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

| **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 | **NOTE PARTICULIÈRE /** Special comment | **Date de la révision /** Date of revision |
|---|---|---|---|---|---|---|---|
| 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 | May also order Leukotriene E4 | 24-May-19 |
| 2,3-Dinor-11Beta-Prostaglandin F2 Alpha, Urine [23BPG] | 1 | | Preferred: 24-hour urine collection<br>Container/Tube: Plastic, 5-mL tube (T465)<br>Specimen Volume: 4 mL<br>Collection Instructions:<br>1. Collect urine for 24 hours.<br>2. No preservative preferred. | Mayo Clinical Laboratories (www.mayomedicallaboratories.com) | mast cell activation disorders including systemic mastocytosis; Replaces 11 BETA-PROSTAGLANDIN F2 ALPHA | May also order Leukotriene E4 | 24-May-19 |
| 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 | | 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 | | 06-Sep-18 |
| ABCC6 Gene Analysis in Pseudoxanthoma Elasticum [2642] | | ABCC6 (603234) | Lavender top (EDTA) | GeneDx (www.genedx.com) | Pseudoxanthoma Elasticum (264800) | | 09-Dec-19 |
| Acetaminophen [ACETA] | | | Serum | In-Common Laboratories | quantitation; to determine clearance | | 06-Sep-18 |
| Acetylcholine Receptor Antibodies by RIPA (AchR Ab, 91020/91021 only) | 1465 | | Serum (Gold SST) or CSF. Store at -20°C. | BC Neuroimmunology (bcneuro.ca) | myasthenia gravis | | 31-Oct-20 |
| Acetylcholine Receptor Antibodies with reflex to Muscle Specific Tyrosine Kinase Antibodies (MuSK Ab) | 1465 | | Serum (Gold SST) or CSF. Store at -20°C. | BC Neuroimmunology (bcneuro.ca) | myasthenia gravis | | 31-Oct-20 |
| Acyclovir, Serum/Plasma [FACY] | | | Red Top or Lavender Top (NOT SST) | Mayo Clinical Laboratories (www.mayomedicallaboratories.com) | | | |
| 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 | NGS Panel available | 14-May-20 |
| Adenosine Deaminase, Pleural Fluid [FADPL] | 11 | | Must be frozen within 24 hours of collection. No freeze/thaw cycles. | Mayo Clinical Laboratories (www.mayomedicallaboratories.com) | | | 08-Aug-17 |
| 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) | | 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) | Alernate site : MML | 03-Aug-18 |
| Adrenal hyperplasia due to 21-hydroyxlase deficiency (201910) | | CYP21A2 (613815) | Lavender Top (EDTA) | Alberta Mol Dx Laboratory (Calgary) | | | 26-Sep-18 |
| 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<br>◦ X-Linked Agammaglobulinemia<br>◦ SH2D1A-Related Lymphoproliferative Disease, X-Linked<br>◦ Agammaglobulinemia 2, Autosomal Recessive<br>◦ Agammaglobulinemia 3, Autosomal Recessive<br>◦ Agammaglobulinemia 4, Autosomal Recessive<br>◦ Agammaglobulinemia 5, Autosomal Dominant<br>◦ Agammaglobulinemia 6, Autosomal Recessive<br>◦ Agammaglobulinemia 7, Autosomal Recessive | | |

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| AGXT Gene, Full Gene Analysis [AGXMS] | | AGXT (604285) | Lavender Top (EDTA) | Mayo Clinical Laboratories (www.mayomedicallaboratories.com) | Hyperoxaluria, primary, type 1 (259900) | | |
| aHUS Genetic Susceptibility Panel [7409177] | 672 | NGS: C3, CFB, CFH, CFHR5, CFI, MCP, THBD; Sanger: DGKE; MLPA: CFHR1-CFHR3 | Lavender Top (EDTA) | Cinncinatti Children's Hospital (Division of Human Genetics Diagnostic Laboratories) | | new price | 14-Jan-19 |
| 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 | | 14-May-20 |
| 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 | 06-Sep-18 |
| Albendazole | 1917 | | Serum AND EDTA Plasma | Inselspital Bern<br>Freiburgstrasse 10<br>Zentrum für Labormedizin<br>Zentrale Annahme INOF / z. Hd. Y. Aebi<br>CH-3010 Bern, Switzerland | Measures albendazole sulfoxide due to 1st pass effcets | | 17-Aug-17 |
| Albright's hereditary Osteodystrophy (103580) | 440 | GNAS (139320) | Lavender Top (EDTA) 2 x 4 mL | Fulgent Genetics (fulgentdiagnostics.com) | Albright's hereditary Osteodystrophy (103580) | | 28-Feb-20 |
| Alexander Disease via the GFAP Gene [3775] | | GFAP (137780) | Lavender Top (EDTA) | Prevention Genetics (www.preventiongenetics.com) | Alexander disease (203450) | | 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) | | 14-May-20 |
| Alpha aminoadipic semialdehyde (Whole Blood) [AASA] | 12 | Alpha-aminoadipic semialdehyde | Specimen: Whole Blood<br>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.<br>http://seattlechildrenslab.testcatalog.org/ | Pyridoxine-dependent epilepsy (266100) | | 27-Jan-20 |
| ALPHA-1-ANTITRYPSIN DEFICIENCY (613490) | | SERPINA1 (104400) | Lavender Top (EDTA) | Attn: Norine Freedman or Lynn Coleman<br>Special Chemistry Laboratory<br>St Paul's Hospital<br>1081 Burrard Street<br>Vancouver, B.C. V6Z 1Y6. Contact Dr. A. Mattman BEFORE sending sample. | ALPHA-1-ANTITRYPSIN DEFICIENCY (613490) | | |
| 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) | 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 | | |
| Alport Syndrome NGS Panel | 785 | COL4A3, COL4A4, COL4A5 COL4A^ | Lavender Top (EDTA) | Fulgent Genetics (fulgentdiagnostics.com) | | | 03-Feb-20 |
| ALZHEIMER'S DISEASE, FAMILIAL VIA THE APP GENE (604) | | APP (104760) | Lavender Top (EDTA) | Prevention Genetics (www.preventiongenetics.com) | Cerebral Amyloid Angiopathy, App-Related (605714); ALZHEIMER DISEASE, FAMILIAL, 1 (104300) | 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) | 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) | 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) | | |
| 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) | | ADD CHARGE FOR MICRODISSECTION | 06-Jul-18 |

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| Amyotrophic Lateral Sclerosis and Frontotemporal Dementia Panel [1965] | 853 | ANG<br>ANXA11<br>ARHGEF28<br>C9orf72<br>CDH13<br>CHMP2B<br>FUS<br>GRN<br>HNRNPA1<br>HNRNPA2B1<br>KIF5A<br>MAPT<br>OPTN<br>PFN1<br>PSEN1<br>PSEN2<br>SETX<br>SOD1<br>SQSTM1<br>TARDBP<br>TBK1<br>TREM2<br>UBQLN2<br>VAPB<br>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) | | 07-Jan-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) | | Always add del/dupl | 22-Aug-17 |
| 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 | | 07-Oct-18 |
| Angelman Syndrome: Methylation and Copy Number Analysis | 311 | SNRPN | Lavender Top (EDTA) | Genome Diagnostics<br>The Department of Paediatric Laboratory Medicine<br>The Hospital for Sick Children | | | 19-Feb-20 |
| Angelman Syndrome: UPD 15 Analysis | 311 | | Lavender Top (EDTA) | Genome Diagnostics<br>The Department of Paediatric Laboratory Medicine<br>The Hospital for Sick Children | | | 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 | | CSF or Serum (Red Top OR Gold SST). Store at -20°C. | BC Neuroimmunology (bcneuro.ca) | Anti-Aquaporin 4 (Anti-AQP4 Ab) $150; Anti-Myelin Oligodendrocyte Glycoproteins antibody (Anti-MOG) $100 | 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 | | 31-Oct-20 |
| Anti-DNase B (Streptococcal) Antibodies [096289] | 1465 | | Serum (Red Top or SST). Transfer to storage tube. Store and ship at RT. Stable for 9 days at RT. Stable for 14 days frozen. | LabCorp/Dynacare | Test performed in USA | | 26-May-20 |
| Anti-DPPX (dipeptidyl aminopeptidase-like 6) | 1735 | | Gold SST | Mitogen Advanced Diagnostics | encephalitis | | 31-Jul-17 |
| 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 | | | 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 | | | 30-Jun-20 |
| Anti-IgLON5 | 1465 | | Gold SST | Mitogen Advanced Diagnostics | | | 16-Feb-20 |
| Anti-MeCP2 | | | Gold SST | Mitogen Advanced Diagnostics | | Part of Arthitis Panel? | 29-Nov-17 |
| Anti-myelin associated glycoproteins (MAG) | 40 | | Gold SST | Mitogen Advanced Diagnostics | | | 04-Apr-19 |
| 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 | Same price for all anti-TB drugs per determination | 2020-20-29 |

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| Anti-THSD7A | 1465 | | Gold SST; store serum at -20°C | Mitogen Advanced Diagnostics | primary membranous nephropathy | | 15-May-17 |
| 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) | | |
| Aripiprazole (Abilify) | 1142 | | Red top tube ( Gold top SST tube not accepted) | Mayo Clinical Laboratories (www.mayomedicallaboratories.com) | | | 05-Apr-19 |
| 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/ | | | 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) | | 05-Apr-19 |
| 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), MFSD8 (611124), MKKS (604896), MKS1 (609883), MLC1 (605908), MMADHC (611935), MPV17 (137960), MPZ (159440), MR1 (600764), MRE11A (NA), mtDNA (NA), MTFMT (611766), MTPAP (613669), MTTP (157147), MVK (251170), NAGLU (609701), NDUFA1 (300078), NDUFA10 (603835), NDUFA11 (612638), NDUFA12 (614530), NDUFA2 (602137), NDUFA9 | Lavender Top (EDTA) | MNG Laboratories (www.medicalneurogenetics.com) | | | 14-May-20 |

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| 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), MFSD8 (611124), MKKS (604896), MKS1 (609883), MLC1 (605908), MMADHC (611935), MPV17 (137960), MPZ (159440), MR1 (600764), MRE11A (NA), mtDNA (NA), MTFMT (611766), MTPAP (613669), MTTP (157147), MVK (251170), NAGLU (609701), NDUFA1 (300078), NDUFA10 (603835), NDUFA11 (612638), NDUFA12 (614530), NDUFA2 (602137), NDUFA9 | Lavender Top (EDTA) | MNG Laboratories (www.medicalneurogenetics.com)/Dynacare | | | 14-May-20 |
| Atypical Hemolytic Uremic Syndrome and Membranoproliferative Glomerulonephritis Panel: Sequencing | | CD46<br>CFB<br>CFH<br>CFHR5<br>CFI<br>C3<br>THBD<br>APLN | Lavender Top (EDTA) 2 x 4 mL | The Hospital for Sick Children<br>Rapid Response Laboratory<br>170 Elizabeth Street, Room 3642<br>Toronto, ON<br>M5G 2G3<br>Canada<br>Phone: 416-813-7200<br>Phone: 1-855-381-3212 | aHUS<br>Familial Hemolytic-Uremic syndrome<br>Hereditary Hemolytic-Uremic syndrome<br>MPGN; Mesangiocapillary glomerulonephritis | | |
| 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,<br>Familial Hemolytic-Uremic syndrome,<br>Hereditary Hemolytic-Uremic syndrome,<br>MPGN; Mesangiocapillary glomerulonephritis | 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) | | 02-Feb-18 |
| Autism/ID Xpanded Panel [952] | 717 | | Lavender Top (EDTA) | GeneDx (www.genedx.com) | | | 09-Dec-19 |
| Autoimmune Dysautonomia Evaluation, Serum (ADE) | | | Red Top | Mayo Clinical Laboratories (www.mayomedicallaboratories.com) | | | |

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| Autoimmune Liver Diseases Profile | 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 3EBPO LC-1 Ro52/TRIM21 | | 23-Aug-18 |
| 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 | | 03-Apr-19 |
| Autoimmune Myopathy/Myositis Profile | 153 | | Gold SST | Mitogen Advanced Diagnostics | Jo-1, Mi2, Mi2-α, Mi2β, MDA5, NXP2, TIF1γ PL7, PL12, PM/Scl75, PM/Scl100, Ku, SRP, Ro52, EJ, OJ, Ro52. | | 29-Nov-17 |
| Autoimmune Polyendocrinopathy Syndrome Type 1 via the AIRE Gene [1224] | | AIRE (607358) | Lavender Top (EDTA) | Prevention Genetics (www.preventiongenetics.com) | | | 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) | | 11-Apr-20 |
| Autoinflammatory Syndromes Panel [08120] | 838 | | Lavender Top (EDTA) | Invitae (www.invitae.com) | | | 03-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) | | | |
| Avian IgG Antibodies Panel, Serum (Budgie and Pigeon) | 1419 | | Red Top; Gold SST | In-Common Laboratories | budgie = parakeet | | 10-Mar-20 |
| Avian precipitins: Pigeon IgG Antibodies | 1419 | | Red Top; Gold SST | In-Common Laboratories | | | 08-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) | | 09-Dec-19 |
| Baller-Gerold syndrome (218600) | | RECQL4 (603780) | Lavender Top (EDTA) | Centogene AG (www.centogene.com) | | | |
| 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 |

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| Basal Ganglia CalcificationNGS 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) | | 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) | | 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) | | 19-Feb-20 |
| Beta-Thalassemia (613985) | 823 | HBB (141900) | Lavender Top (EDTA) | Molecular Genetics Laboratory - McMaster University Medical Centre | | | |
| 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. | Available at MCL ($153.70) [BAFS] | 08-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 | | NOT AVAILABLE AT MCL | 08-Aug-17 |
| Birt-Hogg-Dubé Syndrome (135150) | 740 | FLCN (607273) | Lavender Top (EDTA) | Alberta Mol Dx Laboratory (Calgary) | | | 16-Jun-20 |
| Blastomyces Antibody by EIA, Serum [BLAST] | 491 | | Red Top | Mayo Clinical Laboratories (www.mayomedicallaboratories.com) | | | 31-Oct-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) | | 09-Dec-19 |
| Bone Marrow Failure Syndrome Panel [HE0801] | 1427 | 135 genes | Lavender Top (EDTA) [2 tubes] | Blueprint Genetics http://blueprintgenetics.com/ | | | 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 | | $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) | | | 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. | | 06-Feb-20 |

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| 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) | | 07-Oct-18 |
| Bullous Autoimmune Skin Disease Profile | 750 | | Serum gel (Gold top) | Mitogen Advanced Diagnostics | BP180, BP230, Desmoglein 1, Desmoglein 3 | | 07-Aug-19 |
| 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) | Available as Facteur C3 nephritique (activité) at CHUQ (HEJ) | 07-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 | | 18-Sep-19 |
| 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 [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 | 29-Jun-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) | | 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) | | 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) | | |
| Carbohydrate Deficient Transferrin for Congenital Disorders of Glycosylation, Serum [CDG] | 406 | | Red Top. Store serum frozen. | Mayo Clinical Laboratories (www.mayomedicallaboratories.com) | | | 07-Oct-20 |
| Carbohydrate, Urine [CHOU] | 1572 | | Random urine. Store frozen. Stable 21 days. | Mayo Clinical Laboratories (www.mayomedicallaboratories.com) | Replaces "reducing substances" | | 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) | | |
| 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) | | |
| CDKN2A (p16) & CDK4 (Exon2) Sequencing [2021] | | CDKN2A (600160); CDK4 (123829) | Lavender Top (EDTA) | GeneDx (www.genedx.com) | Melanoma, cutaneous malignant, 3 | | 09-Dec-19 |

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| Central Hypoventilation And Apnea Panel [PU0401] | | CHAT, CHRNA1,CHRNB1, CHRND, CHRNE, COLQ, EDN3, GLRA1, MEP2, PHOX2B, PAPSN, RET, SCN4A, SLC6A5, ZEB2 | Lavender Top (EDTA) | Blueprint Genetics http://blueprintgenetics.com/ | | | 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) | | 12-Nov-19 |
| CEREBRAL CAVERNOUS MALFORMATIONS VIA THE CCM2 GENE (603284) [122] | | CCM2 (607929) | Lavender Top (EDTA) | Prevention Genetics (www.preventiongenetics.com) | Cerebral calvernous malformations-2 | | |
| CEREBRAL CAVERNOUS MALFORMATIONS VIA THE KRIT1/CCM1 GENE [121] | | KRIT1 (604241) | Lavender Top (EDTA) | Prevention Genetics (www.preventiongenetics.com) | Cerebral calvernous malformations-1 (116860) | | |
| CEREBRAL CAVERNOUS MALFORMATIONS VIA THE PDCD10/CCM3 (603285) [123] | | PDCD10 (609118) | Lavender Top (EDTA) | Prevention Genetics (www.preventiongenetics.com) | Cerebral calvernous malformation-3 | | |
| CerebroTendinous Xanthomatosis (CTX) via the CYP27A1 Gene [1670] | | CYP27A1 (606530) | Lavender Top (EDTA) | Prevention Genetics (www.preventiongenetics.com) | CerebroTendinous Xanthomatosis (213700) | | 31-Mar-17 |
| 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 (610826), SLC35A2 (314375), GPR172A (607882), SLC6A8 (300036), SPG11 (610844), SPTAN1 (182810), SUCLA2 (603921), SUMF1 (607939), SURF1 (185620), TBC1D24 (613577), TDP1 (607198), TFG (602498), TMEM126A (612988), TMTC3 (617218), TRIM2 (614141), TRMT5 (611023), TRPV4 (605427), TSFM | Lavender Top (EDTA) | MNG Laboratories (www.medicalneurogenetics.com)/Dynacare | Charcot-Marie tooth Disease (302800) | 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) | | | 28-Feb-20 |
| Cholestanol [FCHO] | 400 | | Lavender top (EDTA) | Mayo Clinical Laboatories (Kennedy Kreiger Institute (Biochemical Genetics)) | Cerebrotendinous xanthomatosis | | 18-Sep-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 | **NOTE PARTICULIÈRE /** Special comment | **Date de la révision /** Date of revision |
|---|---|---|---|---|---|---|---|
| Cholestasis Panel | 400 | ABCB11<br>MYO5B*<br>NOTCH2*<br>NR 21 27<br>NPHP1<br>PEX1<br>PEX10<br>PEX12<br>PEX2<br>PEX26<br>PEX5<br>PEX6<br>SERPINA1<br>SLC25A13 SLC26A3<br>SMPD1<br>SPINT2<br>TJP2<br>TMEM216<br>TRMU L<br>TTC37<br>UGT1A1<br>VIPAS39<br>VPS33B<br>ABCB4<br>ABCC2<br>AKR1D1<br>ATP8B1<br>BAAT<br>CFTR<br>CREB3L3<br>CYP7B1<br>DCDC2<br>DGUOK<br>EPCAM 80<br>FAH<br>HSD3B7<br>JAG1<br>LCT | Lavender Top (EDTA) | Blueprint Genetics http://blueprintgenetics.com/ | | | 28-May-19 |
| Choreoacanthocytosis (200150) | | VPS13A (605978) | Lavender Top (EDTA) | North York General | | | |
| 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) | | |
| Chronic Granulomatous Disease NextGen Sequencing (NGS) Panel [1971] | | CYBA ( 608508); CYBB (300481); NCF2 (608515); NCF4 (601488) | Lavender Top (EDTA) | Fulgent Genetics (fulgentdiagnostics.com) | | | |
| Chronic Inflammatory Demyelinating Polyneuropathy (CIDP) Test Panel - Nodal and Paraneodal 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) | | 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) | | | 08-Feb-19 |
| CLCN1 Full Gene sequencing Analysis [MOL356]; Paramyotonia Congenita (168300) | | CLCN1 (118425) | Lavender Top (EDTA) | MNG Laboratories (www.medicalneurogenetics.com)/Dynacare | Paramyotonia Congenita (168300) | | 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) | | |
| 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) | | | 15-May-19 |
| Collagen VII IgG Antibody Level | 1399 | | Red Top or Gold SST. Stability<br>Ambient: 7 days Refrigerated: 14 days Frozen: Indefinitely | Immunodermatology Laboratory<br>Department of Dermatology<br>30 North 1900 East, 4A330 SOM<br>Salt Lake City, Utah 84132<br>Email: immunoderm@hsc.utah.edu | | | 06-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) | | | 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) | | |
| ColoVantage (Methylated Septin 9) [16983] | 1395 | SEPT9 | Plasma (EDTA); handle at 4°C; freeze plasma in plastic container. Minimum 10 mL. | Quest Diagnostics Chantilly<br>14225 Newbrook Dr.<br>Chantilly, VA 20153-0841 | | | 11-Apr-18 |

| **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 | **NOTE PARTICULIÈRE /** Special comment | **Date de la révision /** Date of revision |
|---|---|---|---|---|---|---|---|
| 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), COX412, 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), | | 09-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) | | | 02-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 |
| Complement System Disorder Panel Plus Anlaysis [IM0701] | 803 | ADIPOQ Complement system AD/AR 2 8<br>ADIPOR1* Complement system AD/AR 4<br>ADIPOR2 Complement system AD/AR<br>ARMC4* Ciliary dyskinesia AR 13 15<br>C1QA C1q deficiency AR 2 7<br>C1QB C1q deficiency AR 4 7<br>C1QBP Primary immunodeficiency AD/AR 6<br>C1QC C1q deficiency AR 4 7<br>C1R Immunodeficiency AD/AR 14 16<br>C1S Complement component C1s deficiency AR 4 8<br>C2* Complement component 2 deficiency AR 4 6<br>C3 Hemolytic uremic syndrome, atypical, Complement component 3 deficiency AD/AR 5 82<br>C3AR1 Complement system AD/AR 1 3<br>C4A* Blood group, Chido/Rodgers system BG 1 5<br>C4B* Complement component 4B deficiency AR 8<br>C4BPA Complement system AD/AR 2<br>C4BPB Complement system AD/AR<br>C5 Eculizumab, poor response to, Complement component 5 deficiency AD/AR 5 17<br>C5AR1 Complement system AD/AR<br>C5AR2 Complement system AD/AR 2<br>C6 Complement component 6 deficiency AR 7 11<br>C7 Complement component 7 deficiency AR 14 29<br>C8A Complement component 8 deficiency AR 2 5<br>C8B Complement component 8 deficiency AR 7 7<br>C8G Immunodeficiency AD/AR<br>C9 Complement component 9 deficiency AR 7 7<br>CCDC39 Ciliary dyskinesia AR 16 38<br>CCDC40 Ciliary dyskinesia AR 19 32<br>CCDC65 Ciliary dyskinesia AR 1<br>CCDC103 Ciliary dyskinesia AR 3 4<br>CCDC114 Ciliary dyskinesia AR 6 7<br>CCNO Ciliary dyskinesia AR 9 9<br>CD46* Hemolytic uremic syndrome, atypical AD/AR 4 64<br>CD55 Blood group, Cromer system BG 7 6<br>CD59 CD59 deficiency AR 3 6<br>CD93 Complement system AD/AR<br>CFB Complement factor B deficiency, Hemolytic uremic syndrome, atypical AD/AR 2 21<br>CFD Complement factor D deficiency AR 2 3<br>CFH* Hemolytic uremic syndrome, atypical, Complement factor H deficiency AD/AR 17 259<br>CFI Hemolytic uremic syndrome, atypical, Complement factor I deficiency AD/AR 7 123<br>CFP Properdin deficiency XL 5 17<br>CLU Complement system AD/AR 16<br>COLEC11 3MC syndrome AR 6 10<br>CR1* Blood group, Knops system BG 1 15<br>CR2 Common variable immunodeficiency AR 2 5<br>CRP Complement system AD/AR | Lavender Top (EDTA) | Blueprint Genetics http://blueprintgenetics.com/ | | | 02-Feb-18 |

| **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 | **NOTE PARTICULIÈRE /** Special comment | **Date de la révision /** Date of revision |
|---|---|---|---|---|---|---|---|
| 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), COX4I1 (123864), COX4I2 (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), ECI1 (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), GFM2 (606544), GPAM (602395), GPD1 (138420), GPD2 (138430), GPHN (603930), GPI (172400), GPT2 (138210), GSK3B (605004), GSS (601002), GTPBP3 (608536), GYG1 (603942), GYS1 (138570), GYS2 (138571), HADH (601609), HADHA (600890), HADHB (143450), HARS2 (600783), HCCS (300056), HK1 (142600), HK2 (601125), HK3 (142570), HLCS (609018), | Lavender Top (EDTA) | MNG Laboratories (www.medicalneurogenetics.com)/Dynacare | | | 14-May-20 |
| 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 | | | 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 | **NOTE PARTICULIÈRE /** Special comment | **Date de la révision /** Date of revision |
|---|---|---|---|---|---|---|---|
| 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), TSC2 (191092), UBE3A (601623), UBR5 (601068), WFS1 (222300), ZEB2 (605802).<br><br>ABAT (137150), ABCC8 (600509), ACADSB (600301), ACSF3 (614245), ADSL (608222), ALDH5A1 (610045), ALDH7A1 | Lavender Top (EDTA) | MNG Laboratories (www.medicalneurogenetics.com)/Dynacare | | Alternative: Fulgent Epilepsy NGS Panel | 28-Jul-20 |
| 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, RFT1, RNASEH2A, RNASEH2B, RNASEH2C, ROGDI, RPGRIP1L, RYR3, SAMHD1, SCARB2, SCN10A, SCN1A, SCN1B, SCN2A, SCN2B, SCN3A, SCN3B, SCN4A, SCN4B, SCN5A, SCN8A, SCN9A, SCO2, SDHA, SERPINI1, SETBP1, SGSH, SHH, SHOC2, SIK1, SIX3, SLC12A5, | Lavender Top (2 x 4 mL) | Fulgent Genetics (fulgentdiagnostics.com) | | | |
| Comprehensive Leukodystrophy/Leukoencephalopathy (NGS Panel and Copy Number Analysis + mtDNA) [NGS372] | 964 | | Lavender Top (EDTA) | MNG Laboratories (www.medicalneurogenetics.com)/Dynacare | | | 29-Sep-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 | **NOTE PARTICULIÈRE /** Special comment | **Date de la révision /** Date of revision |
|---|---|---|---|---|---|---|---|
| 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, STEB, SUCLA2, SUCLG1, SUMF1, SUOX, TAZ, TBC1D4, TCF4, TCN2, TFR2, TIMM8A, TK2, TMEM126A, TMEM165, TMEM70, TPP1, TREX1, TRPM6, TRPM7, TUSC3, TWNK, TYMP, UCP2, UROD, UROS, WFS1, ZMPSTE24 | Lavender Top (EDTA) 2 x 4 mL | | | | 28-Feb-20 |
| 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 (606598), GFER (600924), GFM1 (606639), GFPT1 (138292), GJB1 (304040), GJC2 (608803), GK (300474), GLE1 (603371), GMPPB (615320), GNB4 (610863), GNB5 (604447), GNE (603824), GPI (172400), GRID2 (602368), GRIN2D (602717), GTF2H5 (608780), GYG1 (603942), GYS1 (138570), HADHA | Lavender Top (EDTA) | MNG Laboratories (www.medicalneurogenetics.com)/Dynacare | | See Prevention Genetics Panel | 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 | **NOTE PARTICULIÈRE /** Special comment | **Date de la révision /** Date of revision |
|---|---|---|---|---|---|---|---|
| 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 KCNJ6 KCNQ2 KCNT1 KDM1A KDM5C KIAA0196 KIAA0226 KIAA0586 KIAA1033 KIAA2022 KIDINS220 KIF1A KIF1B KIF1C KIF2A KIF5A KIF5C KLC2 KRIT1 KY L1CAM L2HGDH LAGE3 LAMA2 LAMB1 LAMB2 LARGE LARS2 LDB3 LDHA LEP LIAS LIFR LINS LIPT1 LIPT2 | Lavender Top (EDTA) | MNG Laboratories (www.medicalneurogenetics.com)/Dynacare | | | 14-May-20 |
| 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 | | | 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) | | |
| 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/ | | | 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 | **NOTE PARTICULIÈRE /** Special comment | **Date de la révision /** Date of revision |
|---|---|---|---|---|---|---|---|
| Congenital Hyperinsulinism Sequencing Panel [1939] | 541 | ABCC8 (600509)<br>GCK (138079)<br>GLUD1 (138130)<br>HADH (601609)<br>HNF1A (142410)<br>HNF4A (600281)<br>KCNJ11 (600937)<br>SLC16A1 (600682)<br>UCP2 (601693) | Lavender Top (EDTA) | Prevention Genetics (www.preventiongenetics.com) | Hyperinsulinemic Hypoglycemia, Familial 3 602485<br>Hyperinsulinemic Hypoglycemia, Familial 6 606762<br>Hyperinsulinemic Hypoglycemia, Familial, 1 256450<br>Hyperinsulinemic Hypoglycemia, Familial, 2 601820<br>Hyperinsulinemic Hypoglycemia, Familial, 4 609975<br>Hyperinsulinemic Hypoglycemia, Familial, 7 610021<br>Maturity-Onset Diabetes Of The Young, Type 1 125850<br>Maturity-Onset Diabetes Of The Young, Type 3 600496<br>Related Tests | | 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)<br>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 (613152); Merosin | 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 | **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 (2532900; 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- | | 25-Jul-17 |
| CONGENITAL MYOPATHY NEXTGEN SEQUENCING (NGS) PANEL [1365] | 631 | | Lavender Top (EDTA) | Prevention Genetics (www.preventiongenetics.com) | | | |
| Congenital Neutropenia Panel [IM0501] | 836 | ACTB* CLPB<br>CSF2RA#* CSF3R<br>CTSC EFL1*<br>ELANE G6PC3<br>GATA2 GFI1<br>GINS1 HAX1<br>IFNGR2<br>JAGN1 LAMTOR2<br>LYST MKL1<br>PGM3 RAC2<br>SBDS* SLC37A4<br>SMARCD2 SRP54<br>SRP72* VPS13B<br>VPS45# WAS WDR1 | Lavender Top (2 x 4 mL) | Blueprint Genetics http://blueprintgenetics.com/ | | | 28-Feb-20 |
| Copeptin & Osmolality | 1465 | | Serum or Plasma (Li heparin). Store and send frozen. | In-Common Laboratories | replaces ADH | | 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) | | |
| 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 | | 09-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 | | 2017/03/09 | |
| 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 burnettii IgG et IgM) at CHUS Fleurimont | | 31-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 | **NOTE PARTICULIÈRE /** Special comment | **Date de la révision /** Date of revision |
|---|---|---|---|---|---|---|---|
| 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 | | |
| 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) | | |
| 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) | | |
| Cryoglobulin and Cryofibrinogen Panel, Serum and Plasma [CRGSP] | | | **Cryofibrinogen**<br>**Collection** Container/Tube: Lavender top (EDTA)<br>Submission Container/Tube: Plastic vial<br>Specimen Volume: 1 mL<br>Collection Instructions:<br>1. Tube must remain at 37 degrees C.<br>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.<br>3. Place plasma into an appropriately labeled plastic vial.<br>**Cryoglobulin**<br>**Collection** Container/Tube: Red top<br>Submission Container/Tube: Plastic vial<br>Specimen Volume: 5 mL<br>Collection Instructions:<br>1. Tube must remain at 37 degrees C.<br>2. Allow blood to clot at 37 degrees C.<br>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.<br>4. Place serum into an appropriately labeled plastic vial. | Mayo Clinical Laboratories (www.mayomedicallaboratories.com) | Evaluating patients with vasculitis, glomerulonephritis, and lymphoproliferative diseases<br><br>Evaluating patients with macroglobulinemia or myeloma in whom symptoms occur with cold exposure | | |
| 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 | | |
| 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) | 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) | | |

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| Currarino syndrome | | MNX1 (142994) | Lavender top (EDTA) | Centogene AG (www.centogene.com) | Currarino syndrome (176450) | | |
| Custom Panel - Cystinuria | | SLC3A1 (104614); SLC7A9 (604144); PREPL (609577) | Lavender Top (2 x 4 mL) | Fulgent Genetics (fulgentdiagnostics.com) | Cystinuria (220100) | | |
| Cyclic AMP, Urinary Excretion [GRP] | | | Serum (Red Top) AND random urine resquired. Store frozen. | Mayo Clinical Laboratories (www.mayomedicallaboratories.com) | | | 19-May-17 |
| Cystatin C [CYSTC] | 78 | | Serum. Store and send frozen | In-Common Laboratories | | | 06-Sep-18 |
| Cystic Fibrosis: CFTR Deletion Duplication Analysis | 763 | CFTR (602421) | Lavender Top (EDTA) | Genome Diagnostics<br>The Department of Paediatric Laboratory Medicine<br>The Hospital for Sick Children<br>(www.sickkids.ca/paediatriclabmedicinems/lab-divisions/genome-diagnostics/genome-diagnostics.html#genome) | Cystic Fibrosis (219700) | | |
| 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)<br>Alternate name: DARS2: Mitochondrial Aspartyl-tRNA Synthetase Deficiency | | 14-May-20 |
| Deletion 1p | | | Green Top; RT only | Cytogenetics Laboratory, Hospital for Sick Children | Chromosome1p36 deletion syndrome (607872) | | |
| 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 |
| Dexamethasone [FDXM] | | | Red Top | Mayo Clinical Laboratories (www.mayomedicallaboratories.com) | | | 31-Oct-20 |
| DGUOK Sequence Analysis<br>(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) | | |
| Diabetes Insipidus Panel [KI1801] | 1457 | AQP2 AVP AVPR2 | Lavender Top (EDTA) 2 x 4 mL | Blueprint Genetics http://blueprintgenetics.com/ | | | 28-Feb-20 |
| DICER1 single gene test [S00555] | 246 | DICER1 | Lavender Top (EDTA) 2 x 4 mL | Blueprint Genetics http://blueprintgenetics.com/ | | | 28-Feb-20 |
| Dihydrotestosterone, Serum [DHT] | 1459 | | Red Top or Gold Top. Store serum and send frozen | In-Common Laboratories | | | 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)<br>(https://www.nemours.org/pediatric-research/labservices/diagnostic/gastroenterology-lab.html) | | | 24-Aug-17 |

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| Disorders of Sex Development Sequencing Panel with CNV Detection [4509] | 1395 | AMH 600957<br>AMHR2 600956<br>ANOS1 300836<br>AR 313700<br>ARL6 608845<br>ARX 300382<br>ATRX 300032<br>BBS1 209901<br>BBS10 610148<br>BBS12 610683<br>BBS2 606151<br>BBS4 600374<br>BBS5 603650<br>BBS7 607590<br>BBS9 607968<br>CBX2 602770<br>CHD7 608892<br>CYP11A1 118485<br>CYP17A1 609300<br>CYP19A1 107910<br>DHH 605423<br>DMRT1 602424<br>DMRT2 604935<br>FGF8 600483<br>FGFR1 136350<br>FGFR2 176943<br>FOXL2 605597<br>FSHB 136530<br>GATA4 600576<br>GNRH1 152760<br>GNRHR 138850<br>HESX1 601802<br>HFE 613609<br>HS6ST1 604846<br>HSD17B3 605573<br>KISS1 603286<br>KISS1R 604161<br>LEP 164160<br>LEPR 601007<br>LHCGR 152790<br>LHX3 600577<br>LHX4 602146<br>MAMLD1 300120<br>MAP3K1 600982<br>MKKS 604896<br>NR0B1 300473<br>NR5A1 184757<br>NSMF 608137<br>PCSK1 162150<br>POR 124015 | Lavender Top (EDTA) | Prevention Genetics (www.preventiongenetics.com) | 3-Oxo-5 Alpha-Steroid Delta 4-Dehydrogenase Deficiency AD 264600<br>46,XX Sex Reversal, Type 1 AR 400045<br>46,XY Sex Reversal, Type 3 AD 612965<br>46,XY Sex Reversal, Type 5 AD 613080<br>46,XY Sex Reversal, Type 6 AD 613762<br>46,XY Sex Reversal, Type 7 AR 233420<br>Adrenal Insufficiency, Congenital, With 46,XY Sex Reversal, Partial Or Complete AR 613743<br>Androgen Resistance Syndrome XL 300068<br>Antley-Bixler Syndrome AR 207410<br>Antley-Bixler Syndrome With Genital Anomalies And Disordered Steroidogenesis AR 201750<br>ATR-X Syndrome XL 301040<br>Bardet-Biedl Syndrome 1 AR 209900<br>Bardet-Biedl Syndrome 10 AR 615987<br>Bardet-Biedl Syndrome 11 AR 615988<br>Bardet-Biedl Syndrome 12 AR 615989<br>Bardet-Biedl Syndrome 2 AR 615981<br>Bardet-Biedl Syndrome 3 AR 600151<br>Bardet-Biedl Syndrome 4 AR 615982<br>Bardet-Biedl Syndrome 5 AR 615983<br>Bardet-Biedl Syndrome 6 AR 605231<br>Bardet-Biedl Syndrome 7 AR 615984<br>Bardet-Biedl Syndrome 8 AR 615985<br>Bardet-Biedl Syndrome 9 AR 615986<br>Blepharophimosis, Ptosis, And Epicanthus Inversus AR 110100<br>Camptomelic Dysplasia AD 114290<br>Cholesterol Monooxygenase (Side-Chain Cleaving) Deficiency AR 201710<br>Deficiency Of Steroid 17-Alpha-Monooxygenase AD,AR 202110<br>Familial Gynecomastia, Due To Increased Aromatase Activity AR 139300<br>Follicle-Stimulating Hormone Deficiency, Isolated XL 229070<br>Gonadotropin-Independent Familial Sexual Precocity AR 176410<br>Hemochromatosis Type 1 AR 235200<br>Hypogonadotropic Hypogonadism 10 with or without Anosmia AR 614839<br>Hypogonadotropic Hypogonadism 11 with or without Anosmia AR 614840<br>Hypogonadotropic Hypogonadism 12 with or without Anosmia AR 614841<br>Hypogonadotropic Hypogonadism 13 with or without Anosmia AR 614842<br>Hypogonadotropic Hypogonadism 14 with or without Anosmia AD 614858<br>Hypogonadotropic Hypogonadism 15 with or | | 06-Jun-19 |
| DISTAL HEREDITARY MOTOR NEUROPATHY NGS PANEL | | ATP7A (300011); BSCL2 (606158); DCTN1 (601143); DNMT1 (126375); FIG4 (609390); GAN (605379); GARS (600287); HSPB1 (602195); HSPB8 (6080140; 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) | | |
| 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] | | GCA1 (600225) | Lavender Top (EDTA) | Prevention Genetics (www.preventiongenetics.com) | Dystonia 5, Dopa-Responsive Type (128230) | | |

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| 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) | | | 04-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) | See Med Neurogenetics Panel | 21-Mar-19 |
| Dystonia/Parkinson Panel [T402] | 1096 | | Lavender Top (EDTA) | GeneDx (www.genedx.com) | | | 03-Oct-19 |
| 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) | | | 12-Jul-19 |
| 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) | | | |

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| 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 | 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) | | |
| 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 |
| Factor I | | | 2ml frozen serum. | Cincinnati Children's Hospital Medical Center 3333 Burnet Avenue NRB 1042 Cincinnati, OH 45229 (www.cincinnatichildrens.org) | | | 15-Nov-17 |
| 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 | 03-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 | | 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) | | | 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) | Replaces: UofA targeted mutation testing | |
| Fatty Acid Oxidation Syndrome Panel [ME1701] | 1320 | | Lavender Top (2 x 4 mL) | Blueprint Genetics http://blueprintgenetics.com/ | | | 29-Feb-20 |

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|---|---|---|---|---|---|---|---|
| FH Autoantibody Testing | 64 | | 2ml frozen serum. | Cincinnati Children's Hospital Medical Center 3333 Burnet Avenue NRB 1042 Cincinnati, OH 45229 (www.cincinnatichildrens.org) | | | 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) | | 31-Oct-17 |
| Fibrodysplasia ossificans progressiva (135100) | | ACVR1 (102576) | Lavender Top (EDTA) | University of Pennsylvania School of Medicine | Fibrodysplasia ossificans progressiva (135100) | | |
| Fluphenazine (Prolixin), Serum [PROLX] | 929 | | Serum<br>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 | | 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) | | 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) | | 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) | | 09-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) | | | 20-Jan-20 |
| Gabapentin [GABA] | 107 | | Red Top. Store and send frozen. | In-Common Laboratories | | | 06-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 | 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) | | 09-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) | | |
| 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) | | 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 | | |
| 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 | | |

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|---|---|---|---|---|---|---|---|
| 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 | | 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) | | |
| 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) | | |
| Glucagon, Plasma [GLP] | 110 | | Collection Container/Tube: Lavender top (EDTA)<br>Submission Container/Tube: Plastic vial<br>Specimen Volume: 2 mL<br>Collection Instructions:<br>1. Fasting.<br>2. Prechill tube at 4 degrees C before drawing the specimen.<br>3. Draw the prechilled tube, and process as follows:<br>a. After drawing specimen, chill tube in wet ice for 10 minutes.<br>b. Centrifuge in a refrigerated centrifuge or in chilled centrifuge cup.<br>c. Immediately after centrifugation, remove plasma, place in a plastic transport vial (Supply T465), and freeze. | Mayo Clinical Laboratories (www.mayomedicallaboratories.com) | | | 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) | | | 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) | | |
| GLUT1 Deficiency Syndrome (SLC2A1 Single Gene Test) | 262 | SLC2A1 (138140) | Lavender Top (EDTA) [2 tubes] | Fulgent Genetics (fulgentdiagnostics.com) | | | 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<br>ALDOA 103850<br>ALDOB 612724<br>ENO3 131370<br>G6PC 613742<br>GAA 606800<br>GBE1 607839<br>GYG1 603942<br>GYS1 138570<br>GYS2 138571<br>LAMP2 309060<br>LDHA 150000<br>PC 608786<br>PCK1 614168<br>PCK2 614095<br>PFKM 610681<br>PGAM2 612931<br>PGM1 171900<br>PHKA1 311870<br>PHKA2 300798<br>PHKB 172490<br>PHKG2 172471<br>PRKAG2 602743<br>PYGL 613741<br>PYGM 608455<br>SLC16A1 600682<br>SLC2A2 138160<br>SLC37A4 602671 | 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) | | 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) | | |
| 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) | | |

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|---|---|---|---|---|---|---|---|
| Goose Feather (e70) IgE, Serum | 1419 | | Gold SST | Check if available at CHUM | | | 07-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) | | |
| 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 | | | 20-Sep-19 |
| 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) | | | |
| 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) | | | |
| 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 | | | 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) | | 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) | | 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) | | 05-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) | | |

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| 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) | | | |
| 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 | | | 09-Dec-19 |
| Hereditary Spastic Paraplegia: Autosomal Dominant [HSP-Panel 1] | 657 | ALDH18A1<br>ATL1<br>BSCL2<br>C10orf2<br>HSPD1<br>KIAA0196<br>KIF5A<br>NIPA1<br>POLG<br>POLG2<br>REEP1<br>SPAST<br>RTN2<br>SLC33A1<br>SETX | Lavender Top (EDTA) | Molecular Genetics Laboratory, Hospital for Sick Children (www.sickkids.ca/PaediatricLaboratoryMedicine/Laboratories-Services/Molecular-Genetics-Laboratory/index.html) | | Price of individual genes: $500 (seq) + 500$ (del/dupl) | 21-Aug-19 |
| Hereditary Spastic Paraplegia: Autosomal Recessive [HSP-Panel 2] | 657 | ALDH18A1<br>ALS2<br>AP4B1<br>AP4E1<br>AP4M1<br>AP4S1<br>AP5Z1<br>C10orf2<br>C12orf65<br>C19orf12<br>CYP2U1<br>CYP7B1<br>DDHD1<br>DDHD2<br>ENTPD1<br>ERLIN1<br>ERLIN2<br>FA2H<br>GBA2<br>GJC2<br>KIF1A<br>KIF1C<br>NT5C2<br>PGAP1<br>PNPLA6<br>POLG<br>SACS<br>SPG11<br>SPG20<br>SPG21<br>SPG7<br>TECPR2<br>VPS37A<br>ZFYVE26 | Lavender Top (EDTA) | Molecular Genetics Laboratory, Hospital for Sick Children (www.sickkids.ca/PaediatricLaboratoryMedicine/Laboratories-Services/Molecular-Genetics-Laboratory/index.html) | | Price of individual genes: $500 (seq) + 500$ (del/dupl) | 21-Aug-19 |

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|---|---|---|---|---|---|---|---|
| Hereditary Spastic Paraplegia: Comprehensive Testing [HSP-COMP] | 657 | ALDH18A1<br>ATL1<br>BSCL2<br>C10orf2<br>HSPD1<br>KIAA0196<br>KIF5A<br>NIPA1<br>POLG<br>POLG2<br>REEP1<br>SPAST<br>RTN2<br>SLC33A1<br>SETX<br>ALS2<br>AP4B1<br>AP4E1<br>AP4M1<br>AP4S1<br>AP5Z1<br>C10orf2<br>C12orf65<br>C19orf12<br>CYP2U1<br>CYP7B1<br>DDHD1<br>DDHD2<br>ENTPD1<br>ERLIN1<br>ERLIN2<br>FA2H<br>GBA2<br>GJC2<br>KIF1A<br>KIF1C<br>NT5C2<br>PGAP1<br>PNPLA6<br>SACS<br>SPG11<br>SPG20<br>SPG21<br>SPG7<br>TECPR2<br>VPS37A<br>ZFYVE26<br>L1CAM<br>PLP1<br>SLC16A2 | Lavender Top (EDTA) | Molecular Genetics Laboratory, Hospital for Sick Children (www.sickkids.ca/PaediatricLaboratoryMedicine/Laboratories-Services/Molecular-Genetics-Laboratory/index.html) | | 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) | | | 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) | | | |
| 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 | | 28-Feb-19 |
| 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) | | | 28-Feb-19 |
| Histoplasma Antibody, Serum [SHSTO] | | | Red Top | Mayo Clinical Laboratories (www.mayomedicallaboratories.com) | Histoplasmosis | | 20-Oct-20 |
| 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) | | |
| HTRA1 DNA Sequencing Test (CARASIL) [442] | | HTRA1 (602194) | Lavender Top (EDTA) | Athena Diagnostics (www.athenadiagnostics.com) | CARASIL (600142) | | |
| 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) | | | 22-Sep-19 |

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|---|---|---|---|---|---|---|---|
| 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) | | 01-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) | | 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) | 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) | | | 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 | | | 09-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) | | 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) | | |
| 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 | | 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. | | 20-Nov-17 |

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|---|---|---|---|---|---|---|---|
| Idiopathic Ataxia | 1465 | | Gold SST or Red Top | Mitogen Advanced Diagnostics | | | 13-Feb-20 |
| 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) | | | 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) | | | 14-Oct-20 |
| 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) | | |
| 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) | | |
| INGγR12 (CD119) [ILFGPATHWAY] | 817 | | Heparin | Alberta Precision Laboratories (Flow Cytometry - Calgary) | | | 14-Oct-20 |
| Insulin Antibodies [INSAB] | 853 | | Gold SST or Red Top | In Common Laboratories | | | 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) | | |
| 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) | | Not M-R | 20-Oct-20 |

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|---|---|---|---|---|---|---|---|
| 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, PTPN11, PYCR1, PYGL, RAB39B, RAB40AL, RAI1, RAPSN, RBBP8, RBFOX1, RBM10, RELN, RFX6, RPGRIP1L, RPL10, RPS6KA3, SACS, SAMHD1, SATB2, SCN1A, SCN2A, SCN8A, SDCCAG8, SGCA, SGSH, SHANK2, SHANK3, SHROOM4, SIL1, SLC16A2, | Lavender Top (2 x 4 mL) | Fulgent Genetics (fulgentdiagnostics.com) | | | 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] | | | 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) | | | |
| Interstitial Lung Disease Antibody (ILD) Profile | 1724 | | Gold SST or Red Top | Mitogen Advanced Diagnostics | | | 19-Mar-20 |
| 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) | | 30-Apr-18 |
| ITGA9 - Single Gene Testing | | ITGA9 (603963) | Lavender Top (2 x 4 mL) | Fulgent Genetics (fulgentdiagnostics.com) | Integrin alpha 9 | | 30-Apr-18 |
| JAG1 Gene Sequencing & Del/Dup [1004] | | JAG1 (601920) | Lavender Top (EDTA) | GeneDx (www.genedx.com) | Alagille syndrome (118450) | | 09-Dec-19 |
| Jaundice Chip | | | Lavender Top (EDTA) | Cinncinatti Children's Hospital (Division of Human Genetics Diagnostic Laboratories) | multiple genes | | |
| JOUBERT AND MECKEL-GRUBER SYNDROMES NEXTGEN SEQUENCING (NGS) PANEL [1057] | 332 | HI1 (608894)<br>ARL13B (608922)<br>B9D1 (614144)<br>B9D2 (611951)<br>C5orf42 (614571)<br>CC2D2A (612013)<br>CEP290 (610142)<br>CEP41 (610523)<br>INPP5E (613037)<br>KIF7 (611254)<br>MKS1 (609883)<br>NPHP1 (607100)<br>NPHP3 (608002)<br>OFD1 (300170)<br>RPGRIP1L (610937)<br>TCTN1 (609863)<br>TCTN2 (613846)<br>TCTN3 (613847)<br>TMEM138 (614459)<br>TMEM216 (613277)<br>TMEM231 (614949)<br>TMEM237 (614423)<br>TMEM67 (609884)<br>TTC21B (612014)<br>ZNF423 (604557) | Lavender Top (EDTA) | Prevention Genetics (www.preventiongenetics.com) | | | |
| 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) | | | 09-Dec-19 |
| Juvenile Polyposis Syndrome (610069 and 174900) [537] | | BMPR1A (601299) | Lavender Top (EDTA) | GeneDx (www.genedx.com) | | | 09-Dec-19 |

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| Juvenile Polyposis Syndrome (610069 and 174900) [538] | | BMPR1A (601299); SMAD4 (600993) | Lavender Top (EDTA) | GeneDx (www.genedx.com) | | | 09-Dec-19 |
| KCNA1 Full Gene Sequencing Analysis [MOL064] | | KCNA1 (176260) | Lavender Top (EDTA) | MNG Laboratories (www.medicalneurogenetics.com)/Dynacare | Episodic ataxia/myokymia syndrome | | 09-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) | | | 28-Feb-20 |
| KIT Single Gene | 1537 | cKit (164920) | Lavender Top (2 x 4 mL) | Fulgent Genetics (fulgentdiagnostics.com) | Systemic mastocytosis (154800) | | 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) | 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) | | 09-Dec-09 |
| Lacosamide, Serum [LACO] | 1572 | | RED OR GOLD TOP. Store frozen | Mayo Clinical Laboratories (www.mayomedicallaboratories.com) | | | 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)t; Restrictive Dermopathy, Lethal (275210); Hutchinson-Gilford Syndrome (176670) | | 08-May-17 |
| Lebers hereditary optic neuropathy (LHON) (535000) | | | Lavender Top (2 x 4 mL) | London Health Sciences Centre, Molecular Diagnostic Laboratory | Also available: Alberta Mol Dx Laboratory (Edmonton) | See: MNG Laboratories [MOL021] | |
| 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 | | | 09-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) | | 09-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) | | | 12-Jun-20 |
| Leukemia, Philadelphia chromosome-positive, resistant to imatinib | | ABL1 (189980) | Lavender Top (EDTA) | University Health Network (Toronto General Hospital) | | | |

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| 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 | | | 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) | | | |
| LIMB GIRDLE MUSCULAR DYSTROPHY (LGMD) NEXTGEN SEQUENCING (NGS) PANEL [1345] | | ANO5 (608662); CAPN3 (114240); CAV3 (6012530; 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 2J (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) | | |
| LIMB GIRDLE MUSCULAR DYSTROPHY TYPE 2B AND MIYOSHI MYOPATHY VIA THE DYSF GENE [342] | | 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) | | |
| 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) | | |

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|---|---|---|---|---|---|---|---|
| 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 (606090); 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 | | | 09-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) | | | |
| 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) | | | |
| 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) | | 06-Jan-20 |
| 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 | | 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) | | |
| 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 | | | 09-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 | | | 09-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) | | 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) | | 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) | | |

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|---|---|---|---|---|---|---|---|
| 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) | | |
| MECP2 Analysis | | MECP2 (300005) | Lavender Top (EDTA) 2-5 cc | Alberta Children's hospital http://www.medicalgenetics.ca/molecular.html | Rett's disorder (312750) | | 04-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) | aCGH [600] $690 | 15-Mar-07 |
| MERRF (545000), MELAS (540000), NARP (551500) | | | Lavender Top (EDTA) | Alberta Mol Dx Laboratory (Edmonton) | | | |
| Metabolic Hypoglycemia Sequencing Panel with CNV Detection [10365] | 1517 | ACADM 607008<br>ACADVL 609575<br>ACAT1 607809<br>ACSF3 614245<br>AGL 610860<br>ALDOB 612724<br>CA5A 114761<br>DGUOK 601465<br>ETFA 608053<br>ETFB 130410<br>ETFDH 231675<br>FBP1 611570<br>G6PC 613742<br>GALT 606999<br>GK 300474<br>GYS2 138571<br>HADH 601609<br>HMGCL 613898<br>HMGCS2 600234<br>MLYCD 606761<br>MPV17 137960<br>NNT 607878<br>OXCT1 601424<br>PC 608786<br>PCK1 614168<br>PCK2 614095<br>PGM1 171900<br>PHKA2 300798<br>PHKB 172490<br>PHKG2 172471 | Lavender Top (EDTA) | Prevention Genetics (www.preventiongenetics.com) | 3-Hydroxy-3-Methylglutaryl-CoA Lyase Deficiency AR 246450<br>Alpha-Methylacetoacetic Aciduria AR 203750<br>Combined Malonic And Methylmalonic Aciduria AR 614265<br>Congenital Disorder of Glycosylation, Type It AR 614921<br>Fanconi-Bickel Syndrome AR 227810<br>Fructose-Biphosphatase Deficiency AR 229700<br>Galactosemia AR 230400<br>Glycogen Storage Disease 0, Liver AR 240600<br>Glycogen Storage Disease Type Ia AR 232200<br>Glycogen Storage Disease Type Ib AR 232220<br>Glycogen Storage Disease Type Ic AR 232240<br>Glycogen Storage Disease Type III AR 232400<br>Glycogen Storage Disease Type IXa1 XL 306000<br>Glycogen Storage Disease Type IXc AR 613027<br>Glycogen Storage Disease Type VI AR 232700<br>Glycogen Storage DiseaseType IXb AR 261750<br>Hereditary Fructose Intolerance AR 229600<br>Malonyl-CoA Decarboxylase Deficiency AR 248360<br>Monocarboxylate Transporter 1 Deficiency AD,AR 616095<br>Phosphoenolpyruvate Carboxykinase Deficiency, Cytosolic AR 261680<br>Phosphoenolpyruvate Carboxykinase Deficiency, Mitochondrial AR 261650<br>Pyruvate Carboxylase Deficiency AR 266150<br>Succinyl-CoA Acetoacetate Transferase Deficiency AR 245050 | | 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) | | | 23-Oct-17 |
| Migraine Panel [NE1201] | 581 | | Lavender Top (EDTA) | Blueprint Genetics http://blueprintgenetics.com/ | | | 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 | | | 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. | | 04-Aug-20 |

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|---|---|---|---|---|---|---|---|
| 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); | Lavender Top (EDTA) | MNG Laboratories (www.medicalneurogenetics.com)/Dynacare | Mitochondrial DNA Deletion Syndromes<br>Mitochondrial DNA Depletion Syndrome<br>Mitochondrial DNA Depletion Syndrome<br>Mitochondrial DNA Depletion | | 09-May-20 |
| Mitochondrial Genome Sequencing + Deletion Analysis [MOL189] | 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); MTCYB (516020); MTFO1 | 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 | | 14-May-20 |
| 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); | Lavender Top (EDTA) | MNG Laboratories (www.medicalneurogenetics.com)/Dynacare | Mitochondrial diseases including:<br>Leber hereditary optic neuropathy (LHON); Neuropath, ataxia, and retinitis pigmentosa (NARP); Myoclonic epilepsy associated with ragged-red fibers (MERRF); Mitochondrial myopathy, | | 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); | 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:<br>Leber hereditary optic neuropathy (LHON)<br>Neuropath, ataxia, and retinitis pigmentosa (NARP)<br>Myoclonic epilepsy associated with ragged-red fibers (MERRF) | | 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) | | 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) | | | 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 | | |
| Monogenic Autoimmunity Panel [08150] | 817 | ACP5 ADA2 ADAR AICDA AIRE AP3B1 BLOC1S6 BTKCASP10 CASP8 CD27 CD40LG CR2 CTLA4 CYBA CYBBDOCK8 FADD F AS FASLG FOXP3 ICOS IFIH1 IL10IL10RA IL10RB IL21 IL21R IL 2RA ITCH ITK LRBALYST MAGT1 NCF2 NCF4 NFAT5 NFKB2 N FKBIA ORAI1PIK3CD PIK3R1 PLCG2 PNP PRF1 PRKCD RAB27 A RAC2RFX5 RFXANK RFXAP RMRP RNASEH2A RNASEH2B RNASEH2C SAMHD1SH2D1A SLC7A7 STAT1 STAT3 STAT5B S TIM1 STX11 STXBP2TBX1 TMEM173 TNFRSF13B TNFRSF13C T NFSF12 TPP2 TREX1 UNC13DUNG WAS XIAP | Lavender Top (EDTA) [2 tubes] | Invitae (www.invitae.com) | | | 20-Feb-20 |

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|---|---|---|---|---|---|---|---|
| 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 | | 07-Oct-18 |
| MOSIAC-6 Autoimmine Encephalitis Panel | 1417 | NMDAR, $GABA_B$, DPPX, LGI, CASPR, AMPAR | Serum or CSF. Store at -20°C. | 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. | | 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) | 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) | | | |
| 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 | | | 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 | **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 | | 09-May-20 |
| Nephrotic Syndrome (NS)/Focal Segmental Glomerulosclerosis (FSGS) Sequencing Panel with CNV Detection [10417] | 100 | ACTN4 604638<br>ANLN 616027<br>ARHGAP24 610586<br>ARHGDIA 601925<br>CD2AP 604241<br>COL4A3 120070<br>COL4A4 120131<br>COL4A5 303630<br>COL4A6 303631<br>COQ2 609825<br>COQ6 614647<br>COQ8B 615567<br>CRB2 609720<br>CUBN 602997<br>DGKE 601440<br>EMP2 602334<br>FAT1 600976<br>INF2 610982<br>ITGA3 605025<br>ITGB4 147557<br>KANK1 607704<br>KANK2 614610<br>KANK4 614612<br>LAGE3 300060<br>LAMA5 601033<br>LAMB2 150325<br>LMX1B 602575<br>MAGI2 606382<br>MYO1E 601479<br>NPHS1 602716<br>NPHS2 604766<br>NUP107 607617<br>NUP205 614352<br>NUP93 614351<br>OSGEP 610107<br>PAX2 167409<br>PDSS2 610564<br>PLCE1 608414<br>PTPRO 600579<br>SCARB2 602257<br>SGPL1 603729<br>SMARCAL1 606622<br>TP53RK 608679<br>TPRKB 608680<br>TRPC6 603652<br>TTC21B 612014<br>WDR73 616144<br>WT1 607102<br>XPO5 607845 | Lavender Top (EDTA) | Prevention Genetics (www.preventiongenetics.com) | Alport Syndrome, Autosomal Dominant AD 104200<br>Alport Syndrome, Autosomal Recessive AR 203780<br>Alport Syndrome, X-Linked Recessive XL 301050<br>Cerebral Palsy, Spastic Quadriplegic, 2 AR 612900<br>Coenzyme Q10 Deficiency AR 607426<br>Coenzyme Q10 deficiency, primary, 3 AR 614652<br>Coenzyme Q10 deficiency, primary, 6 AR 614650<br>Deafness, X-linked 6 XL 300914<br>Epidermolysis Bullosa With Pyloric Atresia AR 226730<br>Epilepsy, Progressive Myoclonic 4, With Or Without Renal Failure AR 254900<br>Finnish Congenital Nephrotic Syndrome AR 256300<br>Focal Segmental Glomerulosclerosis 1 AD 603278<br>Focal Segmental Glomerulosclerosis 2 AD 603965<br>Focal Segmental Glomerulosclerosis 3 | | 23-Dec-18 |
| 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 | | | 09-May-20 |
| Neurofilament (pNf-H) | 1465 | | CSF or Serum | Mitogen Advanced Diagnostics | | | 28-Jul-20 |
| Neurological Disease Profile (IgG+IgM) | 32 | | Gold SST | Mitogen Advanced Diagnostics | Anti- GM1, GM2, GM3, GD1a, GD1b, GT1b, GQ1b | | 23-Aug-18 |

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|---|---|---|---|---|---|---|---|
| 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) | | 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 | | | 06-Jul-20 |
| 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) | | 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 | | 12-Jun-20 |
| 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 | | | 09-May-20 |
| NOTCH3-CADASIL (125310) | 65 | NOTCH3 (600276) | Lavender Top (EDTA) | London Laboratories Service Group | | To change to NGS with exon copy number in Oct 2015 | |

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|---|---|---|---|---|---|---|---|
| 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 ATP8B1 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 COX4I1 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 ECI1 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 PDHB PDHX PDP1 PDSS1 PDSS2 PDX1 PEX1 PEX10 PEX11B PEX12 PEX13 PEX14 PEX16 PEX19 PEX2 PEX26 PEX3 PEX5 PEX6 PEX7 PGAM2 PGK1 PHB PHYH PKLR PNKD PNMT POLG POLG2 POLRMT PPARGC1A PPARGC1B PPOX PPT1 PREPL PRODH PTGES2 PTS PUS1 | Lavender Top (2 x 4 mL) | Fulgent Genetics (fulgentdiagnostics.com) | | | 10-Jul-19 |
| Oculophyaryngeal Muscular Dystrophy | 613 | PABPN1 (602279) | Lavender Top | Molecular Genetics Laboratory - BC Children's Hospital & BC Women's Hospital | | | 25-Jun-20 |
| Oncotype DX | | | FFPE | Genomic Health | | | |
| Orexin-A/Hypocretin-1, Spinal fluid [ORXNA] | | | CSF. Store frozen. | Mayo Clinical Laboratories (www.mayomedicallaboratories.com) | | | 03-Jun-19 |
| 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) | | | 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) | | | 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) | | | 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 |
| 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 NFIA NFIXNPR2 NSD1 OFD1 PDGFRB PHF6 PIK3CA PIK3R2 PPP2R5B PPP2R5C PPP2R5D PTCH1 PTEN RAB39B RNF125 RNF135 SETD2 STRADA SUZ12 TBC1D7 TMEM94 UPF3BZBTB20 | 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 |

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|---|---|---|---|---|---|---|---|
| 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); CA5A (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); COX411 (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); MRP63 (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); MRPL33 (610059); MRPL34 (611840); MRPL35 (611841); MRPL36 (611842); MRPL37 (611843); MRPL38 (611844); MRPL39 (611845); MRPL4 (611823); MRPL40 (605089); MRPL41 (611846); MRPL42 (611847); MRPL43 (611848); MRPL44 (611849); MRPL45 (611850); MRPL46 (611851); MRPL47 (611852); MRPL48 (611853); MRPL49 (606866); MRPL50 (611854); MRPL51 (611855); MRPL52 | Lavender Top (EDTA) | MNG Laboratories (www.medicalneurogenetics.com)/Dynacare | | | 09-May-20 |
| Oxysterols, Plasma [OXNP] | | | 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 | | 17-May-18 |
| Paliperidone (Invecta) [91895] | | | Red Top (only) | Quest Diagnostics Nichols Institute - California, Molecular Genetics Laboratory | 9-hydroxyrespiridone | | 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) | | | |
| 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) | | | |
| Paraglioma | | SDHB (185470) | Lavender Top (EDTA) | Alberta Mol Dx Laboratory (Calgary) | | | |
| Paraglioma | | SDHC (602413) | Lavender Top (EDTA) | Alberta Mol Dx Laboratory (Calgary) | | | |
| Paraglioma | | SDHD (602690) | Lavender Top (EDTA) | Alberta Mol Dx Laboratory (Calgary) | | | |
| 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) | Alt @MUHC on serum: Anti-Neuronal Ab (Anti:HU,RI,YO,CV2,PNMA2 ,Amphiphysin) | 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 special approval.* | | 31-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 | **NOTE PARTICULIÈRE /** Special comment | **Date de la révision /** Date of revision |
|---|---|---|---|---|---|---|---|
| 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 | | 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) | | |
| Parkinsons Disease/Parkinsonism [NGS357] | 665 | | Lavender Top (EDTA) | MNG Laboratories (www.medicalneurogenetics.com)/Dynacare | | | 09-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. | | |
| 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. | | |
| Perforin | | PRF1 (1780280) | Lavender Top (EDTA) | Hospital for Sick Children (Rapid Response Laboratory) | hemophagocytic lymphohistiocytosis (HLH), familial 2 (603553) | | |
| 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) | | | 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) | Add on: TNFAIP3 | 10-Aug-17 |

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|---|---|---|---|---|---|---|---|
| Phosphatidylserine Antibodies (IgG, IgA, IgM) [10062] | | | Preferred Specimen(s)<br>1 mL plasma collected in a 3.2% sodium citrate (light blue-top) tube<br><br>Minimum Volume<br>0.5 mL<br><br>Collection Instructions<br>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).<br><br>Transport Container<br>Plastic screw-cap vial<br><br>Transport Temperature<br>Room temperature<br><br>Specimen Stability<br>Room temperature: 7 days<br>Refrigerated: 28 days<br>Frozen: 28 days | Quest Diagnostics/Nichols Institute | thrombosis; pregnancy loss | | |
| Phosphoglycerate kinase 1 deficiency (300653) | | PGK1 (311800) | Lavender Top (EDTA) | Centogene AG (www.centogene.com) | | | |
| 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) | | | 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) | | 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) | | |
| 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) | | 04-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) | | |

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|---|---|---|---|---|---|---|---|
| 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) | | |
| Pontocerebellar Hypoplasia NGS Panel | 886 | CASK, OPHN1, RARS2, SEPSECS, TSEN2, TSEN34, TSEN54, VRK1 | Lavender Top (EDTA) | Fulgent Genetics (fulgentdiagnostics.com) | | 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) | See Fulgent Panel | |
| Porencephaly 1 (175780) | | COL4A1 (120130) | Lavender Top (EDTA) | Centogene AG (www.centogene.com) | | | |
| Porphobilinogen Deaminase (PBGD), Whole Blood [PBGD_] | 1324 | | Green Top, 2 mL, 4°C only | Mayo Clinical Laboratories (www.mayomedicallaboratories.com) | Acute Intermittent Porphyria (176000) | | |
| 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) | | | 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) | | 07-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) | 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 | | | 07-Oct-18 |

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|---|---|---|---|---|---|---|---|
| Procainamide and N-acetylprocainamide, Serum [PROCG] | | | Gold SST | Mayo Clinical Laboratories (www.mayomedicallaboratories.com) | | | 21-Feb-18 |
| Procollagen I Intact N-Terminal, Serum [PINP] | | | 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 | | 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) | | |
| Progressive Familial Intrahepatic Cholestasis Type 1 (211600) | | ATP8B1 (602397) | Lavender Top (EDTA) | Cinncinatti Children's Hospital (Division of Human Genetics Diagnostic Laboratories) | | | |
| 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) | | | 29-Jan-19 |
| 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<br>Children (>2 years): 3-5 mL<br>Older Children & Adults: 5-10 mL | https://www.allelediagnostics.com/ | | Alternate: Blueprint Short Stature Syndrome Panel [MA2101]. Does not include methylation study. | 02-Dec-17 |
| 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) | | |
| 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) | 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 \| | | 09-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) | | |

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

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| 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) | | |
| RETINOBLASTOMA VIA THE RB1 GENE [795] | | RB1 (614041) | Lavender Top (EDTA) | Prevention Genetics (www.preventiongenetics.com) | Retinoblastoma (180200) | | |
| 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 | | | 09-Nov-17 |
| RNA Polymerase III Antibodies, IgG, Serum [RNAP] | | | Serum (Gold SST or Red Top) | Mayo Clinical Laboratories (www.mayomedicallaboratories.com) | systemic sclerosis | | 04-Jul-19 |
| Russel Silver Syndrome: Methylation and Copy Number Analysis | 992 | | Lavender Top (EDTA) | Genome Diagnostics<br>The Department of Paediatric Laboratory Medicine<br>The Hospital for Sick Children | Russel-Silver syndrome (180860) | | 19-Feb-20 |
| Russel Silver Syndrome: UPD7 Analysis | 992 | | Lavender Top (EDTA) | Genome Diagnostics<br>The Department of Paediatric Laboratory Medicine<br>The Hospital for Sick Children | Russel-Silver syndrome (180860) | | 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) | | 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) | | |
| SANDHOFF DISEASE VIA THE HEXB GENE [476] | | HEXB (606873) | Lavender Top (EDTA) | Prevention Genetics (www.preventiongenetics.com) | Sandhoff Disease (268800) | | |
| SAP Protein Expression [XLP1] | 817 | SH2D1A (300490) | Heparin | Alberta Precision Laboratories (Flow Cytometry - Calgary) | X-linked lymphoproliferative syndrome (XLP1) (308240) | | 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 | | | 18-Apr-19 |
| SCN4A Full Gene sequencing Analysis [MOL355]; Paramyotonia Congenita (168300) | | SCN4A (603967) | Lavender Top (EDTA) | MNG Laboratories (www.medicalneurogenetics.com)/Dynacare | Paramyotonia Congenita (168300) | | 09-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) | | |
| 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 | |
| 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);<br>Leri Weill Dyschondrosteosis (127300);<br>Short Stature, Idiopathic, X-Linked (300582) | | 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 | | 09-Nov-17 |
| SLC12A3 DNA Sequencing Test [766] | | SLC12A3 (600968) | Lavender Top (EDTA) | Athena Diagnostics (www.athenadiagnostics.com) | Gitelman syndrome (9263800) | | |
| SLC27A4 Gene Sequencing [712] | | SLC27A4 (604194) | Lavender Top (EDTA) | GeneDx (www.genedx.com) | ichthyosis prematurity syndrome (608649( | | 09-Dec-19 |

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|---|---|---|---|---|---|---|---|
| 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) | Offered for free by MNG (The testing costs is billed directly to Ultragenyx Pharmaceutical who is developping a new treatment) | 09-May-20 |
| SLC40A1-Related Hereditary Hemochromatosis (606069) | | SLC40A1 (604653) | Lavender Top (EDTA) | Centogene AG (www.centogene.com) | ferroportin; Hereditary Hemochromatosis type IV (606069) | | |
| 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) | | |
| 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 | | | 13-Sep-18 |
| SOTOS SYNDROME VIA THE NSD1 GENE [132] | | NSD1 (606681) | Lavender Top (EDTA) | Prevention Genetics (www.preventiongenetics.com) | Sotos' Syndrome (117550) | | |
| 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 | | 09-May-20 |
| Spastic paraplegia 3A, autosomal dominant (182600) | 657 | ATL1 (606439) | Lavender Top (EDTA) | Centogene AG (www.centogene.com) | | | |

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|---|---|---|---|---|---|---|---|
| Spermatogenic Failure-6 (102530) | | PGF6 (SPATA16) (609856) | Lavender Top (EDTA) | Centogene AG (www.centogene.com) | | | |
| Spermatogenic Failure-9 (613958) | | DPY19L2 (613893) | Lavender Top (EDTA) | Centogene AG (www.centogene.com) | | | |
| 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 | | 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 | | | 09-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) | | | |
| Spinocerebellar Ataxia (SCA) Panel (1,2,3,6,7,8,17) | 584 | | Lavender Top (EDTA) | North York General | individuals tests $3780 | 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 | | 05-Aug-20 |
| Spinocerebellar ataxia 11 (604432) | 584 | TTBK2 (611695) | Lavender Top (EDTA) | Centogene AG (www.centogene.com) | | | |
| Spinocerebellar ataxia 17 (607136) | 584 | TBP (600075) | Lavender Top (EDTA) | North York General | SCA Panel available | | |
| Spinocerebellar ataxia 2 (183090) | 584 | ATXN2 (601517) | Lavender Top (EDTA) | North York General | SCA Panel available | | 05-Aug-20 |
| Spinocerebellar ataxia 6 (183086) | 584 | CACNA1A (601011) | Lavender Top (EDTA) | North York General | SCA Panel available | | |
| Spinocerebellar ataxia 7 (607640) | 584 | ATXN7 (607640) | Lavender Top (EDTA) | North York General | SCA Panel available | | |
| Spinocerebellar ataxia 8 (608768) | 584 | ATXN8 (613289) | Lavender Top (EDTA) | North York General | SCA Panel available | | |
| Spinocerebellar ataxia type 27 (609307) | 584 | FGF14 (601515) | Lavender Top (EDTA) | Centogene AG (www.centogene.com) | | | |
| 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) | | |
| 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) | | |

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|---|---|---|---|---|---|---|---|
| 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) | | | 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) | | | |
| 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) | | |
| 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) | | | 03-Apr-19 |
| 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) | | |
| 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) | | | 25-Jan-19 |
| 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°Ç or -20°C. | NMS Labs | | | 06-May-19 |
| Thiosulfate, Urine [4472U] | 920 | | Plastic container (no preservatives). **Store only at 4°C.** Stable for 30 days. Rejected if frozen. | NMS Labs | | | 06-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/Interpace Diagnostics | TERT available at no added cost | ON HOLD | 13-Nov-17 |
| ThyroSeq v.3 | 1680 | | Fine needle aspirate | Groupe TMTC/UPMC | | ON HOLD | 28-Nov-18 |
| 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) | | | 01-Feb-18 |
| Tissue Transglutaminase Abs (IgA & IgG) assay [TTIGAG] | | | Serum (Gold SST or Red Top) | Hospitals-In-Common | | | 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) | | | | 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) | | |
| TPSAB1 Copy Number Analysis | 1395 | | Lavender Top (EDTA) | Gene by Gene | | | 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 |

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| 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 | price per sample | 14-Jan-19 |
| Trypsinogen [TRGEN] | | | Serum. Store and send frozen | In-Common Laboratories | pancreatic dysfunction (e.g. CF) | | |
| 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) | | 05-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) | | 05-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) | | 05-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) | | 05-Feb-20 |
| UBE3A Full Gene Sequencing Analysis [MOL093] | | UBE3A (601623) | Lavender Top (EDTA) | MNG Laboratories (www.medicalneurogenetics.com)/Dynacare | Angelman Syndrome (105830) | Requires local methylation study | 14-May-20 |
| 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 | | 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) | | 02-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) | | | 28-Feb-19 |
| 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 | | 04-Apr-18 |
| 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 Verner-Morrison's (Watery Diarrhea) Syndrome | If paired to another test going to ISI | 09-Nov-17 |
| VGKC antibodies | 54 | LGI1 and Caspr2 (Voltage gated potassium chanel) | Gold SST; CSF | Mitogen Advanced Diagnostics | Paraneoplastic Disease | Not available: LGI1 and CASPR2 Antibodies Immunology Lab - Oxford U. Hospitals 80 UKP | 10-Apr-19 |
| Vitamin B1 [VITB1 WB] | 224 | | Whole Blood (EDTA); not SST. Collect fasting specimen. Protect from light within 1 hour of collection and during storage and transport. Store and send frozen. If the specimen thaws it is unsuitable for analysis. This assay measures the primary active form of Vitamin B1 (Thiamine diphosphate). | In-Common Laboratories | thiamin diphosphate; to assess body stores | plasma at CHUM | |
| Vitamin B3 Niacin in Plasma [FNIAC] | 218 | | Plasma EDTA. Store frozen. | Mayo Clinical Laboratories (www.mayomedicallaboratories.com) | | | 16-Apr-18 |

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| 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) | | | 12-Jun-20 |
| Vitamine D dependant rickets type1 (264700) | | CYP27B1 (609506) | Lavender Top (2 x 4 mL) | Fulgent Genetics (fulgentdiagnostics.com) | Vitamine D dependant rickets type1 (264700) | | |
| 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) | | 31-Oct-20 |
| Von Hippel-Lindau Disease via VHL Gene [7523] | 261 | VHL (608537) | Lavender Top (EDTA) | Prevention Genetics (www.preventiongenetics.com) | | | 31-Oct-20 |
| von Willebrand disease (193400) | | VWF (613160) | Lavender Top (EDTA) | ARUP Laboratories | | | |
| 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) | | |
| Weaver Syndrome (277590) | | EZH2 (601573) | Lavender Top (2 x 4 mL) | Fulgent Genetics (fulgentdiagnostics.com) | | | |
| WILSON DISEASE / HEPATOLENTICULAR DEGENERATION VIA THE ATP7B GENE [7871] | 1185 | ATP7B (606882) | Lavender Top (EDTA) | Prevention Genetics (www.preventiongenetics.com) | Wilson Disease (277900) | | 31-Oct-20 |
| Wolfram syndrome (222300) | | WFS1 (606201) | Lavender Top (EDTA) | GeneDx | | | 09-Dec-19 |
| XIAP Protein Expression [XLP2] | 817 | | Heparin | Alberta Precision Laboratories (Flow Cytometry - Calgary) | X-linked lymphoproliferative syndrome (XLP1) (308240) | | 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) | | | |
| 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) | | 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 |