q deficiency AR 4 7 C1R Immunodeficiency AD/AR 14 16 C1S Complement component C1s deficiency AR 4 8 C2* Complement component 2 deficiency AR 4 6 C3 Hemolytic uremic syndrome, atypical, Complement component 3 deficiency AD/AR 5 82 C3AR1 Complement system AD/AR 1 3 C4A* Blood group, Chido/Rodgers system BG 1 5 C4B* Complement component 4B deficiency AR 8 C4BPA Complement system AD/AR 2 C4BPB Complement system AD/AR C5 Eculizumab, poor response to, Complement component 5 deficiency AD/AR 5 17 C5AR1 Complement system AD/AR C5AR2 Complement system AD/AR 2 C6 Complement component 6 deficiency AR 7 11 C7 Complement component 7 deficiency AR 14 29 C8A Complement component 8 deficiency AR 2 5 C8B Complement component 8 deficiency AR 7 7 C8G Immunodeficiency AD/AR C9 Complement component 9 deficiency AR 7 7 CCDC39 Ciliary dyskinesia AR 16 38 CCDC40 Ciliary dyskinesia AR 19 32 CCDC65 Ciliary dyskinesia AR 1 CCDC103 Ciliary dyskinesia AR 3 4 CCDC114 Ciliary dyskinesia AR 6 7 CCNO Ciliary dyskinesia AR 9 9 CD46* Hemolytic uremic syndrome, atypical AD/AR 4 64 CD55 Blood group, Cromer system BG 7 6 CD59 CD59 deficiency AR 3 6 CD93 Complement system AD/AR CFB Complement factor B deficiency, Hemolytic uremic syndrome, atypical AD/AR 2 21 CFD Complement factor D deficiency AR 2 3 CFH* Hemolytic uremic syndrome, atypical, Complement factor H deficiency AD/AR 17 259 CFI Hemolytic uremic syndrome, atypical, Complement factor I	Lavender Top (EDTA)	Blueprint Genetics http://blueprintgenetics.com/				2-Feb-18
Comprehensive Cardiomyopathy NGS Panel	289		Lavender Top (EDTA)	Fulgent Genetics (fulgentdiagnostics.com)				3-Nov-20
Comprehensive Cardiovascular NGS Panel	286		Lavender Top (EDTA)	Fulgent Genetics (fulgentdiagnostics.com)				3-Nov-20
Comprehensive Cellular Energetics Defects (NextGen Sequencing Panel and Copy Number Analysis; 320 Genes + mtDNA) [NGS301]	636	AARS2 (612035), ABCB7 (300135), ABCB8 (605464), ABCD1 (300371), ABCF2 (612510), ABHD5 (604780), ACAA1 (604054), ACACA (200350), ACACB (601557), ACAD8 (604773), ACAD9 (611103), ACADL (609576), ACADM (607008), ACADS (606885), ACADSB (600301), ACADVL (609575), ACAT1 (607809), ACAT2 (100678), ACLY (108728), ACO1 (100880), ACO2 (100850), ACOX1 (609751), ACOX2 (601641), ACP6 (611471), ACSBG1 (614362), ACSF3 (614245), ACSL1 (152425), ACSL3 (602371), ACSL4 (300157), ACSM1 (614357), ACSM2B (614359), ACSM3 (145505), ADCK3 (606980), ADHFE1 (611083), AFG3L2 (604581), AGK (610345), AGL (610860), AIFM1 (300169), AK2 (103020), AK3 (609290), ALAD (125270), ALAS2 (301300), ALDH3A2 (609523), ALDOA (103850), ALDOB (612724), ALDOC (103870), ANO10 (613728), APOPT1 (616003), APTX (606350), ARMS2 (611313), ARX (300382), ATAD3B (612317), ATP5A1 (164360), ATP5B (102910), ATP5E (606153), ATP7B (606882), ATPAF2 (608918), AUH (600529), BAAT (602938), BAX (600040), BBOX1 (603312), BCKDHA (608348), BCKDHB (248611), BCS1L (603647), BOLA3 (613183), BPGM (613896), BTD (609019), C10ORF2 (606075), C12orf65 (613541), C21orf33 (607962), CA5A (114761), CACNA1S (114208), CALM1 (114180), CALM2 (114182), CALM3 (114183), CARS2 (612800), CCT7 (605140), CHCHD10 (615903), CHKB (612395), CISD2 (611507), CKMT1B (123290), CKMT2 (123295), CLPP (601119), COA5 (613920), COQ2 (609825), COQ4 (612898), COQ6 (614647), COQ7 (601683), COQ9 (612837), COX10 (602125), C12ORF62 (614478), COX15 (603646), FAM36A (614698), COX411 (123864), COX412 (607976), COX6A1 (602072), COX6B1 (124089), COX7A1 (123995), COX7B (603792), COX8A (123870), CPOX (612732), CPT1A (600528), CPT1B (601987), CPT1C (608846), CPT2 (600650), CS (118950), CYC1 (123980), CYCS (123970), CYP4A11 (601310), DARS (603084), DARS2 (610956), DBT (248610), DGUOK (601465), DHTKD1 (614984), DLAT (608770), DLD (238331), DLST (126063), DNA2 (601810), DNAJC19 (608977), DNM1L (603850), EARS2 (612799), ECHS1 (602292), EC11 (600305), ECSIT (608388), EHHADH (607037), ELOVL4 (605512), ELOVL5 (611805), ENO1 (172430), ENO2 (131360), ENO3 (131370), ETFA (608053), ETFB (130410), ETFDH (231675), ETHE1 (608451), FABP1 (134650), FABP2 (134640), FABP3 (134651), FABP7 (602965), FADS2 (606149), FARS2 (611592), FASN (600212), FASTKD2 (612322), FBP1 (611570), FBXL4 (605654), FDPS (134629), FECH (612386), FH (136850), FOLR1 (136430), FOXRED1 (613622), FXN (606829), G6PC (613742), G6PC3 (611045), G6PD (305900), GAA (606800), GAMT (601240), GAPDH (138400), GARS (600287), GATM (602360), GBE1 (607839), GCDH (608801), GCK (138079), GFER (600924), GFM1 (606639), GEM2 (606544), GPAM (602395), GPD1 (138420), GPD2	Lavender Top (EDTA)	MNG Laboratories (www.medicalneurogenetics.com)/Dynacare		Next-Generation sequencing		14-May-20

Appelation approuvée de l'analyse ou maladie associée [avec numéro de phénotype MIM] / Approved test name or Associated Disease [with MIM Phenotype Number]	**Code Test** / Test Code **(Dictionnaire MSSS)**	**SYMBOL HGNC DU GÈNE et NOM OMIM DU GÈNE** / HGNC Gene Symbol and OMIM Gene Number	**RÈGLES DE MANUTENTION /** Handling Conditions	**LABORATOIRE DÉSIGNÉ** / Designated laboratory	**RENSEIGNEMENTS SUR L'ANALYSE** / Test details	**MÉTHODE /** Methode	**NOTE PARTICULIÈRE /** Special comment	**Date de la révision /** Date of revision
Comprehensive Dystonia (NGS Panel and Copy Number + mtDNA) [NGS358]	1096	AARS AARS2 AASS ABCA7 ABCD1 ABCD4 ABHD12 ACOX1 ACP2 ACP5 ACTB ACVR1 ADAMTS13 ADAR ADCY5 ADD3 ADH1C ADRA2B AFG3L2 ALDH6A1 ALS2 ANO3 AP1S2 AP3B2 AP3D1 AP4B1 APTX ARHGAP31 ARSA ARV1 ARX ATCAY ATM ATP13A2 ATP1A2 ATP1A3 ATP2B3 ATP7B ATR AUH B4GALNT1 BCAP31 BCL11B BCS1L BICD2 BRAT1 BSCL2 C11orf73 C19orf12 CA2 CACNA1A CACNA1B CACNA1D CARS2 CHMP2B CIT CKAP2L CLPB COASY COL4A1 COL6A3 COQ9 COX10 COX15 CP CRLF1 CTC1 CYB5R3 CYP27A1 DAG1 DCAF17 DDC DDX3X DLAT DLD DMXL2 DNAJC12 DNAJC6 DOCK6 DPYS DRD5 DYNC1H1 EARS2 ECHS1 ECM1 ELAC2 ELP2 EMC1 ERCC6 ERCC8 ERLIN1 FA2H FAR1 FASTKD2 FBXL4 FBXO7 FGFR1 FOXG1 FOXRED1 FRRS1L FTL GAMT GBA GCDH GCH1 GJA1 GJC2 GLB1 GLUD2 GLYCTK GM2A GNA11 GNAL GNAO1 GNAS GPR88 GRIK2 GTF2E2 GTF2H5 HACE1 HIBCH HNRNPH2 HPCA HPRT1 HTRA2 HTT IFIH1 IFNG ISG15 JAM3 JPH3 KCNJ10 KCNMA1 KCNQ2 KCTD17 KMT2B KRIT1 L2HGDH LIPT2 MAPT MARS2 MAT1A MCCC2 MCOLN1 MDH2 MECP2 MECR MFF MGP MICU1 MMADHC MORC2 MPV17 MRE11A MRPS34 MTO1 NADK2 NALCN NDUFA10 NDUFA12 NDUFA2 NDUFA9 NDUFAF2 NDUFAF6 NDUFS3 NDUFS4 NDUFS7 NDUFS8 NHP2 NKX2-1 NME1 NPC1 NPC2 NUP62 OBFC1 OCLN PAH PANK2 PARK2 PCCA PCCB PDE10A PDE8B PDGFB PDGFRB PDHA1 PDHX PET100 PINK1 PLA2G6 PLEKHG2 PLP1 PNKD PNKP PNP PNPLA8 PNPT1 POLR1C POLR3A PPP1R15B PRKCG PRKRA PRRT2 PSEN1 PSMB8 PTEN PTS QDPR RAB18 RBBP8 RLIM RNASEH1 RNASEH2A RNASEH2B RNASEH2C RNASET2 RNF125 RNF216 SAMHD1 SCP2 SDHA SDHAF1 SDHD SERAC1 SETX SFXN4 SGCE SLC19A3 SLC20A2 SLC25A15 SLC25A26 SLC2A1 SLC30A10 SLC30A9 SLC39A14 SLC39A4 SLC46A1 SLC6A17 SLC6A3 SLC6A8 SLC9A1 SNCA SNORD118 SPG20 SPR SQSTM1 STAMBP STUB1 SUCLA2 SUOX SURF1 SYNJ1 TAF1 TAF2 TANGO2 TBC1D24 TELO2 TENM4 TH THAP1 THOC2 TIMM8A TIMMDC1 TINF2 TMEM126B TOR1A TPI1 TPK1 TRAPPC11 TREM2 TREX1 TSEN2 TSFM TTC19 TUBB4A TXN2 TYROBP UBA5 UBQLN2 UBTF UPB1 UQCRQ UROC1 USP18 VAC14 VAMP1 VCP VPS13A VPS13C VPS37A VWA3B WDR45 WDR73 XK XPNPEP3 XPR1 XRCC4 ZBTB20 ZC4H2 ZNF592	Lavender Top (2 x 4 mL)	MNG Laboratories (www.medicalneurogenetics.com)/Dynacare		NextGen Sequencing		14-May-20
Comprehensive Epilepsy (813 genes + Del/Dup + mtDNA) [NGS385]	356	ABAT (137150), ABCC8 (600509), ACADSB (600301), ACSF3 (614245), ADSL (608222), ALDH5A1 (610045), ALDH7A1 (107323), ALG11 (613666), AMT (238310), ARHGEF15 (608504), ARHGEF9 (300429), ARX (300382), ASAH1 (613468), ATIC (601731), ATP6AP2 (300556), AUTS2 (607270), BOLA3 (613183), C10ORF2 (606075), CACNA1A (601011), CACNA1G (604065), CACNA1H (607904), CACNA2D2 (607082), CACNB4 (601949), CACNG3 (606403), CADPS2 (609978), CDKL5 (300203), CERS1 (606919), CHD2 (602119), CHRNA2 (118502), CHRNA4 (118504), CHRNB2 (118507), CLCN2 (600570), CLCN4 (302910), CLN3 (607042), CLN5 (608102), CLN6 (606725), CLN8 (607837), CNTNAP2 (604569), COG7 (606978), COG8 (606979), COL6A2 (120240), COQ2 (609825), COQ9 (612837), CPA6 (609562), CSTB (601145), CTSD (116840), D2HGDH (609186), DEPDC5 (614191), DNAJC5 (611203), DNM1 (602377), DOCK7 (615730), DPAGT1 (191350), DYRK1A (600855), EEF1A2 (602959), EFHC1 (608815), EPM2A (607566), ETHE1 (608451), FARS2 (611592), FASTKD2 (612322), FH (136850), FOLR1 (136430), FOXG1 (164874), GABRA1 (137160), GABRB3 (137192), GABRD (137163), GABRG2 (137164), GAMT (601240), GATM (602360), GCK (138079), GCSH (238330), GFM1 (606639), GJA1 (600309), GLUD1 (606762), GLUL (610015), GNAO1 (139311), GOSR2 (604027), GPHN (603930), GRIN1 (614254), GRIN2A (138253), GRIN2B (138252), GRN (138945), HCFC1 (300019), HCN1 (602780), HCN2 (602781), HSD17B10 (300256), IBA57 (615316), IER3IP1 (609382), INHA (147380), INPP4A (600916), KCNB1 (600397), KCNH2 (152427), KCNH5 (605716), KCNJ11 (600937), KCNMA1 (600150), KCNQ2 (602235), KCNQ3 (602232), KCNT1 (608167), KCTD7 (611725), LAMC3 (604439), LGI1 (604619), LGI4 (608303), LIAS (607031), MANBA (609489), MAPK10 (602897), ME2 (154270), MECP2 (300005), MFSD8 (611124), MLYCD (606761), MOCS1 (603707), MOCS2 (603708), MRPL12 (602375), MTHFR (607093), NECAP1 (611623), NEU1 (608272), NHLRC1 (608072), NOL3 (605235), NRXN1 (600565), OCLN (602876), PCDH19 (300460), PDSS2 (610564), PGK1 (311800), PHGDH (606879), PIGM (610273), PIGO (614730), PLCB1 (607120), PNKP (605610), PNPO (603287), POLG (174763), PPT1 (600722), PRICKLE1 (608500), PRICKLE2 (608501), PRRT2 (614386), PURA (600473), RMND1 (614917), ROGDI (614574), SCARB2 (254900), SCN1A (182389), SCN1B (600235), SCN2A (182390), SCN8A (600702), SCN9A (603415), SERPINI1 (602445), SGCE (604149), SLC13A5 (608305), SLC25A22 (609302), SLC2A1 (138140), SLC35A2 (314375), SLC6A8 (300036), SLC9A6 (300231), SMC1A (300040), SPR (182125), SPTAN1 (182810), ST3GAL3 (615006), ST3GAL5 (604402), STXBP1 (602926), SUOX (606887), SYN1 (313440), SYNGAP1 (603384), SZT2 (615463), TBC1D24 (613577), TCF4 (602272), TPP1 (607998), TSC1 (605284)	Lavender Top (EDTA)	MNG Laboratories (www.medicalneurogenetics.com)/Dynacare		NextGen Sequencing	Alternative: Fulgent Epilepsy NGS Panel	28-Jul-20

Appelation approuvée de l'analyse ou maladie associée [avec numéro de phénotype MIM] / Approved test name or Associated Disease [with MIM Phenotype Number]	**Code Test** / Test Code **(Dictionnaire MSSS)**	**SYMBOL HGNC DU GÈNE et NOM OMIM DU GÈNE** / HGNC Gene Symbol and OMIM Gene Number	**RÈGLES DE MANUTENTION /** Handling Conditions	**LABORATOIRE DÉSIGNÉ** / Designated laboratory	**RENSEIGNEMENTS SUR L'ANALYSE** / Test details	**MÉTHODE /** Methode	**NOTE PARTICULIÈRE /** Special comment	**Date de la révision /** Date of revision
Comprehensive Epilepsy NGS Panel	356	ABAT, ABCB1, ABCC8, ACY1, ADAR, ADCK3, ADGRG1, ADGRV1, ADSL, AGA, AHI1, AKT3, ALDH4A1, ALDH5A1, ALDH7A1, ALG1, ALG12, ALG13, ALG2, ALG3, ALG6, ALG8, ALG9, AMACR, AMT, ANK3, APTX, ARFGEF2, ARG1, ARHGEF15, ARHGEF9, ARL13B, ARSA, ARSB, ARX, ASNS, ASPA, ASPM, ATIC, ATP13A2, ATP1A2, ATP2A2, ATP6AP2, ATP6V0A2, ATPAF2, ATR, ATRX, AUH, B4GALT1, BCKDK, BCS1L, BOLA3, BRAF, BRAT1, BRD2, BTD, BUB1B, C10orf2, C12orf57, C12orf65, CACNA1A, CACNA1H, CACNA2D2, CACNB4, CASK, CASR, CBL, CC2D2A, CCDC88C, CCL2, CDK5RAP2, CDKL5, CDON, CELSR1, CENPJ, CEP152, CEP290, CHD2, CHRNA2, CHRNA4, CHRNA7, CHRNB2, CLCN2, CLCN4, CLCNKA, CLCNKB, CLN3, CLN5, CLN6, CLN8, CNTN2, CNTNAP2, COG1, COG7, COG8, COL18A1, COL4A1, COQ2, COQ9, COX10, COX15, CPA6, CPT2, CRH, CSTB, CTSA, CTSD, CTSF, CUL4B, DCX, DEPDC5, DHCR7, DHFR, DLD, DNAJC5, DNM1, DOCK7, DOLK, DPAGT1, DPM1, DPM2, DPM3, DPYD, DYNC1H1, DYRK1A, EEF1A2, EFHC1, EFHC2, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EMX2, EPM2A, ETFA, ETFB, ETFDH, FARS2, FASN, FGD1, FGF8, FGFR3, FH, FKRP, FKTN, FLNA, FLVCR2, FOLR1, FOXG1, FUCA1, GABBR2, GABRA1, GABRB2, GABRB3, GABRD, GABRG2, GALC, GALNS, GAMT, GATM, GCDH, GCSH, GFAP, GLB1, GLDC, GLI2, GLI3, GLRA1, GLRB, GLUD1, GNAO1, GNE, GNPTAB, GNPTG, GNS, GOSR2, GPC3, GPHN, GRIA3, GRIN1, GRIN2A, GRIN2B, GRN, GUSB, HCN1, HCN2, HCN4, HDAC4, HEXA, HEXB, HGSNAT, HNRNPH1, HNRNPU, HPD, HRAS, HSD17B10, IDS, IDUA, IQSEC2, KANSL1, KAT6B, KCNA1, KCNA2, KCNAB2, KCNB1, KCNC1, KCNH2, KCNH5, KCNJ1, KCNJ10, KCNJ11, KCNMA1, KCNQ2, KCNQ3, KCNT1, KCNV2, KCTD7, KDM5C, KDM6A, KIAA2022, KIF1BP, KMT2D, KPNA7, KRAS, L2HGDH, LAMA2, LARGE, LBR, LGI1, LIAS, LIG4, LRPPRC, MAGI2, MAP2K1, MAP2K2, MAPK10, MBD5, MCOLN1, MCPH1, ME2, MECP2, MED12, MED17, MEF2C, MFSD8, MGAT2, MLC1, MOCS1, MOCS2, MOGS, MPDU1, MPI, MTHFR, MTOR, NAGLU, NDE1, NDUFA1, NDUFA2, NDUFS1, NDUFS3, NDUFS4, NDUFS7, NDUFS8, NDUFV1, NEDD4L, NEU1, NF1, NHEJ1, NHLRC1, NIPA2, NIPBL, NODAL, NOTCH3, NPC1, NPC2, NPHP1, NR2F1, NRAS, NRXN1, NSD1, NTNG1, OFD1, OPHN1, PAFAH1B1, PAK3, PANK2, PAX6, PC, PCDH19, PCNT, PDHA1, PDSS1, PDSS2, PEX1, PEX12, PEX14, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PGK1, PHF6, PIGA, PIGO, PIGV, PIK3AP1, PLA2G6, PLCB1, PLP1, PMM2, PNKP, PNPO, POLG, POMGNT1, POMT1, POMT2, PPT1, PQBP1, PRICKLE1, PRICKLE2, PRODH, PRRT2, PSAP, PTCH1, PTPN11, PURA, QARS, QDPR, RAB39B, RAB3GAP1, RAF1, RAI1, RANGAP1, RARS2, RBFOX1, RBFOX3, RELN, RET1, RNASEH2A, RNASEH2B,	Lavender Top (2 x 4 mL)	Fulgent Genetics (fulgentdiagnostics.com)		NextGen Sequencing		
Comprehensive Leukodystrophy/Leukoencephalopathy (NGS Panel and Copy Number Analysis + mtDNA) [NGS372]	964		Lavender Top (EDTA)	MNG Laboratories (www.medicalneurogenetics.com)/Dynacare		Next-Generation sequencing		29-Sep-20
Comprehensive Metabolism NGS Panel	1891	ABCC8, ABCD1, ABCD4, ACAA1, ACAD9, ACADL, ACADM, ACADS, ACADVL, ACAT1, ACOX1, ACSF3, ACY1, ADAMTSL2, ADAR, ADSL, AGA, AGK, AGL, AGPAT2, AKT2, ALAD, ALAS2, ALDH5A1, ALDH6A1, ALDH7A1, ALDOA, ALG1, ALG11, ALG12, ALG13, ALG2, ALG3, ALG6, ALG8, ALG9, AMACR, AMT, ANO10, ANTXR2, APTX, ARG1, ARSA, ARSB, ASAH1, ASL, ASPA, ASS1, ATP13A2, ATP6V0A2, AUH, B3GLCT, B4GALT1, BCKDHA, BCKDHB, BSCL2, BTD, C12orf65, CA2, CACNA1S, CAV1, CAV3, CAVIN1, CBS, CD320, CKMT1A, CKMT1B, CKMT2, CLCN1, CLDN16, CLDN19, CLN3, CLN5, CLN6, CLN8, CNNM1, CNNM2, CNNM4, COG1, COG4, COG5, COG6, COG7, COG8, COL11A2, COL2A1, COQ2, COQ6, COQ8A, COQ9, CPOX, CPS1, CPT1A, CPT1B, CPT2, CTNS, CTSA, CTSC, CTSD, CTSK, DBT, DDOST, DGUOK, DHCR7, DHDDS, DLD, DOLK, DPAGT1, DPM1, DPM2, DPM3, DPYD, DYM, ECHS1, EGF, ENO3, EPM2A, ETFA, ETFB, ETFDH, FAH, FBP1, FBXL4, FECH, FH, FLNA, FLNB, FOLR1, FUCA1, FXYD2, G6PC, GAA, GALC, GALNS, GAMT, GATM, GBA, GBE1, GCDH, GCH1, GCK, GIF, GLA, GLB1, GLDC, GLUD1, GMPPA, GNE, GNPAT, GNPTAB, GNPTG, GNS, GPC3, GPHN, GUSB, GYG1, GYS1, GYS2, HADH, HADHA, HADHB, HAMP, HCFC1, HEXA, HEXB, HFE, HGSNAT, HJV, HLCS, HMBS, HMGCL, HMGCS2, HNF1A, HNF1B, HNF4A, HPD, HRAS, HSD17B10, HSD17B4, HYAL1, IDS, IDUA, IFIH1, INSR, ISCU, IVD, KCNA1, KCNJ11, KCNJ2, L2HGDH, LAMA2, LAMP2, LDB3, LDHA, LIPA, LMBRD1, LMNA, LPIN1, MAGT1, MAN1B1, MANBA, MCCC1, MCCC2, MCEE, MCOLN1, MFN2, MFSD8, MGAT2, MMAA, MMAB, MMACHC, MMADHC, MMGT1, MOCS1, MOCS2, MOGS, MPDU1, MPI, MPV17, MTHFR, MTR, MTRR, MUT, MYOT, NAGLU, NAGS, NDUFS1, NEU1, NHLRC1, NIPA2, NPC1, NPC2, OAT, OPA1, OPA3, OTC, OXCT1, PAH, PC, PCBD1, PCCA, PCCB, PCK1, PCK2, PDHA1, PDHB, PDHX, PDSS1, PDSS2, PDX1, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX16, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PFKM, PGAM2, PGK1, PGM1, PHKA1, PHKA2, PHKB, PHKG1, PHKG2, PHYH, PLIN1, PLPBP, PMM2, POLG, POLG2, PPARG, PPOX, PPT1, PRKAG2, PRKAG3, PRODH, PSAP, PTS, PYGL, PYGM, QDPR, RAI1, RBCK1, RFT1, RNASEH2A, RNASEH2B, RNASEH2C, RPN2, RRM2B, RYR1, SAMHD1, SCN4A, SEC23B, SGSH, SLC12A3, SLC16A1, SLC17A5, SLC22A5, SLC25A13, SLC25A15, SLC25A20, SLC25A3, SLC25A4, SLC2A2, SLC30A10, SLC35A1, SLC35A2, SLC35C1, SLC37A4, SLC39A4, SLC3A1, SLC40A1, SLC41A2, SLC41A3, SLC46A1, SLC6A19, SLC6A8, SLC7A7, SLC7A9, SMPD1, SPG7, SRD5A3, SSR4, STT3A, STT3B, SUCLA2, SUCLG1, SUMF1, SUOX, TAZ, TBC1D4, TCF4, TCN2, TFR2, TIMM8A, TK2, TMEM126A, TMEM165, TMEM70,	Lavender Top (EDTA) 2 x 4 mL			NextGen Sequencing		28-Feb-20

Appelation approuvée de l'analyse ou maladie associée [avec numéro de phénotype MIM] / Approved test name or Associated Disease [with MIM Phenotype Number]	**Code Test** / Test Code **(Dictionnaire MSSS)**	**SYMBOL HGNC DU GÈNE et NOM OMIM DU GÈNE** / HGNC Gene Symbol and OMIM Gene Number	**RÈGLES DE MANUTENTION** / Handling Conditions	**LABORATOIRE DÉSIGNÉ** / Designated laboratory	**RENSEIGNEMENTS SUR L'ANALYSE** / Test details	**MÉTHODE** / Methode	**NOTE PARTICULIÈRE** / Special comment	**Date de la révision** / Date of revision
Comprehensive Muscular Dystrophy/Myopathy (NextGen Sequencing Panel and Copy Number Analysis; 909 Genes + mtDNA) [NGS330]	636	AAAS (605378), AARS (601065), AARS2 (612035), ABCC8 (600509), ABHD5 (604780), ACACA (200350), ACAD9 (611103), ACADS (606885), ACADVL (609575), ACTA1 (102610), ACY1 (104620), ADCK3 (606980), ADSSL1 (612498), AFG3L2 (604581), AGK (610345), AGL (610860), AGRN (103320), AIFM1 (300169), AIMP1 (603605), AIP (605555), ALAD (125270), ALDH18A1 (138250), ALDOA (103850), ALG2 (607905), ALS2 (606352), AMMECR1 (300195), AMPD1 (102770), ANO5 (608662), AP3B2 (602166), AP3D1 (607246), KIAA0415 (613653), APOA1 (107680), APOPT1 (616003), APTX (606350), AR (313700), ARSA (607574), ASAH1 (613468), ASXL2 (612991), ATL1 (606439), ATP1A3 (182350), ATP2A1 (108730), ATP7A (300011), B2M (109700), B3GALNT2 (610194), B3GNT1 (605517), B4GALT1 (137060), BAG3 (603883), BCL11B (606558), BCS1L (603647), BICD2 (609797), BIN1 (601248), BSCL2 (606158), C10ORF2 (606075), C12orf65 (613541), C19orf12 (614297), C9orf72 (614260), CACNA1A (601011), CACNA1S (114208), CAPN3 (114240), CASK (300172), CASQ1 (114250), CAV3 (601253), CCDC78 (614666), CCDC88A (609736), CFL2 (601443), CHAT (118490), CHCHD10 (615903), CHD4 (603277), CHKB (612395), CHMP2B (609512), CHRNA1 (100690), CHRNB1 (100710), CHRND (100720), CHRNE (100725), CHRNG (100730), CIT (605629), CLCN1 (118425), CLCNKB (602023), CNBP (116955), CNTN1 (600016), COL4A1 (120130), COL6A1 (120220), COL6A2 (120240), COL6A3 (120250), COLQ (603033), COQ2 (609825), COQ9 (612837), COX10 (602125), C12ORF62 (614478), COX15 (603646), FAM36A (614698), COX6A1 (602072), COX6B1 (124089), COX8A (123870), CPT1C (608846), CPT2 (600650), CRYAB (123590), CTNS (606272), CYP7B1 (603711), D2HGDH (609186), DARS2 (610956), DCPS (610534), DCTN1 (601143), DDHD1 (614603), DDHD2 (615003), DES (125660), DGUOK (601465), DHTKD1 (614984), DMD (300377), DMPK (605377), DNA2 (601810), DNAJB2 (604139), DNAJB6 (611332), DNAJC12 (606060), DNAJC19 (608977), DNM2 (602378), DOK7 (610285), DPAGT1 (191350), DPM1 (603503), DPM2 (603564), DPM3 (605951), DYNC1H1 (600112), DYSF (603009), EARS2 (612799), EGR2 (129010), EIF2B2 (606454), EIF2B4 (606687), EMD (300384), ENO3 (131370), ERBB4 (600543), ERCC6 (609413), ERLIN2 (611605), ETFA (608053), ETFB (130410), ETFDH (231675), EXOSC3 (606489), FAH (613871), FAM126A (610531), FAM134B (613114), FARS2 (611592), FASTKD2 (612322), FBLN5 (604580), FBN1 (134797), FGA (134820), FGD4 (611104), FGF12 (601513), FHL1 (300163), FIG4 (609390), FKBP14 (614505), FKRP (606596), FKTN (607440), FLAD1 (610595), FLNC (102565), FLVCR1 (609144), FOXRED1 (613622), C9ORF4 (604574), GAA (606800), GAN (605379), GARS (600287), GATM (602360), GBA2 (609471), GBE1 (607839), GCK (138079), GDAP1	Lavender Top (EDTA)	MNG Laboratories (www.medicalneurogenetics.com)/Dynacare		Next-Generation sequencing	See Prevention Genetics Panel	14-May-20
Comprehensive Neuropathies (NextGen Sequencing Panel and Copy Number Analysis; 869 genes + mtDNA) [NGS445]	643 (445)	43717 AAAS AARS AASS ABAT ABCA1 ABCB7 ABCC8 ABCD1 ABCD4 ABHD12 ACO2 ACOX1 ACP5 ACTA1 ACTB ACVR1 ACVRL1 ACY1 ADAMTS2 ADCK3 ADCY5 ADCY6 ADSL ADSSL1 AFG3L2 AGXT AIFM1 AIMP1 AKT1 ALAD ALDH18A1 ALDH3A2 ALDH5A1 ALDH6A1 ALG2 ALOX5AP ALS2 AMACR AMER1 AMN AMPD1 AMPD2 ANG ANKH ANKLE2 ANO5 AP1S1 AP1S2 AP3D1 AP4B1 AP4E1 AP4M1 AP4S1 AP5Z1 APOA1BP APOB APTX AR ARCN1 ARHGEF10 ARL13B ARL6IP1 ARNT2 ARSA ARX ASAH1 ASCC1 ASNS ASPA ASXL1 ATAD3A ATL1 ATL3 ATP13A2 ATP1A3 ATP2A1 ATP2B3 ATP6AP2 ATP7A ATP7B ATRX AUH B3GALT6 B3GALTL B4GALNT1 BAG3 BCAP31 BCOR BCS1L BICD2 BIN1 BMP2 BOLA3 BRAT1 BRPF1 BSCL2 BTNL2 BUB1B C10ORF2 C11orf73 C12orf65 C19orf12 CACNA1A CACNA1D CACNA1G CAPN1 CAPN3 CARS2 CASK CAV1 CAV3 CCDC88C CCND1 CCT5 CD59 CDK5 CECR1 CEP120 CEP55 CFH CFHR1 CFHR3 CFL2 CHCHD10 CHMP1A CHMP2B CHRNA1 CHRND CHRNG CISD2 CIT CKAP2L CLCF1 CLCN1 CLCN7 CLP1 CLPB CLPP CNBP CNTN2 CNTNAP1 COASY COG2 COL18A1 COL4A2 COL6A1 COL6A2 COL6A3 COMP COQ2 COQ4 COQ7 COQ9 COX10 COX15 COX6A1 COX7B CPOX CPT1C CREBBP CRYAB CSF1R CSPP1 CTC1 CTDP1 CTNNB1 CTSD CTSF CUBN CWF19L1 CYB5R3 CYP27A1 CYP2U1 CYP7B1 D2HGDH DARS DARS2 DCAF8 DCTN1 DDC DDHD1 DDHD2 DDOST DDR2 DDX3X DES DGUOK DHCR24 DHFR DHH DHTKD1 DKC1 DLAT DMXL2 DNA2 DNAJB2 DNAJC12 DNAJC3 DNM1L DNM2 DNMT1 DOCK6 DPAGT1 DPH1 DPM1 DSE DST DYNC1H1 EARS2 EBF3 ECE1 EGR2 EIF2B1 EIF2B2 EIF2B3 EIF2B4 EIF2B5 ELOVL4 ELOVL5 ELP2 EMC1 EML1 ENTPD1 EPG5 ERBB3 ERBB4 ERCC1 ERCC2 ERCC3 ERCC6 ERCC8 ERLIN1 ERLIN2 ESCO2 EVC EVC2 EXOSC3 EXOSC8 EXT1 FA2H FADD FAH FAM126A FAM134B FAR1 FARS2 FAT2 FBLN5 FBN1 FBXL4 FBXO38 FBXO7 FGD4 FGF14 FGFR1 FHL1 FIG4 FKRP FKTN FLNA FLNB FLNC FLVCR1 FLVCR2 FOLR1 FOXG1 FOXP1 FOXRED1 FTL FTO FUCA1 FUS FXN FXYD2 GAA GABRB3 GAD1 GALC GAN GARS GBA GBA2 GBE1 GCDH GCH1 GCLC GCSH GDAP1 GFAP GFM1 GIF GJA1 GJB1 GJC2 GLA GLB1 GLDC GLDN GLRX5 GLUD2 GLUL GLYCTK GM2A GMNN GNA11 GNAO1 GNB4 GOSR2 GPR56 GPT2 GPX4 GRIA3 GRID2 GRM1 GSN GSS GTF2E2 GUCY1A3 H19 HACE1 HADHA HADHB HARS HARS2 HERC1 HEXB HFE HINT1 HIVEP2 HK1 HLA-DQA1 HLA-DQB1 HLA-DRB1 HMBS HNRNPA1 HNRNPDL HOXB1 HPCA HPRT1 HSD17B4 HSPB1 HSPB3 HSPB8 HSPD1 HTRA1 HTT HYLS1 IARS2 IBA57 IDS IER3IP1 IFIH1 IGHMBP2 IKBKAP IKBKG INF2 IRF3 ISCA2 ISPD ITPA ITPR1 JAM3 JPH1 JPH3 KARS KATNB1 KCNA1 KCNC3 KCND3 KCNJ10	Lavender Top (EDTA)	MNG Laboratories (www.medicalneurogenetics.com)/Dynacare		NextGen Sequencing		14-May-20

Appelation approuvée de l'analyse ou maladie associée [avec numéro de phénotype MIM] / Approved test name or Associated Disease [with MIM Phenotype Number]	**Code Test /** Test Code **(Dictionnaire MSSS)**	**SYMBOL HGNC DU GÈNE et NOM OMIM DU GÈNE /** HGNC Gene Symbol and OMIM Gene Number	**RÈGLES DE MANUTENTION /** Handling Conditions	**LABORATOIRE DÉSIGNÉ /** Designated laboratory	**RENSEIGNEMENTS SUR L'ANALYSE /** Test details	**MÉTHODE /** Methode	**NOTE PARTICULIÈRE /** Special comment	**Date de la révision /** Date of revision
Comprehensive Ophthalmoplegia Syndromes (NextGen Sequencing Panel and Copy Number Analysis; 55 Genes + mtDNA) [NGS352]	1648	ACADS (606885), ACTA1 (102610), AFG3L2 (604581), AGRN (103320), APTX (606350), ATXN1 (601556), ATXN2 (601517), ATXN3 (607047), ATXN7 (607640), BIN1 (601248), C10ORF2 (606075), C12orf65 (613541), CHAT (118490), CHRNA1 (100690), CHRNB1 (100710), CHRND (100720), CHRNE (100725), CLPP (601119), COLQ (603033), DNM2 (602378), DOK7 (610285), EARS2 (612799), FOXE3 (601094), FOXRED1 (613622), GBA (606463), GRM1 (614831), HARS2 (600783), HCCS (300056), HSD17B4 (601860), KIF21A (608283), LARS2 (604544), C20ORF72 (615076), MPV17 (137960), MTM1 (300415), MTMR14 (611089), MUSK (601296), MYH2 (160740), OPA1 (605290), OPA3 (606580), PABPN1 (602279), POLG (174763), POLG2 (604983), RAPSN (601592), RRM2B (604712), RYR1 (180901), SCO2 (604272), SDHAF1 (612848), SEPN1 (606210), SLC19A3 (606152), SLC9A6 (300231), SPEG (615950), TPM3 (191030), TUBB3 (602661), TYMP (131222), ZNF592 (606937).	Lavender Top (EDTA)	MNG Laboratories (www.medicalneurogenetics.com)/Dynacare		NextGen Sequencing		14-May-20
Cone-Rod Dystrophy (CORDX3) via the CACNA1F gene []	1444	CACNA1F (300110)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Aland Island Eye Disease (300600); Cone-Rod Dystrophy X-Linked 3 (300476); Congenital Stationary Night Blindness, Type 2A (300071)	NextGen Sequencing		
Congenital Adrenal Hyperplasia NGS Panel	1326	ARMC5, CYP11B1, CYP11B2, CYP17A1, CYP21A2, HSD3B2, POR, PRKAR1A, STAR	Lavender Top (EDTA) 2 x 4 mL	Fulgent Genetics (fulgentdiagnostics.com)				28-Feb-20
Congenital Adrenal Hyperplasia Panel [EN0801]	1326	ARMC5 CYP11A1 CYP11B1* CYP11B2* CYP17A1 CYP21A2* HSBD3B2 PDE11A PDE8B POR PRKAR1A STAR	Lavender Top (EDTA) 2 x 4 mL	Blueprint Genetics http://blueprintgenetics.com/		NextGen Sequencing		28-Feb-20
Congenital Hyperinsulinism Sequencing Panel [1939]	541	ABCC8 (600509) GCK (138079) GLUD1 (138130) HADH (601609) HNF1A (142410) HNF4A (600281) KCNJ11 (600937) SLC16A1 (600682) UCP2 (601693)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Hyperinsulinemic Hypoglycemia, Familial 3 602485 Hyperinsulinemic Hypoglycemia, Familial 6 606762 Hyperinsulinemic Hypoglycemia, Familial, 1 256450 Hyperinsulinemic Hypoglycemia, Familial, 2 601820 Hyperinsulinemic Hypoglycemia, Familial, 4 609975 Hyperinsulinemic Hypoglycemia, Familial, 7 610021 Maturity-Onset Diabetes Of The Young, Type 1 125850 Maturity-Onset Diabetes Of The Young, Type 3 600496 Related Tests	Next-Generation sequencing		16-Aug-17
CONGENITAL MUSCULAR DYSTROPHY NEXTGEN SEQUENCING (NGS) PANEL [1301]	606	ITGA7 (600536); FKTN (607440); FKRP (606596); (LAMA2 (156225); LARGE (603590); POMT1 (607426); POMT2 (607439); POMGNT1 (606822); DAG1 (128239); DPM1 (603503); DPM3 (605951); CHKB (612395); ISPD (614631); LMNA (150330); GTDC2 (614828); TMEM5 (605862); B3GALNT2 (610194); GMPPB (615320); B3GNT1 (605517); GOSP2 (604027); SGK196 (615247); ST3GAL4 (104240)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Muscular Dystrophy-Dystroglycanopathy (Congenital with Brain and Eye Anomalies), Type A, 10; MDDGA10 (615041); Congenital Disorder Of Glycosylation Type 1E (608799); Congenital Disorder Of Glycosylation Type 1O(612937); Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 14(615352);Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 12 (615249) Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 13 (615287); Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies, type A, 11 (615181); Congenital Muscular Dystrophy-Dystroglycanopathy (With Brain And Eye Anomalies) Type A5 (613153); Merosin Deficient Congenital Muscular Dystrophy (607855); Muscular Dystrophy-Dystroglycanopathy (Limb-Girdle), Type C, 9 (613818); Walker-Warburg Congenital Muscular Dystrophy (614830); Muscular Dystrophy-Dystroglycanopathy (Limb-Girdle), Type C, 2 (613158); Muscular Dystrophy-Dystroglycanopathy (Congenital With Brain And Eye Anomalies), Type A, 6 (613154); Walker-Warburg Congenital Muscular Dystrophy 236670 Muscle Eye Brain Disease (253280); Fukuyama Congenital Muscular Dystrophy (253800); Muscular Dystrophy, Congenital, LMNA-Related (613205); Muscular Dystrophy, Congenital, Due To Integrin Alpha-7 Deficiency (613204);	NextGen Sequencing (22 genes)	See Med Neurogenetics Panel	

Appelation approuvée de l'analyse ou maladie associée [avec numéro de phénotype MIM] / Approved test name or Associated Disease [with MIM Phenotype Number]	**Code Test /** Test Code **(Dictionnaire MSSS)**	**SYMBOL HGNC DU GÈNE et NOM OMIM DU GÈNE /** HGNC Gene Symbol and OMIM Gene Number	**RÈGLES DE MANUTENTION /** Handling Conditions	**LABORATOIRE DÉSIGNÉ /** Designated laboratory	**RENSEIGNEMENTS SUR L'ANALYSE /** Test details	**MÉTHODE /** Methode	**NOTE PARTICULIÈRE /** Special comment	**Date de la révision /** Date of revision
Congenital Myasthenic Syndrome NGS Panel	606	AGRN, ALG14, ALG2, CHAT, CHRNA1, CHRNB1, CHRND, CHRNE, CHRNG, COL13A1, COLQ, DOK7, DPAGT1, GFPT1, GMPPB, LAMB2, LRP4, MUSK, MYO9A, PLEC, PREPL, RAPSN, SCN4A, SLC25A1, SLC5A7, SNAP25, STIM1, SYT2 (28 genes)	Lavender Top (2 x 4 mL)	Fulgent Genetics (fulgentdiagnostics.com)	Myasthenic syndrome, congenital, with pre- and postsynaptic defect s 615120); Myasthenic syndrome, congenital, associated with episodic apnea (254210); Multiple pterygium syndrome, lethal type (253290); Myasthenic syndrome, fast-channel congenital (608930); Myasthenic syndrome, slow-channel congenital (601462); Myasthenic syndrome, congenital, associated with acetylcholine receptor deficiency (608931); Myasthenic syndrome, slow-channel congenital (601462); Multiple pterygium syndrome, lethal type (253290); Myasthenic syndrome, fast-channel congenital (608930); Myasthenic syndrome, slow-channel congenital (601462); Myasthenic syndrome, congenital, associated with acetylcholine receptor deficiency (6089310; Myasthenic syndrome, fast-channel congenital (608930); Myasthenic syndrome, slow-channel congenital (601462); Endplate acetylcholinesterase deficiency (603034); Fetal akinesia deformation sequence (208150); Myasthenia, limb-girdle, familial (254300); Myasthenia, congenital, with tubular aggregates 1 (610542); Myasthenic syndrome, congenital, associated with acetylcholine receptor deficiency (608931); Fetal akinesia deformation sequence (208150); Myasthenic syndrome, congenital, associated with acetylcholine receptor deficiency (608931); Myasthenic syndrome, congenital, associated with facial dysmorphism and acetylcholine receptor deficiency (608931); Hyperkalemic periodic paralysis, type 2 (170500); Hyperkalemic periodic paralysis, type 2	Next-Generation sequencing		25-Jul-17
CONGENITAL MYOPATHY NEXTGEN SEQUENCING (NGS) PANEL [1365]	631		Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)		Next-Generation sequencing		
Congenital Neutropenia Panel [IM0501]	836	ACTB* CLPB CSF2RA#* CSF3R CTSC EFL1* ELANE G6PC3 GATA2 GFI1 GINS1 HAX1 IFNGR2 JAGN1 LAMTOR2 LYST MKL1 PGM3 RAC2 SBDS* SLC37A4 SMARCD2 SRP54 SRP72* VPS13B VPS45# WAS WDR1	Lavender Top (2 x 4 mL)	Blueprint Genetics http://blueprintgenetics.com/		NextGen Sequencing		28-Feb-20
Copeptin & Osmolality	1465		Serum or Plasma (Li heparin). Store and send frozen.	In-Common Laboratories	replaces vasopressin/ADH	Fluorescent immunoassay		29-Apr-19
Cornelia de Lange Syndrome		NIPBL (608667); SMC1A (300040); SMC3 (606062); HDAC8 (300269); RAD21 (606462)	Lavender Top (2 x 4 mL)	Fulgent Genetics (fulgentdiagnostics.com)	Cornelia de Lange syndrome 1 (122470); Cornelia de Lange syndrome 2 (300590); Cornelia de Lange syndrome 3 (610759); Cornelia de Lange syndrome 4 (614701); Cornelia de Lange syndrome 5 (300882)	Next-Generation sequencing		
Cortical Brain Malformations Panel [698]	346	ARFGEF2 (605371), ARX (300382), DCX (300121), EOMES (604615), FKRP (606596), FKTN (607440), FLNA (300017), GPR56 (604110), LAMC3 (604349), LARGE (603590), NDE1 (609449), OCLN (602876), PAFAH1B1 (601545), POMGNT1 (606822), POMT1 (607423), POMT2 (607439), RELN (600514), SRPX2 (300642), TUBA1A (602529), TUBA8 (605742), TUBB2B (612850), TUBB3 (602661), VLDLR(192977)	Lavender Top (EDTA)	GeneDx (www.genedx.com)	Lisencephaly: Lissencephaly and subcortical band heterotopia (SBH), Alpha- dystroglycanopathies, Periventricular nodular heterotopia (PVNH); Polymicrogyria	Exon Array CGH, Next-gen Sequencing		9-Dec-19
Corticotropin Releasing Factor (CRF, CRH)			3 ml EDTA plasma should be collected and separated as soon as possible. Plasma should be frozen immediately after separation	Inter Science Institute - 944 West Hyde Park Blvd, Inglewood, CA 90302		RIA	3/9/2017	
Cortisol, free [CORTF]			Red Top or Lavender Top	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)				31-Oct-20
Coxiella burnetii (Q fever), Molecular Detection, PCR, Blood [CBBRP]			Whole blood EDTA; Stable 7 days only at at 4°C or -20°C.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	Fièvre Q (Coxiella burnetii IgG et IgM) at CHUS Fleurimont			31-Oct-20
Craniosynostosis Non-Syndromic (select exons of FGFR3 gene)		FGFR3 (134934)	Lavender Top (EDTA)	Molecular Genetics Laboratory, Hospital for Sick Children (www.sickkids.ca/PaediatricLaboratoryMedicine/Laboratories-Services/Molecular-Genetics-Laboratory/index.html)	Craniosynostosis	Sanger sequencing; FGFR3 (p.Pro250Arg)		
Creatine Disorders Panel, Urine [CRDPU]		GATM (602360); SLC6A8 (300036)	Random urine	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	arginine:glycine amidinotransferase deficiency (602360/612718), guanidinoacetate methyltransferase deficiency (601240/612736), creatine transporter (SLC6A8) defect (300036/300352)			

Appelation approuvée de l'analyse ou maladie associée [avec numéro de phénotype MIM] / Approved test name or Associated Disease [with MIM Phenotype Number]	**Code Test** / Test Code **(Dictionnaire MSSS)**	**SYMBOL HGNC DU GÈNE et NOM OMIM DU GÈNE** / HGNC Gene Symbol and OMIM Gene Number	**RÈGLES DE MANUTENTION** / Handling Conditions	**LABORATOIRE DÉSIGNÉ** / Designated laboratory	**RENSEIGNEMENTS SUR L'ANALYSE** / Test details	**MÉTHODE** / Methode	**NOTE PARTICULIÈRE** / Special comment	**Date de la révision** / Date of revision
Crouzon Syndrome (select exons of FGFR2 and FGFR3 gene)		FGFR2 (176943); FGFR3 (134934)	Lavender Top (EDTA)	Molecular Genetics Laboratory, Hospital for Sick Children (www.sickkids.ca/PaediatricLaboratoryMedicine/Laboratories-Services/Molecular-Genetics-Laboratory/index.html)	Craniosynostosis: Crouzon syndrome (123500)	Sanger sequencing: FGFR2 (exon 7 and 8); FGFR3 (p.Pro250Arg)		
Cryoglobulin and Cryofibrinogen Panel, Serum and Plasma [CRGSP]			**Cryofibrinogen** **Collection** Container/Tube: Lavender top (EDTA) Submission Container/Tube: Plastic vial Specimen Volume: 1 mL Collection Instructions: 1. Tube must remain at 37 degrees C. 2. Centrifuge at 37 degrees C. (Do not use a refrigerated centrifuge. If absolutely necessary, ambient temperature is acceptable.) It is very important that the specimen remain at 37 degrees C until after separation of plasma from red cells. 3. Place plasma into an appropriately labeled plastic vial. **Cryoglobulin** **Collection** Container/Tube: Red top Submission Container/Tube: Plastic vial Specimen Volume: 5 mL Collection Instructions: 1. Tube must remain at 37 degrees C. 2. Allow blood to clot at 37 degrees C. 3. Centrifuge at 37 degrees C. (Do not use a refrigerated centrifuge. If absolutely necessary, ambient temperature is acceptable.) It is very important that the specimen remain at 37 degrees C until after separation of serum from red cells. 4. Place serum into an appropriately labeled plastic vial.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	Evaluating patients with vasculitis, glomerulonephritis, and lymphoproliferative diseases Evaluating patients with macroglobulinemia or myeloma in whom symptoms occur with cold exposure	immunofixation		
Cryopyrin-Associated Periodic Syndromes via the NLRP3 Gene [1638]		NLRP3 (606416)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Chronic Infantile Neurological, Cutaneous And Articular Syndrome (607115); Familial Amyloid Nephropathy With Urticaria And Deafness (191900); Familial Cold Urticaria	Sanger sequencing		
CSF Protein Immunoassay Panel (CJD Protein Test Panel)	1383		2.0 mL CSF; Freeze sample as soon as possible after collection. Ship frozen on dry ice.	National Microbiology Laboratory, Health Canada (Winnipeg)	Creutzfeldt-Jakob Disease (CJD)	14-3-3 protein testing of cerebrospinal fluid (CSF) (ELISA); PrPd (QuIC); Tau protein (ELISA)	Should be registerd with surveillance.	24-May-17
CSTB dodecmer repeat expansion [7084]		CSTB (601145)	Lavender Top (EDTA)	Ambry Genetics	Epilepsy, progressive myoclonic 1A (Unverricht and Lundborg) (254800)			
Currarino syndrome		MNX1 (142994)	Lavender top (EDTA)	Diagenos (www.diagenos.com)	Currarino syndrome (176450)	Sanger sequencing		
Currarino syndrome		MNX1 (142994)	Lavender top (EDTA)	Centogene AG (www.centogene.com)	Currarino syndrome (176450)	Sanger sequencing		
Cyclic AMP, Urinary Excretion [GRP]			Serum (Red Top) AND random urine resquired. Store frozen.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)				19-May-17
CYP27B1 Single Gene Test	897	CYP27B1 (609506)	Lavender Top (2 x 4 mL)	Fulgent Genetics (fulgentdiagnostics.com)	Vitamine D dependant rickets type1 (264700)	NextGen Sequencing		23-Mar-21
Cystatin C [CYSTC]	78		Serum. Store and send frozen	In-Common Laboratories		Immunonephelometry		6-Sep-18
Cystic Fibrosis: CFTR Deletion Duplication Analysis	763	CFTR (602421)	Lavender Top (EDTA)	Genome Diagnostics The Department of Paediatric Laboratory Medicine The Hospital for Sick Children (www.sickkids.ca/paediatriclabmedicinems/lab-divisions/genome-diagnostics/genome-diagnostics.html#genome)	Cystic Fibrosis (219700)	MLPA		
Cystinuria NGS Panel	807	SLC3A1 (104614); SLC7A9 (604144); PREPL (609577)	Lavender Top (2 x 4 mL)	Fulgent Genetics (fulgentdiagnostics.com)	Cystinuria (220100)	NextGen Sequencing		22-Dec-20
Cytochrome P450 2D6 (CYP2D6) Comprehensive Cascade [2D6CV]	77	CYP2D6 (124030)	Lavender Top (EDTA). Stable for 30 days at 4°C.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)				31-Oct-20
Dabigratan			Serum (Light blue top)	Quest Diagnostics				
DARS2 Full Gene Sequencing Analysis [MOL094]		DARS2 (610956)	Lavender Top (EDTA)	MNG Laboratories (www.medicalneurogenetics.com)/Dynacare	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation (611105) Alternate name: DARS2: Mitochondrial Aspartyl-tRNA Synthetase Deficiency	Sanger sequencing		14-May-20
Deletion 1p			Green Top; RT only	Cytogenetics Laboratory, Hospital for Sick Children	Chromosome1p36 deletion syndrome (607872)	microarray		
Dementia Panel [10309]	598	APP C9orf72 CHMP2B FUS GRN MAPT PSEN1 PSEN2 SQSTM1 TARDBP TREM2 TYROBP UBQLN2	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)				20-Oct-20

Appelation approuvée de l'analyse ou maladie associée [avec numéro de phénotype MIM] / Approved test name or Associated Disease [with MIM Phenotype Number]	**Code Test** / Test Code **(Dictionnaire MSSS)**	**SYMBOL HGNC DU GÈNE et NOM OMIM DU GÈNE** / HGNC Gene Symbol and OMIM Gene Number	**RÈGLES DE MANUTENTION** / Handling Conditions	**LABORATOIRE DÉSIGNÉ** / Designated laboratory	**RENSEIGNEMENTS SUR L'ANALYSE** / Test details	**MÉTHODE** / Methode	**NOTE PARTICULIÈRE** / Special comment	**Date de la révision** / Date of revision
Dexamethasone [FDXM]			Red Top	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)				31-Oct-20
DGUOK Sequence Analysis (Familial Mutation/Variant Analysis) [3076]		DGUOK (601465)	Lavender Top (EDTA)	Medical Genetics Laboratories Baylor College of Medicine (www.bcmgeneticlabs.org)	Mitochondrial DNA depletion syndrome 3 (hepatocerebral type) (251880)	Sanger sequencing		
Diabetes Insipidus Panel [KI1801]	1457	AQP2 AVP AVPR2	Lavender Top (EDTA) 2 x 4 mL	Blueprint Genetics http://blueprintgenetics.com/		NextGen Sequencing		28-Feb-20
DICER1 single gene test [S00555]	246	DICER1	Lavender Top (EDTA) 2 x 4 mL	Blueprint Genetics http://blueprintgenetics.com/		NextGen Sequencing		28-Feb-20
Dihydrotestosterone, Serum [DHT]	1459		Red Top or Gold Top. Store serum and send frozen	In-Common Laboratories		ELISA		30-Jan-20
Disaccharidase Determination, Small Bowel Biopsy	85		Intestinal biopsy, 2-5 mg wet weight. Store at -70°C until shipping and send on dry ice	Gastroenterology Clinical Lab. Nemours/Alfred I.Dupont Hospital for Children, Wilmington, DE (www.nemours.org) (https://www.nemours.org/pediatric-research/labservices/diagnostic/gastroenterology-lab.html)		lactase, maltase, sucrase, palatinase and glucoamylase		24-Aug-17
Disorders of Sex Development Sequencing Panel with CNV Detection [4509]	1395	AMH 600957 AMHR2 600956 ANOS1 300836 AR 313700 ARL6 608845 ARX 300382 ATRX 300032 BBS1 209901 BBS10 610148 BBS12 610683 BBS2 606151 BBS4 600374 BBS5 603650 BBS7 607590 BBS9 607968 CBX2 602770 CHD7 608892 CYP11A1 118485 CYP17A1 609300 CYP19A1 107910 DHH 605423 DMRT1 602424 DMRT2 604935 FGF8 600483 FGFR1 136350 FGFR2 176943 FOXL2 605597 FSHB 136530 GATA4 600576 GNRH1 152760 GNRHR 138850 HESX1 601802 HFE 613609 HS6ST1 604846 HSD17B3 605573 KISS1 603286 KISS1R 604161 LEP 164160 LEPR 601007 LHCGR 152790 LHX3 600577 LHX4 602146 MAML1D1 300120	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	3-Oxo-5 Alpha-Steroid Delta 4-Dehydrogenase Deficiency AD 264600 46,XX Sex Reversal, Type 1 AR 400045 46,XY Sex Reversal, Type 3 AD 612965 46,XY Sex Reversal, Type 5 AD 613080 46,XY Sex Reversal, Type 6 AD 613762 46,XY Sex Reversal, Type 7 AR 233420 Adrenal Insufficiency, Congenital, With 46,XY Sex Reversal, Partial Or Complete AR 613743 Androgen Resistance Syndrome XL 300068 Antley-Bixler Syndrome AR 207410 Antley-Bixler Syndrome With Genital Anomalies And Disordered Steroidogenesis AR 201750 ATR-X Syndrome XL 301040 Bardet-Biedl Syndrome 1 AR 209900 Bardet-Biedl Syndrome 10 AR 615987 Bardet-Biedl Syndrome 11 AR 615988 Bardet-Biedl Syndrome 12 AR 615989 Bardet-Biedl Syndrome 2 AR 615981 Bardet-Biedl Syndrome 3 AR 600151 Bardet-Biedl Syndrome 4 AR 615982 Bardet-Biedl Syndrome 5 AR 615983 Bardet-Biedl Syndrome 6 AR 605231 Bardet-Biedl Syndrome 7 AR 615984 Bardet-Biedl Syndrome 8 AR 615985 Bardet-Biedl Syndrome 9 AR 615986 Blepharophimosis, Ptosis, And Epicanthus Inversus AR 110100 Camptomelic Dysplasia AD 114290 Cholesterol Monooxygenase (Side-Chain Cleaving) Deficiency AR 201710 Deficiency Of Steroid 17-Alpha-Monooxygenase AD,AR 202110 Familial Gynecomastia, Due To Increased Aromatase Activity AR	Exome sequencing with CNV detection		6-Jun-19

Appelation approuvée de l'analyse ou maladie associée [avec numéro de phénotype MIM] / Approved test name or Associated Disease [with MIM Phenotype Number]	**Code Test /** Test Code **(Dictionnaire MSSS)**	**SYMBOL HGNC DU GÈNE et NOM OMIM DU GÈNE /** HGNC Gene Symbol and OMIM Gene Number	**RÈGLES DE MANUTENTION /** Handling Conditions	**LABORATOIRE DÉSIGNÉ /** Designated laboratory	**RENSEIGNEMENTS SUR L'ANALYSE /** Test details	**MÉTHODE /** Methode	**NOTE PARTICULIÈRE /** Special comment	**Date de la révision /** Date of revision
DISTAL HEREDITARY MOTOR NEUROPATHY NGS PANEL		ATP7A (300011); BSCL2 (606158); DCTN1 (601143); DNMT1 (126375); FIG4 (609390); GAN (605379); GARS (600287); HSPB1 (602195); HSPB8 (608014 0; IGHMBP2 (600502); MEGF10 (612453); REEP1 (609139); SETX (608465); SLC5A7 (608761); TRPV4 (605427)	Lavender Top (2 x 4 mL)	Fulgent Genetics (fulgentdiagnostics.com)	Distal Hereditary Motor Neuronopathy Type 2B (608634); Charcot-Marie-Tooth Disease Type 2F (606595); Distal Hereditary Motor Neuronopathy Type 2A (158590); Charcot-Marie-Tooth Disease, Type 2L (608673); Distal Hereditary Motor Neuronopathy Type 5 (600794); Charcot-Marie-Tooth Disease Type 2D (601472); Spastic Paraplegia 17 (270685); Spinocerebellar Ataxia Autosomal Recessive 1 (606002);Charcot-Marie-Tooth Disease Type 2C (606071); Spinal Muscular Atrophy, Distal, Congenital Nonprogressive (600175) ; Scapuloperoneal Spinal Muscular Atrophy (181405); NEURONOPATHY, DISTAL HEREDITARY MOTOR, TYPE VIIB; HMN7B (607641);Perry Syndrome (168605); Charcot-Marie-Tooth Disease, Type 4J (611228); Neuropathy, Hereditary Sensory, Type Ie (614116); Myopathy, Early-Onset, Areflexia, Respiratory Distress, And Dysphagia (614399); Neuronopathy, distal hereditary motor, type VIIA (158580); Giant Axonal Neuropathy (256850); Spinal Muscular Atrophy, Distal, X-Linked 3 (300489); Neuromyotonia and axonal neuropathy, autosomal recessive (137200); Neuronopathy, distal hereditary motor, type VB (614751); Spinal Muscular Atrophy With Respiratory Distress 1 (604320)	Next Generation Sequencing (NGS) and Sanger sequencing technologies		
Diuretic Screen, Urine [FDIRU]			Random urine	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	benzthiazide, bumetanide, chlorothiazide, chlorthalidone, furosemide, hydrochlorothiazide, hydroflumethiazide, and metolazone			
DOPA-RESPONSIVE DYSTONIA VIA THE GCH1 GENE [161]	1096	GCA1 (600225)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Dystonia 5, Dopa-Responsive Type (128230)	Sanger sequencing		
Doxycycline [94093]	1465		Serum (2 mL). Store frozen.	Quest Diagnostics				21-Feb-20
Drug Dependent Platelet Antibody [9000]			Gold SST	Blood Center of Wisconsin				
Drug Screen, Prescription/OTC, Urine [PDSU]			Random urine	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	Trifluoperazine (stelazine)			20-Oct-20
Duchenne and Becker Muscular Dystrophy (310200 and 300376)	617	DMD (300377)	Lavender Top (EDTA)	Molecular Genetics Laboratory, Hospital for Sick Children (www.sickkids.ca/PaediatricLaboratoryMedicine/Laboratories-Services/Molecular-Genetics-Laboratory/index.html)		1. Sanger sequencing 2. gene dosage		4-May-17
Dystonia Dyskinesia NGS Panel	1096	ANO3, ATP1A3, CACNA1B, CIZ1, COL6A3, DRD2, DRD5, GCH1, GNAL, HPCA, KCTD17, PNKD, PRKN, PRKRA, PRRT2, SCP2, SGCE, SLC2A1, SLC6A3, SPR, TAF1, TH, THAP1, TOR1A, TUBB4A	Lavender Top (2 x 4 mL)	Fulgent Genetics (fulgentdiagnostics.com)	Alternating hemiplegia of childhood 2 (614820); CAPOS syndrome (601338); Dystonia-12 (128235); Dystonia, myoclonic (159900); Dystonia, primary cervical; Dystonia 16 (612067); Convulsions, familial infantile, with paroxysmal choreoathetosis (602066); Episodic kinesigenic dyskinesia 1 (128200); Seizures, benign familial infantile, 2 (605751); Leukoencephalopathy with dystonia and motor neuropathy (613724); Dystonia-11, myoclonic (159900); Parkinsonism-dystonia, infantile (613315); Dystonia, dopa-responsive, due to sepiapterin reductase deficiency (612716); Dystonia-Parkinsonism, X-linked (314250); Dystonia 6, torsion (602629); Dystonia-1, torsion (128100)	Next-Generation sequencing	See Med Neurogenetics Panel	21-Mar-19
Dystonia/Parkinson Panel [T402]	1096		Lavender Top (EDTA)	GeneDx (www.genedx.com)		Next-Generation sequencing		3-Oct-19

Appelation approuvée de l'analyse ou maladie associée [avec numéro de phénotype MIM] / Approved test name or Associated Disease [with MIM Phenotype Number]	**Code Test** / Test Code **(Dictionnaire MSSS)**	**SYMBOL HGNC DU GÈNE et NOM OMIM DU GÈNE** / HGNC Gene Symbol and OMIM Gene Number	**RÈGLES DE MANUTENTION** / Handling Conditions	**LABORATOIRE DÉSIGNÉ** / Designated laboratory	**RENSEIGNEMENTS SUR L'ANALYSE** / Test details	**MÉTHODE** / Methode	**NOTE PARTICULIÈRE** / Special comment	**Date de la révision** / Date of revision
Early-Onset Epileptic Encephalopathy Panel		ACY1, ADGRV1, ADSL, ALDH7A1, ALG13, ARFGEF2, ARHGEF15, ARHGEF9, ARX, ATP1A2, ATP6AP2, AUH, BCKDK, CACNA1A, CACNA2D2, CACNB4, CDKL5, CHD2, CHRNA2, CHRNA4, CHRNA7, CHRNB2, CLCN4, CLN3, CLN5, CLN6, CLN8, CNTNAP2, CPA6, CSTB, CTSD, CTSF, DEPDC5, DNAJC5, DNM1, DYNC1H1, DYRK1A, EEF1A2, EFHC1, EPM2A, FARS2, FOLR1, FOXG1, GABRA1, GABRB2, GABRB3, GABRD, GABRG2, GAMT, GATM, GNAO1, GOSR2, GRIN1, GRIN2A, GRIN2B, HCN1, HCN2, HNRNPU, IQSEC2, KANSL1, KCNB1, KCNH2, KCNH5, KCNJ10, KCNQ2, KCNQ3, KCNT1, KCTD7, KPNA7, LGI1, LIAS, MAGI2, MBD5, MECP2, MEF2C, MFSD8, MTOR, NEDD4L, NHLRC1, NR2F1, NRXN1, PCDH19, PIGA, PIGO, PIGV, PLCB1, PNKP, PNPO, POLG, PPT1, PRICKLE1, PRRT2, QARS, RBFOX1, RBFOX3, ROGDI, SCARB2, SCN1A, SCN1B, SCN2A, SCN3A, SCN5A, SCN8A, SCN9A, SIK1, SLC12A5, SLC13A5, SLC19A3, SLC25A12, SLC25A22, SLC2A1, SLC35A2, SLC6A8, SLC9A6, SPTAN1, SRPX2, ST3GAL3, ST3GAL5, STX1B, STXBP1, SYN1, SYNGAP1, SZT2, TBC1D24, TCF4, TNK2, TPP1, TSC1, TSC2, UBE3A, WDR45, WWOX, ZEB2	Lavender Top (2 x 4 mL)	Fulgent Genetics (fulgentdiagnostics.com)				
Ehlers-Danlos syndrome NGS panel - Dominant & Recessive [5069]	575	ADAMTS2, ATP7A, CHST14, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2, DSE, FKBP14, FLNA, PLOD1, SLC39A13	Lavender Top (EDTA)	Connective Tissue Gene Tests (www.ctgt.net)		NextGen Sequencing; Del Dupl		3-Nov-20
Encephalopathy, Autoimmune Evaluation [ENCES CSF]			CSF minimum 3 cc	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	IFA, RIA, CBA, WB			
Encephalopathy, Autoimmune Evaluation [ENCES serum]			Red top, 7-10 cc of blood	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	IFA, RIA, CBA, WB			
Epidermolysis bullosa (226600)		COL7A1 (120120)	Lavender Top (EDTA)	Centogene AG (www.centogene.com)		1. single exon testing 2. full gene sequencing 3. deletion/duplication testing		
Essential Epilepsey Panel	356	ACY1, ADSL, ALDH7A1, ALG13, ARHGEF9, ARX, CDKL5, CHD2, CHRNA2, CHRNA4, CHRNB2, CNTNAP2, DEPDC5, DNM1, FOLR1, FOXG1, GABRA1, GABRB3, GABRG2, GAMT, GNAO1, GRIN2A, GRIN2B, HCN1, HNRNPU, KCNA2, KCNB1, KCNQ2, KCNQ3, KCNT1, LGI1, MAPK10, MBD5, MECP2, MEF2C, NRXN1, PCDH19, PLCB1, PNKP, PNPO, POLG, PRRT2, RNASEH2A, RNASEH2B, RNASEH2C, ROGDI, SAMHD1, SCN1A, SCN1B, SCN2A, SCN8A, SCN9A, SIK1, SLC19A3, SLC25A22, SLC2A1, SLC35A2, SLC9A6, SPTAN1, ST3GAL3, ST3GAL5, STXBP1, SYNGAP1, SZT2, TBC1D24, TCF4, TREX1, TSC1, TSC2, UBE3A, ZEB2	Lavender Top (2 x 4 mL)	Fulgent Genetics (fulgentdiagnostics.com)				
Expanded Neuromuscular Disorders: Sequencing and Deletion/Duplication Panel[MM360]		ACTA1, AMPD1, AMPD3, ANO5, BAG3, BIN1, BSCL2, CAPN3, CAV3, CFL2, CHAT, CHRNA1, CHRNB1, CHRND, CHRNE, CHRNG, COL6A1, COL6A2, COL6A3, COLQ, CRYAB, DAG1, DES, DMD, DNM2, DOK7, DYSF, EMD, FHL1, FKRP, FKTN, FLNC, GAA, GLE1, GNE, IGHMBP2, ISPD, ITGA7, LAMA2, LARGE, LDB3, LMNA, MTM1, MTMR14, MUSK, MYH2, MYH7, MYOT, NEB, PABPN1, PLEC, PLEKHG5, PMM2, POMGNT1, POMT1, POMT2, PTRF, PYGM, RAPSN, RYR1, RYR2, SCN4A, SEPN1, SGCA, SGCB, SGCD, SGCE, SGCG, SIL1, SYNE1, SYNE2, TCAP, TNNI2, TNNT1, TPM2, TPM3, TRIM32, TTN, VRK1	Lavender Top (EDTA)	EGL Genetics Laboratory 2460 Mountain Industrial, Tucker GA 30084 (geneticslab.emory.edu)	Muscular dystrophies, Congenital myopathies, Congenital myasthenic syndrome, Nemaline myopathy, Limb girdle muscular dystrophy, Emery-Dreifuss muscular dystrophy, Congenital muscular dystrophy, Cardiomyopathies, Myoadenylate deaminase deficiency, Erythrocyte AMP deaminase deficiency, Myofibrillar myopathy, Duchenne/Becker muscular dystrophy, Congenital disorder of glycosylation type 1a, Malignant hyperthermia susceptibility, Myoclonus dystonia, Marinesco-Sjogren syndrome, Distal arthrogryposis, NMD, Neuromuscular disorders	Next-Generation sequencing and gene-targeted CGH array (deletion/duplication analysis included in this panel: BIN1, BSCL2, CFL2, CHAT, CHRNA1, CHRNB1, CHRND, CHRNE, CHRNG, COLQ, DNM2, DOK7, FHL1, FLNC, GLE1, GNE, IGHMBP2, ISPD, MTM1, MTMR14, MUSK, MYH2, PLEKHG5, PTRF, PYGM, RAPSN, SCN4A, SYNE1, SYNE2, and VRK1)	Fomerly Emory	
F12 Select Exons Sequencing [388]	1832	F12 (610619)	Lavender Top (2 x 4 mL)	GeneDx (www.genedx.com)	Hereditay Angioedema (HAE) Type III (610618)			14-Aug-17
Facioscapulohumeral Muscular Dystrophy	984	DUX4 (D4Z4) (606009)	Lavender Top (EDTA)	Childern's Hospital of Eastern Ontario	FACIOSCAPULOHUMERAL MUSCULAR DYSTROPHY 1 (158900)	deletion and haplotype		
Factor B			0.5 mL red top serum- spun, separated, frozen within 2 hrs of collection; ship on dry ice	Cincinnati Children's Hospital Medical Center 3333 Burnet Avenue NRB 1042 Cincinnati, OH 45229 (www.cincinnatichildrens.org)				15-Nov-17
Factor H autoantibodies	64		2ml frozen serum.	Cincinnati Children's Hospital Medical Center 3333 Burnet Avenue NRB 1042 Cincinnati, OH 45229 (www.cincinnatichildrens.org)				28-Jun-19

Appelation approuvée de l'analyse ou maladie associée [avec numéro de phénotype MIM] / Approved test name or Associated Disease [with MIM Phenotype Number]	**Code Test** / Test Code **(Dictionnaire MSSS)**	**SYMBOL HGNC DU GÈNE et NOM OMIM DU GÈNE** / HGNC Gene Symbol and OMIM Gene Number	**RÈGLES DE MANUTENTION** / Handling Conditions	**LABORATOIRE DÉSIGNÉ** / Designated laboratory	**RENSEIGNEMENTS SUR L'ANALYSE** / Test details	**MÉTHODE** / Methode	**NOTE PARTICULIÈRE** / Special comment	**Date de la révision** / Date of revision
Factor I			2ml frozen serum.	Cincinnati Children's Hospital Medical Center 3333 Burnet Avenue NRB 1042 Cincinnati, OH 45229 (www.cincinnatichildrens.org)		complement-mediated renal diseases FI		15-Nov-17
Familial Hemiplegic Migraine (NGS Panel and Copy Number Analysis + mtDNA) [NGS429]	581	ATP1A2, ATP1A3, BAP1, CACNA1A, PRRT2, SCN1A	Lavender Top	MNG Laboratories (www.medicalneurogenetics.com)/Dynacare		Next-Generation sequencing	Replaced by Blueprint panel	21-Aug-20
Familial Hemophagocytic Lymphohistiocytosis (FHL), Autosomal Recessive; type 2 (603553), type 3 (608898), type 4 (603552) and type 5 (613101)		PRF1 (170280); UNC13D (MUNC13-4) (608897); STXBP2 (601717); RAB27A (603868); STX11 (605014)	Lavender Top (EDTA)	Cinncinatti Children's Hospital (Division of Human Genetics Diagnostic Laboratories)	PRF1 and STX11 also at: Hospital for Sick Children		Available at Fulgent for 1450 $ or 1950$ with del/du	
Familial Limb Girdle Myasthenia Syndrome via DOK7 Gene Sequencing with CNV Detection [7629]		DOK7 (610285)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Myasthenia, Limb-Girdle, Familial (254300)		Available at MNG	3-May-19
Farmer's Lung IgG Antibodies, Serum [FLGAB]	1465		Serum (2 mL). Store frozen.	In-Common Laboratories	M. faeni IgG Ab; T. vulgaris IgG Ab	FEIA		10-Mar-20
Fat, Feces [FATF]	1465		For a random collection, a minimum of 5 g (do not send entire collection) is required. Store frozen.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)		Nuclear Magnetic Resonance (NMR) Spectroscopy		10-Mar-20
Fatty Acid Oxidation Deficiency NGS Panel	528	ACAD9, ACADL, ACADM, ACADS, ACADVL, CPT1A, CPT1B, CPT2, ETFA, ETFB, ETFDH, GLUD1, HADH, HADHA, HADHB, HMGCL, HMGCS2, HSD17B10, LPIN1, SLC22A5, SLC25A20, TAZ	Lavender Top (2 x 4 mL)	Fulgent Genetics (fulgentdiagnostics.com)	Includes: Fatty liver, acute, of pregnancy (609016); HELLP syndrome, maternal, of pregnancy (609016)	NextGen Sequencing	Replaces: UofA targeted mutation testing	
Fatty Acid Oxidation Syndrome Panel [ME1701]	1320		Lavender Top (2 x 4 mL)	Blueprint Genetics http://blueprintgenetics.com/		Next-Generation sequencing		29-Feb-20
FH Autoantibody Testing	64		2ml frozen serum.	Cincinnati Children's Hospital Medical Center 3333 Burnet Avenue NRB 1042 Cincinnati, OH 45229 (www.cincinnatichildrens.org)		complement-mediated renal diseases FI		15-Nov-17
FH Autoantibody Testing	64		Panel requires at least 2ml frozen serum.	Molecular Otolaryngology & Renal Research Laboratory, U. of Iowa Carver College of Medicine (www.medicine.uiowa.edu/morl)	Hemolytic uremic syndrome, atypical, susceptibility to, 5 (612925)	ELSIA and other assays		31-Oct-17
Fibrodysplasia ossificans progressiva (135100)		ACVR1 (102576)	Lavender Top (EDTA)	University of Pennsylvania School of Medicine	Fibrodysplasia ossificans progressiva (135100)	ACVR1 point mutation C617 G-A		
Fluphenazine (Prolixin), Serum [PROLX]	929		Serum Draw blood in a plain red-top tube(s), serum gel tube is not acceptable. Spin down and send 3 mL of serum refrigerated in a plastic vial.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	Modecate	Liquid Chromatography/Tandem Mass Spectrometry (LC/MS/MS)		26-Jan-20
Focused Pharmacogenomics Panel [PGXFP]	1582	*CYP1A2, CYP2C9, CYP2C19, CYP2D6, CYP3A4, CYP3A5, SLCO1B1, VKORC1, CYP4F2* , and rs12777823	Lavender Top (EDTA); Saliva; DNA	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)		TaqMan	Replaces ONEOME (oneome.com)	26-Oct-17
FSHD - Detection of Abnormal Alleles with Interpretation (FSHD1 and FSHD2)	102	DUX4 (D4Z4) (606009); SMCHD1 (614982)	Lavender Top (EDTA)	University of Iowa Diagnostic Laboratories (http://www.healthcare.uiowa.edu/path_handbook/rhandbook/test127.html)	FACIOSCAPULOHUMERAL MUSCULAR DYSTROPHY 1 (158900); Facioscapulohumeral Muscular Dystrophy 2 (158901)	Southern blot, 4qA.4qVhaplotyping; methylation; SMCHD1 sequencing		13-May-19
Fumarase deficiency (606812) [713]	1034	FH (136850)	Lavender Top (EDTA)	GeneDx (www.genedx.com)	fumarate hydratase; Includes: Hereditary Leiomyomatosis and Renal cell Cancer (HLRCC) (150800)	Sanger sequencing and del/dupl		9-Dec-19
Fungitell, Serum [FUNGS]	489		Collect 3-5 mL blood in a serum separator gel tube (SST). Centrifuge specimen within 2 hours. Ship serum gel tube frozen	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)		Replaced by: 1,3-Beta-D-Glucan (Fungitell), Serum		20-Jan-20
Gabapentin [GABA]	107		Red Top. Store and send frozen.	In-Common Laboratories		LC/UV		6-Sep-18
Galactose-alpha-1,3-galactose (Alpha-Gal) IgE [FGA13]	1761		Collect RED or GOLD SST. Store at 4°C for 28 days or at -20°C (1 year).	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	Phadia ImmunoCAP	Phadia ImmunoCAP	Available at ICL $120	19-Jun-19
GARS Full Gene Sequencing Analysis [MOL167]		GARS (600287)	Lavender Top (EDTA)	MNG Laboratories (www.medicalneurogenetics.com)/Dynacare	Charcot-Marie-Tooth disease, type 2D (601427); Neuropathy, distal hereditary motor, type V (600794)	Sanger sequencing		9-May-20
Gaucher Disease (recurrent mutations)		GBA (606463)	Lavender Top (EDTA)	Molecular Genetics Laboratory, Hospital for Sick Children (www.sickkids.ca/PaediatricLaboratoryMedicine/Laboratories-Services/Molecular-Genetics-Laboratory/index.html)	Gaucher's Disease, Type 1 (230800); Gaucher Disease, Perinatal Lethal (608013); Subacute Neuronopathic Gaucher's Disease (230900); Gaucher Disease, Type IIIc (231005)	Direct mutation analysis (9 mutations);AKJ 90% sensitivity; others 50-60%		
GAUCHER DISEASE VIA THE GBA GENE [479]	464	GBA (606463)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Gaucher's Disease, Type 1 (230800); Gaucher Disease, Perinatal Lethal (608013); Subacute Neuronopathic Gaucher's Disease (230900); Gaucher Disease, Type IIIc (231005)	Sanger sequencing		28-Jan-20
Gene dosage for FGFR2, FGFR3 & TWIST		FGFR2 (176943); FGFR3 (134934); TWIST1 (601622)	Lavender Top (EDTA)	Molecular Genetics Laboratory, Hospital for Sick Children (www.sickkids.ca/PaediatricLaboratoryMedicine/Laboratories-Services/Molecular-Genetics-Laboratory/index.html)	Craniosynostosis	del/dupl		
Genetic Eye Disease Panel for **Strabismus** (Gedi-S)		ROBO3, PHOX2A, HOXA1, SALL4, CHN1, TUBB3, KIF21A, HOXB1	Lavender Top (EDTA) 2-5 cc	Ocular genomics (https://oculargenomics.meei.harvard.edu/index.php/gdt)	Strabismus	NextGen Sequencing		

Appelation approuvée de l'analyse ou maladie associée [avec numéro de phénotype MIM] / Approved test name or Associated Disease [with MIM Phenotype Number]	**Code Test** / Test Code **(Dictionnaire MSSS)**	**SYMBOL HGNC DU GÈNE et NOM OMIM DU GÈNE** / HGNC Gene Symbol and OMIM Gene Number	**RÈGLES DE MANUTENTION** / Handling Conditions	**LABORATOIRE DÉSIGNÉ** / Designated laboratory	**RENSEIGNEMENTS SUR L'ANALYSE** / Test details	**MÉTHODE** / Methode	**NOTE PARTICULIÈRE** / Special comment	**Date de la révision** / Date of revision
Genetic Renal Panel	1285	CFH (134370), CFI (217030), MCP (120920), CFB (138470), CFHR5 (608593), C3 (120700), THBD (188040), ADAMTS13 (604134), DGKE (601440), PLG (173350); CFHR3-CFHR1 (605336/134371)	Lavender Top (EDTA)	Molecular Otolaryngology & Renal Research Laboratory, U. of Iowa Carver College of Medicine (www.medicine.uiowa.edu/morl)	thrombotic microangiopathies: Hemolytic Uremic Syndrome, atypical Hemolytic Uremic Syndrome and Thrombotic Thrombocytopenic Purpura	Next-Generation sequencing; MLPA		23-Mar-20
GH-RH			Serum or EDTA plasma	InterScience Institute				
GLA gene dosage	469	GLA (300644)	Lavender top (EDTA); Store at RT or 4°C for up to 48 h after drawing. At 4°C for >48 h.	Molecular Genetics Laboratory, Hospital for Sick Children (www.sickkids.ca/PaediatricLaboratoryMedicine/Laboratories-Services/Molecular-Genetics-Laboratory/index.html)	Fabry Disease (301500)	MLPA		
GLA gene sequencing	469	GLA (300644)	Lavender top (EDTA); Store at RT or 4°C for up to 48 h after drawing. At 4°C for >48 h.	Molecular Genetics Laboratory, Hospital for Sick Children (www.sickkids.ca/PaediatricLaboratoryMedicine/Laboratories-Services/Molecular-Genetics-Laboratory/index.html)	Fabry Disease (301500)	Sanger sequencing		
Glucagon, Plasma [GLP]	110		Collection Container/Tube: Lavender top (EDTA) Submission Container/Tube: Plastic vial Specimen Volume: 2 mL Collection Instructions: 1. Fasting. 2. Prechill tube at 4 degrees C before drawing the specimen. 3. Draw the prechilled tube, and process as follows: a. After drawing specimen, chill tube in wet ice for 10 minutes. b. Centrifuge in a refrigerated centrifuge or in chilled centrifuge cup. c. Immediately after centrifugation, remove plasma, place in a plastic transport vial (Supply T465), and freeze.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)		RIA		17-Mar-20
GLUCOSE TRANSPORTER TYPE 1 DEFICIENCY SYNDROME (06777)		SLC2A1 (138140)	Lavender Top (EDTA)	BC Children's Hospital & BC Women's Hospital, Canada (http://www.genebc.ca)		Sanger sequencing & reflex MLPA		19-Oct-17
GLUCOSE-6-PHOSPHATE DEHYDROGENASE DEFICIENCY VIA THE G6PD GENE		G6PD (305900)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Hemolytic anemia due to G6PD deficiency (300908)	Sanger sequencing		
GLUT1 Deficiency Syndrome (SLC2A1 Single Gene Test)	262	SLC2A1 (138140)	Lavender Top (EDTA) [2 tubes]	Fulgent Genetics (fulgentdiagnostics.com)		NextGen Sequencing		28-Feb-20
Glutamate Receptor R1							NOT AVAILABLE: Wash U, Mayo, Oxford	21-Apr-17
Glycogen Storage Disease and Disorders of Glucose Metabolism Sequencing Panel with CNV Detection [10385]	476	AGL 610860 ALDOA 103850 ALDOB 612724 ENO3 131370 G6PC 613742 GAA 606800 GBE1 607839 GYG1 603942 GYS1 138570 GYS2 138571 LAMP2 309060 LDHA 150000 PC 608786 PCK1 614168 PCK2 614095 PFKM 610681 PGAM2 612931 PGM1 171900 PHKA1 311870 PHKA2 300798 PHKB 172490 PHKG2 172471 PRKAG2 602743 PYGL 613741 PYGM 608455 SLC16A1 600682 SLC2A2 138160	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Glycogen Storage Disease Type III (232400); Glycogen Storage Disease Type IA (232200); Glycogen Storage Disease Type II (232300); Glycogen Storage Disease Type IV (232500); Glycogen Storage Disease Type 0 (240600); Glycogen Storage Disease Type VII (232800); Glycogen Storage Disease Type IXd (300559); Glycogen Storage Disease Type IXa1 (306000); Glycogen Storage Disease Type IX (261750); Glycogen Storage Disease Type IXc (613027); Glycogen Storage Disease Type VI (232700); Glycogen Storage Disease Type V (232600); Glycogen Storage Disease Type Ib (232200); Glycogen Storage Disease Type Ic (232240); Fanconi-Bickel Syndrome (227810)	Next Generation Sequencing (NGS) and Sanger sequencing technologies		15-Aug-19
GLYCOGEN STORAGE DISEASE TYPE III VIA THE AGL GENE [224]	476	AGL (610680)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Glycogen Storage Disease Type III (GSDIII) (232400)	Sanger sequencing		
GLYCOGEN STORAGE DISEASE TYPE IV VIA THE GBE1 GENE [225]	476	GBE1 (607893)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Glycogen Storage Disease Type IV (232500)	Sanger sequencing		
Goose Feather (e70) IgE, Serum	1419		Gold SST	Check if available at CHUM				7-Oct-19
Granulocyte Ab, Serum [LAGGT]	1836		Gold SST or Red Top	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)				28-Feb-19
GRHPR Gene, Full Gene Analysis [GRHMS]		GRHPR (604296)	Lavender top (EDTA)	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	Hyperoxaluria, primary, type II (296000)	Sanger sequencing and. MLPA		
Growth Hormone Releasing Hormone (GH-RH)			3 ml serum (Red or Gold Top) or EDTA plasma should be collected and separated as soon as possible. Freeze the plasma immediately after separation. Minimum specimen size is 1 ml.	InterScience Institute				
Haloperidol			Red Top or Li heparin (light Green Top)	In-Common Laboratories		LC-MS		20-Sep-19

Appelation approuvée de l'analyse ou maladie associée [avec numéro de phénotype MIM] / Approved test name or Associated Disease [with MIM Phenotype Number]	**Code Test** / Test Code **(Dictionnaire MSSS)**	**SYMBOL HGNC DU GÈNE et NOM OMIM DU GÈNE** / HGNC Gene Symbol and OMIM Gene Number	**RÈGLES DE MANUTENTION /** Handling Conditions	**LABORATOIRE DÉSIGNÉ** / Designated laboratory	**RENSEIGNEMENTS SUR L'ANALYSE** / Test details	**MÉTHODE /** Methode	**NOTE PARTICULIÈRE /** Special comment	**Date de la révision /** Date of revision
Hearing Loss: Branchio-Oto-Renal (BOR) Syndrome (115630)		EYA1 (601653)	Lavender Top (EDTA)	Molecular Genetics Laboratory, Hospital for Sick Children (www.sickkids.ca/PaediatricLaboratoryMedicine/Laboratories-Services/Molecular-Genetics-Laboratory/index.html)		1. EYA1 gene sequencing 2. EYA1 gene dossage		
Hearing Loss: Non-Syndromic (Connexin 26 & 30)		GJB2 (121011); GJB6 (604418)	Lavender Top (EDTA)	Molecular Genetics Laboratory, Hospital for Sick Children (www.sickkids.ca/PaediatricLaboratoryMedicine/Laboratories-Services/Molecular-Genetics-Laboratory/index.html)		1. GJB2 sequencing and GJB6 deletion 2. GJB2 sequencing only		
Heparin Induced Thrombocytopenia Testing [HIT]	116		1) RED TOP. 4 mL. Draw blood into red-top vacutainer and allow to clot. Centrifuge and transfer serum to two plastic tube. 2) Freeze promptly. Ship frozen	Platelet Immunology Laboratory, McMaster University		HIT Confirmatory Test (Serotonin Release Assay; SRA) $300 and HIT Screen test (Anti-PF4/heparin EIA $130		31-Oct-19
HEREDITARY ANGIOEDEMA VIA THE SERPING1 /C1NH GENE [8425]	1505	SERPING1 (606860)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Hereditary Angioneurotic Edema (106100)	Next-Generation sequencing		24-Oct-17
HEREDITARY DIFFUSE GASTRIC CANCER (137215) VIA THE CDH1 GENE [798]		CDH1 (192090)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)				24-Oct-17
Hereditary Erythrocytosis Mutations [HEMP]		EPOR (133171), VHL (608537), EGLN1(PHD2) (606425), EPAS1 (HIF2A) (603349)	Lavender Top (EDTA)	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	Erythrocytosis, familial, 1 (133100); Erythrocytosis, familial, 2 (263400); Erythrocytosis, familial, 3 (609820); Erythrocytosis, familial, 4 (611783)	Sanger sequencing. Panel: EPOR: 133171, EGLN1: 606425, EPAS1: 603349, As reflex VHL: 608537		20-Oct-20
HEREDITARY HEMOCHROMATOSIS PANEL [10243]	1655	FTH1 (134770); FTL (134790); HAMP (606464); HFE (613609); HJV (608374); SLC40A1 (60463); TFR2 (604720)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Hemochromatosis Type 5 (615517); Hyperferritinemia Cataract Syndrome (600886); Hemochromatosis Type 4 (606069); Hemochromatosis Type 2 (602390); Hemochromatosis Type 2B (613313); Hemochromatosis Type 3 (604250); Hemochromatosis Type 1 (235200)	Next-Generation sequencing		5-Jun-20
Hereditary Hemorrhagic Telangiectasia	242	ENG (131195), ACVRL1 (ALK1) (601284), SMAD4 (600993)	Lavender Top (EDTA)	Molecular Genetics Laboratory, Hospital for Sick Children (www.sickkids.ca/PaediatricLaboratoryMedicine/Laboratories-Services/Molecular-Genetics-Laboratory/index.html)	Telangiectasia, hereditary hemorrhagic, type 1 (187300) (OSLER-RENDU-WEBER DISEASE);Telangiectasia, hereditary hemorrhagic, type 2 (600376); Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome (175050)	Sanger sequencing and MLPA		
Hereditary Hemorrhagic Telangiectasia (HHT), type 1 (187300) and type 2 (600376)	242	ENG (131195); ACVRL1(ALK1) (601284)	Lavender Top (EDTA)	Molecular Genetics Laboratory, Hospital for Sick Children (www.sickkids.ca/PaediatricLaboratoryMedicine/Laboratories-Services/Molecular-Genetics-Laboratory/index.html)		1. ACVRL1 gene sequencing 2.ENG gene sequencing 3. SMAD4 gene sequencing 4. ACVRL1 and ENG gene dosage		
Hereditary Melanoma and Skin Cancer Panel [ON0501]	243		Lavender Top (EDTA)	Blueprint Genetics http://blueprintgenetics.com/		NextGen Sequencing		15-Dec-20
Hereditary Neuropathy Sequencing & Del/Dup Panel [737]	656	AARS, ATL1, ATP7A, BSCL2, DNAJB2, DNM2, DNMT1, DYNC1H1, EGR2, FAM134B, FGD4, FIG4, GAN, GARS, GDAP1, GJB1, GLA, HINT1, HSPB1, HSPB8, IGHMBP2, IKBKAP, INF2, KIF1A, KIF5A, LITAF, LMNA, LRSAM1, MFN2, MPZ, MTMR2, NDRG1, NEFL, NGF, NTRK1, PLEKHG5, PMP22, PRPS1, PRX, RAB7A, REEP1 (C2ORF23), SBF2, SCN9A, SH3TC2, SLC12A6, SLC52A2, SPTLC1, SPTLC2, TFG, TRPV4, TTR, WNK1 (exon 10 only), YARS	Lavender Top (EDTA)	GeneDx		NGS		9-Dec-19
Hereditary Spastic Paraplegia: Autosomal Dominant [HSP-Panel 1]	657	ALDH18A1 ATL1 BSCL2 C10orf2 HSPD1 KIAA0196 KIF5A NIPA1 POLG POLG2 REEP1 SPAST RTN2 SLC33A1 SETX	Lavender Top (EDTA)	Molecular Genetics Laboratory, Hospital for Sick Children (www.sickkids.ca/PaediatricLaboratoryMedicine/Laboratories-Services/Molecular-Genetics-Laboratory/index.html)		NextGen Sequencing	Price of individual genes: $500 (seq) + 500$ (del/dupl)	21-Aug-19

Appelation approuvée de l'analyse ou maladie associée [avec numéro de phénotype MIM] / Approved test name or Associated Disease [with MIM Phenotype Number]	**Code Test** / Test Code **(Dictionnaire MSSS)**	**SYMBOL HGNC DU GÈNE et NOM OMIM DU GÈNE** / HGNC Gene Symbol and OMIM Gene Number	**RÈGLES DE MANUTENTION** / Handling Conditions	**LABORATOIRE DÉSIGNÉ** / Designated laboratory	**RENSEIGNEMENTS SUR L'ANALYSE** / Test details	**MÉTHODE** / Methode	**NOTE PARTICULIÈRE** / Special comment	**Date de la révision** / Date of revision
Hereditary Spastic Paraplegia: Autosomal Recessive [HSP-Panel 2]	657	ALDH18A1 ALS2 AP4B1 AP4E1 AP4M1 AP4S1 AP5Z1 C10orf2 C12orf65 C19orf12 CYP2U1 CYP7B1 DDHD1 DDHD2 ENTPD1 ERLIN1 ERLIN2 FA2H GBA2 GJC2 KIF1A KIF1C NT5C2 PGAP1 PNPLA6 POLG SACS SPG11 SPG20 SPG21 SPG7 TECPR2 VPS37A	Lavender Top (EDTA)	Molecular Genetics Laboratory, Hospital for Sick Children (www.sickkids.ca/PaediatricLaboratoryMedicine/Laboratories-Services/Molecular-Genetics-Laboratory/index.html)		NextGen Sequencing	Price of individual genes: $500 (seq) + 500$ (del/dupl)	21-Aug-19
Hereditary Spastic Paraplegia: Comprehensive Testing [HSP-COMP]	657	ALDH18A1 ATL1 BSCL2 C10orf2 HSPD1 KIAA0196 KIF5A NIPA1 POLG POLG2 REEP1 SPAST RTN2 SLC33A1 SETX ALS2 AP4B1 AP4E1 AP4M1 AP4S1 AP5Z1 C10orf2 C12orf65 C19orf12 CYP2U1 CYP7B1 DDHD1 DDHD2 ENTPD1 ERLIN1 ERLIN2 FA2H GBA2 GJC2 KIF1A KIF1C NT5C2 PGAP1 PNPLA6 SACS SPG11 SPG20 SPG21 SPG7	Lavender Top (EDTA)	Molecular Genetics Laboratory, Hospital for Sick Children (www.sickkids.ca/PaediatricLaboratoryMedicine/Laboratories-Services/Molecular-Genetics-Laboratory/index.html)		NextGen Sequencing	Price of individual genes: $500 (seq) + 500$ (del/dupl)	21-Aug-19
Hereditary Spastic Paraplegia: Deletion & Duplication Analysis [HSP-DOSAGE]	657		Lavender Top (EDTA)	Molecular Genetics Laboratory, Hospital for Sick Children (www.sickkids.ca/PaediatricLaboratoryMedicine/Laboratories-Services/Molecular-Genetics-Laboratory/index.html)		Deletion & Duplication analysis by exon targeted microarray		21-Aug-19
Heterotaxy and Situs Inversus NGS Panel	385	CVR2B, CCDC39, CCDC40, CFC1, DNAAF1, DNAAF2, DNAAF3, DNAH11, DNAH5, DNAI1, DNAI2, DNAL1, FOXH1, GDF1, INVS, LEFTY2, NKX2-5, NME8, NODAL, ZIC3	Lavender Top (2 x 4 mL)	Fulgent Genetics (fulgentdiagnostics.com)		NextGen Sequencing		
Histamine, 24-Hour Urine [FH24U]	117		4 mL urine from a 24-hour collection containing 10 mL 6N HCl; Altenate: No presevative. Specimen Stability: Room temperature: 48 hours, Refrigerated: 14 days, Frozen: 14 days. Patient should refrain from taking allergy causing drugs, antihistamines, oral corticosteroids, and substances which block H2 receptors for at least 24 hours prior to specimen collection.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	carcinoid	Immunoassay		28-Feb-19

Appelation approuvée de l'analyse ou maladie associée [avec numéro de phénotype MIM] / Approved test name or Associated Disease [with MIM Phenotype Number]	**Code Test** / Test Code **(Dictionnaire MSSS)**	**SYMBOL HGNC DU GÈNE et NOM OMIM DU GÈNE** / HGNC Gene Symbol and OMIM Gene Number	**RÈGLES DE MANUTENTION /** Handling Conditions	**LABORATOIRE DÉSIGNÉ** / Designated laboratory	**RENSEIGNEMENTS SUR L'ANALYSE** / Test details	**MÉTHODE /** Methode	**NOTE PARTICULIÈRE /** Special comment	**Date de la révision /** Date of revision
Histamine, Plasma [FHSPL]	117		Draw 3 mL blood in a lavender-top (EDTA) tube(s). Cool immediately on ice. Centrifuge at 1500 rpm for 10 minutes at 4 degrees C. The centrifugation should be performed within 20 minutes of collection. Carefully remove 1 mL of EDTA plasma from the upper part of the tube. Freeze and send frozen in a plastic vial.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)		EIA		28-Feb-19
Histoplasma Sereology			Gold SST	Alberta Provincial Laboratory				
HLA-B 1502 Genotype, Carbamazepine Hypersensitivity, Blood [HLA15]		HLA-B (142830)	Lavender Top (EDTA)	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	Stevens-Johnson syndrome (608579) and toxic epidermal necrolysis (608579)			
Homocystinuria [1563]		CBS (613381)	Lavender top (EDTA)	Connective Tissue Gene Tests (www.ctgt.net)	Homocystinuria, B6-responsive and nonresponsive types (236200)	1.Sanger sequencing 2.HD array		
HTRA1 DNA Sequencing Test (CARASIL) [442]		HTRA1 (602194)	Lavender Top (EDTA)	Athena Diagnostics (www.athenadiagnostics.com)	CARASIL (600142)	Sanger sequencing		
Hydroxychloroquine, Serum [HCQ]	1689		Red Top ONLY. Store and send serum at 4°C or frozen (stable 28 d).	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)		LC-MS/MS		22-Sep-19
Hyper IgE Syndrome Panel [1969]	1258	DOCK8 (614443), SPINK5 (605010), STAT3 (102582), TYK2 (176941)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Hyper-IgE recurrent infection syndrome (147060); Hyper-IgE recurrent infection syndrome, autosomal recessive (243700); Netherton syndrome (256500); Tyrosine kinase 2 deficiency (611521)	Exon Array CGH, Next-gen Sequencing		1-Aug-07
Hyperglycemia and Hypoglycemia via the GCK Gene [1220]		GCK (138079)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Hyperinsulinemic Hypoglycemia, Familial 3 (602485); Maturity-Onset Diabetes Of The Young, Type 2 (125851); Permanent Neonatal Diabetes Mellitus (606176)	Sanger sequencing		24-Oct-17
Hyperglycosylated hCG			Gold SST	Quest Laboratories	First trimester screen for Down syndrome and trisomy 18			
Hyperlipidemia Panel Plus Analysis [CA1101]	1533	ABCA1 (600046), ABCG5 (605459); ABC8 (603076); APOA1 (107680); APOB (143890); APOC3 (107720); APOE (107741); LDLR (144010); LDLRAP1 (81479); LPL (699708); PCSK9 (603776)	Lavender Top (EDTA)	Blueprint Genetics http://blueprintgenetics.com/	Familial Hypercholesterolemia (143890); Hypercholesterolemia, Autosomal Dominant, Type B (144010); Hypercholesterolemia, Autosomal Dominant, 3 (603776); Hypercholesterolemia, Autosomal Recessive (603813)	Next Generation DNA Sequencing + Del/Dupl	Family Member testing $450	16-Jun-17
Hypertrophic Cardiomyopathy NGS Panel	287	ABCC9, ACTC1, ACTN2, ANKRD1, BAG3, BRAF, CAV3, CRYAB, CSRP3, DES, DMD, DSC2, DSG2, DSP, DTNA, EMD, FKTN, GATAD1, GLA, HRAS, ILK, JPH2, JUP, KRAS, LAMA4, LAMP2, LDB3, LMNA, MAP2K1, MAP2K2, MYBPC3, MYH7, MYL2, MYL3, MYLK2, MYOZ2, MYPN, NEBL, NEXN, NRAS, PDLIM3, PKP2, PLN, PRKAG2, PTPN11, RAF1, RBM20, RYR2, SCN5A, SGCD, SOS1, TAZ, TCAP, TMEM43, TMPO, TNNC1, TNNI3, TNNT2, TPM1, TTN, TTR, VCL	Lavender Top (2 x 4 mL)	Fulgent Genetics (fulgentdiagnostics.com)		NextGen Sequencing (24 genes)		11-Nov-19
Hypokalemic and Hyperkalemic Periodic Paralysis Disorders (NextGen Sequencing Panel and Copy Number Analysis; 7 Genes) [NGS332]	591	AIP (605555), AMMECR1 (300195), CACNA1S (114208), CLCN1 (118425), KCNJ2 (600681), SCN4A (603967), SLC12A3 (600968).	Lavender Top (EDTA)	MNG Laboratories (www.medicalneurogenetics.com)/Dynacare		NextGen Sequencing		9-May-20
Hypomyelinating leukodystrophy 7 and 8 (4H syndrome, 607694 and 614381)	964	POLR3A (614258); POLR3B (614366)	Lavender Top (2 x 4 mL)	Fulgent Genetics (fulgentdiagnostics.com)		NextGen Sequencing	Custom Panel; specific both genes	
Hypomyelinating Leukodystrophy 9 (616140)	964	RARS (107820)	Lavender Top (2 x 4 mL)	Alfred I Dupont Laboratory http://www.nemours.org/pediatric-research/labservices/diagnostic/molecular-diagnostics-lab.html				
Hypomyelinating Leukodystrophy With Atrophy of the Basal Ganglia and Cerebellum (H ABC)	964	TUBB4A (602662)	Lavender Top (2 x 4 mL)	Alfred I Dupont Laboratory http://www.nemours.org/pediatric-research/labservices/diagnostic/molecular-diagnostics-lab.html	Dystonia 4, torsion, autosomal dominant (128101); Leukodystrophy, hypomyelinating, 6 (612438)			
HYPOMYELINATION AND CONGENITAL CATARACT (HCC) VIA THE FAM126A GENE [1669]		FAM126A (610531)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Hypomyelination And Congenital Cataract (610532)	Sanger sequencing		
Hypophosphatasia via the ALPL Gene [851]		ALPL (171760)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Adult Hypophosphatasia (146300); Childhood Hypophosphatasia (241510); Infantile Hypophosphatasia (241500)			11-Jun-18
IA-2 Antibody [37933]	35		Gold SST or Red Top. Specimen Stability - Room temperature: 7 days Refrigerated: 7 days Frozen: 30 days	In-Common Laboraotories	anti-tyrosine phosphatase or anti-Islet antibody	ELISA		19-Jun-19
IBD sgi Diagnostic [1800]	120		2.0 mL Serum (Red Top or SST) AND 2.0 mL Whole Blood EDTA / Lavender Top Tube. Store at 4°C.	Prometheus Biosciences	differentiate among IBD types			
Ibuprofen (Motrin, Advil, Nuprin), serum [FIBUP]			Collect Plain Red. Also acceptable: Green top. NO GEL. Specimen Preparation Separate from cells. Transfer 1 mL serum or plasma to plastic vial. Store frozen.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	Optimize drug therapy and monitor patient adherence.	HPLC-UV		20-Nov-17

Appelation approuvée de l'analyse ou maladie associée [avec numéro de phénotype MIM] / Approved test name or Associated Disease [with MIM Phenotype Number]	**Code Test** / Test Code **(Dictionnaire MSSS)**	**SYMBOL HGNC DU GÈNE et NOM OMIM DU GÈNE** / HGNC Gene Symbol and OMIM Gene Number	**RÈGLES DE MANUTENTION** / Handling Conditions	**LABORATOIRE DÉSIGNÉ** / Designated laboratory	**RENSEIGNEMENTS SUR L'ANALYSE** / Test details	**MÉTHODE** / Methode	**NOTE PARTICULIÈRE** / Special comment	**Date de la révision** / Date of revision
Idiopathic Ataxia anti MPP-1	1465		Gold SST or Red Top	Mitogen Advanced Diagnostics				2-Jun-21
Idiopathic Generalized Epilepsy Panel		ADSL, ALDH7A1, ARHGEF9, ARX, ATP6AP2, ATRX, CACNA1A, CACNA1H, CACNB4, CASK, CASR, CDKL5, CHD2, CHRNA2, CHRNA4, CHRNB2, CLCN2, CNTN2, CNTNAP2, CPA6, CSTB, CUL4B, DCX, DEPDC5, DHFR, DNAJC5, DYNC1H1, EFHC1, EPM2A, FGD1, FOXG1, GABRA1, GABRB3, GABRD, GABRG2, GOSR2, GPC3, GRIA3, GRIN2A, HSD17B10, KANSL1, KCNC1, KCNJ10, KCNMA1, KCNQ2, KCNQ3, KCNT1, KCTD7, KDM5C, LGI1, MBD5, ME2, MECP2, MEF2C, NHLRC1, NIPA2, NRXN1, OFD1, OPHN1, PAK3, PCDH19, PHF6, PIGA, PLP1, PQBP1, PRICKLE1, PRICKLE2, PRRT2, RAB39B, ROGDI, SCARB2, SCN1A, SCN1B, SCN2A, SCN8A, SCN9A, SLC2A1, SLC9A6, SMC1A, SRPX2, STX1B, STXBP1, SYN1, SYNGAP1, SYP, TBC1D24, TCF4, UBE3A, ZEB2	Lavender Top (2 x 4 mL)	Fulgent Genetics (fulgentdiagnostics.com)				
IGF Binding Protein-1 (IGFBP-1) [FIGBP]			Draw blood in a plain, red-top tube(s). Spin down and separate within one hour. Ship 0.5 mL frozen in a plastic vial.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)		RIA		20-Nov-17
IGF-II [FIGF2]	123		Draw blood in a plain, red-top tube(s). SST acceptable. Separate within 1 hour of collection,freeze immediately	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)				30-Jul-20
IL12Rß1 (CD212) [IL12PATHWAY]	817		Heparin	Alberta Precision Laboratories (Flow Cytometry - Calgary)			To be accompanied by normal control sample	14-Oct-20
IL-2 by Luminex [LIL2]	1523		Plasma from a lavender top (EDTA) tube is the only acceptable sample type. Mix sample thoroughly. Specimen Preparation: Centrifuge at room temp within one half hour of collection; preferably immediately after venipuncture. Transfer the cell-free plasma to a clean tube and immediately freeze the cell-free plasma on dry ice or at -70°C.	National Jewish Health		Luminex		18-Mar-21
Inclusion body myopathy and autosomal recessive, ealrly onset myopathy via the MYH2 gene [361]		MYH2 (160740)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Inclusion Body Myopathy 3 (605637)	Sanger sequencing		
INCLUSION BODY MYOPATHY-2 (AUTOSOMAL RECESSIVE) AND NONAKA MYOPATHY VIA THE GNE GENE [367]		GNE (6603824)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Inclusion Body Myopathy 2 (600737); Nonaka Myopathy (605820)	Sanger sequencing		
INGγR12 (CD119) [ILFGPATHWAY]	817		Heparin	Alberta Precision Laboratories (Flow Cytometry - Calgary)			To be accompanied by normal control sample	14-Oct-20
Insulin Antibodies [INSAB]	853		Gold SST or Red Top	In Common Laboratories		RIA		19-Jun-19
Insulin Receptor		INSR (147670)	Lavender Top (EDTA) 2-4 cc (1-2 cc for children less than 1year old)	Fulgent Genetics (fulgentdiagnostics.com)	Hyperinsulinemic hypoglycemia familial 5 (609968); Insulin-resistant diabetes mellitus AND acanthosis nigricans (610549); Leprechaunism syndrome (246200); Pineal hyperplasia AND diabetes mellitus syndrome (262190)	NGS and CGH		
Insulin-Like Growth Factor 1 (IGF1), LC-MS and Insulin-Like Growth Factor-Binding Protein 3 (IGFBP3) Growth Panel [IGFGP]			Red Top. Split into 2 plastic vials.Store and send serum frozen.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)		IGFMS: Liquid Chromatography-Mass Spectrometry (LC-MS) IGFB3: Enzyme-Labeled Chemiluminescent Immunometric Assay	Not M-R	20-Oct-20

Appelation approuvée de l'analyse ou maladie associée [avec numéro de phénotype MIM] / Approved test name or Associated Disease [with MIM Phenotype Number]	**Code Test** / Test Code **(Dictionnaire MSSS)**	**SYMBOL HGNC DU GÈNE et NOM OMIM DU GÈNE** / HGNC Gene Symbol and OMIM Gene Number	**RÈGLES DE MANUTENTION** / Handling Conditions	**LABORATOIRE DÉSIGNÉ** / Designated laboratory	**RENSEIGNEMENTS SUR L'ANALYSE** / Test details	**MÉTHODE** / Methode	**NOTE PARTICULIÈRE** / Special comment	**Date de la révision** / Date of revision
Intellectual Disability NGS Panel	920	ABCC6, ABCD1, ABCG5, ACAT1, ACOX1, ACSL4, ACY1, ADAR, ADSL, AFF2, AGL, AGT, AGTR2, AHI1, AIFM1, ALDH18A1, ALDH4A1, ALDH5A1, ALG11, ALG12, ALG6, ALX4, AMER1, ANK3, ANKRD11, AP1S1, AP1S2, AP3B1, AP4B1, AP4E1, AP4M1, AP4S1, AR, ARG1, ARHGEF6, ARHGEF9, ARID1A, ARID1B, ARX, ASPM, ASS1, ATL1, ATP10A, ATP13A2, ATP1A2, ATP6AP2, ATP7A, ATRX, AUH, AUTS2, AVP, AVPR1A, AVPR2, BBS9, BCOR, BCS1L, BDNF, BIN1, BRAF, BRIP1, BRWD3, BUB1B, CACNA1C, CACNG2, CAMTA1, CANT1, CASK, CBS, CC2D1A, CC2D2A, CCDC22, CCDC88C, CDH15, CDK16, CDKL5, CDKN1C, CEP290, CEP41, CEP57, CHD7, CHD8, CHRNA4, CLCN4, CLIC2, CLN3, CNKSR2, CNTNAP2, CNTNAP5, COG5, COG7, COL1A2, CP, CPA6, CPS1, CRADD, CRBN, CREBBP, CTC1, CTNNB1, CTSA, CUL4B, CYB5R3, CYP27A1, D2HGDH, DARS2, DBT, DCX, DHCR24, DHCR7, DKC1, DLG3, DLGAP2, DMD, DOCK4, DPP10, DPP6, DPYD, DYNC1H1, DYRK1A, EBP, EFNB1, EHMT1, EIF2S3, ELOVL4, ERCC2, ERCC3, ERCC5, ERCC6, ERCC8, FAAH2, FAM126A, FANCB, FANCG, FBLN5, FBN1, FGD1, FGF14, FGFR1, FGFR2, FGFR3, FKRP, FKTN, FLNA, FMR1, FOLR1, FOXG1, FOXP1, FOXP2, FRMPD4, FTO, FTSJ1, G6PC3, GABRB3, GABRG1, GABRG2, GALE, GAMT, GAN, GBA, GBE1, GCK, GDI1, GFAP, GFM1, GHR, GK, GLI3, GLRA1, GLUL, GLYCTK, GM2A, GNA14, GNAS, GNPAT, GNPTAB, GNPTG, GPC3, GRIA3, GRIK2, GRIN1, GRIN2A, GRIN2B, GRM1, GRPR, GSPT2, GSS, GUSB, GYS2, HAX1, HCCS, HCFC1, HDAC4, HDAC8, HECW2, HEPACAM, HEXB, HOXA1, HOXD10, HPD, HPRT1, HSD17B10, HSPD1, HUWE1, IDS, IGBP1, IGF1, IGF1R, IL1RAPL1, IMMP2L, INSR, IQSEC2, IRX5, ITGA7, KATNAL2, KCNJ10, KCNJ11, KCNK9, KCNQ2, KCTD13, KCTD7, KDM5C, KDM6A, KIAA2022, KIF11, KIF1A, KIF21A, KIF5A, KIF7, KIRREL3, KLF8, KMT2D, KRAS, L1CAM, LAMA2, LAMC3, LAMP2, LARGE1, LAS1L, LBR, LHX3, LIG4, LMBRD1, LRP5, LYST, MAGT1, MAN1B1, MAN2B1, MANBA, MAOA, MAT1A, MBD5, MBTPS2, MCCC1, MCCC2, MCOLN1, MCPH1, MECP2, MED12, MED17, MED23, MEF2C, MET, MFSD8, MGAT2, MID1, MKKS, MMADHC, MOCS2, MPI, MPZ, MRAP, MTFMT, MTHFR, MTM1, MTR, MYCN, MYO5A, MYO7A, NAA10, NAGA, NBN, NDP, NDUFA1, NDUFAF5, NDUFS1, NEGR1, NF1, NGF, NGLY1, NHEJ1, NHP2, NHS, NIPBL, NLGN3, NLGN4X, NPC1, NPC2, NPHP3, NRXN1, NSD1, NSDHL, NSUN2, NTNG1, OCRL, OFD1, OGT, OPHN1, ORC1, OTC, PAFAH1B1, PAH, PAK3, PAX6, PCDH19, PCDH9, PCNT, PDE10A, PDE4D, PDHA1, PDHX, PDSS1, PEX7, PGK1, PHF6, PHF8, PHKA2, PHKG2, PIGL, PIGO, PIGV, PIP5K1B, PLA2G6, PLP1, PNKP, POMGNT1, POMT1, POMT2, PON3, PORCN, POU1F1, PPOX, PQBP1, PRICKLE1, PRKAR1A, PRPS1, PRSS12, PTCHD1, PTEN	Lavender Top (2 x 4 mL)	Fulgent Genetics (fulgentdiagnostics.com)		NextGen Sequencing		28-Jul-07
Interferon Beta Neutralizing Antibodies In MS Patient Treated With IFN [P91858]			Gold SST	Neuro-Immunology Laboratories (UBC)				
Interleukin-2 (IL-2), ELISA [FIL2]	1523		Draw sufficient blood in a plain, red-top tube(s). Spin down and pour off into a plastic, screw-capped tube. Freeze immediately.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)		ELISA		18-Mar-21
Interstitial Lung Disease Antibody (ILD) Profile	1724		Gold SST or Red Top	Mitogen Advanced Diagnostics		LIA		2-Jun-21
Iron-refractory Iron Deficiency Anemia (TMPRSS6 Single Gene Test)		TMPRSS6 (609862)	Lavender Top (2 x 4 mL)	Fulgent Genetics (fulgentdiagnostics.com)	Iron refractory iron deficiency anemia (206200)	NextGen Sequencing		30-Apr-18
ITGA9 - Single Gene Testing		ITGA9 (603963)	Lavender Top (2 x 4 mL)	Fulgent Genetics (fulgentdiagnostics.com)	Integrin alpha 9	NextGen Sequencing		30-Apr-18
JAG1 Gene Sequencing & Del/Dup [1004]		JAG1 (601920)	Lavender Top (EDTA)	GeneDx (www.genedx.com)	Alagille syndrome (118450)			9-Dec-19
Jaundice Chip			Lavender Top (EDTA)	Cinncinatti Children's Hospital (Division of Human Genetics Diagnostic Laboratories)	multiple genes	gene chip		
JOUBERT AND MECKEL-GRUBER SYNDROMES NEXTGEN SEQUENCING (NGS) PANEL [1057]	332	HJT (608894) ARL13B (608922) B9D1 (614144) B9D2 (611951) C5orf42 (614571) CC2D2A (612013) CEP290 (610142) CEP41 (610523)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)		NextGen Sequencing		
JUNCTIONAL EPIDERMOLYSIS BULLOSA (JEB) SANGER sequencing panel [975]		LAMC2 (150292); LAMA3 (600805); LAMB3 (150310); COL17A1 (113811)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)				
Juvenile Polyposis Syndrome (610069 and 174900) [536]		BMPR1A (601299); SMAD4 (600993)	Lavender Top (EDTA)	GeneDx (www.genedx.com)		Tier 1: Sanger Sequencing, Exon Array CGH		9-Dec-19
Juvenile Polyposis Syndrome (610069 and 174900) [537]		BMPR1A (601299)	Lavender Top (EDTA)	GeneDx (www.genedx.com)		Tier 2: sequencing		9-Dec-19
Juvenile Polyposis Syndrome (610069 and 174900) [538]		BMPR1A (601299); SMAD4 (600993)	Lavender Top (EDTA)	GeneDx (www.genedx.com)		Tier 3: Exon Array CGH		9-Dec-19

Appelation approuvée de l'analyse ou maladie associée [avec numéro de phénotype MIM] / Approved test name or Associated Disease [with MIM Phenotype Number]	**Code Test** / Test Code **(Dictionnaire MSSS)**	**SYMBOL HGNC DU GÈNE et NOM OMIM DU GÈNE** / HGNC Gene Symbol and OMIM Gene Number	**RÈGLES DE MANUTENTION** / Handling Conditions	**LABORATOIRE DÉSIGNÉ** / Designated laboratory	**RENSEIGNEMENTS SUR L'ANALYSE** / Test details	**MÉTHODE** / Methode	**NOTE PARTICULIÈRE** / Special comment	**Date de la révision** / Date of revision
KCNA1 Full Gene Sequencing Analysis [MOL064]		KCNA1 (176260)	Lavender Top (EDTA)	MNG Laboratories (www.medicalneurogenetics.com)/Dynacare	Episodic ataxia/myokymia syndrome			9-May-20
Kidney Dysplasia NGS Panel	1615	ACE, AGT, AGTR1, ANOS1, BMP4, BMP7, CDC5L, CHD1L, DSTYK, EYA1, FGF20, FGFR2, FRAS1, FREM1, FREM2, GATA3, GLI3, GRIP1, HNF1B, HOXA13, HOXA4, HOXB6, HPSE2, ITGA8, LRP4, MUC1, MYH9, NIPBL, PAX2, REN, RET, ROBO2, SALL1, SIX1, SIX2, SIX5, SOX17, TRAP1, UMOD, UPK3A, WNT4	Lavender Top (EDTA) [2 tubes]	Fulgent Genetics (fulgentdiagnostics.com)		NextGen Sequencing		28-Feb-20
KIT Single Gene	1537	cKit (164920)	Lavender Top (2 x 4 mL)	Fulgent Genetics (fulgentdiagnostics.com)	Systemic mastocytosis (154800)	NextGen Sequencing		Januray 30, 2019
KRABBE DISEASE VIA THE GALC GENE [631]		GALC (606890)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Galactosylceramide Beta-Galactosidase Deficiency (245200)	Sanger sequencing	enzyme activity at HSJ	
L1CAM (fetal sexing)		L1CAM (308840)	Gold SST	Laboratoire Cerba (www.lab-cerba.com)				
L1CAM Gene Sequencing & Del/Dup [552]		L1CAM (308840)	Lavender Top (EDTA)	GeneDx (www.genedx.com)	X-Linked Hydrocephalus Syndrome (307000); Spastic Paraplegia 1 (303350); Corpus Callosum, Partial Agenesis Of, X-Linked (304100)	1. Sanger sequencing and aCGH 2. targeted mutation		9-Dec-09
Lacosamide, Serum [LACO]	1572		RED OR GOLD TOP. Store frozen	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)		HPLC-MS		28-Jun-18
LAMINOPATHIES VIA THE LMNA GENE [347]		LMNA (150330)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Charcot-Marie-Tooth Disease Type 2B1 (605588); Limb-Girdle Muscular Dystrophy, Type 1B (159001); Lipodystrophy, Familial Partial, Dunnigan Type (151660); Dilated Cardiomyopathy 1A (115200); Emery-Dreifuss Muscular Dystrophy, Autosomal Dominan (151350); Restrictive Dermopathy, Lethal (275210); Hutchinson-Gilford Syndrome (176670)	Sanger sequencing		8-May-17
Left Ventricular Noncompaction Cardiomyopathy Syndromes (NextGen Sequencing Panel and Copy Number Analysis; 8 genes) [NGS364]		ACTC1 (102540), DTNA (601239), ITGA7 (600536), LDB3 (605906), MYH7 (160760), MYH7B (609928), PRDM16 (605557), TNNT2 (191045).	Lavender Top (EDTA)	MNG Laboratories (www.medicalneurogenetics.com)/Dynacare		NextGen Sequencing		9-May-20
Leigh disease and Leigh-like syndromes NextGen DNA Sequencing Panel (75 genes) [NGS351]		AIFM1 (300169); ALDH5A1 (610045); ARX (300382); BCS1L (603647); C12orf65 (613541); COA5 (613920); COX10 (602125); COX14 (614478); COX15 (603646); COX6B1 (124089); CPT2 (600650); DLAT (608770); DLD (238331); ETHE1 (608451); FARS2 (611592); FASTKD2 (612322); FOXRED1 (613622); GCDH (608801); KCNQ2 (602235); LIAS (607031); LRPPRC (607544); MTFMT (611766); MUT (609058); NDUFA1 (300078); NDUFA10 (603835); NDUFA11 (612638); NDUFA12 (614530); NDUFA2 (602137); NDUFA9 (603834); NDUFAF1 (606934); NDUFAF2 (609653); NDUFAF3 (612911); NDUFAF4 (611776); NDUFAF5 (612360); NDUFAF6 (612392); NDUFB3 (603839); NDUFS1 (157655); NDUFS2 (602985); NDUFS3 (603846); NDUFS4 (602694); NDUFS6 (603848); NDUFS7 (601825); NDUFS8 (602141); NDUFV1 (161015); NDUFV2 (600532); NDUFV3 (602184); NUBPL (613621); PC (608786); PDHA1 (300502); PDHA2 (179061); PDHB (179060); PDHX (608769); PDP1 (605993); PDSS2 (610564); PNPT1 (610316); POLG (174763); RANBP2 (601181); SCO1 (603644); SCO2 (604272); SCP2 (184755); SDHA (600857); SDHAF1 (612848); SERAC1 (614725); SLC19A3 (606152); SLC25A19 (606521); SUCLA2 (603921); SUCLG1 (611224); SUOX (606887); SURF1 (185620); TACO1 (612958); TLR3 (603002); TPK1 (606370); TTC19 (613814); UQCRB (191330); UQCRQ (612080)	Lavender Top (EDTA)	MNG Laboratories (www.medicalneurogenetics.com)/Dynacare	Leigh syndrome (256000); Leigh syndrome, X-linked (308930)	NextGen Sequencing		9-May-20
Leptin [FLEP]	1465		Draw blood in a plain, red-top tube(s). (Serum gel tube is acceptable.) Separate and freeze within one hour. Send 1 mL of serum frozen in a plastic vial. Store frozen.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)		ELISA		12-Jun-20
Leukemia, Philadelphia chromosome-positive, resistant to imatinib		ABL1 (189980)	Lavender Top (EDTA)	University Health Network (Toronto General Hospital)				
Leukemia/Lymphoma Immunophenotyping (PNH and ZAP-70 available) [2001]	1465		Na heparin tube. Store and send at RT. Send immediately overnight.	Hematologics, Inc. 3161 Elliot Ave. Suite 200, Seattle WA 98121 1800-860-0934		Flow cytometry		23-Jun-20
Leukodystrophy with dysmyelination and spastic paraparesis with or without dystonia (SPG35) (612319)	964	FA2H (611026)	Lavender Top (EDTA)	Centogene AG (www.centogene.com)		Sanger sequencing		
Leukotriene E4, Urine [LTE4]	1465		Preferred: 24-hour urine collection Container/Tube: Plastic, 5-mL tube (T465) Specimen Volume: 4 mL Collection Instructions: 1. Collect urine for 24 hours. 2. Refrigerate specimen during collection, aliquot 4 mL of urine into plastic tube, and send specimen refrigerated. Store frozen (30 days).	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)		LC-MS/MS		12-Jan-21

Appelation approuvée de l'analyse ou maladie associée [avec numéro de phénotype MIM] / Approved test name or Associated Disease [with MIM Phenotype Number]	**Code Test** / Test Code **(Dictionnaire MSSS)**	**SYMBOL HGNC DU GÈNE et NOM OMIM DU GÈNE** / HGNC Gene Symbol and OMIM Gene Number	**RÈGLES DE MANUTENTION** / Handling Conditions	**LABORATOIRE DÉSIGNÉ** / Designated laboratory	**RENSEIGNEMENTS SUR L'ANALYSE** / Test details	**MÉTHODE** / Methode	**NOTE PARTICULIÈRE** / Special comment	**Date de la révision** / Date of revision
LIMB GIRDLE MUSCULAR DYSTROPHY (LGMD) NEXTGEN SEQUENCING (NGS) PANEL [1345]		ANO5 (608662); CAPN3 (114240); CAV3 (601253O; DES (125660); DNAJB6 (611332); DYSF (603009); FKRP (606596); GMPPB (615320); ISPD (614631); LIMS2 (607908); LMNA (150330); MYOT (604103); PNPLA2 (609059); SGCA (600119); SGCB (600900); SGCD (601411); SGCG (608896); SMCDH1 (614982); TCAP (604488); TNOP3 (610032); TOR1AIP1 (614512); TRAPPC11 (614138); TRIM32 (602290); TTN (188840)	Lavender Top (EDTA) [2 tubes]	Prevention Genetics (www.preventiongenetics.com)	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 5 (607155); Limb-Girdle Muscular Dystrophy, Type 2H (254110);Limb-Girdle Muscular Dystrophy, Type 2G (601954);Limb-Girdle Muscular Dystrophy, Type 2C (253700); Limb-Girdle Muscular Dystrophy, Type 2F (601287); Limb-Girdle Muscular Dystrophy, Type 2E (604286); Limb-Girdle Muscular Dystrophy, Type 2D (608099); Limb-Girdle Muscular Dystrophy, Type 2B (253601); Limb-Girdle Muscular Dystrophy, Type 2A (253600); Limb-Girdle Muscular Dystrophy, Type 2I (608807) ; Limb-Girdle Muscular Dystrophy, Type 2L (611307); Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 14 (615352); Muscular dystrophy, limb-girdle, type 2S (615356); Limb-Girdle Muscular Dystrophy, Type 1E (603511); Muscular Dystrophy, Limb-Girdle, Type 1C (607801); Limb-Girdle Muscular Dystrophy, Type 1B (159001); Limb-Girdle Muscular Dystrophy, Type 1A (159000); Facioscapulohumeral Muscular Dystrophy 2 (158901); Neutral Lipid Storage Disease With Myopathy (610717); Muscular dystrophy, limb-girdle, type 2R (615325); Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7 (614643)	NextGen Sequencing (24 genes)		
LIMB GIRDLE MUSCULAR DYSTROPHY TYPE 2B AND MIYOSHI MYOPATHY VIA THE DYSF GENE [3123]	606	DYSF (603009)	Lavender Top (EDTA) [2 tubes]	Prevention Genetics (www.preventiongenetics.com)	Limb-Girdle Muscular Dystrophy, Type 2B (253601); Miyoshi Myopathy (254130), Myopathy, Distal, With Anterior Tibial Onset (606768)			9-Mar-21
LIMB GIRDLE MUSCULAR DYSTROPHY, TYPE 2L (LGMD2L) AND DISTAL MIYOSHI MYOPATHY (MMD3) VIA THE ANO5 GENE [469]	606	ANO5 (608662)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Miyoshi Muscular Dystrophy 3 (613319); Limb-Girdle Muscular Dystrophy, Type 2L (611307)	Sanger sequencing		
Lipid Metabolism Deficiency NGS Sequencing Panel (71 genes) + Del/Dup + mtDNA [NGS303]		ABCD1 (300371); ABCD2 (601081); ACAA1 (604054); ACAA2 (604770); ACACA (200350); ACACB (601557); ACAD10 (611181); ACAD11 (614288); ACAD9 (611103); ACADL (609576); ACADM (607008); ACADS (606885); ACADSB (600301); ACADVL (609575); ACAT1 (607809); ACAT2 (100678); ACLY (108728); ACOT1 (614313); ACOT12 (614315); ACOT2 (609972); ACOT4 (614314); ACOT6 (614267); ACOT7 (602587); ACOT8 (608123); ACOT9 (300862); ACOX1 (609751); ACOX2 (601641); ACOX3 (603402); ACP6 (611471); ACSBG1 (614362); ACSBG2 (614363); ACSF2 (610465); ACSF3 (614245); ACSL1 (152425); ACSL3 (602371); ACSL4 (300157); ACSL5 (605677); ACSL6 (604443); ACSM1 (614357); ACSM2A (614358); ACSM2B (614359); ACSM3 (145505); ACSM4 (614360); ACSM5 (614361); ACSS1 (614355); ACSS2 (605832); ADHFE1 (611083); ALDH3A2 (609523); BAAT (602938); BBOX1 (603312); BDH1 (603063); BTD (609019); CHKB (612395); CPT1A (600528); CPT1B (601987); CPT1C (608846); CPT2 (600650); CRAT (600184); CROT (606098); CYP4A11 (601310); DECR1 (222745); DHRS1 (610410); ECHS1 (602292); ECI1 (600305); ECI2 (608024); EHHADH (607037); ELOVL1 (611813); ELOVL2 (611814); ELOVL3 (611815); ELOVL4 (605512); ELOVL5 (611805); ELOVL6 (611546); ELOVL7 (614451); FAAH2 (300654); FABP1 (134650); FABP2 (134640); FABP3 (134651); FABP4 (600434); FABP5 (605168); FABP6 (600422); FABP7 (602965); FADS1 (606148); FADS2 (606149); FADS3 (606150); FASN (600212); GPAM (602395); GPD1 (138420); GPD2 (138430); HADH (601609); HADHA (600890); HADHB (143450); HLCS (609018); HMGCL (613898); HMGCS2 (600234); HSD17B12 (609574); LIPE (151750); MCAT (614479); MECR (608205); MLYCD (606761); OPA3 (606580); OXCT1 (601424); OXCT2 (610289); OXSM (610324); PECR (605843); PNPLA2 (609059); PPARGC1A (604517); PPARGC1B (608886); PRKAA1 (602739); PRKAA2 (600497); PTPLA (610467); SCP2 (184755); SLC22A5 (603377); SLC25A20 (613698); SLC27A1 (600691); SLC27A2 (603247); SLC27A3 (604193); SLC27A4 (604194); SLC27A5 (603314); SLC27A6 (604196); SLC33A1 (603690); TECR (610057)	Lavender Top (EDTA)	MNG Laboratories (www.medicalneurogenetics.com)/Dynacare		NextGen Sequencing		9-May-20
Lipodystrophy		AGPAT2 (603100), AKT2 (164731), BSCL2 (606158), CAV1 (601047), CIDEC (612210) , LMNA (150330), PPARG (601487), PTRF (603198), TBC1D4 (612465), ZMPSTE24 (606480)	Lavender Top (2 x 4 mL)	Fulgent Genetics (fulgentdiagnostics.com)		NextGen Sequencing		
Lissencephaly NGS Panel		ACTB, ACTG1, ARX, DCX, FKRP, FKTN, LARGE, PAFAH1B1, POMGNT1, POMT1, POMT2, RELN, TUBA1A, VLDLR	Lavender Top (2 x 4 mL)	Fulgent Genetics (fulgentdiagnostics.com)		NextGen Sequencing		

Appelation approuvée de l'analyse ou maladie associée [avec numéro de phénotype MIM] / Approved test name or Associated Disease [with MIM Phenotype Number]	**Code Test** / Test Code **(Dictionnaire MSSS)**	**SYMBOL HGNC DU GÈNE et NOM OMIM DU GÈNE** / HGNC Gene Symbol and OMIM Gene Number	**RÈGLES DE MANUTENTION** / Handling Conditions	**LABORATOIRE DÉSIGNÉ** / Designated laboratory	**RENSEIGNEMENTS SUR L'ANALYSE** / Test details	**MÉTHODE** / Methode	**NOTE PARTICULIÈRE** / Special comment	**Date de la révision** / Date of revision
Long QT/Brugada Syndrome NGS Panel	285	AKAP9 (604001), ANK2 (106410), CACNA1C (114205), CAV3 (601253), KCNE1 (176261), KCNE2 (603796), KCNH2 (152427), KCNJ2 (600681), KCNJ5 (600734), KCNQ1 (607542), SCN4B (608256), SCN5A (600163), SNTA1 (601017)	Lavender Top (2 x 4 mL)	Fulgent Genetics (fulgentdiagnostics.com)	Long QT Syndrome (192500)	NextGen Sequencing		6-Jan-20
LRBA	1248		Na heparin tube. Store and send at RT. Send immediately overnight.	Alberta Precision Laboratories (Flow Cytometry - Calgary)		Flow cytometry		1-Jun-21
LRP4 Autoantibody Test [1483]	18		Serum (Gold SST or Red Top). Store at -20°C	Athena Diagnostics (www.athenadiagnostics.com)	For AcR and MUSK negative subjects	IFA		21-Aug-20
LYMPHEDEMA-DISTICHIASIS SYNDROME VIA THE FOXC2 GENE [280]		FOXC2 [602402]	Lavender top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Lymphedema-Distichiasis Syndrome (153400)	Sanger sequencing and del/dupl		
Lysosomal Disease (NextGen Sequencing Panel and Copy Number Analysis; 72 Genes) [NGS313]	474	ADAMTSL2 (612277), AGA (613228), ANTXR2 (608041), ARSA (607574), ARSB (611542), ASAH1 (613468), ATP13A2 (610513), ATP7A (300011), ATP7B (606882), CERS1 (606919), CLN3 (607042), CLN5 (608102), CLN6 (606725), CLN8 (607837), COL11A2 (120290), COL2A1 (120140), CTNS (606272), CTSA (613111), CTSC (602365), CTSD (116840), CTSF (603539), CTSK (601105), DHCR7 (602858), DNAJC5 (611203), DYM (607461), FUCA1 (612280), GAA (606800), GALC (606890), GALNS (612222), GBA (606463), GLA (300644), GLB1 (611458), GM2A (613109), GNE (603824), GNPTAB (607840), GNPTG (607838), GNS (607664), GPC3 (300037), GRN (138945), GUSB (611499), HEXA (606869), HEXB (606873), HGSNAT (610453), HRAS (190020), HYAL1 (607071), IDS (300823), IDUA (252800), KCTD7 (611725), KDM6A (300128), MLL2 (602113), LAMP2 (309060), LIPA (613497), LMBRD1 (612625), LYST (606897), MAN2B1 (609458), MANBA (609489), MCOLN1 (605248), MFSD8 (611124), NAGA (104170), NAGLU (609701), NEU1 (608272), NPC1 (607623), NPC2 (601015), PPT1 (600722), PSAP (176801), RAI1 (607642), SGSH (605270), SLC17A5 (604322), SMPD1 (607608), SUMF1 (607939), TCF4 (602272), TPP1 (607998).	Lavender Top (EDTA)	MNG Laboratories (www.medicalneurogenetics.com)/Dynacare		Next Generation DNA Sequencing		9-May-20
Malignant Hyperthermia (NextGen Sequencing Panel and Copy Number Analysis; 2 Genes) [NGS333]		CACNA1S (114208), RYR1 (180901)	Lavender Top (EDTA)	MNG Laboratories (www.medicalneurogenetics.com)/Dynacare		NextGen Sequencing		9-May-20
Mannose-Binding Lectin Deficiency NGS Test		MBL2 (154545)	Lavender Top (2 x 4 mL)	Fulgent Genetics (fulgentdiagnostics.com)	Mannose-binding protein deficiency (614372)	Next Generation DNA Sequencing		23-Oct-17
Marfan Syndrome and Thoracic Aortic Aneurysm and Dissection NGS Panel	279	ACTA2, CBS, COL3A1, COL5A1, COL5A2, FBN1, FBN2, FLNA, MED12, MYH11, MYLK, NOTCH1, PLOD1, PRKG1, SKI, SLC2A10, SMAD3, SMAD4, TGFB2, TGFB3, TGFBR1, TGFBR2 (22 genes)	Lavender Top (2 x 4 mL)	Fulgent Genetics (fulgentdiagnostics.com)	Marfan syndrome, type I (154700)	NextGen Sequencing		17-Feb-19
MARINESCO-SJOGREN SYNDROME VIA THE SIL1 GENE [1674]		SIL1 (608005)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Marinesco-Sjogren Syndrome (248800)	Sanger sequencing		
Meckel-Gruber syndrome Sequencing Panel		CC2D2A (612013); CEP290 (610142); MKS (609883); RPGRIP1L (610937); TCTN (613846)2; TMEM67 (609884); TMEM216 (613877)	Lavender Top (EDTA)	University of Chicago Genetic Services Laboratories (dnatesting.uchicago.edu)	Meckel Syndrome, Type 8 (613885); Meckel Syndrome, Type 6 (312284); Meckel Syndrome, Type1 (249000); Meckel Syndrome, Type 4 (611134); Meckel Syndrome, Type 3 (607361); Meckel Syndrome, Type 5 (611561); Meckel syndrome 2 (603194)	Sanger sequencing		
MECP2 Analysis		MECP2 (300005)	Lavender Top (EDTA) 2-5 cc	Alberta Children's hospital http://www.medicalgenetics.ca/molecular.html	Rett's disorder (312750)	Sanger sequencing		4-May-17
Medullary Cystic Kidney Disease type 2 and Familial Juvenile Hyperuricemic Nephropathy type 1 via the UMOD Gene [1681]		UMOD (191845)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Familial Juvenile Hyperuricemic Nephropathy (162000); Medullary Cystic Kidney Disease 2(603860)	Sanger sequencing	aCGH [600] $690	15-Mar-07
MERRF (545000), MELAS (540000), NARP (551500)			Lavender Top (EDTA)	Alberta Mol Dx Laboratory (Edmonton)		Targeted mutation analysis		

Appelation approuvée de l'analyse ou maladie associée [avec numéro de phénotype MIM] / Approved test name or Associated Disease [with MIM Phenotype Number]	**Code Test** / Test Code **(Dictionnaire MSSS)**	**SYMBOL HGNC DU GÈNE et NOM OMIM DU GÈNE** / HGNC Gene Symbol and OMIM Gene Number	**RÈGLES DE MANUTENTION /** Handling Conditions	**LABORATOIRE DÉSIGNÉ** / Designated laboratory	**RENSEIGNEMENTS SUR L'ANALYSE** / Test details	**MÉTHODE /** Methode	**NOTE PARTICULIÈRE /** Special comment	**Date de la révision /** Date of revision
Metabolic Hypoglycemia Sequencing Panel with CNV Detection [10365]	1517	ACADM 607008 ACADVL 609575 ACAT1 607809 ACSF3 614245 AGL 610860 ALDOB 612724 CASA 114761 DGUOK 601465 ETFA 608053 ETFB 130410 ETFDH 231675 FBP1 611570 G6PC 613742 GALT 606999 GK 300474 GYS2 138571 HADH 601609 HMGCL 613898 HMGCS2 600234 MLYCD 606761 MPV17 137960 NNT 607878 OXCT1 601424 PC 608786 PCK1 614168 PCK2 614095 PGM1 171900 PHKA2 300798 PHKB 172490 PHKG2 172471	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	3-Hydroxy-3-Methylglutaryl-CoA Lyase Deficiency AR 246450 Alpha-Methylacetoacetic Aciduria AR 203750 Combined Malonic And Methylmalonic Aciduria AR 614265 Congenital Disorder of Glycosylation, Type It AR 614921 Fanconi-Bickel Syndrome AR 227810 Fructose-Biphosphatase Deficiency AR 229700 Galactosemia AR 230400 Glycogen Storage Disease 0, Liver AR 240600 Glycogen Storage Disease Type Ia AR 232200 Glycogen Storage Disease Type Ib AR 232220 Glycogen Storage Disease Type Ic AR 232240 Glycogen Storage Disease Type III AR 232400 Glycogen Storage Disease Type IXa1 XL 306000 Glycogen Storage Disease Type IXc AR 613027 Glycogen Storage Disease Type VI AR 232700 Glycogen Storage DiseaseType IXb AR 261750 Hereditary Fructose Intolerance AR 229600 Malonyl-CoA Decarboxylase Deficiency AR 248360 Monocarboxylate Transporter 1 Deficiency AD,AR 616095 Phosphoenolpyruvate Carboxykinase Deficiency, Cytosolic AR 261680 Phosphoenolpyruvate Carboxykinase Deficiency, Mitochondrial AR 261650 Pyruvate Carboxylase Deficiency AR 266150 Succinyl-CoA Acetoacetate Transferase Deficiency AR 245050	NextGen Sequencing		15-Aug-19
Metformin, Serum/Plasma [FMETF]			Red Top or Lavender Top only. Store and send serum/plasma frozen.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)				
Microcephaly NGS Panel	350	AKT3, ANKLE2, ARFGEF2, ASPM, ATR, ATRIP, BUB1B, CASK, CDK5RAP2, CDK6, CENPE, CENPF, CENPJ, CEP135, CEP152, CEP63, CKAP2L, COX7B, CRIPT, DIAPH1, DNM1L, EFTUD2, HMGB3, IER3IP1, KATNB1, KIF11, KNL1, LIG4, MCPH1, MED17, MFSD2A, MIR17HG, MRE11, MSMO1, MYCN, NBN, NDE1, NHEJ1, NIN, NR2E1, PAFAH1B1, PCLO, PCNT, PHC1, PLEKHG2, PLK4, PNKP, POMT1, PPP1R15B, PQBP1, QARS, RARS2, RBBP8, RTTN, SASS6, SLC1A4, SLC25A19, SLC9A6, SPATA5, STAMBP, STIL, THOC6, TRMT10A, TSEN2, TSEN34, TSEN54, TUBB2B, TUBGCP4, TUBGCP6, VRK1, WDR62, WDR73, XRCC4, ZEB2, ZNF335 (75 genes)	Lavender Top (EDTA)	Fulgent Genetics (fulgentdiagnostics.com)		NextGen Sequencing		23-Oct-17
Migraine Panel [NE1201]	581		Lavender Top (EDTA)	Blueprint Genetics http://blueprintgenetics.com/		Next-Generation sequencing		21-Aug-20
Minimal Residual Disease Testing (MRD) T cell [2002]	1084		Na heparin tube. Store and send at RT. Send immediately overnight.	Hematologics, Inc. 3161 Elliot Ave. Suite 200, Seattle WA 98121 1800-860-0934		Flow cytometry		23-Jun-20
Minimal Residual Disease Testing for Acute Lymphoblastic Leukemia	1084		EDTA Whole Blood. Keeep at room temperature.	Flow Cytometry Laboratory. London Health Sciences Centre.	For Friday samples email Ben Hedley with ID information & location where sample is being sent.	Flow cytometry		4-Aug-20
Mitochondrial Genome Sequencing [MOL021]	518	The human mitochondrial genome is 16569 base pairs in length and encodes 37 genes including 2 ribosomal RNA genes, 22 transfer RNA genes, and 13 protein-coding genes. Our mitochondrial DNA (mtDNA) panel includes complete sequencing of the following genes (MIM#): MTRNR1 (561000), MTRNR2 (561010); MTTA (590000); MTTR (590005); MTTN (590010); MTTD (590015); MTTC (590020); MTTE (590025); MTTQ (590030); MTTG (590035); MTTH (590040); MTTI (590045); MTTL1 (590050); MTTL2 (590055); MTTK (590060); MTTM (590065); MTTF (590070); MTTP (590075); MTTS1 (590080); MTTS2 (590085); MTTT (590090); MTTW (590095); MTTY (590100); MTTV (590105); MTND1 (516000); MTND2 (516001); MTND3 (516002); MTND4 (516003); MTND4L (516004); MTND5 (516005); MTND6 (516006); MYCYB (516020); MTCO1 (516030); MTCO2 (516040); MTCO3 (516050); MTATP6 (516060); MTATP8 (516070).	Lavender Top (EDTA)	MNG Laboratories (www.medicalneurogenetics.com)/Dynacare	Mitochondrial DNA Deletion Syndromes Mitochondrial DNA Depletion Syndrome Mitochondrial DNA Depletion Syndrome Mitochondrial DNA Depletion Syndrome, Encephalomyopathic Form Mitochondrial DNA Depletion Syndrome, Encephalomyopathic Form Mitochondrial DNA Depletion Syndrome, Hepatocerebral Form Mitochondrial DNA Depletion Syndrome, MNGIE Form Mitochondrial DNA Depletion Syndrome, Myopathic Form mtDNA Deletion Syndromes	NextGen Sequencing		9-May-20
Mitochondrial Genome Sequencing + Deletion Analysis [MOL189]	518	MTRNR1 (561000), MTRNR2 (561010); MTTA (590000); MTTR (590005); MTTN (590010); MTTD (590015); MTTC (590020); MTTE (590025); MTTQ (590030); MTTG (590035); MTTH (590040); MTTI (590045); MTTL1 (590050); MTTL2 (590055); MTTK (590060); MTTM (590065); MTTF (590070); MTTP (590075); MTTS1 (590080); MTTS2 (590085); MTTT (590090); MTTW (590095); MTTY (590100); MTTV (590105); MTND1 (516000); MTND2 (516001); MTND3 (516002); MTND4 (516003); MTND4L (516004); MTND5 (516005); MTND6 (516006); MYCYB (516020); MTCO1 (516030); MTCO2 (516040); MTCO3 (516050); MTATP6 (516060); MTATP8 (516070).	Blood | Fibroblasts | Muscle | Extracted DNA | Buccal Cells	MNG Laboratories (www.medicalneurogenetics.com)/Dynacare	Leber hereditary optic neuropathy (LHON); Neuropath, ataxia, and retinitis pigmentosa (NARP); Myoclonic epilepsy associated with ragged-red fibers (MERRF); Mitochondrial myopathy, encephalopathy, lactic acidosis, and stroke-like episodes (MELAS)	Deletion; Capillary DNA Sequencing		14-May-20

Appelation approuvée de l'analyse ou maladie associée [avec numéro de phénotype MIM] / Approved test name or Associated Disease [with MIM Phenotype Number]	**Code Test** / Test Code **(Dictionnaire MSSS)**	**SYMBOL HGNC DU GÈNE et NOM OMIM DU GÈNE** / HGNC Gene Symbol and OMIM Gene Number	**RÈGLES DE MANUTENTION /** Handling Conditions	**LABORATOIRE DÉSIGNÉ** / Designated laboratory	**RENSEIGNEMENTS SUR L'ANALYSE** / Test details	**MÉTHODE /** Methode	**NOTE PARTICULIÈRE /** Special comment	**Date de la révision /** Date of revision
Mitochondrial Genome Sequencing + Deletion Analysis + Depletion Testing (Leukocyte) [MOL232]	518	The human mitochondrial genome is 16569 base pairs in length and encodes 37 genes including 2 ribosomal RNA genes, 22 transfer RNA genes, and 13 protein-coding genes. Our mitochondrial DNA (mtDNA) panel includes complete sequencing of the following genes (MIM#): MTRNR1 (561000), MTRNR2 (561010); MTTA (590000); MTTR (590005); MTTN (590010); MTTD (590015); MTTC (590020); MTTE (590025); MTTQ (590030); MTTG (590035); MTTH (590040); MTTI (590045); MTTL1 (590050); MTTL2 (590055); MTTK (590060); MTTM (590065); MTTF (590070); MTTP (590075); MTTS1 (590080); MTTS2 (590085); MTTT (590090); MTTW (590095); MTTY (590100); MTTV (590105); MTND1 (516000); MTND2 (516001); MTND3 (516002); MTND4 (516003); MTND4L (516004); MTND5 (516005); MTND6 (516006); MYCYB (516020); MTCO1 (516030); MTCO2 (516040); MTCO3 (516050); MTATP6 (516060); MTATP8 (516070).	Lavender Top (EDTA)	MNG Laboratories (www.medicalneurogenetics.com)/Dynacare	Mitochondrial diseases including: Leber hereditary optic neuropathy (LHON); Neuropath, ataxia, and retinitis pigmentosa (NARP); Myoclonic epilepsy associated with ragged-red fibers (MERRF); Mitochondrial myopathy, encephalopathy, lactic acidosis, and stroke-like episodes (MELAS)	Deletion; Real-time Quantitative PCR analysis; Sequencing		14-May-20
Mitochondrial Genome Sequencing + Deletion Analysis + Depletion Testing (Muscle) [MOL340]	518	The human mitochondrial genome is 16569 base pairs in length and encodes 37 genes including 2 ribosomal RNA genes, 22 transfer RNA genes, and 13 protein-coding genes. Our mitochondrial DNA (mtDNA) panel includes complete sequencing of the following genes (MIM#): MTRNR1 (561000), MTRNR2 (561010); MTTA (590000); MTTR (590005); MTTN (590010); MTTD (590015); MTTC (590020); MTTE (590025); MTTQ (590030); MTTG (590035); MTTH (590040); MTTI (590045); MTTL1 (590050); MTTL2 (590055); MTTK (590060); MTTM (590065); MTTF (590070); MTTP (590075); MTTS1 (590080); MTTS2 (590085); MTTT (590090); MTTW (590095); MTTY (590100); MTTV (590105); MTND1 (516000); MTND2 (516001); MTND3 (516002); MTND4 (516003); MTND4L (516004); MTND5 (516005); MTND6 (516006); MYCYB (516020); MTCO1 (516030); MTCO2 (516040); MTCO3 (516050); MTATP6 (516060); MTATP8 (516070).	50-75 milligrams muscle snap frozen in liquid nitrogen and maintained at -80°Celsius or below. (2) DNA extracted from muscle	MNG Laboratories (www.medicalneurogenetics.com)/Dynacare	Mitochondrial diseases including: Leber hereditary optic neuropathy (LHON) Neuropath, ataxia, and retinitis pigmentosa (NARP) Myoclonic epilepsy associated with ragged-red fibers (MERRF) Mitochondrial myopathy, encephalopathy, lactic acidosis, and stroke-like episodes (MELAS)	Polymerase Chain Reaction (PCR) followed by DNA sequencing analysis; Polymerase Chain Reaction (PCR) followed by Restriction Endonuclease digestion and gel separation; Real-time Quantitative PCR analysis		14-May-20
MLPA (screen for deletions of CFHR1-CFHR3)		CFHR1 (134371); CFHR3 (605336)	Lavender Top (EDTA)	Molecular Otolaryngology & Renal Research Laboratory, U. of Iowa Carver College of Medicine (www.medicine.uiowa.edu/morl)	Hemolytic uremic syndrome, atypical, susceptibility to (235400)	MLPA		31-Oct-17
MODY Neonatal Diabetes NGS Panel	481	ABCC8, AKT2, BLK, CEL, CISD2, CP, EIF2AK3, FOXP3, GATA6, GCK, GLIS3, GLUD1, HADH, HNF1A, HNF1B, HNF4A, IER3IP1, INS, INSR, KCNJ11, KLF11, NEUROD1, NEUROG3, PAX4, PDX1, PTF1A, RFX6, SLC2A2, WFS1, ZFP57 (30 genes)	Lavender Top (2 x 4 mL)	Fulgent Genetics (fulgentdiagnostics.com)		NextGen Sequencing		13-May-19
Mold Mix 1	1465		Serum (Gold SST or Red Top)	In-Common Laboratories				12-Mar-20
Mold Mix 2	1465		Serum (Gold SST or Red Top)	In-Common Laboratories				12-Mar-20
Molecular Testing for Lissencephaly		DCX (300121)	Lavender Top (EDTA)	University of Chicago Genetic Services Laboratories (dnatesting.uchicago.edu)	Lissencephaly, X-linked (300067);Subcortical laminal heteropia, X-link (300067); testing for a known mutation	Sanger sequencing		
Monogenic Autoimmunity Panel [08150]	817	ACP5 ADA2 ADAR AICDA AIRE AP3B1 BLOC1S6 BTKCASP10 CASP8 CD27 CD40LG CR2 CTLA4 CYBA CYBBDOCK8 FADD FAS FASLG FOXP3 ICOS IFIH1 IL10IL10RA IL10RB IL21 IL21R IL2RA ITCH ITK LRBALYST MAGT1 NCF2 NCF4 NFAT5 NFKB2 NFKBIA ORAI1PIK3CD PIK3R1 PLCG2 PNP PRF1 PRKCD RAB27A RAC2RFX5 RFXANK RFXAP RMRP RNASEH2A RNASEH2B RNASEH2C SAMHD1SH2D1A SLC7A7 STAT1 STAT3 STAT5B STIM1 STX11 STXBP2TBX1 TMEM173 TNFRSF13B TNFRSF13C TNFSF12 TPP2 TREX1 UNC13DUNG WAS XIAP	Lavender Top (EDTA) [2 tubes]	Invitae (www.invitae.com)		NextGen Sequencing		20-Feb-20

Appelation approuvée de l'analyse ou maladie associée [avec numéro de phénotype MIM] / Approved test name or Associated Disease [with MIM Phenotype Number]	**Code Test** / Test Code **(Dictionnaire MSSS)**	**SYMBOL HGNC DU GÈNE et NOM OMIM DU GÈNE** / HGNC Gene Symbol and OMIM Gene Number	**RÈGLES DE MANUTENTION /** Handling Conditions	**LABORATOIRE DÉSIGNÉ** / Designated laboratory	**RENSEIGNEMENTS SUR L'ANALYSE** / Test details	**MÉTHODE /** Methode	**NOTE PARTICULIÈRE /** Special comment	**Date de la révision /** Date of revision
Monogenic Inflammatory Bowel Disease Panel (Invitae) [08122]	1151	ADA, ADAM17, AICDA, BTK, CD3G, CD40LG, CTLA4, CYBA, CYBB, DCLRE1C, DKC1, DOCK8, FOXP3, G6PC3, ICOS, IL10, IL10RA, IL10RB, IL21, IL2RA, IL2RG, ITGB2, LIG4, LRBA, MEFV, MVK, NCF2, NCF4, NFAT5, NLRC4, PIK3CD, PIK3R1, PLCG2, RAG1, RAG2, RTEL1, SH2D1A, SLC37A4, STAT1, STAT3, STIM1, STXBP2, TTC7A, WAS, XIAP, ZAP70	Lavender Top (EDTA)	Invitae (www.invitae.com)	XIAP X-linked lymphoproliferative syndrome 2; ZAP70 Severe combined immunodeficiency, Omenn syndrome; WAS Wiskott–Aldrich syndrome; TTC7A Gastrointestinal defects and immunodeficiency (GIDID) syndrome; STXBP2 Familial hemophagocytic lymphohistiocytosis type 5; STIM1 STIM1 deficiency; STAT3 IPEX-like syndrome; STAT1 IPEX-like syndrome; SLC37A4 Glycogen storage disease type Ib; SH2D1A X-linked lymphoproliferative syndrome 1; RTEL1 Dyskeratosis congenita; RAG2 Severe combined immunodeficiency, Omenn syndrome; RAG1 Severe combined immunodeficiency, Omenn syndrome; PLCG2 Familial cold autoinflammatory syndrome; PIK3R1 Agammaglobulinemia; PIK3CD Activated PI3K-delta; PIK3CD Activated PI3K-delta; NLRC4 NLRC4 defect; NFAT5 NFAT5 haploinsufficiency; NCF4 Chronic granulomatous disease (CGD); NCF2 Chronic granulomatous disease (CGD); NCF2 Chronic granulomatous disease (CGD); MVK Mevalonate kinase deficiency; MEFV Familial Mediterranean fever; LRBA Common variable immunodeficiency (CVID); LIG4 LIG4 syndrome; ITGB2 Leukocyte adhesion deficiency 1; IL2RG Severe combined immunodeficiency, Omenn syndrome; denosine deaminase deficiency; ADAM17 ADAM17 deficiency; AICDA Hyper IgM syndrom; BTK Agammaglobulinemia; CD3G Severe combined immunodeficiency, Omenn syndrome; CD40LG Hyper IgM	Next-Generation sequencing		7-Oct-18
MOSAIC-6 Autoimmune Encephalitis Panel	1417	NMDAR, $GABA_B$, DPPX, LGI, CASPR, AMPAR	Serum or CSF. Store at -20°C. (CSF only NMDA as Ab is made by plasma cells in CSF).	BC Neuroimmunolgy (bcneuro.ca)				31-Oct-20
Muscle Specific Tyrosine Kinase Antibodies (MuSK Ab) [P91022]	43		Gold SST. Centrifuge and transfer serum to plastic container. Store at -20°C. Transport frozen.	BC Neuroimmunology (bcneuro.ca)	myasthenia gravis			31-Oct-20
Myelin Basic Protein [663]	1465		CSF. Store frozen. Specimen Stability : Room temperature: 7 days. Refrigerated: 14 days. Frozen: 21 days	Quest Laboratories	The pesence of myelin basic protein in the spinal fluid is supportive evidence for the diagnosis of multiple sclerosis and other demyelinating diseases, although it is a non-specific finding and present in other causes of damage to CNS myelin.	RIA		17-Sep-19
MYH9-RELATED DISORDERS VIA THE MYH9 GENE [11491]	839	MYH9 (160775)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	May-Hegglin Anomaly (155100); Epstein Syndrome (153650); Fechtner Syndrome (153640); Sebastian Syndrome (605249); Deafness, Autosomal Dominant 17 (603622); May-Hegglin Anomaly (155100); Epstein Syndrome (153650); Fechtner Syndrome (15364); Sebastian Syndrome (605249); Deafness, Autosomal Dominant 17 (603622)	Next Generation DNA Sequencing	NOT available at HMR	30-Sep-20
Myopathy-Rhabdomyolysis		ACAD9, ACADL, ACADM, ACADVL, AGL, C10ORF2, CPT1B, CPT2, GAA, GYS1, HADHA, HADHB, OPA1, OPA3, PFKM, PGAM2, PGM1, PHKA1, POLG, POLG2, RRM2B, SUCLA2, TK2, TYMP	Lavender Top (EDTA)	Fulgent Genetics (fulgentdiagnostics.com)		NextGen Sequencing		
Myotonia Congenita via the CLCN1 Gene [1179]	607	CLCN1 (118425)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Congenital Myotonia, Autosomal Dominant Form (160800), Myotonia Congenita Autosomal Recessive (255700)			11-Aug-20
Myotonic Dystrophy 2 (ZNF9/CNBP) Genetic Testing (Repeat Expansion) [MOL303]	607	CNBP (ZNF9) (116955)	Lavender Top (EDTA)	MNG Laboratories (www.medicalneurogenetics.com)/Dynacare		Repeat-primed PCR (QP-PCR)		25-Jun-20

Appelation approuvée de l'analyse ou maladie associée [avec numéro de phénotype MIM] / Approved test name or Associated Disease [with MIM Phenotype Number]	**Code Test** / Test Code **(Dictionnaire MSSS)**	**SYMBOL HGNC DU GÈNE et NOM OMIM DU GÈNE** / HGNC Gene Symbol and OMIM Gene Number	**RÈGLES DE MANUTENTION /** Handling Conditions	**LABORATOIRE DÉSIGNÉ** / Designated laboratory	**RENSEIGNEMENTS SUR L'ANALYSE** / Test details	**MÉTHODE /** Methode	**NOTE PARTICULIÈRE /** Special comment	**Date de la révision /** Date of revision
Neopterin/**Tetrahydrobiopterin** (CSF) [NC03]	1224	BH4, Neopterine	Collection tube requirement : Call laboratory to obtain appropriate sample collection containers. Each sample collection set consists of 5 numbered centrifuge tubes in a small plastic bag. Tube #3 contains antioxidants necessary to protect the sample from oxidation. One set of tubes is required per patient. The total CSF volume required is 3.5 milliliters. Collection of sample : CSF should be collected directly from the tap needle: Collect from the first drop in to the containers in numerical order. Fill each tube to the marked line (0.5 milliliters in tubes 1, 2, & 5 – 1.0 ml in tubes 3 & 4). Attach patient identifiers to each tube without covering the tube number. If there is no blood contamination, place the tubes into a biohazard bag and place on ice (or dry ice if available) at the bedside. Transfer the samples to a -80°C freezer ASAP. Ship on dry ice. If the sample is blood contaminated, the tubes should immediately be centrifuged (prior to freezing) and the clear CSF transferred to new similarly labeled tubes then frozen and stored at -80°C ASAP.	MNG Laboratories (www.medicalneurogenetics.com)/Dynacare	Dystonia, dopamine responsive | DRD | GCH1 | Dystonia, dopa responsive, due to sepiapterin reductase deficiency | SPR | Hyperphenylalaninemia, BH4 deficient, B | HPABH4 | Hyperphenylalaninemia, BH4 deficient, A | HPABHA | PTS | Hyperphenylalaninemia, BH4 deficient, C | QDPR | HPABH4C | Hyperphenylalaninemia, BH4 deficient, D | HPABH4D | PCBD1			9-May-20
Nephrotic Syndrome (NS)/Focal Segmental Glomerulosclerosis (FSGS) Sequencing Panel with CNV Detection [10417]	100	ACTN4 604638 ANLN 616027 ARHGAP24 610586 ARHGDIA 601925 CD2AP 604241 COL4A3 120070 COL4A4 120131 COL4A5 303630 COL4A6 303631 COQ2 609825 COQ6 614647 COQ8B 615567 CRB2 609720 CUBN 602997 DGKE 601440 EMP2 602334 FAT1 600976 INF2 610982 ITGA3 605025 ITGB4 147557 KANK1 607704 KANK2 614610 KANK4 614612 LAGE3 300060 LAMA5 601033 LAMB2 150325 LMX1B 602575 MAGI2 606382 MYO1E 601479 NPHS1 602716 NPHS2 604766 NUP107 607617 NUP205 614352 NUP93 614351 OSGEP 610107 PAX2 167409 PDSS2 610564 PLCE1 608414 PTPRO 600579 SCARB2 602257 SGPL1 603729 SMARCAL1 606622 TP53RK 608679 TPRKB 608680	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Alport Syndrome, Autosomal Dominant AD 104200 Alport Syndrome, Autosomal Recessive AR 203780 Alport Syndrome, X-Linked Recessive XL 301050 Cerebral Palsy, Spastic Quadriplegic, 2 AR 612900 Coenzyme Q10 Deficiency AR 607426 Coenzyme Q10 deficiency, primary, 3 AR 614652 Coenzyme Q10 deficiency, primary, 6 AR 614650 Deafness, X-linked 6 XL 300914 Epidermolysis Bullosa With Pyloric Atresia AR 226730 Epilepsy, Progressive Myoclonic 4, With Or Without Renal Failure AR 254900 Finnish Congenital Nephrotic Syndrome AR 256300 Focal Segmental Glomerulosclerosis 1 AD 603278 Focal Segmental Glomerulosclerosis 2 AD 603965 Focal Segmental Glomerulosclerosis 3, Susceptibility To AR, AD 607832 Focal Segmental Glomerulosclerosis 5 AD 613237 Focal Segmental Glomerulosclerosis 6 AR 614131 Focal Segmental Glomerulosclerosis 8 AD 616032 Focal Segmental Glomerulosclerosis 9 AR 616220 Galloway-Mowat Syndrome AR 251300 Galloway-Mowat Syndrome 2, X-linked XL 301006 Galloway-Mowat Syndrome 3 AR 617729 Galloway-Mowat Syndrome 4 AR 617730 Galloway-Mowat Syndrome 5 AR	Next Generation DNA Sequencing		5-May-21
Neurofibromatosis (NextGen Sequencing Panel and Copy Number Analysis; 21 Genes) [NGS335]	776	ATM (607585), BRAF (164757), CBL (165360), HRAS (190020), KRAS (190070), MAP2K1 (176872), NF1 (613113), NF2 (607379), NRAS (164790), PTEN (601728), PTPN11 (176876), RAF1 (164760), RIT1 (609591), SDHAF2 (613019), SDHB (185470), SDHC (602413), SDHD (602690), SH3BP2 (602104), SHOC2 (602775), SOS1 (182530), SPRED1 (609291).	Lavender Top (EDTA)	MNG Laboratories (www.medicalneurogenetics.com)/Dynacare		Next Generation DNA Sequencing		9-May-20
Neurofilament (pNf-H)	1465		CSF or Serum	Mitogen Advanced Diagnostics		ELISA		2-Jun-21
Neurofilament (pNf-L)	1465		CSF or Serum	Mitogen Advanced Diagnostics		SIMOA		2-Jun-21
Neurological Disease Profile (IgG+IgM)	32		Serum/Plasma only	Mitogen Advanced Diagnostics	Anti- GM1, GM2, GM3, GD1a, GD1b, GT1b, GQ1b	LIA		2-Jun-21
Neutropenia NGS Panel	836	AP3B1, CSF3R, CXCR4, ELANE, G6PC3, GATA1, GATA2, GFI1, HAX1, LAMTOR2, LYST, RAB27A, RAC2, SBDS, SLC37A4, TAZ, USB1, VPS13B, VPS45, WAS, WIPF1	Lavender Top (2 x 4 mL)	Fulgent Genetics (fulgentdiagnostics.com)	Cyclical neutropenia (162800), Severe congenital neutropenia autosomal dominant (202700), Severe congenital neutropenia X-linked (300299), Severe congenital neutropenia 4, autosomal recessive (612541), Severe congenital neutropenia 2, autosomal dominant (613107), Severe congenital neutropenia 3, autosomal recessive (610738)	NextGen Sequencing		28-Feb-20
Neutrophil Antibody, Flow Cytometry [1606]	1572		Serum. Red Top (only). Room temperature: 7 days Refrigerated: 14 days Frozen: 30 days	Quest Diagnostics				6-Jul-20

Appelation approuvée de l'analyse ou maladie associée [avec numéro de phénotype MIM] / Approved test name or Associated Disease [with MIM Phenotype Number]	**Code Test** / Test Code **(Dictionnaire MSSS)**	**SYMBOL HGNC DU GÈNE et NOM OMIM DU GÈNE** / HGNC Gene Symbol and OMIM Gene Number	**RÈGLES DE MANUTENTION /** Handling Conditions	**LABORATOIRE DÉSIGNÉ** / Designated laboratory	**RENSEIGNEMENTS SUR L'ANALYSE** / Test details	**MÉTHODE /** Methode	**NOTE PARTICULIÈRE /** Special comment	**Date de la révision /** Date of revision
Niemann-Pick Disease Type C Sequencing Panel [3425]	468	NPC1 (607623); NPC2 (601015)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Niemann-Pick Disease Type C1 (257220)	Next Generation DNA Sequencing		12-Nov-19
N-Methylhistamine, 24 Hour, Urine [NMH24]	117		Collection Instructions: Collect 24-hour urine with no preservatives. Total volume required. Record on both the specimen container and the request form. Urine Preservative Collection Options: Note: The addition of preservative or application of temperature controls must occur within 4 hours of completion of the collection: 6N HCl, 50% Acetic Acid, Na(2)CO(3), Toluene, 6N HNO(3), Boric Acid, Thymol. Store frozen. **Stable 14 days.**	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	mastocytosis	Liquid Chromatography-Tandem Mass Spectrometry (LC-MS/MS)		4-Mar-21
Noonan Syndrome (NextGen Sequencing Panel and Copy Number Analysis; 10 Genes) [NGS414]	778	BRAF KRAS LZTR1 MAP2K1 NRAS PTPN11 RAF1 RIT1 SOS1 SOS2	Lavender top (EDTA) [2 tubes]	MNG Laboratories (www.medicalneurogenetics.com)/Dynacare		NextGen Sequencing		9-May-20
NOTCH3-CADASIL (125310)	65	NOTCH3 (600276)	Lavender Top (EDTA)	London Laboratories Service Group		Sanger sequencing; 33 coding exons of NOTCH3+ 20nt intronic sequence around each exon	To change to NGS with exon copy number in Oct 2015	
Nuclear-Mito NGS Panel	518	AAAS AARS2 AASS ABAT ABCB6 ABCB7 ABCC8 ABCC9 ABCD1 ABCD3 ACACA ACACB ACAD8 ACAD9 ACADL ACADM ACADS ACADSB ACADVL ACAT1 ACAT2 ACHE ACLY ACO2 ACSF3 ACSL4 ACSL5 ACSM3 ADSL AFG3L2 AGK AGPS AGXT AGXT2 AIFM1 AK2 AKAP10 AKR7A2 AKT1 AKT2 ALAS2 ALDH18A1 ALDH2 ALDH3A2 ALDH4A1 ALDH5A1 ALDH6A1 ALDH7A1 AMACR AMT ANK2 ANKRD26 APTX ARMS2 AS3MT ASS1 ATIC ATP10D ATP5E ATP5SL ATP7B ATPBD1 ATPAF2 ATXN7 AUH BAX BCAT1 BCAT2 BCKDHA BCKDHB BCL2 BCS1L BOLA3 C10orf2 C12orf65 CACNA1A CACNA1S CACNA2D1 CASP8 CDC42BPB CDKL5 CFTR CHAT CHDH CHRNA4 CHRNB2 CISD2 CKM CLCN1 CLCN2 CLCN5 CLCN7 CLCNKB CLN3 CLN5 CLN6 CLN8 CLYBL CNR1 COA5 COMT COQ2 COQ4 COQ5 COQ6 COQ9 COX10 COX14 COX15 COX411 COX4I2 COX6B1 COX7A2 CPOX CPS1 CPT1A CPT1B CPT2 CTSD CYB5A CYB5R3 CYBA CYBB CYCS CYP11A1 CYP11B1 CYP11B2 CYP24A1 CYP27A1 CYP27B1 D2HGDH DARS2 DBT DDAH1 DDC DECR1 DGUOK DHODH DIABLO DISC1 DLAT DLD DMGDH DMPK DNAJC19 DNAJC5 DNM1L DTNBP1 EARS2 EC11 ECSIT ELAC2 ELN ENO1 ENO3 ETFA ETFB ETFDH ETHE1 FAAH FARS2 FASN FASTKD2 FBP1 FECH FH FOLR1 FOXC1 FOXG1 FOXRED1 FPGS FTH1 FXN G6PC G6PD GAD1 GAD2 GALC GARS GATM GCDH GCK GCSH GDAP1 GFER GFM1 GK GLDC GLO1 GLRA1 GLRX5 GLS GLUD1 GLYCTK GNAS GNPAT GPAM GPD1 GPD2 GPI GPX1 GPX4 GYS1 GYS2 H6PD HADH HADHA HADHB HARS HARS2 HCCS HIBCH HIGD2A HK1 HK2 HLCS HMGCL HMGCS2 HOGA1 HSD17B10 HSD17B4 HSD3B1 HSD3B2 HSPA9 HSPB7 HSPD1 HTRA2 HTT IDE IDH1 IDH2 IDH3B IMMP2L IMMT INPP5E INSR ISCU IVD KARS KCNA1 KCNE1 KCNE2 KCNH2 KCNJ11 KCNJ2 KCNQ1 KCNQ2 KCNQ3 KIF1B KRT5 KYNU L2HGDH LARS2 LDHA LDHB LETM1 LIAS LRPPRC LRRK2 MAOA MAOB MARS2 MAVS MCCC1 MCCC2 MCEE MDH1 MECP2 MED23 MEN1 MFN2 MFSD8 MGLL MGST3 MLYCD MMAA MMAB MMACHC MMADHC MOCOS MOCS1 MOCS2 MOGS MPV17 MRPL3 MRPL48 MRPS16 MRPS22 MRRF MTCH2 MTFMT MTHFD1 MTHFD1L MTHFS MTO1 MTPAP MTRR MUT MUTYH NAGS NARS2 NDUFA1 NDUFA10 NDUFA11 NDUFA12 NDUFA13 NDUFA2 NDUFA4 NDUFA6 NDUFA7 NDUFA8 NDUFA9 NDUFAF1 NDUFAF2 NDUFAF3 NDUFAF4 NDUFAF5 NDUFAF6 NDUFB1 NDUFB3 NDUFB6 NDUFB9 NDUFC2 NDUFS1 NDUFS2 NDUFS3 NDUFS4 NDUFS5 NDUFS6 NDUFS7 NDUFS8 NDUFV1 NDUFV2 NDUFV3 NFU1 NIPSNAP1 NIPSNAP3A NLRX1 NME1 NOS3 NPL NRXN1 NTHL1 NUBPL OAT OGG1 OPA1 OPA3 OTC OXCT1 PACRG PAH PAK5 PANK2 PARK2 PARL PARP1 PC PCCA PCCB PCK1 PCK2 PDHA1	Lavender Top (2 x 4 mL)	Fulgent Genetics (fulgentdiagnostics.com)		NextGen Sequencing		10-Jul-19
Oculopharyngeal Muscular Dystrophy	613	PABPN1 (602279)	Lavender Top	Molecular Genetics Laboratory - BC Children's Hospital & BC Women's Hospital		repeat sizing		25-Jun-20
Oncotype DX			FFPE	Genomic Health		gene expression		
Orexin-A/Hypocretin-1, Spinal fluid [ORXNA]	1691		CSF. Store frozen.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)		RIA		8-Dec-20
Osteogenesis imperfecta NGS panel - Dominant & Recessive [5102]	533	ALPL, BMP1, COL1A1, COL1A2, CREB3L1, CRTAP, FKBP10, IFITM5, LEPRE1, LRP5, PLOD2, PLS3, PPIB, SERPINF1, SERPINH1, SP7, TMEM38B, WNT1	Lavender Top (EDTA)	Connective Tissue Gene Test (www.ctgt.net)		NGS with del/dup analysis		25-Oct-19
Osteogenesis imperfecta NGS panel - Dominant [5099]	533	ALPL, COL1A1, COL1A2, IFITM5, PLS3	Lavender Top (EDTA)	Connective Tissue Gene Test (www.ctgt.net)		NGS with del/dup analysis		25-Oct-19
Osteogenesis imperfecta NGS panel - Recessive [5105]	533	ALPL, BMP1, CREB3L1, CRTAP, FKBP10, LEPRE1, LPR5, PLOD2, PPIB, SERPINF1, SERPINH1, SP7, TMEM38B, WNT1	Lavender Top (EDTA)	Connective Tissue Gene Test (www.ctgt.net)		NGS with del/dup analysis		25-Oct-19
Ovarian Dysgenesis via the FSHR gene [732]		FSHR (136435)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Ovarian dysgenesis 1 (233300)			
Overgrowth and Macrocephaly Syndromes Panel [04501]	968	AKT2 AKT3 CDKN1C CUL4B DIS3L2 DNMT3A EZH2 GLI3 GPC3 KPTN MED12 MTOR NF1 NFIX NPR2 NSD1 PHF6 PIK3R2 PTEN SETD2 SPRED1	Lavender top (EDTA) [2 tubes]	Invitae (www.invitae.com)				28-Feb-20

Appelation approuvée de l'analyse ou maladie associée [avec numéro de phénotype MIM] / Approved test name or Associated Disease [with MIM Phenotype Number]	**Code Test** / Test Code **(Dictionnaire MSSS)**	**SYMBOL HGNC DU GÈNE et NOM OMIM DU GÈNE** / HGNC Gene Symbol and OMIM Gene Number	**RÈGLES DE MANUTENTION /** Handling Conditions	**LABORATOIRE DÉSIGNÉ** / Designated laboratory	**RENSEIGNEMENTS SUR L'ANALYSE** / Test details	**MÉTHODE /** Methode	**NOTE PARTICULIÈRE /** Special comment	**Date de la révision /** Date of revision
Overgrowth and Macrocephaly Syndromes Panel [3449]	968	ABCC9 AKT1 AKT2 AKT3 ASPABRWD3 CCND2 CDKN1C CHD8 CUL4B DIS3L2 DNMT3A EED EZH2 GFAP GLI3 GPC3 HEPACAM HERC1 HUWE1 KIF7 KPTN MED12 MLC1 MPDZ MTOR NF1A NFIXNPR2 NSD1 OFD1 PDGFRB PHF6 PIK3CA PIK3R2 PPP2R5B PPP2R5C PPP2R5D PTCH1 PTEN RAB39B RNF125 RNF135 SETD2 STRADA SUZ12 TBC1D7 TMEM94	Lavender top (EDTA) [2 tubes]	Prevention Genetics (www.preventiongenetics.com)				28-Feb-20
Oxalate, Plasma [POXA1]	1176		Green Top; sample must be acidified	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)				20-Oct-20
Oxidative Phosphorylation (OXPHOS) Defects NGS Screening Panel (232 genes) + Del/Dup + mtDNA [NGS306]		AARS2 (612035); ABCB10 (605454); ABCB7 (300135); ABCB8 (605464); ABHD5 (604780); ACADS (606885); ADCK3 (606980); AFG3L2 (604581); AGK (610345); AIFM1 (300169); AK2 (103020); AK3 (609290); APTX (606350); ARMS2 (611313); ARX (300382); ATAD3A (612316); ATAD3B (612317); ATP5A1 (164360); ATP5B (102910); ATP5C1 (108729); ATP5D (603150); ATP5E (606153); ATP5F1 (603270); ATP5G1 (603192); ATP5G2 (603193); ATP5G3 (602736); ATP5H (); ATP5I (601519); ATP5J (603152); ATP5O (600828); ATP5S (); ATP7B (606882); ATPAF1 (608917); ATPAF2 (608918); BCS1L (603647); BOLA3 (613183); C10ORF2 (606075); C12orf65 (613541); C21orf33 (607962); CASA (114761); CARS2 (612800); CCT7 (605140); CHCHD3 (613748); CHCHD4 (611077); CISD2 (611507); CLPP (601119); COA5 (613920); COQ2 (609825); COQ3 (605196); COQ4 (612898); COQ6 (614647); COQ7 (601683); COQ9 (612837); COX10 (602125); COX11 (603648); COX14 (614478); COX15 (603646); COX17 (604813); COX18 (610428); COX19 (610429); COX4I1 (123864); COX4I2 (607976); COX5A (603773); COX5B (123866); COX6A1 (602072); COX6A2 (602009); COX6B1 (124089); COX6C (124090); COX7A1 (123995); COX7A2 (123996); COX7A2L (605771); COX7B (603792); COX7B2 (609811); COX7C (603774); COX8A (123870); CRLS1 (608188); CYC1 (123980); CYCS (123970); DARS2 (610956); DDX28 (607618); DGUOK (601465); DHRS1 (610410); DHTKD1 (614984); DNAJC19 (608977); DNM1L (603850); DUS2L (609707); DUT (601266); EARS2 (612799); ECSIT (608388); ETHE1 (608451); EXOG (604051); FARS2 (611592); FASTKD2 (612322); FDPS (134629); FIS1 (609003); FOXRED1 (613622); FTMT (608847); FXC1 (607388); FXN (606829); GARS (600287); GFER (600924); GFM1 (606639); GFM2 (606544); GRPEL1 (606173); GRSF1 (604851); GTPBP3 (608536); HACL1 (604300); HARS2 (600783); HCCS (300056); HOGA1 (613597); HSPA9 (600548); HSPD1 (118190); HSPE1 (600141); IARS2 (612801); ICT1 (603000); IDI1 (604055); IMMP2L (605977); IMMT (600378); IREB2 (147582); ISCA1 (611006); ISCU (611911); KARS (601421); KIF1B (605995); LACTB (608440); LARS2 (604544); LCLAT1 (614241); LDHD (607490); LETM1 (604407); LRPPRC (607544); LYRM4 (613311); MARS (156560); MARS2 (609728); MCU (614197); METAP1D (610267); MFN1 (608506); MFN2 (608507); MGME1 (615076); MIPEP (602241); MNF1 (614461); MPV17 (137960); MRPS3 (611997); MRPL1 (611821); MRPL10 (611825); MRPL11 (611826); MRPL12 (602375); MRPL13 (610200); MRPL14 (611827); MRPL15 (611828); MRPL16 (611829); MRPL17 (611830); MRPL18 (611831); MRPL19 (611832); MRPL2 (611822); MRPL20 (611833); MRPL21 (611834); MRPL22 (611835); MRPL24 (611836); MRPL27 (611837); MRPL28 (604853); MRPL3 (607118); MRPL30 (611838); MRPL32 (611839);	Lavender Top (EDTA)	MNG Laboratories (www.medicalneurogenetics.com)/Dynacare		NextGen Sequencing		9-May-20
Oxysterols, Plasma [OXNP]	1579		Lavender Top (EDTA). Store plasma frozen. Prolonged storage at RT can lead to autooxidation (FP).	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	Niemann-Pick types A, B, and C disease	Liquid Chromatography-Tandem Mass Spectrometry (LC-MS/MS)		17-May-18
Paliperidone (Invecta) [91895]			Red Top (only)	Quest Diagnostics Nichols Institute - California, Molecular Genetics Laboratory	9-hydroxyrespiridone	Liquid Chromatography-Tandem Mass Spectrometry (LC-MS/MS)		19-Feb-18
Pancreastatin			Collect 10 mL EDTA plasma in special tube containing the Z-tube and separate as soon as possible. Freeze plasma immediately after separation. Special Z-tube is available from Inter Science Institute (ISI). Minimum specimen size is 1 ml. Ship frozen.	InterScience Institute (www.interscienceinstitute.com)		radioimmunoassay		
Pancreatic Polypeptide, Plasma [HPP]	1284		Plasma EDTA, fasting, handle 4°C	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)				17-Mar-20
Paraglioma		SDHB (185470); SDHC (602413); SDHD (602690)	Lavender Top (EDTA)	Alberta Mol Dx Laboratory (Calgary)		del/dupl		
Paraglioma		SDHB (185470)	Lavender Top (EDTA)	Alberta Mol Dx Laboratory (Calgary)		Sanger sequencing		
Paraglioma		SDHC (602413)	Lavender Top (EDTA)	Alberta Mol Dx Laboratory (Calgary)		Sanger sequencing		
Paraglioma		SDHD (602690)	Lavender Top (EDTA)	Alberta Mol Dx Laboratory (Calgary)		Sanger sequencing		

Appelation approuvée de l'analyse ou maladie associée [avec numéro de phénotype MIM] / Approved test name or Associated Disease [with MIM Phenotype Number]	**Code Test** / Test Code **(Dictionnaire MSSS)**	**SYMBOL HGNC DU GÈNE et NOM OMIM DU GÈNE** / HGNC Gene Symbol and OMIM Gene Number	**RÈGLES DE MANUTENTION /** Handling Conditions	**LABORATOIRE DÉSIGNÉ** / Designated laboratory	**RENSEIGNEMENTS SUR L'ANALYSE** / Test details	**MÉTHODE /** Methode	**NOTE PARTICULIÈRE /** Special comment	**Date de la révision /** Date of revision
Paraneoplastic (Neuronal) Antibody Panel	61		Serum (Gold SST or Red Top). Store at -20°C	BC Neuroimmunology (bcneuro.ca)	Amphiphysin, CV2 (CRMP5), PNMA2 (Ma2/Ta), Ri, Yo, Hu, Recoverin, SOX1, Tintin, Zic4, GAD65, Tr (DNER)	Immunoblot. Confirmed by CBA fixed.	Alt @MUHC on serum: Paraneoplastic/Anti-Neuronal Ab (Anti:HU,RI,YO,CV2,PNMA2, Amphiphysin). These are biomarkers of cytotoxic T-cells for disease and not disease causing.	31-Oct-20
Paraneoplastic Autoantibody Evaluation, Serum [PAVAL]	61		Red Top (preferred); Gold SST. Store at 4°C (14 days) or frozen.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	*Requires specia l approval.*	RIA; IFA; EIA; Western blot; CBA		31-Oct-20
Parkinson-Alzheimer-Dementia NGS Panel	597	A2M, AAAS, ACE, APOE, APP, ATP13A2, ATP1A3, C9orf72, CSF1R, DCTN1, DNMT1, EIF4G1, FBXO7, GBA, GCH1, GRN, HTRA2, LRRK2, MAPT, MPO, PARK2, PARK7, PINK1, PLA2G6, POLG, PRKRA, PRNP, PSEN1, PSEN2, SLC6A3, SNCA, SNCB, TAF1, TH, TREM2, TYROBP, UCHL1, VPS35 (38 genes)	Lavender Top (EDTA)	Fulgent Genetics (fulgentdiagnostics.com)	Includes: Dystonia, DOPA-responsive (BRD) (128230) and C9orf72 repeat analysis	NextGen Sequencing		23-Nov-18
PARKINSON'S DISEASE, JUVENILE VIA THE PARK2 GENE [1027]	664	PARK2 (602544)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Parkinson disease, juvenile, type 2 (600116)	Sanger sequencing		
Parkinsons Disease/Parkinsonism [NGS357]	665		Lavender Top (EDTA)	MNG Laboratories (www.medicalneurogenetics.com)/Dynacare		Next Generation DNA Sequencing		9-May-20
Pendred Syndrome		SLC26A4	Lavender Top (EDTA)	Molecular Genetics Laboratory, Hospital for Sick Children (www.sickkids.ca/PaediatricLaboratoryMedicine/Laboratories-Services/Molecular-Genetics-Laboratory/index.html)				
Pepsinogen I	1748		3 ml serum or EDTA plasma should be collected and separated as soon as possible. Freeze specimens immediately after separation. Minimum specimen size is 1 ml.	InterScience Institute (www.interscienceinstitute.com)	Patient should be fasting 10 - 12 hours prior to collection of specimen. Antacids or other medications affecting stomach acidity or gastrointestinal motility should be discontinued, if possible, for at least 48 hours prior to collection.	RIA		
Pepsinogen I [FPEPS]	1748		Red Top or Gold SST or Lavender (EDTA). Separate immediately and freeze.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)			Sent to Interscience Institute. PSN-2 at ISI.	20-Oct-20
Pepsinogen II	1748		3 ml serum or EDTA plasma should be collected and separated as soon as possible. Freeze specimens immediately after separation. Minimum specimen size is 1 ml.	InterScience Institute (www.interscienceinstitute.com)	Patient should be fasting 10 - 12 hours prior to collection of specimen. Antacids or other medications affecting stomach acidity or gastrointestinal motility should be discontinued, if possible, for at least 48 hours prior to collection.	RIA		
Perforin		PRF1 (1780280)	Lavender Top (EDTA)	Hospital for Sick Children (Rapid Response Laboratory)	hemophagocytic lymphohistiocytosis (HLH), familial 2 (603553)	Sanger sequencing		
Perforin protein expression			Lavender Top (EDTA)	Hospital for Sick Children (Rapid Response Laboratory)	hemophagocytic lymphohistiocytosis (HLH) (603553)			
Perforin/Granzyme [HLH]	817		Heparin	Alberta Precision Laboratories (Flow Cytometry - Calgary)			To be accompanied by normal control sample	14-Oct-20
Periodic Fever/Autoinflammatory Disorders NGS Panel	838	AP1S3, CARD14, CECR1, ELANE, HAX1, IL10, IL10RA, IL10RB, IL1RN, IL36RN, LPIN2, MEFV, MVK, NLRC4, NLRP12, NLRP3, NLRP7, NOD2, PLCG2, PSMB8, PSTPIP1, RBCK1, SH3BP2, SLC29A3, TMEM173, TNFRSF11A, TNFRSF1A, NLRP1 (28 genes)	Lavender Top (2 x 4 mL)	Fulgent Genetics (fulgentdiagnostics.com)	Familial Mediterranean Fever (FMF) (249100 and 134610); TNF receptor-associated periodic syndrome (TRAPS (142680); Hyperimmunoglobulin D Syndrome (HIDS) (260920)	NextGen Sequencing	Add on: TNFAIP3	10-Aug-17
Phosphatidylserine Antibodies (IgG, IgA, IgM) [10062]			Preferred Specimen(s) 1 mL plasma collected in a 3.2% sodium citrate (light blue-top) tube Minimum Volume 0.5 mL Collection Instructions Platelet-poor plasma: Centrifuge light blue-top tube 15 minutes at approximately 1500 g within 60 minutes of collection. Using a plastic pipette, remove plasma, taking care to avoid the WBC/platelet buffy layer and place into a plastic vial. Centrifuge a second time and transfer platelet-poor plasma into a new plastic vial. Plasma must be free of platelets (<10,000/µL). Transport Container Plastic screw-cap vial Transport Temperature Room temperature Specimen Stability Room temperature: 7 days Refrigerated: 28 days Frozen: 28 days	Quest Diagnostics/Nichols Institute	thrombosis; pregnancy loss	immunoassay		

Appelation approuvée de l'analyse ou maladie associée [avec numéro de phénotype MIM] / Approved test name or Associated Disease [with MIM Phenotype Number]	**Code Test** / Test Code **(Dictionnaire MSSS)**	**SYMBOL HGNC DU GÈNE et NOM OMIM DU GÈNE** / HGNC Gene Symbol and OMIM Gene Number	**RÈGLES DE MANUTENTION /** Handling Conditions	**LABORATOIRE DÉSIGNÉ** / Designated laboratory	**RENSEIGNEMENTS SUR L'ANALYSE** / Test details	**MÉTHODE /** Methode	**NOTE PARTICULIÈRE /** Special comment	**Date de la révision /** Date of revision
Phosphoglycerate kinase 1 deficiency (300653)		PGK1 (311800)	Lavender Top (EDTA)	Centogene AG (www.centogene.com)		Sanger sequencing		
Phosphomannomutase (PMM) and Phosphomannose Isomerase (PMI), Leukocytes [PMMIL]			Yellow top (ACD solution B) or Yellow top (ACD solution A). DO NOT CONFUSE WITH STANDARD GOLD SST tube. Do not transfer contents. Store at 4°C. Send immediately.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)		Enzyme assay.		20-Oct-20
Phosphorylase b Kinase enzyme activity			Lavender Top (EDTA) 10 cc OR dried blood spot; other sample types available	Glycogen Storage Disease Laboratory. Duke University Hospital. http://pediatrics.duke.edu/divisions/medical-genetics/biochemical-genetics-laboratory	Glycogen Storage Disease, Type IX, Liver form (306000)	GSDIX phosphorylase b kinase enzyme assay on red blood cells; other enzyme assays available		18-Jul-17
PLP1-RELATED DISORDERS VIA THE PLP1 GENE [307]		PLP1 (300401)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Pelizaeus-Merzbacher disease (312080) and spastic paraplegia (312920)	Sanger sequencing/aCGH		
Polycystic Kidney Disease NGS Panel	795	PKD1 (601313); PKD2 (173910), PKHD1	Lavender Top (2 x 4 mL)	Fulgent Genetics (fulgentdiagnostics.com)	Polycystic kidney disease, adult type 1 (173900); Polycystic kidney disease 2 (613095)	NextGen Sequencing		4-Dec-18
Polymicrogyria Deletion/Duplication Panel (11 genes deletion/duplication analysis)		GPR56 (604110), KIAA1279 (609367), OCLN (602876), RTTN (610436), TUB1A (602529), TUBA8 (605742),TUBB2b (612850), TUBB3 (602661), RAP18 (602207), RAB3GAP1 (602536), RAB3GAP2 (609275)	Lavender Top (EDTA)	University of Chicago Genetic Services Laboratories	Bilateral frontoparietal polymicrogyria (606854), Goldberg-Shprintzen Magacolon Synddrome (609460), Band-like Calcification with Simplified glyration and polymicrogyria (251290), Polymicrogyria with seizures (614833), polymicrogyria with optic nerve hypoplasia (612180), asymmetric polymicrogyria (610031), Complex cortical dysplasia with other brain malformations (614039), Warburg Micro syndrome (600118)	oligonucleotide array-CGH		
Polymicrogyria Sequencing Panel (12 genes sequecning)		GPR56 (604110), KIAA1279 (609367), OCLN (602876), RTTN (610436), TUB1A (602529), TUBA8 (605742),TUBB2b (612850), TUBB3 (602661), RAP18 (602207), RAB3GAP1 (602536), RAB3GAP2 (609275), WDR62 (613583)	Lavender Top (EDTA)	University of Chicago Genetic Services Laboratories	Bilateral frontoparietal polymicrogyria (606854), Goldberg-Shprintzen Magacolon Synddrome (609460), Band-like Calcification with Simplified glyration and polymicrogyria (251290), Polymicrogyria with seizures (614833), polymicrogyria with optic nerve hypoplasia (612180), asymmetric polymicrogyria (610031), Complex cortical dysplasia with other brain malformations (614039), Warburg Micro syndrome (600118)	1. Next-gen Sequencing 2. Sanger sequencing (confirmation)		
Pontocerebellar Hypoplasia NGS Panel	886	CASK, OPHN1, RARS2, SEPSECS, TSEN2, TSEN34, TSEN54, VRK1	Lavender Top (EDTA)	Fulgent Genetics (fulgentdiagnostics.com)		NextGen Sequencing	EXOSC3 can be added at no cost	
PONTOCEREBELLAR HYPOPLASIA TYPE 1B VIA THE EXOSC3 GENE [1208]		EXOSC3 (606489)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Pontocerebellar hypoplasia, type 1B (614678)	Sanger sequencing	See Fulgent Panel	
Porencephaly 1 (175780)		COL4A1 (120130)	Lavender Top (EDTA)	Centogene AG (www.centogene.com)		1. single exon testing 2. full gene sequencing 3. deletion/duplication testing		
Porphobilinogen Deaminase (PBGD), Whole Blood [PBGD_]	1324		Green Top, 2 mL, 4°C only	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	Acute Intermittent Porphyria (176000)			

Appelation approuvée de l'analyse ou maladie associée [avec numéro de phénotype MIM] / Approved test name or Associated Disease [with MIM Phenotype Number]	**Code Test** / Test Code **(Dictionnaire MSSS)**	**SYMBOL HGNC DU GÈNE et NOM OMIM DU GÈNE** / HGNC Gene Symbol and OMIM Gene Number	**RÈGLES DE MANUTENTION** / Handling Conditions	**LABORATOIRE DÉSIGNÉ** / Designated laboratory	**RENSEIGNEMENTS SUR L'ANALYSE** / Test details	**MÉTHODE** / Methode	**NOTE PARTICULIÈRE** / Special comment	**Date de la révision** / Date of revision
Porphyria Disorders NGS Panel	1324	ALAD (125270), ALAS2 (301300) C15ORF41 (615626), CPOX (612732), FECH (612386), HFE (613609), HMBS (609806), PPOX (600923), SLC19A2 (603941), UROD (613521), UROS (606938).	Lavender Top (2 x 4 mL)	Fulgent Genetics (fulgentdiagnostics.com)		NextGen Sequencing		31-Aug-19
Porphyrins Evaluation, Whole Blood [PEE]	1324		Green Top (heparin), fasting, handle 4°C. Must arrive within 3 days of drawing. Alternate: washed erthyrocytes	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	erythropoietic protoporphyria (177000) and congenital erythropoietic porphyria (163700)			20-Oct-20
Porphyrins, Total, plasma [PTP]	1324		Green top (heparin); protect from light; transfer to amber vial	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	Porphyria Cutanea Tarda (176100)			20-Oct-20
Primary Ciliary Dyskinesia panel (Invitae) [04101]	771	CCDC65, DNAH11, RSPH4A, RPGR, CCDC40, ZMYND10, DNAAF1, DNAAF2, DNAAF3, DYX1C1, DNAI1, CCDC103, DNAI2, RSPH3, RSPH1, DNAAF5, RSPH9, OFD1, DRC1, CCDC39, LRRC6, SPAG1, CCDC151, MCIDAS, ARMC4, C21orf59, DNAH1, DNAH5, DNAH8, NME8, GAS8, CCNO, CCDC114, DNAL1	Lavender Top (EDTA) 2-5 cc	Invitae (https://www.invitae.com 475 Brannan St. Ste. 230) San Francisco, CA, 94107	Primary Ciliary Dyskinesia panel (244400)	NGS with del/dup analysis		7-Oct-18
PRIMARY FAMILIAL AND CONGENITAL POLYCYTHEMIA (PFCP) VIA THE EPOR GENE [1649]		EPOR (133171)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Familial Erythrocytosis, 1 (133100)	Sanger sequencing	NOT available at HMR	
Primary Immunodeficiency Panel (Invitae) [08100]	1042	ACD, ACP5, ACTB, ADA, ADA2, ADAM17, ADAR, AICDA, AIRE, AK2, AP3B1, ATM, B2M, BCL10, BLNK, BLOC1S6, BTK, CARD11, CARD14, CARD9, CASP10, CASP8, CD247, CD27, CD3D, CD3E, CD3G, CD40LG, CD79A, CD79B, CD8A, CEBPE, CHD7, CIITA, CLPB, COPA, CORO1A, CR2, CSF2RA, CSF3R, CTC1, CTLA4, CTPS1, CTSC, CXCR4, CYBA, CYBB, DCLRE1B, DCLRE1C, DKC1, DNMT3B, DOCK2, DOCK8, ELANE, EPG5, FADD, FAS, FASLG, FERMT3, FOXN1, FOXP3, FPR1, G6PC3, GATA2, GFI1, HAX1, ICOS, IFIH1, IFNGR1, IFNGR2, IGLL1, IKBKB, IL10, IL10RA, IL10RB, IL12B, IL12RB1, IL17F, IL17RA, IL17RC, IL1RN, IL21, IL21R, IL2RA, IL2RG, IL36RN, IL7R, IRAK4, IRF7, IRF8, ISG15, ITCH, ITGB2, ITK, JAGN1, JAK3, LAMTOR2, LCK, LIG4, LPIN2, LRBA, LYST, MAGT1, MALT1, MAP3K14, MEFV, MOGS, MVK, MYD88, NBN, NCF2, NCF4, NFAT5, NFKB2, NFKBIA, NHEJ1, NHP2, NLRC4, NLRP12, NLRP3, NOD2, NOP10, ORAI1, PARN, PGM3, PIK3CD, PIK3R1, PLCG2, PMS2, PNP, POLE, PRF1, PRKCD, PRKDC, PSMB8, PSTPIP1, PTPRC, RAB27A, RAC2, RAG1, RAG2, RBCK1, RFX5, RFXANK, RFXAP, RHOH, RMRP, RNASEH2A, RNASEH2B, RNASEH2C, RORC, RTEL1, SAMHD1, SEMA3E, SH2D1A, SH3BP2, SLC29A3, SLC35C1, SLC37A4, SLC7A7, SMARCAL1, SP110, SPINK5, STAT1, STAT2, STAT3, STAT5B, STIM1, STK4, STX11, STXBP2, TAP1, TAP2, TAPBP, TAZ, TBK1, TCN2, TERC, TERT, TFNSF12, TICAM1, TINF2, TLR3, TMC6, TMC8, TMEM173, TNFRSF13B, TNFRSF13C, TNFRSF1A, TNFRSF4, TPP2, TRAF3, TRAF3IP2, TREX1, TRNT1, TTC7A, TYK2, UNC13D, UNC93B1, UNG, VPS13B, VPS45, WAS, WIPF1, XIAP, ZAP70, ZBTB24	Lavender Top (2 x 4 mL)	Invitae (https://www.invitae.com 475 Brannan St. Ste. 230) San Francisco, CA, 94107		NGS with del/dup analysis		7-Oct-18
Procainamide and N-acetylprocainamide, Serum [PROCG]			Gold SST	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)		immunoassay		21-Feb-18
Procollagen I Intact N-Terminal, Serum [PINP]	178		Red Top or Gold SST. Send serum. Stable frozen for 14 days.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	An aid in monitoring antiresorptive and anabolic therapy in patients with osteoporosis; An adjunct in the assessment of conditions associated with increased bone turnover such as Paget disease	competitive radioimmunoassay		21-Feb-17
PROGRESSIVE BULBAR PALSY WITH OR WITHOUT SENSORINEURAL DEAFNESS [BROWN-VIALETTO-VAN LAERE SYNDROME (211530) AND FAZIO-LONDE DISEASE (211500)] VIA THE SLC52A3 (C20ORF54) [417]		SLC52A3 (613350)	Lavender top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Not available: SLC52A2 (607882) and SLC52A1 (607883)	Sanger sequencing		
Progressive Familial Intrahepatic Cholestasis Type 1 (211600)		ATP8B1 (602397)	Lavender Top (EDTA)	Cinncinatti Children's Hospital (Division of Human Genetics Diagnostic Laboratories)		Sanger sequencing		
Proinsulin, Plasmat [PINS]	179		Draw blood in a ice-cooled purple-top EDTA. (Plasma gel tube is not acceptable.) Chill on ice for 10 minutes. Spin down and send 1 mL of EDTA plasma frozen.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)		Quantitative Chemiluminescent Immunoassay		4-Mar-21
Proportionate short stature/small for gestational age [8760]		ATRX, BLM, BTK, CREBBP, CUL7, DHCR7, EP300, ERCC6, ERCC8, FGD1, GH1, GHR, GHRHR, GLI2, HESX1, IGF1, IGF1R, INSR, KDM6A, KMT2D, KRAS, LHX3, NBN, NIPBL, PITX2, POU1F1, PROP1, PTPN11, RAF1, ROR2, RPS6KA3, SHOX, SMARCAL1, SMC1A, SMC3, SOS1, SOX2, SOX3, SRCAP, STAT5B, TBCE, THRB, TRIM37, WRN	Infants (<2 years): 2-3 mL Children (>2 years): 3-5 mL Older Children & Adults: 5-10 mL	https://www.allelediagnostics.com/		Performed using Next Generation Sequencing, Methylation-specific MLPA (MS-MLPA), and DNA methylation specific PCR.	Alternate: Blueprint Short Stature Syndrome Panel [MA2101]. Does not include methylation study.	2-Dec-17
Psychotropic Pharmcogenomics Gene Panel, Varies [PSYGP]	1582		Lavender Top (EDTA)	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)				4-Mar-21
PTEN Hamartoma Tumor Syndrome via the PTEN Gene [707]		PTEN (601728)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Bannayan-Riley-Ruvalcaba Syndrome (153480); Cowden Disease (158350); Cutaneous Malignant Melanoma 1 (155600); Macrocephaly/Autism Syndrome (605309); Vacterl Association With Hydrocephalus (276950)	Sanger sequencing		

Appelation approuvée de l'analyse ou maladie associée [avec numéro de phénotype MIM] / Approved test name or Associated Disease [with MIM Phenotype Number]	**Code Test** / Test Code **(Dictionnaire MSSS)**	**SYMBOL HGNC DU GÈNE et NOM OMIM DU GÈNE** / HGNC Gene Symbol and OMIM Gene Number	**RÈGLES DE MANUTENTION /** Handling Conditions	**LABORATOIRE DÉSIGNÉ** / Designated laboratory	**RENSEIGNEMENTS SUR L'ANALYSE** / Test details	**MÉTHODE /** Methode	**NOTE PARTICULIÈRE /** Special comment	**Date de la révision /** Date of revision
Pulmonary Hypertension NGS Panel		ACVRL1 (601284), BMPR2 (600799), CAV1 (601047), ENG (131195), SMAD9 (603295)	Lavender Top (2 x 4 mL)	Fulgent Genetics (fulgentdiagnostics.com)	Telangiectasia, hereditary hemorrhagic, type 2 (600376); Pulmonary hypertension, familial primary, 1, with or without HHT/Pulmonary hypertension, primary, fenfluramine or dexfenfluramine-associated (178600); Pulmonary hypertension, primary, 3 (615343); Telangiectasia, hereditary hemorrhagic, type 1 (187300); Pulmonary hypertension, primary, 2 (615342)	NextGen Sequencing	Alt: CTGT PPH 4 genes & del/dupl	
Pyridoxal 5'-phosphate (CSF) [NC05]			Collect 1 milliliter of CSF. Spin sample if contaminated with red blood cells and freeze the clear CSF at -80°Celsius. Store frozen at -80°Celsius.	MNG Laboratories (www.medicalneurogenetics.com)/Dynacare	Pyridoxamine 5-prime-phosphate oxidase deficiency |			9-May-20
RAPSYN-related disorders via the RAPSYN gene [466]		RAPSN (601592)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Congenital myasthenic syndromes (608931); Fetal Akinesia Deformation Sequence (208150)	Sanger sequencing		
Recombx Zic4 Antibody Test [127]			Red Top	Athena Diagnostics (www.athenadiagnostics.com)	paraneoplastic syndrome			
Renal Tubular Acidosis NGS Panel	806	ATP6V0A4, ATP6V1B1, CA2, SLC4A1, SLC4A4	Lavender Top (EDTA) [2 tubes]	Fulgent Genetics (fulgentdiagnostics.com)				28-Feb-20
Resistance to Thyroid Hormone (RTH) Mutation Analysis [16053(X)]		THRB (190160)	Lavender Top (EDTA)	Quest Diagnostics Nichols Institute - California, Molecular Genetics Laboratory	Thyroid hormone resistance (188570)	Sanger sequencing		
RETINOBLASTOMA VIA THE RB1 GENE [795]		RB1 (614041)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Retinoblastoma (180200)	1. Sanger sequencing 2. aCGH [600]		
Retinol Binding Protein			Red-top tube(s) or a serum gel tube or urine	Quest Diagnostics/Nichols Institute				
Rifampin/Ethambutol [NTM9]	58		Isolate	National Jewish Health				9-Nov-17
RNA Polymerase III Antibodies, IgG, Serum [RNAP]	1078		Serum (Gold SST or Red Top)	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	systemic sclerosis	ELISA		4-Jul-19
Russel Silver Syndrome: Methylation and Copy Number Analysis	992		Lavender Top (EDTA)	Genome Diagnostics The Department of Paediatric Laboratory Medicine The Hospital for Sick Children	Russel-Silver syndrome (180860)	Methylation-Specific-MLPA of imprinting center 1 (H19)		19-Feb-20
Russel Silver Syndrome: UPD7 Analysis	992		Lavender Top (EDTA)	Genome Diagnostics The Department of Paediatric Laboratory Medicine The Hospital for Sick Children	Russel-Silver syndrome (180860)	UPD7 studies via STR (short tandem repeat) analysis		19-Feb-20
SACS Single Gene testing		SACS (604490)	Lavender Top (EDTA)	Molecular Genetics Laboratory, Hospital for Sick Children (www.sickkids.ca/PaediatricLaboratoryMedicine/Laboratories-Services/Molecular-Genetics-Laboratory/index.html)	Spastic ataxia, Charlevoix-Saguenay type (270550)	NextGen Sequencing		21-Aug-19
Saethre-Chotzen Syndrome (TWIST seq & select exons in FGFR3)		TWIST1 (601622); FGFR3 (134934)	Lavender Top (EDTA)	Molecular Genetics Laboratory, Hospital for Sick Children (www.sickkids.ca/PaediatricLaboratoryMedicine/Laboratories-Services/Molecular-Genetics-Laboratory/index.html)	Craniosynostosis: Saethre-Chotzen syndrome (101400)	Sanger sequencing: TWIST1; FGFR3 (p.Pro250Arg)		
SANDHOFF DISEASE VIA THE HEXB GENE [476]		HEXB (606873)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Sandhoff Disease (268800)	Sanger sequencing		
SAP Protein Expression [XLP1]	816	SH2D1A (300490)	Heparin	Alberta Precision Laboratories (Flow Cytometry - Calgary)	X-linked lymphoproliferative syndrome (XLP1) (308240)		To be accompanied by normal control sample	14-Oct-20
sC5b-9 (MAC) [7304502]	188		0.5mL of EDTA plasma, separated and frozen within 2 hours of collection; ship on dry ice	Cincinnati Children's Hospital Medical Center 3333 Burnet Avenue NRB 1042 Cincinnati, OH 45229 (www.cincinnatichildrens.org)	complement-mediated renal diseases , C5b-9			14-Jan-19
SCA3/Machado Joseph disease (109150)		ATNX3 (607047)	Lavender Top (EDTA)	North York General	SCA Panel available			
Scleroderma/Systemic Sclerosis Profile	1179	Anti-CENP A + B, Topo-I/Scl-70, RNA polymerase III, fibrillarin, Th/To, Ku, PDGFR, Ro52/TRIM21, PM/Scl-75, PM/Scl-100, NOR90/hUBF	Gold SST	Mitogen Advanced Diagnostics		LIA		2-Jun-21
SCN4A Full Gene sequencing Analysis [MOL356]; Paramyotonia Congenita (168300)		SCN4A (603967)	Lavender Top (EDTA)	MNG Laboratories (www.medicalneurogenetics.com)/Dynacare	Paramyotonia Congenita (168300)	Sanger sequencing		9-May-20
Secretin			EDTA plasma containing the G.I. Preservative Freeze immediately	InterScience Institute				
Selenoprotein N, 1 via the SEPN1 gene [330]		SEPN1 (606210)		Prevention Genetics (www.preventiongenetics.com)	Muscular dystrophy, rigid spine, 1 (602771)	Sanger sequencing		
Serum Amyloid (SAA)	1123		Serum samples (blood collection on a red or yellow tube, centrifuged and decanted). Frozen at -20°C on dry ice	Dynacare Next			May be free if specifiy medication	

Appelation approuvée de l'analyse ou maladie associée [avec numéro de phénotype MIM] / Approved test name or Associated Disease [with MIM Phenotype Number]	**Code Test** / Test Code **(Dictionnaire MSSS)**	**SYMBOL HGNC DU GÈNE et NOM OMIM DU GÈNE** / HGNC Gene Symbol and OMIM Gene Number	**RÈGLES DE MANUTENTION /** Handling Conditions	**LABORATOIRE DÉSIGNÉ** / Designated laboratory	**RENSEIGNEMENTS SUR L'ANALYSE** / Test details	**MÉTHODE /** Methode	**NOTE PARTICULIÈRE /** Special comment	**Date de la révision /** Date of revision
Short stature Homeobox (SHOX)-related Haploinsufficiency Disorder via the SHOX Gene [626]		SHOX	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Langer Mesomelic Dysplasia Syndrome (249700); Leri Weill Dyschondrosteosis (127300); Short Stature, Idiopathic, X-Linked (300582)	Sanger sequencing		28-Sep-17
Single Drug MIC [NTM3]		NTM	Isolate	National Jewish Health	RIF, EMB, CIP, MXF, AMK, LZD, CLR, CLF, RFB, STR, ETH, LVX, AZM OFX, KAN, CSI			9-Nov-17
Single Gene Repeat Expansion Analysis RFC1	1395	RFC1	Lavender Top (EDTA)	U. of Chicago Genetic Services Laboratory	CANVAS			24-Mar-21
SLC12A3 DNA Sequencing Test [766]		SLC12A3 (600968)	Lavender Top (EDTA)	Athena Diagnostics (www.athenadiagnostics.com)	Gitelman syndrome (9263800)	Sanger sequencing		
SLC27A4 Gene Sequencing [712]		SLC27A4 (604194)	Lavender Top (EDTA)	GeneDx (www.genedx.com)	ichthyosis prematurity syndrome (608649(	Sanger sequencing		9-Dec-19
SLC2A1 Full Gene Sequencing + MLPA Duplication/Deletion Analysis [MOL231] (GLUT1 deficiency syndrome 1)		SLC2A1 (138140)	Lavender Top (EDTA)	MNG Laboratories (www.medicalneurogenetics.com)/Dynacare	GLUT1 deficiency syndrome 1 (GLUT1DS1); Dystonia 9 (DYT9); GLUT1 deficiency syndrome 2 (GLUT1DS2); Epilepsy, idiopathic generalized, susceptibility to, 12 (EIG1)	Polymerase Chain Reaction (PCR) followed by DNA sequencing analysis; Multiplex Ligation-dependent Probe Amplification (MLPA) analysis	Offered for free by MNG (The testing costs is billed directly to Ultragenyx Pharmaceutical who is developping a new treatment)	9-May-20
SLC40A1-Related Hereditary Hemochromatosis (606069)		SLC40A1 (604653)	Lavender Top (EDTA)	Centogene AG (www.centogene.com)	ferroportin; Hereditary Hemochromatosis type IV (606069)	1. single exon testing 2. full gene sequencing 3. deletion/duplication testing		
SLC4A1 Gene Analysis		SLC4A1 (109270)	Lavender Top (EDTA)	Laboratory for Molecular Diagnostics: Center for Nephrology and Metabolic Disorders	Renal tubular acidosis, distal, AD (179800); Renal tubular acidosis (267200)	Sanger sequencing		
Soluble CD 163			Lavender Top (EDTA)	Hospital for Sick Children (Rapid Response Laboratory)	hemophagocytic lymphohistiocytosis (HLH)			
Soluble IL-2 Receptor (CD25)			Lavender Top (EDTA)	Hospital for Sick Children (Rapid Response Laboratory)	hemophagocytic lymphohistiocytosis (HLH)		See also HSJ "Cellules T regulatrices"/Change to ARUP	
Somatostatin (Somatotropin Release-Inhibiting Factor, SRIF)	193		EDTA plasma containing the G.I. Preservative ($30). Freeze immediately.	InterScience Institute		Also at Mayo Clin Lab		13-Sep-18
SOTOS SYNDROME VIA THE NSD1 GENE [132]		NSD1 (606681)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Sotos' Syndrome (117550)	Sanger sequencing		
Spastic Paraplegia (NextGen Sequencing Panel and Copy Number Analysis; 380 Genes + mtDNA) [NGS337]	657	ABCD1 (300371), ACOX1 (609751), ACTB (102630), AFG3L2 (604581), ALDH18A1 (138250), ALS2 (606352), AMPD2 (102771), AP4B1 (607245), AP4E1 (607244), AP4M1 (602296), AP4S1 (607243), KIAA0415 (613653), ARSA (607574), ARX (300382), ASNS (615574), ATL1 (606439), ATXN2 (601517), AUH (600529), B3GALT6 (615291), B4GALNT1 (601873), BCOR (300485), BSCL2 (606158), C12orf65 (613541), C19orf12 (614298), CCDC88C (611204), CCT5 (610150), CLPP (601119), COASY (609855), CPT1C (608846), CTNNB1 (116806), CYP2U1 (610670), CYP7B1 (603711), DARS (603084), DCTN1 (601143), DDHD1 (614603), DDHD2 (615003), DYNC1H1 (600112), EARS2 (612799), ELOVL4 (605512), ERLIN2 (611605), FA2H (611026), FARS2 (611592), FBXO7 (605648), FLNA (300017), FUCA1 (612280), GAD1 (605363), GAN (605379), GBA (606463), GBA2 (609471), GBE1 (607839), GCDH (608801), GJC2 (608803), GLB1 (611458), GM2A (613109), GSS (601002), HARS2 (600783), HSD17B4 (601860), HSPD1 (118190), IBA57 (615316), IFIH1 (606951), KCNQ2 (602235), KDM5C (314690), KIAA0196 (610657), KIF1A (601255), KIF2A (602591), KIF5A (602821), L1CAM (308840), L2HGDH (609584), LAMB1 (150240), LARS2 (604544), LIAS (607031), LIPT1 (610284), LYRM7 (615831), MARS2 (609728), MCOLN1 (605248), MOCS1 (603707), MOCS2 (603708), MRE11A (NA), MRPS22 (605810), mtDNA (NA), MTPAP (613669), NIPA1 (608145), NT5C2 (600417), OPA3 (606580), PAFAH1B1 (601545), PDHX (608769), PHGDH (606879), PLA2G6 (603604), PLP1 (300401), PNPLA6 (603197), PSAP (176801), PSEN1 (104311), REEP1 (609139), REEP2 (609347), RNASEH2B (610326), RTN2 (603183), SACS (604490), SCN2A (182390), SEPSECS (613009), SLC16A2 (300095), SLC19A3 (606152), SLC2A1 (138140), SLC30A10 (611146), SLC33A1 (603690), SOD1 (147450), SOX10 (602229), SPAST (604277), SPG11 (610844), SPG20 (607111), SPG21 (608181), SPG7 (602783), SPTAN1 (182810), STXBP1 (602926), TAF1 (313650), TARDBP (605078), TBC1D20 (611663), TFG (602498), TREX1 (606609), TTC19 (613814), TTR (176300), TUBA1A (602529), TUBG1 (191135), UBQLN2 (300264), UCHL1 (191342), VAMP1 (185880), VPS37A (609927), VPS53 (615850), WDR45 (300526), WDR62 (613583), ZFYVE26 (612012), ZFYVE27 (610243).	Lavender Top (EDTA)	MNG Laboratories (www.medicalneurogenetics.com)/Dynacare	= paraparesis	NextGen Sequencing		9-May-20

Appelation approuvée de l'analyse ou maladie associée [avec numéro de phénotype MIM] / Approved test name or Associated Disease [with MIM Phenotype Number]	**Code Test** / Test Code **(Dictionnaire MSSS)**	**SYMBOL HGNC DU GÈNE et NOM OMIM DU GÈNE** / HGNC Gene Symbol and OMIM Gene Number	**RÈGLES DE MANUTENTION** / Handling Conditions	**LABORATOIRE DÉSIGNÉ** / Designated laboratory	**RENSEIGNEMENTS SUR L'ANALYSE** / Test details	**MÉTHODE** / Methode	**NOTE PARTICULIÈRE** / Special comment	**Date de la révision** / Date of revision
Spastic paraplegia 3A, autosomal dominant (182600)	657	ATL1 (606439)	Lavender Top (EDTA)	Centogene AG (www.centogene.com)		1. single exon testing 2. full gene sequencing 3. deletion/duplication testing		
Spermatogenic Failure-6 (102530)		PGF6 (SPATA16) (609856)	Lavender Top (EDTA)	Centogene AG (www.centogene.com)		1. single exon testing 2. full gene sequencing 3. deletion/duplication testing		
Spermatogenic Failure-9 (613958)		DPY19L2 (613893)	Lavender Top (EDTA)	Centogene AG (www.centogene.com)		1. single exon testing 2. full gene sequencing 3. deletion/duplication testing		
Spinal and Bulbar Muscular Atrophy: AR Trinucleotide Repeat Analysis	958	AR (313700)	Lavender Top (EDTA)	Molecular Genetics Laboratory, Hospital for Sick Children (www.sickkids.ca/PaediatricLaboratoryMedicine/Laboratories-Services/Molecular-Genetics-Laboratory/index.html)	Spinal and bulbar muscular atrophy of Kennedy	AR exon 1 trinucleotide (CAG) repeat analysis		25-Feb-19
Spinal Muscular Atrophy (NextGen Sequencing Panel and Copy Number Analysis; 21 Genes) [NGS347]	958	AR (313700), ASAH1 (613468), ATP7A (300011), BICD2 (609797), DNAJB2 (604139), DYNC1H1 (600112), HSPB1 (602195), HSPB3 (604624), HSPB8 (608014), IGHMBP2 (600502), PLEKHG5 (611101), SIGMAR1 (601978), GPR172A (607882), SLC5A7 (608761), SMN1 (600354), SMN2 (601627), TBCD (604649), TRPV4 (605427), UBA1 (314370), VAPB (605704), VRK1 (602168)	Lavender Top (EDTA)	MNG Laboratories (www.medicalneurogenetics.com)/Dynacare		NextGen Sequencing		9-May-20
Spinal Muscular Atrophy: SMN1 and SMN2 deletion/Duplication Analysis	958	SMN1 (600354); SMN2 (601627)	Lavender Top (EDTA)	Molecular Genetics Laboratory, Hospital for Sick Children (www.sickkids.ca/PaediatricLaboratoryMedicine/Laboratories-Services/Molecular-Genetics-Laboratory/index.html)		Deletion /duplication via MLPA		
Spinocerebellar Ataxia (SCA) Panel (1,2,3,6,7,8,17)	584		Lavender Top (EDTA)	North York General	individuals tests $378	Sanger sequencing	no del/dupl; See also MNG's MOL380	14-May-18
Spinocerebellar ataxia 1 (601556)	584	ATXN1 (601556)	Lavender Top (EDTA)	North York General	SCA Panel available	Sanger sequencing		5-Aug-20
Spinocerebellar ataxia 11 (604432)	584	TTBK2 (611695)	Lavender Top (EDTA)	Centogene AG (www.centogene.com)		1. single exon testing 2. full gene sequencing 3. deletion/duplication testing		
Spinocerebellar ataxia 17 (607136)	584	TBP (600075)	Lavender Top (EDTA)	North York General	SCA Panel available	Sanger sequencing		
Spinocerebellar ataxia 2 (183090)	584	ATXN2 (601517)	Lavender Top (EDTA)	North York General	SCA Panel available	Sanger sequencing		5-Aug-20
Spinocerebellar ataxia 6 (183086)	584	CACNA1A (601011)	Lavender Top (EDTA)	North York General	SCA Panel available	triplet expansion		
Spinocerebellar ataxia 7 (607640)	584	ATXN7 (607640)	Lavender Top (EDTA)	North York General	SCA Panel available	Sanger sequencing		
Spinocerebellar ataxia 8 (608768)	584	ATXN8 (613289)	Lavender Top (EDTA)	North York General	SCA Panel available	Sanger sequencing		
Spinocerebellar Ataxia Type 2 via the ATXN2 CAG Repeat Expansion [12976]	584	ATXN2 (601517)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)		FFLA		5-Jan-21
Spinocerebellar ataxia type 27 (609307)	584	FGF14 (601515)	Lavender Top (EDTA)	Centogene AG (www.centogene.com)		1. single exon testing 2. full gene sequencing 3. deletion/duplication testing		
Spinocerebellar ataxia, autosomal recessive-8 (French Canadian mutation Sanger sequencing panel) [247]	584	SYNE1 (608441)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Autosomal recessive cerebellar ataxia 1 (ARCA1; OMIM 610743)	Sanger sequencing		
SPINOCEREBELLAR ATAXIA, AUTOSOMAL RECESSIVE-8 VIA THE SYNE1 GENE EXONS 2-146 [246]	584	SYNE1 (608441)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Spinocerebellar Ataxia, Autosomal Recessive 8 (610743); Emery-Dreifuss Muscular Dystrophy 4, Autosomal Dominant (612998)	Sanger sequencing		
STAT Epilepsy Panel [814]	356	ALDH7A1, ARX, BRAT1, CDKL5, FOLR1, GLDC, KCNQ2, KCNQ3, KCNT1, MECP2, MEF2C, PCDH19, PNPO, POLG, SCN1A, SCN1B, SCN2A, SCN8A, SLC19A3, SLC2A1, SLC6A8, SPTAN1, STXBP1, TPP1, TSC1, TSC2	Lavender Top (EDTA) [2 tubes]	GeneDx (www.genedx.com)		NextGen Sequencing		28-Feb-20
Stickler syndrome NGS panel [5127]	567	COL2A1, COL9A1, COL9A2, COL9A3, COL11A1, COL11A2, VCAN	Lavender Top (EDTA)	Connective Tissue Gene Test (www.ctgt.net)		NextGen Sequencing		
Surfactant NGS Panel		ABCA3 (601615), NKX2-1 (600635), SFTPB (178640), SFTPC (178620)	Lavender Top (EDTA)	Fulgent Genetics (fulgentdiagnostics.com)	Surfactant metabolism dysfunction, pulmonary, 2 (610913)			
Syntaxin 11		STX11 (605014)	Lavender Top (EDTA)	Hospital for Sick Children (Rapid Response Laboratory)	hemophagocytic lymphohistiocytosis (HLH) (603552)	Sanger sequencing		
T3 (Triiodothyronine), Reverse, Serum [RT3]	1649		Red Top (preferred); Gold SST. Store at 4°C (7 days) or frozen.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)		LC-MS/MS		4-Mar-21

Appelation approuvée de l'analyse ou maladie associée [avec numéro de phénotype MIM] / Approved test name or Associated Disease [with MIM Phenotype Number]	**Code Test** / Test Code **(Dictionnaire MSSS)**	**SYMBOL HGNC DU GÈNE et NOM OMIM DU GÈNE** / HGNC Gene Symbol and OMIM Gene Number	**RÈGLES DE MANUTENTION /** Handling Conditions	**LABORATOIRE DÉSIGNÉ** / Designated laboratory	**RENSEIGNEMENTS SUR L'ANALYSE** / Test details	**MÉTHODE /** Methode	**NOTE PARTICULIÈRE /** Special comment	**Date de la révision /** Date of revision
T3 (Triiodothyronine), Total, Serum [T3]			Gold SST	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)				
T4 (Thyroxine), Free, Serum [FRT4]			Red top	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)				
TAY-SACHS DISEASE VIA THE HEXA GENE [475]		HEXA (606869)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Tay-Sachs Disease (272800)	Sanger sequencing		
Telomere Length Measurement	1166		Lavender Top (EDTA) - 10 mL. Store at RT. Sample OK for 2 days only - same day send out.	Repeat Diagnostics (www.repeatdiagnostics.com)		Flow FISH		25-Jan-19
Thiamin (Vitamin B1), Whole Blood [TDP]	224		Fast overnight. Whole Blood (EDTA). Protect from light. Store at -20°C.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	Measures TPP as this provides stores.	LC-MS/MS		4-Mar-21
Thiosulfate, Serum/Plasma [4472SP]	92		**Red Top (only)**. Promplty separate into screw capped vial. No other tube type accepted. Serum stable for 24 days at 4°C or -20°C.	NMS Labs		LC-MS/MS		6-May-19
Thiosulfate, Urine [4472U]	920		Plastic container (no preservatives). **Store only at 4°C.** Stable for 30 days. Rejected if frozen.	NMS Labs		LC-MS/MS		6-May-19
Thrombocytopenia NGS Panel	1076	ADAMTS13, ANKRD26, CYCS, ETV6, GATA1, GP1BA, GP1BB, GP9, ITGA2B, ITGB3, MASTL, MPL, MYH9, RUNX1, WAS	Lavender Top (EDTA)	Fulgent Genetics (fulgentdiagnostics.com)				24-Oct-18
ThyGenX	1680	Point mutations - KRAS, HRAS, NRAS, BRAF, PIK3CA Rearrangements/translocations – RET/PTC1,3, PAX8, PPARgamma	Fine needle aspirate	Groupe TMTC (https://grouptmtc.com)	TERT available at no added cost		ON HOLD	21-Jan-21
ThyGenX/ThyraMIR (reflex)	1680	Point mutations - KRAS, HRAS, NRAS, BRAF, PIK3CA Rearrangements/translocations – RET/PTC1,3, PAX8, PPARgamma	Fine needle aspirate	Groupe TMTC (https://grouptmtc.com)	TERT available at no added cost		ON HOLD	21-Jan-21
ThyroSeq v.3	1680		Fine needle aspirate	Groupe TMTC (https://grouptmtc.com)		NextGen Sequencing		21-Jan-21
Thyrotropin Releasing hormone	1572		Thyrotropin Releasing Hormone must be collected with the TRH Preservative tune ($30). Ship specimens frozen in dry ice.	InterScience Institute (www.interscienceinstitute.com)		RIA		1-Feb-18
Tissue Transglutaminase Abs (IgA & IgG) assay [TTIGAG]			Serum (Gold SST or Red Top)	Hospitals-In-Common		CLIA		16-Mar-17
Titanium, Plasma	202		Royal Blue EDTA only. No gel tubes. Separate within30 min. Transfer plasma to plastic container.	Hospitals-In-Common				28-Jul-20
TMA Functional Panel (serologies for TTP, aHUS, HUS)	1306		2 ml frozen serum (RED TOP ONLY) and 2ml forzen EDTA plasma. Samples are stable indefinitely at -80°C.	Molecular Otolaryngology & Renal Research Laboratory, U. of Iowa Carver College of Medicine (www.medicine.uiowa.edu/morl)	complement-mediated renal diseases (FH autoantibody, hemolytic assay, CH50eq, APFA, FH, FI, C3, C4, FB, Bb, sC5b-9)			13-Jan-20
TMA Profile aHUS/TTP [7215217] (ADAMTS13 Activiy; C3; C4; Factor B; Factor I; Factor H; Factor H Autoantibody)	1306			Cinncinatti Children's Hospital (Diagnostic Immunology Laboratories)				January 14, 209
TP63-RELATED DISORDERS VIA THE TP63 GENE [834]		TP63 (603273)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	ADULT Syndrome (103285); Limb-Mammary Syndrome (603543); Hay-Wells Syndrome (106260); Rapp-Hodgkin Ectodermal Dysplasia Syndrome (129400);Split-Hand/Foot Malformation 4 (605289); Ectrodactyly, Ectodermal Dysplasia, And Cleft Lip/Palate Syndrome 3 (604292)	Sanger sequencing		
TPSAB1 Copy Number Analysis	1395		Lavender Top (EDTA)	Gene by Gene		ddPCR		11-Jul-18
Transthyretin amyloidosis (105210)	642	TTR (176300)	Lavender Top (EDTA)	Molecular GeneticsLlaboratory - BC Children's Hospital & BC Women's Hospital				12-Feb-20
Trimethylamine (TMA) and TMA N-oxide (TMAO),Urine (quantitative) [L6949]	1468		Pre-load: Morning void urine. Freeze immediately. Post-load (5 g choleine): collect urine 12-h after lead. Freeze immediately.	Denver Genetics Laboratories (www.DenverGenetics.org)	trimethylaminuria	MS/MS	price per sample	14-Jan-19
Trypsinogen [TRGEN]			Serum. Store and send frozen	In-Common Laboratories	pancreatic dysfunction (e.g. CF)	RIA		
Tuberous Sclerosis Complex Panel [10661]	783	TSC1 (605284); TSC2 (191092)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Tuberous sclerosis-1 (191100); Tuberous sclerosis-2 (613254)	NextGen Sequencing + del/dupl (MLPA)		5-Feb-20
TYPE IV VOLTAGE-GATED SODIUM CHANNEL (ALPHA SUBUNIT)-RELATED DISORDERS VIA THE SCN4A GENE [11645]		SCN4A (603967)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Congenital Myasthenic Syndrome, Acetazolamide-Responsive (614198); Potassium Aggravated Myotonia (608390); Paramyotonia Congenita Of Von Eulenburg (168300); Hypokalemic Periodic Paralysis, Type 2 (613345); Hyperkalemic Periodic Paralysis; HYPP (170500)	Replaces Sanger sequencing [416]		5-Feb-20
Type VI Collagenopathy via the COL6A1 gene [11197]		COL6A1 (120220)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Ullrich congenital muscular dystrophy (254090); Bethlem myopathy (158810)	Replaces Sanger sequencing [487]		5-Feb-20
Type VI-Related Collagenopathy Panel [10183]		COL12A1, COL6A1 (120220), COL6A2 (120240), COL6A3 (120250)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Ullrich congenital muscular dystrophy (254090) and Bethlem myopathy (158810)	NextGen Sequencing + del/dupl		5-Feb-20
UBE3A Full Gene Sequencing Analysis [MOL093]		UBE3A (601623)	Lavender Top (EDTA)	MNG Laboratories (www.medicalneurogenetics.com)/Dynacare	Angelman Syndrome (105830)	Sanger sequencing	Requires local methylation study	14-May-20

OPTILAB Montréal-CUSM

LISTE DES ANALYSES HORS-QUEBEC
List for testing done out of Québec
LABORATOIRE CLINIQUES CUSM
MUHC CLINICAL LABS

Centre universitaire de santé McGill — McGill University Health Centre

Appelation approuvée de l'analyse ou maladie associée [avec numéro de phénotype MIM] / Approved test name or Associated Disease [with MIM Phenotype Number]	**Code Test** / Test Code **(Dictionnaire MSSS)**	**SYMBOL HGNC DU GÈNE et NOM OMIM DU GÈNE** / HGNC Gene Symbol and OMIM Gene Number	**RÈGLES DE MANUTENTION /** Handling Conditions	**LABORATOIRE DÉSIGNÉ** / Designated laboratory	**RENSEIGNEMENTS SUR L'ANALYSE** / Test details	**MÉTHODE /** Methode	**NOTE PARTICULIÈRE /** Special comment	**Date de la révision /** Date of revision
UDP-Glucuronosyl Transferase 1A1 (UGT1A1), Full Gene Sequencing, Hyperbilirubinemia [UGTFG]	1340	UGT1A1 (191740)	Lavender Top (EDTA)	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	diagnosis of Gilbert (143500) or Crigler-Najjar syndromes (218800 and 606785); irinotecan & others sensitivity	Sanger sequencing		Dec. 23, 2018
Unknown alpha-chain Hemoglobin Variants	820	HBA1 (141800)/HBA2 (141850)	Lavender Top (EDTA)	McMaster University Medical Centre, Molecular Genetics Laboratory	also availble at CHUM			13-Nov-19
Unknown beta-chain Hemoglobin Variants	820	HBB (141900)	Lavender Top (EDTA)	McMaster University Medical Centre, Molecular Genetics Laboratory	also availble at MUHC			13-Nov-19
Uroporphyrinogen Decarboxylase (UPG D), Whole Blood [UPGD]			Green top, 2 mL, 4°C only	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	Porphyria Cutanea Tarda (176100)			
VALOSIN-CONTAINING PROTEIN-RELATED DISORDERS VIA THE VCP GENE [4807]		VCP (601023)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Inclusion body myopathy with early-onset Paget disease and frontotemporal dementia (167320)	NextGen Sequencing		2-May-07
Vascular Endothelial Growth Factor (VEGF), Plasma [VEGF]	213		Lavender Top (EDTA); Draw blood in a lavender-top (EDTA) tube(s). Spin down and send 1 mL of EDTA plasma frozen in a plastic vial.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)		ELISA		4-Mar-21
Vascular Endothelial Growth Factor D (VEGF-D)	213		Gold SST. Centrifuge and transfer serum to plastic container. Store at -20°C. Transport frozen.	Cincinatti Children's Hospital (Translational Trials Development and Support Laboratory (TTDSL))	LAM, TSC	ELISA		24-Feb-21
Vasoactive Intestinal Polypeptide (VIP)	211		10 ml EDTA plasma containing the **G.I. Preservative** should be collected and separated as soon as possible. Freeze plasma immediately after separation.	InterScience Institute (www.interscienceinstitute.com)	VIPomas, hepatic cirrhosis, and the Vemer-Morrison's (Watery Diarrhea) Syndrome	radioimmunoassay	If paired to another test going to ISI	9-Nov-17
VGKC antibodies	54	LGI1 and Caspr2 (Voltage gated potassium chanel)		BC Neuroimmunology (bcneuro.ca)				5-Jan-21
Vitamin B3 Niacin in Plasma [FNIAC]	218		Plasma EDTA. Store frozen.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)		LC-MS/MS		16-Apr-18
Vitamin B6	219		Plasma (EDTA or Li heparin). Separate and freeze within 1 hour. Protect specimen from light. Specimen must be labelled inside and outside light-protecting wrap. Gel-separator tubes not acceptable. Store and send frozen. If the specimen thaws, it is unsuitable for analysis	In-Common Laboratories	pyridoxine		Stopped doing at CHUM	19-Mar-20
Vitamin K1, serum [VITK1]	1464		Red top; Collection Instructions: Fasting-overnight (12-14 hours) (infants-draw prior to next feeding). Store at 4°C or frozen (14 d). Send frozen.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)		LC-MS/MS		12-Jun-20
Voltage Gated Calcium Channel Antibodies (VGCC) [P91861]	53		Gold SST. Centrifuge and transfer serum to plastic container. Store at -20°C. Transport frozen.	BC Neuroimmunology (bcneuro.ca)	Lambert-Eaton Myasthenic Syndrome (LEMS)	RIPA		31-Oct-20
Von Hippel-Lindau Disease via VHL Gene [7523]	261	VHL (608537)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)		NGS		31-Oct-20
von Willebrand disease (193400)		VWF (613160)	Lavender Top (EDTA)	ARUP Laboratories		Sequence analysis of select exons		
Warburg Micro Syndrome (600118)		RAB3GAP2 (609275)	Lavender Top (EDTA)	University of Chicago Genetic Services Laboratories	Also available: 1. Warburg Micro syndrome Sequencing panel (RAB3GAP1, RAB3GAP2 and RAB18 sequencing) 2. Warburg Micro syndrome Deletion/Duplication panel (RAB3GAP1, RAB3GAP2 and RAB18 del/dup)	Sanger sequencing		
Weaver Syndrome (277590)		EZH2 (601573)	Lavender Top (2 x 4 mL)	Fulgent Genetics (fulgentdiagnostics.com)		NextGen Sequencing + del/dupl		
WILSON DISEASE / HEPATOLENTICULAR DEGENERATION VIA THE ATP7B GENE [7871]	1185	ATP7B (606882)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Wilson Disease (277900)	NextGen Sequencing		31-Oct-20
Wolfram syndrome (222300)		WFS1 (606201)	Lavender Top (EDTA)	GeneDx		Part of NGS panel: Combined Mito Genome Plus Mito 140 Nuclear Gene Panel [615]		9-Dec-19
XIAP Protein Expression [XLP2]	817		Heparin	Alberta Precision Laboratories (Flow Cytometry - Calgary)	X-linked lymphoproliferative syndrome (XLP1) (308240)		To be accompanied by normal control sample	14-Oct-20
X-linked Adrenoleukodystrophy (300100)	849	ABCD1 (300371)	Lavender Top (EDTA)	EGL Genetics Laboratory 2460 Mountain Industrial, Tucker GA 30084 (geneticslab.emory.edu)		Sanger sequencing and del/dupl		
X-Linked Complete Congenital Stationary Night Blindness (CSNB1) via the NYX Gene [8705]		NYX (300278)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Congenital Stationary night blindness, X-Linked, type 1A (310500)	NextGen Sequencing		31-Oct-20
Zinc Transporter 8 (ZnT8) Antibody, Serum [EZNT8]	1819		SST gel tube 3 cc	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)				31-Oct-20