

Appellation 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 (Déclaration MSSS)	SYMBOL HGNC DU GÈNE et/ou NOM OMIM DU GÈNE / HGNC Gene Symbol and OMIM Gene Number	RÈGLES DE MANUTENTION / Handling Conditions	LABORATOIRE DÉSIGNÉ / Designated laboratory	RENSEIGNEMENTS SUR L'ANALYSE / Test details	MÉTHODE / Methode	NOTE PARTICULIÈRE / Special comment	Date de la révision / Date of revision
1,5-anhydroglucitol (GlycoMark) [500115] [GLYC]	1465		Gold Top or Red Top (Serum) or Lavender (plasma). Centrifuge and aliquot serum to transport tube within one hour of collection. Store and ship frozen. Stable at -20°C for 14 days.	LabCorp/Dynacare (https://www.labcorp.com/)		Enzymatic, colorimetric assay.		10-Jan-21
15 Mold Panel	1465			In-Common Laboratories	Done at Alltest			12-Mar-20
2,3-Dinor-11Beta-Prostaglandin F2 Alpha, 24 Hour, Urine [23BPT]	1		Urine. No preservative. Collect for 24-h or random sample. Refrigerate 4°C during collection. Store frozen.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	systemic mastocytosis; mast cell activation disorders including systemic mastocytosis; Replaces 11 BETA-PROSTAGLANDIN F2 ALPHA	LC-MS/MS	May also order Leukotriene E4	04-Mar-21
5-Methyltetrahydrofolate (CSF) [NC01]			Collect 1 milliliter of CSF. Spin sample if contaminated with red blood cells and freeze the clear CSF at -80°Celsius. Store frozen at -80°Celsius.	MNG Laboratories (www.medicalneurogenetics.com)/Dynacare	Cerebral folate deficiency			14-May-20
5'Nucleotidase	4		Serum. Stable 4h at RT, 7 days at 4°C or 14 days at -20°C.	Dynacare		enzymatic	Requires preppova as tets is of limited utility	22-Oct-20
68KD (HSP-70) Antibodies [F68KD]	1280		Red Top (preferred); Gold SST. Store at -20°C.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	autoimmune hearing loss	No longer available at CHUQ: Anticorps anti-oreille interne (anticochleë)		03-May-22
8-hydrox acyclovir and 9-carboxymethy guanosine	1465		Serum. Red Top preferred over gel.	Karolinska University Hospital at Huddinge Clinical Pharmacology Laboratory, C2:69 SE-141 86 Stockholm		LC-MS/MS		08-Jan-22
a1-Acid glycoprotein [A1AGP]			Serum. Freeze if > 72 hours	In-Common Laboratories	Other names: Orosomucoid, Acid Glycoprotein, Alpha 1 Acid Glycoprotein	Nephelometry		06-Sep-18
ABCC6 Gene Analysis in Pseudoxanthoma Elasticum [2642]	1323	ABCC6(603234)	Lavender top (EDTA)	GeneDx (www.genedx.com)	Pseudoxanthoma Elasticum (264800)	Tier 1: sequencing for common mutations [2641]; Tier 2: full gene sequencing [2642]; del/dupl (MLPA) [906]		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)	18		Serum (Gold SST) ONLY. Store at -20°C.	BC Neuroimmunology (bcneuro.ca)	myasthenia gravis	RIPA - first performed 91020 qualitative test. If positive than 91021 quantitative test		09-Aug-22
Acetylcholine Receptor Antibodies with reflex to Muscle Specific Tyrosine Kinase Antibodies (MuSK Ab)	18		Serum (Gold SST) ONLY. Store at -20°C.	BC Neuroimmunology (bcneuro.ca)	myasthenia gravis	RIPA		09-Aug-22
Acyclovir, Serum/Plasma [0158SP]	1325		Red Top or Lavender Top (NOT SST). Store frozen at -20°C.	NMS Laboratories		LC-MS/MS		27-Apr-22
ADAR Full Gene Sequencing Analysis [MOL309]		ADAR (146920)	Lavender Top (EDTA)	MNG Laboratories (www.medicalneurogenetics.com)/Dynacare	Aicardi-goutieres syndrome 6 (615010); Dyschromatosis symmetrica hereditaria	Sanger sequencing	NGS Panel available	14-May-20
Adenosine Deaminase and Purine Nucleoside Phosphorylase	11		Guthere card + 1 mL EDTA plasma. Keep at RT.	Michael Hershfield, M.D. 418 Sands Building 303 Research Dr, Duke Univ Med Cent Durham, North Carolina 27710 USA Tel: 919-684-4184				12-Aug-22
Adenosine Deaminase, Pleural Fluid [FADPF]	11		Must be frozen within 24 hours of collection. No freeze/thaw cycles.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)		Ultraviolet Spectrophotometry		11-Feb-21
Adiponectin [FADIO]			Draw blood in a plain red-top or Gold SST, serum gel tube(s) is acceptable. Spin down and send 1 mL of serum refrigerated in a plastic vial. Store frozen. Overnight fasting is required.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)		ELISA	HIC: RUO	31-Oct-20
ADmark® Phospho-Tau/Total-Tau/Ab42 CSF Analysis & Interpretation (Symptomatic) [177]	596		Cerebrospinal Fluid (CSF), 2 mL. POLYPROPYLENE TUBES ONLY. Store at -20°C.	Athena Diagnostics (www.athenadiagnostics.com)	Alzheimer Disease (104300)	ELISA	Alertna site : MML Vancouver	03-Aug-18
Adrenal hyperplasia due to 21-hydroxylase deficiency (201910)	1326	CYP21A2(613815)	Lavender Top (EDTA)	Alberta Mol Dx Laboratory (Calgary)		sequencing & MLPA		07-Mar-22
Afirma GEC	1680		Fine needle aspirate	Groupe TMTC (https://grouptmtc.com)			ON HOLD. Requires approval by head of OOP	21-Jan-21

Appellation 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 / IGENC DU GÈNE et/ou NOM OMIM DU GÈNE / IGENC Gene Symbol and OMIM Gene Number	RÈGLES DE MANUTENTION / Handling Conditions	LABORATOIRE DÉSIGNÉ / Designated laboratory	RENSEIGNEMENTS SUR L'ANALYSE / Test details	MÉTHODE / Methode	NOTE PARTICULIÈRE / Special comment	Date de la révision / Date of revision
1,5-anhydroglucitol (GlycoMark) [500115] [GLYC]	1465		Gold Top or Red Top (Serum) or Lavender (plasma). Centrifuge and aliquot serum to transport tube within one hour of collection. Store and ship frozen. Stable at -20°C for 14 days.	LabCorp/Dynacare (https://www.labcorp.com/)		Enzymatic, colorimetric assay.		10-Jan-21
Agammaglobulinemia Panel, Sequencing (9 Genes) and Deletion/Duplication (6 Genes) [2011151]	999	BLNK, BTK, CD79B, CD79B, IGHM, IGLL1, LRRC5A, PIK3R1, SH2D1A	Lavender Top (EDTA)	ARUP Laboratories (www.aruplab.com)	Agammaglobulinemia 1, Autosomal Recessive - X-Linked Agammaglobulinemia - SH2D1A-Related Lymphoproliferative Disease, X-Linked - Agammaglobulinemia 2, Autosomal Recessive - Agammaglobulinemia 3, Autosomal Recessive - Agammaglobulinemia 4, Autosomal Recessive - Agammaglobulinemia 5, Autosomal Dominant - Agammaglobulinemia 6, Autosomal Recessive - Agammaglobulinemia 7, Autosomal Recessive	Massively Parallel Sequencing/Exonic Oligonucleotide-based CGH Microarray		
AGXT Gene, Full Gene Analysis [AGXMS]		AGXT (60428)	Lavender Top (EDTA)	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	Hyperoxaluria, primary, type 1 (259900)	Sanger sequencing and MLPA		
Aicardi-Goutieres Syndrome (NextGen Sequencing Panel and Copy Number Analysis; 6 Genes) [NGS344]		ADAR (608099); ALDH7A1 (107323); RNASEH2A (608046); RNASEH2B (610326); RNASEH2C (608336); SAMHD1 (606754); TREM1 (606699)	Lavender Top (EDTA)	MNG Laboratories (www.medicalneurogenetics.com)/Dynacare	Aicardi-Goutieres Syndrome	NextGen Sequencing		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		RED TOP ONLY. Serum without gel, centrifuge immediately after collection, freeze and ship with dry ice	Inselspital Bern Freiburgstrasse 10 Zentrum für Labormedizin Zentrale Annahme INOF / z. Hd. Y. Aebi CH-3010 Bern, Switzerland	Measures albendazole sulfoxide due to 1st pass effects			06-Apr-21
Albright's hereditary Osteodystrophy (103580)	440	GNAS (13923)	Lavender Top (EDTA) 2 x 4 mL	Fulgent Genetics (fulgentdiagnostics.com)	Albright's hereditary Osteodystrophy (103580)	NextGen Sequencing		28-Feb-20
Alexander Disease via the GFAP Gene [3775]		GFAP (137780)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Alexander disease (203450)	Sanger sequencing		17-Mar-17
Alpha aminoacidic semialdehyde (Urine) [MET20]	12	Alpha-aminoacidic semialdehyde	Collect 1 milliliter urine (random) and freeze at -20°Celsius. Store frozen at -20°Celsius and ship frozen.	MNG Laboratories (www.medicalneurogenetics.com)/Dynacare	Pyridoxine-dependent epilepsy (266100)	MS/MS		14-May-20
Alpha aminoacidic semialdehyde (Whole Blood) [AASA]	12	Alpha-aminoacidic semialdehyde	Specimen: Whole Blood Container(s): Dark Green/Sodium Heparin or Lt. Green/Lithium Heparin Tube Reject due to: If sample is not spun and frozen within 1 hour of collection. 48 hr storage at -20 C is acceptable. Store plasma at -70°C.	Seattle Children's Hospital. http://seattlechildrens.testcatalog.org/	Pyridoxine-dependent epilepsy (266100)			27-Jan-20
ALPHA-1-ANTITRYPSIN DEFICIENCY (613490)	1395	SERPINA1 (104400)	Lavender Top (EDTA)	Attn: Norine Freedman or Lynn Coleman Special Chemistry Laboratory St Paul's Hospital 1081 Burrard Street Vancouver, B.C. V6Z 1Y6. Contact Dr. A. Mattman BEFORE sending sample.	ALPHA-1-ANTITRYPSIN DEFICIENCY (613490)	Sanger sequencing		
ALPHA-1-ANTITRYPSIN DEFICIENCY (613490)	1395	SERPINA1 (104400)	Lavender Top (EDTA)	Fulgent Genetics (fulgentdiagnostics.com)	ALPHA-1-ANTITRYPSIN DEFICIENCY (613490)		Preferred	03-Feb-22
alpha-actin (skeletal muscle form)-related myopathy via the ACTA1 gene [358]		ACTA1 (102610)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	neuromuscular myopathy (NEM); OMIM #161800) and congenital fiber-type disproportion (CFTD); OMIM #255310)			
Alpha-aminoacidic semialdehyde (CSF) [NC08]	12	Alpha-aminoacidic 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)/Dynacare	Pyridoxine-dependent epilepsy (266100)	MS/MS	Seattle for Heparin plasma	14-May-20
Alpha-Subunit Pituitary Tumor Marker, Serum [APGH]	55		Red Top	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	Alternate name: Alpha Glycoprotein Subunit			12-Sep-22
Alpha-Thalassemia (604131)	826	HBA1 (141800); HBA2 (141850)	Lavender Top (EDTA)	Molecular Genetics Laboratory - McMaster University Medical Centre	targeted mutations at HSI	Sanger sequencing & del/dupl		
Alport Syndrome NGS Panel	785	COL4A3, COL4A4, COL4A5, COL4A6	Lavender Top (EDTA)	Fulgent Genetics (fulgentdiagnostics.com)		NextGen Sequencing		03-Feb-20
ALZHEIMER'S DISEASE, FAMILIAL VIA THE APP GENE (604)		APP (104760)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)		Sanger sequencing	For known mutations only. Otherwise use Fulgent panel.	
ALZHEIMER'S DISEASE, FAMILIAL VIA THE PSEN1 GENE [1414]		PSEN1 (104311)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Alzheimer Disease, Type 3 (607882); Frontotemporal Dementia (600274)	Sanger sequencing	For known mutations only. Otherwise use Fulgent panel.	
ALZHEIMER'S DISEASE, FAMILIAL VIA THE PSEN2 GENE (1415)		PSEN2 (600799)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Alzheimer Disease, Type 4 (606889)	Sanger sequencing	For known mutations only. Otherwise use Fulgent panel.	
AMELOGENESIS IMPERFECTA VIA THE DLX3 GENE [1603]		DLX3 (600525)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Tricho-Dento-Osseous Syndrome (190320); Amelogenesis Imperfecta, Type IV (104510)	Sanger sequencing		
Aminolevulinic Acid Dehydratase (ALAD), Whole Blood [ALAD]			Green Top, 4°C only	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	Aminolevulinic Acid Dehydratase Deficiency Porphyria (612740)			31-Oct-20

Appellation 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/ou NOM OMIM DU GÈNE / HGNC Gene Symbol et/ou OMIM Gene Number	RÈGLES DE MANUTENTION / Handling Conditions	LABORATOIRE DÉSIGNÉ / Designated laboratory	RENSEIGNEMENTS SUR L'ANALYSE / Test details	MÉTHODE / Methode	NOTE PARTICULIÈRE / Special comment	Date de la révision / Date of revision
1,5-anhydroglucitol (GlycoMark) [500115] [GLYC]	1465		Gold Top or Red Top (Serum) or Lavender (plasma). Centrifuge and aliquot serum to transport tube within one hour of collection. Store and ship frozen. Stable at -20°C for 14 days.	LabCorp/Dynacare (https://www.labcorp.com/)		Enzymatic, colorimetric assay.		10-Jan-21
AMYLOID PROTEIN ID, PAR, LC MS/MS [AMPIP]	1123		Paraffin section	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)		LC-MS/MS	ADD CHARGE FOR MICRODISSECTION	06-Jul-18
Amyotrophic Lateral Sclerosis and Frontotemporal Dementia Panel [10359]	853	ANG ANXA1APP ARHGEP28 ATXN2 C9orf72 CFAP10 CHCHD10 CHMP2B DAO DCTN1 ERBB4 FUS FUS GEN HSNRP1 HSNRP2 HSNRP3 HSNRP4 HSNRP5 HSNRP6 HSNRP7 HSNRP8 HSNRP9 HSNRP10 HSNRP11 HSNRP12 HSNRP13 HSNRP14 HSNRP15 HSNRP16 HSNRP17 HSNRP18 HSNRP19 HSNRP20 HSNRP21 HSNRP22 HSNRP23 HSNRP24 HSNRP25 HSNRP26 HSNRP27 HSNRP28 HSNRP29 HSNRP30 HSNRP31 HSNRP32 HSNRP33 HSNRP34 HSNRP35 HSNRP36 HSNRP37 HSNRP38 HSNRP39 HSNRP40 HSNRP41 HSNRP42 HSNRP43 HSNRP44 HSNRP45 HSNRP46 HSNRP47 HSNRP48 HSNRP49 HSNRP50 HSNRP51 HSNRP52 HSNRP53 HSNRP54 HSNRP55 HSNRP56 HSNRP57 HSNRP58 HSNRP59 HSNRP60 HSNRP61 HSNRP62 HSNRP63 HSNRP64 HSNRP65 HSNRP66 HSNRP67 HSNRP68 HSNRP69 HSNRP70 HSNRP71 HSNRP72 HSNRP73 HSNRP74 HSNRP75 HSNRP76 HSNRP77 HSNRP78 HSNRP79 HSNRP80 HSNRP81 HSNRP82 HSNRP83 HSNRP84 HSNRP85 HSNRP86 HSNRP87 HSNRP88 HSNRP89 HSNRP90 HSNRP91 HSNRP92 HSNRP93 HSNRP94 HSNRP95 HSNRP96 HSNRP97 HSNRP98 HSNRP99 HSNRP100 HSNRP101 HSNRP102 HSNRP103 HSNRP104 HSNRP105 HSNRP106 HSNRP107 HSNRP108 HSNRP109 HSNRP110 HSNRP111 HSNRP112 HSNRP113 HSNRP114 HSNRP115 HSNRP116 HSNRP117 HSNRP118 HSNRP119 HSNRP120 HSNRP121 HSNRP122 HSNRP123 HSNRP124 HSNRP125 HSNRP126 HSNRP127 HSNRP128 HSNRP129 HSNRP130 HSNRP131 HSNRP132 HSNRP133 HSNRP134 HSNRP135 HSNRP136 HSNRP137 HSNRP138 HSNRP139 HSNRP140 HSNRP141 HSNRP142 HSNRP143 HSNRP144 HSNRP145 HSNRP146 HSNRP147 HSNRP148 HSNRP149 HSNRP150 HSNRP151 HSNRP152 HSNRP153 HSNRP154 HSNRP155 HSNRP156 HSNRP157 HSNRP158 HSNRP159 HSNRP160 HSNRP161 HSNRP162 HSNRP163 HSNRP164 HSNRP165 HSNRP166 HSNRP167 HSNRP168 HSNRP169 HSNRP170 HSNRP171 HSNRP172 HSNRP173 HSNRP174 HSNRP175 HSNRP176 HSNRP177 HSNRP178 HSNRP179 HSNRP180 HSNRP181 HSNRP182 HSNRP183 HSNRP184 HSNRP185 HSNRP186 HSNRP187 HSNRP188 HSNRP189 HSNRP190 HSNRP191 HSNRP192 HSNRP193 HSNRP194 HSNRP195 HSNRP196 HSNRP197 HSNRP198 HSNRP199 HSNRP200 HSNRP201 HSNRP202 HSNRP203 HSNRP204 HSNRP205 HSNRP206 HSNRP207 HSNRP208 HSNRP209 HSNRP210 HSNRP211 HSNRP212 HSNRP213 HSNRP214 HSNRP215 HSNRP216 HSNRP217 HSNRP218 HSNRP219 HSNRP220 HSNRP221 HSNRP222 HSNRP223 HSNRP224 HSNRP225 HSNRP226 HSNRP227 HSNRP228 HSNRP229 HSNRP230 HSNRP231 HSNRP232 HSNRP233 HSNRP234 HSNRP235 HSNRP236 HSNRP237 HSNRP238 HSNRP239 HSNRP240 HSNRP241 HSNRP242 HSNRP243 HSNRP244 HSNRP245 HSNRP246 HSNRP247 HSNRP248 HSNRP249 HSNRP250 HSNRP251 HSNRP252 HSNRP253 HSNRP254 HSNRP255 HSNRP256 HSNRP257 HSNRP258 HSNRP259 HSNRP260 HSNRP261 HSNRP262 HSNRP263 HSNRP264 HSNRP265 HSNRP266 HSNRP267 HSNRP268 HSNRP269 HSNRP270 HSNRP271 HSNRP272 HSNRP273 HSNRP274 HSNRP275 HSNRP276 HSNRP277 HSNRP278 HSNRP279 HSNRP280 HSNRP281 HSNRP282 HSNRP283 HSNRP284 HSNRP285 HSNRP286 HSNRP287 HSNRP288 HSNRP289 HSNRP290 HSNRP291 HSNRP292 HSNRP293 HSNRP294 HSNRP295 HSNRP296 HSNRP297 HSNRP298 HSNRP299 HSNRP300 HSNRP301 HSNRP302 HSNRP303 HSNRP304 HSNRP305 HSNRP306 HSNRP307 HSNRP308 HSNRP309 HSNRP310 HSNRP311 HSNRP312 HSNRP313 HSNRP314 HSNRP315 HSNRP316 HSNRP317 HSNRP318 HSNRP319 HSNRP320 HSNRP321 HSNRP322 HSNRP323 HSNRP324 HSNRP325 HSNRP326 HSNRP327 HSNRP328 HSNRP329 HSNRP330 HSNRP331 HSNRP332 HSNRP333 HSNRP334 HSNRP335 HSNRP336 HSNRP337 HSNRP338 HSNRP339 HSNRP340 HSNRP341 HSNRP342 HSNRP343 HSNRP344 HSNRP345 HSNRP346 HSNRP347 HSNRP348 HSNRP349 HSNRP350 HSNRP351 HSNRP352 HSNRP353 HSNRP354 HSNRP355 HSNRP356 HSNRP357 HSNRP358 HSNRP359 HSNRP360 HSNRP361 HSNRP362 HSNRP363 HSNRP364 HSNRP365 HSNRP366 HSNRP367 HSNRP368 HSNRP369 HSNRP370 HSNRP371 HSNRP372 HSNRP373 HSNRP374 HSNRP375 HSNRP376 HSNRP377 HSNRP378 HSNRP379 HSNRP380 HSNRP381 HSNRP382 HSNRP383 HSNRP384 HSNRP385 HSNRP386 HSNRP387 HSNRP388 HSNRP389 HSNRP390 HSNRP391 HSNRP392 HSNRP393 HSNRP394 HSNRP395 HSNRP396 HSNRP397 HSNRP398 HSNRP399 HSNRP400 HSNRP401 HSNRP402 HSNRP403 HSNRP404 HSNRP405 HSNRP406 HSNRP407 HSNRP408 HSNRP409 HSNRP410 HSNRP411 HSNRP412 HSNRP413 HSNRP414 HSNRP415 HSNRP416 HSNRP417 HSNRP418 HSNRP419 HSNRP420 HSNRP421 HSNRP422 HSNRP423 HSNRP424 HSNRP425 HSNRP426 HSNRP427 HSNRP428 HSNRP429 HSNRP430 HSNRP431 HSNRP432 HSNRP433 HSNRP434 HSNRP435 HSNRP436 HSNRP437 HSNRP438 HSNRP439 HSNRP440 HSNRP441 HSNRP442 HSNRP443 HSNRP444 HSNRP445 HSNRP446 HSNRP447 HSNRP448 HSNRP449 HSNRP450 HSNRP451 HSNRP452 HSNRP453 HSNRP454 HSNRP455 HSNRP456 HSNRP457 HSNRP458 HSNRP459 HSNRP460 HSNRP461 HSNRP462 HSNRP463 HSNRP464 HSNRP465 HSNRP466 HSNRP467 HSNRP468 HSNRP469 HSNRP470 HSNRP471 HSNRP472 HSNRP473 HSNRP474 HSNRP475 HSNRP476 HSNRP477 HSNRP478 HSNRP479 HSNRP480 HSNRP481 HSNRP482 HSNRP483 HSNRP484 HSNRP485 HSNRP486 HSNRP487 HSNRP488 HSNRP489 HSNRP490 HSNRP491 HSNRP492 HSNRP493 HSNRP494 HSNRP495 HSNRP496 HSNRP497 HSNRP498 HSNRP499 HSNRP500 HSNRP501 HSNRP502 HSNRP503 HSNRP504 HSNRP505 HSNRP506 HSNRP507 HSNRP508 HSNRP509 HSNRP510 HSNRP511 HSNRP512 HSNRP513 HSNRP514 HSNRP515 HSNRP516 HSNRP517 HSNRP518 HSNRP519 HSNRP520 HSNRP521 HSNRP522 HSNRP523 HSNRP524 HSNRP525 HSNRP526 HSNRP527 HSNRP528 HSNRP529 HSNRP530 HSNRP531 HSNRP532 HSNRP533 HSNRP534 HSNRP535 HSNRP536 HSNRP537 HSNRP538 HSNRP539 HSNRP540 HSNRP541 HSNRP542 HSNRP543 HSNRP544 HSNRP545 HSNRP546 HSNRP547 HSNRP548 HSNRP549 HSNRP550 HSNRP551 HSNRP552 HSNRP553 HSNRP554 HSNRP555 HSNRP556 HSNRP557 HSNRP558 HSNRP559 HSNRP560 HSNRP561 HSNRP562 HSNRP563 HSNRP564 HSNRP565 HSNRP566 HSNRP567 HSNRP568 HSNRP569 HSNRP570 HSNRP571 HSNRP572 HSNRP573 HSNRP574 HSNRP575 HSNRP576 HSNRP577 HSNRP578 HSNRP579 HSNRP580 HSNRP581 HSNRP582 HSNRP583 HSNRP584 HSNRP585 HSNRP586 HSNRP587 HSNRP588 HSNRP589 HSNRP590 HSNRP591 HSNRP592 HSNRP593 HSNRP594 HSNRP595 HSNRP596 HSNRP597 HSNRP598 HSNRP599 HSNRP600 HSNRP601 HSNRP602 HSNRP603 HSNRP604 HSNRP605 HSNRP606 HSNRP607 HSNRP608 HSNRP609 HSNRP610 HSNRP611 HSNRP612 HSNRP613 HSNRP614 HSNRP615 HSNRP616 HSNRP617 HSNRP618 HSNRP619 HSNRP620 HSNRP621 HSNRP622 HSNRP623 HSNRP624 HSNRP625 HSNRP626 HSNRP627 HSNRP628 HSNRP629 HSNRP630 HSNRP631 HSNRP632 HSNRP633 HSNRP634 HSNRP635 HSNRP636 HSNRP637 HSNRP638 HSNRP639 HSNRP640 HSNRP641 HSNRP642 HSNRP643 HSNRP644 HSNRP645 HSNRP646 HSNRP647 HSNRP648 HSNRP649 HSNRP650 HSNRP651 HSNRP652 HSNRP653 HSNRP654 HSNRP655 HSNRP656 HSNRP657 HSNRP658 HSNRP659 HSNRP660 HSNRP661 HSNRP662 HSNRP663 HSNRP664 HSNRP665 HSNRP666 HSNRP667 HSNRP668 HSNRP669 HSNRP670 HSNRP671 HSNRP672 HSNRP673 HSNRP674 HSNRP675 HSNRP676 HSNRP677 HSNRP678 HSNRP679 HSNRP680 HSNRP681 HSNRP682 HSNRP683 HSNRP684 HSNRP685 HSNRP686 HSNRP687 HSNRP688 HSNRP689 HSNRP690 HSNRP691 HSNRP692 HSNRP693 HSNRP694 HSNRP695 HSNRP696 HSNRP697 HSNRP698 HSNRP699 HSNRP700 HSNRP701 HSNRP702 HSNRP703 HSNRP704 HSNRP705 HSNRP706 HSNRP707 HSNRP708 HSNRP709 HSNRP710 HSNRP711 HSNRP712 HSNRP713 HSNRP714 HSNRP715 HSNRP716 HSNRP717 HSNRP718 HSNRP719 HSNRP720 HSNRP721 HSNRP722 HSNRP723 HSNRP724 HSNRP725 HSNRP726 HSNRP727 HSNRP728 HSNRP729 HSNRP730 HSNRP731 HSNRP732 HSNRP733 HSNRP734 HSNRP735 HSNRP736 HSNRP737 HSNRP738 HSNRP739 HSNRP740 HSNRP741 HSNRP742 HSNRP743 HSNRP744 HSNRP745 HSNRP746 HSNRP747 HSNRP748 HSNRP749 HSNRP750 HSNRP751 HSNRP752 HSNRP753 HSNRP754 HSNRP755 HSNRP756 HSNRP757 HSNRP758 HSNRP759 HSNRP760 HSNRP761 HSNRP762 HSNRP763 HSNRP764 HSNRP765 HSNRP766 HSNRP767 HSNRP768 HSNRP769 HSNRP770 HSNRP771 HSNRP772 HSNRP773 HSNRP774 HSNRP775 HSNRP776 HSNRP777 HSNRP778 HSNRP779 HSNRP780 HSNRP781 HSNRP782 HSNRP783 HSNRP784 HSNRP785 HSNRP786 HSNRP787 HSNRP788 HSNRP789 HSNRP790 HSNRP791 HSNRP792 HSNRP793 HSNRP794 HSNRP795 HSNRP796 HSNRP797 HSNRP798 HSNRP799 HSNRP800 HSNRP801 HSNRP802 HSNRP803 HSNRP804 HSNRP805 HSNRP806 HSNRP807 HSNRP808 HSNRP809 HSNRP810 HSNRP811 HSNRP812 HSNRP813 HSNRP814 HSNRP815 HSNRP816 HSNRP817 HSNRP818 HSNRP819 HSNRP820 HSNRP821 HSNRP822 HSNRP823 HSNRP824 HSNRP825 HSNRP826 HSNRP827 HSNRP828 HSNRP829 HSNRP830 HSNRP831 HSNRP832 HSNRP833 HSNRP834 HSNRP835 HSNRP836 HSNRP837 HSNRP838 HSNRP839 HSNRP840 HSNRP841 HSNRP842 HSNRP843 HSNRP844 HSNRP845 HSNRP846 HSNRP847 HSNRP848 HSNRP849 HSNRP850 HSNRP851 HSNRP852 HSNRP853 HSNRP854 HSNRP855 HSNRP856 HSNRP857 HSNRP858 HSNRP859 HSNRP860 HSNRP861 HSNRP862 HSNRP863 HSNRP864 HSNRP865 HSNRP866 HSNRP867 HSNRP868 HSNRP869 HSNRP870 HSNRP871 HSNRP872 HSNRP873 HSNRP874 HSNRP875 HSNRP876 HSNRP877 HSNRP878 HSNRP879 HSNRP880 HSNRP881 HSNRP882 HSNRP883 HSNRP884 HSNRP885 HSNRP886 HSNRP887 HSNRP888 HSNRP889 HSNRP890 HSNRP891 HSNRP892 HSNRP893 HSNRP894 HSNRP895 HSNRP896 HSNRP897 HSNRP898 HSNRP899 HSNRP900 HSNRP901 HSNRP902 HSNRP903 HSNRP904 HSNRP905 HSNRP906 HSNRP907 HSNRP908 HSNRP909 HSNRP910 HSNRP911 HSNRP912 HSNRP913 HSNRP914 HSNRP915 HSNRP916 HSNRP917 HSNRP918 HSNRP919 HSNRP920 HSNRP921 HSNRP922 HSNRP923 HSNRP924 HSNRP925 HSNRP926 HSNRP927 HSNRP928 HSNRP929 HSNRP930 HSNRP931 HSNRP932 HSNRP933 HSNRP934 HSNRP935 HSNRP936 HSNRP937 HSNRP938 HSNRP939 HSNRP940 HSNRP941 HSNRP942 HSNRP943 HSNRP944 HSNRP945 HSNRP946 HSNRP947 HSNRP948 HSNRP949 HSNRP950 HSNRP951 HSNRP952 HSNRP953 HSNRP954 HSNRP955 HSNRP956 HSNRP957 HSNRP958 HSNRP959 HSNRP960 HSNRP961 HSNRP962 HSNRP963 HSNRP964 HSNRP965 HSNRP966 HSNRP967 HSNRP968 HSNRP969 HSNRP970 HSNRP971 HSNRP972 HSNRP973 HSNRP974 HSNRP975 HSNRP976 HSNRP977 HSNRP978 HSNRP979 HSNRP980 HSNRP981 HSNRP982 HSNRP983 HSNRP984 HSNRP985 HSNRP986 HSNRP987 HSNRP988 HSNRP989 HSNRP990 HSNRP991 HSNRP992 HSNRP993 HSNRP994 HSNRP995 HSNRP996 HSNRP997 HSNRP998 HSNRP999 HSNRP1000	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Frontotemporal Dementia And/Or Amyotrophic Lateral Sclerosis (105550); Amyotrophic Lateral Sclerosis Type 6 (608030); Amyotrophic Lateral Sclerosis Type 9 (611895); Amyotrophic Lateral Sclerosis Type 1 (105400); Amyotrophic Lateral Sclerosis Type 10 (612069); Amyotrophic Lateral Sclerosis Type 12 (613435)	NextGen Sequencing		02-Nov-20
Amyotrophic Lateral Sclerosis NGS Panel	853	ABCD1, ABHD12, ALS2, ANG, ARHGEP28, ATXN1, ATXN2, C9orf72, CHCHD10, CHER, CHMP2B, CFAP10, DAO, DCTN1, ERBB4, FUS, FUS, GEN, HSNRP1, HSNRP2, HSNRP3, HSNRP4, HSNRP5, HSNRP6, HSNRP7, HSNRP8, HSNRP9, HSNRP10, HSNRP11, HSNRP12, HSNRP13, HSNRP14, HSNRP15, HSNRP16, HSNRP17, HSNRP18, HSNRP19, HSNRP20, HSNRP21, HSNRP22, HSNRP23, HSNRP24, HSNRP25, HSNRP26, HSNRP27, HSNRP28, HSNRP29, HSNRP30, HSNRP31, HSNRP32, HSNRP33, HSNRP34, HSNRP35, HSNRP36, HSNRP37, HSNRP38, HSNRP39, HSNRP40, HSNRP41, HSNRP42, HSNRP43, HSNRP44, HSNRP45, HSNRP46, HSNRP47, HSNRP48, HSNRP49, HSNRP50, HSNRP51, HSNRP52, HSNRP53, HSNRP54, HSNRP55, HSNRP56, HSNRP57, HSNRP58, HSNRP59, HSNRP60, HSNRP61, HSNRP62, HSNRP63, HSNRP64, HSNRP65, HSNRP66, HSNRP67, HSNRP68, HSNRP69, HSNRP70, HSNRP71, HSNRP72, HSNRP73, HSNRP74, HSNRP75, HSNRP76, HSNRP77, HSNRP78, HSNRP79, HSNRP80, HSNRP81, HSNRP82, HSNRP83, HSNRP84, HSNRP85, HSNRP86, HSNRP87, HSNRP88, HSNRP89, HSNRP90, HSNRP91, HSNRP92, HSNRP93, HSNRP94, HSNRP95, HSNRP96, HSNRP97, HSNRP98, HSNRP99, HSNRP100, HSNRP101, HSNRP102, HSNRP103, HSNRP104, HSNRP105, HSNRP106, HSNRP107, HSNRP108, HSNRP109, HSNRP110, HSNRP111, HSNRP112, HSNRP113, HSNRP114, HSNRP115, HSNRP116, HSNRP117, HSNRP118, HSNRP119, HSNRP120, HSNRP121, HSNRP122, HSNRP123, HSNRP124, HSNRP125, HSNRP126, HSNRP127, HSNRP128, HSNRP129, HSNRP130, HSNRP131, HSNRP132, HSNRP133, HSNRP134, HSNRP135, HSNRP136, HSNRP137, HSNRP138, HSNRP139, HSNRP140, HSNRP141, HSNRP142, HSNRP143, HSNRP144, HSNRP145, HSNRP146, HSNRP147, HSNRP148, HSNRP149, HSNRP150, HSNRP151, HSNRP152, HSNRP153, HSNRP154, HSNRP155, HSNRP156, HSNRP157, HSNRP158, HSNRP159, HSNRP160, HSNRP161, HSNRP162, HSNRP163, HSNRP164, HSNRP165, HSNRP166, HSNRP167, HSNRP168, HSNRP169, HSNRP170, HSNRP171, HSNRP172, HSNRP173, HSNRP174, HSNRP175, HSNRP176, HSNRP177, HSNRP178, HSNRP179, HSNRP180, HSNRP181, HSNRP182, HSNRP183, HSNRP184, HSNRP185, HSNRP186, HSNRP187, HSNRP188, HSNRP189, HSNRP190, HSNRP191, HSNRP192, HSNRP193, HSNRP194, HSNRP195, HSNRP196, HSNRP197, HSNRP198, HSNRP199, HSNRP200, HSNRP201, HSNRP202, HSNRP203, HSNRP204, HSNRP205, HSNRP206, HSNRP207, HSNRP208, HSNRP209, HSNRP210, HSNRP211, HSNRP212, HSNRP213, HSNRP214, HSNRP215, HSNRP216, HSNRP217, HSNRP218, HSNRP219, HSNRP220, HSNRP221, HSNRP222, HSNRP223, HSNRP224, HSNRP225, HSNRP226, HSNRP227, HSNRP228, HSNRP229, HSNRP230, HSNRP231, HSNRP232, HSNRP233, HSNRP234, HSNRP235, HSNRP236, HSNRP237, HSNRP238, HSNRP239, HSNRP240, HSNRP241, HSNRP242, HSNRP243, HSNRP244, HSNRP245, HSNRP246, HSNRP247, HSNRP248, HSNRP249, HSNRP250, HSNRP251, HSNRP252, HSNRP253, HSNRP254, HSNRP255, HSNRP256, HSNRP257, HSNRP258, HSNRP259, HSNRP260, HSNRP261, HSNRP262, HSNRP263, HSNRP264, HSNRP265, HSNRP266, HSNRP267, HSNRP268, HSNRP269, HSNRP270, HSNRP271, HSNRP272, HSNRP273, HSNRP274, HSNRP275, HSNRP276, HSNRP277, HSNRP278, HSNRP279, HSNRP280, HSNRP281, HSNRP282, HSNRP283, HSNRP284, HSNRP285, HSNRP286, HSNRP287, HSNRP288, HSNRP289, HSNRP290, HSNRP291, HSNRP292, HSNRP293, HSNRP294, HSNRP295, HSNRP296, HSNRP297, HSNRP298, HSNRP299, HSNRP300, HSNRP301, HSNRP302, HSNRP303, HSNRP304, HSNRP305, HSNRP306, HSNRP307, HSNRP308, HSNRP309, HSNRP310, HSNRP311, HSNRP312, HSNRP313, HSNRP314, HSNRP315, HSNRP316, HSNRP317, HSNRP318, HSNRP319, HSNRP320, HSNRP321, HSNRP322, HSNRP323, HSNRP324, HSNRP325, HSNRP326, HSNRP327, HSNRP328, HSNRP329, HSNRP330, HSNRP331, HSNRP332, HSNRP333, HSNRP334, HSNRP335, HSNRP336, HSNRP337, HSNRP338, HSNRP339, HSNRP340, HSNRP341, HSNRP342, HSNRP343, HSNRP344, HSNRP345, HSNRP346, HSNRP347, HSNRP348, HSNRP349, HSNRP350, HSNRP351, HSNRP352, HSNRP353, HSNRP354, HSNRP355, HSNRP356, HSNRP357, HSNRP358, HSNRP359, HSNRP360, HSNRP361, HSNRP362, HSNRP363, HSNRP364, HSNRP365, HSNRP366, HSNRP367, HSNRP368, HSNRP369, HSNRP370, HSNRP371, HSNRP372, HSNRP373, HSNRP374, HSNRP375, HSNRP376, HSNRP377, HSNRP378, HSNRP379, HSNRP380, HSNRP381, HSNRP382, HSNRP383, HSNRP384, HSNRP385, HSNRP386, HSNRP387, HSNRP388, HSNRP389, HSNRP390, HSNRP391, HSNRP392, HSNRP393, HSNRP394, HSNRP395, HSNRP396, HSNRP397, HSNRP398, HSNRP399, HSNRP400, HSNRP401, HSNRP402, HSNRP403, HSNRP404, HSNRP405, HSNRP406, HSNRP407, HSNRP408, HSNRP409, HSNRP410, HSNRP411, HSNRP412, HSNRP413, HSNRP414, HSNRP415, HSNRP416, HSNRP417, HSNRP418, HSNRP419, HSNRP420, HSNRP421, HSNRP422, HSNRP423, HSNRP424, HSNRP425, HSNRP426, HSNRP427, HSNRP428, HSNRP429, HSNRP430, HSNRP431, HSNRP432, HSNRP433, HSNRP434, HSNRP435, HSNRP436, HSNRP437, HSNRP438, HSNRP439, HSNRP440, HSNRP441, HSNRP442, HSNRP443, HSNRP444, HSNRP445, HSNRP446, HSNRP447, HSNRP448, HSNRP449, HSNRP450, HSNRP451, HSNRP452, HSNRP453, HSNRP454, HSNRP455, HSNRP456, HSNRP457, HSNRP458, HSNRP459, HSNRP460, HSNRP461, HSNRP462, HSNRP463, HSNRP464, HSNRP465, HSNRP466, HSNRP467, HSNRP468, HSNRP469, HSNRP470, HSNRP471, HSNRP472, HSNRP473, HSNRP474, HSNRP475, HSNRP476, HSNRP477, HSNRP478, HSNRP479, HSNRP480, HSNRP481, HSNRP482, HSNRP483, HSNRP484, HSNRP485, HSNRP486, HSNRP487, HSNRP488, HSNRP489, HSNRP490, HSNRP491, HSNRP492, HSNRP493, HSNRP494, HSNRP495, HSNRP496, HSNRP497, HSNRP498, HSNRP499, HSNRP500, HSNRP501, HSNRP502, HSNRP503, HSNRP504, HSNRP505, HSNRP506, HSNRP507, HSNRP508, HSNRP509, HSNRP510, HSNRP511, HSNRP512, HSNRP513, HSNRP514, HSNRP515, HSNRP516, HSNRP517, HSNRP518, HSNRP519, HSNRP520, HSNRP521, HSNRP522, HSNRP523, HSNRP524, HSNRP525, HSNRP526, HSNRP527, HSNRP528, HSNRP529, HSNRP530, HSNRP531, HSNRP532, HSNRP533, HSNRP534, HSNRP535, HSNRP536, HSNRP537, HSNRP538, HSNRP539, HSNRP540, HSNRP541, HSNRP542, HSNRP543, HSNRP544, HSNRP545, HSNRP546, HSNRP547, HSNRP548, HSNRP549, HSNRP550, HSNRP551, HSNRP552, HSNRP553, HSNRP554, HSNRP555, HSNRP556, HSNRP557, HSNRP558, HSNRP559, HSNRP560, HSNRP561, HSNRP562, HSNRP563, HSNRP564, HSNRP565, HSNRP566, HSNRP567, HSNRP568, HSNRP569, HSNRP570, HSNRP571, HSNRP572, HSNRP573, HSNRP574, HSNRP575, HSNRP576, HSNRP577, HSNRP578, HSNRP579, HSNRP580, HSNRP581, HSNRP582, HSNRP583, HSNRP584, HSNRP585, HSNRP586, HSNRP587, HSNRP588, HSNRP589, HSNRP590, HSNRP591, HSNRP592, HSNRP593, HSNRP594, HSNRP595, HSNRP596, HSNRP597, HSNRP598, HSNRP599, HSNRP600, HSNRP601, HSNRP602, HSNRP603, HSNRP604, HSNRP605, HSNRP606, HSNRP607, HSNRP608, HSNRP609, HSNRP610, HSNRP611, HSNRP612, HSNRP613, HSNRP614, HSNRP615, HSNRP616, HSNRP617, HSNRP618, HSNRP619, HSNRP620, HSNRP621, HSNRP622, HSNRP623, HSNRP624, HSNRP625, HSNRP626, HSNRP627, HSNRP628, HSNRP629, HSNRP630, HSNRP631, HSNRP632, HSNRP633, HSNRP634, HSNRP635, HSNRP636, HSNRP637, HSNRP638, HSNRP639, HSNRP640, HSNRP641, HSNRP642, HSNRP643, HSNRP644, HSNRP645, HSNRP646, HSNRP647, HSNRP648, HSNRP649, HSNRP650, HSNRP651, HSNRP652, HSNRP653, HSNRP654, HSNRP655, HSNRP656, HSNRP657, HSNRP658, HSNRP659, HSNRP660, HSNRP661, HSNRP662, HSNRP663, HSNRP664, HSNRP665, HSNRP666, HSNRP667, HSNRP668, HSNRP669, HSNRP670, HSNRP671, HSNRP672, HSNRP673, HSNRP674, HSNRP675, HSNRP676, HSNRP677, HSNRP678, HSNRP679, HSNRP680, HSNRP681, HSNRP682, HSNRP683, HSNRP684, HSNRP685, HSNRP686, HSNRP687, HSNRP688, HSNRP689, HSNRP690, HSNRP691, HSNRP692, HSNRP693, HSNRP694, HSNRP695, HSNRP696, HSNRP697, HSNRP698, HSNRP699, HSNRP700, HSNRP701, HSNRP702, HSNRP703, HSNRP704, HSNRP705, HSNRP706, HSNRP707, HSNRP708, HSNRP709, HSNRP710, HSNRP711, HSNRP712, HSNRP713, HSNRP714, HSNRP715, HSNRP716, HSNRP717, HSNRP718, HSNRP719, HSNRP720, HSNRP721, HSNRP722, HSNRP723, HSNRP724, HSNRP725, HSNRP726, HSNRP727, HSNRP728, HSNRP729, HSNRP730, HSNRP731, HSNRP732, HSNRP733, HSNRP734, HSNRP735, HSNRP736, HSNRP737, HSNRP738, HSNRP739, HSNRP740, HSNRP741, HSNRP742, HSNRP743, HSNRP744, HSNRP745, HSNRP746, HSNRP747, HSNRP748, HSNRP749, HSNRP750, HSNRP751, HSNRP752, HSNRP753, HSNRP754, HSNRP755, HSNRP756, HSNRP757, HSNRP758, HSNRP759, HSNRP760, HSNRP761, HSNRP762, HSNRP763, HSNRP764, HSNRP765, HSNRP766, HSNRP767, HSNRP768, HSNRP769, HSNRP770, HSNRP771, HSNRP772, HSNRP773, HSNRP774, HSNRP775, HSNRP776, H						

Appellation 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/BIEN CÉ GÈNE et/ou NUM OMDM DU GÈNE / HGNC Gene Symbol and/or OMIM Gene Number	RÈGLES DE MANUTENTION / Handling Conditions	LABORATOIRE DÉSIGNÉ / Designated laboratory	RENSEIGNEMENTS SUR L'ANALYSE / Test details	MÉTHODE / Methode	NOTE PARTICULIÈRE / Special comment	Date de la révision / Date of revision
1,5-anhydroglucitol (GlycoMark) [500115] [GLYC]	1465		Gold Top or Red Top (Serum) or Lavender (plasma). Centrifuge and aliquot serum to transport tube within one hour of collection. Store and ship frozen. Stable at -20°C for 14 days.	LabCorp/Dynacare (https://www.labcorp.com/)		Enzymatic, colorimetric assay.		10-Jan-21
Apert/Pfeiffer Syndrome (recurrent mutation in FGFR1 gene)		FGFR1 (136350; FGFR2 (2964))	Lavender Top (EDTA)	Molecular Genetics Laboratory, Hospital for Sick Children (www.sickkids.ca/PaediatricLaboratoryMedicine/Laboratories-Services/Molecular-Genetics-Laboratory/index.html)	Craniosynostosis: Apert syndrome (101200); Pfeiffer syndrome (101600)	Sanger sequencing: FGFR2 (exon 7); FGFR1 (p.Pro252Arg)		
Aripiprazole (Ablify) [FARI]	1142		Red top tube (Gold top SST tube not accepted)	Mayo Medical Laboratories (www.mayomedicallaboratories.com)		LC-MS/MS		05-May-21
Arrhythmia Panel [CA1601]	289	ARCC9 AKAP9 ANK2 CACNA1C CACNB2 CALM1 CALM2 CALM3 CASQ2 CAV3 CDH2 CTNNA1 DBP1 DSC2 DSG2 DSP C FLCN1 GATA6 HADHA HCN4 KCNA5 KCNE1 KCNE2 KCNE3 KCNQ1 LDB1 LEM2 LMAN1 MYH7 MYH7 MYL4 NKX2-5 NGLAP PRRF1 PLSN PLSN2 RYR2 SALL4 SCN10A SCN5B SCN5B SCN5A TBX3 TBC1D17 TCF7L3 TCF7L3-AS1 TNN3 TNN3K1 TNN3Z TROSD1 TRPM4 TTN1	Lavender Top (EDTA) [2 tubes]	Blueprint Genetics http://blueprintgenetics.com/		NextGen Sequencing		20-Feb-20
Arthritis Panel	1465		Serum gel (Gold top)	Mitogen Advanced Diagnostics				05-Aug-22
Aspergillus Antibody by Immunodiffusion [324]			Serum: Collect serum specimens in serum separator or red top tube. Allow blood to clot for 30 minutes, then centrifuge. Pipette serum into a plastic screw cap vial. STABILITY Refrigerated: 14 days Frozen: 6 months	MiraVista Diagnostics				08-Sep-22
Ataxia with Oculomotor Apraxia Type 1/2 NGS Panel		APTX (60630); SETX (60845)	Lavender Top (2 x 4 mL)	Fulgent Genetics (fulgentdiagnostics.com)	Ataxia, early-onset, with oculomotor apraxia and hypochromemia (209220); Amyotrophic lateral sclerosis 4, juvenile (602433); Ataxia-ocular apraxia-2 (606002)	NextGen Sequencing		05-Apr-19
Ataxia/Episodic Ataxia Disorders (NextGen Sequencing Panel and Copy Number Analysis + mtDNA + FRDA Repeat Expansion Analysis) [NGS419]	583	ACAS1 (60678); ADCK2 (612018); AFB1 (100158); AFB2 (60680); AFB3 (60071); AFB4 (60678); AFB5 (60678); AFB6 (60678); AFB7 (60678); AFB8 (60678); AFB9 (60678); AFB10 (60678); AFB11 (60678); AFB12 (60678); AFB13 (60678); AFB14 (60678); AFB15 (60678); AFB16 (60678); AFB17 (60678); AFB18 (60678); AFB19 (60678); AFB20 (60678); AFB21 (60678); AFB22 (60678); AFB23 (60678); AFB24 (60678); AFB25 (60678); AFB26 (60678); AFB27 (60678); AFB28 (60678); AFB29 (60678); AFB30 (60678); AFB31 (60678); AFB32 (60678); AFB33 (60678); AFB34 (60678); AFB35 (60678); AFB36 (60678); AFB37 (60678); AFB38 (60678); AFB39 (60678); AFB40 (60678); AFB41 (60678); AFB42 (60678); AFB43 (60678); AFB44 (60678); AFB45 (60678); AFB46 (60678); AFB47 (60678); AFB48 (60678); AFB49 (60678); AFB50 (60678); AFB51 (60678); AFB52 (60678); AFB53 (60678); AFB54 (60678); AFB55 (60678); AFB56 (60678); AFB57 (60678); AFB58 (60678); AFB59 (60678); AFB60 (60678); AFB61 (60678); AFB62 (60678); AFB63 (60678); AFB64 (60678); AFB65 (60678); AFB66 (60678); AFB67 (60678); AFB68 (60678); AFB69 (60678); AFB70 (60678); AFB71 (60678); AFB72 (60678); AFB73 (60678); AFB74 (60678); AFB75 (60678); AFB76 (60678); AFB77 (60678); AFB78 (60678); AFB79 (60678); AFB80 (60678); AFB81 (60678); AFB82 (60678); AFB83 (60678); AFB84 (60678); AFB85 (60678); AFB86 (60678); AFB87 (60678); AFB88 (60678); AFB89 (60678); AFB90 (60678); AFB91 (60678); AFB92 (60678); AFB93 (60678); AFB94 (60678); AFB95 (60678); AFB96 (60678); AFB97 (60678); AFB98 (60678); AFB99 (60678); AFB100 (60678); AFB101 (60678); AFB102 (60678); AFB103 (60678); AFB104 (60678); AFB105 (60678); AFB106 (60678); AFB107 (60678); AFB108 (60678); AFB109 (60678); AFB110 (60678); AFB111 (60678); AFB112 (60678); AFB113 (60678); AFB114 (60678); AFB115 (60678); AFB116 (60678); AFB117 (60678); AFB118 (60678); AFB119 (60678); AFB120 (60678); AFB121 (60678); AFB122 (60678); AFB123 (60678); AFB124 (60678); AFB125 (60678); AFB126 (60678); AFB127 (60678); AFB128 (60678); AFB129 (60678); AFB130 (60678); AFB131 (60678); AFB132 (60678); AFB133 (60678); AFB134 (60678); AFB135 (60678); AFB136 (60678); AFB137 (60678); AFB138 (60678); AFB139 (60678); AFB140 (60678); AFB141 (60678); AFB142 (60678); AFB143 (60678); AFB144 (60678); AFB145 (60678); AFB146 (60678); AFB147 (60678); AFB148 (60678); AFB149 (60678); AFB150 (60678); AFB151 (60678); AFB152 (60678); AFB153 (60678); AFB154 (60678); AFB155 (60678); AFB156 (60678); AFB157 (60678); AFB158 (60678); AFB159 (60678); AFB160 (60678); AFB161 (60678); AFB162 (60678); AFB163 (60678); AFB164 (60678); AFB165 (60678); AFB166 (60678); AFB167 (60678); AFB168 (60678); AFB169 (60678); AFB170 (60678); AFB171 (60678); AFB172 (60678); AFB173 (60678); AFB174 (60678); AFB175 (60678); AFB176 (60678); AFB177 (60678); AFB178 (60678); AFB179 (60678); AFB180 (60678); AFB181 (60678); AFB182 (60678); AFB183 (60678); AFB184 (60678); AFB185 (60678); AFB186 (60678); AFB187 (60678); AFB188 (60678); AFB189 (60678); AFB190 (60678); AFB191 (60678); AFB192 (60678); AFB193 (60678); AFB194 (60678); AFB195 (60678); AFB196 (60678); AFB197 (60678); AFB198 (60678); AFB199 (60678); AFB200 (60678); AFB201 (60678); AFB202 (60678); AFB203 (60678); AFB204 (60678); AFB205 (60678); AFB206 (60678); AFB207 (60678); AFB208 (60678); AFB209 (60678); AFB210 (60678); AFB211 (60678); AFB212 (60678); AFB213 (60678); AFB214 (60678); AFB215 (60678); AFB216 (60678); AFB217 (60678); AFB218 (60678); AFB219 (60678); AFB220 (60678); AFB221 (60678); AFB222 (60678); AFB223 (60678); AFB224 (60678); AFB225 (60678); AFB226 (60678); AFB227 (60678); AFB228 (60678); AFB229 (60678); AFB230 (60678); AFB231 (60678); AFB232 (60678); AFB233 (60678); AFB234 (60678); AFB235 (60678); AFB236 (60678); AFB237 (60678); AFB238 (60678); AFB239 (60678); AFB240 (60678); AFB241 (60678); AFB242 (60678); AFB243 (60678); AFB244 (60678); AFB245 (60678); AFB246 (60678); AFB247 (60678); AFB248 (60678); AFB249 (60678); AFB250 (60678); AFB251 (60678); AFB252 (60678); AFB253 (60678); AFB254 (60678); AFB255 (60678); AFB256 (60678); AFB257 (60678); AFB258 (60678); AFB259 (60678); AFB260 (60678); AFB261 (60678); AFB262 (60678); AFB263 (60678); AFB264 (60678); AFB265 (60678); AFB266 (60678); AFB267 (60678); AFB268 (60678); AFB269 (60678); AFB270 (60678); AFB271 (60678); AFB272 (60678); AFB273 (60678); AFB274 (60678); AFB275 (60678); AFB276 (60678); AFB277 (60678); AFB278 (60678); AFB279 (60678); AFB280 (60678); AFB281 (60678); AFB282 (60678); AFB283 (60678); AFB284 (60678); AFB285 (60678); AFB286 (60678); AFB287 (60678); AFB288 (60678); AFB289 (60678); AFB290 (60678); AFB291 (60678); AFB292 (60678); AFB293 (60678); AFB294 (60678); AFB295 (60678); AFB296 (60678); AFB297 (60678); AFB298 (60678); AFB299 (60678); AFB300 (60678); AFB301 (60678); AFB302 (60678); AFB303 (60678); AFB304 (60678); AFB305 (60678); AFB306 (60678); AFB307 (60678); AFB308 (60678); AFB309 (60678); AFB310 (60678); AFB311 (60678); AFB312 (60678); AFB313 (60678); AFB314 (60678); AFB315 (60678); AFB316 (60678); AFB317 (60678); AFB318 (60678); AFB319 (60678); AFB320 (60678); AFB321 (60678); AFB322 (60678); AFB323 (60678); AFB324 (60678); AFB325 (60678); AFB326 (60678); AFB327 (60678); AFB328 (60678); AFB329 (60678); AFB330 (60678); AFB331 (60678); AFB332 (60678); AFB333 (60678); AFB334 (60678); AFB335 (60678); AFB336 (60678); AFB337 (60678); AFB338 (60678); AFB339 (60678); AFB340 (60678); AFB341 (60678); AFB342 (60678); AFB343 (60678); AFB344 (60678); AFB345 (60678); AFB346 (60678); AFB347 (60678); AFB348 (60678); AFB349 (60678); AFB350 (60678); AFB351 (60678); AFB352 (60678); AFB353 (60678); AFB354 (60678); AFB355 (60678); AFB356 (60678); AFB357 (60678); AFB358 (60678); AFB359 (60678); AFB360 (60678); AFB361 (60678); AFB362 (60678); AFB363 (60678); AFB364 (60678); AFB365 (60678); AFB366 (60678); AFB367 (60678); AFB368 (60678); AFB369 (60678); AFB370 (60678); AFB371 (60678); AFB372 (60678); AFB373 (60678); AFB374 (60678); AFB375 (60678); AFB376 (60678); AFB377 (60678); AFB378 (60678); AFB379 (60678); AFB380 (60678); AFB381 (60678); AFB382 (60678); AFB383 (60678); AFB384 (60678); AFB385 (60678); AFB386 (60678); AFB387 (60678); AFB388 (60678); AFB389 (60678); AFB390 (60678); AFB391 (60678); AFB392 (60678); AFB393 (60678); AFB394 (60678); AFB395 (60678); AFB396 (60678); AFB397 (60678); AFB398 (60678); AFB399 (60678); AFB400 (60678); AFB401 (60678); AFB402 (60678); AFB403 (60678); AFB404 (60678); AFB405 (60678); AFB406 (60678); AFB407 (60678); AFB408 (60678); AFB409 (60678); AFB410 (60678); AFB411 (60678); AFB412 (60678); AFB413 (60678); AFB414 (60678); AFB415 (60678); AFB416 (60678); AFB417 (60678); AFB418 (60678); AFB419 (60678); AFB420 (60678); AFB421 (60678); AFB422 (60678); AFB423 (60678); AFB424 (60678); AFB425 (60678); AFB426 (60678); AFB427 (60678); AFB428 (60678); AFB429 (60678); AFB430 (60678); AFB431 (60678); AFB432 (60678); AFB433 (60678); AFB434 (60678); AFB435 (60678); AFB436 (60678); AFB437 (60678); AFB438 (60678); AFB439 (60678); AFB440 (60678); AFB441 (60678); AFB442 (60678); AFB443 (60678); AFB444 (60678); AFB445 (60678); AFB446 (60678); AFB447 (60678); AFB448 (60678); AFB449 (60678); AFB450 (60678); AFB451 (60678); AFB452 (60678); AFB453 (60678); AFB454 (60678); AFB455 (60678); AFB456 (60678); AFB457 (60678); AFB458 (60678); AFB459 (60678); AFB460 (60678); AFB461 (60678); AFB462 (60678); AFB463 (60678); AFB464 (60678); AFB465 (60678); AFB466 (60678); AFB467 (60678); AFB468 (60678); AFB469 (60678); AFB470 (60678); AFB471 (60678); AFB472 (60678); AFB473 (60678); AFB474 (60678); AFB475 (60678); AFB476 (60678); AFB477 (60678); AFB478 (60678); AFB479 (60678); AFB480 (60678); AFB481 (60678); AFB482 (60678); AFB483 (60678); AFB484 (60678); AFB485 (60678); AFB486 (60678); AFB487 (60678); AFB488 (60678); AFB489 (60678); AFB490 (60678); AFB491 (60678); AFB492 (60678); AFB493 (60678); AFB494 (60678); AFB495 (60678); AFB496 (60678); AFB497 (60678); AFB498 (60678); AFB499 (60678); AFB500 (60678); AFB501 (60678); AFB502 (60678); AFB503 (60678); AFB504 (60678); AFB505 (60678); AFB506 (60678); AFB507 (60678); AFB508 (60678); AFB509 (60678); AFB510 (60678); AFB511 (60678); AFB512 (60678); AFB513 (60678); AFB514 (60678); AFB515 (60678); AFB516 (60678); AFB517 (60678); AFB518 (60678); AFB519 (60678); AFB520 (60678); AFB521 (60678); AFB522 (60678); AFB523 (60678); AFB524 (60678); AFB525 (60678); AFB526 (60678); AFB527 (60678); AFB528 (60678); AFB529 (60678); AFB530 (60678); AFB531 (60678); AFB532 (60678); AFB533 (60678); AFB534 (60678); AFB535 (60678); AFB536 (60678); AFB537 (60678); AFB538 (60678); AFB539 (60678); AFB540 (60678); AFB541 (60678); AFB542 (60678); AFB543 (60678); AFB544 (60678); AFB545 (60678); AFB546 (60678); AFB547 (60678); AFB548 (60678); AFB549 (60678); AFB550 (60678); AFB551 (60678); AFB552 (60678); AFB553 (60678); AFB554 (60678); AFB555 (60678); AFB556 (60678); AFB557 (60678); AFB558 (60678); AFB559 (60678); AFB560 (60678); AFB561 (60678); AFB562 (60678); AFB563 (60678); AFB564 (60678); AFB565 (60678); AFB566 (60678); AFB567 (60678); AFB568 (60678); AFB569 (60678); AFB570 (60678); AFB571 (60678); AFB572 (60678); AFB573 (60678); AFB574 (60678); AFB575 (60678); AFB576 (60678); AFB577 (60678); AFB578 (60678); AFB579 (60678); AFB580 (60678); AFB581 (60678); AFB582 (60678); AFB583 (60678); AFB584 (60678); AFB585 (60678); AFB586 (60678); AFB587 (60678); AFB588 (60678); AFB589 (60678); AFB590 (60678); AFB591 (60678); AFB592 (60678); AFB593 (60678); AFB594 (60678); AFB595 (60678); AFB596 (60678); AFB597 (60678); AFB598 (60678); AFB599 (60678); AFB600 (60678); AFB601 (60678); AFB602 (60678); AFB603 (60678); AFB604 (60678); AFB605 (60678); AFB606 (60678); AFB607 (60678); AFB608 (60678); AFB609 (60678); AFB610 (60678); AFB611 (60678); AFB612 (60678); AFB613 (60678); AFB614 (60678); AFB615 (60678); AFB616 (60678); AFB617 (60678); AFB618 (60678); AFB619 (60678); AFB620 (60678); AFB621 (60678); AFB622 (60678); AFB623 (60678); AFB624 (60678); AFB625 (60678); AFB626 (60678); AFB627 (60678); AFB628 (60678); AFB629 (60678); AFB630 (60678); AFB631 (60678); AFB632 (60678); AFB633 (60678); AFB634 (60678); AFB635 (60678); AFB636 (60678); AFB637 (60678); AFB638 (60678); AFB639 (60678); AFB640 (60678); AFB641 (60678); AFB642 (60678); AFB643 (60678); AFB644 (60678); AFB645 (60678); AFB646 (60678); AFB647 (60678); AFB648 (60678); AFB649 (60678); AFB650 (60678); AFB651 (60678); AFB652 (60678); AFB653 (60678); AFB654 (60678); AFB655 (60678); AFB656 (60678); AFB657 (60678); AFB658 (60678); AFB659 (60678); AFB660 (60678); AFB661 (60678); AFB662 (60678); AFB663 (60678); AFB664 (60678); AFB665 (60678); AFB666 (60678); AFB667 (60678); AFB668 (60678); AFB669 (60678); AFB670 (60678); AFB671 (60678); AFB672 (60678); AFB673 (60678); AFB674 (60678); AFB675 (60678); AFB676 (60678); AFB677 (60678); AFB678 (60678); AFB679 (60678); AFB680 (60678); AFB681 (60678); AFB682 (60678); AFB683 (60678); AFB684 (60678); AFB685 (60678); AFB686 (60678); AFB687 (60678); AFB688 (60678); AFB689 (60678); AFB690 (60678); AFB691 (60678); AFB692 (60678); AFB693 (60678); AFB694 (60678); AFB695 (60678); AFB696 (60678); AFB697 (60678); AFB698 (60678); AFB699 (60678); AFB700 (60678); AFB701 (60678); AFB702 (60678); AFB703 (60678); AFB704 (60678); AFB705 (60678); AFB706 (60678); AFB707 (60678); AFB708 (60678); AFB709 (60678); AFB710 (60678); AFB711 (60678); AFB712 (60678); AFB713 (60678); AFB714 (60678); AFB715 (60678); AFB716 (60678); AFB717 (60678); AFB718 (60678); AFB719 (60678); AFB720 (60678); AFB721 (60678); AFB722 (60678); AFB723 (60678); AFB724 (60678); AFB725 (60678); AFB726 (60678); AFB727 (60678); AFB728 (60678); AFB729 (60678); AFB730 (60678); AFB731 (60678); AFB732 (60678); AFB733 (60678); AFB734 (60678); AFB735 (60678); AFB736 (60678); AFB737 (60678); AFB738 (60678); AFB739 (60678); AFB740 (60678); AFB741 (60678); AFB742 (60678); AFB743 (60678); AFB744 (60678); AFB745 (60678); AFB746 (60678); AFB747 (60678); AFB748 (60678); AFB749 (60678); AFB750 (60678); AFB751 (60678); AFB752 (60678); AFB753 (60678); AFB754 (60678); AFB755 (60678); AFB756 (60678); AFB757 (60678); AFB758 (60678); AFB759 (60678); AFB760 (60678); AFB761 (60678); AFB762 (60678); AFB763 (60678); AFB764 (60678); AFB765 (60678); AFB766 (60678); AFB767 (60678); AFB768 (60678); AFB769 (60678); AFB770 (60678); AFB771 (60678); AFB772 (60678); AFB773 (60678); AFB774 (60678); AFB775 (60678); AFB776 (60678); AFB777 (60678); AFB778 (60678); AFB779 (60678); AFB780 (60678); AFB781 (60678); AFB782 (60678); AFB783 (60678); AFB784 (60678); AFB785 (60678); AFB786 (60678); AFB787 (60678); AFB788 (60678); AFB789 (60678); AFB790 (60678); AFB791 (60678); AFB792 (60678); AFB793 (60678); AFB794 (60678); AFB795 (60678); AFB796 (60678); AFB797 (60678); AFB798 (60678); AFB799 (60678); AFB800 (60678); AFB801 (60678); AFB802 (60678); AFB803 (60678); AFB804 (60678); AFB805 (60678); AFB806 (60678); AFB807 (60678); AFB808 (60678); AFB809 (60678); AFB810 (60678); AFB811 (60678); AFB812 (60678); AFB813 (60678); AFB814 (60678); AFB815 (60678); AFB816 (60678); AFB817 (60678); AFB818 (60678); AFB819 (60678); AFB820 (60678); AFB821 (60678); AFB822 (60678); AFB823 (60678); AFB824 (60678); AFB825 (60678); AFB826 (60678); AFB827 (60678); AFB828 (60678); AFB829 (60678); AFB830 (60678); AFB831 (60678); AFB832 (60678); AFB833 (60678); AFB834 (60678); AFB835 (60678); AFB836 (60678); AFB837 (60678); AFB838 (60678); AFB839 (60678); AFB840 (60678); AFB841 (60678); AFB842 (60678); AFB843 (60678); AFB844 (60678); AFB845 (60678); AFB846 (60678); AFB847 (60678); AFB848 (60678); AFB849 (60678); AFB850 (60678); AFB851 (60678); AFB852 (60678); AFB853 (60678); AFB854 (60678); AFB855 (60678); AFB856 (60678); AFB857 (60678); AFB858 (60678); AFB859 (60678); AFB860 (60678); AFB861 (60678); AFB862 (60678); AFB863 (60678); AFB864 (60678); AFB865 (60678); AFB866 (60678); AFB867 (60678); AFB868 (60678); AFB869 (60678); AFB870 (60678); AFB871 (60678); AFB872 (60678); AFB873 (60678); AFB874 (60678); AFB875 (60678); AFB876 (60678); AFB877 (60678); AFB878 (60678); AFB879 (60678); AFB880 (60678); AFB881 (60678); AFB882 (60678); AFB883 (60678); AFB884 (60678); AFB885 (60678); AFB886 (60678); AFB887 (60678); AFB888 (60678); AFB889 (60678); AFB890 (60678); AFB891 (60678); AFB892 (60678); AFB893 (60678); AFB89						

Appellation 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/BIEN CÉ DÉ GÈNE et/ou NOM OMIM DU GÈNE / HGNC Gene Symbol and OMIM Gene Number	RÈGLES DE MANUTENTION / Handling Conditions	LABORATOIRE DÉSIGNÉ / Designated laboratory	RENSEIGNEMENTS SUR L'ANALYSE / Test details	MÉTHODE / Methode	NOTE PARTICULIÈRE / Special comment	Date de la révision / Date of revision
1,5-anhydroglucitol (GlycoMark) [500115] [GLYC]	1465		Gold Top or Red Top (Serum) or Lavender (plasma). Centrifuge and aliquot serum to transport tube within one hour of collection. Store and ship frozen. Stable at -20°C for 14 days.	LabCorp/Dynacare (https://www.labcorp.com/)		Enzymatic, colorimetric assay.		10-Jan-21
Ataxia/Episodic Ataxia Disorders (NextGen Sequencing Panel and Copy Number Analysis + mtDNA) [NGS324]	583	ACAN (608776), AKOR2 (602407), ARHGAP1 (609536), ARMC8 (608999), ARMC9 (609171), ARID5 (606786), ACOX2 (608830), ALKBH3 (608223), ALGRL2 (604851), AHI1 (608894), ALDH5A1 (610045), ALG6 (604566), AMACR (NA), ANO10 (613726), APOPT1 (616003), APTX (606350), ARL13B (609022), ARHGAP (608485), ARSA (607574), ASL (606710), ASS1 (607670), ATM (607585), ATN1 (607462), ATP1A2 (182340), ATP1A3 (182350), ATP8A2 (605870), ATPAF2 (608918), ATXN1 (601556), ATXN10 (605106), ATXN2 (601517), ATXN3 (607047), ATXN7 (607646), ATR (606529), BDNF (614144), BBS1 (209601), BBS10 (610148), BBS12 (610063), BBS2 (606151), BBS4 (603734), BBS5 (605650), BBS7 (607590), BBS9 (607908), BCKDHA (608248), BCKDHL1 (248611), BCS1L (603647), BEAN1 (612651), BOLA3 (613183), BCL2 (606558), BDNF (609419), CBOR1 (608075), C12orf85 (613541), C19orf12 (614296), CSORF42 (614571), CAS (114815), CACNA1A (601011), CACNA1G (604905), CACNB4 (603949), CAMTA1 (611501), CASK (607121), CEB2A (612013), CTRCF2 (601622), CTRCF3 (611264), CECR1 (607575), CEP290 (610142), TSGA14 (610523), CHCHD10 (615903), CLCN2 (606570), CLN3 (608020), CLN6 (606725), CLN8 (607871), CLPF (601119), COG4 (606976), COQ2 (608025), COX6B (610125), C10orf102 (614458), COX3 (605446), FAM36A (614698), COX6B1 (124089), COX6A (123870), CP (117700), CPS1 (608307), CSPP1 (611454), CSTB (601145), CTFP1 (604927), CTSD (116840), CTF (605526), CYP27A1 (606510), DARS2 (605065), DMT (248040), DCN (300121), DHRF (126060), DKC1 (300126), DLAT (608770), DLD (238331), DNAAF19 (608977), DNAAF3 (611201), DNMT1 (125375), DP94 (605103), DYRK1A (608855), EEF2 (136610), EGR2 (129610), ELAVL4 (605124), ELAVL5 (611805), ERCC2 (126348), ERCC5 (133530), ERCC6 (133540), ETHE1 (608451), FA2H (611026), FASTK1L2 (612122), FXR1 (609554), FGF4 (601515), FVCK1 (601044), FMR1 (602506), FOXR1 (613622), FTL (134700), FXN (606629), GABRB3 (137192), GALT (601240), GBA (604645), GBA2 (609471), GCH1 (600225), GFAP (137780), GJA1 (606399), GJB1 (304040), GIC2 (608803), GLEB1 (611458), GMPBP (615520), GOS2 (606073), GPI (173400), GRP94 (604110), GRM1 (614831), GRN (138945), GSS (610102), HARS (142810), HARS2 (607075), HCN1 (602780), HEPACAM (611642), HEXB (606873), HTR1A (109700), HTR1A1 (602344), HTT (603004), INPPE1 (613817), ITIH3B (605940), ITPI1 (147265), KCNA1 (176269), KCNC1 (176258), KCNC3 (176264), KCND3 (605411), KCYD1 (611725), KIAA1 (602255), KIA2B (609996), KIA5A (602821), KIF7 (611254), LARS (604541), LMB1 (159340), LRRPC (607544), LYRM1 (615011), MAN2B1 (609458), MARS2 (609726), MBD5 (611472), MECP2 (300005), MED14 (608771), MFSB8 (611124), MKS3 (604896), MKS1 (609853), MLC1 (609908), MMRADRC (611955), MPP1 (137960), MPP2 (154040), MRL1 (601766), MRE11A (NA), mtDNA (NA), MTFMT (611766), MTPAP (613669), MTPP (157147), MVK (251170), NAGAL (609700), NDUFA1 (300078), NDUFA10 (603835), NDUFA11 (602038), NDUFA12 (614530), NDUFA2 (602137), NDUFA9 (603834), NDUFA9 (600934), NDUFAF2 (609653), NDUFAF3 (612911), NDUFA4 (611776), C20ORF7 (612360), C20ORF8 (612362), NFE1 (608272), NFE2 (607739), NFE3 (606252), NOP56 (614154), NPC1 (607823), NPC2 (610103), NPSP1 (607100), NR1B1 (613211), OREN (300170), OPA1 (605290), OPA3 (606580), OPN1 (300127), OTC (300461), PANK2 (600157), PANG (607100), FHRA1 (300502), PDKX (608769), PDPN (151340), PEX3 (602899), PES2 (266510), PEST (601757), PGR1 (313800), PGM1 (173100), PHVH (600226), PLA2G6 (603604), PLP1 (300401), PMA6 (601785), PMP22 (602530), PMP23 (602531), PMP24 (602532), PMP25 (602533), PMP26 (602534), PMP27 (602535), PMP28 (602536), PMP29 (602537), PMP30 (602538), PMP31 (602539), PMP32 (602540), PMP33 (602541), PMP34 (602542), PMP35 (602543), PMP36 (602544), PMP37 (602545), PMP38 (602546), PMP39 (602547), PMP40 (602548), PMP41 (602549), PMP42 (602550), PMP43 (602551), PMP44 (602552), PMP45 (602553), PMP46 (602554), PMP47 (602555), PMP48 (602556), PMP49 (602557), PMP50 (602558), PMP51 (602559), PMP52 (602560), PMP53 (602561), PMP54 (602562), PMP55 (602563), PMP56 (602564), PMP57 (602565), PMP58 (602566), PMP59 (602567), PMP60 (602568), PMP61 (602569), PMP62 (602570), PMP63 (602571), PMP64 (602572), PMP65 (602573), PMP66 (602574), PMP67 (602575), PMP68 (602576), PMP69 (602577), PMP70 (602578), PMP71 (602579), PMP72 (602580), PMP73 (602581), PMP74 (602582), PMP75 (602583), PMP76 (602584), PMP77 (602585), PMP78 (602586), PMP79 (602587), PMP80 (602588), PMP81 (602589), PMP82 (602590), PMP83 (602591), PMP84 (602592), PMP85 (602593), PMP86 (602594), PMP87 (602595), PMP88 (602596), PMP89 (602597), PMP90 (602598), PMP91 (602599), PMP92 (602600), PMP93 (602601), PMP94 (602602), PMP95 (602603), PMP96 (602604), PMP97 (602605), PMP98 (602606), PMP99 (602607), PMP100 (602608), PMP101 (602609), PMP102 (602610), PMP103 (602611), PMP104 (602612), PMP105 (602613), PMP106 (602614), PMP107 (602615), PMP108 (602616), PMP109 (602617), PMP110 (602618), PMP111 (602619), PMP112 (602620), PMP113 (602621), PMP114 (602622), PMP115 (602623), PMP116 (602624), PMP117 (602625), PMP118 (602626), PMP119 (602627), PMP120 (602628), PMP121 (602629), PMP122 (602630), PMP123 (602631), PMP124 (602632), PMP125 (602633), PMP126 (602634), PMP127 (602635), PMP128 (602636), PMP129 (602637), PMP130 (602638), PMP131 (602639), PMP132 (602640), PMP133 (602641), PMP134 (602642), PMP135 (602643), PMP136 (602644), PMP137 (602645), PMP138 (602646), PMP139 (602647), PMP140 (602648), PMP141 (602649), PMP142 (602650), PMP143 (602651), PMP144 (602652), PMP145 (602653), PMP146 (602654), PMP147 (602655), PMP148 (602656), PMP149 (602657), PMP150 (602658), PMP151 (602659), PMP152 (602660), PMP153 (602661), PMP154 (602662), PMP155 (602663), PMP156 (602664), PMP157 (602665), PMP158 (602666), PMP159 (602667), PMP160 (602668), PMP161 (602669), PMP162 (602670), PMP163 (602671), PMP164 (602672), PMP165 (602673), PMP166 (602674), PMP167 (602675), PMP168 (602676), PMP169 (602677), PMP170 (602678), PMP171 (602679), PMP172 (602680), PMP173 (602681), PMP174 (602682), PMP175 (602683), PMP176 (602684), PMP177 (602685), PMP178 (602686), PMP179 (602687), PMP180 (602688), PMP181 (602689), PMP182 (602690), PMP183 (602691), PMP184 (602692), PMP185 (602693), PMP186 (602694), PMP187 (602695), PMP188 (602696), PMP189 (602697), PMP190 (602698), PMP191 (602699), PMP192 (602700), PMP193 (602701), PMP194 (602702), PMP195 (602703), PMP196 (602704), PMP197 (602705), PMP198 (602706), PMP199 (602707), PMP200 (602708), PMP201 (602709), PMP202 (602710), PMP203 (602711), PMP204 (602712), PMP205 (602713), PMP206 (602714), PMP207 (602715), PMP208 (602716), PMP209 (602717), PMP210 (602718), PMP211 (602719), PMP212 (602720), PMP213 (602721), PMP214 (602722), PMP215 (602723), PMP216 (602724), PMP217 (602725), PMP218 (602726), PMP219 (602727), PMP220 (602728), PMP221 (602729), PMP222 (602730), PMP223 (602731), PMP224 (602732), PMP225 (602733), PMP226 (602734), PMP227 (602735), PMP228 (602736), PMP229 (602737), PMP230 (602738), PMP231 (602739), PMP232 (602740), PMP233 (602741), PMP234 (602742), PMP235 (602743), PMP236 (602744), PMP237 (602745), PMP238 (602746), PMP239 (602747), PMP240 (602748), PMP241 (602749), PMP242 (602750), PMP243 (602751), PMP244 (602752), PMP245 (602753), PMP246 (602754), PMP247 (602755), PMP248 (602756), PMP249 (602757), PMP250 (602758), PMP251 (602759), PMP252 (602760), PMP253 (602761), PMP254 (602762), PMP255 (602763), PMP256 (602764), PMP257 (602765), PMP258 (602766), PMP259 (602767), PMP260 (602768), PMP261 (602769), PMP262 (602770), PMP263 (602771), PMP264 (602772), PMP265 (602773), PMP266 (602774), PMP267 (602775), PMP268 (602776), PMP269 (602777), PMP270 (602778), PMP271 (602779), PMP272 (602780), PMP273 (602781), PMP274 (602782), PMP275 (602783), PMP276 (602784), PMP277 (602785), PMP278 (602786), PMP279 (602787), PMP280 (602788), PMP281 (602789), PMP282 (602790), PMP283 (602791), PMP284 (602792), PMP285 (602793), PMP286 (602794), PMP287 (602795), PMP288 (602796), PMP289 (602797), PMP290 (602798), PMP291 (602799), PMP292 (602800), PMP293 (602801), PMP294 (602802), PMP295 (602803), PMP296 (602804), PMP297 (602805), PMP298 (602806), PMP299 (602807), PMP300 (602808), PMP301 (602809), PMP302 (602810), PMP303 (602811), PMP304 (602812), PMP305 (602813), PMP306 (602814), PMP307 (602815), PMP308 (602816), PMP309 (602817), PMP310 (602818), PMP311 (602819), PMP312 (602820), PMP313 (602821), PMP314 (602822), PMP315 (602823), PMP316 (602824), PMP317 (602825), PMP318 (602826), PMP319 (602827), PMP320 (602828), PMP321 (602829), PMP322 (602830), PMP323 (602831), PMP324 (602832), PMP325 (602833), PMP326 (602834), PMP327 (602835), PMP328 (602836), PMP329 (602837), PMP330 (602838), PMP331 (602839), PMP332 (602840), PMP333 (602841), PMP334 (602842), PMP335 (602843), PMP336 (602844), PMP337 (602845), PMP338 (602846), PMP339 (602847), PMP340 (602848), PMP341 (602849), PMP342 (602850), PMP343 (602851), PMP344 (602852), PMP345 (602853), PMP346 (602854), PMP347 (602855), PMP348 (602856), PMP349 (602857), PMP350 (602858), PMP351 (602859), PMP352 (602860), PMP353 (602861), PMP354 (602862), PMP355 (602863), PMP356 (602864), PMP357 (602865), PMP358 (602866), PMP359 (602867), PMP360 (602868), PMP361 (602869), PMP362 (602870), PMP363 (602871), PMP364 (602872), PMP365 (602873), PMP366 (602874), PMP367 (602875), PMP368 (602876), PMP369 (602877), PMP370 (602878), PMP371 (602879), PMP372 (602880), PMP373 (602881), PMP374 (602882), PMP375 (602883), PMP376 (602884), PMP377 (602885), PMP378 (602886), PMP379 (602887), PMP380 (602888), PMP381 (602889), PMP382 (602890), PMP383 (602891), PMP384 (602892), PMP385 (602893), PMP386 (602894), PMP387 (602895), PMP388 (602896), PMP389 (602897), PMP390 (602898), PMP391 (602899), PMP392 (602900), PMP393 (602901), PMP394 (602902), PMP395 (602903), PMP396 (602904), PMP397 (602905), PMP398 (602906), PMP399 (602907), PMP400 (602908), PMP401 (602909), PMP402 (602910), PMP403 (602911), PMP404 (602912), PMP405 (602913), PMP406 (602914), PMP407 (602915), PMP408 (602916), PMP409 (602917), PMP410 (602918), PMP411 (602919), PMP412 (602920), PMP413 (602921), PMP414 (602922), PMP415 (602923), PMP416 (602924), PMP417 (602925), PMP418 (602926), PMP419 (602927), PMP420 (602928), PMP421 (602929), PMP422 (602930), PMP423 (602931), PMP424 (602932), PMP425 (602933), PMP426 (602934), PMP427 (602935), PMP428 (602936), PMP429 (602937), PMP430 (602938), PMP431 (602939), PMP432 (602940), PMP433 (602941), PMP434 (602942), PMP435 (602943), PMP436 (602944), PMP437 (602945), PMP438 (602946), PMP439 (602947), PMP440 (602948), PMP441 (602949), PMP442 (602950), PMP443 (602951), PMP444 (602952), PMP445 (602953), PMP446 (602954), PMP447 (602955), PMP448 (602956), PMP449 (602957), PMP450 (602958), PMP451 (602959), PMP452 (602960), PMP453 (602961), PMP454 (602962), PMP455 (602963), PMP456 (602964), PMP457 (602965), PMP458 (602966), PMP459 (602967), PMP460 (602968), PMP461 (602969), PMP462 (602970), PMP463 (602971), PMP464 (602972), PMP465 (602973), PMP466 (602974), PMP467 (602975), PMP468 (602976), PMP469 (602977), PMP470 (602978), PMP471 (602979), PMP472 (602980), PMP473 (602981), PMP474 (602982), PMP475 (602983), PMP476 (602984), PMP477 (602985), PMP478 (602986), PMP479 (602987), PMP480 (602988), PMP481 (602989), PMP482 (602990), PMP483 (602991), PMP484 (602992), PMP485 (602993), PMP486 (602994), PMP487 (602995), PMP488 (602996), PMP489 (602997), PMP490 (602998), PMP491 (602999), PMP492 (603000), PMP493 (603001), PMP494 (603002), PMP495 (603003), PMP496 (603004), PMP497 (603005), PMP498 (603006), PMP499 (603007), PMP500 (603008), PMP501 (603009), PMP502 (603010), PMP503 (603011), PMP504 (603012), PMP505 (603013), PMP506 (603014), PMP507 (603015), PMP508 (603016), PMP509 (603017), PMP510 (603018), PMP511 (603019), PMP512 (603020), PMP513 (603021), PMP514 (603022), PMP515 (603023), PMP516 (603024), PMP517 (603025), PMP518 (603026), PMP519 (603027), PMP520 (603028), PMP521 (603029), PMP522 (603030), PMP523 (603031), PMP524 (603032), PMP525 (603033), PMP526 (603034), PMP527 (603035), PMP528 (603036), PMP529 (603037), PMP530 (603038), PMP531 (603039), PMP532 (603040), PMP533 (603041), PMP534 (603042), PMP535 (603043), PMP536 (603044), PMP537 (603045), PMP538 (603046), PMP539 (603047), PMP540 (603048), PMP541 (603049), PMP542 (603050), PMP543 (603051), PMP544 (603052), PMP545 (603053), PMP546 (603054), PMP547 (603055), PMP548 (603056), PMP549 (603057), PMP550 (603058), PMP551 (603059), PMP552 (603060), PMP553 (603061), PMP554 (603062), PMP555 (603063), PMP556 (603064), PMP557 (603065), PMP558 (603066), PMP559 (603067), PMP560 (603068), PMP561 (603069), PMP562 (603070), PMP563 (603071), PMP564 (603072), PMP565 (603073), PMP566 (603074), PMP567 (603075), PMP568 (603076), PMP569 (603077), PMP570 (603078), PMP571 (603079), PMP572 (603080), PMP573 (603081), PMP574 (603082), PMP575 (603083), PMP576 (603084), PMP577 (603085), PMP578 (603086), PMP579 (603087), PMP580 (603088), PMP581 (603089), PMP582 (603090), PMP583 (603091), PMP584 (603092), PMP585 (603093), PMP586 (603094), PMP587 (603095), PMP588 (603096), PMP589 (603097), PMP590 (603098), PMP591 (603099), PMP592 (603100), PMP593 (603101), PMP594 (603102), PMP595 (603103), PMP596 (603104), PMP597 (603105), PMP598 (603106), PMP599 (603107), PMP600 (603108), PMP601 (603109), PMP602 (603110), PMP603 (603111), PMP604 (603112), PMP605 (603113), PMP606 (603114), PMP607 (603115), PMP608 (603116), PMP609 (603117), PMP610 (603118), PMP611 (603119), PMP612 (603120), PMP613 (603121), PMP614 (603122), PMP615 (603123), PMP616 (603124), PMP617 (603125), PMP618 (603126), PMP619 (603127), PMP620 (603128), PMP621 (603129), PMP622 (603130), PMP623 (603131), PMP624 (603132), PMP625 (603133), PMP626 (603134), PMP627 (603135), PMP628 (603136), PMP629 (603137), PMP630 (603138), PMP631 (603139), PMP632 (603140), PMP633 (603141), PMP634 (603142), PMP635 (603143), PMP636 (603144), PMP637 (603145), PMP638 (603146), PMP639 (603147), PMP640 (603148), PMP641 (603149), PMP642 (603150), PMP643 (603151), PMP644 (603152), PMP645 (603153), PMP646 (603154), PMP647 (603155), PMP648 (603156), PMP649 (603157), PMP650 (603158), PMP651 (603159), PMP652 (603160), PMP653 (603161), PMP654 (603162), PMP655 (603163), PMP656 (603164), PMP657 (603165), PMP658 (603166), PMP659 (603167), PMP660 (603168), PMP661 (603169), PMP662 (603170), PMP663 (603171), PMP664 (603172), PMP665 (603173), PMP666 (603174), PMP667 (603175), PMP668 (603176), PMP669 (603177), PMP670 (603178), PMP671 (603179), PMP672 (603180), PMP673 (603181), PMP674 (603182), PMP675 (603183), PMP676 (603184), PMP677 (603185), PMP678 (603186), PMP679 (603187), PMP680 (603188), PMP681 (603189), PMP682 (603190), PMP683 (603191), PMP684 (603192), PMP685 (603193), PMP686 (603194), PMP687 (603195), PMP688 (603196), PMP689 (603197), PMP690 (603198), PMP691 (603199), PMP692 (603200), PMP693 (603201), PMP694 (603202), PMP695 (603203), PMP696 (603204), PMP697 (603205), PMP698 (603206), PMP699 (603207), PMP700 (603208), PMP701 (603209), PMP702 (603210), PMP703 (603211), PMP704 (603212), PMP705 (603213), PMP706 (603214), PMP707 (603215), PMP708 (603216), PMP709 (603217), PMP710 (603218), PMP711 (603219), PMP712 (603220), PMP713 (603221), PMP714 (603222), PMP715 (603223), PMP716 (603224), PMP717 (603225), PMP718 (603226), PMP719 (603227), PMP720 (603228), PMP721 (603229), PMP722 (603230), PMP723 (603231), PMP724 (603232), PMP725 (603233), PMP726 (603234), PMP727 (603235), PMP728 (603236), PMP729 (603237), PMP730 (603238), PMP731 (603239), PMP732 (603240), PMP733 (603241), PMP734 (603242), PMP735 (603243), PMP736 (603244), PMP737 (603245), PMP738 (603246), PMP739 (603247), PMP740 (603						

Appellation 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/ou NOM OMIM DU GÈNE / HGNC Gene Symbol and OMIM Gene Number	RÈGLES DE MANUTENTION / Handling Conditions	LABORATOIRE DÉSIGNÉ / Designated laboratory	RENSEIGNEMENTS SUR L'ANALYSE / Test details	MÉTHODE / Methode	NOTE PARTICULIÈRE / Special comment	Date de la révision / Date of revision
1,5-anhydroglucitol (GlycoMark) [500115] [GLYC]	1465		Gold Top or Red Top (Serum) or Lavender (plasma). Centrifuge and aliquot serum to transport tube within one hour of collection. Store and ship frozen. Stable at -20°C for 14 days.	LabCorp/Dynacare (https://www.labcorp.com/)		Enzymatic, colorimetric assay.		10-Jan-21
Autoimmune Retinopathy Panel by Immunoblot [ARP]	720		Serum. Store at 4°C. DO NOT FREEZE.	Ocular Immunology Laboratory, OHSU Biomedical Research Building, Room 253 3181 SW Sam Jackson Park Road Portland, OR 97239, USA	CAII (carbonic anhydrase II), HSP27 (heat shock protein 27), GAPDH (glyceraldehyde 3-phosphate dehydrogenase), Aldolase, Enolase, Arrestin, Tubulin, PKM2 (pyruvate kinase M2)	Immunoblot		21-Jun-22
Autoinflammatory and Autoimmunity Syndromes Panel [08120]	838		Lavender Top (EDTA)	Invitae (www.invitae.com)		NextGen Sequencing		03-Oct-19
Autosomal dominant lateral temporal lobe epilepsy (Epilepsy, familial temporal lobe, 1) (600512)		LEI-1 (604619)	Lavender Top (EDTA)	Centogene AG (www.centogene.com)		1. single exon testing 2. full gene sequencing 3. deletion /duplication testing		
Avian IgG Antibodies Panel, Serum (Budgie and Pigeon)	1419		Red Top; Gold SST	In-Common Laboratories	budgie = parakeet	FEIA		10-Mar-20
Avian precipitins: Pigeon IgG Antibodies	1419		Red Top; Gold SST	In-Common Laboratories		FEIA		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)	Sanger sequencing		09-Dec-19
Baller-Gerold syndrome (218600)		RECQL4 (603780)	Lavender Top (EDTA)	Centogene AG (www.centogene.com)		1. single exon testing 2. full gene sequencing 3. deletion /duplication testing		
Barter Syndrome NGS Panel	804	ATP9B1, BSSND, CA2, CASR, CLCNKA, CLCNKB, CLDN16, CLDN19, PKXD2, GNA11, HNF1B, HSD11B2, KCNJ1, KCNJ10, KLF13, MAGED2, NR3C2, SCNN1A, SCNN1B, SCNN1G, SLC12A1, SLC12A2, SLC12A3, SLC12A4, SLC24A4, WDR61, WDR84	Lavender Top (EDTA) 2 x 4 mL	Fulgent Genetics (fulgentdiagnostics.com)				28-Feb-20
Basal Ganglia Calcification NGS Panel	1192	CA2, MYO6B, PDGFBR (190040), PDGFRR, SLC22A2 (158178), XPR1	Lavender Top (EDTA) 2 x 4 mL	Fulgent Genetics (fulgentdiagnostics.com)	Basal ganglia calcification, idiopathic, 5 (615483); Basal ganglia calcification, idiopathic, 1 (213600)	NextGen Sequencing		13-Feb-20
Bb	792		0.5 mL EDTA plasma (serum also accepted) – spun, separated, frozen within 2 hrs of collection, separate aliquot each test; ship on dry ice	Cincinnati Children's Hospital Medical Center 3333 Burnet Avenue NRB 1042 Cincinnati, OH 45229 (www.cincinnatichildrens.org)				15-Nov-17
Beckwith-Wiedemann syndrome: CDKN1C Sequencing (Step 3)	571	CDKN1C (608856)	Lavender Top (EDTA) [2 tubes]	Genome Diagnostics The Department of Paediatric Laboratory Medicine The Hospital for Sick Children	Wiedemann-Beckwith Syndrome (130650)			19-Feb-20
Beckwith-Wiedemann syndrome: Methylation & Copy Number (Step1)	571		Lavender Top (EDTA) [2 tubes]	Genome Diagnostics The Department of Paediatric Laboratory Medicine The Hospital for Sick Children	Wiedemann-Beckwith Syndrome (130650)	Methylation-Specific-MLPA of imprinting centers 1 and 2		19-Feb-20
Beckwith-Wiedemann syndrome: UPD11 Analysis (Step 2)	571	H19 (102280), KCNQ1OT1 (604115), CDKN1C (608856)	Lavender Top (EDTA) [2 tubes]	Genome Diagnostics The Department of Paediatric Laboratory Medicine The Hospital for Sick Children	Wiedemann-Beckwith Syndrome (130650)	UPD11 studies via STR (short tandem repeat) analysis.		19-Feb-20
Beta-Thalassemia (613985)	823	HBB (141900)	Lavender Top (EDTA)	Molecular Genetics Laboratory - McMaster University Medical Centre		Sanger sequencing		
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 reflex test from Bile Acids, Urine. Both sample should be sent if possible.	FAB-MS		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		FAB-MS	NOT AVAILABLE AT MCL	08-Aug-17
Bile Acids for Peroxisomal Disorders, Serum [BAIPD]	9		Serum. Store at 4°C or -20°C. Stable for 90 days.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)		LC-MS/MS		12-Jul-22
Birt-Hogg-Dubé Syndrome (135150)	740	FLCN (607273)	Lavender Top (EDTA)	Alberta Mol Dx Laboratory (Calgary)		NGS		16-Jun-20

Appellation 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 (Déclaration MSSS)	SYMBOL/ICDNC DU GÈNE et NOM OMIM DU GÈNE / ICDNC Gene Symbol and OMIM Gene Number	RÈGLES DE MANUTENTION / Handling Conditions	LABORATOIRE DÉSIGNÉ / Designated laboratory	RENSEIGNEMENTS SUR L'ANALYSE / Test details	MÉTHODE / Methode	NOTE PARTICULIÈRE / Special comment	Date de la révision / Date of revision
I,5-anhydroglucitol (GlycoMark) [500115] [GLYC]	1465		Gold Top or Red Top (Serum) or Lavender (plasma). Centrifuge and aliquot serum to transport tube within one hour of collection. Store and ship frozen. Stable at -20°C for 14 days.	LabCorp/Dynacare (https://www.labcorp.com/)		Enzymatic, colorimetric assay.		10-Jan-21
Blastomyces Antibody by Immunodiffusion [322]	491		Serum: Collect serum specimens in serum separator or red top tube. Allow blood to clot for 30 minutes, then centrifuge. Pipette serum into a plastic screw cap vial. STABILITY: Refrigerated: 14 days Frozen: 6 months	MiraVista Diagnostics				08-Sep-22
Blastomyces Quantitative EIA [316]	491		Serum: Collect serum specimens in serum separator or red top tube. Allow blood to clot for 30 minutes, then centrifuge. Pipette serum into a plastic screw cap vial. Plasma: Collect plasma specimens in an EDTA, heparin or sodium citrate tube. Centrifuge for 15 minutes and pipette plasma into a plastic screw cap vial. Urine/CSF/BAL/Other Body Fluid: Submit urine, CSF, BAL and all other body fluids in a sterile screw cap container. STABILITY Room Temperature: 2 weeks Refrigerated: 2 weeks Frozen: Indefinitely	MiraVista Diagnostics				08-Sep-22
BMPR1A, SMAD4 Gene Sequencing & Del/Dup [717]		SMAD4 (600993); BMPR1A (601299)	Lavender Top (EDTA)	GeneDx (www.genedx.com)	174900 (juvenile polyposis syndrome); 175050 (juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome)	Sanger Sequencing, Exon Array CGH		09-Dec-19
Bone Marrow Failure Syndrome Panel [HE0801]	1427	135 genes	Lavender Top (EDTA) [2 tubes]	Blueprint Genetics http://blueprintgenetics.com/		NextGen Sequencing		28-Feb-20
BP 180 and BP230	750		Serum	Immunodermatology Laboratory Department of Dermatology 30 North 1900 East, 4A330 SOM Salt Lake City, Utah 84132 Email: immunoderm@hsc.utah.edu		ELISA		14-Sep-20
Brain-Iron Accumulation NGS Panel	11213	ATP13A2, C19orf12, COASY, CP, DCAF17, FA2H, FTL, GTPBP2, PANK2, PLA2G6, SCFD3, WDR45	Lavender Top (EDTA) 2 x 4 mL	Fulgent Genetics (fulgentdiagnostics.com)		NextGen Sequencing		28-Feb-20
Breast and Gynaecological Cancer Specific Panel	227		Lavender Top (EDTA)	Invitae (www.invitae.com)	This is a special panel from the Cedars cancer Centre.	Next-Generation sequencing		06-Feb-20
Breast Cancer Panel (Invitae) [01202]	227	ATM (1), BARD1 (1), BRCA1 (113705), BRCA2 (600185), BRIP1 (1), CDH1 (192990), CHEK2 (1), NBN (1), NFI (1), PALB2 (601726), PTEN (601726), RAD50 (1), STK11 (602216), TP53 (91170)	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 (1); Pancreatic cancer (260350); Peutz-Jeghers syndrome (175200); Breast cancer (114480)	Next-Generation sequencing		07-Oct-18
Brivacetam [0718SP]	1465		Red Top or Lavender Top only. Store and send serum/plasma frozen.	NMS laboratories		LC-MS/MS		17-Mar-22
Bromine - Total Blood [FBROM]	1465		Royal Blue (EDTA) top tube	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)		ICP/MS	INSPQ?? TO BE VERIFIED - NO	30-Apr-21
Bullous Autoimmune Skin Disease Profile	750		Serum gel (Gold top)	Mitogen Advanced Diagnostics	BP180, BP230, Desmoglein 1, Desmoglein 3	CBA		02-Jun-21

Appellation 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 (Déclaration MSSS)	SYMBOL HGENC DU GÈNE et NOM OMIM DU GÈNE / HGENC Gene Symbol and OMIM Gene Number	RÈGLES DE MANUTENTION / Handling Conditions	LABORATOIRE DÉSIGNÉ / Designated laboratory	RENSEIGNEMENTS SUR L'ANALYSE / Test details	MÉTHODE / Methode	NOTE PARTICULIÈRE / Special comment	Date de la révision / Date of revision
I,5-anhydroglucitol (GlycoMark) [500115] [GLYC]	1465		Gold Top or Red Top (Serum) or Lavender (plasma). Centrifuge and aliquot serum to transport tube within one hour of collection. Store and ship frozen. Stable at -20°C for 14 days.	LabCorp/Dynacare (https://www.labcorp.com/)		Enzymatic, colorimetric assay.		10-Jan-21
C3 Nephritic Factor	792		1 ml frozen serum	Molecular Otolaryngology & Renal Research Laboratory, U. of Iowa Carver College of Medicine (www.medicine.uiowa.edu/mor/)	Dense Deposit Disease (DDD, aka Membranoproliferative Glomerulonephritis Type II, MPGNII)	1. Immunofixation electrophoresis (IFE) combines the techniques of electrophoresis with immunofixation to detect C3 degradation products, an indirect measure of dysregulation of C3 convertase. Conversion of C3 to C3c is quantitated (Koch, et al., 1981). 2. C3 Convertase Stabilizing Assay (C3CSA) measures the ability of C3Nefs to stabilize C3 convertase on sheep erythrocytes. 3. C3 Convertase Stabilizing Assay with Properdin (C3CSAP) measures the ability of C3Nefs to stabilize C3 convertase built with properdin on sheep erythrocytes.	Available as Facteur C3 néphritique (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 néphritique (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/mor/)	DDD & C3GN	serology		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 and ATXN2 [13039]	853	C9orf72 (644260); SOD1 (147450)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Antyoptic lateral sclerosis		If C9orf72 is only performed, then the price is only \$250 USD	02-Nov-20
CACNA1A Full Gene Sequencing Analysis [MOL033]		CACNA1A (60101)	Lavender Top (EDTA)	MNG Laboratories (www.medicalneurogenetics.com)/Dynacare	Familial hemiplegic migraine 1 (141500); episodic ataxia type 2 (108500)	Sanger sequencing		14-May-20
CACNB4 Full Gene Sequencing Analysis [MOL227]		CACNB4 (601949)	Lavender Top (EDTA)	MNG Laboratories (www.medicalneurogenetics.com)/Dynacare	Episodic ataxia, type 5 (613855)	Sanger sequencing		14-May-20
Calpain 3 DNA Sequencing Test [563]		CAPN3 (114240)	Lavender Top (EDTA)	Athena Diagnostics (www.athenadiagnostics.com)	LGM2A (253600), Calpainopathy			
CAMURATI-ENGELMANN DISEASE (CED) VIA THE TGFB1 GENE [787]		TGFB1 (190180)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Camurati-Engelmann Disease (131300)	Sanger sequencing		
CAR Panel by Immunoblot and Immunohistochemistry [CARP]	720		Serum. Store at 4°C. Stable for 7 d only. DO NOT FREEZE.	Ocular Immunology Laboratory, OHSU Biomedical Research Building, Room 253 3181 SW Sam Jackson Park Road Portland, OR 97239, USA				03-Aug-22
Carbohydrate Deficient Transferin for Congenital Disorders of Glycosylation, Serum [CDG]	406		Red Top. Store serum frozen.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)		Affinity Chromatography-Mass Spectrometry	This is a confirmatory test following the screening test for CDT testing at the MUHC.	16-Dec-20
Carbohydrate, Urine [CHOU]	1572		Random urine. Store frozen. Stable 21 days.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	Replaces "reducing substances"	TLC		31-Oct-20
Cardio-Facio-Cutaneous Syndrome NGS Panel (5 genes)		BRAF (164757), KRAS (190079), MAP2K1 (176872), MAP2K2 (601263), SOS1 (182580)	Lavender Top (EDTA) 2 x 4 mL	Fulgent Genetics (fulgentdiagnostics.com)	Cardiofaciocutaneous syndrome (115150); Cardiofaciocutaneous syndrome 2 (615278); Cardiofaciocutaneous syndrome 3 (615279); Cardiofaciocutaneous syndrome 4 (615280); Fibromatosis, gingival (135300)	NextGen Sequencing		
Cathartic Laxatives Profile, Stool [FCLPS]			Stool	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	Mg, Pi			20-Oct-20

Appellation 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 HUGO DU GÈNE et NOM GDM DU GÈNE / HGNC Gene Symbol/OMIM Gene Number	RÈGLES DE MANUTENTION / Handling Conditions	LABORATOIRE DÉSIGNÉ / Designated laboratory	RENSEIGNEMENTS SUR L'ANALYSE / Test details	MÉTHODE / Methode	NOTE PARTICULIÈRE / Special comment	Date de la révision / Date of revision
1,5-anhydroglucitol (GlycoMark) [500115] [GLYC]	1465		Gold Top or Red Top (Serum) or Lavender (plasma). Centrifuge and aliquot serum to transport tube within one hour of collection. Store and ship frozen. Stable at -20°C for 14 days.	LabCorp/Dynacare (https://www.labcorp.com/)		Enzymatic, colorimetric assay.		10-Jan-21
CAVEOLINOPATHY TESTING VIA THE CAV3 GENE [467]		CAV3 (601253)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Muscular Dystrophy, Limb-Girdle, Type 1C (607801)	Sanger sequencing		
CDKN2A (p16) & CDK4 (Exon2) Sequencing [2021]		CDKN2A (601606); CDK4 (12829)	Lavender Top (EDTA)	GeneDx (www.genedx.com)	Melanoma, cutaneous malignant, 3			09-Dec-19
Central Hypoventilation And Apnea Panel [PU0401]	775	CHAT, CHRNA3, CHRNA1, CHRNA2, CHRNA4, CHRNA5, CHRNA6, CHRNA7, CHRNA8, CHRNA9, CHRNA10, CHRNA11, CHRNA12, CHRNA13, CHRNA14, CHRNA15, CHRNA16, CHRNA17, CHRNA18, CHRNA19, CHRNA20, CHRNA21, CHRNA22, CHRNA23, CHRNA24, CHRNA25, CHRNA26, CHRNA27, CHRNA28, CHRNA29, CHRNA30, CHRNA31, CHRNA32, CHRNA33, CHRNA34, CHRNA35, CHRNA36, CHRNA37, CHRNA38, CHRNA39, CHRNA40, CHRNA41, CHRNA42, CHRNA43, CHRNA44, CHRNA45, CHRNA46, CHRNA47, CHRNA48, CHRNA49, CHRNA50, CHRNA51, CHRNA52, CHRNA53, CHRNA54, CHRNA55, CHRNA56, CHRNA57, CHRNA58, CHRNA59, CHRNA60, CHRNA61, CHRNA62, CHRNA63, CHRNA64, CHRNA65, CHRNA66, CHRNA67, CHRNA68, CHRNA69, CHRNA70, CHRNA71, CHRNA72, CHRNA73, CHRNA74, CHRNA75, CHRNA76, CHRNA77, CHRNA78, CHRNA79, CHRNA80, CHRNA81, CHRNA82, CHRNA83, CHRNA84, CHRNA85, CHRNA86, CHRNA87, CHRNA88, CHRNA89, CHRNA90, CHRNA91, CHRNA92, CHRNA93, CHRNA94, CHRNA95, CHRNA96, CHRNA97, CHRNA98, CHRNA99, CHRNA100	Lavender Top (EDTA)	Blueprint Genetics http://blueprintgenetics.com/		Next-Generation sequencing		26-Sep-18
CEREBRAL CAVERNOUS MALFORMATIONS PANEL [1943]	823	KRT1 (602134), CCM2 (607292), PCCD10 (609118)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Cerebral Cavernous Malformations 1 (116860), Cerebral Cavernous Malformations 2 (603284), Cerebral Cavernous Malformations 3 (603285)	Sequencing and CNV Detection via NextGen Sequencing using PG-Select Capture Probes		02-Feb-20
CEREBRAL CAVERNOUS MALFORMATIONS VIA THE CCM2 GENE (603284) [122]		CCM2 (607292)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Cerebral cavernous malformations-2	Sanger sequencing		
CEREBRAL CAVERNOUS MALFORMATIONS VIA THE KRT1/CCM1 GENE [121]		KRT1 (602134)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Cerebral cavernous malformations-1 (116860)	Sanger sequencing		
CEREBRAL CAVERNOUS MALFORMATIONS VIA THE PCCD10/CCM3 (603285) [123]		PCCD10 (609118)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Cerebral cavernous malformation-3	Sanger sequencing		
Cerebral Small Vessel Diseases Panel [12637]	1582	COL4A1, COL4A2, COLGALT1, CTCC1, CTSX, FOXO1, GLA, HTRA1, NOTCH3, TRKH1	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)		Next-Generation sequencing		16-Nov-21
CerebroTendinous Xanthomatosis (CTX) via the CYP27A1 Gene [1670]		CYP27A1 (606330)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	CerebroTendinous Xanthomatosis (213700)	Sanger sequencing		31-Mar-17
Charcot-Marie-Tooth Disease (NextGen Sequencing Panel and Copy Number Analysis + mtDNA) [NGS345]	643	AAAS (605758), AARS (601005), ABCD1 (300371), ACO2 (10880), ACOX1 (609751), APGL2 (605881), APM1 (300169), ALDH3A2 (609523), ALDH3A1 (601376), ALG2 (607965), AMPD2 (602771), AP2E2 (606036), AP2E3 (607246), AP4M1 (602296), APTX (606350), ARCN1 (608820), ARHGAP10 (608136), ARNT2 (609036), ARSA (607574), ANS1 (608750), ASPA (608014), ATAD1A (602316), ATP13A1 (601051), ATP7B (606082), BCL2L1 (600783), BAGO1 (603883), BRF1 (602410), BSLC2 (606158), C10ORF2 (600075), C12A5 (601354), C19orf12 (614279), CLP1 (609575), COQ7 (601681), CUX1 (602072), CSFR1 (604778), CTDP1 (609073), CYBB (116086), CYP21A1 (601676), DARS2 (610956), DDHD1 (614403), DDOST (602252), DGCR8 (601405), DHRF1 (126069), DHTKD1 (614984), DNAAF2 (604139), DNAAF3 (601184), DNAAF4 (609530), DNAAF5 (602378), DNAAF6 (608810), DNAAF7 (600112), EGR2 (126016), ELOVL4 (605512), ELOVL5 (611805), ERCC3 (133510), ERCC5 (609413), ERCC8 (609412), ERCC9 (608489), FBLN5 (604580), FGD4 (611104), FGF14 (601515), FGF4 (605950), FLYC1E1 (609144), FOLR1 (136430), FOXG1 (166874), FOXO2 (605153), FTL (134750), GAA (606800), GABRB3 (137192), GALT (606890), GAN (605379), GARS (600287), GBE1 (607879), GDAP1 (606398), GIP (609342), GJB1 (300400), GJC2 (608803), GJL1 (138250), GNDA (610061), GP6B (604110), HADHA (143450), HARS (142810), HNT1 (601314), HK1 (142600), HSD17B4 (601800), HSPB1 (602195), HSPB8 (608044), HTRA1 (602194), IARS2 (612801), IBA57 (615150), IERBP1 (609082), IERBP2 (609082), INP2 (609882), ITPA (145720), KARS (601421), KIDINS220 (615799), KIAA1 (602155), KIF1B (605995), KIF5A (602821), LITAF (607795), LMAN1 (150310), LRXAM1 (601919), MAN1A (609489), MARS1 (166560), MATA1 (610550), MED22 (610177), MENC (601807), MICU1 (605684), MME1 (128230), MOC51 (607507), MOC52 (607390), MPZ (119440), MRPS22 (608810), MTRMR2 (601357), MTPP1 (157147), NAGA (104170), NAE1 (607910), NARS2 (612803), NARG1 (605252), NEFH (142230), NEFL (162280), OFAI1 (602290), PC (608786), PDK3 (300906), PDYN (131340), PEX30 (602599), PEX11B (601867), PEX16 (601360), PEX19 (602279), PEX2 (170993), PEX5 (604140), PEX7 (607157), PFBP3 (606419), PFKFB3 (172110), PFKFB2 (606879), PKD1 (602744), PLA2G6 (603004), PLEKHG5 (611101), PMP22 (601097), PNP1A6 (603197), SGK196 (615247), PRDM12 (616458), PRICKLE1 (608800), PRMT7 (601867), PRPF8 (111880), PRC5 (607270), PRPAP (170810), PURA (600473), PYROXD1 (617220), RAB7A (602298), RANBP1 (614917), RNASEH1 (604123), SBF1 (601360), SBF2 (607097), SEPT9 (604061), SH3TC2 (608206), SLC12A1 (604870), SLC12A12 (609095), SLC12A14 (606229), SLC25A1 (190315), SLC25A12 (603667), SLC25A19 (608521), SLC25A22 (609302), SLC25A46 (609826), SLC35A2 (314375), SPRY17A (607882), SLC6A8 (300036), SPGL1 (609844), SPPL1 (158310), SUCLA2 (609251), SUCRA1 (607930), SURF1 (185620), TBC1D24 (613577), TDP1 (607198), TFG (602498), TMEM126A (612988), TMTC3 (617210), TRIM2 (614414), TRMT5 (611023), TRPV4 (605427), TRPM (60723), TRK (176300), TUBB3 (602614), TYMP (112221), UBA5 (605052), VCP (601023), VPS3A (610034), YARS (603623), ZC4H2 (300897), ZNF315 (610827)	Lavender Top (EDTA)	MNG Laboratories (www.medicalneurogenetics.com)/Dynacare	Charcot-Marie tooth Disease (302800)	Next-Generation sequencing	Alt: CTGT 42 genes & del/dupl	14-May-20
Childhood-Onset Epilepsy Panel [542]	1031	ADSL, ALDH5A1, ATP1A3, ATRX, CACNA1A, CASK, CDKL5, CHD2, CHRNA2, CHRNA3, CHRNA4, CHRNA5, CHRNA6, CHRNA7, CHRNA8, CHRNA9, CHRNA10, CHRNA11, CHRNA12, CHRNA13, CHRNA14, CHRNA15, CHRNA16, CHRNA17, CHRNA18, CHRNA19, CHRNA20, CHRNA21, CHRNA22, CHRNA23, CHRNA24, CHRNA25, CHRNA26, CHRNA27, CHRNA28, CHRNA29, CHRNA30, CHRNA31, CHRNA32, CHRNA33, CHRNA34, CHRNA35, CHRNA36, CHRNA37, CHRNA38, CHRNA39, CHRNA40, CHRNA41, CHRNA42, CHRNA43, CHRNA44, CHRNA45, CHRNA46, CHRNA47, CHRNA48, CHRNA49, CHRNA50, CHRNA51, CHRNA52, CHRNA53, CHRNA54, CHRNA55, CHRNA56, CHRNA57, CHRNA58, CHRNA59, CHRNA60, CHRNA61, CHRNA62, CHRNA63, CHRNA64, CHRNA65, CHRNA66, CHRNA67, CHRNA68, CHRNA69, CHRNA70, CHRNA71, CHRNA72, CHRNA73, CHRNA74, CHRNA75, CHRNA76, CHRNA77, CHRNA78, CHRNA79, CHRNA80, CHRNA81, CHRNA82, CHRNA83, CHRNA84, CHRNA85, CHRNA86, CHRNA87, CHRNA88, CHRNA89, CHRNA90, CHRNA91, CHRNA92, CHRNA93, CHRNA94, CHRNA95, CHRNA96, CHRNA97, CHRNA98, CHRNA99, CHRNA100	Lavender Top (EDTA) [2 tubes]	GeneDx (www.genedx.com)		NextGen Sequencing		28-Feb-20

Appellation 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 (Déclencheur MSSS)	SYMBOL HGNC DU GÈNE et NOM OMIM DU GÈNE / HGNC Gene Symbol and OMIM Gene Number	RÈGLES DE MANUTENTION / Handling Conditions	LABORATOIRE DÉSIGNÉ / Designated laboratory	RENSEIGNEMENTS SUR L'ANALYSE / Test details	MÉTHODE / Methode	NOTE PARTICULIÈRE / Special comment	Date de la révision / Date of revision
1,5-anhydroglucitol (GlycoMark) [500115] [GLYC]	1465		Gold Top or Red Top (Serum) or Lavender (plasma). Centrifuge and aliquot serum to transport tube within one hour of collection. Store and ship frozen. Stable at -20°C for 14 days.	LabCorp/Dynacare (https://www.labcorp.com/)		Enzymatic, colorimetric assay.		10-Jan-21
Cholestasis Panel	400	ABCB11 MYO8B* NOTCH2* NR1H2 NPH1 PEX1 PEX2 PEX3 PEX4 PEX5 PEX6 SERPINA1 SLC2A11 SLC26A3 SMPD1 SPT2 TP2 TMEM216 TRMT1L TTC37 UGT1A1 VPS33B VPS33B ABCB4 ABCB2 AKR1D1 ATP8B1 BAAT CFTR CREB3L3 CYP8B1 DCDC2 DGLK1 EPCAM 80 FAH HSD17B7 JAG1 LCT	Lavender Top (EDTA)	Blueprint Genetics http://blueprintgenetics.com/		NextGen Sequencing		28-May-19
Choreoacanthocytosis (200150)		VPS11A (605976)	Lavender Top (EDTA)	North York General		2 mutations		
Christianson Type X-Linked Mental Retardation via the SLC9A6 Gene [562]		SLC9A6 (30231)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Mental Retardation, X-Linked, Syndromic, Christianson Type (30231)	Sanger sequencing		
Chronic Granulomatous Disease NextGen Sequencing (NGS) Panel [1971]		CYBA (60858); CYBB (30081); NCF2 (608515); NCF4 (601488)	Lavender Top (EDTA)	Fulgent Genetics (fulgentdiagnostics.com)		NextGen Sequencing		
Chronic Inflammatory Demyelinating Polyneuropathy (CIDP) Test Panel - Nodal and Paranodal Antibodies	336	Neurofascin 140 (NF140), Neurofascin 186 (NF186), Contactin-1 (CNTN1), Contactin-associated protein 1 (CASPR1), and Neurofascin 155 (NF155)	Serum ONLY. Store at -20°C.	BC Neuroimmunology (bcneuro.ca)		CBA fixed	Replaces contactin and neurofascin testing at Washington U.	09-Aug-22
Chronic Pancreatitis NGS Panel	402	GASR, CFTR, CTSC, PRSS1, SPINK1	Lavender Top (EDTA) 2 x 4 mL	Fulgent Genetics (fulgentdiagnostics.com)		NextGen Sequencing		08-Feb-19
Chyluria Screen, Random Urine [CSU]	1465		First morning random urine. Store frozen. Stable 10 days.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)		electrophoresis		13-Sep-21
CLCN1 Full Gene sequencing Analysis [MOL:355]; Paramyotonia Congenita (168300)	607	CLCN1 (118425)	Lavender Top (EDTA)	MNG Laboratories (www.medicalneurogenetics.com)/Dynacare	Paramyotonia Congenita (168300)	Sanger sequencing		14-May-20
Coccidioides Antibody by Immunodiffusion [320]	493		Serum: Collect serum specimens in serum separator or red top tube. Allow blood to clot for 30 minutes, then centrifuge. Pipette serum into a plastic screw cap vial. STABILITY Refrigerated: 14 days Frozen: 6 months	MiraVista Diagnostics				08-Sep-22
Coccidioides Antibody IgG, IgM EIA [325]	493		Serum: Collect serum specimens in serum separator or red top tube. Allow blood to clot for 30 minutes, then centrifuge. Pipette serum into a plastic screw cap vial. CSF: Sterile transport tube STABILITY Refrigerated: 14 days Frozen: 14 days	MiraVista Diagnostics				08-Sep-22
Coccidioides Quantitative EIA [315]	493		Serum: Collect serum specimens in serum separator or red top tube. Allow blood to clot for 30 minutes, then centrifuge. Pipette serum into a plastic screw cap vial. Plasma: Collect plasma specimens in an EDTA, heparin or sodium citrate tube. Centrifuge for 15 minutes and pipette plasma into a plastic screw cap vial. Urine/CSF/BAL/Other Body Fluids: Submit urine, CSF, BAL, and all other body fluids in a sterile screw cap container. STABILITY Room Temperature: 14 days Refrigerated: 14 days Frozen: Indefinitely	MiraVista Diagnostics				08-Sep-22
COCKAYNE SYNDROME VIA THE ERCC6 GENE [1008]		ERCC6 (69413)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Cockayne Syndrome, Type B (133540)	Sanger sequencing		

Appellation 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 (Déclassement MSSS)	SYMBOL / IGENC DU GÈNE et NOM OMIM DU GÈNE / IGENC Gene Symbol and OMIM Gene Number	RÈGLES DE MANUTENTION / Handling Conditions	LABORATOIRE DÉSIGNÉ / Designated laboratory	RENSEIGNEMENTS SUR L'ANALYSE / Test details	MÉTHODE / Methode	NOTE PARTICULIÈRE / Special comment	Date de la révision / Date of revision
1,5-anhydroglucitol (GlycoMark) [500115] [GLYC]	1465		Gold Top or Red Top (Serum) or Lavender (plasma). Centrifuge and aliquot serum to transport tube within one hour of collection. Store and ship frozen. Stable at -20°C for 14 days.	LabCorp/Dynacare (https://www.labcorp.com/)		Enzymatic, colorimetric assay.		10-Jan-21
Collagen Type II Antibodies [FFTYC]	1399		Red Top; Gold SST. Stable for 7 d at 4°C or longer at -20°C.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)		Enzyme Linked Immunosorbent Assay (ELISA)		15-May-19
Collagen VII IgG Antibody Level	1399		Red Top or Gold SST. Stability Ambient: 7 days Refrigerated: 14 days Frozen: Indefinitely	Immunodermatology Laboratory Department of Dermatology 30 North 1900 East, 4A330 SOM Salt Lake City, Utah 84132 Email: immunoderm@hsc.utah.edu		Enzyme Linked Immunosorbent Assay (ELISA)		06-Jul-20
Colon Cancer NGS Panel	231	APC, AMN2, BMP3A, BRIB1, BRIB2, CDH1, CDKN2A, CHEK2, EPCAM, EXO1, F1CN, GALNT1, MLH1, MSH2, MSH6, MUTH1, PMS1, PMS2, PTEN, SMAD4, STK11, TP53	Lavender Top (EDTA) 2 x 4 mL	Fulgent Genetics (fulgentdiagnostics.com)		NextGen Sequencing		December 12, 2018
Colorectal cancer (Li-Fraumeni syndrome)	231	TP53 (191170)	Lavender Top (EDTA)	Molecular Genetics Laboratory, Hospital for Sick Children (www.sickkids.ca/PaediatricLaboratoryMedicine/Laboratories-Services/Molecular-Genetics-Laboratory/index.html)	Colorectal cancer (114500); Li-Fraumeni syndrome (151623)	1. Sanger sequencing 2. gene dosage		
ColoVantage (Methylated Septin 9) [16983]	1395	SEPT9	Plasma (EDTA); handle at 4°C; freeze plasma in plastic container. Minimum 10 mL.	Quest Diagnostics Chantilly 14225 Newbrook Dr. Chantilly, VA 20153-0841				11-Apr-18
Combined Mito Genome Plus Mito Nuclear Gene Panel [615]	518	AARS, AARS2, ABCB11, ABCB4, ABCB7, ABCB9, ACAD9, ACADM, ACADVL, ACAD12, ACSF3, ACOX1, CACR1, COX6B, ACOX4, AGLL2, AGL3, AGL, AIFM1, ALAS2, ALDOA, ALDOB, ALG1, ALG11, ALG2, ALG3, ALG4, ALG9, AMACR, APOPT1, APTX, ARGL, ASL, ASS1, ATP5A1, ATP5E, ATP7B, ATP8A1, ATP9A2, ATP12, AXL, BBS1, BBS2, BBS3, BBS4, BBS5, BBS6, BBS7, BBS8, BBS9, BBS10, BBS11, BBS12, BBS13, BBS14, BBS15, BBS16, BBS17, BBS18, BBS19, BBS20, BBS21, BBS22, BBS23, BBS24, BBS25, BBS26, BBS27, BBS28, BBS29, BBS30, BBS31, BBS32, BBS33, BBS34, BBS35, BBS36, BBS37, BBS38, BBS39, BBS40, BBS41, BBS42, BBS43, BBS44, BBS45, BBS46, BBS47, BBS48, BBS49, BBS50, BBS51, BBS52, BBS53, BBS54, BBS55, BBS56, BBS57, BBS58, BBS59, BBS60, BBS61, BBS62, BBS63, BBS64, BBS65, BBS66, BBS67, BBS68, BBS69, BBS70, BBS71, BBS72, BBS73, BBS74, BBS75, BBS76, BBS77, BBS78, BBS79, BBS80, BBS81, BBS82, BBS83, BBS84, BBS85, BBS86, BBS87, BBS88, BBS89, BBS90, BBS91, BBS92, BBS93, BBS94, BBS95, BBS96, BBS97, BBS98, BBS99, BBS100, BBS101, BBS102, BBS103, BBS104, BBS105, BBS106, BBS107, BBS108, BBS109, BBS110, BBS111, BBS112, BBS113, BBS114, BBS115, BBS116, BBS117, BBS118, BBS119, BBS120, BBS121, BBS122, BBS123, BBS124, BBS125, BBS126, BBS127, BBS128, BBS129, BBS130, BBS131, BBS132, BBS133, BBS134, BBS135, BBS136, BBS137, BBS138, BBS139, BBS140, BBS141, BBS142, BBS143, BBS144, BBS145, BBS146, BBS147, BBS148, BBS149, BBS150, BBS151, BBS152, BBS153, BBS154, BBS155, BBS156, BBS157, BBS158, BBS159, BBS160, BBS161, BBS162, BBS163, BBS164, BBS165, BBS166, BBS167, BBS168, BBS169, BBS170, BBS171, BBS172, BBS173, BBS174, BBS175, BBS176, BBS177, BBS178, BBS179, BBS180, BBS181, BBS182, BBS183, BBS184, BBS185, BBS186, BBS187, BBS188, BBS189, BBS190, BBS191, BBS192, BBS193, BBS194, BBS195, BBS196, BBS197, BBS198, BBS199, BBS200, BBS201, BBS202, BBS203, BBS204, BBS205, BBS206, BBS207, BBS208, BBS209, BBS210, BBS211, BBS212, BBS213, BBS214, BBS215, BBS216, BBS217, BBS218, BBS219, BBS220, BBS221, BBS222, BBS223, BBS224, BBS225, BBS226, BBS227, BBS228, BBS229, BBS230, BBS231, BBS232, BBS233, BBS234, BBS235, BBS236, BBS237, BBS238, BBS239, BBS240, BBS241, BBS242, BBS243, BBS244, BBS245, BBS246, BBS247, BBS248, BBS249, BBS250, BBS251, BBS252, BBS253, BBS254, BBS255, BBS256, BBS257, BBS258, BBS259, BBS260, BBS261, BBS262, BBS263, BBS264, BBS265, BBS266, BBS267, BBS268, BBS269, BBS270, BBS271, BBS272, BBS273, BBS274, BBS275, BBS276, BBS277, BBS278, BBS279, BBS280, BBS281, BBS282, BBS283, BBS284, BBS285, BBS286, BBS287, BBS288, BBS289, BBS290, BBS291, BBS292, BBS293, BBS294, BBS295, BBS296, BBS297, BBS298, BBS299, BBS300, BBS301, BBS302, BBS303, BBS304, BBS305, BBS306, BBS307, BBS308, BBS309, BBS310, BBS311, BBS312, BBS313, BBS314, BBS315, BBS316, BBS317, BBS318, BBS319, BBS320, BBS321, BBS322, BBS323, BBS324, BBS325, BBS326, BBS327, BBS328, BBS329, BBS330, BBS331, BBS332, BBS333, BBS334, BBS335, BBS336, BBS337, BBS338, BBS339, BBS340, BBS341, BBS342, BBS343, BBS344, BBS345, BBS346, BBS347, BBS348, BBS349, BBS350, BBS351, BBS352, BBS353, BBS354, BBS355, BBS356, BBS357, BBS358, BBS359, BBS360, BBS361, BBS362, BBS363, BBS364, BBS365, BBS366, BBS367, BBS368, BBS369, BBS370, BBS371, BBS372, BBS373, BBS374, BBS375, BBS376, BBS377, BBS378, BBS379, BBS380, BBS381, BBS382, BBS383, BBS384, BBS385, BBS386, BBS387, BBS388, BBS389, BBS390, BBS391, BBS392, BBS393, BBS394, BBS395, BBS396, BBS397, BBS398, BBS399, BBS400, BBS401, BBS402, BBS403, BBS404, BBS405, BBS406, BBS407, BBS408, BBS409, BBS410, BBS411, BBS412, BBS413, BBS414, BBS415, BBS416, BBS417, BBS418, BBS419, BBS420, BBS421, BBS422, BBS423, BBS424, BBS425, BBS426, BBS427, BBS428, BBS429, BBS430, BBS431, BBS432, BBS433, BBS434, BBS435, BBS436, BBS437, BBS438, BBS439, BBS440, BBS441, BBS442, BBS443, BBS444, BBS445, BBS446, BBS447, BBS448, BBS449, BBS450, BBS451, BBS452, BBS453, BBS454, BBS455, BBS456, BBS457, BBS458, BBS459, BBS460, BBS461, BBS462, BBS463, BBS464, BBS465, BBS466, BBS467, BBS468, BBS469, BBS470, BBS471, BBS472, BBS473, BBS474, BBS475, BBS476, BBS477, BBS478, BBS479, BBS480, BBS481, BBS482, BBS483, BBS484, BBS485, BBS486, BBS487, BBS488, BBS489, BBS490, BBS491, BBS492, BBS493, BBS494, BBS495, BBS496, BBS497, BBS498, BBS499, BBS500, BBS501, BBS502, BBS503, BBS504, BBS505, BBS506, BBS507, BBS508, BBS509, BBS510, BBS511, BBS512, BBS513, BBS514, BBS515, BBS516, BBS517, BBS518, BBS519, BBS520, BBS521, BBS522, BBS523, BBS524, BBS525, BBS526, BBS527, BBS528, BBS529, BBS530, BBS531, BBS532, BBS533, BBS534, BBS535, BBS536, BBS537, BBS538, BBS539, BBS540, BBS541, BBS542, BBS543, BBS544, BBS545, BBS546, BBS547, BBS548, BBS549, BBS550, BBS551, BBS552, BBS553, BBS554, BBS555, BBS556, BBS557, BBS558, BBS559, BBS560, BBS561, BBS562, BBS563, BBS564, BBS565, BBS566, BBS567, BBS568, BBS569, BBS570, BBS571, BBS572, BBS573, BBS574, BBS575, BBS576, BBS577, BBS578, BBS579, BBS580, BBS581, BBS582, BBS583, BBS584, BBS585, BBS586, BBS587, BBS588, BBS589, BBS590, BBS591, BBS592, BBS593, BBS594, BBS595, BBS596, BBS597, BBS598, BBS599, BBS600, BBS601, BBS602, BBS603, BBS604, BBS605, BBS606, BBS607, BBS608, BBS609, BBS610, BBS611, BBS612, BBS613, BBS614, BBS615, BBS616, BBS617, BBS618, BBS619, BBS620, BBS621, BBS622, BBS623, BBS624, BBS625, BBS626, BBS627, BBS628, BBS629, BBS630, BBS631, BBS632, BBS633, BBS634, BBS635, BBS636, BBS637, BBS638, BBS639, BBS640, BBS641, BBS642, BBS643, BBS644, BBS645, BBS646, BBS647, BBS648, BBS649, BBS650, BBS651, BBS652, BBS653, BBS654, BBS655, BBS656, BBS657, BBS658, BBS659, BBS660, BBS661, BBS662, BBS663, BBS664, BBS665, BBS666, BBS667, BBS668, BBS669, BBS670, BBS671, BBS672, BBS673, BBS674, BBS675, BBS676, BBS677, BBS678, BBS679, BBS680, BBS681, BBS682, BBS683, BBS684, BBS685, BBS686, BBS687, BBS688, BBS689, BBS690, BBS691, BBS692, BBS693, BBS694, BBS695, BBS696, BBS697, BBS698, BBS699, BBS700, BBS701, BBS702, BBS703, BBS704, BBS705, BBS706, BBS707, BBS708, BBS709, BBS710, BBS711, BBS712, BBS713, BBS714, BBS715, BBS716, BBS717, BBS718, BBS719, BBS720, BBS721, BBS722, BBS723, BBS724, BBS725, BBS726, BBS727, BBS728, BBS729, BBS730, BBS731, BBS732, BBS733, BBS734, BBS735, BBS736, BBS737, BBS738, BBS739, BBS740, BBS741, BBS742, BBS743, BBS744, BBS745, BBS746, BBS747, BBS748, BBS749, BBS750, BBS751, BBS752, BBS753, BBS754, BBS755, BBS756, BBS757, BBS758, BBS759, BBS760, BBS761, BBS762, BBS763, BBS764, BBS765, BBS766, BBS767, BBS768, BBS769, BBS770, BBS771, BBS772, BBS773, BBS774, BBS775, BBS776, BBS777, BBS778, BBS779, BBS780, BBS781, BBS782, BBS783, BBS784, BBS785, BBS786, BBS787, BBS788, BBS789, BBS790, BBS791, BBS792, BBS793, BBS794, BBS795, BBS796, BBS797, BBS798, BBS799, BBS800, BBS801, BBS802, BBS803, BBS804, BBS805, BBS806, BBS807, BBS808, BBS809, BBS810, BBS811, BBS812, BBS813, BBS814, BBS815, BBS816, BBS817, BBS818, BBS819, BBS820, BBS821, BBS822, BBS823, BBS824, BBS825, BBS826, BBS827, BBS828, BBS829, BBS830, BBS831, BBS832, BBS833, BBS834, BBS835, BBS836, BBS837, BBS838, BBS839, BBS840, BBS841, BBS842, BBS843, BBS844, BBS845, BBS846, BBS847, BBS848, BBS849, BBS850, BBS851, BBS852, BBS853, BBS854, BBS855, BBS856, BBS857, BBS858, BBS859, BBS860, BBS861, BBS862, BBS863, BBS864, BBS865, BBS866, BBS867, BBS868, BBS869, BBS870, BBS871, BBS872, BBS873, BBS874, BBS875, BBS876, BBS877, BBS878, BBS879, BBS880, BBS881, BBS882, BBS883, BBS884, BBS885, BBS886, BBS887, BBS888, BBS889, BBS890, BBS891, BBS892, BBS893, BBS894, BBS895, BBS896, BBS897, BBS898, BBS899, BBS900, BBS901, BBS902, BBS903, BBS904, BBS905, BBS906, BBS907, BBS908, BBS909, BBS910, BBS911, BBS912, BBS913, BBS914, BBS915, BBS916, BBS917, BBS918, BBS919, BBS920, BBS921, BBS922, BBS923, BBS924, BBS925, BBS926, BBS927, BBS928, BBS929, BBS930, BBS931, BBS932, BBS933, BBS934, BBS935, BBS936, BBS937, BBS938, BBS939, BBS940, BBS941, BBS942, BBS943, BBS944, BBS945, BBS946, BBS947, BBS948, BBS949, BBS950, BBS951, BBS952, BBS953, BBS954, BBS955, BBS956, BBS957, BBS958, BBS959, BBS960, BBS961, BBS962, BBS963, BBS964, BBS965, BBS966, BBS967, BBS968, BBS969, BBS970, BBS971, BBS972, BBS973, BBS974, BBS975, BBS976, BBS977, BBS978, BBS979, BBS980, BBS981, BBS982, BBS983, BBS984, BBS985, BBS986, BBS987, BBS988, BBS989, BBS990, BBS991, BBS992, BBS993, BBS994, BBS995, BBS996, BBS997, BBS998, BBS999, BBS1000, BBS1001, BBS1002, BBS1003, BBS1004, BBS1005, BBS1006, BBS1007, BBS1008, BBS1009, BBS1010, BBS1011, BBS1012, BBS1013, BBS1014, BBS1015, BBS1016, BBS1017, BBS1018, BBS1019, BBS1020, BBS1021, BBS1022, BBS1023, BBS1024, BBS1025, BBS1026, BBS1027, BBS1028, BBS1029, BBS1030, BBS1031, BBS1032, BBS1033, BBS1034, BBS1035, BBS1036, BBS1037, BBS1038, BBS1039, BBS1040, BBS1041, BBS1042, BBS1043, BBS1044, BBS1045, BBS1046, BBS1047, BBS1048, BBS1049, BBS1050, BBS1051, BBS1052, BBS1053, BBS1054, BBS1055, BBS1056, BBS1057, BBS1058, BBS1059, BBS1060, BBS1061, BBS1062, BBS1063, BBS1064, BBS1065, BBS1066, BBS1067, BBS1068, BBS1069, BBS1070, BBS1071, BBS1072, BBS1073, BBS1074, BBS1075, BBS1076, BBS1077, BBS1078, BBS1079, BBS1080, BBS1081, BBS1082, BBS1083, BBS1084, BBS1085, BBS1086, BBS1087, BBS1088, BBS1089, BBS1090, BBS1091, BBS1092, BBS1093, BBS1094, BBS1095, BBS1096, BBS1097, BBS1098, BBS1099, BBS1100, BBS1101, BBS1102, BBS1103, BBS1104, BBS1105, BBS1106, BBS1107, BBS1108, BBS1109, BBS1110, BBS1111, BBS1112, BBS1113, BBS1114, BBS1115, BBS1116, BBS1117, BBS1118, BBS1119, BBS1120, BBS1121, BBS1122, BBS1123, BBS1124, BBS1125, BBS1126, BBS1127, BBS1128, BBS1129, BBS1130, BBS1131, BBS1132, BBS1133, BBS1134, BBS1135, BBS1136, BBS1137, BBS1138, BBS1139, BBS1140, BBS1141, BBS1142, BBS1143, BBS1144, BBS1145, BBS1146, BBS1147, BBS1148, BBS1149, BBS1150, BBS1151, BBS1152, BBS1153, BBS1154, BBS1155, BBS1156, BBS1157, BBS1158, BBS1159, BBS1160, BBS1161, BBS1162, BBS1163, BBS1164, BBS1165, BBS1166, BBS1167, BBS1168, BBS1169, BBS1170, BBS1171, BBS1172, BBS1173, BBS1174, BBS1175, BBS1176, BBS1177, BBS1178, BBS1179, BBS1180, BBS1181, BBS1182, BBS1183, BBS1184, BBS1185, BBS1186, BBS1187, BBS1188, BBS1189, BBS1190, BBS1191, BBS1192, BBS1193, BBS1194, BBS1195, BBS1196, BBS1197, BBS1198, BBS1199, BBS1200, BBS1201, BBS1202, BBS1203, BBS1204, BBS1205, BBS1206, BBS1207, BBS1208, BBS1209, BBS1210, BBS1211, BBS1212, BBS1213, BBS1214, BBS1215, BBS1216, BBS1217, BBS1218, BBS1219, BBS1220, BBS1221, BBS1222, BBS1223, BBS1224, BBS1225, BBS1226, BBS1227, BBS1228, BBS1229, BBS1230, BBS1231, BBS1232, BBS1233, BBS1234, BBS1235, BBS1236, BBS1237, BBS1238, BBS1239, BBS1240, BBS1241, BBS1242, BBS1243, BBS1244, BBS1245, BBS1246, BBS1247, BBS1248, BBS1249, BBS1250, BBS1251, BBS1252, BBS1253, BBS1254, BBS1255, BBS1256, BBS1257, BBS1258, BBS1259, BBS1260, BBS1261, BBS1262, BBS1263, BBS1264, BBS1265, BBS1266, BBS1267, BBS1268, BBS1269, BBS1270, BBS1271, BBS1272, BBS1273, BBS1274, BBS1275, BBS1276, BBS1277, BBS1278, BBS1279, BBS1280, BBS1281, BBS1282, BBS1283, BBS1284, BBS1285, BBS1286, BBS1287, BBS1288, BBS1289, BBS1290, BBS1291, BBS1292, BBS1293, BBS1294, BBS1295, BBS1296, BBS1297, BBS1298, BBS1299, BBS1300, BBS1301, BBS1302, BBS1303, BBS1304, BBS1305, BBS1306, BBS1307, BBS1308, BBS1309, BBS1310, BBS1311, BBS1312, BBS1313, BBS1314, BBS1315, BBS1316, BBS1317, BBS1318, BBS1319, BBS1320, BBS1321, BBS1322, BBS1323, BBS1324, BBS1325, BBS1326, BBS1327, BBS1328, BBS1329, BBS1330, BBS1331, BBS1332, BBS1333, BBS1334, BBS1335, BBS1336, BBS1337, BBS1338, BBS1339, BBS1340, BBS1341, BBS1342, BBS1343, BBS1344, BBS1345, BBS1346, BBS1347, BBS1348, BBS1349, BBS1350, BBS1351, BBS1352, BBS1353, BBS1354, BBS1355, BBS1356, BBS1357, BBS1358, BBS1359, BBS1360, BBS1361, BBS1362, BBS1363, BBS1364, BBS1365, BBS1366, BBS1367, BBS1368, BBS1369, BBS1370, BBS1371, BBS1372, BBS1373, BBS1374, BBS1375, BBS1376, BBS1377, BBS1378, BBS1379, BBS1380, BBS1381, BBS1382, BBS1383, BBS1384, BBS1385, BBS1386, BBS1387, BBS1388, BBS1389, BBS1390, BBS1391, BBS1392, BBS1393, BBS1394, BBS1395, BBS1396, BBS1397, BBS1398, BBS1399, BBS1400, BBS1401, BBS1402, BBS1403, BBS1404, BBS1405, BBS1406, BBS1407, BBS1408, BBS1409, BBS1410, BBS1411, BBS1412, BBS1413, BBS1414, BBS1415, BBS1416, BBS1417, BBS1418, BBS1419, BBS1420, BBS1421, BBS1422, BBS1423, BBS1424, BBS1425, BBS1426, BBS1427, BBS1428, BBS1429, BBS1430, BBS1431, BBS1432, BBS1433, BBS1434, BBS1435, BBS1436, BBS1437, BBS1438, BBS1439, BBS1440, BBS1441, BBS1442, BBS1443, BBS1444, BBS1445, BBS1446, BBS1447, BBS1448, BBS1449, BBS1450, BBS1451, BBS1452, BBS1453, BBS1454, BBS1455, BBS1456, BBS1457, BBS1458, BBS1459, BBS1460, BBS1461, BBS1462, BBS1463, BBS1464, BBS1465, BBS1466, BBS1467, BBS1468, BBS1469, BBS1470, BBS1471, BBS1472, BBS1473, BBS1474, BBS1475, BBS1476, BBS1477, BBS1478, BBS1479, BBS1480, BBS1481, BBS1482, BBS1483, BBS1484, BBS1485, BBS1486, BBS1487, BBS1488, BBS1489, BBS1490, BBS1491, BBS1492, BBS1493, BBS1494, BBS1495, BBS1496, BBS1497, BBS1498, BBS1499, BBS1500, BBS1501, BBS1502, BBS1503, BBS1504, BBS1505, BBS1506, BBS1507, BBS1508, BBS1509, BBS1510, BBS1511, BBS1512, BBS1513, BBS1514, BBS1515, BBS1516, BBS1517, BBS1518, BBS1519, BBS1520, BBS1521, BBS1522, BBS1523, BBS1524, BBS1525, BBS1526, BBS1527, BBS1528, BBS1529, BBS1530, BBS1531, BBS1532, BBS1533, BBS1534, BBS1535, BBS1536, BBS1537, BBS1538, BBS1539, BBS1540, BBS1541, BBS1542, BBS1543, BBS1544, BBS1545, BBS1546, BBS1547, BBS1548, BBS1549, BBS1550, BBS1551, BBS1552, BBS1553, BBS1554, BBS1555, BBS1556, BBS1557, BBS1558, BBS1559, BBS1560, BBS1561, BBS1562, BBS1563, BBS1564, BBS1565, BBS1566, BBS1567, BBS1568, BBS1569, BBS1570, BBS1571, BBS1572, BBS1573, BBS1574, BBS1575, BBS1576, BBS1577, BBS1578, BBS1579, BBS1580, BBS1581, BBS1582, BBS1583, BBS1584, BBS1585, BBS1586, BBS1587, BBS1588, BBS1589, BBS1590, BBS1591, BBS1592, BBS1593, BBS1594, BBS1595, BBS1596, BBS1597, BBS1598, BBS1599, BBS1600, BBS1601, BBS1602, BBS1603, BBS1604, BBS1605, BBS1606, BBS1607, BBS1608, BBS1609, BBS1610, BBS1611, BBS1612, BBS1613, BBS1614, BBS1615, BBS1616, BBS1617, BBS1618, BBS1619, BBS1620, BBS1621, BBS1622, BBS1623, BBS1624, BBS1625, BBS1626, BBS1627, BBS1628, BBS1629, BBS1630, BBS1631, BBS1632, BBS1633, BBS1634, BBS1635, BBS1636, BBS1637, BBS1638, BBS1639, BBS1640, BBS1641, BBS1642, BBS1643, BBS1644, BBS1645, BBS1646, BBS1647, BBS1648, BBS1649, BBS1650, BBS1651, BBS1652, BBS1653, BBS1654, BBS1655, BBS1656, BBS1657, BBS1658, BBS1659, BBS1660, BBS1661, BBS1662, BBS1663, BBS1664, BBS1665, BBS1666, BBS1667, BBS1668, BBS1669, BBS1670, BBS1671, BBS1672, BBS1673, BBS1674, BBS1675, BBS1676, BBS1677, BBS1678, BBS1679, BBS1680, BBS1681, BBS1682, BBS16						

Appellation 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 HUGO DU GÈNE et NOM OMIM DU GÈNE / HUGO Gene Symbol and OMIM Gene Number	RÈGLES DE MANUTENTION / Handling Conditions	LABORATOIRE DÉSIGNÉ / Designated laboratory	RENSEIGNEMENTS SUR L'ANALYSE / Test details	MÉTHODE / Methode	NOTE PARTICULIÈRE / Special comment	Date de la révision / Date of revision
1,5-anhydroglucitol (GlycoMark) [500115] [GLYC]	1465		Gold Top or Red Top (Serum) or Lavender (plasma). Centrifuge and aliquot serum to transport tube within one hour of collection. Store and ship frozen. Stable at -20°C for 14 days.	LabCorp/Dynacare (https://www.labcorp.com/)		Enzymatic, colorimetric assay.		10-Jan-21
Complement System Disorder Panel Plus Anlysis [IM0701]	803	<p>ADDPOR2 Complement system AD/AR 4</p> <p>ADDPOR1 Complement system AD/AR 4</p> <p>ADDPOR2 Complement system AD/AR</p> <p>ARMC4* Claryr dyslexia AR 13 15</p> <p>C1QA C1q deficiency AR 21</p> <p>C1QB C1q deficiency AR 4 7</p> <p>C1QB* Primary immunodeficiency AD/AR 6</p> <p>C1QC C1q deficiency AR 4 7</p> <p>C1R Immunodeficiency AD/AR 14 16</p> <p>C1S Complement component C1s deficiency AR 4 8</p> <p>C2* Complement component 2 deficiency AR 4 6</p> <p>C3 Hemolytic uremic syndrome, atypical. Complement component 3 deficiency AD/AR 5 2</p> <p>CSAR1 Complement system AD/AR 1 3</p> <p>CAA* Blood group, Chido/Rodgers system BG 1 5</p> <p>C4B* Complement component 4B deficiency AR 8</p> <p>C4BPA Complement system AD/AR 2</p> <p>C4BPB Complement system AD/AR</p> <p>C5 Esculnab, poor response to. Complement component 5 deficiency AD/AR 5 17</p> <p>CSAR1 Complement system AD/AR</p> <p>CSAR2 Complement system AD/AR 2</p> <p>C6 Complement component 6 deficiency AR 7 11</p> <p>C7 Complement component 7 deficiency AR 14 29</p> <p>C8A Complement component 8 deficiency AR 2 5</p> <p>C8B Complement component 8 deficiency AR 7 7</p> <p>C9C Immunodeficiency AD/AR</p> <p>C9 Complement component 9 deficiency AR 7 7</p> <p>CCDC39 Claryr dyslexia AR 16 38</p> <p>CCDC39* Claryr dyslexia AR 16 32</p> <p>CCDC65 Claryr dyslexia AR 1</p> <p>CCDC103 Claryr dyslexia AR 3 4</p> <p>CCDC114 Claryr dyslexia AR 6 7</p> <p>CCNO Claryr dyslexia AR 9 9</p> <p>CD40* Hemolytic uremic syndrome, atypical AD/AR 4 64</p> <p>CD59 Blood group, Cosner system BG 7 6</p> <p>CD59* CD59 deficiency AR 3 6</p> <p>CD59 Complement system AD/AR</p> <p>CFB Complement factor B deficiency. Hemolytic uremic syndrome, atypical AD/AR 2 21</p> <p>CFD Complement factor D deficiency AR 2 3</p> <p>CFH* Hemolytic uremic syndrome, atypical. Complement factor H deficiency AD/AR 17 259</p> <p>CFI Hemolytic uremic syndrome, atypical. Complement factor I deficiency AD/AR 7 123</p> <p>CFP Prosephin deficiency XL 5 17</p> <p>CLL Complement system AD/AR 16</p> <p>CRLC11 BMC syndrome AR 6 10</p> <p>CR1* Blood group, Knop system BG 1 15</p> <p>CR2 Common variable immunodeficiency AR 2 5</p> <p>CRP Complement system AD/AR</p> <p>DGKE Nephrotic syndrome AR 13 27</p>	Lavender Top (EDTA)	Blueprint Genetics http://blueprintgenetics.com/				02-Feb-18
Comprehensive Cardiomyopathy NGS Panel	289		Lavender Top (EDTA)	Fulgent Genetics (fulgentdiagnostics.com)				03-Nov-20
Comprehensive Cardiovascular NGS Panel	286		Lavender Top (EDTA)	Fulgent Genetics (fulgentdiagnostics.com)				03-Nov-20
Comprehensive Cellular Energetics Defects (NextGen Sequencing Panel and Copy Number Analysis; 320 Genes + mtDNA) [NGS301]	636	<p>AAAS2 (61205), ABLB1 (400185), ABLB2 (605464), ABCD1 (180371), ABCP2 (612510), ABHD15 (604780), ACA1A1 (604054), ACACA (200590), ACACB (601557), ACADM (600773), ACAD9 (611610), ACADL (609570), ACADM (607008), ACADS (606885), ACADSB (60301), ACADVL (609575), ACAT1 (607809), ACAT2 (100678), ACTL1 (109728), ACTD1 (10880), ACC2 (100850), ACCN1 (609751), ACCX2 (601641), ACPE (611671), ACSB1 (614062), ACSF3 (614245), ACSL1 (152425), ACSL3 (602371), ACSL4 (600157), ACSM1 (614357), ACSMB (614399), ACSM1 (145595), ADCK3 (606980), ADHBE1 (611030), AFDL2 (604810), AGR (601050), AGL (601060), AFM1 (601069), AK1 (100320), AK3 (609290), ALAD (125270), ALAS2 (301300), ALDH3A2 (609521), ALDOA (103850), ALDOB (612724), ALDOC (103870), ANO10 (613728), APOPT1 (610405), APTX (600598), ARMS2 (611371), ARK (300382), ATAD1 (612171), ATP5A1 (164340), ATP5B (102910), ATP5E (606153), ATP7B (606882), ATPAF2 (608918), ATRX (606259), BAAT (602938), BAX (600040), BBOX1 (60312), BCKRBA (608548), BCKRBD (610611), BCL11 (601647), BOLA1 (613165), BPGM (613896), BTBD (609019), C10ORF2 (606075), C12orf65 (61541), C21orf53 (607962), CACNA1 (114761), CACNA1S (114238), CALM1 (114180), CALM2 (114182), CALM3 (114183), CALS2 (612360), CACT1 (601540), CHECD10 (615963), CHKB (612395), C10orf159, CKMT1B (123260), CKMT2 (123295), CLPP (601119), COA5 (613920), COQ2 (609825), COQ4 (612898), COQ6 (614647), COQ7 (601603), COQ9 (612675), COX3 (602125), C12orf62 (614478), COX5B (603446), FAM56A (614698), COX41 (123864), COX42 (607976), COX6A1 (603072), COX6B1 (120499), COX7A1 (123995), COX7B (60792), COX8A (123870), CPKX (612712), CPT1A (600230), CPT1B (601987), CPTIC (608840), CPT2 (600650), CS (118990), CYC1 (123980), CYCS (123970), CYP4A11 (601310), DARS (60384), DARS2 (609956), DBT (248610), DCAF1 (601405), DHTKD1 (610964), DLAT (600770), DLEC1 (23810), DLEU1 (123665), DNAI2 (601010), DNAAF1 (608977), DNAAF2 (608950), EARS2 (612599), ECHS1 (602262), ECH1 (601005), ECFM2 (60888), EHDAD (60707), ELAVL1 (60512), ELOVL5 (611865), ENO1 (172438), ENO2 (121366), ENO3 (131170), EPHA (60885), EYFB (130410), EYFDH (231075), ETHE1 (608451), FARP1 (13460), FARP2 (134640), FARP3 (13461), FARP7 (602965), FARS2 (606149), FARS2 (611929), FASN (108212), FSTL3 (612322), FBP1 (611576), FBXL4 (605644), FIPB (134629), FECH (612386), FH (13650), FOLR1 (136430), FOXRED1 (61562), FNN (608829), GPC3 (613142), GPC3 (611045), GPD (30990), GAA (600800), GAMT (601240), GAPDH (138480), GARS (60237), GATM (60230), GBE1 (60789), GCDH (60880), GCK (138079), GER (60924), GPM1 (60639), GPM2 (606544), GPM3 (602395), GPD1 (138420), GPR2 (138430), GPHN (601930), GPI (172400), GPT2 (138210), GSK3B (609084), GSK3L1 (601062), GTPBP1 (609536), GYCI (603942), GYS1 (138570), GYS2 (138571), HADH (601609), HADHA (600890), HADHB (143450), HARS2 (60978), HCCS (30056), HK1 (14260), HK2 (601125), HK3 (142570), HLC5 (60910), HMB1 (60966), HMGCL1 (61308), HMGCS1 (142940), HMGCS2 (60024), HOGA1 (61197), HSPA9 (600548), HSPD1 (118790), HSPH1 (60041), HTT (61304), IARS2 (61280), IBA57 (61516), IBA1 (14700), IBA2 (14700), IBA3 (60820), IBA4 (30009), IBA5 (600378), ISCU (611911), KARS (601421), KIK1 (61450), KIF1B (609995), L1H3DH (60994), LAMP2 (10900), LARS (60544), LDHA (15000), LDHB (15000), LDHC (15010), LIL1 (60701), LIPF (15770), LIFT1 (60264), LRPPRC (60754), LRRK2 (60907), LYRM4 (61511), LYRM7 (61581), MARKS (15060), MARKS2 (60978), MCEE (608419), MDH1 (15420), MDH2 (15410), ME1 (15429), ME2 (15427), ME3 (60807), C12orf62 (614478), COX5B (603446), FAM56A (614698), COX41 (123864), COX42 (607976), COX6A1 (603072), COX6B1 (120499), COX7A1 (123995), COX7B (60792), COX8A (123870), CPKX (612712), CPT1A (600230), CPT1B (601987), CPTIC (608840), CPT2 (600650), CS (118990), CYC1 (123980), CYCS (123970), CYP4A11 (601310), DARS (60384), DARS2 (609956), DBT (248610), DCAF1 (601405), DHTKD1 (610964), DLAT (600770), DLEC1 (23810), DLEU1 (123665), DNAI2 (601010), DNAAF1 (608977), DNAAF2 (608950), EARS2 (612599), ECHS1 (602262), ECH1 (601005), ECFM2 (60888), EHDAD (60707), ELAVL1 (60512), ELOVL5 (611865), ENO1 (172438), ENO2 (121366), ENO3 (131170), EPHA (60885), EYFB (130410), EYFDH (231075), ETHE1 (608451), FARP1 (13460), FARP2 (134640), FARP3 (13461), FARP7 (602965), FARS2 (606149), FARS2 (611929), FASN (108212), FSTL3 (612322), FBP1 (611576), FBXL4 (605644), FIPB (134629), FECH (612386), FH (13650), FOLR1 (136430), FOXRED1 (61562), FNN (608829), GPC3 (613142), GPC3 (611045), GPD (30990), GAA (600800), GAMT (601240), GAPDH (138480), GARS (60237), GATM (60230), GBE1 (60789), GCDH (60880), GCK (138079), GER (60924), GPM1 (60639), GPM2 (606544), GPM3 (602395), GPD1 (138420), GPR2 (138430), GPHN (601930), GPI (172400), GPT2 (138210), GSK3B (609084), GSK3L1 (601062), GTPBP1 (609536), GYCI (603942), GYS1 (138570), GYS2 (138571), HADH (601609), HADHA (600890), HADHB (143450), HARS2 (60978), HCCS (30056), HK1 (14260), HK2 (601125), HK3 (142570), HLC5 (60910), HMB1 (60966), HMGCL1 (61308), HMGCS1 (142940), HMGCS2 (60024), HOGA1 (61197), HSPA9 (600548), HSPD1 (118790), HSPH1 (60041), HTT (61304), IARS2 (61280), IBA57 (61516), IBA1 (14700), IBA2 (14700), IBA3 (60820), IBA4 (30009), IBA5 (600378), ISCU (611911), KARS (601421), KIK1 (61450), KIF1B (609995), L1H3DH (60994), LAMP2 (10900), LARS (60544), LDHA (15000), LDHB (15000), LDHC (15010), LIL1 (60701), LIPF (15770), LIFT1 (60264), LRPPRC (60754), LRRK2 (60907), LYRM4 (61511), LYRM7 (61581), MARKS (15060), MARKS2 (60978), MCEE (608419), MDH1 (15420), MDH2 (15410), ME1 (15429), ME2 (15427), ME3 (60807), C12orf62 (614478), COX5B (603446), FAM56A (614698), COX41 (123864), COX42 (607976), COX6A1 (603072), COX6B1 (120499), COX7A1 (123995), COX7B (60792), COX8A (123870), CPKX (612712), CPT1A (600230), CPT1B (601987), CPTIC (608840), CPT2 (600650), CS (118990), CYC1 (123980), CYCS (123970), CYP4A11 (601310), DARS (60384), DARS2 (609956), DBT (248610), DCAF1 (601405), DHTKD1 (610964), DLAT (600770), DLEC1 (23810), DLEU1 (123665), DNAI2 (601010), DNAAF1 (608977), DNAAF2 (608950), EARS2 (612599), ECHS1 (602262), ECH1 (601005), ECFM2 (60888), EHDAD (60707), ELAVL1 (60512), ELOVL5 (611865), ENO1 (172438), ENO2 (121366), ENO3 (131170), EPHA (60885), EYFB (130410), EYFDH (231075), ETHE1 (608451), FARP1 (13460), FARP2 (134640), FARP3 (13461), FARP7 (602965), FARS2 (606149), FARS2 (611929), FASN (108212), FSTL3 (612322), FBP1 (611576), FBXL4 (605644), FIPB (134629), FECH (612386), FH (13650), FOLR1 (136430), FOXRED1 (61562), FNN (608829), GPC3 (613142), GPC3 (611045), GPD (30990), GAA (600800), GAMT (601240), GAPDH (138480), GARS (60237), GATM (60230), GBE1 (60789), GCDH (60880), GCK (138079), GER (60924), GPM1 (60639), GPM2 (606544), GPM3 (602395), GPD1 (138420), GPR2 (138430), GPHN (601930), GPI (172400), GPT2 (138210), GSK3B (609084), GSK3L1 (601062), GTPBP1 (609536), GYCI (603942), GYS1 (138570), GYS2 (138571), HADH (601609), HADHA (600890), HADHB (143450), HARS2 (60978), HCCS (30056), HK1 (14260), HK2 (601125), HK3 (142570), HLC5 (60910), HMB1 (60966), HMGCL1 (61308), HMGCS1 (142940), HMGCS2 (60024), HOGA1 (61197), HSPA9 (600548), HSPD1 (118790), HSPH1 (60041), HTT (61304), IARS2 (61280), IBA57 (61516), IBA1 (14700), IBA2 (14700), IBA3 (60820), IBA4 (30009), IBA5 (600378), ISCU (611911), KARS (601421), KIK1 (61450), KIF1B (609995), L1H3DH (60994), LAMP2 (10900), LARS (60544), LDHA (15000), LDHB (15000), LDHC (15010), LIL1 (60701), LIPF (15770), LIFT1 (60264), LRPPRC (60754), LRRK2 (60907), LYRM4 (61511), LYRM7 (61581), MARKS (15060), MARKS2 (60978), MCEE (608419), MDH1 (15420), MDH2 (15410), ME1 (15429), ME2 (15427), ME3 (60807), C12orf62 (614478), COX5B (603446), FAM56A (614698), COX41 (123864), COX42 (607976), COX6A1 (603072), COX6B1 (120499), COX7A1 (123995), COX7B (60792), COX8A (123870), CPKX (612712), CPT1A (600230), CPT1B (601987), CPTIC (608840), CPT2 (600650), CS (118990), CYC1 (123980), CYCS (123970), CYP4A11 (601310), DARS (60384), DARS2 (609956), DBT (248610), DCAF1 (601405), DHTKD1 (610964), DLAT (600770), DLEC1 (23810), DLEU1 (123665), DNAI2 (601010), DNAAF1 (608977), DNAAF2 (608950), EARS2 (612599), ECHS1 (602262), ECH1 (601005), ECFM2 (60888), EHDAD (60707), ELAVL1 (60512), ELOVL5 (611865), ENO1 (172438), ENO2 (121366), ENO3 (131170), EPHA (60885), EYFB (130410), EYFDH (231075), ETHE1 (608451), FARP1 (13460), FARP2 (134640), FARP3 (13461), FARP7 (602965), FARS2 (606149), FARS2 (611929), FASN (108212), FSTL3 (612322), FBP1 (611576), FBXL4 (605644), FIPB (134629), FECH (612386), FH (13650), FOLR1 (136430), FOXRED1 (61562), FNN (608829), GPC3 (613142), GPC3 (611045), GPD (30990), GAA (600800), GAMT (601240), GAPDH (138480), GARS (60237), GATM (60230), GBE1 (60789), GCDH (60880), GCK (138079), GER (60924), GPM1 (60639), GPM2 (606544), GPM3 (602395), GPD1 (138420), GPR2 (138430), GPHN (601930), GPI (172400), GPT2 (138210), GSK3B (609084), GSK3L1 (601062), GTPBP1 (609536), GYCI (603942), GYS1 (138570), GYS2 (138571), HADH (601609), HADHA (600890), HADHB (143450), HARS2 (60978), HCCS (30056), HK1 (14260), HK2 (601125), HK3 (142570), HLC5 (60910), HMB1 (60966), HMGCL1 (61308), HMGCS1 (142940), HMGCS2 (60024), HOGA1 (61197), HSPA9 (600548), HSPD1 (118790), HSPH1 (60041), HTT (61304), IARS2 (61280), IBA57 (61516), IBA1 (14700), IBA2 (14700), IBA3 (60820), IBA4 (30009), IBA5 (600378), ISCU (611911), KARS (601421), KIK1 (61450), KIF1B (609995), L1H3DH (60994), LAMP2 (10900), LARS (60544), LDHA (15000), LDHB (15000), LDHC (15010), LIL1 (60701), LIPF (15770), LIFT1 (60264), LRPPRC (60754), LRRK2 (60907), LYRM4 (61511), LYRM7 (61581), MARKS (15060), MARKS2 (60978), MCEE (608419), MDH1 (15420), MDH2 (15410), ME1 (15429), ME2 (15427), ME3 (60807), C12orf62 (614478), COX5B (603446), FAM56A (614698), COX41 (123864), COX42 (607976), COX6A1 (603072), COX6B1 (120499), COX7A1 (123995), COX7B (60792), COX8A (123870), CPKX (612712), CPT1A (600230), CPT1B (601987), CPTIC (608840), CPT2 (600650), CS (118990), CYC1 (123980), CYCS (123970), CYP4A11 (601310), DARS (60384), DARS2 (609956), DBT (248610), DCAF1 (601405), DHTKD1 (610964), DLAT (600770), DLEC1 (23810), DLEU1 (123665), DNAI2 (601010), DNAAF1 (608977), DNAAF2 (608950), EARS2 (612599), ECHS1 (602262), ECH1 (601005), ECFM2 (60888), EHDAD (60707), ELAVL1 (60512), ELOVL5 (611865), ENO1 (172438), ENO2 (121366), ENO3 (131170), EPHA (60885), EYFB (130410), EYFDH (231075), ETHE1 (608451), FARP1 (13460), FARP2 (134640), FARP3 (13461), FARP7 (602965), FARS2 (606149), FARS2 (611929), FASN (108212), FSTL3 (612322), FBP1 (611576), FBXL4 (605644), FIPB (134629), FECH (612386), FH (13650), FOLR1 (136430), FOXRED1 (61562), FNN (608829), GPC3 (613142), GPC3 (611045), GPD (30990), GAA (600800), GAMT (601240), GAPDH (138480), GARS (60237), GATM (60230), GBE1 (60789), GCDH (60880), GCK (138079), GER (60924), GPM1 (60639), GPM2 (606544), GPM3 (602395), GPD1 (138420), GPR2 (138430), GPHN (601930), GPI (172400), GPT2 (138210), GSK3B (609084), GSK3L1 (601062), GTPBP1 (609536), GYCI (603942), GYS1 (138570), GYS2 (138571), HADH (601609), HADHA (600890), HADHB (143450), HARS2 (60978), HCCS (30056), HK1 (14260), HK2 (601125), HK3 (142570), HLC5 (60910), HMB1 (60966), HMGCL1 (61308), HMGCS1 (142940), HMGCS2 (60024), HOGA1 (61197), HSPA9 (600548), HSPD1 (118790), HSPH1 (60041), HTT (61304), IARS2 (61280), IBA57 (61516), IBA1 (14700), IBA2 (14700), IBA3 (60820), IBA4 (30009), IBA5 (600378), ISCU (611911), KARS (601421), KIK1 (61450), KIF1B (609995), L1H3DH (60994), LAMP2 (10900), LARS (60544), LDHA (15000), LDHB (15000), LDHC (15010), LIL1 (60701), LIPF (15770), LIFT1 (60264), LRPPRC (60754), LRRK2 (60907), LYRM4 (61511), LYRM7 (61581), MARKS (15060), MARKS2 (60978), MCEE (608419), MDH1 (15420), MDH2 (15410), ME1 (15429), ME2 (15427), ME3 (60807), C12orf62 (614478), COX5B (603446), FAM56A (614698), COX41 (123864), COX42 (607976), COX6A1 (603072), COX6B1 (120499), COX7A1 (123995), COX7B (60792), COX8A (123870), CPKX (612712), CPT1A (600230), CPT1B (601987), CPTIC (608840), CPT2 (600650), CS (118990), CYC1 (123980), CYCS (123970), CYP4A11 (601310), DARS (60384), DARS2 (609956), DBT (248610), DCAF1 (601405), DHTKD1 (610964), DLAT (600770), DLEC1 (23810), DLEU1 (123665), DNAI2 (601010), DNAAF1 (608977), DNAAF2 (608950), EARS2 (612599), ECHS1 (602262), ECH1 (601005), ECFM2 (60888), EHDAD (60707), ELAVL1 (60512), ELOVL5 (611865), ENO1 (172438), ENO2 (121366), ENO3 (131170), EPHA (60885), EYFB (130410), EYFDH (231075), ETHE1 (608451), FARP1 (13460), FARP2 (134640), FARP3 (13461), FARP7 (602965), FARS2 (606149), FARS2 (611929), FASN (108212), FSTL3 (612322), FBP1 (611576), FBXL4 (605644), FIPB (134629), FECH (612386), FH (13650), FOLR1 (136430), FOXRED1 (61562), FNN (608829), GPC3 (613142), GPC3 (611045), GPD (30990), GAA (600800), GAMT (601240), GAPDH (138480), GARS (60237), GATM (60230), GBE1 (60789), GCDH (60880), GCK (138079), GER (60924), GPM1 (60639), GPM2 (606544), GPM3 (602395), GPD1 (138420), GPR2 (138430), GPHN (601930), GPI (172400), GPT2 (138210), GSK3B (609084), GSK3L1 (601062), GTPBP1 (609536), GYCI (603942), GYS1 (138570), GYS2 (138571), HADH (601609), HADHA (600890), HADHB (143450), HARS2 (60978), HCCS (30056), HK1 (14260), HK2 (601125), HK3 (142570), HLC5 (60910), HMB1 (60966), HMGCL1 (61308), HMGCS1 (142940), HMGCS2 (60024), HOGA1 (61197), HSPA9 (600548), HSPD1 (118790), HSPH1 (60041), HTT (61304), IARS2 (61280), IBA57 (61516), IBA1 (14700), IBA2 (14700), IBA3 (60820), IBA4 (30009), IBA5 (600378), ISCU (611911), KARS (601421), KIK1 (61450), KIF1B (609995), L1H3DH (60994), LAMP2 (10900), LARS (60544), LDHA (15000), LDHB (15000), LDHC (15010), LIL1 (60701), LIPF (15770), LIFT1 (60264), LRPPRC (60754), LRRK2 (60907), LYRM4 (61511), LYRM7 (61581), MARKS (15060), MARKS2 (60978), MCEE (608419), MDH1 (15420), MDH2 (15410), ME1 (15429), ME2 (15427), ME3 (60807), C12orf62 (614478), COX5B (603446), FAM56A (614698), COX41 (123864), COX42 (607976), COX6A1 (603072), COX6B1 (120499), COX7A1 (123995), COX7B (60792), COX8A (123870), CPKX (612712), CPT1A (600230), CPT1B (601987), CPTIC (608840), CPT2 (600650), CS (118990), CYC1 (123980), CYCS (123970), CYP4A11 (601310), DARS (60384), DARS2 (609956), DBT (248610), DCAF1 (601405), DHTKD1 (610964), DLAT (600770), DLEC1 (23810), DLEU1 (123665), DNAI2 (601010), DNAAF1 (608977), DNAAF2 (608950), EARS2 (612599), ECHS1 (602262), ECH1 (601005), ECFM2 (60888), EHDAD (60707), ELAVL1 (60512), ELOVL5 (611865), ENO1 (172438), ENO2 (121366), ENO3 (131170), EPHA (60885), EYFB (130410), EYFDH (231075), ETHE1 (608451), FARP1 (13460), FARP2 (134640), FARP3 (13461), FARP7 (602965), FARS2 (606149), FARS2 (611929), FASN (108212), FSTL3 (612322), FBP1 (611576), FBXL4 (605644), FIPB (134629), FECH (612386), FH (13650), FOLR1 (136430), FOXRED1 (61562), FNN (608829), GPC3 (613142), GPC3 (611045), GPD (30990), GAA (600800), GAMT (601240), GAPDH (138480), GARS (60237), GATM (60230), GBE1 (60789), GCDH (60880), GCK (138079),</p>						

Appellation 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 (Déclaratoire MSSS)	SYMBOL HUGO DU GÈNE et NOM OMDM DU GÈNE / HUGO Gene Symbol and OMDM Gene Name	RÈGLES DE MANUTENTION / Handling Conditions	LABORATOIRE DÉSIGNÉ / Designated laboratory	RENSEIGNEMENTS SUR L'ANALYSE / Test details	MÉTHODE / Methode	NOTE PARTICULIÈRE / Special comment	Date de la révision / Date of revision
1,5-anhydroglucitol (GlycoMark) [500115] [GLYC]	1465		Gold Top or Red Top (Serum) or Lavender (plasma). Centrifuge and aliquot serum to transport tube within one hour of collection. Store and ship frozen. Stable at -20°C for 14 days.	LabCorp/Dynacare (https://www.labcorp.com/)		Enzymatic, colorimetric assay.		10-Jan-21
Comprehensive Ophthalmoplegia Syndromes (NextGen Sequencing Panel and Copy Number Analysis; 55 Genes + mtDNA) [NGS352]	1648	ACADS (60885), ACTA1 (102016), AFG3L2 (604581), AGRN (103220), APTX (60658), ATXN1 (601556), ATXN2 (601517), ATXN3 (607047), ATXN7 (607649), BIN1 (601248), C10ORF2 (606075), C12orf65 (615841), CHAT (118490), CHRNA1 (100690), CHRNA3 (100719), CHRNA4 (100723), CHRNA5 (100725), CLF1 (601119), COLQ (603033), DNMG (602738), DOK7 (602628), EARS2 (612799), FOXE3 (601094), FOXRED1 (613622), GBA (606463), GRM1 (614831), HARS2 (608793), HCCS (606556), HSD17B4 (601980), KIF21A (608263), LARS2 (604540), C20ORF72 (605076), MPV17 (157960), MTM1 (604155), MTRR4 (611089), MUSK (601290), MYH2 (160740), OPA1 (602950), OPA3 (606880), PABPN1 (602279), PDLG1 (157670), POLG2 (604960), RAPS8 (601092), RAB29 (604712), RYR1 (188901), SCY2 (604272), SHAF1 (612848), SEPN1 (606210), SLC19A3 (606152), SLC9A6 (602231), SPEG (615950), TPM1 (191010), TUBB3 (602061), TYMP (131222), ZNF592 (606957).	Lavender Top (EDTA)	MNG Laboratories (www.medicalneurogenetics.com/) / Dynacare		NextGen Sequencing		14-May-20
Cone-Rod Dystrophy (CORDX3) via the CACNA1F gene []	1444	CACNA1F (60110)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Aland Island Eye Disease (300600); Cone-Rod Dystrophy X-Linked 3 (300476); Congenital Stationary Night Blindness, Type 2A (300071)	NextGen Sequencing		
Congenital Adrenal Hyperplasia NGS Panel	1326	ARMC5, CYP11B1, CYP11B2, CYP17A1, CYP21A2, HSD11B2, 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, HSD11B2, PDE11A, PDE8B, POR, PRKAR1A, STAR	Lavender Top (EDTA) 2 x 4 mL	Blueprint Genetics http://blueprintgenetics.com/		NextGen Sequencing		28-Feb-20
Congenital Hyperinsulinism Sequencing Panel [1939]	541	ABCC8 (60509), GCK (118079), GLUD1 (118130), HADH (601049), HNF1A (142410), HNF1A (602281), KCNJ11 (600087), SLC16A1 (600082), UCP2 (601693)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Hyperinsulinemic Hypoglycemia, Familial 3 (612485) Hyperinsulinemic Hypoglycemia, Familial 6 (606762) Hyperinsulinemic Hypoglycemia, Familial 1 (256450) Hyperinsulinemic Hypoglycemia, Familial 2 (601820) Hyperinsulinemic Hypoglycemia, Familial 4 (609975) Hyperinsulinemic Hypoglycemia, Familial 7 (610021) Maturity-Onset Diabetes Of The Young, Type 1 (125850) Maturity-Onset Diabetes Of The Young, Type 3 (610406) Beard's Triad	Next-Generation sequencing	16-Aug-17	
CONGENITAL MUSCULAR DYSTROPHY NEXTGEN SEQUENCING (NGS) PANEL [1301]	606	ITGA7 (600516), FKTN (607440), FKBP (606596), LAMA2 (156225), LARGE (601598), POMT1 (607326), POMT2 (607109), POMT3 (100622), DAG1 (128209), DPM1 (601503), DPM3 (605951), CHKB (612295), SPD (644631), LMNA (150300), C10C2 (614826), TMEM8 (608862), B3GALNT2 (610594), GMPY8 (615320), B3GNT1 (605171), GORBP2 (604027), SGK196 (615247), STXAL4 (104240)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Muscular Dystrophy-Dystroglycanopathy (Congenital with Brain and Eye Anomalies), Type A, 10, MDDGA10 (615041); Congenital Disorder Of Glycosylation Type 1E (608799); Congenital Disorder Of Glycosylation Type 1O (612937); Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 14 (615352); Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 12 (615249) Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 11 (615181); Congenital Muscular Dystrophy-Dystroglycanopathy (With Brain And Eye Anomalies) Type A5 (613153); Merosin Deficient Congenital Muscular Dystrophy (607855); Muscular Dystrophy-Dystroglycanopathy (Limb-Girdle), Type C, 9 (613818); Walker-Warburg Congenital Muscular Dystrophy (614830); Muscular Dystrophy-Dystroglycanopathy (Limb-Girdle), Type C, 2 (613158); Muscular Dystrophy-Dystroglycanopathy (Congenital With Brain And Eye Anomalies), Type A, 6 (613154); Walker-Warburg Congenital Muscular Dystrophy 256670 Muscle Eye Brain Disease (253280); Fukuyama Congenital Muscular Dystrophy (253800); Muscular Dystrophy, Congenital, LMNA-Related (613205); Muscular Dystrophy, Congenital, Due To Integrin Alpha-7 Deficiency (613204); Muscular Dystrophy, Congenital, Megaconial Type (602541); Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 7 (614643)	NextGen Sequencing (22 genes)	See Med Neurogenetics Panel	

Appellation 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 (Déclencheur MSSS)	SYMBOL HUGO DU GÈNE et NOM OMIM DU GÈNE / HUGO Gene Symbol and OMIM Gene Number	RÈGLES DE MANUTENTION / Handling Conditions	LABORATOIRE DÉSIGNÉ / Designated laboratory	RENSEIGNEMENTS SUR L'ANALYSE / Test details	MÉTHODE / Methode	NOTE PARTICULIÈRE / Special comment	Date de la révision / Date of revision
I,5-anhydroglucitol (GlycoMark) [500115] [GLYC]	1465		Gold Top or Red Top (Serum) or Lavender (plasma). Centrifuge and aliquot serum to transport tube within one hour of collection. Store and ship frozen. Stable at -20°C for 14 days.	LabCorp/Dynacare (https://www.labcorp.com/)		Enzymatic, colorimetric assay.		10-Jan-21
Congenital Myasthenic Syndrome NGS Panel	606	AGRN, ALGH, ALGE, CHAT, CHRNL, CHRND1, CHRND, CHRNE, CHRNG, COL11A1, COL4, DOK7, DRAGT1, GPT1, GNPBB, LA-MBL2, LEP3A, MURK, MYO9A, PLEC, PREPL, RAPS3, SCN4A, SLC25A1, SLC5A7, SNAF25, STIM1, SYT2 (28 genes)	Lavender Top (2 x 4 mL)	Fulgent Genetics (fulgentdiagnostics.com)	Myasthenic syndrome, congenital, with pre- and postsynaptic defect (615120); Myasthenic syndrome, congenital, associated with episodic apnea (254210); Multiple pterygium syndrome, lethal type (253290); Myasthenic syndrome, fast-channel congenital (608930); Myasthenic syndrome, slow-channel congenital (601462); Myasthenic syndrome, congenital, associated with acetylcholine receptor deficiency (608931); Myasthenic syndrome, slow-channel congenital (601462); Multiple pterygium syndrome, lethal type (253290); Myasthenic syndrome, fast-channel congenital (608930); Myasthenic syndrome, slow-channel congenital (601462); Myasthenic syndrome, congenital, associated with acetylcholine receptor deficiency (608931); Myasthenic syndrome, fast-channel congenital (608930); Myasthenic syndrome, slow-channel congenital (601462); Endplate acetylcholinesterase deficiency (603034); Fetal akinesia deformation sequence (208150); Myasthenia, limb-girdle, familial (254300); Myasthenia, congenital, with tubular aggregates 1 (610542); Myasthenic syndrome, congenital, associated with acetylcholine receptor deficiency (608931); Fetal akinesia deformation sequence (208150); Myasthenic syndrome, congenital, associated with acetylcholine receptor deficiency (608931); Myasthenic syndrome, congenital, associated with facial dysmorphism and acetylcholine receptor deficiency (608931); Hypokalemic periodic paralysis, type 2 (170500); Hypokalemic periodic paralysis, type 2 (613345); Myasthenic syndrome, acetazolamide-responsive (614198); Myotonia congenita, atypical, acetazolamide-responsive (608300); Paramyotonia congenita (1168300)	Next-Generation sequencing		25-Jul-17
CONGENITAL MYOPATHY NEXTGEN SEQUENCING (NGS) PANEL [1365]	631		Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)			Next-Generation sequencing	
Congenital Neutropenia Panel [IM0501]	836	ACTB, CLPB, CSFR3A, CSFR3B, CTSC, EFL1, ELANE, GPC3, GATA2, GR1, GNS1, HAX1, JNCR2, JAGN1, LAMTOR2, LYST, MKL1, PCDH16, RAC2, SRSF5, SLC37A4, SMARCD2, SRP54, SRP72, VPS33B, VPS45, WAS, WDR1	Lavender Top (2 x 4 mL)	Blueprint Genetics http://blueprintgenetics.com/			NextGen Sequencing	28-Feb-20
Copeptin, Serum/Plasma	1465		Prefered Plasma (Li heparin). Serum OK. Store and send frozen (90 days).	In-Common Laboratories	Replaces vasopressin/ADH. Suggest ordering osmolality at same time.	Fluorescent immunoassay		05-Sep-22
Cornelia de Lange Syndrome		NIPBL (608667), SMC1A (300040), SMC3 (600662), HDAC8 (300209), RAD21 (606462)	Lavender Top (2 x 4 mL)	Fulgent Genetics (fulgentdiagnostics.com)	Cornelia de Lange syndrome 1 (122470); Cornelia de Lange syndrome 2 (300599); Cornelia de Lange syndrome 3 (610759); Cornelia de Lange syndrome 4 (614701); Cornelia de Lange syndrome 5 (300882)	Next-Generation sequencing		
Cortical Brain Malformations Panel [698]	346	ARFGAP2 (605771), ARX (300382), DCX (300211), EOMES (604615), FKBP (605996), FKTN (607440), FLNA (300077), GPR56 (604100), LAMC3 (604349), LARGC (605590), NDE1 (609406), OCLN (602670), PAPDA1B1 (601545), POMGN1 (606622), POMT1 (607423), POMT2 (607439), RELN (602514), SRFX2 (300642), TUBA1A (602520), TUBA8 (605742), TUBB2B (612830), TUBB3 (602661), VLDLR (92977)	Lavender Top (EDTA)	GeneDx (www.genedx.com)	Lisencephaly; Lisencephaly and subcortical band heterotopia (SBL); Alpha-dystroglycanopathies; Periventricular nodular heterotopia (PVNH); Polymicrogyria	Exon Array CGH, Next-gen Sequencing		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		RIA	2017/03/09	
Cortisol, free [CORIF]			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 4°C or -20°C.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	Flèvre Q (Coxiella burnetii IgG et IgM) at CHUS Fleurance			31-Oct-20
Craniosynostosis Non-Syndromic (select exons of FGFR3 gene)		FGFR3 (134934)	Lavender Top (EDTA)	Molecular Genetics Laboratory, Hospital for Sick Children (www.sickkids.ca/PaediatricLaboratoryMedicine/Laboratories-Services/Molecular-Genetics-Laboratory/index.html)	Craniosynostosis	Sanger sequencing; FGFR3 (p.Pro250Arg)		

Appellation 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 (Déclaré dans MSSS)	SYMBOL HGNC DU GÈNE et NOM OMIM DU GÈNE / HGNC Gene Symbol and OMIM Gene Number	RÈGLES DE MANUTENTION / Handling Conditions	LABORATOIRE DÉSIGNÉ / Designated laboratory	RENSEIGNEMENTS SUR L'ANALYSE / Test details	MÉTHODE / Methode	NOTE PARTICULIÈRE / Special comment	Date de la révision / Date of revision
1,5-anhydroglucitol (GlycoMark) [500115] [GLYC]	1465		Gold Top or Red Top (Serum) or Lavender (plasma). Centrifuge and aliquot serum to transport tube within one hour of collection. Store and ship frozen. Stable at -20°C for 14 days.	LabCorp/Dynacare (https://www.labcorp.com/)		Enzymatic, colorimetric assay.		10-Jan-21
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 (170943); FGFR3 (134934)	Lavender Top (EDTA)	Molecular Genetics Laboratory, Hospital for Sick Children (www.sickkids.ca/PaediatricLaboratoryMedicine/Laboratories-Services/Molecular-Genetics-Laboratory/index.html)	Craniosynostosis; Crouzon syndrome (123500)	Sanger sequencing: FGFR2 (exon 7 and 8); FGFR3 (p.Pro250Arg)		
Cryoglobulin and Cryofibrinogen Panel, Serum and Plasma [CRGSP]			Cryofibrinogen Collection Container/Tube: Lavender top (EDTA) Submission Container/Tube: Plastic vial Specimen Volume: 1 mL Collection Instructions: 1. Tube must remain at 37 degrees C. 2. Centrifuge at 37 degrees C. (Do not use a refrigerated centrifuge. If absolutely necessary, ambient temperature is acceptable.) It is very important that the specimen remain at 37 degrees C until after separation of plasma from red cells. 3. Place plasma into an appropriately labeled plastic vial. Cryoglobulin Collection Container/Tube: Red top Submission Container/Tube: Plastic vial Specimen Volume: 5 mL Collection Instructions: 1. Tube must remain at 37 degrees C. 2. Allow blood to clot at 37 degrees C. 3. Centrifuge at 37 degrees C. (Do not use a refrigerated centrifuge. If absolutely necessary, ambient temperature is acceptable.) It is very important that the specimen remain at 37 degrees C until after separation of serum from red cells. 4. Place serum into an appropriately labeled plastic vial.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	Evaluating patients with vasculitis, glomerulonephritis, and lymphoproliferative diseases Evaluating patients with macroglobulinemia or myeloma in whom symptoms occur with cold exposure	immunofixation		
Cryopyrin-Associated Periodic Syndromes via the NLRP3 Gene [1638]		NLRP3 (606416)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Chronic Infantile Neurological, Cutaneous And Articular Syndrome (607115); Familial Amyloid Nephropathy With Urticaria And Deafness (191900); Familial Cold Urticaria	Sanger sequencing		
Cryptococcal Antigen, latex agglutination [319]	2020		Serum: Collect serum specimens in serum separator or red top tube. Allow blood to clot for 30 minutes, then centrifuge. Pipette serum into a plastic screw cap vial. CSF: Collect in sterile container. Centrifuge to remove white cells and particulate matter. STABILITY refrigerated/Frozen: 2 months	MiraVista Diagnostics				08-Sep-22
CSF Protein Immunoassay Panel (CID 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 (CID)	14-3-3 protein testing of cerebrospinal fluid (CSF) (ELISA); PpPd (QuC); Tau protein (ELISA)	Should be registered with surveillance.	24-May-17
CSTB dodecmer repeat expansion [7084]		CSTB (601145)	Lavender Top (EDTA)	Amby Genetics	Epilepsy, progressive myoclonic 1A (Luvierriicht and Lundborg) (254800)			
Curarino syndrome		MCN1 (142994)	Lavender top (EDTA)	Diagenos (www.diagenos.com)	Curarino syndrome (176450)	Sanger sequencing		
Curarino syndrome		MCN1 (142994)	Lavender top (EDTA)	Centogene AG (www.centogene.com)	Curarino syndrome (176450)	Sanger sequencing		
Cyclic AMP, Urinary Excretion [GRP]			Serum (Red Top) AND random urine required. Store frozen.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)				19-May-17
CYP27B1 Single Gene Test	897	CYP27B1 (609506)	Lavender Top (2 x 4 mL)	Fulgent Genetics (fulgentdiagnostics.com)	Vitamin D dependant rickets type1 (264700)	NextGen Sequencing		23-Mar-21
Cystatin C [CYSTC]	78		Serum. Store and send frozen	In-Common Laboratories		Immunonephelometry		06-Sep-18

Appellation 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 (Déclaration MSSS)	SYMBOL, HGNC ID, GENE et NOM OMIM DU GÈNE / HGNC Gene Symbol and OMIM Gene Number	RÈGLES DE MANUTENTION / Handling Conditions	LABORATOIRE DÉSIGNÉ / Designated laboratory	RENSEIGNEMENTS SUR L'ANALYSE / Test details	MÉTHODE / Methode	NOTE PARTICULIÈRE / Special comment	Date de la révision / Date of revision
I,5-anhydroglucitol (GlycoMark) [500115] [GLYC]	1465		Gold Top or Red Top (Serum) or Lavender (plasma). Centrifuge and aliquot serum to transport tube within one hour of collection. Store and ship frozen. Stable at -20°C for 14 days.	LabCorp/Dynacare (https://www.labcorp.com/)		Enzymatic, colorimetric assay.		10-Jan-21
Cystic Fibrosis: CFTR Deletion Duplication Analysis	763	CFTR (602421)	Lavender Top (EDTA)	Genome Diagnostics The Department of Paediatric Laboratory Medicine The Hospital for Sick Children (www.sickkids.ca/paediatriclabmedicinems/lab-divisions/genome-diagnostics/genome-diagnostics.htm#genome)	Cystic Fibrosis (219700)	MLPA		
Cystinuria NGS Panel	807	SLC3A1 (108642), SLC7A9 (608144), PREPL (609577)	Lavender Top (2 x 4 mL)	Fulgent Genetics (fulgentdiagnostics.com)	Cystinuria (220100)	NextGen Sequencing		22-Dec-20
Cytochrome P450 2D6 (CYP2D6) Comprehensive Cascade [2D6CV]	77	CYP2D6 (124030)	Lavender Top (EDTA). Stable for 30 days at 4°C.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)				31-Oct-20
Cytokine Panel [CYPAN]	1523		Lavender Top (EDTA). Stable for 30 days at 4°C.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	includes soluble interleukin-2 receptor	bead-based multiplex immunoassay		28-Dec-21
Dabigatran			Serum (Light blue top)	Quest Diagnostics				
DARS2 Full Gene Sequencing Analysis [MOL094]		DARS2 (610956)	Lavender Top (EDTA)	MNG Laboratories (www.medicalneurogenetics.com)/Dynacare	Leukoencephalopathy with brain stem and spinal cord involvement and lactate elevation (611105) Alternate name: DARS2: Mitochondrial Aspartyl-tRNA Synthetase Deficiency	Sanger sequencing		14-May-20
Deletion 1p			Green Top; RT only	Cytogenetics Laboratory, Hospital for Sick Children	Chromosome 1p36 deletion syndrome (607872)	microarray		
Dementia Panel [I0309]	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 (Familial Mutation/Variant Analysis) [3076]		DGUOK (601405)	Lavender Top (EDTA)	Medical Genetics Laboratories Baylor College of Medicine (www.bcmgeneticlabs.org)	Mitochondrial DNA depletion syndrome 3 (hepatocerebral type) (251880)	Sanger sequencing		
Diabetes Insipidus Panel [KI1801]	1457	AQP2 AVP AVPR2	Lavender Top (EDTA) 2 x 4 mL	Blueprint Genetics http://blueprintgenetics.com/		NextGen Sequencing		28-Feb-20
DICER1 single gene test [S00555]	246	DICER1	Lavender Top (EDTA) 2 x 4 mL	Blueprint Genetics http://blueprintgenetics.com/		NextGen Sequencing		28-Feb-20
Dihydrotestosterone, Serum [DHT]	1459		Red Top or Gold Top. Store serum and send frozen	In-Common Laboratories		ELISA		30-Jan-20
Dilated Cardiomyopathy NGS Panel	286		Lavender Top (EDTA) 2 x 4 mL	Fulgent Genetics (fulgentdiagnostics.com)				31-Oct-21
Disaccharidase Determination, Small Bowel Biopsy	85		Intestinal biopsy, 2-5 mg wet weight. Store at -70°C until shipping and send on dry ice	Gastroenterology Clinical Lab, Nemours/Alfred I. Dupont Hospital for Children, Wilmington, DE (www.nemours.org) (https://www.nemours.org/pediatric-research/labservices/diagnostic/gastroenterology-lab.html)		lactase, maltase, sucrase, palatinase and glucoamylase		06-Apr-22

Appellation 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 (Déclaratoire MSSS)	SYMBOL/IGENE DU GÈNE et NOM OMDM DU GÈNE / IGENE Gene Symbol/and OMDM Gene Name	RÈGLES DE MANUTENTION / Handling Conditions	LABORATOIRE DÉSIGNÉ / Designated laboratory	RENSEIGNEMENTS SUR L'ANALYSE / Test details	MÉTHODE / Methode	NOTE PARTICULIÈRE / Special comment	Date de la révision / Date of revision
1,5-anhydroglucitol (GlycoMark) [500115] [GLYC]	1465		Gold Top or Red Top (Serum) or Lavender (plasma). Centrifuge and aliquot serum to transport tube within one hour of collection. Store and ship frozen. Stable at -20°C for 14 days.	LabCorp/Dynacare (https://www.labcorp.com/)		Enzymatic, colorimetric assay.		10-Jan-21
Disorders of Sex Development Sequencing Panel with CNV Detection [4509]	1395	<p>AMH1 60097</p> <p>AMHR2 60096</p> <p>ANOS1 30036</p> <p>AR 11700</p> <p>ARL4 00885</p> <p>ARX 30082</p> <p>ATRX 30002</p> <p>BBS1 20901</p> <p>BBS10 61048</p> <p>BBS2 61063</p> <p>BBS2 60615</p> <p>BBS4 60374</p> <p>BBS5 60350</p> <p>BBS7 60790</p> <p>BBS9 60798</p> <p>CBX2 60270</p> <p>CHD7 60892</p> <p>CYP11A1 11845</p> <p>CYP17A1 60930</p> <p>CYP19A1 107010</p> <p>DMH1 60543</p> <p>DMRT1 60242</p> <p>DMRT2 60493</p> <p>FCF8 00483</p> <p>FCFR1 13050</p> <p>FCFR2 17643</p> <p>FOXL2 60597</p> <p>FSH3 13030</p> <p>GATA4 60076</p> <p>GNRH1 15270</p> <p>GNRHR 13850</p> <p>HESX1 60180</p> <p>HFE 61309</p> <p>HSD17B3 60573</p> <p>KISS1 60236</p> <p>KISS1B 60416</p> <p>LEP 16410</p> <p>LEPR 61007</p> <p>LHCOR 15290</p> <p>LUX3 60577</p> <p>LUX4 60216</p> <p>MAP3K1 30020</p> <p>MAP3K1 60982</p> <p>MKKS 60496</p> <p>NROB1 30475</p> <p>NROB1 184757</p> <p>NSMF 60817</p> <p>PCSK1 60250</p> <p>PCSK1 12405</p> <p>PCSK1 60900</p>	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	<p>3-Oxo-5 Alpha-Steroid Delta 4-Dehydrogenase Deficiency AD 264600</p> <p>46,XX Sex Reversal, Type 1 AR 400045</p> <p>46,XY Sex Reversal, Type 5 AD 612965</p> <p>46,XY Sex Reversal, Type 6 AD 613080</p> <p>46,XY Sex Reversal, Type 7 AD 613762</p> <p>46,XY Sex Reversal, Type 7 AR 233420</p> <p>Adrenal Insufficiency, Congenital, With 46,XY Sex Reversal, Partial Or Complete AR 613743</p> <p>Androgen Resistance Syndrome XL 300068</p> <p>Antley-Bixler Syndrome AR 207410</p> <p>Antley-Bixler Syndrome With Genital Anomalies And Disordered Steroidogenesis AR 201750</p> <p>ATR-X Syndrome XL 301040</p> <p>Bardet-Biedl Syndrome 1 AR 209900</p> <p>Bardet-Biedl Syndrome 10 AR 615987</p> <p>Bardet-Biedl Syndrome 11 AR 615988</p> <p>Bardet-Biedl Syndrome 12 AR 615989</p> <p>Bardet-Biedl Syndrome 2 AR 615981</p> <p>Bardet-Biedl Syndrome 3 AR 600151</p> <p>Bardet-Biedl Syndrome 4 AR 615982</p> <p>Bardet-Biedl Syndrome 5 AR 615983</p> <p>Bardet-Biedl Syndrome 6 AR 605231</p> <p>Bardet-Biedl Syndrome 7 AR 615984</p> <p>Bardet-Biedl Syndrome 8 AR 615985</p> <p>Bardet-Biedl Syndrome 9 AR 615986</p> <p>Blepharophimosis, Ptosis, And Ectocantus Inversus AR 110100</p> <p>Camptomic Dysplasia AD 114290</p> <p>Cholesterol Monoxygenase (Side-Chain Cleaving) Deficiency AR 201710</p> <p>Deficiency Of Steroid 17-Alpha-Monoxygenase AD,AR 202110</p> <p>Familial Gynecomastia, Due To Increased Aromatase Activity AR 139300</p> <p>Follicle-Stimulating Hormone Deficiency, Isolated XL 229070</p> <p>Gonadotropin-Independent Familial Sexual Precocity AR 176410</p> <p>Hemochromatosis Type 1 AR 235200</p> <p>Hypogonadotropic Hypogonadism 10 with or without Anosmia AR 614839</p> <p>Hypogonadotropic Hypogonadism 11 with or without Anosmia AR 614840</p>	Exome sequencing with CNV detection		06-Jun-19
DISTAL HEREDITARY MOTOR NEUROPATHY NGS PANEL		<p>ATP7A (30001); BSCL2 (606158); DCTN1 (601143); DNMT1 (126275); FIG4 (60390); GAN (605379); GARS (60227); HSPB1 (602195); HSPB8 (608014); IGHMBP2 (609002); MEEF3 (612453); REEP1 (609139); SETX (608465); SLC5A7 (606761); TRPV4 (605427)</p>	Lavender Top (2 x 4 mL)	Fulgent Genetics (fulgentdiagnostics.com)	<p>Distal Hereditary Motor Neuropathy Type 2B (608634); Charcot-Marie-Tooth Disease Type 2F (606595); Distal Hereditary Motor Neuropathy Type 2A (158590); Charcot-Marie-Tooth Disease, Type 2L (608673); Distal Hereditary Motor Neuropathy 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); Scapulohumeral Spinal Muscular Atrophy (181405); NEURONOPATHY, DISTAL HEREDITARY MOTOR, TYPE VIII; HMN7B (607641); Perry Syndrome (168605); Charcot-Marie-Tooth Disease, Type 4J (611228); Neuropathy, Hereditary Sensory, Type 1e (614116); Myopathy, Early-Onset, Areflexia, Respiratory Distress, And Dysphagia (614399); Neuropathy, distal hereditary motor, type VIIA (158380); Giant Axonal Neuropathy (256850); Spinal Muscular Atrophy, Distal, X-Linked 3 (300489); Neurotonia and axonal neuropathy, autosomal recessive (137200); Neuropathy, distal hereditary motor, type VIb (614751); Spinal Muscular Atrophy With Respiratory Distress 1 (604320)</p>	Next Generation Sequencing (NGS) and Sanger sequencing technologies		
Diuretic Screen, Urine [FDIRU]			Random urine	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	benzthiazide, bumetanide, chlorothiazide, chlorthalidone, furosemide, hydrochlorothiazide, hydroflumethiazide, and metolazone			
DNA Double-Stranded Antibodies, IgG, Serum [ADNA]	1465		Serum SST. Stable at -20°C for 21 days.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)		ELISA		20-Aug-21
DOPA-RESPONSIVE DYSTONIA VIA THE GCH1 GENE [161]	1096	GCA1 (60225)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Dystonia 5, Dopa-Responsive Type (128230)	Sanger sequencing		
Doxycycline [94093]	1465		Serum (2 mL). Store frozen.	Quest Diagnostics				21-Feb-20
Drug Dependent Platelet Antibody [9000]	1465		Gold SST	Blood Center of Wisconsin				
Drug Screen, Prescription/OTC, Urine [PDSU]	1465		Random urine	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	Trifluoperazine (stelazine)			20-Oct-20

Appellation 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 (Déclaratoire MSSS)	SYMBOL / IGENC DU GÈNE et NOM OMIM DU GÈNE / IGENC Gene Symbol and OMIM Gene Number	RÈGLES DE MANUTENTION / Handling Conditions	LABORATOIRE DÉSIGNÉ / Designated laboratory	RENSEIGNEMENTS SUR L'ANALYSE / Test details	MÉTHODE / Methode	NOTE PARTICULIÈRE / Special comment	Date de la révision / Date of revision
1,5-anhydroglucitol (GlycoMark) [500115] [GLYC]	1465		Gold Top or Red Top (Serum) or Lavender (plasma). Centrifuge and aliquot serum to transport tube within one hour of collection. Store and ship frozen. Stable at -20°C for 14 days.	LabCorp/Dynacare (https://www.labcorp.com/)		Enzymatic, colorimetric assay.		10-Jan-21
Duchenne and Becker Muscular Dystrophy (310200 and 300376)	617	DMD (00077)	Lavender Top (EDTA)	Molecular Genetics Laboratory, Hospital for Sick Children (www.sickkids.ca/PaediatricLaboratoryMedicine/Laboratories-Services/Molecular-Genetics-Laboratory/index.html)		1. Sanger sequencing 2. gene dosage		04-May-17
Dystonia Dyskinesia NGS Panel	1096	AND3, ATP1A3, CACNA1B, CIZ1, COL4A3, DRD2, DRD3, OCH1, GNAL, HPCA, KCTD17, PKNOX, PRKN, PRKRA, PRRT2, SLC6A3, SLC6A4, SLC6A5, SFR, TAF1, TH, THAP1, TOR1A, TUBB8A	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 (603751); Leukoencephalopathy with dystonia and motor neuropathy (613724); Dystonia-11, myoclonic (159900); Parkinsonism-dystonia, infantile (613315); Dystonia, dipa-responsive, due to sepiapterin reductase deficiency (612716); Dystonia-Parkinsonism, X-linked (314250); Dystonia 6, torsion (602629); Dystonia-1, torsion (128100)	Next-Generation sequencing	See Med Neurogenetics Panel	13-Jul-21
Dystonia/Parkinson Panel [T402]	1096		Lavender Top (EDTA)	GeneDx (www.genedx.com)		Next-Generation sequencing		04-Apr-22
Early-Onset Epileptic Encephalopathy Panel		ACV1, ADGRV1, ADSL, ALDH1A1, ALG1, ARHGAP2, ARHGAP5, ARHGAP9, ARK, ATP1A1, ATP1A2, ATR, BCKR1, CACNA1A, CACNA1B, CACNA1C, CACNA1E, CACNA1F, CACNA1G, CACNA1H, CACNA1I, CACNA1J, CACNA1K, CACNA1L, CACNA1M, CACNA1N, CACNA1O, CACNA1P, CACNA1Q, CACNA1R, CACNA1S, CACNA1T, CACNA1U, CACNA1V, CACNA1W, CACNA1X, CACNA1Y, CACNA1Z, CACNA2D1, CACNA2D2, CACNA2D3, CACNA2D4, CACNA2D5, CACNA2D6, CACNA2D7, CACNA2D8, CACNA2D9, CACNA2D10, CACNA2D11, CACNA2D12, CACNA2D13, CACNA2D14, CACNA2D15, CACNA2D16, CACNA2D17, CACNA2D18, CACNA2D19, CACNA2D20, CACNA2D21, CACNA2D22, CACNA2D23, CACNA2D24, CACNA2D25, CACNA2D26, CACNA2D27, CACNA2D28, CACNA2D29, CACNA2D30, CACNA2D31, CACNA2D32, CACNA2D33, CACNA2D34, CACNA2D35, CACNA2D36, CACNA2D37, CACNA2D38, CACNA2D39, CACNA2D40, CACNA2D41, CACNA2D42, CACNA2D43, CACNA2D44, CACNA2D45, CACNA2D46, CACNA2D47, CACNA2D48, CACNA2D49, CACNA2D50, CACNA2D51, CACNA2D52, CACNA2D53, CACNA2D54, CACNA2D55, CACNA2D56, CACNA2D57, CACNA2D58, CACNA2D59, CACNA2D60, CACNA2D61, CACNA2D62, CACNA2D63, CACNA2D64, CACNA2D65, CACNA2D66, CACNA2D67, CACNA2D68, CACNA2D69, CACNA2D70, CACNA2D71, CACNA2D72, CACNA2D73, CACNA2D74, CACNA2D75, CACNA2D76, CACNA2D77, CACNA2D78, CACNA2D79, CACNA2D80, CACNA2D81, CACNA2D82, CACNA2D83, CACNA2D84, CACNA2D85, CACNA2D86, CACNA2D87, CACNA2D88, CACNA2D89, CACNA2D90, CACNA2D91, CACNA2D92, CACNA2D93, CACNA2D94, CACNA2D95, CACNA2D96, CACNA2D97, CACNA2D98, CACNA2D99, CACNA2D100, CACNA2D101, CACNA2D102, CACNA2D103, CACNA2D104, CACNA2D105, CACNA2D106, CACNA2D107, CACNA2D108, CACNA2D109, CACNA2D110, CACNA2D111, CACNA2D112, CACNA2D113, CACNA2D114, CACNA2D115, CACNA2D116, CACNA2D117, CACNA2D118, CACNA2D119, CACNA2D120, CACNA2D121, CACNA2D122, CACNA2D123, CACNA2D124, CACNA2D125, CACNA2D126, CACNA2D127, CACNA2D128, CACNA2D129, CACNA2D130, CACNA2D131, CACNA2D132, CACNA2D133, CACNA2D134, CACNA2D135, CACNA2D136, CACNA2D137, CACNA2D138, CACNA2D139, CACNA2D140, CACNA2D141, CACNA2D142, CACNA2D143, CACNA2D144, CACNA2D145, CACNA2D146, CACNA2D147, CACNA2D148, CACNA2D149, CACNA2D150, CACNA2D151, CACNA2D152, CACNA2D153, CACNA2D154, CACNA2D155, CACNA2D156, CACNA2D157, CACNA2D158, CACNA2D159, CACNA2D160, CACNA2D161, CACNA2D162, CACNA2D163, CACNA2D164, CACNA2D165, CACNA2D166, CACNA2D167, CACNA2D168, CACNA2D169, CACNA2D170, CACNA2D171, CACNA2D172, CACNA2D173, CACNA2D174, CACNA2D175, CACNA2D176, CACNA2D177, CACNA2D178, CACNA2D179, CACNA2D180, CACNA2D181, CACNA2D182, CACNA2D183, CACNA2D184, CACNA2D185, CACNA2D186, CACNA2D187, CACNA2D188, CACNA2D189, CACNA2D190, CACNA2D191, CACNA2D192, CACNA2D193, CACNA2D194, CACNA2D195, CACNA2D196, CACNA2D197, CACNA2D198, CACNA2D199, CACNA2D200, CACNA2D201, CACNA2D202, CACNA2D203, CACNA2D204, CACNA2D205, CACNA2D206, CACNA2D207, CACNA2D208, CACNA2D209, CACNA2D210, CACNA2D211, CACNA2D212, CACNA2D213, CACNA2D214, CACNA2D215, CACNA2D216, CACNA2D217, CACNA2D218, CACNA2D219, CACNA2D220, CACNA2D221, CACNA2D222, CACNA2D223, CACNA2D224, CACNA2D225, CACNA2D226, CACNA2D227, CACNA2D228, CACNA2D229, CACNA2D230, CACNA2D231, CACNA2D232, CACNA2D233, CACNA2D234, CACNA2D235, CACNA2D236, CACNA2D237, CACNA2D238, CACNA2D239, CACNA2D240, CACNA2D241, CACNA2D242, CACNA2D243, CACNA2D244, CACNA2D245, CACNA2D246, CACNA2D247, CACNA2D248, CACNA2D249, CACNA2D250, CACNA2D251, CACNA2D252, CACNA2D253, CACNA2D254, CACNA2D255, CACNA2D256, CACNA2D257, CACNA2D258, CACNA2D259, CACNA2D260, CACNA2D261, CACNA2D262, CACNA2D263, CACNA2D264, CACNA2D265, CACNA2D266, CACNA2D267, CACNA2D268, CACNA2D269, CACNA2D270, CACNA2D271, CACNA2D272, CACNA2D273, CACNA2D274, CACNA2D275, CACNA2D276, CACNA2D277, CACNA2D278, CACNA2D279, CACNA2D280, CACNA2D281, CACNA2D282, CACNA2D283, CACNA2D284, CACNA2D285, CACNA2D286, CACNA2D287, CACNA2D288, CACNA2D289, CACNA2D290, CACNA2D291, CACNA2D292, CACNA2D293, CACNA2D294, CACNA2D295, CACNA2D296, CACNA2D297, CACNA2D298, CACNA2D299, CACNA2D300, CACNA2D301, CACNA2D302, CACNA2D303, CACNA2D304, CACNA2D305, CACNA2D306, CACNA2D307, CACNA2D308, CACNA2D309, CACNA2D310, CACNA2D311, CACNA2D312, CACNA2D313, CACNA2D314, CACNA2D315, CACNA2D316, CACNA2D317, CACNA2D318, CACNA2D319, CACNA2D320, CACNA2D321, CACNA2D322, CACNA2D323, CACNA2D324, CACNA2D325, CACNA2D326, CACNA2D327, CACNA2D328, CACNA2D329, CACNA2D330, CACNA2D331, CACNA2D332, CACNA2D333, CACNA2D334, CACNA2D335, CACNA2D336, CACNA2D337, CACNA2D338, CACNA2D339, CACNA2D340, CACNA2D341, CACNA2D342, CACNA2D343, CACNA2D344, CACNA2D345, CACNA2D346, CACNA2D347, CACNA2D348, CACNA2D349, CACNA2D350, CACNA2D351, CACNA2D352, CACNA2D353, CACNA2D354, CACNA2D355, CACNA2D356, CACNA2D357, CACNA2D358, CACNA2D359, CACNA2D360, CACNA2D361, CACNA2D362, CACNA2D363, CACNA2D364, CACNA2D365, CACNA2D366, CACNA2D367, CACNA2D368, CACNA2D369, CACNA2D370, CACNA2D371, CACNA2D372, CACNA2D373, CACNA2D374, CACNA2D375, CACNA2D376, CACNA2D377, CACNA2D378, CACNA2D379, CACNA2D380, CACNA2D381, CACNA2D382, CACNA2D383, CACNA2D384, CACNA2D385, CACNA2D386, CACNA2D387, CACNA2D388, CACNA2D389, CACNA2D390, CACNA2D391, CACNA2D392, CACNA2D393, CACNA2D394, CACNA2D395, CACNA2D396, CACNA2D397, CACNA2D398, CACNA2D399, CACNA2D400, CACNA2D401, CACNA2D402, CACNA2D403, CACNA2D404, CACNA2D405, CACNA2D406, CACNA2D407, CACNA2D408, CACNA2D409, CACNA2D410, CACNA2D411, CACNA2D412, CACNA2D413, CACNA2D414, CACNA2D415, CACNA2D416, CACNA2D417, CACNA2D418, CACNA2D419, CACNA2D420, CACNA2D421, CACNA2D422, CACNA2D423, CACNA2D424, CACNA2D425, CACNA2D426, CACNA2D427, CACNA2D428, CACNA2D429, CACNA2D430, CACNA2D431, CACNA2D432, CACNA2D433, CACNA2D434, CACNA2D435, CACNA2D436, CACNA2D437, CACNA2D438, CACNA2D439, CACNA2D440, CACNA2D441, CACNA2D442, CACNA2D443, CACNA2D444, CACNA2D445, CACNA2D446, CACNA2D447, CACNA2D448, CACNA2D449, CACNA2D450, CACNA2D451, CACNA2D452, CACNA2D453, CACNA2D454, CACNA2D455, CACNA2D456, CACNA2D457, CACNA2D458, CACNA2D459, CACNA2D460, CACNA2D461, CACNA2D462, CACNA2D463, CACNA2D464, CACNA2D465, CACNA2D466, CACNA2D467, CACNA2D468, CACNA2D469, CACNA2D470, CACNA2D471, CACNA2D472, CACNA2D473, CACNA2D474, CACNA2D475, CACNA2D476, CACNA2D477, CACNA2D478, CACNA2D479, CACNA2D480, CACNA2D481, CACNA2D482, CACNA2D483, CACNA2D484, CACNA2D485, CACNA2D486, CACNA2D487, CACNA2D488, CACNA2D489, CACNA2D490, CACNA2D491, CACNA2D492, CACNA2D493, CACNA2D494, CACNA2D495, CACNA2D496, CACNA2D497, CACNA2D498, CACNA2D499, CACNA2D500, CACNA2D501, CACNA2D502, CACNA2D503, CACNA2D504, CACNA2D505, CACNA2D506, CACNA2D507, CACNA2D508, CACNA2D509, CACNA2D510, CACNA2D511, CACNA2D512, CACNA2D513, CACNA2D514, CACNA2D515, CACNA2D516, CACNA2D517, CACNA2D518, CACNA2D519, CACNA2D520, CACNA2D521, CACNA2D522, CACNA2D523, CACNA2D524, CACNA2D525, CACNA2D526, CACNA2D527, CACNA2D528, CACNA2D529, CACNA2D530, CACNA2D531, CACNA2D532, CACNA2D533, CACNA2D534, CACNA2D535, CACNA2D536, CACNA2D537, CACNA2D538, CACNA2D539, CACNA2D540, CACNA2D541, CACNA2D542, CACNA2D543, CACNA2D544, CACNA2D545, CACNA2D546, CACNA2D547, CACNA2D548, CACNA2D549, CACNA2D550, CACNA2D551, CACNA2D552, CACNA2D553, CACNA2D554, CACNA2D555, CACNA2D556, CACNA2D557, CACNA2D558, CACNA2D559, CACNA2D560, CACNA2D561, CACNA2D562, CACNA2D563, CACNA2D564, CACNA2D565, CACNA2D566, CACNA2D567, CACNA2D568, CACNA2D569, CACNA2D570, CACNA2D571, CACNA2D572, CACNA2D573, CACNA2D574, CACNA2D575, CACNA2D576, CACNA2D577, CACNA2D578, CACNA2D579, CACNA2D580, CACNA2D581, CACNA2D582, CACNA2D583, CACNA2D584, CACNA2D585, CACNA2D586, CACNA2D587, CACNA2D588, CACNA2D589, CACNA2D590, CACNA2D591, CACNA2D592, CACNA2D593, CACNA2D594, CACNA2D595, CACNA2D596, CACNA2D597, CACNA2D598, CACNA2D599, CACNA2D600, CACNA2D601, CACNA2D602, CACNA2D603, CACNA2D604, CACNA2D605, CACNA2D606, CACNA2D607, CACNA2D608, CACNA2D609, CACNA2D610, CACNA2D611, CACNA2D612, CACNA2D613, CACNA2D614, CACNA2D615, CACNA2D616, CACNA2D617, CACNA2D618, CACNA2D619, CACNA2D620, CACNA2D621, CACNA2D622, CACNA2D623, CACNA2D624, CACNA2D625, CACNA2D626, CACNA2D627, CACNA2D628, CACNA2D629, CACNA2D630, CACNA2D631, CACNA2D632, CACNA2D633, CACNA2D634, CACNA2D635, CACNA2D636, CACNA2D637, CACNA2D638, CACNA2D639, CACNA2D640, CACNA2D641, CACNA2D642, CACNA2D643, CACNA2D644, CACNA2D645, CACNA2D646, CACNA2D647, CACNA2D648, CACNA2D649, CACNA2D650, CACNA2D651, CACNA2D652, CACNA2D653, CACNA2D654, CACNA2D655, CACNA2D656, CACNA2D657, CACNA2D658, CACNA2D659, CACNA2D660, CACNA2D661, CACNA2D662, CACNA2D663, CACNA2D664, CACNA2D665, CACNA2D666, CACNA2D667, CACNA2D668, CACNA2D669, CACNA2D670, CACNA2D671, CACNA2D672, CACNA2D673, CACNA2D674, CACNA2D675, CACNA2D676, CACNA2D677, CACNA2D678, CACNA2D679, CACNA2D680, CACNA2D681, CACNA2D682, CACNA2D683, CACNA2D684, CACNA2D685, CACNA2D686, CACNA2D687, CACNA2D688, CACNA2D689, CACNA2D690, CACNA2D691, CACNA2D692, CACNA2D693, CACNA2D694, CACNA2D695, CACNA2D696, CACNA2D697, CACNA2D698, CACNA2D699, CACNA2D700, CACNA2D701, CACNA2D702, CACNA2D703, CACNA2D704, CACNA2D705, CACNA2D706, CACNA2D707, CACNA2D708, CACNA2D709, CACNA2D710, CACNA2D711, CACNA2D712, CACNA2D713, CACNA2D714, CACNA2D715, CACNA2D716, CACNA2D717, CACNA2D718, CACNA2D719, CACNA2D720, CACNA2D721, CACNA2D722, CACNA2D723, CACNA2D724, CACNA2D725, CACNA2D726, CACNA2D727, CACNA2D728, CACNA2D729, CACNA2D730, CACNA2D731, CACNA2D732, CACNA2D733, CACNA2D734, CACNA2D735, CACNA2D736, CACNA2D737, CACNA2D738, CACNA2D739, CACNA2D740, CACNA2D741, CACNA2D742, CACNA2D743, CACNA2D744, CACNA2D745, CACNA2D746, CACNA2D747, CACNA2D748, CACNA2D749, CACNA2D750, CACNA2D751, CACNA2D752, CACNA2D753, CACNA2D754, CACNA2D755, CACNA2D756, CACNA2D757, CACNA2D758, CACNA2D759, CACNA2D760, CACNA2D761, CACNA2D762, CACNA2D763, CACNA2D764, CACNA2D765, CACNA2D766, CACNA2D767, CACNA2D768, CACNA2D769, CACNA2D770, CACNA2D771, CACNA2D772, CACNA2D773, CACNA2D774, CACNA2D775, CACNA2D776, CACNA2D777, CACNA2D778, CACNA2D779, CACNA2D780, CACNA2D781, CACNA2D782, CACNA2D783, CACNA2D784, CACNA2D785, CACNA2D786, CACNA2D787, CACNA2D788, CACNA2D789, CACNA2D790, CACNA2D791, CACNA2D792, CACNA2D793, CACNA2D794, CACNA2D795, CACNA2D796, CACNA2D797, CACNA2D798, CACNA2D799, CACNA2D800, CACNA2D801, CACNA2D802, CACNA2D803, CACNA2D804, CACNA2D805, CACNA2D806, CACNA2D807, CACNA2D808, CACNA2D809, CACNA2D810, CACNA2D811, CACNA2D812, CACNA2D813, CACNA2D814, CACNA2D815, CACNA2D816, CACNA2D817, CACNA2D818, CACNA2D819, CACNA2D820, CACNA2D821, CACNA2D822, CACNA2D823, CACNA2D824, CACNA2D825, CACNA2D826, CACNA2D827, CACNA2D828, CACNA2D829, CACNA2D830, CACNA2D831, CACNA2D832, CACNA2D833, CACNA2D834, CACNA2D835, CACNA2D836, CACNA2D837, CACNA2D838, CACNA2D839, CACNA2D840, CACNA2D841, CACNA2D842, CACNA2D843, CACNA2D844, CACNA2D845, CACNA2D846, CACNA2D847, CACNA2D848, CACNA2D849, CACNA2D850, CACNA2D851, CACNA2D852, CACNA2D853, CACNA2D854, CACNA2D855, CACNA2D856, CACNA2D857, CACNA2D858, CACNA2D859, CACNA2D860, CACNA2D861, CACNA2D862, CACNA2D863, CACNA2D864, CACNA2D865, CACNA2D866, CACNA2D867, CACNA2D868, CACNA2D869, CACNA2D870, CACNA2D871, CACNA2D872, CACNA2D873, CACNA2D874, CACNA2D875, CACNA2D876, CACNA2D877, CACNA2D878, CACNA2D879, CACNA2D880, CACNA2D881, CACNA2D882, CACNA2D883, CACNA2D884, CACNA2D885, CACNA2D886, CACNA2D887, CACNA2D888, CACNA2D889, CACNA2D890, CACNA2D891, CACNA2D892, CACNA2D893, CACNA2D894, CACNA2D895, CACNA2D896, CACNA2D897, CACNA2D898, CACNA2D899, CACNA2D900, CACNA2D901, CACNA2D902, CACNA2D903, CACNA2D904, CACNA2D905, CACNA2D906, CACNA2D907, CACNA2D908, CACNA2D909, CACNA2D910, CACNA2D911, CACNA2D912, CACNA2D913, CACNA2D914, CACNA2D915, CACNA2D916, CACNA2D917, CACNA2D918, CACNA2D919, CACNA2D920, CACNA2D921, CACNA2D922, CACNA2D923, CACNA2D924, CACNA2D925, CACNA2D926, CACNA2D927, CACNA2D928, CACNA2D929, CACNA2D930, CACNA2D931, CACNA2D932, CACNA2D933, CACNA2D934, CACNA2D935, CACNA2D936, CACNA2D937, CACNA2D938, CACNA2D939, CACNA2D940, CACNA2D941, CACNA2D942, CACNA2D943, CACNA2D944, CACNA2D945, CACNA2D946, CACNA2D947, CACNA2D948, CACNA2D949, CACNA2D950, CACNA2D951, CACNA2D952, CACNA2D953, CACNA2D954, CACNA2D955, CACNA2D956, CACNA2D957, CACNA2D958, CACNA2D959, CACNA2D960, CACNA2D961, CACNA2D962, CACNA2D963, CACNA2D964, CACNA2D965, CACNA2D966, CACNA2D967, CACNA2D968, CACNA2D969, CACNA2D970, CACNA2D971, CACNA2D972, CACNA2D973, CACNA2D974, CACNA2D975, CACNA2D976, CACNA2D977, CACNA2D978, CACNA2D979, CACNA2D980, CACNA2D981, CACNA2D982, CACNA2D983, CACNA2D984, CACNA2D985, CACNA2D986, CACNA2D987, CACNA2D988, CACNA2D989, CACNA2D990, CACNA2D991, CACNA2D992, CACNA2D993, CACNA2D994, CACNA2D995, CACNA2D996, CACNA2D997, CACNA2D998, CACNA2D999, CACNA2D1000, CACNA2D1001, CACNA2D1002, CACNA2D1003, CACNA2D1004, CACNA2D1005, CACNA2D1006, CACNA2D1007, CACNA2D1008, CACNA2D1009, CACNA2D1010, CACNA2D1011, CACNA2D1012, CACNA2D1013, CACNA2D1014, CACNA2D1015, CACNA2D1016, CACNA2D1017, CACNA2D1018, CACNA2D1019, CACNA2D1020, CACNA2D1021, CACNA2D1022, CACNA2D1023, CACNA2D1024, CACNA2D1025, CACNA2D1026, CACNA2D1027, CACNA2D1028, CACNA2D1029, CACNA2D1030, CACNA2D1031, CACNA2D1032, CACNA2D1033, CACNA2D1034, CACNA2D1035, CACNA2D1036, CACNA2D1037, CACNA2D1038, CACNA2D1039, CACNA2D1040, CACNA2D1041, CACNA2D1042, CACNA2D1043, CACNA2D1044, CACNA2D1045, CACNA2D1046, CACNA2D1047, CACNA2D1048, CACNA2D1049, CACNA2D1050, CACNA2D1051, CACNA2D1052, CACNA2D1053, CACNA2D1054, CACNA2D1055, CACNA2D1056, CACNA2D1057, CACNA2D1058, CACNA2D1059, CACNA2D1060, CACNA2D1061, CACNA2D1062, CACNA2D1063, CACNA2D1064, CACNA2D1065, CACNA2D1066, CACNA2D1067, CACNA2D1068, CACNA2D1069, CACNA2D1070, CACNA2D1071, CACNA2D1072, CACNA2D1073, CACNA2D1074, CACNA2D1075, CACNA2D1076, CACNA2D1077, CACNA2D1078, CACNA2D1079, CACNA2D1080, CACNA2D1081, CACNA2D1082, CACNA2D1083, CACNA2D1084, CACNA2D1085, CACNA2D1086, CACNA2D1087, CACNA2D1088, CACNA2D1089, CACNA2D1090, CACNA2D1091, CACNA2D1092, CACNA2D1093, CACNA2D1094, CACNA2D1095, CACNA2D1096, CACNA2D1097, CACNA2D1098, CACNA2D1099, CACNA2D1100, CACNA2D1101, CACNA2D1102, CACNA2D1103, CACNA2D1104, CACNA2D1105, CACNA2D1106, CACNA2D1107, CACNA2D1108, CACNA2D1109, CACNA2D1110, CACNA2D1111, CACNA2D1112, CACNA2D1113, CACNA2D1114, CACNA2D1115, CACNA2D1116, CACNA2D1117, CACNA2D1118, CACNA2D1119, CACNA2D1120, CACNA2D1121, CACNA2D1122, CACNA2D1123, CACNA2D1124, CACNA2D1125, CACNA2D1126, CACNA2D1127, CACNA2D1128, CACNA2D1129, CACNA2D1130, CACNA2D1131, CACNA2D1132, CACNA2D1133, CACNA2D1134, CACNA2D1135, CACNA2D1136, CACNA2D1137, CACNA2D1138, CACNA2D1139, CACNA2D1140, CACNA2D1141, CACNA2D1142, CACNA2D1143, CACNA2D1144, CACNA2D1145, CACNA2D1146, CACNA2D1147, CACNA2D1148, CACNA2D1149, CACNA2D1150, CACNA2D1151, CACNA2D1152, CACNA2D1153, CACNA2D1154, CACNA2D1155, CACNA2D1156, CACNA2D1157, CACNA2D1158, CACNA2D1159, CACNA2D1160, CACNA2D1161, CACNA2D1162, CACNA2D1163, CACNA2D1164, CACNA2D1165, CACNA2D1166, CACNA2D1167, CACNA2D1168, CACNA2D1169, CACNA2D1170, CACNA2D1171, CACNA2D1172, CACNA2D1173, CACNA2D1174, CACNA2D1175, CACNA2D1176, CACNA2D1177, CACNA2D1178, CACNA2D1179, CACNA2D1180, CACNA2D1181, CAC						

Appellation 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 (Déclinaire MSSS)	SYMBOL/IGENE DU GÈNE et NOM OMIM DU GÈNE / IGENE Gene Symbol and OMIM Gene Number	RÈGLES DE MANUTENTION / Handling Conditions	LABORATOIRE DÉSIGNÉ / Designated laboratory	RENSEIGNEMENTS SUR L'ANALYSE / Test details	MÉTHODE / Methode	NOTE PARTICULIÈRE / Special comment	Date de la révision / Date of revision
1,5-anhydroglucitol (GlycoMark) [500115] [GLYC]	1465		Gold Top or Red Top (Serum) or Lavender (plasma). Centrifuge and aliquot serum to transport tube within one hour of collection. Store and ship frozen. Stable at -20°C for 14 days.	LabCorp/Dynacare (https://www.labcorp.com/)		Enzymatic, colorimetric assay.		10-Jan-21
Expanded Neuromuscular Disorders: Sequencing and Deletion/Duplication Panel[MM360]		ACTA1, AMPD1, AMPD3, ANOS, BAG1, BIN1, BSCL2, CAPN1, CAV3, CFL2, CHAT, CHRNA1, CHRN1, CHRN2, CHRN3, CHRN4, COL4A1, COL4A2, COL4A3, COL4, CRYAB, DAG1, DES, DMD, DOK6, DOK7, DYX5, EBD, FHL1, FKBP, FKBP1, FLNC, GAA, GLE1, GNE, IGHMBP2, ISPD, ITGA7, LAMA2, LARG1, LDB1, LMNA, MTM1, MTMR14, MUSK, MYH1, MYH7, MYO1, NER, PABPN1, PLEC, PLEKHG5, PPM1A, PPM1B, PPM1L, PPM1R, PTRF, PYGM, RAPS1, RYR1, RYR2, SCN1A, SEPN1, SGCA, SGCB, SGCD, SGCE, SGCG, SIL1, SYNE1, SYNE2, TCAF, TNIN2, TNNT1, TPM2, TPM3, TRIM2, TRN, VRR1	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, Marfan/Sjogren syndrome, Dialal arthropropis, NMD, Neuromuscular disorders		Formerly Emory	
F12 Select Exons Sequencing [388]	1832	F12 (600019)	Lavender Top (2 x 4 mL)	GeneDx (www.genedx.com)	Hereditary Angioedema (HAE) Type III (610618)			14-Aug-17
Facioscapulohumeral Muscular Dystrophy	984	DUXA (D424) (600099)	Lavender Top (EDTA)	Children's Hospital of Eastern Ontario	FACEOSCAPULOHUMERAL MUSCULAR DYSTROPHY 1 (158900)	deletion and haplotype		
Factor B			0.5 mL red top serum- spun, separated, frozen within 2 hrs of collection; ship on dry ice	Cincinnati Children's Hospital Medical Center 3333 Burnet Avenue NRB 1042 Cincinnati, OH 45229 (www.cincinnatichildrens.org)				15-Nov-17
Factor H autoantibodies	64		2ml frozen serum.	Cincinnati Children's Hospital Medical Center 3333 Burnet Avenue NRB 1042 Cincinnati, OH 45229 (www.cincinnatichildrens.org)				28-Jun-19
Factor I			2ml frozen serum.	Cincinnati Children's Hospital Medical Center 3333 Burnet Avenue NRB 1042 Cincinnati, OH 45229 (www.cincinnatichildrens.org)		complement-mediated renal diseases F1		15-Nov-17
Familial Hemiplegic Migraine (NGS Panel and Copy Number Analysis + mtDNA) [NGS429]	581	ATP1A2, ATP1A3, BAP1, CACNA1A, PRR12, SCN1A	Lavender Top	MNG Laboratories (www.medicalneurogenetics.com)/Dynacare		Next-Generation sequencing	Replaced by Blueprint panel	21-Aug-20
Familial Hemophagocytic Lymphohistiocytosis (FHL), Autosomal Recessive; type 2 (603553), type 3 (608898), type 4 (603552) and type 5 (613101)		PRF1 (170280), UNC119 (MIM:12114) (608897), STXBP2 (601717), RAB27A (603866), STX11 (605914)	Lavender Top (EDTA)	Cincinnati 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 Hypocalcemic Hypercalcemia (FHH) Panel [10125]	805	AP2S1, CASR, GNA11	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)		Next-Generation sequencing		23-Sep-21
Familial Limb Girdle Myasthenia Syndrome via DOK7 Gene [7629]	626	DOK7 (602085)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Myasthenia, Limb-Girdle, Familial (254300)		Available at MNG	20-Aug-21
Farmer's Lung IgG Antibodies, Serum [FLGAB]	1465		Serum (2 mL). Store frozen.	In-Common Laboratories	M. faeni IgG Ab; T. vulgaris IgG Ab	FEIA		10-Mar-20
Fat, Feces [FATF]	1465		For a random collection, a minimum of 5 g (do not send entire collection) is required. Store frozen.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)		Nuclear Magnetic Resonance (NMR) Spectroscopy		03-Aug-22
Fatty Acid Oxidation Deficiency NGS Panel	528	ACAD9, ACADL, ACADM, ACADS, ACADVL, CPT1A, CPT1B, CPT2, ETFB, ETFD, ETLCD, GLE1, HADHA, HADHB, HADH, HADHCE1, HADHCE2, HSD17B10, LPIN1, SLC22A5, SLC25A20, TAZ	Lavender Top (2 x 4 mL)	Fulgent Genetics (fulgentdiagnostics.com)	Includes: Fatty liver, acute, of pregnancy (609016); HELLP syndrome, maternal, of pregnancy (609016)	NextGen Sequencing	Replaces: UofA targeted mutation testing	
Fatty Acid Oxidation Syndrome Panel [ME1701]	1320		Lavender Top (2 x 4 mL)	Blueprint Genetics http://blueprintgenetics.com/		Next-Generation sequencing		29-Feb-20
FH Autoantibody Testing	64		2ml frozen serum.	Cincinnati Children's Hospital Medical Center 3333 Burnet Avenue NRB 1042 Cincinnati, OH 45229 (www.cincinnatichildrens.org)		complement-mediated renal diseases F1		15-Nov-17
FH Autoantibody Testing	64		Panel requires at least 2ml frozen serum.	Molecular Otolaryngology & Renal Research Laboratory, U. of Iowa Carver College of Medicine (www.medicine.uiowa.edu/morl)	Hemolytic uremic syndrome, atypical, susceptibility to, 5 (612925)	ELSIA and other assays		31-Oct-17
Fibrodysplasia ossificans progressiva (135100)		ACVR1 (102570)	Lavender Top (EDTA)	University of Pennsylvania School of Medicine	Fibrodysplasia ossificans progressiva (135100)	ACVR1 point mutation C617 G-A		
FLT3/NPM1	1395		Lavender Top (EDTA). Bone marrow. Stable 24-48 h at RT after draw. Do not freeze.	Dept of Clinical Laboratory Genetics, Genome Diagnostics, Toronto General Hospital.		RNA		10-May-22
Fluconazole, Serum or Plasma [2089SP]	1118		Serum: Collect sample in Red top tube Plasma: Collect sample in Lavender top tube (EDTA)	NMS Laboratories		Liquid Chromatography/Tandem Mass Spectrometry (LC/MS/MS)		09-Jun-22

Appellation 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)	SYMBOLIÈRE DU GÈNE et NOM OMIM DU GÈNE / HGNC Gene Symbol and OMIM Gene Number	RÈGLES DE MANUTENTION / Handling Conditions	LABORATOIRE DÉSIGNÉ / Designated laboratory	RENSEIGNEMENTS SUR L'ANALYSE / Test details	MÉTHODE / Methode	NOTE PARTICULIÈRE / Special comment	Date de la révision / Date of revision
I,5-anhydroglucitol (GlycoMark) [500115] [GLYC]	1465		Gold Top or Red Top (Serum) or Lavender (plasma). Centrifuge and aliquot serum to transport tube within one hour of collection. Store and ship frozen. Stable at -20°C for 14 days.	LabCorp/Dynacare (https://www.labcorp.com/)		Enzymatic, colorimetric assay.		10-Jan-21
Fluphenazine (Prolixin), Serum [PROLX]	929		Serum Draw blood in a plain red-top tube(s), serum gel tube is not acceptable. Spin down and send 3 mL of serum refrigerated in a plastic vial.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	Moderate	Liquid Chromatography/Tandem Mass Spectrometry (LC/MS/MS)		26-Jan-20
FSHD - Detection of Abnormal Alleles with Interpretation (FSHD1 and FSHD2)	102	DUX4 (D4Z4) (606009); SMCHD1 (614982)	Lavender Top (EDTA). Rejection: Specimens collected more than 5 days before receipt by the laboratory.	University of Iowa Diagnostic Laboratories (http://www.healthcare.uiowa.edu/path_handbook/rhandbook/test127.html)	Facioscapulohumeral Muscular Dystrophy 1 (158900); Facioscapulohumeral Muscular Dystrophy 2 (158901)	Southern blot, 4qA.4qVhaplotyping; methylation; SMCHD1 sequencing		13-May-19
Fumarate deficiency (606812) [713]	1034	FH (136850)	Lavender Top (EDTA)	GeneDx (www.genedx.com)	Fumarate hydratase; Includes: Hereditary Leiomyomatosis and Renal cell Cancer (HLRCC) (150800)	Sanger sequencing and del/dupl		09-Dec-19
FUNGITEL BETA-D -Glucan Colorimetric Assay [317]	489		Serum: Collect serum specimens in serum separator or red top tube. Allow blood to clot for 30 minutes, then centrifuge. Pipette serum into a transport tube without interfering levels of (1→3)-β-D-Glucan. Most sterile polypropylene DNAase and RNase free tubes are acceptable. CSF: Submit CSF in a sterile screw cap container. STABILITY: Room Temperature: Not acceptable. Refrigerated: 5 days Frozen: Indefinitely	MiraVista Diagnostics	Elevated in fungal infection	Colorimetric Fungitell® assay utilizing a (1→3)-β-D-Glucan -specific Limulus Amebocyte Lysate (LAL)	No longer performed in province. AH-612 must be approved by Microbiology.	08-Sep-22
Fungitell, Serum [FUNGS]	489		Collect 3-5 mL blood in a serum separator gel tube (SST). Centrifuge specimen within 2 hours. Ship serum gel tube frozen.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)		Replaced by: 1,3-Beta-D-Glucan (Fungitell), Serum		20-Jan-20
Gabapentin [GABA]	107		Red Top. Store and send frozen.	In-Common Laboratories		LC/UV		06-Sep-18
Galactose-alpha-1,3-galactose (Alpha-Gal) IgE [ALGAL]	1761		Collect RED or GOLD SST. Store at 4°C for 14 days or at -20°C (90 days).	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	Phadia ImmunoCAP	Phadia ImmunoCAP	Available at ICL \$120	19-Aug-21
GARS Full Gene Sequencing Analysis [MOL167]		GARS (60287)	Lavender Top (EDTA)	MNG Laboratories (www.medicalneurogenetics.com)/Dynacare	Charcot-Marie-Tooth disease, type 2D (601427); Neuropathy, distal hereditary motor, type V (607541)	Sanger sequencing		09-May-20
Gaucher Disease (recurrent mutations)		GBA (60646)	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 Neurodegenerative Gaucher's Disease (230900); Gaucher Disease, Type IIIc (231005)	Direct mutation analysis (9 mutations); AKJ 90% sensitivity; others 50-60%		
GAUCHER DISEASE VIA THE GBA GENE [479]	464	GBA (60646)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Gaucher's Disease, Type 1 (230800); Gaucher Disease, Perinatal Lethal (608013); Subacute Neurodegenerative Gaucher's Disease (230900); Gaucher Disease, Type IIIc (231005)	Sanger sequencing		28-Jan-20
Gene dosage for FGFR2, FGFR3 & TWIST		FGFR2 (179943); FGFR3 (134934); TWIST1 (601622)	Lavender Top (EDTA)	Molecular Genetics Laboratory, Hospital for Sick Children (www.sickkids.ca/PaediatricLaboratoryMedicine/Laboratories-Services/Molecular-Genetics-Laboratory/index.html)	Craniosynostosis	del/dupl		
Genetic Eye Disease Panel for Strabismus (Gedi-S)		ROBO3, PHOX2A, HESX1, SALL4, CHN1, TUBB3, KIF21A, HESX1	Lavender Top (EDTA) 2-5 cc	Ocular genomics (https://oculargenomics.meei.harvard.edu/index.php/gdt/)	Strabismus	NextGen Sequencing		
Genetic Renal Panel	1285	CFH (134700), CF (217030), MCP (120920), CFH (138470), CFHR5 (608591), C3 (120700), THBD (188040), ADAMTS1 (604136), BDKFB (601440), PLAG1 (173308), CFHR3-CFHR1 (605336/13471)	Lavender Top (EDTA)	Molecular Otolaryngology & Renal Research Laboratory, U. of Iowa Carver College of Medicine (www.medicine.uiowa.edu/mor/)	thrombotic microangiopathies; Hemolytic Uremic Syndrome, atypical Hemolytic Uremic Syndrome and Thrombotic Thrombocytopenic Purpura	Next-Generation sequencing; MLPA		23-Mar-20
GH-RH			Serum or EDTA plasma	InterScience Institute				
GLA gene dosage	469	GLA (30664)	Lavender top (EDTA); Store at RT or 4°C for up to 48 h after drawing. At 4°C for >48 h.	Molecular Genetics Laboratory, Hospital for Sick Children (www.sickkids.ca/PaediatricLaboratoryMedicine/Laboratories-Services/Molecular-Genetics-Laboratory/index.html)	Fabry Disease (301500)	MLPA		
GLA gene sequencing	469	GLA (30664)	Lavender top (EDTA); Store at RT or 4°C for up to 48 h after drawing. At 4°C for >48 h.	Molecular Genetics Laboratory, Hospital for Sick Children (www.sickkids.ca/PaediatricLaboratoryMedicine/Laboratories-Services/Molecular-Genetics-Laboratory/index.html)	Fabry Disease (301500)	Sanger sequencing		

Appellation 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 (Déclencheur MSSS)	SYMBOL HUGO DU GÈNE et NOM OMDM DU GÈNE / HUGO Gene Symbol and OMDM Gene Name	RÈGLES DE MANUTENTION / Handling Conditions	LABORATOIRE DÉSIGNÉ / Designated laboratory	RENSEIGNEMENTS SUR L'ANALYSE / Test details	MÉTHODE / Methode	NOTE PARTICULIÈRE / Special comment	Date de la révision / Date of revision
1,5-anhydroglucitol (GlycoMark) [500115] [GLYC]	1465		Gold Top or Red Top (Serum) or Lavender (plasma). Centrifuge and aliquot serum to transport tube within one hour of collection. Store and ship frozen. Stable at -20°C for 14 days.	LabCorp/Dynacare (https://www.labcorp.com/)		Enzymatic, colorimetric assay.		10-Jan-21
Glucagon, Plasma [GLP]	110		Collection Container/Tube: Lavender top (EDTA) Submission Container/Tube: Plastic vial Specimen Volume: 2 mL Collection Instructions: 1. Fasting. 2. Prechill tube at 4 degrees C before drawing the specimen. 3. Draw the prechilled tube, and process as follows: a. After drawing specimen, chill tube in wet ice for 10 minutes. b. Centrifuge in a refrigerated centrifuge or in chilled centrifuge cup. c. Immediately after centrifugation, remove plasma, place in a plastic transport vial (Supply T465), and freeze.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)		RIA		17-Mar-20
GLUCOSE TRANSPORTER TYPE 1 DEFICIENCY SYNDROME (06777)		SLC2A1 (138140)	Lavender Top (EDTA)	BC Children's Hospital & BC Women's Hospital, Canada (http://www.genebc.ca)		Sanger sequencing & reflex MLPA		19-Oct-17
GLUCOSE-6-PHOSPHATE DEHYDROGENASE DEFICIENCY VIA THE G6PD GENE		G6PD (095900)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Hemolytic anemia due to G6PD deficiency (300908)	Sanger sequencing		
GLUT1 Deficiency Syndrome (SLC2A1 Single Gene Test)	262	SLC2A1 (138140)	Lavender Top (EDTA) [2 tubes]	Fulgent Genetics (fulgentdiagnostics.com)		NextGen Sequencing		28-Feb-20
Glutamate Receptor R1							NOT AVAILABLE: Wash U, Mayo, Oxford	21-Apr-17
Glycogen Storage Disease and Disorders of Glucose Metabolism Sequencing Panel with CNV Detection [10385]	476	AGL 610860 ALDOA 101890 ALDOB 612724 ENOG 131370 G6PC 613742 G6A 608080 GBE1 607839 GVI 603942 GYS1 138570 GYS2 138571 LAMP2 399000 LDBA 159000 PC 608786 PCK1 614168 PCK2 614095 PFKM 610081 PGAM2 612931 PFMI 171900 PHKA1 311870 PHKA2 300798 PHKB 172090 PHKG2 172471 PKRAG2 602743 PYGL 613751 PYGM 608455 SLC16A1 600082 SLC2A2 138160 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 IXa (300559); Glycogen Storage Disease Type IXb (306000); Glycogen Storage Disease Type IXc (261750); Glycogen Storage Disease Type IXd (613027); Glycogen Storage Disease Type VI (232700); Glycogen Storage Disease Type V (232600); Glycogen Storage Disease Type Ib (232200); Glycogen Storage Disease Type Ic (232240); Fanconi-Bickel Syndrome (227810)	Next Generation Sequencing (NGS) and Sanger sequencing technologies		15-Aug-19
GLYCOGEN STORAGE DISEASE TYPE III VIA THE AGL GENE [224]	476	AGL (610860)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Glycogen Storage Disease Type III (GSDIII) (232400)	Sanger sequencing		
GLYCOGEN STORAGE DISEASE TYPE IV VIA THE GBE1 GENE [225]	476	GBE1 (607839)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Glycogen Storage Disease Type IV (232500)	Sanger sequencing		
Goose Feather (e70) IgE, Serum	1419		Gold SST	Check if available at CHUM				07-Oct-19
Granulocyte Ab, Serum [LAGGT]	1836		Gold SST or Red Top	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)				22-Mar-22
GRHRP Gene, Full Gene Analysis [GRHMS]		GRHRP (604296)	Lavender top (EDTA)	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	Hyperoxaluria, primary, type II (296000)	Sanger sequencing and MLPA		
Growth Hormone Releasing Hormone (GH-RH)			3 ml serum (Red or Gold Top) or EDTA plasma should be collected and separated as soon as possible. Freeze the plasma immediately after separation. Minimum specimen size is 1 ml.	InterScience Institute				
Haloperidol, Serum/Plasma	115		Red Top or Li heparin (light Green Top)	In-Common Laboratories		LC-MS		09-Sep-21
Hearing Loss: Branchio-Oto-Renal (BOR) Syndrome (115630)		EYA1 (601651)	Lavender Top (EDTA)	Molecular Genetics Laboratory, Hospital for Sick Children (www.sickkids.ca/PaediatricLaboratoryMedicine/Laboratories-Services/Molecular-Genetics-Laboratory/index.html)		1. EYA1 gene sequencing 2. EYA1 gene dosage		
Hearing Loss: Non-Syndromic (Connexin 26 & 30)		GJB2 (121011); GJB6 (604418)	Lavender Top (EDTA)	Molecular Genetics Laboratory, Hospital for Sick Children (www.sickkids.ca/PaediatricLaboratoryMedicine/Laboratories-Services/Molecular-Genetics-Laboratory/index.html)		1. GJB2 sequencing and GJB6 deletion 2. GJB2 sequencing only		

Appellation 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 (Déclencheur MSSS)	SYMBOL/IGENE DU GÈNE et NOM OMDM DU GÈNE / IGENE Gene Symbol and OMDM Gene Name	RÈGLES DE MANUTENTION / Handling Conditions	LABORATOIRE DÉSIGNÉ / Designated laboratory	RENSEIGNEMENTS SUR L'ANALYSE / Test details	MÉTHODE / Methode	NOTE PARTICULIÈRE / Special comment	Date de la révision / Date of revision
1,5-anhydroglucitol (GlycoMark) [500115] [GLYC]	1465		Gold Top or Red Top (Serum) or Lavender (plasma). Centrifuge and aliquot serum to transport tube within one hour of collection. Store and ship frozen. Stable at -20°C for 14 days.	LabCorp/Dynacare (https://www.labcorp.com/)		Enzymatic, colorimetric assay.		10-Jan-21
Heparin Induced Thrombocytopenia Testing [HIT]	116		1) RED TOP: 4 mL. Draw blood into red-top vacutainer and allow to clot. Centrifuge and transfer serum to two plastic tube. 2) Freeze promptly. Ship frozen	Platelet Immunology Laboratory, McMaster University		HIT Confirmatory Test (Serotonin Release Assay; SRA) \$300 and HIT Screen test (Anti-PF4-heparin EIA) \$130		31-Oct-19
HEREDITARY DIFFUSE GASTRIC CANCER (137215) VIA THE CDH1 GENE [798]		CDH1 (192990)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)				24-Oct-17
Hereditary Erythrocytosis Mutations [HEMP]		EPOR (133171), VHL (608557), EGLN1 (618D2) (696245), EPAS1 (HIF2A) (603349)	Lavender Top (EDTA)	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	Erythrocytosis, familial, 1 (133140); Erythrocytosis, familial, 2 (263400); Erythrocytosis, familial, 3 (609820); Erythrocytosis, familial, 4 (611783)	Sanger sequencing. Panel: EPOR: 133171, EGLN1: 606425, EPAS1: 603349, As reflex VHL: 608537		20-Oct-20
HEREDITARY HEMOCHROMATOSIS PANEL [10243]	1655	FTH1 (134700); FTL (134700); HAMP (606664); HFE (613609); HJV (608374); SLC40A1 (60660); TFR2 (604720)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Hemochromatosis Type 5 (615517); Hyperferritinemia Cataract Syndrome (600886); Hemochromatosis Type 4 (606609); Hemochromatosis Type 2 (602390); Hemochromatosis Type 2B (613313); Hemochromatosis Type 3 (604250); Hemochromatosis Type 1 (235200)	Next-Generation sequencing		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)	Sanger sequencing and MLPA		
Hereditary Hemorrhagic Telangiectasia (HHT), type 1 (187300) and type 2 (600376)	242	ENG (131195); ACVRL1 (ALK1) (601284)	Lavender Top (EDTA)	Molecular Genetics Laboratory, Hospital for Sick Children (www.sickkids.ca/PaediatricLaboratoryMedicine/Laboratories-Services/Molecular-Genetics-Laboratory/index.html)		1. ACVRL1 gene sequencing 2. ENG gene sequencing 3. SMAD4 gene sequencing 4. ACVRL1 and ENG gene dosage		
Hereditary Melanoma and Skin Cancer Panel [ON0501]	243		Lavender Top (EDTA)	Blueprint Genetics http://blueprintgenetics.com/		NextGen Sequencing		15-Dec-20
Hereditary Neuropathy Sequencing & Del/Dup Panel [737]	656	AARS, ATLI, ATP9A, BSCL2, DNABP2, DNMT2, DNMT3, DYNC1H1, EGR2, FAM137B, FGD4, FGA, GAN, GARS, GDAP1, GIB1, GLA, HINT1, HSPH1, HSPH9, IGHMBP2, IKBKAP, INF2, KIF1A, KIF5A, LITAF, LMNA, LRSM1, MFN2, MPZ, MTMR2, NDRG1, NERF1, NFR, NTRK1, PLEKHA7, PMP22, PRR5L, PRR4, RAB7A, RIEEP1 (CROB2), SH3BP2, SECNA, SH3TC2, SLC12A6, SLC25A2, SPTLC1, SPTLC2, TRG, TRPV4, TRK, WNK1 (exon 10 only), YARS	Lavender Top (EDTA)	GeneDx		NGS		31-May-22
Hereditary Spastic Paraplegia: Autosomal Dominant [HSP-Panel 1]	657	ALDH3A1, ATLI, BSCL2, C10orf2, C10orf2, HSPH1, KIAA0906, KIF5A, NIPA1, POLG, POLG2, RIEEP1, SPAST, RPN2, SLC31A1, SETX	Lavender Top (EDTA)	Molecular Genetics Laboratory, Hospital for Sick Children (www.sickkids.ca/PaediatricLaboratoryMedicine/Laboratories-Services/Molecular-Genetics-Laboratory/index.html)		NextGen Sequencing	Price of individual genes: \$500 (seq) + 500\$ (del/dupl)	21-Aug-19

Appellation 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 (Déclinaire MSSS)	SYMBOL / IGENC DU GÈNE et NOM OMIM DU GÈNE / IGENC Gene Symbol and OMIM Gene Number	RÈGLES DE MANUTENTION / Handling Conditions	LABORATOIRE DÉSIGNÉ / Designated laboratory	RENSEIGNEMENTS SUR L'ANALYSE / Test details	MÉTHODE / Methode	NOTE PARTICULIÈRE / Special comment	Date de la révision / Date of revision
1,5-anhydroglucitol (GlycoMark) [500115] [GLYC]	1465		Gold Top or Red Top (Serum) or Lavender (plasma). Centrifuge and aliquot serum to transport tube within one hour of collection. Store and ship frozen. Stable at -20°C for 14 days.	LabCorp/Dynacare (https://www.labcorp.com/)		Enzymatic, colorimetric assay.		10-Jan-21
Hereditary Spastic Paraplegia: Autosomal Recessive [HSP-Panel 2]	657	ALDH18A1 ALS2 AP4B1 AP4E1 AP4M1 AP4S1 AP5Z1 C10orf2 C12orf65 C19orf12 CYP2U1 CYP7B1 DDHD1 DDHD2 ENTPD1 ERLIN1 ERLIN2 FAL2H GBA2 GJC2 KIF1A KIF1C KIF1E KIF2C NTSC2 PGAP1 PNPLA6 POLG SACS SPG11 SPG12 SPG20 SPG21 SPG7 TECP2 VPS37A ZFYV226	Lavender Top (EDTA)	Molecular Genetics Laboratory, Hospital for Sick Children (www.sickkids.ca/PaediatricLaboratoryMedicine/Laboratories-Services/Molecular-Genetics-Laboratory/index.html)		NextGen Sequencing	Price of individual genes: \$500 (seq) + 500\$ (del/dupl)	21-Aug-19
Hereditary Spastic Paraplegia: Comprehensive Testing [HSP-COMP]	657	ALDH18A1 ATL1 BSC1L C10orf2 HSPD1 KIAA0906 KIF5A NIPA1 POLG POLG2 REEP1 SPAST RTN2 SLC35A1 SETX ALS2 AP4B1 AP4E1 AP4M1 AP4S1 AP5Z1 C10orf2 C12orf65 C19orf12 CYP2U1 CYP7B1 DDHD1 DDHD2 ENTPD1 ERLIN1 ERLIN2 FAL2H GBA2 GJC2 KIF1A KIF1C KIF1E KIF2C PGAP1 PNPLA6 POLG SACS SPG11 SPG12 SPG20 SPG21 SPG7 TECP2 VPS37A ZFYV226 LICAM PLP1 SLC18A2	Lavender Top (EDTA)	Molecular Genetics Laboratory, Hospital for Sick Children (www.sickkids.ca/PaediatricLaboratoryMedicine/Laboratories-Services/Molecular-Genetics-Laboratory/index.html)		NextGen Sequencing	Price of individual genes: \$500 (seq) + 500\$ (del/dupl)	21-Aug-19
Hereditary Spastic Paraplegia: Deletion & Duplication Analysis [HSP-DOSAGE]	657		Lavender Top (EDTA)	Molecular Genetics Laboratory, Hospital for Sick Children (www.sickkids.ca/PaediatricLaboratoryMedicine/Laboratories-Services/Molecular-Genetics-Laboratory/index.html)		Deletion & Duplication analysis by exon targeted microarray		21-Aug-19
Heterotaxy and Situs Inversus NGS Panel	385	CVR2B, CCD39, CCD40, CFC1, DNAAF1, DNAAF2, DNAAF3, DNAH11, DNARS, DNAI1, DNAL2, DNAL1, FOXP1, GDF1, INVS, LEFTY2, NKX2-5, NMB, NODAL, ZIC3	Lavender Top (2 x 4 mL)	Fulgent Genetics (fulgentdiagnostics.com)		NextGen Sequencing		

Appellation 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 (Déclaratoire MSSS)	SYMBOL/BIEN DE GÈNE et NOM OMIM DU GÈNE / HGNC Gene Symbol and OMIM Gene Number	RÈGLES DE MANUTENTION / Handling Conditions	LABORATOIRE DÉSIGNÉ / Designated laboratory	RENSEIGNEMENTS SUR L'ANALYSE / Test details	MÉTHODE / Methode	NOTE PARTICULIÈRE / Special comment	Date de la révision / Date of revision
1,5-anhydroglucitol (GlycoMark) [500115] [GLYC]	1465		Gold Top or Red Top (Serum) or Lavender (plasma). Centrifuge and aliquot serum to transport tube within one hour of collection. Store and ship frozen. Stable at -20°C for 14 days.	LabCorp/Dynacare (https://www.labcorp.com/)		Enzymatic, colorimetric assay.		10-Jan-21
Histamine, 24-Hour Urine [FH24U]	117		4 mL urine from a 24-hour collection containing 10 mL 6N HCl; Alenate; No preservative. Specimen Stability: Room temperature: 48 hours, Refrigerated: 14 days, Frozen: 14 days. Patient should refrain from taking allergy causing drugs, antihistamines, oral corticosteroids, and substances which block H2 receptors for at least 24 hours prior to specimen collection.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	carcinoid	Immunoassay		13-Jul-21
Histamine, Plasma [FHSPL]	117		Draw 3 mL blood in a lavender-top (EDTA) tube(s). Cool immediately on ice. Centrifuge at 1500 rpm for 10 minutes at 4 degrees C. The centrifugation should be performed within 20 minutes of collection. Carefully remove 1 mL of EDTA plasma from the upper part of the tube. Freeze and send frozen in a plastic vial.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)		EIA		04-Apr-22
Histoplasma Antibody IgG, IgM, EIA [326]	396		Serum: Collect serum specimens in serum separator or red top tube. Allow blood to clot for 30 minutes, then centrifuge. Pipette serum into a plastic screw cap vial. CSF: Sterile transport tube. STABILITY Room Temperature: 14 days Refrigerated: 14 days Frozen: 14 days	MiraVista Diagnostics				08-Sep-22
Histoplasma Antigen Quantitative EIA [310]	396		Serum: Collect serum specimens in serum separator or red top tube. Allow blood to clot for 30 minutes, then centrifuge. Pipette serum into a plastic screw cap vial. Plasma: Collect plasma specimens in an EDTA, heparin or sodium citrate tube. Centrifuge for 15 minutes and pipette plasma into a plastic screw cap vial. Urine/CSF/BAL/Other Body Fluid: Submit urine, CSF, BAL and all other body fluids in a sterile screw cap container. STABILITY Room Temperature: 2 weeks Refrigerated: 2 weeks Frozen: Indefinitely	MiraVista Diagnostics				08-Sep-22
Histoplasma Serology			Gold SST	Alberta Provincial Laboratory				
Histoplasma Antibody by Immunodiffusion [321]	396		Serum: Collect serum specimens in serum separator or red top tube. Allow blood to clot for 30 minutes, then centrifuge. Pipette serum into a plastic screw cap vial. STABILITY Refrigerated: 14 days Frozen: 6 months	MiraVista Diagnostics				08-Sep-22
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)		May be done locally.	
Homocystinuria [1563]		CBS (613381)	Lavender top (EDTA)	Connective Tissue Gene Tests (www.ctgt.net)	Homocystinuria, B6-responsive and nonresponsive types (236200)	1.Sanger sequencing 2.HD array		
HTRA1 DNA Sequencing Test (CARASIL) [442]		HTRA1 (602194)	Lavender Top (EDTA)	Athena Diagnostics (www.athenadiagnostics.com)	CARASIL (600142)	Sanger sequencing		
Hydroxychloroquine, Serum [HCQ]	1689		Red Top ONLY. Store and send serum at 4°C or frozen (stable 28 d).	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)		LC-MS/MS		20-Oct-21
Hyper IgE Syndrome Panel [1969]	1258	DOCK8 (614443), SPINK5 (605810), 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); Neuhart syndrome (256500); Tyrosine kinase 2 deficiency (611521)	Exon Array CGH, Next-gen Sequencing		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)	Sanger sequencing		24-Oct-17
Hyperglycosylated hCG			Gold SST	Quest Laboratories	First trimester screen for Down syndrome and trisomy 18			
Hyperlipidemia Panel Plus Analysis [CA1101]	1533	ABCA1 (600446), ABCG5 (605459), ABCG8 (603076), APOA1 (107680), APOB (143890), APOC3 (107720), APOE (107741), LDLR (144010), LDLRAP1 (91479), LPL (609706), PCSK9 (603770)	Lavender Top (EDTA)	Blueprint Genetics http://blueprintgenetics.com/	Familial Hypercholesterolemia (143890); Hypercholesterolemia, Autosomal Dominant, Type B (144010); Hypercholesterolemia, Autosomal Dominant, 3 (603776); Hypercholesterolemia, Autosomal Recessive (603813)	Next Generation DNA Sequencing + Del/Dupl	Family Member testing \$450	16-Jun-17

Appellation 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 (Déclencheur MSSS)	SYMBOL / IGENC DU GÈNE et NOM OMDM DU GÈNE / IGENC Gene Symbol and OMDM Gene Name	RÈGLES DE MANUTENTION / Handling Conditions	LABORATOIRE DÉSIGNÉ / Designated laboratory	RENSEIGNEMENTS SUR L'ANALYSE / Test details	MÉTHODE / Methode	NOTE PARTICULIÈRE / Special comment	Date de la révision / Date of revision	
1,5-anhydroglucitol (GlycoMark) [500115] [GLYC]	1465		Gold Top or Red Top (Serum) or Lavender (plasma). Centrifuge and aliquot serum to transport tube within one hour of collection. Store and ship frozen. Stable at -20°C for 14 days.	LabCorp/Dynacare (https://www.labcorp.com/)		Enzymatic, colorimetric assay.		10-Jan-21	
Hypertrophic Cardiomyopathy NGS Panel	287	ABCC9, ACTC1, ACTN2, ANKRD1, BAG1, BRAF, CAV3, CRYAB, CSRP1, DES, DMD, DSC2, DSP, DTNA, EMD, FKTN, GATAD1, GLA, HRAA, ILK, JPH2, JUP, KRAS, LAMA4, LAMP2, LDB1, LMNA, MAP2K1, MAP2K2, MYBP1, MYH7, MYL2, MYL3, MYL4, MYOZ2, MYOZ3, MYOZ4, MYOZ5, NRAS, PDGfra, PKP2, PLN, PRKAG2, PTPN11, RAS, RBM20, RYR2, SCN5A, SOD3, SOST, TAZ, TCF4, TM6AS3, TMPO, TNNC1, TNNT3, TNNT2, TPM1, TRN, TRN, VCL	Lavender Top (2 x 4 mL)	Fulgent Genetics (fulgentdiagnostics.com)		NextGen Sequencing (24 genes)		11-Nov-19	
Hypokalemic and Hyperkalemic Periodic Paralysis Disorders (NextGen Sequencing Panel and Copy Number Analysis; 7 Genes) [NGS332]	591	AIP (605555), AMMECR1 (800195), CACNA1S (114208), CLCN1 (118425), KCNJ2 (600681), SCN4A (609667), SLC12A3 (600698)	Lavender Top (EDTA)	MNG Laboratories (www.medicalneurogenetics.com/)/Dynacare		NextGen Sequencing		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)		NextGen Sequencing	Custom Panel; specific both genes		
Hypomyelinating Leukodystrophy 9 (616140)	964	RARS (107820)	Lavender Top (2 x 4 mL)	Alfred I Dupont Laboratory http://www.nemours.org/pediatric-research/labservices/diagnostic/molecular-diagnostics-lab.html					
Hypomyelinating Leukodystrophy With Atrophy of the Basal Ganglia and Cerebellum (H ABC)	964	TUBB4A (602662)	Lavender Top (2 x 4 mL)	Alfred I Dupont Laboratory http://www.nemours.org/pediatric-research/labservices/diagnostic/molecular-diagnostics-lab.html	Dystonia 4, torsion, autosomal dominant (128101); Leukodystrophy, hypomyelinating, 6 (612438)				
HYPOMYELINATION AND CONGENITAL CATARACT (HCC) VIA THE FAM126A GENE [1669]		FAM126A (610531)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Hypomyelination And Congenital Cataract (610532)	Sanger sequencing			
Hypophosphatasia via the ALPL Gene [851]		ALPL (171760)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Adult Hypophosphatasia (146300); Childhood Hypophosphatasia (241510); Infantile Hypophosphatasia (241500)			11-Jun-18	
IA-2 Antibody [37933]	35		Gold SST or Red Top. Specimen Stability - Room temperature: 7 days Refrigerated: 7 days Frozen: 30 days	In-Common Laboratories	anti-tyrosine phosphatase or anti-Ikbt antibody	ELISA		19-Jun-19	
IBD sgi Diagnostic [1800]	120		2.0 mL Serum (Red Top or SST) AND 2.0 mL Whole Blood EDTA / Lavender Top Tube. Store at 4°C. Collect	Prometheus Biosciences	differentiate among IBD types				
Ibuprofen (Motrin, Advil, Nuprin), serum [FIBUP]			Plain Red. Also acceptable: Green top. NO GEL. Specimen Preparation Separate from cells. Transfer 1 mL serum or plasma to plastic vial. Store frozen.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	Optimize drug therapy and monitor patient adherence.	HPLC-UV		20-Nov-17	
Idiopathic Ataxia anti MPP-1	1465		Gold SST or Red Top	Mitogen Advanced Diagnostics				02-Jun-21	
Idiopathic Generalized Epilepsy Panel	1031	ADSL, ALDH1A1, ARHGAP9, ARX, ATP6AP2, ATRX, CACNA1A, CACNA1H, CACNA1C, CASK, CASR, CDKL5, CHD2, CHRNA2, CHRNA4, CHRN2B2, CLDN2, CNTN2, CNTNAP2, CPAA, CSTB, CUL4B, DCX, DEAF1C, DMR1, DNAAF3, DYNC1H1, EFHC1, EPMA3, FGF1, FGF3, GABRA1, GABRB3, GABRD, GABRG2, GOSR2, GPC3, GRIA3, GRIN2A, HSD17B10, KANSL1, KIF1A, KIF20B, KIF20A1, KIF20B, KIF20C, KIF20D, KIF20E, KIF20F, KIF20G, KIF20H, KIF20I, KIF20J, KIF20K, KIF20L, KIF20M, KIF20N, KIF20O, KIF20P, KIF20Q, KIF20R, KIF20S, KIF20T, KIF20U, KIF20V, KIF20W, KIF20X, KIF20Y, KIF20Z, KIF21, KIF22, KIF23, KIF24, KIF25, KIF26, KIF27, KIF28, KIF29, KIF30, KIF31, KIF32, KIF33, KIF34, KIF35, KIF36, KIF37, KIF38, KIF39, KIF40, KIF41, KIF42, KIF43, KIF44, KIF45, KIF46, KIF47, KIF48, KIF49, KIF50, KIF51, KIF52, KIF53, KIF54, KIF55, KIF56, KIF57, KIF58, KIF59, KIF60, KIF61, KIF62, KIF63, KIF64, KIF65, KIF66, KIF67, KIF68, KIF69, KIF70, KIF71, KIF72, KIF73, KIF74, KIF75, KIF76, KIF77, KIF78, KIF79, KIF80, KIF81, KIF82, KIF83, KIF84, KIF85, KIF86, KIF87, KIF88, KIF89, KIF90, KIF91, KIF92, KIF93, KIF94, KIF95, KIF96, KIF97, KIF98, KIF99, KIF100, KIF101, KIF102, KIF103, KIF104, KIF105, KIF106, KIF107, KIF108, KIF109, KIF110, KIF111, KIF112, KIF113, KIF114, KIF115, KIF116, KIF117, KIF118, KIF119, KIF120, KIF121, KIF122, KIF123, KIF124, KIF125, KIF126, KIF127, KIF128, KIF129, KIF130, KIF131, KIF132, KIF133, KIF134, KIF135, KIF136, KIF137, KIF138, KIF139, KIF140, KIF141, KIF142, KIF143, KIF144, KIF145, KIF146, KIF147, KIF148, KIF149, KIF150, KIF151, KIF152, KIF153, KIF154, KIF155, KIF156, KIF157, KIF158, KIF159, KIF160, KIF161, KIF162, KIF163, KIF164, KIF165, KIF166, KIF167, KIF168, KIF169, KIF170, KIF171, KIF172, KIF173, KIF174, KIF175, KIF176, KIF177, KIF178, KIF179, KIF180, KIF181, KIF182, KIF183, KIF184, KIF185, KIF186, KIF187, KIF188, KIF189, KIF190, KIF191, KIF192, KIF193, KIF194, KIF195, KIF196, KIF197, KIF198, KIF199, KIF200, KIF201, KIF202, KIF203, KIF204, KIF205, KIF206, KIF207, KIF208, KIF209, KIF210, KIF211, KIF212, KIF213, KIF214, KIF215, KIF216, KIF217, KIF218, KIF219, KIF220, KIF221, KIF222, KIF223, KIF224, KIF225, KIF226, KIF227, KIF228, KIF229, KIF230, KIF231, KIF232, KIF233, KIF234, KIF235, KIF236, KIF237, KIF238, KIF239, KIF240, KIF241, KIF242, KIF243, KIF244, KIF245, KIF246, KIF247, KIF248, KIF249, KIF250, KIF251, KIF252, KIF253, KIF254, KIF255, KIF256, KIF257, KIF258, KIF259, KIF260, KIF261, KIF262, KIF263, KIF264, KIF265, KIF266, KIF267, KIF268, KIF269, KIF270, KIF271, KIF272, KIF273, KIF274, KIF275, KIF276, KIF277, KIF278, KIF279, KIF280, KIF281, KIF282, KIF283, KIF284, KIF285, KIF286, KIF287, KIF288, KIF289, KIF290, KIF291, KIF292, KIF293, KIF294, KIF295, KIF296, KIF297, KIF298, KIF299, KIF300, KIF301, KIF302, KIF303, KIF304, KIF305, KIF306, KIF307, KIF308, KIF309, KIF310, KIF311, KIF312, KIF313, KIF314, KIF315, KIF316, KIF317, KIF318, KIF319, KIF320, KIF321, KIF322, KIF323, KIF324, KIF325, KIF326, KIF327, KIF328, KIF329, KIF330, KIF331, KIF332, KIF333, KIF334, KIF335, KIF336, KIF337, KIF338, KIF339, KIF340, KIF341, KIF342, KIF343, KIF344, KIF345, KIF346, KIF347, KIF348, KIF349, KIF350, KIF351, KIF352, KIF353, KIF354, KIF355, KIF356, KIF357, KIF358, KIF359, KIF360, KIF361, KIF362, KIF363, KIF364, KIF365, KIF366, KIF367, KIF368, KIF369, KIF370, KIF371, KIF372, KIF373, KIF374, KIF375, KIF376, KIF377, KIF378, KIF379, KIF380, KIF381, KIF382, KIF383, KIF384, KIF385, KIF386, KIF387, KIF388, KIF389, KIF390, KIF391, KIF392, KIF393, KIF394, KIF395, KIF396, KIF397, KIF398, KIF399, KIF400, KIF401, KIF402, KIF403, KIF404, KIF405, KIF406, KIF407, KIF408, KIF409, KIF410, KIF411, KIF412, KIF413, KIF414, KIF415, KIF416, KIF417, KIF418, KIF419, KIF420, KIF421, KIF422, KIF423, KIF424, KIF425, KIF426, KIF427, KIF428, KIF429, KIF430, KIF431, KIF432, KIF433, KIF434, KIF435, KIF436, KIF437, KIF438, KIF439, KIF440, KIF441, KIF442, KIF443, KIF444, KIF445, KIF446, KIF447, KIF448, KIF449, KIF450, KIF451, KIF452, KIF453, KIF454, KIF455, KIF456, KIF457, KIF458, KIF459, KIF460, KIF461, KIF462, KIF463, KIF464, KIF465, KIF466, KIF467, KIF468, KIF469, KIF470, KIF471, KIF472, KIF473, KIF474, KIF475, KIF476, KIF477, KIF478, KIF479, KIF480, KIF481, KIF482, KIF483, KIF484, KIF485, KIF486, KIF487, KIF488, KIF489, KIF490, KIF491, KIF492, KIF493, KIF494, KIF495, KIF496, KIF497, KIF498, KIF499, KIF500, KIF501, KIF502, KIF503, KIF504, KIF505, KIF506, KIF507, KIF508, KIF509, KIF510, KIF511, KIF512, KIF513, KIF514, KIF515, KIF516, KIF517, KIF518, KIF519, KIF520, KIF521, KIF522, KIF523, KIF524, KIF525, KIF526, KIF527, KIF528, KIF529, KIF530, KIF531, KIF532, KIF533, KIF534, KIF535, KIF536, KIF537, KIF538, KIF539, KIF540, KIF541, KIF542, KIF543, KIF544, KIF545, KIF546, KIF547, KIF548, KIF549, KIF550, KIF551, KIF552, KIF553, KIF554, KIF555, KIF556, KIF557, KIF558, KIF559, KIF560, KIF561, KIF562, KIF563, KIF564, KIF565, KIF566, KIF567, KIF568, KIF569, KIF570, KIF571, KIF572, KIF573, KIF574, KIF575, KIF576, KIF577, KIF578, KIF579, KIF580, KIF581, KIF582, KIF583, KIF584, KIF585, KIF586, KIF587, KIF588, KIF589, KIF590, KIF591, KIF592, KIF593, KIF594, KIF595, KIF596, KIF597, KIF598, KIF599, KIF600, KIF601, KIF602, KIF603, KIF604, KIF605, KIF606, KIF607, KIF608, KIF609, KIF610, KIF611, KIF612, KIF613, KIF614, KIF615, KIF616, KIF617, KIF618, KIF619, KIF620, KIF621, KIF622, KIF623, KIF624, KIF625, KIF626, KIF627, KIF628, KIF629, KIF630, KIF631, KIF632, KIF633, KIF634, KIF635, KIF636, KIF637, KIF638, KIF639, KIF640, KIF641, KIF642, KIF643, KIF644, KIF645, KIF646, KIF647, KIF648, KIF649, KIF650, KIF651, KIF652, KIF653, KIF654, KIF655, KIF656, KIF657, KIF658, KIF659, KIF660, KIF661, KIF662, KIF663, KIF664, KIF665, KIF666, KIF667, KIF668, KIF669, KIF670, KIF671, KIF672, KIF673, KIF674, KIF675, KIF676, KIF677, KIF678, KIF679, KIF680, KIF681, KIF682, KIF683, KIF684, KIF685, KIF686, KIF687, KIF688, KIF689, KIF690, KIF691, KIF692, KIF693, KIF694, KIF695, KIF696, KIF697, KIF698, KIF699, KIF700, KIF701, KIF702, KIF703, KIF704, KIF705, KIF706, KIF707, KIF708, KIF709, KIF710, KIF711, KIF712, KIF713, KIF714, KIF715, KIF716, KIF717, KIF718, KIF719, KIF720, KIF721, KIF722, KIF723, KIF724, KIF725, KIF726, KIF727, KIF728, KIF729, KIF730, KIF731, KIF732, KIF733, KIF734, KIF735, KIF736, KIF737, KIF738, KIF739, KIF740, KIF741, KIF742, KIF743, KIF744, KIF745, KIF746, KIF747, KIF748, KIF749, KIF750, KIF751, KIF752, KIF753, KIF754, KIF755, KIF756, KIF757, KIF758, KIF759, KIF760, KIF761, KIF762, KIF763, KIF764, KIF765, KIF766, KIF767, KIF768, KIF769, KIF770, KIF771, KIF772, KIF773, KIF774, KIF775, KIF776, KIF777, KIF778, KIF779, KIF780, KIF781, KIF782, KIF783, KIF784, KIF785, KIF786, KIF787, KIF788, KIF789, KIF790, KIF791, KIF792, KIF793, KIF794, KIF795, KIF796, KIF797, KIF798, KIF799, KIF800, KIF801, KIF802, KIF803, KIF804, KIF805, KIF806, KIF807, KIF808, KIF809, KIF810, KIF811, KIF812, KIF813, KIF814, KIF815, KIF816, KIF817, KIF818, KIF819, KIF820, KIF821, KIF822, KIF823, KIF824, KIF825, KIF826, KIF827, KIF828, KIF829, KIF830, KIF831, KIF832, KIF833, KIF834, KIF835, KIF836, KIF837, KIF838, KIF839, KIF840, KIF841, KIF842, KIF843, KIF844, KIF845, KIF846, KIF847, KIF848, KIF849, KIF850, KIF851, KIF852, KIF853, KIF854, KIF855, KIF856, KIF857, KIF858, KIF859, KIF860, KIF861, KIF862, KIF863, KIF864, KIF865, KIF866, KIF867, KIF868, KIF869, KIF870, KIF871, KIF872, KIF873, KIF874, KIF875, KIF876, KIF877, KIF878, KIF879, KIF880, KIF881, KIF882, KIF883, KIF884, KIF885, KIF886, KIF887, KIF888, KIF889, KIF890, KIF891, KIF892, KIF893, KIF894, KIF895, KIF896, KIF897, KIF898, KIF899, KIF900, KIF901, KIF902, KIF903, KIF904, KIF905, KIF906, KIF907, KIF908, KIF909, KIF910, KIF911, KIF912, KIF913, KIF914, KIF915, KIF916, KIF917, KIF918, KIF919, KIF920, KIF921, KIF922, KIF923, KIF924, KIF925, KIF926, KIF927, KIF928, KIF929, KIF930, KIF931, KIF932, KIF933, KIF934, KIF935, KIF936, KIF937, KIF938, KIF939, KIF940, KIF941, KIF942, KIF943, KIF944, KIF945, KIF946, KIF947, KIF948, KIF949, KIF950, KIF951, KIF952, KIF953, KIF954, KIF955, KIF956, KIF957, KIF958, KIF959, KIF960, KIF961, KIF962, KIF963, KIF964, KIF965, KIF966, KIF967, KIF968, KIF969, KIF970, KIF971, KIF972, KIF973, KIF974, KIF975, KIF976, KIF977, KIF978, KIF979, KIF980, KIF981, KIF982, KIF983, KIF984, KIF985, KIF986, KIF987, KIF988, KIF989, KIF990, KIF991, KIF992, KIF993, KIF994, KIF995, KIF996, KIF997, KIF998, KIF999, KIF1000	Lavender Top (2 x 4 mL)	Fulgent Genetics (fulgentdiagnostics.com)					
IGF Binding Protein-1 (IGFBP-1) [FIGBP]			Draw blood in a plain, red-top tube(s). Spin down and separate within one hour. Ship 0.5 mL, frozen in a plastic vial.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)		RIA		20-Nov-17	
IGF-II [FIGF2]	123		Draw blood in a plain, red-top tube(s). SST acceptable. Separate within 1 hour of collection, freeze immediately	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)				30-Jul-20	
IL12RB1 (CD212) [IL12PATHWAY]	817		Heparin	Alberta Precision Laboratories (Flow Cytometry - Calgary)			To be accompanied by normal control sample	14-Oct-20	
IL-2 by Luminex [LIL2]	1523		Plasma from a lavender top (EDTA) tube is the only acceptable sample type. Mix sample thoroughly. Specimen Preparation: Centrifuge at room temp within one half hour of collection; preferably immediately after venipuncture. Transfer the cell-free plasma to a clean tube and immediately freeze the cell-free plasma on dry ice or at -70°C.	National Jewish Health		Luminex		18-Mar-21	
Inclusion body myopathy and autosomal recessive, early onset myopathy via the MYH2 gene [361]		MYH2 (160740)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Inclusion Body Myopathy 3 (605637)	Sanger sequencing			
INCLUSION BODY MYOPATHY-2 (AUTOSOMAL RECESSIVE) AND NONAKA MYOPATHY VIA THE GNE GENE [367]		GNE (600824)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Inclusion Body Myopathy 2 (600737); Nonaka Myopathy (605820)	Sanger sequencing			
ING7R12 (CD119) [ILFPGPATHWAY]	817		Heparin	Alberta Precision Laboratories (Flow Cytometry - Calgary)			To be accompanied by normal control sample	14-Oct-20	

Appellation 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 (Déclaratoire MSSS)	SYMBOL, HUNE DU GÈNE et NOM OMDM DU GÈNE / HGNC Gene Symbol and OMDM Gene Name	RÈGLES DE MANUTENTION / Handling Conditions	LABORATOIRE DÉSIGNÉ / Designated laboratory	RENSEIGNEMENTS SUR L'ANALYSE / Test details	MÉTHODE / Methode	NOTE PARTICULIÈRE / Special comment	Date de la révision / Date of revision
1,5-anhydroglucitol (GlycoMark) [500115] [GLYC]	1465		Gold Top or Red Top (Serum) or Lavender (plasma). Centrifuge and aliquot serum to transport tube within one hour of collection. Store and ship frozen. Stable at -20°C for 14 days.	LabCorp/Dynacare (https://www.labcorp.com/)		Enzymatic, colorimetric assay.		10-Jan-21
Insulin Antibodies [INSAB]	853		Gold SST or Red Top	In Common Laboratories		RIA		19-Jun-19
Insulin Receptor		INSR (147670)	Lavender Top (EDTA) 2-4 cc (1-2 cc for children less than 1 year old)	Fulgent Genetics (fulgentdiagnostics.com)	Hyperinsulinemic hypoglycemia familial 5 (609968); Insulin-resistant diabetes mellitus AND acanthosis nigricans (610549); Leprechaunism syndrome (246200); Pancreal hyperplasia AND diabetes mellitus syndrome (262190)	NGS and CGH		
Insulin-Like Growth Factor 1 (IGF1), LC-MS and Insulin-Like Growth Factor-Binding Protein 3 (IGFBP3) Growth Panel [IGFGP]			Red Top. Split into 2 plastic vials. Store and send serum frozen.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)		IGFMS; Liquid Chromatography-Mass Spectrometry (LC-MS) IGFB3; Enzyme-Labeled Chemiluminescent Immunoassay	Not M-R	20-Oct-20
Intellectual Disability NGS Panel	920	ABCC1, ABCD1, ABCD2, ACN1, ACN2, ACSL4, ACP1, ADRM, ADRM2, APOE, APOE2, APOE3, APOE4, APOE5, APOE6, APOE7, APOE8, APOE9, APOE10, APOE11, APOE12, APOE13, APOE14, APOE15, APOE16, APOE17, APOE18, APOE19, APOE20, APOE21, APOE22, APOE23, APOE24, APOE25, APOE26, APOE27, APOE28, APOE29, APOE30, APOE31, APOE32, APOE33, APOE34, APOE35, APOE36, APOE37, APOE38, APOE39, APOE40, APOE41, APOE42, APOE43, APOE44, APOE45, APOE46, APOE47, APOE48, APOE49, APOE50, APOE51, APOE52, APOE53, APOE54, APOE55, APOE56, APOE57, APOE58, APOE59, APOE60, APOE61, APOE62, APOE63, APOE64, APOE65, APOE66, APOE67, APOE68, APOE69, APOE70, APOE71, APOE72, APOE73, APOE74, APOE75, APOE76, APOE77, APOE78, APOE79, APOE80, APOE81, APOE82, APOE83, APOE84, APOE85, APOE86, APOE87, APOE88, APOE89, APOE90, APOE91, APOE92, APOE93, APOE94, APOE95, APOE96, APOE97, APOE98, APOE99, APOE100, APOE101, APOE102, APOE103, APOE104, APOE105, APOE106, APOE107, APOE108, APOE109, APOE110, APOE111, APOE112, APOE113, APOE114, APOE115, APOE116, APOE117, APOE118, APOE119, APOE120, APOE121, APOE122, APOE123, APOE124, APOE125, APOE126, APOE127, APOE128, APOE129, APOE130, APOE131, APOE132, APOE133, APOE134, APOE135, APOE136, APOE137, APOE138, APOE139, APOE140, APOE141, APOE142, APOE143, APOE144, APOE145, APOE146, APOE147, APOE148, APOE149, APOE150, APOE151, APOE152, APOE153, APOE154, APOE155, APOE156, APOE157, APOE158, APOE159, APOE160, APOE161, APOE162, APOE163, APOE164, APOE165, APOE166, APOE167, APOE168, APOE169, APOE170, APOE171, APOE172, APOE173, APOE174, APOE175, APOE176, APOE177, APOE178, APOE179, APOE180, APOE181, APOE182, APOE183, APOE184, APOE185, APOE186, APOE187, APOE188, APOE189, APOE190, APOE191, APOE192, APOE193, APOE194, APOE195, APOE196, APOE197, APOE198, APOE199, APOE200, APOE201, APOE202, APOE203, APOE204, APOE205, APOE206, APOE207, APOE208, APOE209, APOE210, APOE211, APOE212, APOE213, APOE214, APOE215, APOE216, APOE217, APOE218, APOE219, APOE220, APOE221, APOE222, APOE223, APOE224, APOE225, APOE226, APOE227, APOE228, APOE229, APOE230, APOE231, APOE232, APOE233, APOE234, APOE235, APOE236, APOE237, APOE238, APOE239, APOE240, APOE241, APOE242, APOE243, APOE244, APOE245, APOE246, APOE247, APOE248, APOE249, APOE250, APOE251, APOE252, APOE253, APOE254, APOE255, APOE256, APOE257, APOE258, APOE259, APOE260, APOE261, APOE262, APOE263, APOE264, APOE265, APOE266, APOE267, APOE268, APOE269, APOE270, APOE271, APOE272, APOE273, APOE274, APOE275, APOE276, APOE277, APOE278, APOE279, APOE280, APOE281, APOE282, APOE283, APOE284, APOE285, APOE286, APOE287, APOE288, APOE289, APOE290, APOE291, APOE292, APOE293, APOE294, APOE295, APOE296, APOE297, APOE298, APOE299, APOE300, APOE301, APOE302, APOE303, APOE304, APOE305, APOE306, APOE307, APOE308, APOE309, APOE310, APOE311, APOE312, APOE313, APOE314, APOE315, APOE316, APOE317, APOE318, APOE319, APOE320, APOE321, APOE322, APOE323, APOE324, APOE325, APOE326, APOE327, APOE328, APOE329, APOE330, APOE331, APOE332, APOE333, APOE334, APOE335, APOE336, APOE337, APOE338, APOE339, APOE340, APOE341, APOE342, APOE343, APOE344, APOE345, APOE346, APOE347, APOE348, APOE349, APOE350, APOE351, APOE352, APOE353, APOE354, APOE355, APOE356, APOE357, APOE358, APOE359, APOE360, APOE361, APOE362, APOE363, APOE364, APOE365, APOE366, APOE367, APOE368, APOE369, APOE370, APOE371, APOE372, APOE373, APOE374, APOE375, APOE376, APOE377, APOE378, APOE379, APOE380, APOE381, APOE382, APOE383, APOE384, APOE385, APOE386, APOE387, APOE388, APOE389, APOE390, APOE391, APOE392, APOE393, APOE394, APOE395, APOE396, APOE397, APOE398, APOE399, APOE400, APOE401, APOE402, APOE403, APOE404, APOE405, APOE406, APOE407, APOE408, APOE409, APOE410, APOE411, APOE412, APOE413, APOE414, APOE415, APOE416, APOE417, APOE418, APOE419, APOE420, APOE421, APOE422, APOE423, APOE424, APOE425, APOE426, APOE427, APOE428, APOE429, APOE430, APOE431, APOE432, APOE433, APOE434, APOE435, APOE436, APOE437, APOE438, APOE439, APOE440, APOE441, APOE442, APOE443, APOE444, APOE445, APOE446, APOE447, APOE448, APOE449, APOE450, APOE451, APOE452, APOE453, APOE454, APOE455, APOE456, APOE457, APOE458, APOE459, APOE460, APOE461, APOE462, APOE463, APOE464, APOE465, APOE466, APOE467, APOE468, APOE469, APOE470, APOE471, APOE472, APOE473, APOE474, APOE475, APOE476, APOE477, APOE478, APOE479, APOE480, APOE481, APOE482, APOE483, APOE484, APOE485, APOE486, APOE487, APOE488, APOE489, APOE490, APOE491, APOE492, APOE493, APOE494, APOE495, APOE496, APOE497, APOE498, APOE499, APOE500, APOE501, APOE502, APOE503, APOE504, APOE505, APOE506, APOE507, APOE508, APOE509, APOE510, APOE511, APOE512, APOE513, APOE514, APOE515, APOE516, APOE517, APOE518, APOE519, APOE520, APOE521, APOE522, APOE523, APOE524, APOE525, APOE526, APOE527, APOE528, APOE529, APOE530, APOE531, APOE532, APOE533, APOE534, APOE535, APOE536, APOE537, APOE538, APOE539, APOE540, APOE541, APOE542, APOE543, APOE544, APOE545, APOE546, APOE547, APOE548, APOE549, APOE550, APOE551, APOE552, APOE553, APOE554, APOE555, APOE556, APOE557, APOE558, APOE559, APOE560, APOE561, APOE562, APOE563, APOE564, APOE565, APOE566, APOE567, APOE568, APOE569, APOE570, APOE571, APOE572, APOE573, APOE574, APOE575, APOE576, APOE577, APOE578, APOE579, APOE580, APOE581, APOE582, APOE583, APOE584, APOE585, APOE586, APOE587, APOE588, APOE589, APOE590, APOE591, APOE592, APOE593, APOE594, APOE595, APOE596, APOE597, APOE598, APOE599, APOE600, APOE601, APOE602, APOE603, APOE604, APOE605, APOE606, APOE607, APOE608, APOE609, APOE610, APOE611, APOE612, APOE613, APOE614, APOE615, APOE616, APOE617, APOE618, APOE619, APOE620, APOE621, APOE622, APOE623, APOE624, APOE625, APOE626, APOE627, APOE628, APOE629, APOE630, APOE631, APOE632, APOE633, APOE634, APOE635, APOE636, APOE637, APOE638, APOE639, APOE640, APOE641, APOE642, APOE643, APOE644, APOE645, APOE646, APOE647, APOE648, APOE649, APOE650, APOE651, APOE652, APOE653, APOE654, APOE655, APOE656, APOE657, APOE658, APOE659, APOE660, APOE661, APOE662, APOE663, APOE664, APOE665, APOE666, APOE667, APOE668, APOE669, APOE670, APOE671, APOE672, APOE673, APOE674, APOE675, APOE676, APOE677, APOE678, APOE679, APOE680, APOE681, APOE682, APOE683, APOE684, APOE685, APOE686, APOE687, APOE688, APOE689, APOE690, APOE691, APOE692, APOE693, APOE694, APOE695, APOE696, APOE697, APOE698, APOE699, APOE700, APOE701, APOE702, APOE703, APOE704, APOE705, APOE706, APOE707, APOE708, APOE709, APOE710, APOE711, APOE712, APOE713, APOE714, APOE715, APOE716, APOE717, APOE718, APOE719, APOE720, APOE721, APOE722, APOE723, APOE724, APOE725, APOE726, APOE727, APOE728, APOE729, APOE730, APOE731, APOE732, APOE733, APOE734, APOE735, APOE736, APOE737, APOE738, APOE739, APOE740, APOE741, APOE742, APOE743, APOE744, APOE745, APOE746, APOE747, APOE748, APOE749, APOE750, APOE751, APOE752, APOE753, APOE754, APOE755, APOE756, APOE757, APOE758, APOE759, APOE760, APOE761, APOE762, APOE763, APOE764, APOE765, APOE766, APOE767, APOE768, APOE769, APOE770, APOE771, APOE772, APOE773, APOE774, APOE775, APOE776, APOE777, APOE778, APOE779, APOE780, APOE781, APOE782, APOE783, APOE784, APOE785, APOE786, APOE787, APOE788, APOE789, APOE790, APOE791, APOE792, APOE793, APOE794, APOE795, APOE796, APOE797, APOE798, APOE799, APOE800, APOE801, APOE802, APOE803, APOE804, APOE805, APOE806, APOE807, APOE808, APOE809, APOE810, APOE811, APOE812, APOE813, APOE814, APOE815, APOE816, APOE817, APOE818, APOE819, APOE820, APOE821, APOE822, APOE823, APOE824, APOE825, APOE826, APOE827, APOE828, APOE829, APOE830, APOE831, APOE832, APOE833, APOE834, APOE835, APOE836, APOE837, APOE838, APOE839, APOE840, APOE841, APOE842, APOE843, APOE844, APOE845, APOE846, APOE847, APOE848, APOE849, APOE850, APOE851, APOE852, APOE853, APOE854, APOE855, APOE856, APOE857, APOE858, APOE859, APOE860, APOE861, APOE862, APOE863, APOE864, APOE865, APOE866, APOE867, APOE868, APOE869, APOE870, APOE871, APOE872, APOE873, APOE874, APOE875, APOE876, APOE877, APOE878, APOE879, APOE880, APOE881, APOE882, APOE883, APOE884, APOE885, APOE886, APOE887, APOE888, APOE889, APOE890, APOE891, APOE892, APOE893, APOE894, APOE895, APOE896, APOE897, APOE898, APOE899, APOE900, APOE901, APOE902, APOE903, APOE904, APOE905, APOE906, APOE907, APOE908, APOE909, APOE910, APOE911, APOE912, APOE913, APOE914, APOE915, APOE916, APOE917, APOE918, APOE919, APOE920, APOE921, APOE922, APOE923, APOE924, APOE925, APOE926, APOE927, APOE928, APOE929, APOE930, APOE931, APOE932, APOE933, APOE934, APOE935, APOE936, APOE937, APOE938, APOE939, APOE940, APOE941, APOE942, APOE943, APOE944, APOE945, APOE946, APOE947, APOE948, APOE949, APOE950, APOE951, APOE952, APOE953, APOE954, APOE955, APOE956, APOE957, APOE958, APOE959, APOE960, APOE961, APOE962, APOE963, APOE964, APOE965, APOE966, APOE967, APOE968, APOE969, APOE970, APOE971, APOE972, APOE973, APOE974, APOE975, APOE976, APOE977, APOE978, APOE979, APOE980, APOE981, APOE982, APOE983, APOE984, APOE985, APOE986, APOE987, APOE988, APOE989, APOE990, APOE991, APOE992, APOE993, APOE994, APOE995, APOE996, APOE997, APOE998, APOE999, APOE1000, APOE1001, APOE1002, APOE1003, APOE1004, APOE1005, APOE1006, APOE1007, APOE1008, APOE1009, APOE1010, APOE1011, APOE1012, APOE1013, APOE1014, APOE1015, APOE1016, APOE1017, APOE1018, APOE1019, APOE1020, APOE1021, APOE1022, APOE1023, APOE1024, APOE1025, APOE1026, APOE1027, APOE1028, APOE1029, APOE1030, APOE1031, APOE1032, APOE1033, APOE1034, APOE1035, APOE1036, APOE1037, APOE1038, APOE1039, APOE1040, APOE1041, APOE1042, APOE1043, APOE1044, APOE1045, APOE1046, APOE1047, APOE1048, APOE1049, APOE1050, APOE1051, APOE1052, APOE1053, APOE1054, APOE1055, APOE1056, APOE1057, APOE1058, APOE1059, APOE1060, APOE1061, APOE1062, APOE1063, APOE1064, APOE1065, APOE1066, APOE1067, APOE1068, APOE1069, APOE1070, APOE1071, APOE1072, APOE1073, APOE1074, APOE1075, APOE1076, APOE1077, APOE1078, APOE1079, APOE1080, APOE1081, APOE1082, APOE1083, APOE1084, APOE1085, APOE1086, APOE1087, APOE1088, APOE1089, APOE1090, APOE1091, APOE1092, APOE1093, APOE1094, APOE1095, APOE1096, APOE1097, APOE1098, APOE1099, APOE1100, APOE1101, APOE1102, APOE1103, APOE1104, APOE1105, APOE1106, APOE1107, APOE1108, APOE1109, APOE1110, APOE1111, APOE1112, APOE1113, APOE1114, APOE1115, APOE1116, APOE1117, APOE1118, APOE1119, APOE1120, APOE1121, APOE1122, APOE1123, APOE1124, APOE1125, APOE1126, APOE1127, APOE1128, APOE1129, APOE1130, APOE1131, APOE1132, APOE1133, APOE1134, APOE1135, APOE1136, APOE1137, APOE1138, APOE1139, APOE1140, APOE1141, APOE1142, APOE1143, APOE1144, APOE1145, APOE1146, APOE1147, APOE1148, APOE1149, APOE1150, APOE1151, APOE1152, APOE1153, APOE1154, APOE1155, APOE1156, APOE1157, APOE1158, APOE1159, APOE1160, APOE1161, APOE1162, APOE1163, APOE1164, APOE1165, APOE1166, APOE1167, APOE1168, APOE1169, APOE1170, APOE1171, APOE1172, APOE1173, APOE1174, APOE1175, APOE1176, APOE1177, APOE1178, APOE1179, APOE1180, APOE1181, APOE1182, APOE1183, APOE1184, APOE1185, APOE1186, APOE1187, APOE1188, APOE1189, APOE1190, APOE1191, APOE1192, APOE1193, APOE1194, APOE1195, APOE1196, APOE1197, APOE1198, APOE1199, APOE1200, APOE1201, APOE1202, APOE1203, APOE1204, APOE1205, APOE1206, APOE1207, APOE1208, APOE1209, APOE1210, APOE1211, APOE1212, APOE1213, APOE1214, APOE1215, APOE1216, APOE1217, APOE1218, APOE1219, APOE1220, APOE1221, APOE1222, APOE1223, APOE1224, APOE1225, APOE1226, APOE1227, APOE1228, APOE1229, APOE1230, APOE1231, APOE1232, APOE1233, APOE1234, APOE1235, APOE1236, APOE1237, APOE1238, APOE1239, APOE1240, APOE1241, APOE1242, APOE1243, APOE1244, APOE1245, APOE1246, APOE1247, APOE1248, APOE1249, APOE1250, APOE1251, APOE1252, APOE1253, APOE1254, APOE1255, APOE1256, APOE1257, APOE1258, APOE1259, APOE1260, APOE1261, APOE1262, APOE1263, APOE1264, APOE1265, APOE1266, APOE1267, APOE1268, APOE1269, APOE1270, APOE1271, APOE1272, APOE1273, APOE1274, APOE1275, APOE1276, APOE1277, APOE1278, APOE1279, APOE1280, APOE1281, APOE1282, APOE1283, APOE1284, APOE1285, APOE1286, APOE1287, APOE1288, APOE1289, APOE1290, APOE1291, APOE1292, APOE1293, APOE1294, APOE1295, APOE1296, APOE1297, APOE1298, APOE1299, APOE1300, APOE1301, APOE1302, APOE1303, APOE1304, APOE1305, APOE1306, APOE1307, APOE1308, APOE1309, APOE1310, APOE1311, APOE1312, APOE1313, APOE1314, APOE1315, APOE1316, APOE1317, APOE1318, APOE1319, APOE1320, APOE1321, APOE1322, APOE1323, APOE1324, APOE1325, APOE1326, APOE1327, APOE1328, APOE1329, APOE1330, APOE1331, APOE1332, APOE1333, APOE1334, APOE1335, APOE1336, APOE1337, APOE1338, APOE1339, APOE1340, APOE1341, APOE1342, APOE1343, APOE1344, APOE1345, APOE1346, APOE1347, APOE1348, APOE1349, APOE1350, APOE1351, APOE1352, APOE1353, APOE1354, APOE1355, APOE1356, APOE1357, APOE1358, APOE1359, APOE1360, APOE1361, APOE1362, APOE1363, APOE1364, APOE1365, APOE1366, APOE1367, APOE1368, APOE1369, APOE1370, APOE1371, APOE1372, APOE1373, APOE1374, APOE1375, APOE1376, APOE1377, APOE1378, APOE1379, APOE1380, APOE1381, APOE1382, APOE1383, APOE1384, APOE1385, APOE1386, APOE1387, APOE1388, APOE1389, APOE1390, APOE1391, APOE1392, APOE1393, APOE1394, APOE1395, APOE1396, APOE1397, APOE1398, APOE1399, APOE1400, APOE1401, APOE1402, APOE1403, APOE1404, APOE1405, APOE1406, APOE1407, APOE1408, APOE1409, APOE1410, APOE1411, APOE1412, APOE1413, APOE1414, APOE1415, APOE1416, APOE1417, APOE1418, APOE1419, APOE1420, APOE1421, APOE1422, APOE1423, APOE1424, APOE1425, APOE1426, APOE1427, APOE1428, APOE1429, APOE1430, APOE1431, APOE1432, APOE1433, APOE1434, APOE1435, APOE1436, APOE1437, APOE1438, APOE1439, APOE1440, APOE1441, APOE1442, APOE1443, APOE1444, APOE1445, APOE1446, APOE1447, APOE1448, APOE1449, APOE1450, APOE1451, APOE1452, APOE1453, APOE1454, APOE1455, APOE1456, APOE1457, APOE1458, APOE1459, APOE1460, APOE1461, APOE1462, APOE1463, APOE1464, APOE1465, APOE1466, APOE1467, APOE1468, APOE1469, APOE1470, APOE1471, APOE1472, APOE1473, APOE1474, APOE1475, APOE1476, APOE1477, APOE1478, APOE1479, APOE1480, APOE1481, APOE1482, APOE1483, APOE1484, APOE1485, APOE1486, APOE1487, APOE1488, APOE1489, APOE1490, APOE1491, APOE1492, APOE1493, APOE1494, APOE1495, APOE1496, APOE1497, APOE1498, APOE1499, APOE1500, APOE1501, APOE1502, APOE1503, APOE1504, APOE1505, APOE1506, APOE1507, APOE1508, APOE1509, APOE1510, APOE1511, APOE1512, APOE1513, APOE1514, APOE1515, APOE1516, APOE1517, APOE1518, APOE1519, APOE1520, APOE1521, APOE1522, APOE1523, APOE1524, APOE1525, APOE1526, APOE1527, APOE1528, APOE1529, APOE1530, APOE1531, APOE1532, APOE1533, APOE1534, APOE1535, APOE1536, APOE1537, APOE1538, APOE1539, APOE1540, APOE1541, APOE15						

Appellation 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)	SYMBOLIÈRE DU GÈNE et NOM (OMIM DU GÈNE / HGNC Gene Symbol and OMIM Gene Name)	RÈGLES DE MANUTENTION / Handling Conditions	LABORATOIRE DÉSIGNÉ / Designated laboratory	RENSEIGNEMENTS SUR L'ANALYSE / Test details	MÉTHODE / Methode	NOTE PARTICULIÈRE / Special comment	Date de la révision / Date of revision
1,5-anhydroglucitol (GlycoMark) [500115] [GLYC]	1465		Gold Top or Red Top (Serum) or Lavender (plasma). Centrifuge and aliquot serum to transport tube within one hour of collection. Store and ship frozen. Stable at -20°C for 14 days.	LabCorp/Dynacare (https://www.labcorp.com/)		Enzymatic, colorimetric assay.		10-Jan-21
JOUBERT AND MECKEL-GRUBER SYNDROMES NEXTGEN SEQUENCING (NGS) PANEL [1057]	332	<p>III1 (608864) ARL13B (608922) JPOB1 (614144) JPOB2 (611951) CS642 (614571) CC2D2A (612013) CEP290 (610142) CEP41 (610823) INP96 (613007) KIF7 (611284) MKSI (609883) NRP1 (607100) NRP2 (608002) OFD1 (300170) RORBP1L (610907) TCTN1 (609063) TCTN2 (613846) TCTN3 (613407) TMEI18 (644459) TMEI216 (611277) TMEI221 (613499) TMEI237 (644425) TMEI367 (609884) TTC218 (612014) ZNF823 (604857)</p>	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)		NextGen Sequencing		
JUNCTIONAL EPIDERMOLYSIS BULLOSA (JEB) SANGER sequencing panel [975]		LAMC2 (150292); LAMA3 (600805); LAMB3 (180310); COL17A1 (113811)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)				
Juvenile Polyposis Syndrome (610069 and 174900) [536]		BMPRIA (601299); SMAD4 (600993)	Lavender Top (EDTA)	GeneDx (www.genedx.com)		Tier 1: Sanger Sequencing, Exon Array CGH		09-Dec-19
Juvenile Polyposis Syndrome (610069 and 174900) [537]		BMPRIA (601299)	Lavender Top (EDTA)	GeneDx (www.genedx.com)		Tier 2: sequencing		09-Dec-19
Juvenile Polyposis Syndrome (610069 and 174900) [538]		BMPRIA (601299); SMAD4 (600993)	Lavender Top (EDTA)	GeneDx (www.genedx.com)		Tier 3: Exon Array CGH		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	<p>ACL1, AGT, AGLT1, ANOS1, BMP4, BMP7, CDC3L, CHD1L, DSTYK, EYA1, FGF30, FGFRL2, FRAS1, FREM1, FREM2, GATA3, GLI3, GRIP1, HNF1B, HNF1A1, HNF1A4, HNF1B, HNF1C, HNF1D, HNF1E, HNF1F, HNF1G, HNF1H, HNF1I, HNF1J, HNF1K, HNF1L, HNF1M, HNF1N, HNF1O, HNF1P, HNF1Q, HNF1R, HNF1S, HNF1T, HNF1U, HNF1V, HNF1W, HNF1X, HNF1Y, HNF1Z, HNF2, HNF3, HNF4, HNF5, HNF6, HNF7, HNF8, HNF9, HNF10, HNF11, HNF12, HNF13, HNF14, HNF15, HNF16, HNF17, HNF18, HNF19, HNF20, HNF21, HNF22, HNF23, HNF24, HNF25, HNF26, HNF27, HNF28, HNF29, HNF30, HNF31, HNF32, HNF33, HNF34, HNF35, HNF36, HNF37, HNF38, HNF39, HNF40, HNF41, HNF42, HNF43, HNF44, HNF45, HNF46, HNF47, HNF48, HNF49, HNF50, HNF51, HNF52, HNF53, HNF54, HNF55, HNF56, HNF57, HNF58, HNF59, HNF60, HNF61, HNF62, HNF63, HNF64, HNF65, HNF66, HNF67, HNF68, HNF69, HNF70, HNF71, HNF72, HNF73, HNF74, HNF75, HNF76, HNF77, HNF78, HNF79, HNF80, HNF81, HNF82, HNF83, HNF84, HNF85, HNF86, HNF87, HNF88, HNF89, HNF90, HNF91, HNF92, HNF93, HNF94, HNF95, HNF96, HNF97, HNF98, HNF99, HNF100, HNF101, HNF102, HNF103, HNF104, HNF105, HNF106, HNF107, HNF108, HNF109, HNF110, HNF111, HNF112, HNF113, HNF114, HNF115, HNF116, HNF117, HNF118, HNF119, HNF120, HNF121, HNF122, HNF123, HNF124, HNF125, HNF126, HNF127, HNF128, HNF129, HNF130, HNF131, HNF132, HNF133, HNF134, HNF135, HNF136, HNF137, HNF138, HNF139, HNF140, HNF141, HNF142, HNF143, HNF144, HNF145, HNF146, HNF147, HNF148, HNF149, HNF150, HNF151, HNF152, HNF153, HNF154, HNF155, HNF156, HNF157, HNF158, HNF159, HNF160, HNF161, HNF162, HNF163, HNF164, HNF165, HNF166, HNF167, HNF168, HNF169, HNF170, HNF171, HNF172, HNF173, HNF174, HNF175, HNF176, HNF177, HNF178, HNF179, HNF180, HNF181, HNF182, HNF183, HNF184, HNF185, HNF186, HNF187, HNF188, HNF189, HNF190, HNF191, HNF192, HNF193, HNF194, HNF195, HNF196, HNF197, HNF198, HNF199, HNF200, HNF201, HNF202, HNF203, HNF204, HNF205, HNF206, HNF207, HNF208, HNF209, HNF210, HNF211, HNF212, HNF213, HNF214, HNF215, HNF216, HNF217, HNF218, HNF219, HNF220, HNF221, HNF222, HNF223, HNF224, HNF225, HNF226, HNF227, HNF228, HNF229, HNF230, HNF231, HNF232, HNF233, HNF234, HNF235, HNF236, HNF237, HNF238, HNF239, HNF240, HNF241, HNF242, HNF243, HNF244, HNF245, HNF246, HNF247, HNF248, HNF249, HNF250, HNF251, HNF252, HNF253, HNF254, HNF255, HNF256, HNF257, HNF258, HNF259, HNF260, HNF261, HNF262, HNF263, HNF264, HNF265, HNF266, HNF267, HNF268, HNF269, HNF270, HNF271, HNF272, HNF273, HNF274, HNF275, HNF276, HNF277, HNF278, HNF279, HNF280, HNF281, HNF282, HNF283, HNF284, HNF285, HNF286, HNF287, HNF288, HNF289, HNF290, HNF291, HNF292, HNF293, HNF294, HNF295, HNF296, HNF297, HNF298, HNF299, HNF300, HNF301, HNF302, HNF303, HNF304, HNF305, HNF306, HNF307, HNF308, HNF309, HNF310, HNF311, HNF312, HNF313, HNF314, HNF315, HNF316, HNF317, HNF318, HNF319, HNF320, HNF321, HNF322, HNF323, HNF324, HNF325, HNF326, HNF327, HNF328, HNF329, HNF330, HNF331, HNF332, HNF333, HNF334, HNF335, HNF336, HNF337, HNF338, HNF339, HNF340, HNF341, HNF342, HNF343, HNF344, HNF345, HNF346, HNF347, HNF348, HNF349, HNF350, HNF351, HNF352, HNF353, HNF354, HNF355, HNF356, HNF357, HNF358, HNF359, HNF360, HNF361, HNF362, HNF363, HNF364, HNF365, HNF366, HNF367, HNF368, HNF369, HNF370, HNF371, HNF372, HNF373, HNF374, HNF375, HNF376, HNF377, HNF378, HNF379, HNF380, HNF381, HNF382, HNF383, HNF384, HNF385, HNF386, HNF387, HNF388, HNF389, HNF390, HNF391, HNF392, HNF393, HNF394, HNF395, HNF396, HNF397, HNF398, HNF399, HNF400, HNF401, HNF402, HNF403, HNF404, HNF405, HNF406, HNF407, HNF408, HNF409, HNF410, HNF411, HNF412, HNF413, HNF414, HNF415, HNF416, HNF417, HNF418, HNF419, HNF420, HNF421, HNF422, HNF423, HNF424, HNF425, HNF426, HNF427, HNF428, HNF429, HNF430, HNF431, HNF432, HNF433, HNF434, HNF435, HNF436, HNF437, HNF438, HNF439, HNF440, HNF441, HNF442, HNF443, HNF444, HNF445, HNF446, HNF447, HNF448, HNF449, HNF450, HNF451, HNF452, HNF453, HNF454, HNF455, HNF456, HNF457, HNF458, HNF459, HNF460, HNF461, HNF462, HNF463, HNF464, HNF465, HNF466, HNF467, HNF468, HNF469, HNF470, HNF471, HNF472, HNF473, HNF474, HNF475, HNF476, HNF477, HNF478, HNF479, HNF480, HNF481, HNF482, HNF483, HNF484, HNF485, HNF486, HNF487, HNF488, HNF489, HNF490, HNF491, HNF492, HNF493, HNF494, HNF495, HNF496, HNF497, HNF498, HNF499, HNF500, HNF501, HNF502, HNF503, HNF504, HNF505, HNF506, HNF507, HNF508, HNF509, HNF510, HNF511, HNF512, HNF513, HNF514, HNF515, HNF516, HNF517, HNF518, HNF519, HNF520, HNF521, HNF522, HNF523, HNF524, HNF525, HNF526, HNF527, HNF528, HNF529, HNF530, HNF531, HNF532, HNF533, HNF534, HNF535, HNF536, HNF537, HNF538, HNF539, HNF540, HNF541, HNF542, HNF543, HNF544, HNF545, HNF546, HNF547, HNF548, HNF549, HNF550, HNF551, HNF552, HNF553, HNF554, HNF555, HNF556, HNF557, HNF558, HNF559, HNF560, HNF561, HNF562, HNF563, HNF564, HNF565, HNF566, HNF567, HNF568, HNF569, HNF570, HNF571, HNF572, HNF573, HNF574, HNF575, HNF576, HNF577, HNF578, HNF579, HNF580, HNF581, HNF582, HNF583, HNF584, HNF585, HNF586, HNF587, HNF588, HNF589, HNF590, HNF591, HNF592, HNF593, HNF594, HNF595, HNF596, HNF597, HNF598, HNF599, HNF600, HNF601, HNF602, HNF603, HNF604, HNF605, HNF606, HNF607, HNF608, HNF609, HNF610, HNF611, HNF612, HNF613, HNF614, HNF615, HNF616, HNF617, HNF618, HNF619, HNF620, HNF621, HNF622, HNF623, HNF624, HNF625, HNF626, HNF627, HNF628, HNF629, HNF630, HNF631, HNF632, HNF633, HNF634, HNF635, HNF636, HNF637, HNF638, HNF639, HNF640, HNF641, HNF642, HNF643, HNF644, HNF645, HNF646, HNF647, HNF648, HNF649, HNF650, HNF651, HNF652, HNF653, HNF654, HNF655, HNF656, HNF657, HNF658, HNF659, HNF660, HNF661, HNF662, HNF663, HNF664, HNF665, HNF666, HNF667, HNF668, HNF669, HNF670, HNF671, HNF672, HNF673, HNF674, HNF675, HNF676, HNF677, HNF678, HNF679, HNF680, HNF681, HNF682, HNF683, HNF684, HNF685, HNF686, HNF687, HNF688, HNF689, HNF690, HNF691, HNF692, HNF693, HNF694, HNF695, HNF696, HNF697, HNF698, HNF699, HNF700, HNF701, HNF702, HNF703, HNF704, HNF705, HNF706, HNF707, HNF708, HNF709, HNF710, HNF711, HNF712, HNF713, HNF714, HNF715, HNF716, HNF717, HNF718, HNF719, HNF720, HNF721, HNF722, HNF723, HNF724, HNF725, HNF726, HNF727, HNF728, HNF729, HNF730, HNF731, HNF732, HNF733, HNF734, HNF735, HNF736, HNF737, HNF738, HNF739, HNF740, HNF741, HNF742, HNF743, HNF744, HNF745, HNF746, HNF747, HNF748, HNF749, HNF750, HNF751, HNF752, HNF753, HNF754, HNF755, HNF756, HNF757, HNF758, HNF759, HNF760, HNF761, HNF762, HNF763, HNF764, HNF765, HNF766, HNF767, HNF768, HNF769, HNF770, HNF771, HNF772, HNF773, HNF774, HNF775, HNF776, HNF777, HNF778, HNF779, HNF780, HNF781, HNF782, HNF783, HNF784, HNF785, HNF786, HNF787, HNF788, HNF789, HNF790, HNF791, HNF792, HNF793, HNF794, HNF795, HNF796, HNF797, HNF798, HNF799, HNF800, HNF801, HNF802, HNF803, HNF804, HNF805, HNF806, HNF807, HNF808, HNF809, HNF810, HNF811, HNF812, HNF813, HNF814, HNF815, HNF816, HNF817, HNF818, HNF819, HNF820, HNF821, HNF822, HNF823, HNF824, HNF825, HNF826, HNF827, HNF828, HNF829, HNF830, HNF831, HNF832, HNF833, HNF834, HNF835, HNF836, HNF837, HNF838, HNF839, HNF840, HNF841, HNF842, HNF843, HNF844, HNF845, HNF846, HNF847, HNF848, HNF849, HNF850, HNF851, HNF852, HNF853, HNF854, HNF855, HNF856, HNF857, HNF858, HNF859, HNF860, HNF861, HNF862, HNF863, HNF864, HNF865, HNF866, HNF867, HNF868, HNF869, HNF870, HNF871, HNF872, HNF873, HNF874, HNF875, HNF876, HNF877, HNF878, HNF879, HNF880, HNF881, HNF882, HNF883, HNF884, HNF885, HNF886, HNF887, HNF888, HNF889, HNF890, HNF891, HNF892, HNF893, HNF894, HNF895, HNF896, HNF897, HNF898, HNF899, HNF900, HNF901, HNF902, HNF903, HNF904, HNF905, HNF906, HNF907, HNF908, HNF909, HNF910, HNF911, HNF912, HNF913, HNF914, HNF915, HNF916, HNF917, HNF918, HNF919, HNF920, HNF921, HNF922, HNF923, HNF924, HNF925, HNF926, HNF927, HNF928, HNF929, HNF930, HNF931, HNF932, HNF933, HNF934, HNF935, HNF936, HNF937, HNF938, HNF939, HNF940, HNF941, HNF942, HNF943, HNF944, HNF945, HNF946, HNF947, HNF948, HNF949, HNF950, HNF951, HNF952, HNF953, HNF954, HNF955, HNF956, HNF957, HNF958, HNF959, HNF960, HNF961, HNF962, HNF963, HNF964, HNF965, HNF966, HNF967, HNF968, HNF969, HNF970, HNF971, HNF972, HNF973, HNF974, HNF975, HNF976, HNF977, HNF978, HNF979, HNF980, HNF981, HNF982, HNF983, HNF984, HNF985, HNF986, HNF987, HNF988, HNF989, HNF990, HNF991, HNF992, HNF993, HNF994, HNF995, HNF996, HNF997, HNF998, HNF999, HNF1000, HNF1001, HNF1002, HNF1003, HNF1004, HNF1005, HNF1006, HNF1007, HNF1008, HNF1009, HNF1010, HNF1011, HNF1012, HNF1013, HNF1014, HNF1015, HNF1016, HNF1017, HNF1018, HNF1019, HNF1020, HNF1021, HNF1022, HNF1023, HNF1024, HNF1025, HNF1026, HNF1027, HNF1028, HNF1029, HNF1030, HNF1031, HNF1032, HNF1033, HNF1034, HNF1035, HNF1036, HNF1037, HNF1038, HNF1039, HNF1040, HNF1041, HNF1042, HNF1043, HNF1044, HNF1045, HNF1046, HNF1047, HNF1048, HNF1049, HNF1050, HNF1051, HNF1052, HNF1053, HNF1054, HNF1055, HNF1056, HNF1057, HNF1058, HNF1059, HNF1060, HNF1061, HNF1062, HNF1063, HNF1064, HNF1065, HNF1066, HNF1067, HNF1068, HNF1069, HNF1070, HNF1071, HNF1072, HNF1073, HNF1074, HNF1075, HNF1076, HNF1077, HNF1078, HNF1079, HNF1080, HNF1081, HNF1082, HNF1083, HNF1084, HNF1085, HNF1086, HNF1087, HNF1088, HNF1089, HNF1090, HNF1091, HNF1092, HNF1093, HNF1094, HNF1095, HNF1096, HNF1097, HNF1098, HNF1099, HNF1100, HNF1101, HNF1102, HNF1103, HNF1104, HNF1105, HNF1106, HNF1107, HNF1108, HNF1109, HNF1110, HNF1111, HNF1112, HNF1113, HNF1114, HNF1115, HNF1116, HNF1117, HNF1118, HNF1119, HNF1120, HNF1121, HNF1122, HNF1123, HNF1124, HNF1125, HNF1126, HNF1127, HNF1128, HNF1129, HNF1130, HNF1131, HNF1132, HNF1133, HNF1134, HNF1135, HNF1136, HNF1137, HNF1138, HNF1139, HNF1140, HNF1141, HNF1142, HNF1143, HNF1144, HNF1145, HNF1146, HNF1147, HNF1148, HNF1149, HNF1150, HNF1151, HNF1152, HNF1153, HNF1154, HNF1155, HNF1156, HNF1157, HNF1158, HNF1159, HNF1160, HNF1161, HNF1162, HNF1163, HNF1164, HNF1165, HNF1166, HNF1167, HNF1168, HNF1169, HNF1170, HNF1171, HNF1172, HNF1173, HNF1174, HNF1175, HNF1176, HNF1177, HNF1178, HNF1179, HNF1180, HNF1181, HNF1182, HNF1183, HNF1184, HNF1185, HNF1186, HNF1187, HNF1188, HNF1189, HNF1190, HNF1191, HNF1192, HNF1193, HNF1194, HNF1195, HNF1196, HNF1197, HNF1198, HNF1199, HNF1200, HNF1201, HNF1202, HNF1203, HNF1204, HNF1205, HNF1206, HNF1207, HNF1208, HNF1209, HNF1210, HNF1211, HNF1212, HNF1213, HNF1214, HNF1215, HNF1216, HNF1217, HNF1218, HNF1219, HNF1220, HNF1221, HNF1222, HNF1223, HNF1224, HNF1225, HNF1226, HNF1227, HNF1228, HNF1229, HNF1230, HNF1231, HNF1232, HNF1233, HNF1234, HNF1235, HNF1236, HNF1237, HNF1238, HNF1239, HNF1240, HNF1241, HNF1242, HNF1243, HNF1244, HNF1245, HNF1246, HNF1247, HNF1248, HNF1249, HNF1250, HNF1251, HNF1252, HNF1253, HNF1254, HNF1255, HNF1256, HNF1257, HNF1258, HNF1259, HNF1260, HNF1261, HNF1262, HNF1263, HNF1264, HNF1265, HNF1266, HNF1267, HNF1268, HNF1269, HNF1270, HNF1271, HNF1272, HNF1273, HNF1274, HNF1275, HNF1276, HNF1277, HNF1278, HNF1279, HNF1280, HNF1281, HNF1282, HNF1283, HNF1284, HNF1285, HNF1286, HNF1287, HNF1288, HNF1289, HNF1290, HNF1291, HNF1292, HNF1293, HNF1294, HNF1295, HNF1296, HNF1297, HNF1298, HNF1299, HNF1300, HNF1301, HNF1302, HNF1303, HNF1304, HNF1305, HNF1306, HNF1307, HNF1308, HNF1309, HNF1310, HNF1311, HNF1312, HNF1313, HNF1314, HNF1315, HNF1316, HNF1317, HNF1318, HNF1319, HNF1320, HNF1321, HNF1322, HNF1323, HNF1324, HNF1325, HNF1326, HNF1327, HNF1328, HNF1329, HNF1330, HNF1331, HNF1332, HNF1333, HNF1334, HNF1335, HNF1336, HNF1337, HNF1338, HNF1339, HNF1340, HNF1341, HNF1342, HNF1343, HNF1344, HNF1345, HNF1346, HNF1347, HNF1348, HNF1349, HNF1350, HNF1351, HNF1352, HNF1353, HNF1354, HNF1355, HNF1356, HNF1357, HNF1358, HNF1359, HNF1360, HNF1361, HNF1362, HNF1363, HNF1364, HNF1365, HNF1366, HNF1367, HNF1368, HNF1369, HNF1370, HNF1371, HNF1372, HNF1373, HNF1374, HNF1375, HNF1376, HNF1377, HNF1378, HNF1379, HNF1380, HNF1381, HNF1382, HNF1383, HNF1384, HNF1385, HNF1386, HNF1387, HNF1388, HNF1389, HNF1390, HNF1391, HNF1392, HNF1393, HNF1394, HNF1395, HNF1396, HNF1397, HNF1398, HNF1399, HNF1400, HNF1401, HNF1402, HNF1403, HNF1404, HNF1405, HNF1406, HNF1407, HNF1408, HNF1409, HNF1410, HNF1411, HNF1412, HNF1413, HNF1414, HNF1415, HNF1416, HNF1417, HNF1418, HNF1419, HNF1420, HNF1421, HNF1422, HNF1423, HNF1424, HNF1425, HNF1426, HNF1427, HNF1428, HNF1429, HNF1430, HNF1431, HNF1432, HNF1433, HNF1434, HNF1435, HNF1436, HNF1437, HNF1438, HNF1439, HNF1440, HNF1441, HNF1442, HNF1443, HNF1444, HNF1445, HNF1446, HNF1447, HNF1448, HNF1449, HNF1450, HNF1451, HNF1452, HNF1453, HNF1454, HNF1455, HNF1456, HNF1457, HNF1458, HNF1459, HNF1460, HNF1461, HNF1462, HNF1463, HNF1464, HNF1465, HNF1466, HNF1467, HNF1468, HNF1469, HNF1470, HNF1471, HNF1472, HNF1473, HNF1474, HNF1475, HNF1476, HNF1477, HNF1478, HNF1479, HNF1480, HNF1481, HNF1482, HNF1483, HNF1484, HNF1485, HNF1486, HNF1487, HNF1488, HNF1489, HNF1490, HNF1491, HNF1492, HNF1493, HNF1494, HNF1495, HNF1496, HNF1497, HNF1498, HNF1499, HNF1500, HNF1501, HNF1502, HNF1503, HNF1504, HNF1505, HNF1506, HNF1507, HNF1508, HNF1509, HNF1510, HNF1511, HNF1512, HNF1513, HNF1514, HNF1515, HNF1516, HNF1517, HNF1518, HNF1519, HNF1520, HNF1521, HNF1522, HNF1523, HNF1524, HNF1525, HNF1526, HNF1527, HNF1528, HNF1529, HNF1530, HNF1531, HNF1532, HNF1533, HNF1534, HNF1535, HNF1536, HNF1537, HNF1538, HNF1539, HNF1540, HNF1541, HNF1542, HNF1543, HNF1544, HNF1545, HNF1546, HNF1547, HNF1548, HNF1549, HNF1550, HNF1551, HNF1552, HNF1553, HNF1554, HNF1555, HNF1556, HNF1557, HNF1558, HNF1559, HNF1560, HNF1561, HNF1562, HNF1563, HNF1564, HNF1565, HNF1566, HNF1567, HNF1568, HNF1569, HNF1570, HNF1571, HNF1572, HNF1573, HNF1574, HNF1575, HNF1576, HNF1577, HNF1578, HNF1579, HNF1580, HNF1581, HNF1582, HNF1583, HNF1584, HNF1585, HNF1586, HNF1587, HNF1588, HNF1589, HNF1590, HNF1591, HNF1592, HNF1593, HNF1594, HNF1595, HNF1596, HNF1597, HNF1598, HNF1599, HNF1600, HNF1601, HNF1602, HNF1603, HNF1604, HNF1605, HNF1606, HNF1607, HNF1608, HNF1609, HNF1610, HNF1611, HNF1612, HNF1613, HNF1614, HNF1615, HNF1616, HNF1617, HNF1618, HNF1619, HNF1620, HNF1621, HNF1622, HNF1623, HNF1624, HNF1625, HNF1626, HNF1627, HNF1628, HNF1629, HNF1630, HNF1631, HNF1632, HNF1633, HNF1634, HNF1635, HNF1636, HNF1637, HNF1638, HNF1639, HNF1640, HNF1641, HNF1642, HNF1643</p>						

Appellation approuvée de l'analyse ou maladie associée (avec numéro de phénotype MIM) / Approved test name or Associated Disease (with MIM Phenotype Number)	Code Test / Test Code (Dictionnaire MSSS)	SYMBOL HGNC DU GÈNE et NOM OMIM DU GÈNE / HGNC Gene Symbol and OMIM Gene Number	RÈGLES DE MANUTENTION / Handling Conditions	LABORATOIRE DÉSIGNÉ / Designated laboratory	RENSEIGNEMENTS SUR L'ANALYSE / Test details	MÉTHODE / Methode	NOTE PARTICULIÈRE / Special comment	Date de la révision / Date of revision
1,5-anhydroglucitol (GlycoMark) [500115] [GLYC]	1465		Gold Top or Red Top (Serum) or Lavender (plasma). Centrifuge and aliquot serum to transport tube within one hour of collection. Store and ship frozen. Stable at -20°C for 14 days.	LabCorp/Dynacare (https://www.labcorp.com/)		Enzymatic, colorimetric assay.		10-Jan-21
Leukemia, Philadelphia chromosome-positive, resistant to imatinib		ABL1 (109980)	Lavender Top (EDTA)	University Health Network (Toronto General Hospital)				
Leukemia/Lymphoma Immunophenotyping (PNH and ZAP-70 available) [2001]	1465		Na heparin tube. Store and send at RT. Send immediately overnight.	Hematologies, Inc. 3161 Elliot Ave. Suite 200, Seattle WA 98121 1800-860-0934		Flow cytometry		23-Jun-20
Leukodystrophy with dysmyelination and spastic paraparesis with or without dystonia (SPG35) (612319)	964	FA3H (611026)	Lavender Top (EDTA)	Centogene AG (www.centogene.com)		Sanger sequencing		
Leukotriène E4, 24 Hour, Urine [TL-TE4]	1465		Preferred: 24-hour urine collection Container/Tube: Plastic, 5-mL tube (T465) Specimen Volume: 5 mL Collection Instructions: 1. Collect urine for 24 hours.No preservative. 2. Refrigerate specimen during collection, aliquot 5 mL of urine into plastic tube, and send specimen refrigerated. Store frozen (30 days).	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)		LC-MS/MS	Random test available RLTE4	16-Jun-22
LIMB GIRDLE MUSCULAR DYSTROPHY PANEL [10401]	911	ANOF (60862); CAPN3 (114248); CAV3 (601258); DES (125646); DNAJB1 (611332); DYSF (603009); FKBP (605996); GMPFB (615320); ISPD (614631); LIMS2 (607906); LMNA (150336); MYOT (604163); PNPLA2 (606099); SGCA (600139); SGCB (603900); SGCD (601411); SGCG (608896); SMC2H1 (614982); TCAP (604488); TNOP1 (601032); TOR1AIP1 (614512); TRAPPC11 (644138); 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)	NextGen Sequencing (24 genes)		15-Mar-22
LIMB GIRDLE MUSCULAR DYSTROPHY TYPE 2B AND MIYOSHI MYOPATHY VIA THE DYSF GENE [3123]	606	DYSF (603009)	Lavender Top (EDTA) [2 tubes]	Prevention Genetics (www.preventiongenetics.com)	Limb-Girdle Muscular Dystrophy, Type 2B (253601); Miyoshi Myopathy (254120), Myopathy, Distal, With Anterior Tibial Onset (606768).			09-Mar-21

Appellation 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/HEC DU GÈNE et NOM OMIM DU GÈNE / HEC Gene Symbol and OMIM Gene Number	RÈGLES DE MANUTENTION / Handling Conditions	LABORATOIRE DÉSIGNÉ / Designated laboratory	RENSEIGNEMENTS SUR L'ANALYSE / Test details	MÉTHODE / Methode	NOTE PARTICULIÈRE / Special comment	Date de la révision / Date of revision	
I,5-anhydroglucitol (GlycoMark) [500115] [GLYC]	1465		Gold Top or Red Top (Serum) or Lavender (plasma). Centrifuge and aliquot serum to transport tube within one hour of collection. Store and ship frozen. Stable at -20°C for 14 days.	LabCorp/Dynacore (https://www.labcorp.com/)		Enzymatic, colorimetric assay.		10-Jan-21	
Lipid Metabolism Deficiency NGS Sequencing Panel (71 genes) + Del/Dup + mtDNA [NGS303]		ABCD1 (30017); ABCD2 (60180); ACAA1 (60454); ACAA2 (60470); ACACA (20030); ACACB (60557); ACAD10 (61181); ACAD11 (61428); ACAD9 (61103); ACADL (60976); ACADM (60786); ACADS (60685); ACADSB (60301); ACADVL (60975); ACAT1 (60780); ACAT2 (100678); ACLA1 (100728); ACO1 (61413); ACO12 (61415); ACO12 (60972); ACO14 (61416); ACO15 (61420); ACO17 (60287); ACOT8 (60812); ACOT9 (60862); ACOX1 (60971); ACOX2 (60641); ACOX3 (60340); ACP6 (61471); ACSBG1 (61492); ACSBG2 (61493); ACSF1 (60945); ACSF3 (61424); ACSL1 (15242); ACSL3 (60271); ACSL4 (60957); ACSL5 (60967); ACSL6 (60442); ACSM1 (61457); ACSMDA (61458); ACSMB (61459); ACSM1 (14505); ACSM4 (61456); ACSM5 (61461); ACSN1 (61455); ACSN2 (60952); ADHFE1 (61105); ALDH2A2 (60923); BAA1 (60288); BBOX1 (60212); BRL1 (60363); BTD (60609); CHKB (61295); CPT1A (60528); CPT1B (60187); CPTIC (60846); CPT2 (60950); CRAT (60184); CROT (60990); CYP6A11 (60110); DCR1 (22745); DHRX1 (61040); ECHS1 (60292); ECH1 (60305); ECI2 (60824); EBHADD1 (60707); ELOVL1 (61181); ELOVL2 (61184); ELOVL3 (61185); ELOVL4 (60512); ELOVL5 (61189); ELOVL6 (61154); ELOVL7 (61443); FAHE1 (60644); FAPB1 (13460); FAPB2 (13468); FAPB3 (13465); FAPB4 (60044); FAPB5 (60516); FAPB6 (60022); FAPB7 (60295); FAPB8 (60048); FAPB9 (60049); FAPB10 (60050); FASN (60012); GPAM (60295); GPI1 (13620); GPI2 (13618); HADH (60169); HADHA (60089); HADHB (14345); HLCS (60018); HMGCL1 (61388); HMGCS2 (60024); HSD17B2 (60974); LIPA (15170); MCAT (61479); MCR1 (60820); MLYCD1 (60670); OPA1 (60688); ONX1 (60424); ONX2 (61020); OXSM1 (60124); PECK (60843); PNPLA2 (60609); PPARGC1A (60457); PPARGC1B (60886); PRKAA1 (60279); PRKAA2 (60047); PTPN11 (61047); SCY2 (18475); SLC22A5 (60277); SLC22A10 (61688); SLC22A11 (60901); SLC22A12 (60124); SLC27A3 (60493); SLC27A4 (60494); SLC27A5 (60314); SLC27A6 (60496); SLC35A1 (60369); TECK (61007)	Lavender Top (EDTA)	MNG Laboratories (www.medicalneurogenetics.com)/Dynacore		NextGen Sequencing		09-May-20	
Lipodystrophy		ACRAT2 (60380); AKT1 (10471); BCL2L1 (60456); CAV1 (60047); CBEK1 (61210); LMNA (15010); PPARC (60187); PTF (60198); TRCD4 (61265); ZMPSTE24 (60680)	Lavender Top (2 x 4 mL)	Fulgent Genetics (fulgentdiagnostics.com)		NextGen Sequencing			
Lissencephaly NGS Panel		ACTB; ACTG1; ARX; DCX; FKBP; FKTN; LARGE; PPAH1B1; POMGNT1; POMT1; POMT2; RELN; TUBA1A; VLDLR	Lavender Top (2 x 4 mL)	Fulgent Genetics (fulgentdiagnostics.com)		NextGen Sequencing			
Loeys-Dietz Syndrome Panel [4289]	283	SMAD3; TGFBR2; TGFBR3; TGFBR1; TGFBR2	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)		Next Generation DNA Sequencing		28-Oct-21	
Long QT/Brugada Syndrome NGS Panel	285	AKAP9 (60401); ANK2 (10640); CACNA1C (11420); CAV3 (60123); KCNE1 (17626); KCNE2 (601796); KCNH2 (15247); KCNH3 (60086); KCNH4 (600734); KCNQ1 (60542); KCNQ2 (60256); KCNQ3 (60016); SNTA1 (60117)	Lavender Top (2 x 4 mL)	Fulgent Genetics (fulgentdiagnostics.com)	Long QT Syndrome (192900)	NextGen Sequencing		06-Jan-20	
LRBA	1248		Na heparin tube. Store and send at RT. Send immediately overnight.	Alberta Precision Laboratories (Flow Cytometry - Calgary)		Flow cytometry		01-Jun-21	
LRP4 Autoantibody Test [1483]	18		Serum (Gold SST or Red Top). Store at -20°C	Athena Diagnostics (www.athenadiagnostics.com)	For AcR and MUSK negative subjects	IFA		21-Aug-20	
Lurasidone [2543SP]	1688		Red Top or Lavender Top only. Store and send serum/plasma frozen.	NMS Laboratories		LC-MS/MS		17-Mar-22	
LYMPHEDEMA-DISTICHIASIS SYNDROME VIA THE FOXC2 GENE [280]		FOXC2 (60340)	Lavender top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Lymphedema-Distichiasis Syndrome (153400)	Sanger sequencing and del/dupl			
Lysosomal Disease (NextGen Sequencing Panel and Copy Number Analysis; 72 Genes) [NGS313]	474	ADAMTS12 (61277); AGA (61228); ANTXR2 (60841); ARSA (60754); ARSB (61542); ASAH1 (61348); ATP13A2 (61051); ATP1A (30011); ATP7B (60682); CERS1 (60919); CLN3 (607042); CLN5 (60812); CLN6 (60725); CLN8 (60787); COL11A1 (12098); COL11A1 (21049); CTS5 (60272); CTS5A (61311); CTS6 (60265); CTS6B (116840); CTS6 (60359); CTSK (601105); DHCY7 (60258); DNAAF2 (61210); DYM (60746); FUC1 (61250); GAA (60680); GALC (60990); GALNS (61222); GBA (60643); GBA1 (30044); GLB1 (61458); GMDA (61109); GNE (60824); GNPTAB (60740); GNPTG (60788); GNS (60764); GPC3 (30017); GRN (13045); GUSB (61149); HEXA (60689); HEXB (60673); HES6AT (61061); HRAS (60020); HVAL1 (60701); IIS1 (60821); IDUA (25280); ICTD7 (61175); IDMSA (30028); MLL2 (60211); LAMP2 (60960); LIPA (61347); LMBRD1 (61225); LYST (60897); MAN2B1 (60948); MAN2B2 (60949); MCK1 (60248); MPOB (61124); NAGA (60176); NAGL1 (60970); NEU1 (60872); NPC1 (60763); NPC2 (60103); PPT1 (60072); PSAP (17980); RAI1 (60742); SSB1 (60570); SLC17A5 (60432); SMD1 (60708); SMD2 (60709); TCF4 (60222); TFI1 (60706)	Lavender Top (EDTA)	MNG Laboratories (www.medicalneurogenetics.com)/Dynacore		Next Generation DNA Sequencing			09-May-20
Malignant Hyperthermia Susceptibility Panel [3285]	632	CACNA1S (11420); RYR1 (10961); STAC3		Invitae (www.invitae.com)		NextGen Sequencing		2022/09/12	
Mannose-Binding Lectin Deficiency NGS Test		MBL2 (15448)	Lavender Top (2 x 4 mL)	Fulgent Genetics (fulgentdiagnostics.com)	Mannose-binding protein deficiency (614372)	Next Generation DNA Sequencing		23-Oct-17	
MARFAN SYNDROME AND RELATED AORTOPATHIES PANEL [1212]	279		Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)		NextGen Sequencing		2021/12/07	
Marfan Syndrome and Thoracic Aortic Aneurysm and Dissection NGS Panel	279	ACTA2; CBS; COL1A1; COL5A1; COL5A2; FBN1; FBN2; FLNA; MED12; MYH11; MYLK; NOTCH1; NOTCH2; NOTCH3; PRKG1; SLC22A18; SMAD3; SMAD4; TGFBR2; TGFBR3; TGFBR1; TGFBR2 (22 genes)	Lavender Top (2 x 4 mL)	Fulgent Genetics (fulgentdiagnostics.com)	Marfan syndrome, type 1 (154700)	NextGen Sequencing		17-Feb-19	
MARINESCO-SJOGREN SYNDROME VIA THE SIL1 GENE [1674]		SIL1 (60805)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Marinesco-Sjogren Syndrome (248800)	Sanger sequencing			
Meckel-Gruber syndrome Sequencing Panel		CC2D2A (61013); CEP290 (61042); MKS (60883); RPRGRIP1 (61097); TCTN1 (61346); TMEM67 (60984); TMEM216 (61387)	Lavender Top (EDTA)	University of Chicago Genetic Services Laboratories (dnatesting.uchicago.edu)	Meckel Syndrome, Type 8 (613885); Meckel Syndrome, Type 6 (312284); Meckel Syndrome, Type 1 (249000); Meckel Syndrome, Type 4 (611134); Meckel Syndrome, Type 3 (607361); Meckel Syndrome, Type 5 (611561); Meckel syndrome 2 (603194)	Sanger sequencing			

Application 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 (Déclaratoire MSSS)	SYMBOL/BIEN DU GÈNE et NOM OMDM DU GÈNE / HGNC Gene Symbol/and OMDM Gene Number	RÈGLES DE MANUTENTION / Handling Conditions	LABORATOIRE DÉSIGNÉ / Designated laboratory	RENSEIGNEMENTS SUR L'ANALYSE / Test details	MÉTHODE / Methode	NOTE PARTICULIÈRE / Special comment	Date de la révision / Date of revision
1,5-anhydroglucitol (GlycoMark) [500115] [GLYC]	1465		Gold Top or Red Top (Serum) or Lavender (plasma). Centrifuge and aliquot serum to transport tube within one hour of collection. Store and ship frozen. Stable at -20°C for 14 days.	LabCorp/Dynacare (https://www.labcorp.com/)		Enzymatic, colorimetric assay.		10-Jan-21
MECP2 Analysis		MECP2 (60005)	Lavender Top (EDTA) 2-5 cc	Alberta Children's hospital http://www.medicalgenetics.ca/molecular.html	Ret's disorder (312750)	Sanger sequencing		04-May-17
Medullary Cystic Kidney Disease type 2 and Familial Juvenile Hyperuricemic Nephropathy type 1 via the UMOD Gene [1681]		UMOD (91845)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Familial Juvenile Hyperuricemic Nephropathy (162009); Medullary Cystic Kidney Disease 2(603860)	Sanger sequencing		15-Mar-07
Metabolic Hypoglycemia Sequencing Panel with CNV Detection [10365]	1517	ACADM 60708 ACADVL 60975 ACAT1 60789 ACSF1 61425 AGL 61060 ALDOB 61274 CASA 114761 DGUOK 601465 ETFA 60853 ETFB 13040 ETFDH 21675 FBP1 61570 G6PC 61342 GALT 60999 GK 30474 GYS2 13871 HADH 61069 HMOX1 61398 HMOX2 60234 MLYCD 606761 MPV17 13796 SNT 60789 OXCT1 610424 PC 60876 PKCI 61468 PKC2 61405 POM1 17190 PKAC 20078 PKB 17280 PHKG2 172471	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	3-Hydroxy-3-Methylglutaryl-CoA Lyase Deficiency AR 246450 Alpha-Methylacetoacetic Aciduria AR 203750 Combined Malonic Acid Methylmalonic Aciduria AR 614265 Congenital Disorder of Glycosylation, Type I AR 614921 Fanconi-Bickel Syndrome AR 227810 Fructose-Biphosphatase Deficiency AR 229700 Galactosemia AR 230400 Glycogen Storage Disease 0, Liver AR 240600 Glycogen Storage Disease Type Ia AR 232200 Glycogen Storage Disease Type Ib AR 232220 Glycogen Storage Disease Type Ic AR 232240 Glycogen Storage Disease Type III AR 232400 Glycogen Storage Disease Type IXa1 XL 306000 Glycogen Storage Disease Type IXc AR 613027 Glycogen Storage Disease Type VI AR 232700 Glycogen Storage Disease-Type IXb AR 261750 Hereditary Fructose Intolerance AR 229600 Malonyl-CoA Decarboxylase Deficiency AR 238360 Monocarboxylate Transporter 1 Deficiency AD AR 616095 Phosphoenolpyruvate Carboxykinase Deficiency, Cytosolic AR 261680 Phosphoenolpyruvate Carboxykinase Deficiency, Mitochondrial AR 261650 Pyruvate Carboxylase Deficiency AR 266150 Succinyl-CoA Acetoacetyltransferase Deficiency AR 245050	NextGen Sequencing		15-Aug-19
Metformin, Serum/Plasma [FMETF]			Red Top or Lavender Top only. Store and send serum/plasma frozen.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)				
Microcephaly NGS Panel	350	AKT3, ANKRD2, ARHGAP2, ASPM, ATR, ATRIP, BUB1B, CASK, CDC5RAP2, CDK6, CENPE, CENPF, CENPL, CEP135, CEP150, CEP350, CKA2L1, COX2B, CRIP1, DAAPH1, DNMT1, EFTUD2, HMOX3, IER3IP1, KAT5B1, KIF11, KNL1, LIGA, MEF2L, MED17, MESP2A, MIB1, MIB2, MIB3, MIB4, MIB5, MYCN, NDN, NDEL, NHEH1, NIN, NREL, PAFAH1B1, PCL9, PCNT, PHF1, PLEKHA7, PLK4, PNKP, POMT1, PPP1R15B, PQBP1, QARS, RARS2, RBBP9, RTTN, SASS6, SLC11A4, SLC25A9, SLC26A5, SPATA5, STAMBP, STIL, THOC3, TRMT10A, TSEN1, TSEN2, TSEN3A, TUBB3B, TUBGCPA, TUBGCPA, VRK1, WDR62, WDR73, XRCC4, ZEB2, ZNF338 (75 genes)	Lavender Top (EDTA)	Fulgent Genetics (fulgentdiagnostics.com)		NextGen Sequencing		23-Oct-17
Migraine Panel [NE1201]	581		Lavender Top (EDTA)	Blueprint Genetics http://blueprintgenetics.com/		Next-Generation sequencing		21-Aug-20
Minimal Residual Disease Testing (MRD) T cell [2002]	1084		Na heparin tube. Store and send at RT. Send immediately overnight.	Hematologies, Inc. 3161 Elliot Ave. Suite 200, Seattle WA 98121 1800-860-0934		Flow cytometry		23-Jun-20
Minimal Residual Disease Testing (MRD) T-ALL	1084		Na heparin tube. Store and send at RT. Send immediately overnight.	Johns Hopkins Medical Laboratories		Flow cytometry	Price can vary by number of markers	08-Feb-22
Minimal Residual Disease Testing for Acute Lymphoblastic Leukemia	1084		EDTA Whole Blood. Keep at room temperature.	Flow Cytometry Laboratory, London Health Sciences Centre.	For Friday samples email Ben Hedley with ID information & location where sample is being sent.	Flow cytometry		04-Aug-20
Mitochondrial Genome Sequencing [MLOL021]	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), MTRR (590005), MTN1 (590010), MTND1 (590015), MTTG (590020), MTTT (590025), MTTQ (590030), MTTI (590035), MTR (590040), MTL1 (590045), MTL2 (590050), MTK (590060), MTHA (590065), MTF (590070), MTFP (590075), MTFB1 (590080), MTFB2 (590085), MTF (590090), MTFB (590095), MTFY (590100), MTFV (590105), MTFN1 (516000), MTFND2 (516001), MTFND3 (516002), MTFND4 (516003), MTFND4L (516004), MTFND5 (516005), MTFND6 (516006), MYCYB (516020), MTFCO1 (516030), MTFCO2 (516040), MTFCO3 (516050), MTATP6 (516060), MTATP9 (516070).	Lavender Top (EDTA)	MNG Laboratories (www.medicalneurogenetics.com/)Dynacare	Mitochondrial DNA Deletion Syndromes Mitochondrial DNA Depletion Syndrome Mitochondrial DNA Depletion Syndrome Mitochondrial DNA Depletion Syndrome, Encephalomyopathic Form Mitochondrial DNA Depletion Syndrome, Encephalomyopathic Form Mitochondrial DNA Depletion Syndrome, MNGIE Form Mitochondrial DNA Depletion Syndrome, Hepatoencephalic Form Mitochondrial DNA Depletion Syndrome, MNGIE Form Mitochondrial DNA Depletion Syndrome, Myopathic Form mtDNA Deletion Syndromes	NextGen Sequencing		09-May-20

Appellation 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 HUGO DU GÈNE et NOM OMIM DU GÈNE / HUGO Gene Symbol and OMIM Gene Number	RÈGLES DE MANUTENTION / Handling Conditions	LABORATOIRE DÉSIGNÉ / Designated laboratory	RENSEIGNEMENTS SUR L'ANALYSE / Test details	MÉTHODE / Methode	NOTE PARTICULIÈRE / Special comment	Date de la révision / Date of revision
1,5-anhydroglucitol (GlycoMark) [500115] [GLYC]	1465		Gold Top or Red Top (Serum) or Lavender (plasma). Centrifuge and aliquot serum to transport tube within one hour of collection. Store and ship frozen. Stable at -20°C for 14 days.	LabCorp/Dynacare (https://www.labcorp.com/)		Enzymatic, colorimetric assay.		10-Jan-21
Mitochondrial Genome Sequencing + Deletion Analysis [MOL189]	518	MTRNR1 (561000), MTRNR2 (561010), MTTA (590000), MTR (590005), MTTN (590010), MTTD (590015), MTTK (590020), MTH (590025), MTTQ (590030), MTD (590035), MTH (590040), MTT (590045), MTL1 (590050), MTL2 (590055), MTK (590060), MTTM (590065), MTF (590070), MTP (590075), MTS1 (590080), MTS2 (590085), MTT (590090), MTTW (590095), MTTV (590100), MTTX (590105), MTD1 (516000), MTD2 (516005), MTD3 (516010), MTD4 (516015), MTD5 (516020), MTD6 (516025), MTCYB (516030), MTCO1 (516035), MTCO2 (516040), MTCO3 (516045), MTA1P6 (516050), MTA1P9 (516070).	Blood Fibroblasts Muscle Extracted DNA Buccal Cells	MNG Laboratories (www.medicalneurogenetics.com/) / Dynacare	Leber hereditary optic neuropathy (LHON); Neuropath, ataxia, and retinitis pigmentosa (NARP); Myoclonic epilepsy associated with ragged-red fibers (MERRF); Mitochondrial myopathy, encephalopathy, lactic acidosis, and stroke-like episodes (MELAS)	Deletion; Capillary DNA Sequencing		14-May-20
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), MTR (590005), MTH (590010), MTD (590035), MTK (590060), MTL1 (590050), MTL2 (590055), MTTQ (590030), MTTG (590035), MTH (590040), MTT (590045), MTL1 (590050), MTL2 (590055), MTK (590060), MTTM (590065), MTF (590070), MTP (590075), MTS1 (590080), MTS2 (590085), MTT (590090), MTTW (590095), MTTV (590100), MTTX (590105), MTD1 (516000), MTD2 (516005), MTD3 (516010), MTD4 (516015), MTD5 (516020), MTD6 (516025), MTCYB (516030), MTCO1 (516035), MTCO2 (516040), MTCO3 (516045), MTA1P6 (516050), MTA1P9 (516070).	Lavender Top (EDTA)	MNG Laboratories (www.medicalneurogenetics.com/) / Dynacare	Mitochondrial diseases including: Leber hereditary optic neuropathy (LHON); Neuropath, ataxia, and retinitis pigmentosa (NARP); Myoclonic epilepsy associated with ragged-red fibers (MERRF); Mitochondrial myopathy, encephalopathy, lactic acidosis, and stroke-like episodes (MELAS)	Deletion; Real-time Quantitative PCR analysis; Sequencing		14-May-20
Mitochondrial Genome Sequencing + Deletion Analysis + Depletion Testing (Muscle) [MOL340]	518	The human mitochondrial genome is 16569 base pairs in length and encodes 37 genes including 2 ribosomal RNA genes, 22 transfer RNA genes, and 13 protein-coding genes. Our mitochondrial DNA (mtDNA) panel includes complete sequencing of the following genes (MIM#): MTRNR1 (561000), MTRNR2 (561010), MTTA (590000), MTR (590005), MTH (590010), MTD (590035), MTK (590060), MTL1 (590050), MTL2 (590055), MTTQ (590030), MTTG (590035), MTH (590040), MTT (590045), MTL1 (590050), MTL2 (590055), MTK (590060), MTTM (590065), MTF (590070), MTP (590075), MTS1 (590080), MTS2 (590085), MTT (590090), MTTW (590095), MTTV (590100), MTTX (590105), MTD1 (516000), MTD2 (516005), MTD3 (516010), MTD4 (516015), MTD5 (516020), MTD6 (516025), MTCYB (516030), MTCO1 (516035), MTCO2 (516040), MTCO3 (516045), MTA1P6 (516050), MTA1P9 (516070).	50-75 milligrams muscle snap frozen in liquid nitrogen and maintained at -80°Celsius or below. (2) DNA extracted from muscle	MNG Laboratories (www.medicalneurogenetics.com/) / Dynacare	Mitochondrial diseases including: Leber hereditary optic neuropathy (LHON) Neuropath, ataxia, and retinitis pigmentosa (NARP) Myoclonic epilepsy associated with ragged-red fibers (MERRF) Mitochondrial myopathy, encephalopathy, lactic acidosis, and stroke-like episodes (MELAS)	Polymerase Chain Reaction (PCR) followed by DNA sequencing analysis; Polymerase Chain Reaction (PCR) followed by Restriction Endonuclease digestion and gel separation; Real-time Quantitative PCR analysis		14-May-20
MLPA (screen for deletions of CFHR1-CFHR3)		CFHR1 (134371); CFHR3 (605350)	Lavender Top (EDTA)	Molecular Otolaryngology & Renal Research Laboratory, U. of Iowa Carver College of Medicine (www.medicine.uiowa.edu/mor/)	Hemolytic uremic syndrome, atypical; susceptibility to (235400)	MLPA		31-Oct-17
MODY Neonatal Diabetes NGS Panel	481	ALX1, AKT2, BDK, CEL, CSD2, CP, EIF2AK3, FOXO1, GATA4, GSK, GLIS3, GLE1, HADH, HNF1A, HNF1B, HNF1A, HNF1B, INS, INS2, KCNJ11, KLF11, NEUROD1, NEUROG3, PAX4, PDX1, PTF1A, RFX5, SLC22A2, WFS1, ZFP57 (30 genes)	Lavender Top (2 x 4 mL)	Fulgent Genetics (fulgentdiagnostics.com)		NextGen Sequencing		13-May-19
Mold Mix 1	1465		Serum (Gold SST or Red Top)	In-Common Laboratories				12-Mar-20
Mold Mix 2	1465		Serum (Gold SST or Red Top)	In-Common Laboratories				12-Mar-20
Molecular Testing for Lissencephaly		DCX (30012)	Lavender Top (EDTA)	University of Chicago Genetic Services Laboratories (dnatesting.uchicago.edu)	Lissencephaly, X-linked (300067); Subcortical laminar heteropia, X-link (300067); testing for a known mutation	Sanger sequencing		
Monogenic Autoimmunity Panel [08150]	817	ACPS1, ADA2, ADA3, AICDA, AIRE, AP3B1, BLOC1B6, BTK, CASP10, CASP9, CD27, CD40LG, CR2, CTLA4, CYBA, CYBB, DOCK8, FADD, FAS, FASLG, FOXP3, ICOS1, IHH1, IL10L1, IL10RA, IL10RB, IL21, IL21R, IL2RA, ITCH, ITR, IKBALYST, MAGT1, NCF2, NCF4, NFKB1, NFKB2, NFKB3, ORAI1, PRKCE, PRKCH, PRKCI, PLEKHA7, PLEKHA9, PRKCD, RAB27A, RAC3, RFX5, RFXANK, RFXAP, RMRP, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, SH2B3, SLC7A7, STAT1, STAT3, STAT5B, STIM1, STX11, STXAP3E, TRAF3, TRAF6, TNFRSF10B, TNFRSF10C, TNFRSF12, TNFRSF21, TRIM3, UNCLDUNG, WAS, XIAP	Lavender Top (EDTA) [2 tubes]	Invitae (www.invitae.com)		NextGen Sequencing		20-Feb-20

Appellation 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 (Déclaratoire MSSS)	SYMBOL HGNC DU GÈNE et NOM OMIM DU GÈNE / HGNC Gene Symbol and OMIM Gene Number	RÈGLES DE MANUTENTION / Handling Conditions	LABORATOIRE DÉSIGNÉ / Designated laboratory	RENSEIGNEMENTS SUR L'ANALYSE / Test details	MÉTHODE / Methode	NOTE PARTICULIÈRE / Special comment	Date de la révision / Date of revision
1,5-anhydroglucitol (GlycoMark) [500115] [GLYC]	1465		Gold Top or Red Top (Serum) or Lavender (plasma). Centrifuge and aliquot serum to transport tube within one hour of collection. Store and ship frozen. Stable at -20°C for 14 days.	LabCorp/Dynacare (https://www.labcorp.com/)		Enzymatic, colorimetric assay.		10-Jan-21
Monogenic Inflammatory Bowel Disease Panel (Invitae) [08122]	1151	ADA, ADAM17, AICDA, BTK, CD3G, CD40LG, CTLA4, CYBA, CYBB, DCLRE1C, DOCK8, FOXP3, GATA3, FOXP3, IL10, IL13, IL13RA, IL13RB, IL21, IL2RA, IL2RG, IFNG, IL1A, IL1B, IL1E, IL1F3, IL1F5, IL1F6, IL1F7, IL1F8, IL1F9, IL1F10, IL1F11, IL1F12, IL1F13, IL1F14, IL1F15, IL1F16, IL1F17, IL1F18, IL1F19, IL1F20, IL1F21, IL1F22, IL1F23, IL1F24, IL1F25, IL1F26, IL1F27, IL1F28, IL1F29, IL1F30, IL1F31, IL1F32, IL1F33, IL1F34, IL1F35, IL1F36, IL1F37, IL1F38, IL1F39, IL1F40, IL1F41, IL1F42, IL1F43, IL1F44, IL1F45, IL1F46, IL1F47, IL1F48, IL1F49, IL1F50, IL1F51, IL1F52, IL1F53, IL1F54, IL1F55, IL1F56, IL1F57, IL1F58, IL1F59, IL1F60, IL1F61, IL1F62, IL1F63, IL1F64, IL1F65, IL1F66, IL1F67, IL1F68, IL1F69, IL1F70, IL1F71, IL1F72, IL1F73, IL1F74, IL1F75, IL1F76, IL1F77, IL1F78, IL1F79, IL1F80, IL1F81, IL1F82, IL1F83, IL1F84, IL1F85, IL1F86, IL1F87, IL1F88, IL1F89, IL1F90, IL1F91, IL1F92, IL1F93, IL1F94, IL1F95, IL1F96, IL1F97, IL1F98, IL1F99, IL1F100, IL1F101, IL1F102, IL1F103, IL1F104, IL1F105, IL1F106, IL1F107, IL1F108, IL1F109, IL1F110, IL1F111, IL1F112, IL1F113, IL1F114, IL1F115, IL1F116, IL1F117, IL1F118, IL1F119, IL1F120, IL1F121, IL1F122, IL1F123, IL1F124, IL1F125, IL1F126, IL1F127, IL1F128, IL1F129, IL1F130, IL1F131, IL1F132, IL1F133, IL1F134, IL1F135, IL1F136, IL1F137, IL1F138, IL1F139, IL1F140, IL1F141, IL1F142, IL1F143, IL1F144, IL1F145, IL1F146, IL1F147, IL1F148, IL1F149, IL1F150, IL1F151, IL1F152, IL1F153, IL1F154, IL1F155, IL1F156, IL1F157, IL1F158, IL1F159, IL1F160, IL1F161, IL1F162, IL1F163, IL1F164, IL1F165, IL1F166, IL1F167, IL1F168, IL1F169, IL1F170, IL1F171, IL1F172, IL1F173, IL1F174, IL1F175, IL1F176, IL1F177, IL1F178, IL1F179, IL1F180, IL1F181, IL1F182, IL1F183, IL1F184, IL1F185, IL1F186, IL1F187, IL1F188, IL1F189, IL1F190, IL1F191, IL1F192, IL1F193, IL1F194, IL1F195, IL1F196, IL1F197, IL1F198, IL1F199, IL1F200, IL1F201, IL1F202, IL1F203, IL1F204, IL1F205, IL1F206, IL1F207, IL1F208, IL1F209, IL1F210, IL1F211, IL1F212, IL1F213, IL1F214, IL1F215, IL1F216, IL1F217, IL1F218, IL1F219, IL1F220, IL1F221, IL1F222, IL1F223, IL1F224, IL1F225, IL1F226, IL1F227, IL1F228, IL1F229, IL1F230, IL1F231, IL1F232, IL1F233, IL1F234, IL1F235, IL1F236, IL1F237, IL1F238, IL1F239, IL1F240, IL1F241, IL1F242, IL1F243, IL1F244, IL1F245, IL1F246, IL1F247, IL1F248, IL1F249, IL1F250, IL1F251, IL1F252, IL1F253, IL1F254, IL1F255, IL1F256, IL1F257, IL1F258, IL1F259, IL1F260, IL1F261, IL1F262, IL1F263, IL1F264, IL1F265, IL1F266, IL1F267, IL1F268, IL1F269, IL1F270, IL1F271, IL1F272, IL1F273, IL1F274, IL1F275, IL1F276, IL1F277, IL1F278, IL1F279, IL1F280, IL1F281, IL1F282, IL1F283, IL1F284, IL1F285, IL1F286, IL1F287, IL1F288, IL1F289, IL1F290, IL1F291, IL1F292, IL1F293, IL1F294, IL1F295, IL1F296, IL1F297, IL1F298, IL1F299, IL1F300, IL1F301, IL1F302, IL1F303, IL1F304, IL1F305, IL1F306, IL1F307, IL1F308, IL1F309, IL1F310, IL1F311, IL1F312, IL1F313, IL1F314, IL1F315, IL1F316, IL1F317, IL1F318, IL1F319, IL1F320, IL1F321, IL1F322, IL1F323, IL1F324, IL1F325, IL1F326, IL1F327, IL1F328, IL1F329, IL1F330, IL1F331, IL1F332, IL1F333, IL1F334, IL1F335, IL1F336, IL1F337, IL1F338, IL1F339, IL1F340, IL1F341, IL1F342, IL1F343, IL1F344, IL1F345, IL1F346, IL1F347, IL1F348, IL1F349, IL1F350, IL1F351, IL1F352, IL1F353, IL1F354, IL1F355, IL1F356, IL1F357, IL1F358, IL1F359, IL1F360, IL1F361, IL1F362, IL1F363, IL1F364, IL1F365, IL1F366, IL1F367, IL1F368, IL1F369, IL1F370, IL1F371, IL1F372, IL1F373, IL1F374, IL1F375, IL1F376, IL1F377, IL1F378, IL1F379, IL1F380, IL1F381, IL1F382, IL1F383, IL1F384, IL1F385, IL1F386, IL1F387, IL1F388, IL1F389, IL1F390, IL1F391, IL1F392, IL1F393, IL1F394, IL1F395, IL1F396, IL1F397, IL1F398, IL1F399, IL1F400, IL1F401, IL1F402, IL1F403, IL1F404, IL1F405, IL1F406, IL1F407, IL1F408, IL1F409, IL1F410, IL1F411, IL1F412, IL1F413, IL1F414, IL1F415, IL1F416, IL1F417, IL1F418, IL1F419, IL1F420, IL1F421, IL1F422, IL1F423, IL1F424, IL1F425, IL1F426, IL1F427, IL1F428, IL1F429, IL1F430, IL1F431, IL1F432, IL1F433, IL1F434, IL1F435, IL1F436, IL1F437, IL1F438, IL1F439, IL1F440, IL1F441, IL1F442, IL1F443, IL1F444, IL1F445, IL1F446, IL1F447, IL1F448, IL1F449, IL1F450, IL1F451, IL1F452, IL1F453, IL1F454, IL1F455, IL1F456, IL1F457, IL1F458, IL1F459, IL1F460, IL1F461, IL1F462, IL1F463, IL1F464, IL1F465, IL1F466, IL1F467, IL1F468, IL1F469, IL1F470, IL1F471, IL1F472, IL1F473, IL1F474, IL1F475, IL1F476, IL1F477, IL1F478, IL1F479, IL1F480, IL1F481, IL1F482, IL1F483, IL1F484, IL1F485, IL1F486, IL1F487, IL1F488, IL1F489, IL1F490, IL1F491, IL1F492, IL1F493, IL1F494, IL1F495, IL1F496, IL1F497, IL1F498, IL1F499, IL1F500, IL1F501, IL1F502, IL1F503, IL1F504, IL1F505, IL1F506, IL1F507, IL1F508, IL1F509, IL1F510, IL1F511, IL1F512, IL1F513, IL1F514, IL1F515, IL1F516, IL1F517, IL1F518, IL1F519, IL1F520, IL1F521, IL1F522, IL1F523, IL1F524, IL1F525, IL1F526, IL1F527, IL1F528, IL1F529, IL1F530, IL1F531, IL1F532, IL1F533, IL1F534, IL1F535, IL1F536, IL1F537, IL1F538, IL1F539, IL1F540, IL1F541, IL1F542, IL1F543, IL1F544, IL1F545, IL1F546, IL1F547, IL1F548, IL1F549, IL1F550, IL1F551, IL1F552, IL1F553, IL1F554, IL1F555, IL1F556, IL1F557, IL1F558, IL1F559, IL1F560, IL1F561, IL1F562, IL1F563, IL1F564, IL1F565, IL1F566, IL1F567, IL1F568, IL1F569, IL1F570, IL1F571, IL1F572, IL1F573, IL1F574, IL1F575, IL1F576, IL1F577, IL1F578, IL1F579, IL1F580, IL1F581, IL1F582, IL1F583, IL1F584, IL1F585, IL1F586, IL1F587, IL1F588, IL1F589, IL1F590, IL1F591, IL1F592, IL1F593, IL1F594, IL1F595, IL1F596, IL1F597, IL1F598, IL1F599, IL1F600, IL1F601, IL1F602, IL1F603, IL1F604, IL1F605, IL1F606, IL1F607, IL1F608, IL1F609, IL1F610, IL1F611, IL1F612, IL1F613, IL1F614, IL1F615, IL1F616, IL1F617, IL1F618, IL1F619, IL1F620, IL1F621, IL1F622, IL1F623, IL1F624, IL1F625, IL1F626, IL1F627, IL1F628, IL1F629, IL1F630, IL1F631, IL1F632, IL1F633, IL1F634, IL1F635, IL1F636, IL1F637, IL1F638, IL1F639, IL1F640, IL1F641, IL1F642, IL1F643, IL1F644, IL1F645, IL1F646, IL1F647, IL1F648, IL1F649, IL1F650, IL1F651, IL1F652, IL1F653, IL1F654, IL1F655, IL1F656, IL1F657, IL1F658, IL1F659, IL1F660, IL1F661, IL1F662, IL1F663, IL1F664, IL1F665, IL1F666, IL1F667, IL1F668, IL1F669, IL1F670, IL1F671, IL1F672, IL1F673, IL1F674, IL1F675, IL1F676, IL1F677, IL1F678, IL1F679, IL1F680, IL1F681, IL1F682, IL1F683, IL1F684, IL1F685, IL1F686, IL1F687, IL1F688, IL1F689, IL1F690, IL1F691, IL1F692, IL1F693, IL1F694, IL1F695, IL1F696, IL1F697, IL1F698, IL1F699, IL1F700, IL1F701, IL1F702, IL1F703, IL1F704, IL1F705, IL1F706, IL1F707, IL1F708, IL1F709, IL1F710, IL1F711, IL1F712, IL1F713, IL1F714, IL1F715, IL1F716, IL1F717, IL1F718, IL1F719, IL1F720, IL1F721, IL1F722, IL1F723, IL1F724, IL1F725, IL1F726, IL1F727, IL1F728, IL1F729, IL1F730, IL1F731, IL1F732, IL1F733, IL1F734, IL1F735, IL1F736, IL1F737, IL1F738, IL1F739, IL1F740, IL1F741, IL1F742, IL1F743, IL1F744, IL1F745, IL1F746, IL1F747, IL1F748, IL1F749, IL1F750, IL1F751, IL1F752, IL1F753, IL1F754, IL1F755, IL1F756, IL1F757, IL1F758, IL1F759, IL1F760, IL1F761, IL1F762, IL1F763, IL1F764, IL1F765, IL1F766, IL1F767, IL1F768, IL1F769, IL1F770, IL1F771, IL1F772, IL1F773, IL1F774, IL1F775, IL1F776, IL1F777, IL1F778, IL1F779, IL1F780, IL1F781, IL1F782, IL1F783, IL1F784, IL1F785, IL1F786, IL1F787, IL1F788, IL1F789, IL1F790, IL1F791, IL1F792, IL1F793, IL1F794, IL1F795, IL1F796, IL1F797, IL1F798, IL1F799, IL1F800, IL1F801, IL1F802, IL1F803, IL1F804, IL1F805, IL1F806, IL1F807, IL1F808, IL1F809, IL1F810, IL1F811, IL1F812, IL1F813, IL1F814, IL1F815, IL1F816, IL1F817, IL1F818, IL1F819, IL1F820, IL1F821, IL1F822, IL1F823, IL1F824, IL1F825, IL1F826, IL1F827, IL1F828, IL1F829, IL1F830, IL1F831, IL1F832, IL1F833, IL1F834, IL1F835, IL1F836, IL1F837, IL1F838, IL1F839, IL1F840, IL1F841, IL1F842, IL1F843, IL1F844, IL1F845, IL1F846, IL1F847, IL1F848, IL1F849, IL1F850, IL1F851, IL1F852, IL1F853, IL1F854, IL1F855, IL1F856, IL1F857, IL1F858, IL1F859, IL1F860, IL1F861, IL1F862, IL1F863, IL1F864, IL1F865, IL1F866, IL1F867, IL1F868, IL1F869, IL1F870, IL1F871, IL1F872, IL1F873, IL1F874, IL1F875, IL1F876, IL1F877, IL1F878, IL1F879, IL1F880, IL1F881, IL1F882, IL1F883, IL1F884, IL1F885, IL1F886, IL1F887, IL1F888, IL1F889, IL1F890, IL1F891, IL1F892, IL1F893, IL1F894, IL1F895, IL1F896, IL1F897, IL1F898, IL1F899, IL1F900, IL1F901, IL1F902, IL1F903, IL1F904, IL1F905, IL1F906, IL1F907, IL1F908, IL1F909, IL1F910, IL1F911, IL1F912, IL1F913, IL1F914, IL1F915, IL1F916, IL1F917, IL1F918, IL1F919, IL1F920, IL1F921, IL1F922, IL1F923, IL1F924, IL1F925, IL1F926, IL1F927, IL1F928, IL1F929, IL1F930, IL1F931, IL1F932, IL1F933, IL1F934, IL1F935, IL1F936, IL1F937, IL1F938, IL1F939, IL1F940, IL1F941, IL1F942, IL1F943, IL1F944, IL1F945, IL1F946, IL1F947, IL1F948, IL1F949, IL1F950, IL1F951, IL1F952, IL1F953, IL1F954, IL1F955, IL1F956, IL1F957, IL1F958, IL1F959, IL1F960, IL1F961, IL1F962, IL1F963, IL1F964, IL1F965, IL1F966, IL1F967, IL1F968, IL1F969, IL1F970, IL1F971, IL1F972, IL1F973, IL1F974, IL1F975, IL1F976, IL1F977, IL1F978, IL1F979, IL1F980, IL1F981, IL1F982, IL1F983, IL1F984, IL1F985, IL1F986, IL1F987, IL1F988, IL1F989, IL1F990, IL1F991, IL1F992, IL1F993, IL1F994, IL1F995, IL1F996, IL1F997, IL1F998, IL1F999, IL1F1000, IL1F1001, IL1F1002, IL1F1003, IL1F1004, IL1F1005, IL1F1006, IL1F1007, IL1F1008, IL1F1009, IL1F1010, IL1F1011, IL1F1012, IL1F1013, IL1F1014, IL1F1015, IL1F1016, IL1F1017, IL1F1018, IL1F1019, IL1F1020, IL1F1021, IL1F1022, IL1F1023, IL1F1024, IL1F1025, IL1F1026, IL1F1027, IL1F1028, IL1F1029, IL1F1030, IL1F1031, IL1F1032, IL1F1033, IL1F1034, IL1F1035, IL1F1036, IL1F1037, IL1F1038, IL1F1039, IL1F1040, IL1F1041, IL1F1042, IL1F1043, IL1F1044, IL1F1045, IL1F1046, IL1F1047, IL1F1048, IL1F1049, IL1F1050, IL1F1051, IL1F1052, IL1F1053, IL1F1054, IL1F1055, IL1F1056, IL1F1057, IL1F1058, IL1F1059, IL1F1060, IL1F1061, IL1F1062, IL1F1063, IL1F1064, IL1F1065, IL1F1066, IL1F1067, IL1F1068, IL1F1069, IL1F1070, IL1F1071, IL1F1072, IL1F1073, IL1F1074, IL1F1075, IL1F1076, IL1F1077, IL1F1078, IL1F1079, IL1F1080, IL1F1081, IL1F1082, IL1F1083, IL1F1084, IL1F1085, IL1F1086, IL1F1087, IL1F1088, IL1F1089, IL1F1090, IL1F1091, IL1F1092, IL1F1093, IL1F1094, IL1F1095, IL1F1096, IL1F1097, IL1F1098, IL1F1099, IL1F1100, IL1F1101, IL1F1102, IL1F1103, IL1F1104, IL1F1105, IL1F1106, IL1F1107, IL1F1108, IL1F1109, IL1F1110, IL1F1111, IL1F1112, IL1F1113, IL1F1114, IL1F1115, IL1F1116, IL1F1117, IL1F1118, IL1F1119, IL1F1120, IL1F1121, IL1F1122, IL1F1123, IL1F1124, IL1F1125, IL1F1126, IL1F1127, IL1F1128, IL1F1129, IL1F1130, IL1F1131, IL1F1132, IL1F1133, IL1F1134, IL1F1135, IL1F1136, IL1F1137, IL1F1138, IL1F1139, IL1F1140, IL1F1141, IL1F1142, IL1F1143, IL1F1144, IL1F1145, IL1F1146, IL1F1147, IL1F1148, IL1F1149, IL1F1150, IL1F1151, IL1F1152, IL1F1153, IL1F1154, IL1F1155, IL1F1156, IL1F1157, IL1F1158, IL1F1159, IL1F1160, IL1F1161, IL1F1162, IL1F1163, IL1F1164, IL1F1165, IL1F1166, IL1F1167, IL1F1168, IL1F1169, IL1F1170, IL1F1171, IL1F1172, IL1F1173, IL1F1174, IL1F1175, IL1F1176, IL1F1177, IL1F1178, IL1F1179, IL1F1180, IL1F1181, IL1F1182, IL1F1183, IL1F1184, IL1F1185, IL1F1186, IL1F1187, IL1F1188, IL1F1189, IL1F1190, IL1F1191, IL1F1192, IL1F1193, IL1F1194, IL1F1195, IL1F1196, IL1F1197, IL1F1198, IL1F1199, IL1F1200, IL1F1201, IL1F1202, IL1F1203, IL1F1204, IL1F1205, IL1F1206, IL1F1207, IL1F1208, IL1F1209, IL1F1210, IL1F1211, IL1F1212, IL1F1213, IL1F1214, IL1F1215, IL1F1216, IL1F1217, IL1F1218, IL1F1219, IL1F1220, IL1F1221, IL1F1222, IL1F1223, IL1F1224, IL1F1225, IL1F1226, IL1F1227, IL1F1228, IL1F1229, IL1F1230, IL1F1231, IL1F1232, IL1F1233, IL1F1234, IL1F1235, IL1F1236, IL1F1237, IL1F1238, IL1F1239, IL1F1240, IL1F1241, IL1F1242, IL1F1243, IL1F1244, IL1F1245, IL1F1246, IL1F1247, IL1F1248, IL1F1249, IL1F1250, IL1F1251, IL1F1252, IL1F1253, IL1F1254, IL1F1255, IL1F1256, IL1F1257, IL1F1258, IL1F1259, IL1F1260, IL1F1261, IL1F1262, IL1F1263, IL1F1264, IL1F1265, IL1F1266, IL1F1267, IL1F1268, IL1F1269, IL1F1270, IL1F1271, IL1F1272, IL1F1273, IL1F1274, IL1F1275, IL1F1276, IL1F1277, IL1F1278, IL1F1279, IL1F1280, IL1F1281, IL1F1282, IL1F1283, IL1F1284, IL1F1285, IL1F1286, IL1F1287, IL1F1288, IL1F1289, IL1F1290, IL1F1291, IL1F1292, IL1F1293, IL1F1294, IL1F1295, IL1F1296, IL1F1297, IL1F1298, IL1F1299, IL1F1300, IL1F1301, IL1F1302, IL1F1303, IL1F1304, IL1F1305, IL1F1306, IL1F1307, IL1F1308, IL1F1309, IL1F1310, IL1F1311, IL1F1312, IL1F1313, IL1F1314, IL1F1315, IL1F1316, IL1F1317, IL1F1318, IL1F1319, IL1F1320, IL1F1321, IL1F1322, IL1F1323, IL1F1324, IL1F1325, IL1F1326, IL1F1327, IL1F1328, IL1F1329, IL1F1330, IL1F1331, IL1F1332, IL1F1333, IL1F1334, IL1F1335, IL1F1336, IL1F1337, IL1F1338, IL1F1339, IL1F1340, IL1F1341, IL1F1342, IL1F1343, IL1F1344, IL1F1345, IL1F1346, IL1F1347, IL1F1348, IL1F1349, IL1F1350, IL1F1351, IL1F1352, IL1F1353, IL1F1354, IL1F1355, IL1F1356, IL1F1357, IL1F1358, IL1F1359, IL1F1360, IL1F1361, IL1F1362, IL1F1363, IL1F1364, IL1F1365, IL1F1366, IL1F1367, IL1F1368, IL1F1369, IL1F1370, IL1F1371, IL1F1372, IL1F1373, IL1F1374, IL1F1375, IL1F1376, IL1F1377, IL1F1378, IL1F1379, IL1F1380, IL1F1381, IL1F1382, IL1F1383, IL1F1384, IL1F1385, IL1F1386, IL1F1387, IL1F1388, IL1F1389, IL1F1390, IL1F1391, IL1F1392, IL1F1393, IL1F1394, IL1F1395, IL1F1396, IL1F1397, IL1F1398, IL1F1399, IL1F1400, IL1F1401, IL1F1402, IL1F1403, IL1F1404, IL1F1405, IL1F1406, IL1F1407, IL1F1408, IL1F1409, IL1F1410, IL1F1411, IL1F1412, IL1F1413, IL1F1414, IL1F1415, IL1F1416, IL1F1417, IL1F1418, IL1F1419, IL1F1420, IL1F1421, IL1F1422, IL1F1423, IL1F1424, IL1F1425, IL1F1426, IL1F1427, IL1F1428, IL1F1429, IL1F1430, IL1F1431, IL1F1432, IL1F1433, IL1F1434, IL1F1435, IL1F1436, IL1F1437, IL1F1438, IL1F1439, IL1F1440, IL1F1441, IL1F1442, IL1F1443, IL1F1444, IL1F1445, IL1F1446, IL1F1447, IL1F1448, IL1F1449, IL1F1450, IL1F1451, IL1F1452, IL1F1453, IL1F1454, IL1F1455, IL1F1456, IL1F1457, IL1F1458, IL1F1459, IL1F1460, IL1F1461, IL1F1462, IL1F1463, IL1F1464, IL1F1465, IL1F1466, IL1F1467, IL1F1468, IL1F1469, IL1F1470, IL1F1471, IL1F1472, IL1F1473, IL1F1474, IL1F1475, IL1F1476, IL1F1477, IL1F1478, IL1F1479, IL1F1480, IL1F1481, IL1F1482, IL1F1483, IL1F1484, IL1F1485, IL1F1486, IL1F1487, IL1F1488, IL1F1489, IL1F1490, IL1F1491, IL1F1492, IL1F1493, IL1F1494, IL1F1495, IL1F1496, IL1F1497, IL1F1498, IL1F1499, IL1F1500, IL1F1501, IL1F1502, IL1F1503, IL1F1504, IL1F1505, IL1F1506, IL1F1507, IL1F1508, IL1F1509, IL1F1510, IL1F1511, IL1F1512, IL1F1513, IL1F1514, IL1F1515, IL1F1516, IL1F1517, IL1F1518, IL1F1519, IL1F1520, IL1F1521, IL1F1522, IL1F1523, IL1F1524, IL1F1525, IL1F1526, IL1F1527, IL1F1528, IL1F1529, IL1F1530, IL1F1531, IL1F1532, IL1F1533, IL1F1534, IL1F1535, IL1F1536, IL1F1537, IL1F1538, IL1F1539, IL1F1540, IL1F1541, IL1F1542, IL1F1543, IL1F1544, IL1F1545, IL1F1546, IL1F1547, IL1F1548, IL1F1549, IL1F1550, IL1F1551, IL1F1552, IL1F1553, IL1F1554, IL1F1555, IL1F1556, IL1F1557, IL1F1558, IL1F1559, IL1F1560, IL1F1561, IL1F1562, IL1F1563, IL1F1564, IL1F1565, IL1F1566, IL1F1567, IL1F1568, IL1F1569, IL1F1570, IL1F1571, IL1F1572, IL1F1573, IL1F1574, IL1F1575, IL1F1576, IL1F1577, IL1F1578, IL1F1579, IL1F1580, IL1F1581, IL1F1582, IL1F1583, IL1F1584, IL1F1585, IL1F1586, IL1F1587, IL1F1588, IL1F1589, IL1F1590, IL1F1591, IL1F1592, IL1F159						

Appellation 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 (Déclencheur MSSS)	SYMBOL / IGENC DU GÈNE / NOM OMDM DU GÈNE / IGENC Gene Symbol and OMDM Gene Name	RÈGLES DE MANUTENTION / Handling Conditions	LABORATOIRE DÉSIGNÉ / Designated laboratory	RENSEIGNEMENTS SUR L'ANALYSE / Test details	MÉTHODE / Methode	NOTE PARTICULIÈRE / Special comment	Date de la révision / Date of revision
1,5-anhydroglucitol (GlycoMark) [500115] [GLYC]	1465		Gold Top or Red Top (Serum) or Lavender (plasma). Centrifuge and aliquot serum to transport tube within one hour of collection. Store and ship frozen. Stable at -20°C for 14 days.	LabCorp/Dynacare (https://www.labcorp.com/)		Enzymatic, colorimetric assay.		10-Jan-21
Neopterin/Tetrahydrobiopterin (CSF) [NC03]	1224	BH4, Neopterin	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 HPABHD PCB01		09-May-20	
Nephrotic Syndrome (NS)/Focal Segmental Glomerulosclerosis (FSGS) Panel [10417]	100	ACTN1 604638 ANLN 616027 ARHGAP24 610886 ARHGEA 610928 CDAP 604241 COL4A3 120070 COL4A4 120131 COL4A5 303630 COL4A6 303631 COQ2 609825 COQ6 614647 COQ8B 615567 CRES2 609720 CUBN 602597 DGKE 604440 EMSP 602534 FAT1 600976 INF2 610982 ITGA1 609825 ITGB4 147557 KANK1 607704 KANK2 614610 KANK4 614612 LAGE3 300060 LAMA5 601833 LAMB2 150325 LMO1B 602578 MAGE1 606382 MYO1E 601479 NPHS1 602716 NPHS2 604766 NUP107 607617 NUP205 614132 NUP93 614151 OSGE1 610107 PAX2 167849 PISX2 610664 PLCE1 608414 PTPRO 608579 SCARB2 602257 SEPL1 607129 SMARCA1 606622 TP53K 608679 TPR45 608680 TRPC3 603652 TTC21B 613014 WDR73 616144 WT1 607102 XPO5 607845	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Alport Syndrome, Autosomal Dominant AD 104200 Alport Syndrome, Autosomal Recessive AR 203780 Alport Syndrome, X-Linked Recessive XL 301050 Cerebral Palsy, Spastic Quadriplegic, 2 AR 612900 Coenzyme Q10 Deficiency AR 607426 Coenzyme Q10 deficiency, primary, 3 AR 614652 Coenzyme Q10 deficiency, primary, 6 AR 614650 Deafness, X-linked 6 XL 300914 Epidermolysis Bullosa With Pyloric Anesias AR 226730 Epilepsy, Progressive Myoclonic: 4, With Or Without Renal Failure AR 254900 Finnish Congenital Nephrotic Syndrome AR 256300 Focal Segmental Glomerulosclerosis 1 AD 603278 Focal Segmental Glomerulosclerosis 2 AD 603965 Focal Segmental Glomerulosclerosis 3, Susceptibility To AR, AD 607832 Focal Segmental Glomerulosclerosis 5 AD 613237 Focal Segmental Glomerulosclerosis 6 AR 614131 Focal Segmental Glomerulosclerosis 8 AD 616032 Focal Segmental Glomerulosclerosis 9 AR 616220 Galloway-Mowat Syndrome AR 251300 Galloway-Mowat Syndrome 2, X-linked XL 301006 Galloway-Mowat Syndrome 3 AR 617729 Galloway-Mowat Syndrome 4 AR 617730 Galloway-Mowat Syndrome 5 AR 617731 Glomerulosclerosis, Focal Segmental, 7 AD 616002 Interstitial Lung Disease, Nephrotic Syndrome, and Epidermolysis Bullosa, Congenital AR 614748	Next Generation DNA Sequencing	03-Aug-22	
Neurofibromatosis (NextGen Sequencing Panel and Copy Number Analysis; 21 Genes) [NGS335]	776	ATM (607585), BRAF (164757), CBL (165360), HRAS (190220), KRAS (190076), MAP2K1 (176072), NF1 (603119), NF2 (607579), NRAS (164790), PTEN (601726), PTPN11 (176076), RAS1 (164760), RIT1 (609891), SDHA12 (613019), SDHB (118576), SENN (602113), SMO1 (602360), SMAD2 (602109), SMAD3 (602775), SRSF1 (602530), SPRED1 (609291).	Lavender Top (EDTA)	MNG Laboratories (www.medicalneurogenetics.com/) / Dynacare		Next Generation DNA Sequencing		09-May-20
Neurofilament (pNF-H)	1465		CSF or Serum	Mitogen Advanced Diagnostics		ELISA		02-Jun-21
Neurofilament (pNF-L)	1465		CSF or Serum	Mitogen Advanced Diagnostics		SIMOA	RUO	02-Jun-21
Neurological Disease Profile (IgG+IgM)	32		Serum/Plasma only	Mitogen Advanced Diagnostics	Anti-GM1, GM2, GM3, GD1a, GD1b, GD1c, GQ1b.	LIA		02-Jun-21

Appellation 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/ou NOM OMIM DU GÈNE / HGNC Gene Symbol/OMIM Gene Name	RÈGLES DE MANUTENTION / Handling Conditions	LABORATOIRE DÉSIGNÉ / Designated laboratory	RENSEIGNEMENTS SUR L'ANALYSE / Test details	MÉTHODE / Methode	NOTE PARTICULIÈRE / Special comment	Date de la révision / Date of revision
I,5-anhydroglucitol (GlycoMark) [500115] [GLYC]	1465		Gold Top or Red Top (Serum) or Lavender (plasma). Centrifuge and aliquot serum to transport tube within one hour of collection. Store and ship frozen. Stable at -20°C for 14 days.	LabCorp/Dynacare (https://www.labcorp.com/)		Enzymatic, colorimetric assay.		10-Jan-21
Neutropenia NGS Panel	836	AP1B1, CSF3R, CCR4, ELANE, G6PC1, GATA1, GATA2, GFI1, HAX1, LAMTOR2, LYST, RAR27A, RAC1, SRSB, SLC77A4, TAZ, USH1B, VPS13B, VPS4A, WAS, WIPF1	Lavender Top (2 x 4 mL)	Fulgent Genetics (fulgentdiagnostics.com)	Cyclical neutropenia (162800), Severe congenital neutropenia autosomal dominant (202700), Severe congenital neutropenia X-linked (300299), Severe congenital neutropenia 4, autosomal recessive (612541), Severe congenital neutropenia 2, autosomal dominant (613107), Severe congenital neutropenia 3, autosomal recessive (610738)	NextGen Sequencing		28-Feb-20
Neutrophil Antibody, Flow Cytometry [1606]	1572		Serum, Red Top (only). Room temperature: 7 days Refrigerated: 14 days Frozen: 30 days	Quest Diagnostics				06-Jul-20
Niemann-Pick Disease Type C Sequencing Panel [3425]	468	NPC1 (60762); NPC2 (601015)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Niemann-Pick Disease Type C1 (257220)	Next Generation DNA Sequencing		12-Nov-19
N-Methylhistamine, 24 Hour, Urine [NMH24]	117		Collection Instructions: Collect 24-hour urine with no preservatives. Total volume required. Record on both the specimen container and the request form. Urine Preservative Collection Options: Note: The addition of preservative or application of temperature controls must occur within 4 hours of completion of the collection: 6N HCl, 50% Acetic Acid, Na(2)CO(3), Toluene, 6N HNO(3), Boric Acid, Thymol. Store frozen. Stable 14 days.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	mastocytosis	Liquid Chromatography-Tandem Mass Spectrometry (LC-MS/MS)		04-Mar-21
Noonan Syndrome (NextGen Sequencing Panel and Copy Number Analysis; 10 Genes) [NGS414]	778	BRAF, KRAS, LZTR1, MAP3K1, NRAS, PTPN11, RAF1, RAS1, SOS1, SOS2	Lavender top (EDTA) [2 tubes]	MNG Laboratories (www.medicalneurogenetics.com)/Dynacare		NextGen Sequencing		09-May-20
NOTCH3-CADASIL (125310)	65	NOTCH3 (608276)	Lavender Top (EDTA)	London Laboratories Service Group		Sanger sequencing; 33 coding exons of NOTCH3+ 20nt intronic sequence around each exon	To change to NGS with exon copy number in Oct 2015	
NPM1 MRD	1395	NPM1 (164040)	Lavender Top (EDTA). Bone marrow. Stable 24-48 h at RT after draw. Do not freeze.	Dept of Clinical Laboratory Genetics, Genome Diagnostics, Toronto General Hospital.		RNA based		10-May-22
NPM1 Quantitative RNA PCR	1395	NPM1 (164040)	Lavender Top (EDTA). Stable for only 24 h.	Molecular Genetics Laboratory, University of North Carolina Hospitals. http://labs.unchealthcare.org .		RNA based		10-May-22
Nuclear-Mito NGS Panel	518	ATP5B, ATP5C1, ATP5G1, ATP5G2, ATP5G3, ATP5G4, ATP5G5, ATP5G6, ATP5G7, ATP5G8, ATP5G9, ATP5G10, ATP5G11, ATP5G12, ATP5G13, ATP5G14, ATP5G15, ATP5G16, ATP5G17, ATP5G18, ATP5G19, ATP5G20, ATP5G21, ATP5G22, ATP5G23, ATP5G24, ATP5G25, ATP5G26, ATP5G27, ATP5G28, ATP5G29, ATP5G30, ATP5G31, ATP5G32, ATP5G33, ATP5G34, ATP5G35, ATP5G36, ATP5G37, ATP5G38, ATP5G39, ATP5G40, ATP5G41, ATP5G42, ATP5G43, ATP5G44, ATP5G45, ATP5G46, ATP5G47, ATP5G48, ATP5G49, ATP5G50, ATP5G51, ATP5G52, ATP5G53, ATP5G54, ATP5G55, ATP5G56, ATP5G57, ATP5G58, ATP5G59, ATP5G60, ATP5G61, ATP5G62, ATP5G63, ATP5G64, ATP5G65, ATP5G66, ATP5G67, ATP5G68, ATP5G69, ATP5G70, ATP5G71, ATP5G72, ATP5G73, ATP5G74, ATP5G75, ATP5G76, ATP5G77, ATP5G78, ATP5G79, ATP5G80, ATP5G81, ATP5G82, ATP5G83, ATP5G84, ATP5G85, ATP5G86, ATP5G87, ATP5G88, ATP5G89, ATP5G90, ATP5G91, ATP5G92, ATP5G93, ATP5G94, ATP5G95, ATP5G96, ATP5G97, ATP5G98, ATP5G99, ATP5G100, ATP5G101, ATP5G102, ATP5G103, ATP5G104, ATP5G105, ATP5G106, ATP5G107, ATP5G108, ATP5G109, ATP5G110, ATP5G111, ATP5G112, ATP5G113, ATP5G114, ATP5G115, ATP5G116, ATP5G117, ATP5G118, ATP5G119, ATP5G120, ATP5G121, ATP5G122, ATP5G123, ATP5G124, ATP5G125, ATP5G126, ATP5G127, ATP5G128, ATP5G129, ATP5G130, ATP5G131, ATP5G132, ATP5G133, ATP5G134, ATP5G135, ATP5G136, ATP5G137, ATP5G138, ATP5G139, ATP5G140, ATP5G141, ATP5G142, ATP5G143, ATP5G144, ATP5G145, ATP5G146, ATP5G147, ATP5G148, ATP5G149, ATP5G150, ATP5G151, ATP5G152, ATP5G153, ATP5G154, ATP5G155, ATP5G156, ATP5G157, ATP5G158, ATP5G159, ATP5G160, ATP5G161, ATP5G162, ATP5G163, ATP5G164, ATP5G165, ATP5G166, ATP5G167, ATP5G168, ATP5G169, ATP5G170, ATP5G171, ATP5G172, ATP5G173, ATP5G174, ATP5G175, ATP5G176, ATP5G177, ATP5G178, ATP5G179, ATP5G180, ATP5G181, ATP5G182, ATP5G183, ATP5G184, ATP5G185, ATP5G186, ATP5G187, ATP5G188, ATP5G189, ATP5G190, ATP5G191, ATP5G192, ATP5G193, ATP5G194, ATP5G195, ATP5G196, ATP5G197, ATP5G198, ATP5G199, ATP5G200, ATP5G201, ATP5G202, ATP5G203, ATP5G204, ATP5G205, ATP5G206, ATP5G207, ATP5G208, ATP5G209, ATP5G210, ATP5G211, ATP5G212, ATP5G213, ATP5G214, ATP5G215, ATP5G216, ATP5G217, ATP5G218, ATP5G219, ATP5G220, ATP5G221, ATP5G222, ATP5G223, ATP5G224, ATP5G225, ATP5G226, ATP5G227, ATP5G228, ATP5G229, ATP5G230, ATP5G231, ATP5G232, ATP5G233, ATP5G234, ATP5G235, ATP5G236, ATP5G237, ATP5G238, ATP5G239, ATP5G240, ATP5G241, ATP5G242, ATP5G243, ATP5G244, ATP5G245, ATP5G246, ATP5G247, ATP5G248, ATP5G249, ATP5G250, ATP5G251, ATP5G252, ATP5G253, ATP5G254, ATP5G255, ATP5G256, ATP5G257, ATP5G258, ATP5G259, ATP5G260, ATP5G261, ATP5G262, ATP5G263, ATP5G264, ATP5G265, ATP5G266, ATP5G267, ATP5G268, ATP5G269, ATP5G270, ATP5G271, ATP5G272, ATP5G273, ATP5G274, ATP5G275, ATP5G276, ATP5G277, ATP5G278, ATP5G279, ATP5G280, ATP5G281, ATP5G282, ATP5G283, ATP5G284, ATP5G285, ATP5G286, ATP5G287, ATP5G288, ATP5G289, ATP5G290, ATP5G291, ATP5G292, ATP5G293, ATP5G294, ATP5G295, ATP5G296, ATP5G297, ATP5G298, ATP5G299, ATP5G300, ATP5G301, ATP5G302, ATP5G303, ATP5G304, ATP5G305, ATP5G306, ATP5G307, ATP5G308, ATP5G309, ATP5G310, ATP5G311, ATP5G312, ATP5G313, ATP5G314, ATP5G315, ATP5G316, ATP5G317, ATP5G318, ATP5G319, ATP5G320, ATP5G321, ATP5G322, ATP5G323, ATP5G324, ATP5G325, ATP5G326, ATP5G327, ATP5G328, ATP5G329, ATP5G330, ATP5G331, ATP5G332, ATP5G333, ATP5G334, ATP5G335, ATP5G336, ATP5G337, ATP5G338, ATP5G339, ATP5G340, ATP5G341, ATP5G342, ATP5G343, ATP5G344, ATP5G345, ATP5G346, ATP5G347, ATP5G348, ATP5G349, ATP5G350, ATP5G351, ATP5G352, ATP5G353, ATP5G354, ATP5G355, ATP5G356, ATP5G357, ATP5G358, ATP5G359, ATP5G360, ATP5G361, ATP5G362, ATP5G363, ATP5G364, ATP5G365, ATP5G366, ATP5G367, ATP5G368, ATP5G369, ATP5G370, ATP5G371, ATP5G372, ATP5G373, ATP5G374, ATP5G375, ATP5G376, ATP5G377, ATP5G378, ATP5G379, ATP5G380, ATP5G381, ATP5G382, ATP5G383, ATP5G384, ATP5G385, ATP5G386, ATP5G387, ATP5G388, ATP5G389, ATP5G390, ATP5G391, ATP5G392, ATP5G393, ATP5G394, ATP5G395, ATP5G396, ATP5G397, ATP5G398, ATP5G399, ATP5G400, ATP5G401, ATP5G402, ATP5G403, ATP5G404, ATP5G405, ATP5G406, ATP5G407, ATP5G408, ATP5G409, ATP5G410, ATP5G411, ATP5G412, ATP5G413, ATP5G414, ATP5G415, ATP5G416, ATP5G417, ATP5G418, ATP5G419, ATP5G420, ATP5G421, ATP5G422, ATP5G423, ATP5G424, ATP5G425, ATP5G426, ATP5G427, ATP5G428, ATP5G429, ATP5G430, ATP5G431, ATP5G432, ATP5G433, ATP5G434, ATP5G435, ATP5G436, ATP5G437, ATP5G438, ATP5G439, ATP5G440, ATP5G441, ATP5G442, ATP5G443, ATP5G444, ATP5G445, ATP5G446, ATP5G447, ATP5G448, ATP5G449, ATP5G450, ATP5G451, ATP5G452, ATP5G453, ATP5G454, ATP5G455, ATP5G456, ATP5G457, ATP5G458, ATP5G459, ATP5G460, ATP5G461, ATP5G462, ATP5G463, ATP5G464, ATP5G465, ATP5G466, ATP5G467, ATP5G468, ATP5G469, ATP5G470, ATP5G471, ATP5G472, ATP5G473, ATP5G474, ATP5G475, ATP5G476, ATP5G477, ATP5G478, ATP5G479, ATP5G480, ATP5G481, ATP5G482, ATP5G483, ATP5G484, ATP5G485, ATP5G486, ATP5G487, ATP5G488, ATP5G489, ATP5G490, ATP5G491, ATP5G492, ATP5G493, ATP5G494, ATP5G495, ATP5G496, ATP5G497, ATP5G498, ATP5G499, ATP5G500, ATP5G501, ATP5G502, ATP5G503, ATP5G504, ATP5G505, ATP5G506, ATP5G507, ATP5G508, ATP5G509, ATP5G510, ATP5G511, ATP5G512, ATP5G513, ATP5G514, ATP5G515, ATP5G516, ATP5G517, ATP5G518, ATP5G519, ATP5G520, ATP5G521, ATP5G522, ATP5G523, ATP5G524, ATP5G525, ATP5G526, ATP5G527, ATP5G528, ATP5G529, ATP5G530, ATP5G531, ATP5G532, ATP5G533, ATP5G534, ATP5G535, ATP5G536, ATP5G537, ATP5G538, ATP5G539, ATP5G540, ATP5G541, ATP5G542, ATP5G543, ATP5G544, ATP5G545, ATP5G546, ATP5G547, ATP5G548, ATP5G549, ATP5G550, ATP5G551, ATP5G552, ATP5G553, ATP5G554, ATP5G555, ATP5G556, ATP5G557, ATP5G558, ATP5G559, ATP5G560, ATP5G561, ATP5G562, ATP5G563, ATP5G564, ATP5G565, ATP5G566, ATP5G567, ATP5G568, ATP5G569, ATP5G570, ATP5G571, ATP5G572, ATP5G573, ATP5G574, ATP5G575, ATP5G576, ATP5G577, ATP5G578, ATP5G579, ATP5G580, ATP5G581, ATP5G582, ATP5G583, ATP5G584, ATP5G585, ATP5G586, ATP5G587, ATP5G588, ATP5G589, ATP5G590, ATP5G591, ATP5G592, ATP5G593, ATP5G594, ATP5G595, ATP5G596, ATP5G597, ATP5G598, ATP5G599, ATP5G600, ATP5G601, ATP5G602, ATP5G603, ATP5G604, ATP5G605, ATP5G606, ATP5G607, ATP5G608, ATP5G609, ATP5G610, ATP5G611, ATP5G612, ATP5G613, ATP5G614, ATP5G615, ATP5G616, ATP5G617, ATP5G618, ATP5G619, ATP5G620, ATP5G621, ATP5G622, ATP5G623, ATP5G624, ATP5G625, ATP5G626, ATP5G627, ATP5G628, ATP5G629, ATP5G630, ATP5G631, ATP5G632, ATP5G633, ATP5G634, ATP5G635, ATP5G636, ATP5G637, ATP5G638, ATP5G639, ATP5G640, ATP5G641, ATP5G642, ATP5G643, ATP5G644, ATP5G645, ATP5G646, ATP5G647, ATP5G648, ATP5G649, ATP5G650, ATP5G651, ATP5G652, ATP5G653, ATP5G654, ATP5G655, ATP5G656, ATP5G657, ATP5G658, ATP5G659, ATP5G660, ATP5G661, ATP5G662, ATP5G663, ATP5G664, ATP5G665, ATP5G666, ATP5G667, ATP5G668, ATP5G669, ATP5G670, ATP5G671, ATP5G672, ATP5G673, ATP5G674, ATP5G675, ATP5G676, ATP5G677, ATP5G678, ATP5G679, ATP5G680, ATP5G681, ATP5G682, ATP5G683, ATP5G684, ATP5G685, ATP5G686, ATP5G687, ATP5G688, ATP5G689, ATP5G690, ATP5G691, ATP5G692, ATP5G693, ATP5G694, ATP5G695, ATP5G696, ATP5G697, ATP5G698, ATP5G699, ATP5G700, ATP5G701, ATP5G702, ATP5G703, ATP5G704, ATP5G705, ATP5G706, ATP5G707, ATP5G708, ATP5G709, ATP5G710, ATP5G711, ATP5G712, ATP5G713, ATP5G714, ATP5G715, ATP5G716, ATP5G717, ATP5G718, ATP5G719, ATP5G720, ATP5G721, ATP5G722, ATP5G723, ATP5G724, ATP5G725, ATP5G726, ATP5G727, ATP5G728, ATP5G729, ATP5G730, ATP5G731, ATP5G732, ATP5G733, ATP5G734, ATP5G735, ATP5G736, ATP5G737, ATP5G738, ATP5G739, ATP5G740, ATP5G741, ATP5G742, ATP5G743, ATP5G744, ATP5G745, ATP5G746, ATP5G747, ATP5G748, ATP5G749, ATP5G750, ATP5G751, ATP5G752, ATP5G753, ATP5G754, ATP5G755, ATP5G756, ATP5G757, ATP5G758, ATP5G759, ATP5G760, ATP5G761, ATP5G762, ATP5G763, ATP5G764, ATP5G765, ATP5G766, ATP5G767, ATP5G768, ATP5G769, ATP5G770, ATP5G771, ATP5G772, ATP5G773, ATP5G774, ATP5G775, ATP5G776, ATP5G777, ATP5G778, ATP5G779, ATP5G780, ATP5G781, ATP5G782, ATP5G783, ATP5G784, ATP5G785, ATP5G786, ATP5G787, ATP5G788, ATP5G789, ATP5G790, ATP5G791, ATP5G792, ATP5G793, ATP5G794, ATP5G795, ATP5G796, ATP5G797, ATP5G798, ATP5G799, ATP5G800, ATP5G801, ATP5G802, ATP5G803, ATP5G804, ATP5G805, ATP5G806, ATP5G807, ATP5G808, ATP5G809, ATP5G810, ATP5G811, ATP5G812, ATP5G813, ATP5G814, ATP5G815, ATP5G816, ATP5G817, ATP5G818, ATP5G819, ATP5G820, ATP5G821, ATP5G822, ATP5G823, ATP5G824, ATP5G825, ATP5G826, ATP5G827, ATP5G828, ATP5G829, ATP5G830, ATP5G831, ATP5G832, ATP5G833, ATP5G834, ATP5G835, ATP5G836, ATP5G837, ATP5G838, ATP5G839, ATP5G840, ATP5G841, ATP5G842, ATP5G843, ATP5G844, ATP5G845, ATP5G846, ATP5G847, ATP5G848, ATP5G849, ATP5G850, ATP5G851, ATP5G852, ATP5G853, ATP5G854, ATP5G855, ATP5G856, ATP5G857, ATP5G858, ATP5G859, ATP5G860, ATP5G861, ATP5G862, ATP5G863, ATP5G864, ATP5G865, ATP5G866, ATP5G867, ATP5G868, ATP5G869, ATP5G870, ATP5G871, ATP5G872, ATP5G873, ATP5G874, ATP5G875, ATP5G876, ATP5G877, ATP5G878, ATP5G879, ATP5G880, ATP5G881, ATP5G882, ATP5G883, ATP5G884, ATP5G885, ATP5G886, ATP5G887, ATP5G888, ATP5G889, ATP5G890, ATP5G891, ATP5G892, ATP5G893, ATP5G894, ATP5G895, ATP5G896, ATP5G897, ATP5G898, ATP5G899, ATP5G900, ATP5G901, ATP5G902, ATP5G903, ATP5G904, ATP5G905, ATP5G906, ATP5G907, ATP5G908, ATP5G909, ATP5G910, ATP5G911, ATP5G912, ATP5G913, ATP5G914, ATP5G915, ATP5G916, ATP5G917, ATP5G918, ATP5G919, ATP5G920, ATP5G921, ATP5G922, ATP5G923, ATP5G924, ATP5G925, ATP5G926, ATP5G927, ATP5G928, ATP5G929, ATP5G930, ATP5G931, ATP5G932, ATP5G933, ATP5G934, ATP5G935, ATP5G936, ATP5G937, ATP5G938, ATP5G939, ATP5G940, ATP5G941, ATP5G942, ATP5G943, ATP5G944, ATP5G945, ATP5G946, ATP5G947, ATP5G948, ATP5G949, ATP5G950, ATP5G951, ATP5G952, ATP5G953, ATP5G954, ATP5G955, ATP5G956, ATP5G957, ATP5G958, ATP5G959, ATP5G960, ATP5G961, ATP5G962, ATP5G963, ATP5G964, ATP5G965, ATP5G966, ATP5G967, ATP5G968, ATP5G969, ATP5G970, ATP5G971, ATP5G972, ATP5G973, ATP5G974, ATP5G975, ATP5G976, ATP5G977, ATP5G978, ATP5G979, ATP5G980, ATP5G981, ATP5G982, ATP5G983, ATP5G984, ATP5G985, ATP5G986, ATP5G987, ATP5G988, ATP5G989, ATP5G990, ATP5G991, ATP5G992, ATP5G993, ATP5G994, ATP5G995, ATP5G996, ATP5G997, ATP5G998, ATP5G999, ATP5G1000, ATP5G1001, ATP5G1002, ATP5G1003, ATP5G1004, ATP5G1005, ATP5G1006, ATP5G1007, ATP5G1008, ATP5G1009, ATP5G1010, ATP5G1011, ATP5G1012, ATP5G1013, ATP5G1014, ATP5G1015, ATP5G1016, ATP5G1017, ATP5G1018, ATP5G1019, ATP5G1020, ATP5G1021, ATP5G1022, ATP5G1023, ATP5G1024, ATP5G1025, ATP5G1026, ATP5G1027, ATP5G1028, ATP5G1029, ATP5G1030, ATP5G1031, ATP5G1032, ATP5G1033, ATP5G1034, ATP5G1035, ATP5G1036, ATP5G1037, ATP5G1038, ATP5G1039, ATP5G1040, ATP5G1041, ATP5G1042, ATP5G1043, ATP5G1044, ATP5G1045, ATP5G1046, ATP5G1047, ATP5G1048, ATP5G1049, ATP5G1050, ATP5G1051, ATP5G1052, ATP5G1053, ATP5G1054, ATP5G1055, ATP5G1056, ATP5G1057, ATP5G1058, ATP5G1059, ATP5G1060, ATP5G1061, ATP5G1062, ATP5G1063, ATP5G1064, ATP5G1065, ATP5G1066, ATP5G1067, ATP5G1068, ATP5G1069, ATP5G1070, ATP5G1071, ATP5G1072, ATP5G1073, ATP5G1074, ATP5G1075, ATP5G1076, ATP5G1077, ATP5G1078, ATP5G1079, ATP5G1080, ATP5G1081, ATP5G1082, ATP5G1083, ATP5G1084, ATP5G1085, ATP5G1086, ATP5G1087, ATP5G1088, ATP5G1089, ATP5G1090, ATP5G1091, ATP5G1092, ATP5G1093, ATP5G1094, ATP5G1095, ATP5G1096, ATP5G1097, ATP5G1098, ATP5G1099, ATP5G1100, ATP5G1101, ATP5G1102, ATP5G1103, ATP5G1104, ATP5G1105, ATP5G1106, ATP5G1107, ATP5G1108, ATP5G1109, ATP5G1110, ATP5G1111, ATP5G1112, ATP5G1113, ATP5G1114, ATP5G1115, ATP5G1116, ATP5G1117, ATP5G1118, ATP5G1119, ATP5G1120, ATP5G1121, ATP5G1122, ATP5G1123, ATP5G1124, ATP5G1125, ATP5G1126, ATP5G1127, ATP5G1128, ATP5G1129, ATP5G1130, ATP5G1131, ATP5G1132, ATP5G1133, ATP5G1134, ATP5G1135, ATP5G1136, ATP5G1137, ATP5G1138, ATP5G1139, ATP5G1140, ATP5G1141, ATP5G1142, ATP5G1143, ATP5G1144, ATP5G1145, ATP5G1146, ATP5G1147, ATP5G1148, ATP5G1149, ATP5G1150, ATP5G1151, ATP5G1152, ATP5G1153, ATP5G1154, ATP5G1155, ATP5G1156, ATP5G1157, ATP5G1158, ATP5G1159, ATP5G1160, ATP5G1161, ATP5G1162, ATP5G1163, ATP5G1164, ATP5G1165, ATP5G1166, ATP5G1167, ATP5G1168, ATP5G1169, ATP5G1170, ATP5G1171, ATP5G1172, ATP5G1173, ATP5G1174, ATP5G1175, ATP5G1176, ATP5G1177, ATP5G1178, ATP5G1179, ATP5G1180, ATP5G1181, ATP5G1182, ATP5G1183, ATP5G1184, ATP5G1185, ATP5G1186, ATP5G1187, ATP5G1188, ATP5G1189, ATP5G1190, ATP5G1191, ATP5G1192, ATP5G1193, ATP5G1194, ATP5G1195, ATP5G1196, ATP5G1197, ATP5G1198, ATP5G1199, ATP5G1200, ATP5G1201, ATP5G1202, ATP5G1203, ATP5G1204, ATP5G1205, ATP5G1206, ATP5G1207, ATP5G1208, ATP5G1209, ATP5G1210, ATP5G1211, ATP5G1212, ATP5G1213, ATP5G1214, ATP5G1215, ATP5G1216, ATP5G1217, ATP5G1218, ATP5G1219, ATP5G1220, ATP5G1221, ATP5G1222, ATP5G1223, ATP5G1224, ATP5G1225, ATP5G1226, ATP5G1227, ATP5G1228, ATP5G1229, ATP5G1230, ATP5G1231, ATP5G1232, ATP5G1233, ATP5G1234, ATP5G1235, ATP5G1236, ATP5G1237, ATP5G1238, ATP5G1239, ATP5G1240, ATP5G1241, ATP5G1242, ATP5G1243, ATP5G1244, ATP5G1245, ATP5G1246, ATP5G1247, ATP5G1248, ATP5G1249, ATP5G1250, ATP5G1251, ATP5G1252, ATP5G1253, ATP5G1254, ATP5G1255, ATP5G1256, ATP5G1257, ATP5G1258, ATP5G1259, ATP5G1260, ATP5G1261, ATP5G1262, ATP5G1263, ATP5G1264, ATP5G1265, ATP5G1266, ATP5G1267, ATP5G1268, ATP5G1269, ATP5G1270, ATP5G1271, ATP5G1272, ATP5G1273, ATP5G1274, ATP5G1275, ATP5G1276, ATP5G1277, ATP5G1278, ATP5G1279, ATP5G1280, ATP5G1281, ATP5G1282, ATP5G1283, ATP5G1284, ATP5G1285, ATP5G1286, ATP5G1287, ATP5G1288, ATP5G1289, ATP5G1290, ATP5G1291, ATP5G1292, ATP5G1293, ATP5G1294, ATP5G1295, ATP5G1296, ATP5G1297, ATP5G1298, ATP5G1299, ATP5G1300, ATP5G1301, ATP5G1302, ATP5G1303, ATP5G1304, ATP5G1305, ATP5G1306, ATP5G1307, ATP5G1308, ATP5G1309, ATP5G1310, ATP5G1311, ATP5G1312, ATP5G1313, ATP5G1314, ATP5G1315, ATP5G1316, ATP5G1317, ATP5G1318, ATP5G1319, ATP5G1320, ATP5G1321, ATP5G1322, ATP5G1323, ATP5G1324, ATP5G1325, ATP5G1326, ATP5G1327, ATP5G1328, ATP5G1329, ATP5G1330, ATP5G1331, ATP5G1332, ATP5G1333, ATP5G1334, ATP5G1335, ATP5G1336, ATP5G1337, ATP5G1338, ATP5G1339, ATP5G1340, ATP5G1341, ATP5G1342, ATP5G1343, ATP5G1344, ATP5G1345, ATP5G1346, ATP5G1347, ATP5G1348, ATP5G1349, ATP5G1350, ATP5G1351, ATP5G1352, ATP5G1353, ATP5G1354, ATP5G1355, ATP5G1356, ATP5G1357, ATP5G1358, ATP5G1359, ATP5G1360, ATP5G1361, ATP5G1362, ATP5G1363, ATP5G1364, ATP5G1365, ATP5G1366, ATP5G1367, ATP5G1368, ATP5G1369, ATP5G1370, ATP5G1371, ATP5G1372, ATP5G1373, ATP5G1374, ATP5G1375, ATP5G1376, ATP5G1377, ATP5G1378, ATP5G1379, ATP5G1380, ATP5G1381, ATP5G1382, ATP5G1383, ATP5G1384, ATP5G1385, ATP5G1386, ATP5G1387, ATP5G1388, ATP5G1389, ATP5G1390, ATP5G1391, ATP5G1392, ATP5G1393, ATP5G1394, ATP5G1395, ATP5G1396, ATP5G1397, ATP5G1398, ATP5G1399, ATP5G1400, ATP5G1401, ATP5G1402, ATP5G1403, ATP5G1404, ATP5G1405, ATP5G1406, ATP5G1407, ATP5G1408, ATP5G1409, ATP5G1410, ATP5G1411, ATP5G1412, ATP5G1413, ATP5G1414, ATP5G1415, ATP5G1416, ATP5G1417, ATP5G1418, ATP5G1419, ATP5G1420, ATP5G1421, ATP5G1422, ATP5G1423, ATP5G1424, ATP5G1425, ATP5G1426, ATP5G1427, ATP5G1428, ATP5G1429, ATP5G1						

Appellation 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 MSHS)	SYMBOL HGNC DU GÈNE et NOM OMIM DU GÈNE / HGNC Gene Symbol and OMIM Gene Number	RÈGLES DE MANUTENTION / Handling Conditions	LABORATOIRE DÉSIGNÉ / Designated laboratory	RENSEIGNEMENTS SUR L'ANALYSE / Test details	MÉTHODE / Methode	NOTE PARTICULIÈRE / Special comment	Date de la révision / Date of revision
1,5-anhydroglucitol (GlycoMark) [500115] [GLYC]	1465		Gold Top or Red Top (Serum) or Lavender (plasma). Centrifuge and aliquot serum to transport tube within one hour of collection. Store and ship frozen. Stable at -20°C for 14 days.	LabCorp/Dynacare (https://www.labcorp.com/)		Enzymatic, colorimetric assay.		10-Jan-21
Paliperidone (Invectra) [91895]			Red Top (only)	Quest Diagnostics Nichols Institute - California, Molecular Genetics Laboratory	9-hydroxyrespiridone	Liquid Chromatography-Tandem Mass Spectrometry (LC-MS/MS)		19-Feb-18
Pancreastatin			Collect 10 mL EDTA plasma in special tube containing the Z-tube and separate as soon as possible. Freeze plasma immediately after separation. Special Z-tube is available from Inter Science Institute (ISI). Minimum specimen size is 1 mL. Ship frozen.	InterScience Institute (www.interscienceinstitute.com)		radioimmunoassay		
Pancreatic Cancer Panel [01261]	1284		Lavender Top (EDTA)	Invitae (www.invitae.com)		NGS		17-Mar-22
Pancreatic Polypeptide, Plasma [HPP]	1284		Plasma EDTA, fasting, handle 4°C	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)				17-Mar-20
Paraghioma		SDHB (185470); SDHC (602413); SDHD (602600)	Lavender Top (EDTA)	Alberta Mol Dx Laboratory (Calgary)		del/dupl		
Paraghioma		SDHB (185470)	Lavender Top (EDTA)	Alberta Mol Dx Laboratory (Calgary)		Sanger sequencing		
Paraghioma		SDHC (602413)	Lavender Top (EDTA)	Alberta Mol Dx Laboratory (Calgary)		Sanger sequencing		
Paraghioma		SDHD (602600)	Lavender Top (EDTA)	Alberta Mol Dx Laboratory (Calgary)		Sanger sequencing		
Paraneoplastic (Neuronal) Antibody Panel	61		Serum (Gold SST or Red Top) 1st choice. 2nd choice: CSF. Store at -20°C	BC Neuroimmunology (bcneuro.ca)	Amphiphysin, CV2 (CRMP5), PNMA2 (Ma2/Ta), Ri, Yo, Hu, Recoverin, SOX1, Titin, Zic4, GAD65, Tr (DNER)	IF with reflex to Immunoblot and CBA fixed.	Alt @MUHC on serum: Paraneoplastic/Anti-Neuronal Ab (Anti-HU,RI,YO,CV2,PNMA2, Amphiphysin). These are biomarkers of cytotoxic T-cells for disease and not disease causing.	09-Aug-22
Paraneoplastic Antibody Panel	61		SERUM preferred;Gold SST; 2nd choice CSF	Mitogen Advanced Diagnostics	Amphiphysin, Ri (NOVA-1), Yo, Hu, PNMA2 (Ma2/Ta), CV2-1, Recoverin, SOX1, titin, Zic4, GAD65, Tr (DNER)	LIA	Under special circumstances only; Default is BC Neuroimmunology.	09-Aug-22
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.	RIA; IFA; EIA; Western blot; CBA	Under special circumstances only; Default is BC Neuroimmunology.	09-Aug-22
Parkinson-Alzheimer-Dementia NGS Panel	597	A2M, A4AS, ACE, APOE, APP, ATP10A2, ATP10A3, C9orf72, CSF1R, DCN3, DNMT1, EDH4G, FBN3, GBA, GCH1, GRN, HTRA2, LRRK2, MAP1F, MPO, PARK2, PARK7, PINK1, PLAA36, POLG1, PRKN, PRNP, PSEN1, PSEN2, SLC6A3, SNCA, SNCR, TAF11, TREM2, TYROBP, UCHL1, VPS35 (38 genes)	Lavender Top (EDTA)	Fulgent Genetics (fulgentdiagnostics.com)	Includes: Dystonia, DOPA-responsive (BRD) (128230) and C9orf72 repeat analysis	NextGen Sequencing		23-Nov-18
PARKINSON'S DISEASE, JUVENILE VIA THE PARK2 GENE [1027]	664	PARK2 (602544)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Parkinson disease, juvenile, type 2 (600116)	Sanger sequencing		
Parkinsons Disease/Parkinsonism [NGS357]	665		Lavender Top (EDTA)	MNG Laboratories (www.medicalneurogenetics.com)/Dynacare		Next Generation DNA Sequencing		09-May-20
Pepsinogen I	1748		3 ml serum or EDTA plasma should be collected and separated as soon as possible. Freeze specimens immediately after separation. Minimum specimen size is 1 mL.	InterScience Institute (www.interscienceinstitute.com)	Patient should be fasting 10 - 12 hours prior to collection of specimen. Antacids or other medications affecting stomach acidity or gastrointestinal motility should be discontinued, if possible, for at least 48 hours prior to collection.	RIA		
Pepsinogen I [FPEPS]	1748		Red Top or Gold SST or Lavender (EDTA). Separate immediately and freeze.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)			Sent to Interscience Institute. PSN-2 at ISI.	20-Oct-20
Pepsinogen II	1748		3 ml serum or EDTA plasma should be collected and separated as soon as possible. Freeze specimens immediately after separation. Minimum specimen size is 1 mL.	InterScience Institute (www.interscienceinstitute.com)	Patient should be fasting 10 - 12 hours prior to collection of specimen. Antacids or other medications affecting stomach acidity or gastrointestinal motility should be discontinued, if possible, for at least 48 hours prior to collection.	RIA		
Perforin		PRF1 (178280)	Lavender Top (EDTA)	Hospital for Sick Children (Rapid Response Laboratory)	hemophagocytic lymphohistiocytosis (HLH), familial 2 (603553)	Sanger sequencing		
Perforin protein expression			Lavender Top (EDTA)	Hospital for Sick Children (Rapid Response Laboratory)	hemophagocytic lymphohistiocytosis (HLH) (603553)			
Perforin/Granzyme [HLH]	817		Heparin	Alberta Precision Laboratories (Flow Cytometry - Calgary)			To be accompanied by normal control sample	14-Oct-20
Periodic Fever/Autoinflammatory Disorders NGS Panel	838	AP3S1, CARD14, CECR1, ELANE, HAX1, IL10, IL10RA, IL10RB, IL1RN, IL1RN2, IL1RN3, IL1RN4, IL1RN5, IL1RN6, IL1RN7, IL1RN8, IL1RN9, IL1RN10, IL1RN11, IL1RN12, IL1RN13, IL1RN14, IL1RN15, IL1RN16, IL1RN17, IL1RN18, IL1RN19, IL1RN20, IL1RN21, IL1RN22, IL1RN23, IL1RN24, IL1RN25, IL1RN26, IL1RN27, IL1RN28, IL1RN29, IL1RN30, IL1RN31, IL1RN32, IL1RN33, IL1RN34, IL1RN35, IL1RN36, IL1RN37, IL1RN38, IL1RN39, IL1RN40, IL1RN41, IL1RN42, IL1RN43, IL1RN44, IL1RN45, IL1RN46, IL1RN47, IL1RN48, IL1RN49, IL1RN50, IL1RN51, IL1RN52, IL1RN53, IL1RN54, IL1RN55, IL1RN56, IL1RN57, IL1RN58, IL1RN59, IL1RN60, IL1RN61, IL1RN62, IL1RN63, IL1RN64, IL1RN65, IL1RN66, IL1RN67, IL1RN68, IL1RN69, IL1RN70, IL1RN71, IL1RN72, IL1RN73, IL1RN74, IL1RN75, IL1RN76, IL1RN77, IL1RN78, IL1RN79, IL1RN80, IL1RN81, IL1RN82, IL1RN83, IL1RN84, IL1RN85, IL1RN86, IL1RN87, IL1RN88, IL1RN89, IL1RN90, IL1RN91, IL1RN92, IL1RN93, IL1RN94, IL1RN95, IL1RN96, IL1RN97, IL1RN98, IL1RN99, IL1RN100, IL1RN101, IL1RN102, IL1RN103, IL1RN104, IL1RN105, IL1RN106, IL1RN107, IL1RN108, IL1RN109, IL1RN110, IL1RN111, IL1RN112, IL1RN113, IL1RN114, IL1RN115, IL1RN116, IL1RN117, IL1RN118, IL1RN119, IL1RN120, IL1RN121, IL1RN122, IL1RN123, IL1RN124, IL1RN125, IL1RN126, IL1RN127, IL1RN128, IL1RN129, IL1RN130, IL1RN131, IL1RN132, IL1RN133, IL1RN134, IL1RN135, IL1RN136, IL1RN137, IL1RN138, IL1RN139, IL1RN140, IL1RN141, IL1RN142, IL1RN143, IL1RN144, IL1RN145, IL1RN146, IL1RN147, IL1RN148, IL1RN149, IL1RN150, IL1RN151, IL1RN152, IL1RN153, IL1RN154, IL1RN155, IL1RN156, IL1RN157, IL1RN158, IL1RN159, IL1RN160, IL1RN161, IL1RN162, IL1RN163, IL1RN164, IL1RN165, IL1RN166, IL1RN167, IL1RN168, IL1RN169, IL1RN170, IL1RN171, IL1RN172, IL1RN173, IL1RN174, IL1RN175, IL1RN176, IL1RN177, IL1RN178, IL1RN179, IL1RN180, IL1RN181, IL1RN182, IL1RN183, IL1RN184, IL1RN185, IL1RN186, IL1RN187, IL1RN188, IL1RN189, IL1RN190, IL1RN191, IL1RN192, IL1RN193, IL1RN194, IL1RN195, IL1RN196, IL1RN197, IL1RN198, IL1RN199, IL1RN200, IL1RN201, IL1RN202, IL1RN203, IL1RN204, IL1RN205, IL1RN206, IL1RN207, IL1RN208, IL1RN209, IL1RN210, IL1RN211, IL1RN212, IL1RN213, IL1RN214, IL1RN215, IL1RN216, IL1RN217, IL1RN218, IL1RN219, IL1RN220, IL1RN221, IL1RN222, IL1RN223, IL1RN224, IL1RN225, IL1RN226, IL1RN227, IL1RN228, IL1RN229, IL1RN230, IL1RN231, IL1RN232, IL1RN233, IL1RN234, IL1RN235, IL1RN236, IL1RN237, IL1RN238, IL1RN239, IL1RN240, IL1RN241, IL1RN242, IL1RN243, IL1RN244, IL1RN245, IL1RN246, IL1RN247, IL1RN248, IL1RN249, IL1RN250, IL1RN251, IL1RN252, IL1RN253, IL1RN254, IL1RN255, IL1RN256, IL1RN257, IL1RN258, IL1RN259, IL1RN260, IL1RN261, IL1RN262, IL1RN263, IL1RN264, IL1RN265, IL1RN266, IL1RN267, IL1RN268, IL1RN269, IL1RN270, IL1RN271, IL1RN272, IL1RN273, IL1RN274, IL1RN275, IL1RN276, IL1RN277, IL1RN278, IL1RN279, IL1RN280, IL1RN281, IL1RN282, IL1RN283, IL1RN284, IL1RN285, IL1RN286, IL1RN287, IL1RN288, IL1RN289, IL1RN290, IL1RN291, IL1RN292, IL1RN293, IL1RN294, IL1RN295, IL1RN296, IL1RN297, IL1RN298, IL1RN299, IL1RN300, IL1RN301, IL1RN302, IL1RN303, IL1RN304, IL1RN305, IL1RN306, IL1RN307, IL1RN308, IL1RN309, IL1RN310, IL1RN311, IL1RN312, IL1RN313, IL1RN314, IL1RN315, IL1RN316, IL1RN317, IL1RN318, IL1RN319, IL1RN320, IL1RN321, IL1RN322, IL1RN323, IL1RN324, IL1RN325, IL1RN326, IL1RN327, IL1RN328, IL1RN329, IL1RN330, IL1RN331, IL1RN332, IL1RN333, IL1RN334, IL1RN335, IL1RN336, IL1RN337, IL1RN338, IL1RN339, IL1RN340, IL1RN341, IL1RN342, IL1RN343, IL1RN344, IL1RN345, IL1RN346, IL1RN347, IL1RN348, IL1RN349, IL1RN350, IL1RN351, IL1RN352, IL1RN353, IL1RN354, IL1RN355, IL1RN356, IL1RN357, IL1RN358, IL1RN359, IL1RN360, IL1RN361, IL1RN362, IL1RN363, IL1RN364, IL1RN365, IL1RN366, IL1RN367, IL1RN368, IL1RN369, IL1RN370, IL1RN371, IL1RN372, IL1RN373, IL1RN374, IL1RN375, IL1RN376, IL1RN377, IL1RN378, IL1RN379, IL1RN380, IL1RN381, IL1RN382, IL1RN383, IL1RN384, IL1RN385, IL1RN386, IL1RN387, IL1RN388, IL1RN389, IL1RN390, IL1RN391, IL1RN392, IL1RN393, IL1RN394, IL1RN395, IL1RN396, IL1RN397, IL1RN398, IL1RN399, IL1RN400, IL1RN401, IL1RN402, IL1RN403, IL1RN404, IL1RN405, IL1RN406, IL1RN407, IL1RN408, IL1RN409, IL1RN410, IL1RN411, IL1RN412, IL1RN413, IL1RN414, IL1RN415, IL1RN416, IL1RN417, IL1RN418, IL1RN419, IL1RN420, IL1RN421, IL1RN422, IL1RN423, IL1RN424, IL1RN425, IL1RN426, IL1RN427, IL1RN428, IL1RN429, IL1RN430, IL1RN431, IL1RN432, IL1RN433, IL1RN434, IL1RN435, IL1RN436, IL1RN437, IL1RN438, IL1RN439, IL1RN440, IL1RN441, IL1RN442, IL1RN443, IL1RN444, IL1RN445, IL1RN446, IL1RN447, IL1RN448, IL1RN449, IL1RN450, IL1RN451, IL1RN452, IL1RN453, IL1RN454, IL1RN455, IL1RN456, IL1RN457, IL1RN458, IL1RN459, IL1RN460, IL1RN461, IL1RN462, IL1RN463, IL1RN464, IL1RN465, IL1RN466, IL1RN467, IL1RN468, IL1RN469, IL1RN470, IL1RN471, IL1RN472, IL1RN473, IL1RN474, IL1RN475, IL1RN476, IL1RN477, IL1RN478, IL1RN479, IL1RN480, IL1RN481, IL1RN482, IL1RN483, IL1RN484, IL1RN485, IL1RN486, IL1RN487, IL1RN488, IL1RN489, IL1RN490, IL1RN491, IL1RN492, IL1RN493, IL1RN494, IL1RN495, IL1RN496, IL1RN497, IL1RN498, IL1RN499, IL1RN500, IL1RN501, IL1RN502, IL1RN503, IL1RN504, IL1RN505, IL1RN506, IL1RN507, IL1RN508, IL1RN509, IL1RN510, IL1RN511, IL1RN512, IL1RN513, IL1RN514, IL1RN515, IL1RN516, IL1RN517, IL1RN518, IL1RN519, IL1RN520, IL1RN521, IL1RN522, IL1RN523, IL1RN524, IL1RN525, IL1RN526, IL1RN527, IL1RN528, IL1RN529, IL1RN530, IL1RN531, IL1RN532, IL1RN533, IL1RN534, IL1RN535, IL1RN536, IL1RN537, IL1RN538, IL1RN539, IL1RN540, IL1RN541, IL1RN542, IL1RN543, IL1RN544, IL1RN545, IL1RN546, IL1RN547, IL1RN548, IL1RN549, IL1RN550, IL1RN551, IL1RN552, IL1RN553, IL1RN554, IL1RN555, IL1RN556, IL1RN557, IL1RN558, IL1RN559, IL1RN560, IL1RN561, IL1RN562, IL1RN563, IL1RN564, IL1RN565, IL1RN566, IL1RN567, IL1RN568, IL1RN569, IL1RN570, IL1RN571, IL1RN572, IL1RN573, IL1RN574, IL1RN575, IL1RN576, IL1RN577, IL1RN578, IL1RN579, IL1RN580, IL1RN581, IL1RN582, IL1RN583, IL1RN584, IL1RN585, IL1RN586, IL1RN587, IL1RN588, IL1RN589, IL1RN590, IL1RN591, IL1RN592, IL1RN593, IL1RN594, IL1RN595, IL1RN596, IL1RN597, IL1RN598, IL1RN599, IL1RN600, IL1RN601, IL1RN602, IL1RN603, IL1RN604, IL1RN605, IL1RN606, IL1RN607, IL1RN608, IL1RN609, IL1RN610, IL1RN611, IL1RN612, IL1RN613, IL1RN614, IL1RN615, IL1RN616, IL1RN617, IL1RN618, IL1RN619, IL1RN620, IL1RN621, IL1RN622, IL1RN623, IL1RN624, IL1RN625, IL1RN626, IL1RN627, IL1RN628, IL1RN629, IL1RN630, IL1RN631, IL1RN632, IL1RN633, IL1RN634, IL1RN635, IL1RN636, IL1RN637, IL1RN638, IL1RN639, IL1RN640, IL1RN641, IL1RN642, IL1RN643, IL1RN644, IL1RN645, IL1RN646, IL1RN647, IL1RN648, IL1RN649, IL1RN650, IL1RN651, IL1RN652, IL1RN653, IL1RN654, IL1RN655, IL1RN656, IL1RN657, IL1RN658, IL1RN659, IL1RN660, IL1RN661, IL1RN662, IL1RN663, IL1RN664, IL1RN665, IL1RN666, IL1RN667, IL1RN668, IL1RN669, IL1RN670, IL1RN671, IL1RN672, IL1RN673, IL1RN674, IL1RN675, IL1RN676, IL1RN677, IL1RN678, IL1RN679, IL1RN680, IL1RN681, IL1RN682, IL1RN683, IL1RN684, IL1RN685, IL1RN686, IL1RN687, IL1RN688, IL1RN689, IL1RN690, IL1RN691, IL1RN692, IL1RN693, IL1RN694, IL1RN695, IL1RN696, IL1RN697, IL1RN698, IL1RN699, IL1RN700, IL1RN701, IL1RN702, IL1RN703, IL1RN704, IL1RN705, IL1RN706, IL1RN707, IL1RN708, IL1RN709, IL1RN710, IL1RN711, IL1RN712, IL1RN713, IL1RN714, IL1RN715, IL1RN716, IL1RN717, IL1RN718, IL1RN719, IL1RN720, IL1RN721, IL1RN722, IL1RN723, IL1RN724, IL1RN725, IL1RN726, IL1RN727, IL1RN728, IL1RN729, IL1RN730, IL1RN731, IL1RN732, IL1RN733, IL1RN734, IL1RN735, IL1RN736, IL1RN737, IL1RN738, IL1RN739, IL1RN740, IL1RN741, IL1RN742, IL1RN743, IL1RN744, IL1RN745, IL1RN746, IL1RN747, IL1RN748, IL1RN749, IL1RN750, IL1RN751, IL1RN752, IL1RN753, IL1RN754, IL1RN755, IL1RN756, IL1RN757, IL1RN758, IL1RN759, IL1RN760, IL1RN761, IL1RN762, IL1RN763, IL1RN764, IL1RN765, IL1RN766, IL1RN767, IL1RN768, IL1RN769, IL1RN770, IL1RN771, IL1RN772, IL1RN773, IL1RN774, IL1RN775, IL1RN776, IL1RN777, IL1RN778, IL1RN779, IL1RN780, IL1RN781, IL1RN782, IL1RN783, IL1RN784, IL1RN785, IL1RN786, IL1RN787, IL1RN788, IL1RN789, IL1RN790, IL1RN791, IL1RN792, IL1RN793, IL1RN794, IL1RN795, IL1RN796, IL1RN797, IL1RN798, IL1RN799, IL1RN800, IL1RN801, IL1RN802, IL1RN803, IL1RN804, IL1RN805, IL1RN806, IL1RN807, IL1RN808, IL1RN809, IL1RN810, IL1RN811, IL1RN812, IL1RN813, IL1RN814, IL1RN815, IL1RN816, IL1RN817, IL1RN818, IL1RN819, IL1RN820, IL1RN821, IL1RN822, IL1RN823, IL1RN824, IL1RN825, IL1RN826, IL1RN827, IL1RN828, IL1RN829, IL1RN830, IL1RN831, IL1RN832, IL1RN833, IL1RN834, IL1RN835, IL1RN836, IL1RN837, IL1RN838, IL1RN839, IL1RN840, IL1RN841, IL1RN842, IL1RN843, IL1RN844, IL1RN845, IL1RN846, IL1RN847, IL1RN848, IL1RN849, IL1RN850, IL1RN851, IL1RN852, IL1RN853, IL1RN854, IL1RN855, IL1RN856, IL1RN857, IL1RN858, IL1RN859, IL1RN860, IL1RN861, IL1RN862, IL1RN863, IL1RN864, IL1RN865, IL1RN866, IL1RN867, IL1RN868, IL1RN869, IL1RN870, IL1RN871, IL1RN872, IL1RN873, IL1RN874, IL1RN875, IL1RN876, IL1RN877, IL1RN878, IL1RN879, IL1RN880, IL1RN881, IL1RN882, IL1RN883, IL1RN884, IL1RN885, IL1RN886, IL1RN887, IL1RN888, IL1RN889, IL1RN890, IL1RN891, IL1RN892, IL1RN893, IL1RN894, IL1RN895, IL1RN896, IL1RN897, IL1RN898, IL1RN899, IL1RN900, IL1RN901, IL1RN902, IL1RN903, IL1RN904, IL1RN905, IL1RN906, IL1RN907, IL1RN908, IL1RN909, IL1RN910, IL1RN911, IL1RN912, IL1RN913, IL1RN914, IL1RN915, IL1RN916, IL1RN917, IL1RN918, IL1RN919, IL1RN920, IL1RN921, IL1RN922, IL1RN923, IL1RN924, IL1RN925, IL1RN926, IL1RN927, IL1RN928, IL1RN929, IL1RN930, IL1RN931, IL1RN932, IL1RN933, IL1RN934, IL1RN935, IL1RN936, IL1RN937, IL1RN938, IL1RN939, IL1RN940, IL1RN941, IL1RN942, IL1RN943, IL1RN944, IL1RN945, IL1RN946, IL1RN947, IL1RN948, IL1RN949, IL1RN950, IL1RN951, IL1RN952, IL1RN953, IL1RN954, IL1RN955, IL1RN956, IL1RN957, IL1RN958, IL1RN959, IL1RN960, IL1RN961, IL1RN962, IL1RN963, IL1RN964, IL1RN965, IL1RN966, IL1RN967, IL1RN968, IL1RN969, IL1RN970, IL1RN971, IL1RN972, IL1RN973, IL1RN974, IL1RN975, IL1RN976, IL1RN977, IL1RN978, IL1RN979, IL1RN980, IL1RN981, IL1RN982, IL1RN983, IL1RN984, IL1RN985, IL1RN986, IL1RN987, IL1RN988, IL1RN989, IL1RN990, IL1RN991, IL1RN992, IL1RN993, IL1RN994, IL1RN995, IL1RN996, IL1RN997, IL1RN998, IL1RN999, IL1RN1000, IL1RN1001, IL1RN1002, IL1RN1003, IL1RN1004, IL1RN1005, IL1RN1006, IL1RN1007, IL1RN1008, IL1RN1009, IL1RN1010, IL1RN1011, IL1RN1012, IL1RN1013, IL1RN1014, IL1RN1015, IL1RN1016, IL1RN1017, IL1RN1018, IL1RN1019, IL1RN1020, IL1RN1021, IL1RN1022, IL1RN1023, IL1RN1024, IL1RN1025, IL1RN1026, IL1RN1027, IL1RN1028, IL1RN1029, IL1RN1030, IL1RN1031, IL1RN1032, IL1RN1033, IL1RN1034, IL1RN1035, IL1RN1036, IL1RN1037, IL1RN1038, IL1RN1039, IL1RN1040, IL1RN1041, IL1RN1042, IL1RN1043, IL1RN1044, IL1RN1045, IL1RN1046, IL1RN1047, IL1RN1048, IL1RN1049, IL1RN1050, IL1RN1051, IL1RN1052, IL1RN1053, IL1RN1054, IL1RN1055, IL1RN1056, IL1RN1057, IL1RN1058, IL1RN1059, IL1RN1060, IL1RN1061, IL1RN1062, IL1RN1063, IL1RN1064, IL1RN1065, IL1RN1066, IL1RN1067, IL1RN1068, IL1RN1069, IL1RN1070, IL1RN1071, IL1RN1072, IL1RN1073, IL1RN1074, IL1RN1075, IL1RN1076, IL1RN1077, IL1RN1078, IL1RN1079, IL1RN1080, IL1RN1081, IL1RN1082, IL1RN1083, IL1RN1084, IL1RN1085, IL1RN1086, IL1RN1087, IL1RN1088, IL1RN1089, IL1RN1090, IL1RN1091, IL1RN1092, IL1RN1093, IL1RN1094, IL1RN1095, IL1RN1096, IL1RN1097, IL1RN1098, IL1RN1099, IL1RN1100, IL1RN1101, IL1RN1102, IL1RN1103, IL1RN1104, IL1RN1105, IL1RN1106, IL1RN1107, IL1RN1108, IL1RN1109, IL1RN1110, IL1RN1111, IL1RN1112, IL1RN1113, IL1RN1114, IL1RN1115, IL1RN1116, IL1RN1117, IL1RN1118, IL1RN1119, IL1RN1120, IL1RN1121, IL1RN1122, IL1RN1123, IL1RN1124, IL1RN1125, IL1RN1126, IL1RN1127, IL1RN1128, IL1RN1129, IL1RN1130, IL1RN1131, IL1RN1132, IL1RN1133, IL1RN1134, IL1RN1135, IL1RN1136, IL1RN1137, IL1RN1138, IL1RN1139, IL1RN1140, IL1RN1141, IL1RN1142, IL1RN1143, IL1RN1144, IL1RN1145, IL1RN1146, IL1RN1147, IL1RN1148, IL1RN1149, IL1RN1150, IL1RN1151, IL1RN1152, IL1RN1153, IL1RN1154, IL1RN1155, IL1RN1156, IL1RN1157, IL1RN1158, IL1RN1159, IL1RN1160, IL1RN1161, IL1RN1162, IL1RN1163, IL1RN1164, IL1RN1165, IL1RN1166, IL1RN1167, IL1RN1168, IL1RN1169, IL1RN1170, IL1RN1171, IL1RN1172, IL1RN1173, IL1RN1174, IL1RN1175, IL1RN1176, IL1RN1177, IL1RN1178, IL1RN1179, IL1RN1180, IL1RN1181, IL1RN1182, IL1RN1183, IL1RN1184, IL1RN1185, IL1RN1186, IL1RN1187, IL1RN1188, IL1RN1189, IL1RN1190, IL1RN1191, IL1RN1192, IL1RN1193, IL1RN1194, IL1RN1195, IL1RN1196, IL1RN1197, IL1RN1198, IL1RN1199, IL1RN1200, IL1RN1201, IL1RN1202, IL1RN1203, IL1RN1204, IL1RN1205, IL1RN1206, IL1RN1207, IL1RN1208, IL1RN1209, IL1RN1210, IL1RN1211, IL1RN1212, IL1RN1213, IL1RN1214, IL1RN1215, IL1RN1216, IL1RN1217, IL1RN1218, IL1RN1219, IL1RN1220, IL1RN1221, IL1RN1222, IL1RN1223, IL1RN1224, IL1RN1225, IL1RN1226, IL1RN1227, IL1RN1228, IL1RN1229, IL1RN1230, IL1RN1231, IL1RN1232, IL1RN1233, IL1RN1234, IL1RN1235, IL1RN1236, IL1RN1237, IL1RN1238, IL1RN1239, IL1RN1240, IL1RN1241, IL1RN1242, IL1RN1243, IL1RN1244, IL1RN1245, IL1RN1246, IL1RN1247, IL1RN1248, IL1RN1249, IL1RN1250, IL1RN1251, IL1RN1252, IL1RN1253, IL1RN1254, IL1RN1255, IL1RN1256, IL1RN1257, IL1RN1258, IL1RN1259, IL1RN1260, IL1RN1261, IL1RN1262, IL1RN1263, IL1RN1264, IL1RN1265, IL1RN1266, IL1RN1267, IL1RN1268, IL1RN1269, IL1RN1270, IL1RN1271, IL1RN1272, IL1RN1273, IL1RN1274, IL1RN1275, IL1RN1						

Appellation 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 (Déclaration MSSS)	SYMBOL HGNC DU GÈNE et NOM OMIM DU GÈNE / HGNC Gene Symbol and OMIM Gene Number	RÈGLES DE MANUTENTION / Handling Conditions	LABORATOIRE DÉSIGNÉ / Designated laboratory	RENSEIGNEMENTS SUR L'ANALYSE / Test details	MÉTHODE / Methode	NOTE PARTICULIÈRE / Special comment	Date de la révision / Date of revision
1,5-anhydroglucitol (GlycoMark) [500115] [GLYC]	1465		Gold Top or Red Top (Serum) or Lavender (plasma). Centrifuge and aliquot serum to transport tube within one hour of collection. Store and ship frozen. Stable at -20°C for 14 days.	LabCorp/Dynacare (https://www.labcorp.com/)		Enzymatic, colorimetric assay.		10-Jan-21
Phosphatidylserine Antibodies (IgG, IgA, IgM) [10062]			Preferred Specimen(s) 1 mL plasma collected in a 3.2% sodium citrate (light blue-top) tube Minimum Volume 0.5 mL Collection Instructions Platelet-poor plasma: Centrifuge light blue-top tube 15 minutes at approximately 1500 g within 60 minutes of collection. Using a plastic pipette, remove plasma, taking care to avoid the WBC/platelet buffy layer and place into a plastic vial. Centrifuge a second time and transfer platelet-poor plasma into a new plastic vial. Plasma must be free of platelets (<10,000/µL). Transport Container Plastic screw-cap vial Transport Temperature Room temperature Specimen Stability Room temperature: 7 days Refrigerated: 28 days Frozen: 28 days	Quest Diagnostics/Nichols Institute	thrombosis; pregnancy loss	immunoassay		
Phosphoglycerate kinase 1 deficiency (300653)		PKK1 (311800)	Lavender Top (EDTA)	Centogene AG (www.centogene.com)		Sanger sequencing		
Phosphomannomutase (PMM) and Phosphomannose Isomerase (PMI), Leukocytes [PMMIL]			Yellow top (ACD solution B) or Yellow top (ACD solution A). DO NOT CONFUSE WITH STANDARD GOLD SST tube. Do not transfer contents. Store at 4°C. Send immediately.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)		Enzyme assay.		20-Oct-20
Phosphorylase b Kinase enzyme activity			Lavender Top (EDTA) 10 cc OR dried blood spot; other sample types available	Glycogen Storage Disease Laboratory, Duke University Hospital. http://pediatrics.duke.edu/divisions/medical-genetics/biochemical-genetics-laboratory	Glycogen Storage Disease, Type IX, Liver form (306000)	GSDIX phosphorylase b kinase enzyme assay on red blood cells; other enzyme assays available		18-Jul-17
Platelia Apergillus Galactomannan EIA [309]	1465		Serum: Collect serum specimens in serum separator or red top tube. Allow blood to clot for 30 minutes, then centrifuge. Pipette serum into a plastic screw cap vial. CSF/BAL: Submit CSF and BAL in a sterile screw cap container. STABILITY Room Temperature: 48 hours Refrigerated: 5 days Frozen: 5 months	MiraVista Diagnostics				08-Sep-22
PLP1-RELATED DISORDERS VIA THE PLP1 GENE [307]		PLP1 (30040)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Polkaeus-Merzbacher disease (312080) and spastic paraplegia (312920)	Sanger sequencing/aCGH		
Pneumocystis DNA PCR [402]	1395		Transfer 2 mL of bronchoalveolar lavage (BAL) fluid (preferred) to a sterile, leakproof container. Tracheal aspirate and bronchial wash will be reported with a rare specimen comment. STABILITY Room Temperature/Ambient: 14 days Refrigerated: 14 days Frozen: 28 days	MiraVista Diagnostics				08-Sep-22
Polycystic Kidney Disease NGS Panel	795	PKD1 (60333); PKD2 (172910); PKHD1	Lavender Top (2 x 4 mL)	Fulgent Genetics (fulgentdiagnostics.com)	Polycystic kidney disease, adult type 1 (173900); Polycystic kidney disease 2 (613095)	NextGen Sequencing		04-Dec-18
Polymicrogyria Deletion/Duplication Panel (11 genes deletion/duplication analysis)		GRPS6 (604110), KIAA1279 (609367), OCLN (602976), RTTN (610436), TUBA1A (602529), TUBA8 (605742), TUBB2B (612850), TUBB3 (602661), RAP1B (602307), RAB39A (602536), RAB39B (602775)	Lavender Top (EDTA)	University of Chicago Genetic Services Laboratories	Bilateral frontoparietal polymicrogyria (606854), Goldberg-Shprintzen-Magoclon Syndrome (609460), Band-like Calcification with Simplified gyration and polymicrogyria (251290), Polymicrogyria with seizures (614833), polymicrogyria with optic nerve hypoplasia (612180), asymmetric polymicrogyria (610031), Complex cortical dysplasia with other brain malformations (614039), Warburg Micro syndrome (600118)	oligonucleotide array-CGH		

Application 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 (Déclaratoire MSSS)	SYMBOL HUGO DU GÈNE et NOM OMDM DU GÈNE / HUGO Gene Symbol and OMDM Gene Name	RÈGLES DE MANUTENTION / Handling Conditions	LABORATOIRE DÉSIGNÉ / Designated laboratory	RENSEIGNEMENTS SUR L'ANALYSE / Test details	MÉTHODE / Methode	NOTE PARTICULIÈRE / Special comment	Date de la révision / Date of revision
1,5-anhydroglucitol (GlycoMark) [500115] [GLYC]	1465		Gold Top or Red Top (Serum) or Lavender (plasma). Centrifuge and aliquot serum to transport tube within one hour of collection. Store and ship frozen. Stable at -20°C for 14 days.	LabCorp/Dynacare (https://www.labcorp.com/)		Enzymatic, colorimetric assay.		10-Jan-21
Polymicrogyria Sequencing Panel (12 genes sequencing)		CP956 (60110), KLA129 (609367), CCLN (602576), RTTN (610436), TUBB1A (605259), TUBA8 (605742), TUBB2B (612850), TUBB3 (602661), RAP1B (602207), RAB39A (602536), RAB39B (609775), WDR62 (613583)	Lavender Top (EDTA)	University of Chicago Genetic Services Laboratories	Bilateral frontoparietal polymicrogyria (606854), Goldberg-Shpanzer-Magenchen Syndrome (609460), Band-like Calcification with Simplified gyration and polymicrogyria (251290), Polymicrogyria with seizures (614833), polymicrogyria with optic nerve hypoplasia (612180), asymmetric polymicrogyria (610031), Complex cortical dysplasia with other brain malformations (614039), Warburg Micro syndrome (600118)	1. Next-gen Sequencing 2. Sanger sequencing (confirmation)		
Pontocerebellar Hypoplasia NGS Panel	886	CASK, OPHN1, RARS2, SEFSEC3, TSEN2, TSEN34, TSEN54, VRK1	Lavender Top (EDTA)	Fulgent Genetics (fulgentdiagnostics.com)		NextGen Sequencing	EXOSC3 can be added at no cost	
PONTOCEREBELLAR HYPOPLASIA TYPE 1B VIA THE EXOSC3 GENE [1208]		EXOSC3 (604489)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Pontocerebellar hypoplasia, type 1B (614678)	Sanger sequencing	See Fulgent Panel	
Porencephaly 1 (175780)		COL4A1 (120130)	Lavender Top (EDTA)	Centogene AG (www.centogene.com)		1. single exon testing 2. full gene sequencing 3. deletion/duplication testing		
Porphobilinogen Deaminase (PBGD), Whole Blood [PBGD_]	1324		Green Top, 2 mL, 4°C only	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	Acute Intermittent Porphyria (176000)			
Porphyria Disorders NGS Panel	1324	ALAD (125270), ALAS2 (301300), C10ORF41 (615626), CPOX (612732), FECH (612386), HFE (613609), HMBE5 (609806), PPOX (600522), SLC7A2 (609941), UROD (613521), UROS (606958)	Lavender Top (2 x 4 mL)	Fulgent Genetics (fulgentdiagnostics.com)		NextGen Sequencing		31-Aug-19
Porphyrins Evaluation, Whole Blood [PEE]	1324		Green Top (heparin), fasting, handle 4°C. Must arrive within 3 days of drawing. Alternate: washed erythrocytes	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	CCDC68, DNAH11, RSPH4A, RPR8, C22OR4, ZMYND10, DNAF1, DNAF2, DNAF3, DYX1C1, DNAH1, CCTCH1, DNAH2, RSPH3, RSPH4, DNAF5, RSPH9, OFD1, RRC1, C22OR4, LBRCA, SPAG1, C22OR5, MCDIAB, ARMC9, C22OR9, DNAH1, DNAH5, DNAH8, NME8, GASK, CCNO, C22OR14, DNAH1	Lavender Top (EDTA) 2-5 cc	Invitae (https://www.invitae.com) 475 Brannan St. Ste. 230 San Francisco, CA, 94107	Primary Ciliary Dyskinesia panel (244400)	NGS with del/dup analysis		07-Oct-18
PRIMARY FAMILIAL AND CONGENITAL POLYCYTHEMIA (PFPC) VIA THE EPOR GENE [8441]	1475	EPOR (133171)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Familial Erythrocytosis, 1 (133100)	Sanger sequencing	NOT available at HMR	23-Dec-22
Primary Immunodeficiency Panel (Invitae) [08100]	1042	ACD, ACPS, ACTB, ADA, ADA2, ADAM17, ADAR, AKT2, AIRE, AK2, AFB1, ATR, B2M, BCL2L1, BDNK, BCL2L8, BTK, CARD11, CARD14, CARD9, CASP10, CASP8, CD247, CD27, CD3D, CD3E, CD3G, CD40LG, CD79A, CD79B, CD8A, CEBPE, CHD7, CHTA, CLPLB, COPA, CORO1A, CR2, CS2RA, CS2RB, CTC1, CTLA4, CYP51, CTC5, CXCR4, CYBB, DCLRE1B, DCLRE1C, DKC1, DNMT3B, DOCK2, DOCK8, ELANE, EPOR, FADD, FAS, FASLG, FERMT3, FOXP1, FOXP3, FRR1, GPCR1, GATA2, GFI1, HAX1, ICOS, IFIH1, IPIER1, IPIER2, KELL1, KLRK1, IL11, IL1RA, IL1RB, IL12B, IL12RB1, IL17F, IL17RA, IL17RC, IL18, IL21, IL21R, IL2RA, IL2RG, IL36RN, IL37R, IRAK4, IRF7, IRF8, IRF9, ITCH, ITGAE, ITK, JAGN1, JAK3, LAMTOR2, LCK, LEG1, LPXN2, LIPA, LYST, MAGT1, MALT1, MAP3K4, MEFV, MPOB, MVK, MYD88, NBN, NCF2, NCF4, NFAT5, NFKB2, NFKBIA, NHEJ1, NHP2, NLRCA, NLRP12, NLRP3, NOD2, NOP10, ORAI1, PARN, PDM3, PIK3CD, PIK3R1, PLCG2, PMS2, PNP, PNO1, PPIA, PRKCD1, PRKDC, PSMD9, PSF1P1, PTPRC, RAB27A, RAC2, RAG1, RAG2, RBCK1, RFX5, RFXANK, RFXAP, RHOF, RMRP, RNASEHA, RNASEH2B, RNASEH2C, RORC, RTKL1, SAMBID, SEMA4E, SH2B3, SH2B3IP, SLC25A3, SLC25A3L, SLC27A4, SLC7A2, SMARCC1, SP10, SPINK5, STAT1, STAT2, STAT3, STAT5B, STIM1, STK4, STR11, STXBP2, TAP1, TAP2, TAPBP, TAZ, TBK1, TCN2, TERC, TERT, TNSP2, TRKCA1, TNSF2, TLR3, TRK3, TRK3L, TRIM17, TNFRSF1B, TNFRSF1C, TNFRSF1A, TNFRSF4, TFP2, TRAF3, TRAF3IP2, TREX1, TRNT1, TTC7A, TYK2, UNC119, UNC93B1, UNG, VPS13B, VPS45, WAS, WIPF1, XIAP, ZAP70, ZBTB24	Lavender Top (2 x 4 mL)	Invitae (https://www.invitae.com) 475 Brannan St. Ste. 230 San Francisco, CA, 94107	NGS with del/dup analysis		07-Oct-18	
Procainamide and N-acetylprocainamide, Serum [PA]	1465		Gold SST	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)		immunoassay		10-Aug-21
Procollagen I Intact N-Terminal, Serum [PINP]	178		Red Top or Gold SST. Send serum. Stable frozen for 14 days.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	An aid in monitoring antiresorptive and anabolic therapy in patients with osteoporosis. An adjunct in the assessment of conditions associated with increased bone turnover such as Paget disease	competitive radioimmunoassay		09-Aug-22
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 (613389)	Lavender top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Not available: SLC52A2 (607882) and SLC52A1 (607883)	Sanger sequencing		
Progressive Familial Intrahepatic Cholestasis Type 1 (211600)		ATP9B1 (602397)	Lavender Top (EDTA)	Cincinnati Children's Hospital (Division of Human Genetics Diagnostic Laboratories)		Sanger sequencing		
Proinsulin, Plasmat [PINS]	179		Draw blood in a ice-cooled purple-top EDTA. (Plasma gel tube is not acceptable.) Chill on ice for 10 minutes. Spin down and send 1 mL of EDTA plasma frozen.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)		Quantitative Chemiluminescent Immunoassay		04-Mar-21

Appellation 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/ IGENC DU GÈNE et NOM OMIM DU GÈNE / IGENC Gene Symbol and OMIM Gene Number	RÈGLES DE MANUTENTION / Handling Conditions	LABORATOIRE DÉSIGNÉ / Designated laboratory	RENSEIGNEMENTS SUR L'ANALYSE / Test details	MÉTHODE / Methode	NOTE PARTICULIÈRE / Special comment	Date de la révision / Date of revision
1,5-anhydroglucitol (GlycoMark) [500115] [GLYC]	1465		Gold Top or Red Top (Serum) or Lavender (plasma). Centrifuge and aliquot serum to transport tube within one hour of collection. Store and ship frozen. Stable at -20°C for 14 days.	LabCorp/Dynacare (https://www.labcorp.com/)		Enzymatic, colorimetric assay.		10-Jan-21
Proportionate short stature/small for gestational age [8760]		ATRX, BLM, BTK, CREBBP, CUL7, D1RC7, EP300, ERCC5, ERCC6, FGD1, GH1, GHR, GHRHR, GLI2, HESX1, IGF1, IGF1R, INSR, KDM6A, KMT2D, KRAS, LEX1, NDN, NDR1, PTTX2, POU1F1, PPOF1, PTPN11, RAF1, ROR2, RPS6KA1, SHOX, SMARCA4, SMC1A, SMC3, SOX1, SOX2, SOX3, SRCAP, STASB, TBCE, THRB, TRIM37, WRN	Infants (<2 years): 2-3 mL Children (>2 years): 3-5 mL Older Children & Adults: 5-10 mL	https://www.allelediagnosics.com/		Performed using Next Generation Sequencing, Methylation-specific MLPA (MS-MLPA), and DNA methylation specific PCR.	Alternate: Blueprint Short Stature Syndrome Panel [MA2101]. Does not include methylation study.	02-Dec-17
Psychotropic Pharmacogenomics Gene Panel, Varies [PSYGP]	1582		Lavender Top (EDTA)	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)				04-Mar-21
PTEN Hamartoma Tumor Syndrome via the PTEN Gene [707]		PTEN (601728)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Bannayan-Riley-Ruvalcaba Syndrome (153480); Cowden Disease (158350); Cutaneous Malignant Melanoma 1 (155600); Macrocephaly/Autism Syndrome (605309); Vacterl Association With Hydrocephalus (276950)	Sanger sequencing		
Pulmonary Hypertension NGS Panel		ACVRL1 (601284), BMPR2 (600799), CAV1 (601047), ENG (131195), SMAD9 (601295)	Lavender Top (2 x 4 mL)	Fulgent Genetics (fulgentdiagnostics.com)	Telangiectasia, hereditary hemorrhagic, type 2 (600376); Pulmonary hypertension, familial primary, 1, with or without HHT/Pulmonary hypertension, primary, fenfluramine or dexfenfluramine-associated (178600); Pulmonary hypertension, primary, 3 (615343); Telangiectasia, hereditary hemorrhagic, type 1 (187300); Pulmonary hypertension, primary, 2 (615342)	NextGen Sequencing	Alt: CTGT PPH 4 genes & del/dup1	
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 (600931); Fetal Akinesia Deformation Sequence (208150)	Sanger sequencing		
Recombx Zic4 Antibody Test [127]			Red Top	Athena Diagnostics (www.athenadiagnostics.com)	paraneoplastic syndrome			
Renal Tubular Acidosis NGS Panel	806	ATP6V1A4, ATP6V1B1, CA2, SLC4A4, SLC4A5	Lavender Top (EDTA) [2 tubes]	Fulgent Genetics (fulgentdiagnostics.com)				28-Feb-20
Resistance to Thyroid Hormone (RTH) Mutation Analysis [16053(X)]		THRB (190460)	Lavender Top (EDTA)	Quest Diagnostics Nichols Institute - California, Molecular Genetics Laboratory	Thyroid hormone resistance (188570)	Sanger sequencing		
RETINOBLASTOMA VIA THE RB1 GENE [795]		RB1 (614041)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Retinoblastoma (180200)	1. Sanger sequencing 2. aCGH [600]		
Retinol Binding Protein			Red-top tube(s) or a serum gel tube or urine	Quest Diagnostics/Nichols Institute				
Riboflavin (Vitamin B2), Plasma [VITB2]	1617		Green Top; transfer to amber container; protect from light	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	riboflavin		CHUM no longer	01-Sep-21
Rifampin/Ethambutol [NTM9]	58		Isolate	National Jewish Health				09-Nov-17
RNA Polymerase III Antibodies, IgG, Serum [RNAP]	1078		Serum (Gold SST or Red Top)	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	systemic sclerosis	ELISA		23-Dec-21
Russel Silver Syndrome: Methylation and Copy Number Analysis	992		Lavender Top (EDTA)	Genome Diagnostics The Department of Paediatric Laboratory Medicine The Hospital for Sick Children	Russel-Silver syndrome (180860)	Methylation-Specific-MLPA of imprinting center 1 (H19)		19-Feb-20
Russel Silver Syndrome: UPD7 Analysis	992		Lavender Top (EDTA)	Genome Diagnostics The Department of Paediatric Laboratory Medicine The Hospital for Sick Children	Russel-Silver syndrome (180860)	UPD7 studies via STR (short tandem repeat) analysis		19-Feb-20
SACS Single Gene testing	657	SACS (604490)	Lavender Top (EDTA)	Molecular Genetics Laboratory, Hospital for Sick Children (www.sickkids.ca/PaediatricLaboratoryMedicine/Laboratories-Services/Molecular-Genetics-Laboratory/index.html)	Spastic ataxia, Charlevoix-Saguenay type (270550)	NextGen Sequencing		03-Dec-21
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)	Cranioynostosis; Saethre-Chotzen syndrome (101400)	Sanger sequencing: TWIST1; FGFR3 (p.Pro250Arg)		
SANDHOFF DISEASE VIA THE HEXB GENE [476]		HEXB (606873)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Sandhoff Disease (268800)	Sanger sequencing		
SAP Protein Expression [XLP1]	816	SH2D1A (300406)	Heparin	Alberta Precision Laboratories (Flow Cytometry - Calgary)	X-linked lymphoproliferative syndrome (XLP1) (308240)		To be accompanied by normal control sample	14-Oct-20
sC5b-9 (MAC) [7304502]	188		0.5mL of EDTA plasma, separated and frozen within 2 hours of collection; ship on dry ice	Cincinnati Children's Hospital Medical Center 3333 Burnet Avenue NRB 1042 Cincinnati, OH 45229 (www.cincinnatichildrens.org)	complement-mediated renal diseases - C5b-9			14-Jan-19
SCA3/Machado Joseph disease (109150)		ATNSA3 (607047)	Lavender Top (EDTA)	North York General	SCA Panel available			

Appellation 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 BIOC DU GÈNE et NOM OMIM DU GÈNE / BIOC Gene Symbol/OMIM Gene Number	RÈGLES DE MANUTENTION / Handling Conditions	LABORATOIRE DÉSIGNÉ / Designated laboratory	RENSEIGNEMENTS SUR L'ANALYSE / Test details	MÉTHODE / Methode	NOTE PARTICULIÈRE / Special comment	Date de la révision / Date of revision
I.5-anhydroglucitol (GlycoMark) [500115] [GLYC]	1465		Gold Top or Red Top (Serum) or Lavender (plasma). Centrifuge and aliquot serum to transport tube within one hour of collection. Store and ship frozen. Stable at -20°C for 14 days.	LabCorp/Dynacare (https://www.labcorp.com/)		Enzymatic, colorimetric assay.		10-Jan-21
SCN4A Full Gene sequencing Analysis [MOL356]; Paramyotonia Congenita (168300)		SCN4A (603967)	Lavender Top (EDTA)	MNG Laboratories (www.medicalneurogenetics.com)/Dynacare	Paramyotonia Congenita (168300)	Sanger sequencing		09-May-20
Secretin			EDTA plasma containing the G.I. Preservative Freeze immediately	InterScience Institute				
Selenoprotein N, 1 via the SEPN1 gene [330]		SEPN1 (60210)		Prevention Genetics (www.preventiongenetics.com)	Muscular dystrophy, rigid spine, 1 (602771)	Sanger sequencing		
Serum Amyloid (SAA)	1123		Serum samples (blood collection on a red or yellow tube, centrifuged and decanted). Frozen at -20°C on dry ice	Dynacare Next			May be free if specify medication	
sFlt/PIGF Ratio	1465		Serum (Gold SST or Red Top). Stable 3d at 4°C. Stable 6 months at -20°C.	Sunnybrook Health Sciences Centre Specimen Management/Clinical Chemistry, Room G-CG-01A 2075 Bayview Avenue Toronto Ontario M4N 3M5 Telephone 416 480 2854 Fax: 416-480 4651	Pre-eclampsia	Electrochemiluminescence immunoassay (ECLIA) (Roche)		08-Feb-22
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 (249730); Leri Weill Dyschondroostosis (127300); Short Stature, Idiopathic, X-Linked (300582)	Sanger sequencing		28-Sep-17
Single Drug MIC [NTM3]		NTM	Isolate	National Jewish Health	RIE, EMB, CIP, MXF, AMK, LZD, CLR, CLF, RFB, STR, ETH, LVX, AZM OFX, KAN, CSI			09-Nov-17
Single Gene Repeat Expansion Analysis RFC1 [4103]	1395	RFC1	Lavender Top (EDTA)	U. of Chicago Genetic Services Laboratory	CANVAS			22-Dec-21
SLC27A4 Gene Sequencing [712]		SLC27A4 (604194)	Lavender Top (EDTA)	GeneDx (www.genedx.com)	ichthyosis prematurity syndrome (608649)	Sanger sequencing		09-Dec-19
SLC2A1 Full Gene Sequencing + MLPA Duplication/Deletion Analysis [MOL231] (GLUT1 deficiency syndrome 1)		SLC2A1 (138140)	Lavender Top (EDTA)	MNG Laboratories (www.medicalneurogenetics.com)/Dynacare	GLUT1 deficiency syndrome 1 (GLUT1DS1); Dystonia 9 (DYT9); GLUT1 deficiency syndrome 2 (GLUT1DS2); Epilepsy, idiopathic generalized, susceptibility to, 12 (EIG1)	Offered for free by MNG (The testing costs is billed directly to Ultragenyx Pharmaceutical who is developing a new treatment)		09-May-20
Soluble CD 163			Lavender Top (EDTA)	Hospital for Sick Children (Rapid Response Laboratory)	hemophagocytic lymphohistiocytosis (HLH)			
Soluble IL-2 Receptor Level (CD25)	927		EDTA plasma - 2 aliquots. Store in polypropylene tubes. Send frozen.	Hospital for Sick Children (Rapid Response Laboratory), 555 University, Toronto, CA	hemophagocytic lymphohistiocytosis (HLH)	ELISA	See also HSJ "Cellules T regulatrices"/Change to ARUP	23-Feb-22
Somatostatin (Somatotropin Release-Inhibiting Factor, SRIF)	193		EDTA plasma containing the G.I. Preservative (\$30). Freeze immediately.	InterScience Institute		Also at Mayo Clin Lab		13-Sep-18
SOTOS SYNDROME VIA THE NSD1 GENE [132]		NSD1 (60680)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Sotos' Syndrome (117550)	Sanger sequencing		
Spastic Paraplegia (NextGen Sequencing Panel and Copy Number Analysis; 380 Genes + mtDNA) [NGS337]	657	ABCD1 (300771), ACOX1 (609751), ACTB (102930), AFG3L2 (604881), ALDH3A1 (132936), ALZ1 (60552), AMPD1 (02771), APBB1 (007245), APBB1 (007244), AP4M1 (002296), AP4S1 (007243), KIAA0415 (613653), ARSA (007574), ARX (303582), ASSS (045574), ATLL (006490), ATXN2 (001517), AUIH (000526), BICAL10 (612571), B4GALNT1 (001673), BCRS (004805), BSL2 (000538), C12orf65 (613541), C19orf12 (614298), CCDC88C (611204), CCT3 (601500), CLPP (001190), CDASY (009855), CPTIC (008846), CTNBB1 (116896), CYP211 (010070), CYP7B1 (007111), DARS2 (003984), DCTN1 (601463), EDHD1 (614405), DDHD2 (615003), DYNCH1 (60012), EARS2 (612799), ELOVL4 (005512), ERLIN2 (611005), FA2H (611026), FARS2 (611592), FBXO7 (005646), FLNA (100017), FPC11 (612286), GAD1 (605305), GAN (005279), GBA (006603), GBA2 (604971), GBE1 (007039), GCDH (008801), GIC2 (608803), GLB1 (611458), GM2A (611309), GSS (60002), HARS2 (007078), HSD17B4 (601800), HSPD1 (110190), HRS1 (025136), FH1 (609951), KCNQ3 (002250), KDM5C (314090), KIAA0196 (610057), KIAA1052, KP2A (002591), KIF5A (002821), LICAM (308840), L3HREDH (009584), LAMB1 (150240), LARS2 (004544), LIAS (007031), LIPT1 (610284), LYRM1 (05381), MARS2 (009726), MCOLN1 (005248), MOC31 (603707), MOC32 (603708), MRE11A (NA), MRPS22 (005810), mtDNA (NA), MTPAP (613669), NIPA1 (608145), NTRC2 (000417), OPA3 (600830), PAFAH1B1 (010545), PDE8B (005709), PFKFB1 (009779), PLCA2 (003046), PLEKHA7 (008001), PNPLA6 (003197), PSAP (176801), PSEN1 (104311), REEP1 (009139), REEP2 (009347), RNASEH2B (601236), RTN2 (001832), SACS (004490), SCN2A (118290), SEPRIN3 (013039), SLC16A1 (300195), SLC6A1 (00052), SLC2A1 (138140), SLC3A10 (611146), SLC3A1 (603690), SOD1 (147450), SOX10 (002229), SPAST (004277), SPG11 (608444), SPG20 (007113), SPG21 (008181), SPG7 (002783), SPTAN1 (102300), STXBP1 (002926), TAF1 (213050), TARDBP (000785), TBC1D20 (611663), TEG (002498), TREX1 (006609), TTC19 (613814), TTR (170400), TUBA1A (002529), TUBG1 (191135), UROJLN2 (300264), UCHL1 (191342), VAMP1 (105886), VISTA (009027), VPS3 (610300), WDR61 (000200), WDR62 (611583), ZFYVE26 (612012), ZFYVE27 (610243)	Lavender Top (EDTA)	MNG Laboratories (www.medicalneurogenetics.com)/Dynacare	= paraparesis	NextGen Sequencing		09-May-20
Spastic paraplegia 3A, autosomal dominant (182600)	657	ATL1 (606439)	Lavender Top (EDTA)	Centogene AG (www.centogene.com)		1. single exon testing 2. full gene sequencing 3. deletion/duplication testing		

Appellation approuvée de l'analyse ou maladie associée (avec numéro de phénotype MIM) / Approved test name or Associated Disease (with MIM Phenotype Number)	Code Test / Test Code (Dictionnaire MSSS)	SYMBOL, HGNC DU GÈNE et NOM OMIM DU GÈNE / HGNC Gene Symbol and OMIM Gene Number	RÈGLES DE MANUTENTION / Handling Conditions	LABORATOIRE DÉSIGNÉ / Designated laboratory	RENSEIGNEMENTS SUR L'ANALYSE / Test details	MÉTHODE / Methode	NOTE PARTICULIÈRE / Special comment	Date de la révision / Date of revision
1,5-anhydroglucitol (GlycoMark) [500115] [GLYC]	1465		Gold Top or Red Top (Serum) or Lavender (plasma). Centrifuge and aliquot serum to transport tube within one hour of collection. Store and ship frozen. Stable at -20°C for 14 days.	LabCorp/Dynacare (https://www.labcorp.com/)		Enzymatic, colorimetric assay.		10-Jan-21
Spermatogenic Failure-6 (102530)		PG9 (SPATA16) (609856)	Lavender Top (EDTA)	Centogene AG (www.centogene.com)		1. single exon testing 2. full gene sequencing 3. deletion/duplication testing		
Spermatogenic Failure-9 (613958)		DPY9L2 (613895)	Lavender Top (EDTA)	Centogene AG (www.centogene.com)		1. single exon testing 2. full gene sequencing 3. deletion/duplication testing		
Spinal and Bulbar Muscular Atrophy: AR Trinucleotide Repeat Analysis	958	AR (313700)	Lavender Top (EDTA)	Molecular Genetics Laboratory, Hospital for Sick Children (www.sickkids.ca/PaediatricLaboratoryMedicine/Laboratories-Services/Molecular-Genetics-Laboratory/index.html)	Spinal and bulbar muscular atrophy of Kennedy	AR exon 1 trinucleotide (CAG) repeat analysis		25-Feb-19
Spinal Muscular Atrophy (NextGen Sequencing Panel and Copy Number Analysis; 21 Genes) [NGS347]	958	AR (313700), ASAH1 (613468), APTA (30001), BCL2 (609797), DNAB2 (604176), DYNC1H1 (600112), HSPB1 (602195), HSPB3 (604624), HSPB8 (608016), HSPB9 (606062), PLEKHA7 (61110), SIGIRR1 (605978), GPR172A (607882), SLC3A7 (608761), SMN1 (600354), SMN2 (601627), TBCD (604649), TRPV4 (605427), UBA1 (314370), VAPB (605704), VRI1 (602168)	Lavender Top (EDTA)	MNG Laboratories (www.medicalneurogenetics.com)/Dynacare		NextGen Sequencing		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)		Deletion /duplication via MLPA		
Spinocerebellar Ataxia (SCA) Panel (1,2,3,6,7,8,17)	584		Lavender Top (EDTA)	North York General	individuals tests 5378	Sanger sequencing	no del/dupl; See also MNG's MOL380	14-May-18
Spinocerebellar ataxia 1 (601556)	584	ATXN1 (601556)	Lavender Top (EDTA)	North York General	SCA Panel available	Sanger sequencing		05-Aug-20
Spinocerebellar ataxia 11 (604432)	584	TTBK2 (611695)	Lavender Top (EDTA)	Centogene AG (www.centogene.com)		1. single exon testing 2. full gene sequencing 3. deletion/duplication testing		
Spinocerebellar ataxia 17 (607136)	584	TBP (60075)	Lavender Top (EDTA)	North York General	SCA Panel available	Sanger sequencing		
Spinocerebellar ataxia 2 (183090)	584	ATXN2 (601517)	Lavender Top (EDTA)	North York General	SCA Panel available	Sanger sequencing		05-Aug-20
Spinocerebellar ataxia 6 (183086)	584	CACNA1A (601011)	Lavender Top (EDTA)	North York General	SCA Panel available	triplet expansion		
Spinocerebellar ataxia 7 (607640)	584	ATXN7 (607640)	Lavender Top (EDTA)	North York General	SCA Panel available	Sanger sequencing		
Spinocerebellar ataxia 8 (608768)	584	ATXN8 (613289)	Lavender Top (EDTA)	North York General	SCA Panel available	Sanger sequencing		
Spinocerebellar Ataxia Type 2 via the ATXN2 CAG Repeat Expansion [12976]	584	ATXN2 (601517)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)		FFLA		05-Jan-21
Spinocerebellar ataxia type 27 (609307)	584	PGF4 (60515)	Lavender Top (EDTA)	Centogene AG (www.centogene.com)		1. single exon testing 2. full gene sequencing 3. deletion/duplication testing		
Spinocerebellar ataxia, autosomal recessive-8 (French Canadian mutation Sanger sequencing panel) [247]	584	SYNE1 (68441)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Autosomal recessive cerebellar ataxia 1 (ARCA1; OMIM 610743)	Sanger sequencing		
SPINOCEREBELLAR ATAXIA, AUTOSOMAL RECESSIVE-8 VIA THE SYNE1 GENE EXONS 2-146 [246]	584	SYNE1 (68441)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Spinocerebellar Ataxia, Autosomal Recessive 8 (610743); Emery-Dreifuss Muscular Dystrophy 4, Autosomal Dominant (612998)	Sanger sequencing		
STAT Epilepsy Panel [814]	356	ALDH1A1, ARX, BRAT1, CDKL5, FOLR1, GLDC, KCNG2, KCNQ1, KCNT1, MIEP2, MIEP3, PCDH19, PPO4, POLG, SCN1A, SCN1B, SCN2A, SCN3A, SLC19A3, SLC21A1, SLC6A8, SPTAN1, STXBP1, TFP1, TSC1, TSC2	Lavender Top (EDTA) [2 tubes]	GeneDx (www.genedx.com)		NextGen Sequencing		28-Feb-20
Sterols, Plasma [STER]	1721		Plasma EDTA. Store frozen.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	Includes cholesterol			10-May-22
Stückler syndrome NGS panel [5127]	567	COL2A1, COL3A1, COL3A2, COL3A3, COL11A1, COL11A2, VCAN	Lavender Top (EDTA)	Connective Tissue Gene Test (www.ctgt.net)		NextGen Sequencing		
Sulfatides, Urine	1195		Random urine sample. Store at -20C.	Hospital for Sick Children (Rapid Response Laboratory)				17-May-22
Surfactant NGS Panel		ABCA3 (601645), NCKX2-1 (600655), SFTPB (178640), SFTPC (178620)	Lavender Top (EDTA)	Fulgent Genetics (fulgentdiagnostics.com)	Surfactant metabolism dysfunction, pulmonary, 2 (610913)			
Syntaxin 11		STX11 (605014)	Lavender Top (EDTA)	Hospital for Sick Children (Rapid Response Laboratory)	hemophagocytic lymphohistiocytosis (HLH) (603552)	Sanger sequencing		
T3 (Triiodothyronine), Reverse, Serum [RT3]	1649		Red Top (preferred); Gold SST. Store at 4°C (7 days) or frozen.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)		LC-MS/MS		04-Mar-21
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 (60889)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Tay-Sachs Disease (272800)	Sanger sequencing		
Telomere Length Measurement	1166		Lavender Top (EDTA) - 10 mL. Store at RT. Sample OK for 2 days only - same day send out.	Repeat Diagnostics (www.repeatdiagnostics.com)		Flow FISH		25-Jan-19

Appellation 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 (Déclinaire MSSS)	SYMBOL/BIEN DE GÈNE et NOM OMDM DU GÈNE / HGNC Gene Symbol and OMDM Gene Number	RÈGLES DE MANUTENTION / Handling Conditions	LABORATOIRE DÉSIGNÉ / Designated laboratory	RENSEIGNEMENTS SUR L'ANALYSE / Test details	MÉTHODE / Methode	NOTE PARTICULIÈRE / Special comment	Date de la révision / Date of revision
1,5-anhydroglucitol (GlycoMark) [500115] [GLYC]	1465		Gold Top or Red Top (Serum) or Lavender (plasma). Centrifuge and aliquot serum to transport tube within one hour of collection. Store and ship frozen. Stable at -20°C for 14 days.	LabCorp/Dynacare (https://www.labcorp.com/)		Enzymatic, colorimetric assay.		10-Jan-21
Thiosulfate, Serum/Plasma [4472SP]	92		Red Top (only) . Promptly separate into screw capped vial. No other tube type accepted. Serum stable for 24 days at 4°C or -20°C.	NMS Labs		LC-MS/MS		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		LC-MS/MS		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
Thrombocytopenia Panel [10307]	1076	ADAMTS13, ANKRD26, CYCS, GATA1, GP1BA, GP1BB, GP9, MASTL, MPL, MYH9, RUNX1, WAS	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)		Next Generation DNA Sequencing	MYH9 NOT available at HMR	18-Jan-22
ThyGenX	1680	Point mutations - KRAS, HRAS, NRAS, BRAF, PIK3CA Rearrangements/translocations - RET, PTC1, PAX8, PPARgamma	Fine needle aspirate	Groupe TMTC (https://grouptmtc.com)	TERT available at no added cost		ON HOLD. Requires approval by head of OOP	21-Jan-21
ThyGenX/ThyraMIR (reflex)	1680	Point mutations - KRAS, HRAS, NRAS, BRAF, PIK3CA Rearrangements/translocations - RET, PTC1, PAX8, PPARgamma	Fine needle aspirate	Groupe TMTC (https://grouptmtc.com)	TERT available at no added cost		ON HOLD. Requires approval by head of OOP	21-Jan-21
ThyroSeq v.3	1680		Fine needle aspirate	Groupe TMTC (https://grouptmtc.com) Le Groupe TMTC Molecular testing 181 Boul Hymus suite 200 Pointe Claire P.Q. 1.833.693.8960 Office 514-949-8962 Mobile		NextGen Sequencing	Requires approval by head of OOP	17-Nov-21
Thyrotropin Releasing hormone	1572		Thyrotropin Releasing Hormone must be collected with the TRH Preservative tube (\$30). Ship specimens frozen in dry ice.	InterScience Institute (www.interscienceinstitute.com)		RIA		01-Feb-18
Tissue Transglutaminase Abs (IgA & IgG) assay [TTIGAG]			Serum (Gold SST or Red Top)	Hospitals-In-Common		CLIA		16-Mar-17
Titanium, Plasma	202		Royal Blue EDTA only. No gel tubes. Separate within 30 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 frozen 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 antibody, hemolytic assay, CH50eq, APFA, FH, FI, C3, C4, FB, B1s, sC5b-9)			13-Jan-20
TMA Profile aHUS/TTP [7215217] (ADAMTS13 Activity; C3; C4; Factor B; Factor I; Factor H; Factor H Autoantibody)	1306			Cincinnati Children's Hospital (Diagnostic Immunology Laboratories)				January 14, 209
TP63-RELATED DISORDERS VIA THE TP63 GENE [834]		TP63 (60292)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	ADULT Syndrome (102285); Limb-Mammary Syndrome (603543); Hay-Wells Syndrome (106260); Rapp-Hodgkin Ectodermal Dysplasia Syndrome (129400); Split-Hand/Foot Malformation 4 (605289); Ectrodactyly, Ectodermal Dysplasia, And Clef Lip/Palate Syndrome 3 (604292)	Sanger sequencing		
TPSAB1 (Trypsinase) Copy Number Analysis	1395	TPSAB1	Lavender Top (EDTA)	Gene by Gene		ddPCR		20-Oct-21
Transthyretin amyloidosis (105210)	642	TTR (176300)	Lavender Top (EDTA)	BC Children's Hospital, Division of Genome Diagnostics				22-Mar-22
Trimethylamine (TMA) and TMA N-oxide (TMAO), Urine (quantitative) [L6949]	1468		Pre-load: Morning void urine. Freeze immediately. Post-load (5 g choline); collect urine 12-h after load. Freeze immediately.	Denver Genetics Laboratories (www.DenverGenetics.org)	trimethylaminuria	MS/MS	price per sample	14-Jan-19
Trypsinogen [TRGEN]			Serum. Store and send frozen	In-Common Laboratories	pancreatic dysfunction (e.g. CF)	RIA		
Tuberous Sclerosis Complex Panel [10661]	783	TSC1 (602940); TSC2 (191092)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Tuberous sclerosis-1 (191100); Tuberous sclerosis-2 (613254)	NextGen Sequencing + del/dupl (MLPA)		05-Feb-20
TYPE IV VOLTAGE-GATED SODIUM CHANNEL (ALPHA SUBUNIT)-RELATED DISORDERS VIA THE SCN4A GENE [11645]		SCN4A (603907)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Congenital Myasthenic Syndrome, Acetazolamide-Responsive (614198); Potassium Aggravated Myotonia (608390); Paramyotonia Congenita Of Von Eickenburg (168300); Hypokalemic Periodic Paralysis, Type 2 (613345); Hyperkalemic Periodic Paralysis; HYPP (170500)	Replaces Sanger sequencing [416]		05-Feb-20
Type VI Collagenopathy via the COL6A1 gene [11197]		COL6A1 (130220)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Ulrich congenital muscular dystrophy (254090); Bethlem myopathy (158810)	Replaces Sanger sequencing [487]		05-Feb-20

Appellation 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 BGENC DU GÈNE et NOM OMDM DU GÈNE / BGENC Gene Symbol and OMDM Gene Name	RÈGLES DE MANUTENTION / Handling Conditions	LABORATOIRE DÉSIGNÉ / Designated laboratory	RENSEIGNEMENTS SUR L'ANALYSE / Test details	MÉTHODE / Methode	NOTE PARTICULIÈRE / Special comment	Date de la révision / Date of revision
I,5-anhydroglucitol (GlycoMark) [500115] [GLYC]	1465		Gold Top or Red Top (Serum) or Lavender (plasma). Centrifuge and aliquot serum to transport tube within one hour of collection. Store and ship frozen. Stable at -20°C for 14 days.	LabCorp/Dynacare (https://www.labcorp.com/)		Enzymatic, colorimetric assay.		10-Jan-21
Type VI-Related Collagenopathy Panel [10183]		COL12A1, COL6A1 (120226), COL6A2 (120240), COL6A3 (120250)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Ulrich congenital muscular dystrophy (254090) and Bethlem myopathy (158810)	NextGen Sequencing + del/dupl		05-Feb-20
UBE3A Full Gene Sequencing Analysis [MOL093]		UBE3A (601623)	Lavender Top (EDTA)	MNG Laboratories (www.medicalneurogenetics.com)/Dynacare	Angelman Syndrome (105830)	Sanger sequencing	Requires local methylation study	14-May-20
UDP-Glucuronosyl Transferase 1A1 (UGT1A1), Full Gene Sequencing, Hyperbilirubinemia [UGTFG]	1340	UGT1A1 (191740)	Lavender Top (EDTA)	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)	diagnosis of Gilbert (143500) or Crigler-Najjar syndromes (218800 and 606785); irinotecan & others sensitivity	Sanger sequencing		Dec. 23, 2018
Unknown alpha-chain Hemoglobin Variants	820	HBA1 (141800),HBA2 (141850)	Lavender Top (EDTA)	McMaster University Medical Centre, Molecular Genetics Laboratory	also available at CHUM			13-Nov-19
Unknown beta-chain Hemoglobin Variants	820	HHB (141900)	Lavender Top (EDTA)	McMaster University Medical Centre, Molecular Genetics Laboratory	also available 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)			
Ustekinumab Quantitation with Antibodies, Serum [USTEK]	1465		Serum. Stable 21 d at 4°C (preferred) or -20°C.	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)				12-Jul-22
VALOSIN-CONTAINING PROTEIN-RELATED DISORDERS VIA THE VCP GENE [4807]		VCP (601023)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Inclusion body myopathy with early-onset Paget disease and frontotemporal dementia (167320)	NextGen Sequencing		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)	POEMS	ELISA		04-Mar-21
Vascular Endothelial Growth Factor D (VEGF-D)	213		Gold SST. Centrifuge and transfer serum to plastic container. Store at -20°C. Transport frozen.	Cincinnati Children's Hospital (Translational Trials Development and Support Laboratory (TTDSL))	LAM, TSC	ELISA		24-Feb-21
Vasoactive Intestinal Polypeptide (VIP)	211		SPECIAL BROWN TUBE. 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 Vermer-Morison's (Watery Diarrhea) Syndrome	radioimmunoassay	If paired to another test going to ISI	09-Nov-17
VGKC antibodies	54	LGII and Casp2 (Voltage gated potassium channel)	1st choice CSF; 2nd choice SERUM has reduced sensitivity	BC Neuroimmunology (bcneuro.ca)				09-Aug-22
Vitamin B3 and Metabolites, Plasma [VITB3]	218	nicotinic acid, nicotinamide (main form, NAD precursor), nicotinic acid (inactive metabolite)	Plasma EDTA. Store frozen. Stable for 28 days.	Mayo Clinical Laboratories (https://www.mayocliniclabs.com/index.html)	niacin	LC-MS/MS		09-Jun-22
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 (30 d). Send frozen.	Mayo Clinical Laboratories (https://www.mayocliniclabs.com/index.html)		LC-MS/MS		09-Jun-22
Voltage Gated Calcium Channel Antibodies (VGCC) [P91861]	53		Gold SST. Serum ONLY. Centrifuge and transfer serum to plastic container. Store at -20°C. Transport frozen.	BC Neuroimmunology (bcneuro.ca)	Lambert-Eaton Myasthenic Syndrome (LEMS)	RIPA		09-Aug-22
Von Hippel-Lindau Disease via VHL Gene [7523]	261	VHL (608537)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)		NGS		31-Oct-20
von Willebrand disease (193400)		VWF (617140)	Lavender Top (EDTA)	ARUP Laboratories			Sequence analysis of select exons	
Warburg Micro Syndrome (600118)		RAB3GAP2 (609275)	Lavender Top (EDTA)	University of Chicago Genetic Services Laboratories	Also available: 1. Warburg Micro syndrome Sequencing panel (RAB3GAP1, RAB3GAP2 and RAB18 sequencing) 2. Warburg Micro syndrome Deletion/Duplication panel (RAB3GAP1, RAB3GAP2 and RAB18 del/dup)	Sanger sequencing		
Weaver Syndrome (277590)		EGH2 (601570)	Lavender Top (2 x 4 mL)	Fulgent Genetics (fulgentdiagnostics.com)		NextGen Sequencing + del/dupl		
WILSON DISEASE / HEPATOENTICULAR DEGENERATION VIA THE ATP7B GENE [7871]	1185	ATP7B (608982)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Wilson Disease (277900)	NextGen Sequencing		31-Oct-20
Wolfram syndrome (222300)		WFS1 (606201)	Lavender Top (EDTA)	GeneDx		Part of NGS panel: Combined Mito Genome Plus Mito 140 Nuclear Gene Panel [615]		09-Dec-19
XIAP Protein Expression [XLP2]	817		Heparin	Alberta Precision Laboratories (Flow Cytometry - Calgary)	X-linked lymphoproliferative syndrome (XLP1) (308240)		To be accompanied by normal control sample	14-Oct-20
X-linked Adrenoleukodystrophy (300100)	849	ABCD1 (300371)	Lavender Top (EDTA)	EGL Genetics Laboratory 2460 Mountain Industrial, Tucker GA 30084 (geneticslab.emory.edu)		Sanger sequencing and del/dupl		
X-Linked Complete Congenital Stationary Night Blindness (CSNB1) via the NYX Gene [8705]		NYX (300278)	Lavender Top (EDTA)	Prevention Genetics (www.preventiongenetics.com)	Congenital Stationary night blindness, X-Linked, type 1A (310500)	NextGen Sequencing		31-Oct-20
Zinc Transporter 8 (ZnT8) Antibody, Serum [EZNT8]	1819		SST gel tube 3 cc	Mayo Clinical Laboratories (www.mayomedicallaboratories.com)				31-Oct-20