MOLECULAR TESTS

Catagories	Test	Gene OMIM	Disease	Phenotype OMIM	Comment	Test Specification	Price in €
Molecular Tests	AAAS (ALADIN, ADRACALIN)	605378	ACHALASIA-ADDISONIANISM-ALACRIMA SYNDROME, AAA » TRIPLE-A SYNDROME » ALACRIMA-ACHALASIA-ADRENAL INSUFFICIENCY NEUROLOGIC DISORDER » GLUCOCORTICOID DEFICIENCY AND ACHALASIA » ALLGROVE SYNDROME » ADDISONIAN-ACHALASIA SYNDROME » HYPOADRENALISM WITH ACHALASIA » ALACRIMA-ACHALASIA-ADDISONIANISM » ACTH-RESISTANT ADRENAL INSUFFICIENCY, ACHALASIA AND ALACRIMA	231550		Sequencing	1800
Molecular Tests	ABCA12 (ATP-BINDING CASSETTE, SUBFAMILY A, MEMBER 12)	607800	ICHTHYOSIS, CONGENITA, HARLEQUIN FETUS TYPE » HARLEQUIN ICHTHYOSIS	242500		Sequencing	5280
Molecular Tests	ABCA12 (ATP-BINDING CASSETTE, SUBFAMILY A, MEMBER 12)	607800	ICHTHYOSIS, LAMELLAR, 2, LI2 » LAMELLAR ICHTHYOSIS, TYPE 2 » CATANOSIS CONGENITA, 2A	601277		5 Exons: Exons 28-32	850
Molecular Tests	ABCA3 (ATP-BINDING CASSETTE, SUBFAMILY A, MEMBER 3; ATP-BINDING CASSETTE TRANSPORTER 3)	606615	SURFACTANT METABOLISM DYSFUNCTION, PULMONARY, TYPE 3 » PULMONARY ALVEOLAR PROTEINOSIS DUE TO ABCA3 DEFICIENCY » INTERSTITIAL LUNG DISEASE DUE TO ABCA3 DEFICIENCY	610921		Sequencing	2440
Molecular Tests	ABCA4 (ABCR)		STARGARDT DISEASE, TYPE 1 » MACULAR DEGENERATION, JUVENILE » FUNDUS FLAVIMACULATUS » CANG OF PHOTORECEPTOR OUTER SEGMENT, TYPE 2	248200		Sequencing	820
Molecular Tests	ABCB11 (ATP-BINDING CASSETTE, SUBFAMILY B, MEMBER 11)	605201	CHOLESTASIS, PROGRESSIVE FAMILIAL INTRAHEPATIC 1, PFIC1 » BYLER DISEASE	211600		Sequencing	1900
Molecular Tests	ABCB11 (ATP-BINDING CASSETTE, SUBFAMILY B, MEMBER 11)	605201	CHOLESTASIS, BENIGN RECURRENT INTRAHEPATIC, BRIC » SUMMERSKILL SYNDROME	243300		Sequencing	1900
Molecular Tests	ABCB4 (ATP-BINDING CASSETTE, SUBFAMILY B, MEMBER 4; MULTIDRUG RESISTANCE 3; MDR3; P-GLYCOPROTEIN 3; PGY3)	171060	CHOLESTASIS, PROGRESSIVE FAMILIAL INTRAHEPATIC 3, PFIC3 » MDR3 DEFICIENCY » CHOLESTASIS, PROGRESSIVE FAMILIAL INTRAHEPATIC, WITH ELEVATED GAMMA-GLUTAMYLTRANSFERASE	602347		Sequencing	2090
Molecular Tests	ABCB7 (ATP-BINDING CASSETTE, SUBFAMILY B, MEMBER 7, ABC TRANSPORTER 7)	300135	ANEMIA, SIDEROBLASTIC, AND SPINOCEREBELLAR ATAXIA	301310		Sequencing	850
Molecular Tests	ABCC2 (ATP-BINDING CASSETTE, SUBFAMILY C, MEMBER 2; MULTISPECIFIC ORGANIC ANION TRANSPORTER, CANALICULAR; CMOAT; MULTIDRUG RESISTANCE-ASSOCIATED PROTEIN 2; MRP2)	601107	DUBIN-JOHNSON SYNDROME » HYPERBILIRUBINEMIA, DUBIN-JOHNSON TYPE » MOTHBALL PIGMENTA, TYPE 2	237500		Sequencing	2200
Molecular Tests	ABCC6 (MULTIDRUG RESISTANCE-ASSOCIATED PROTEIN 6, MRP6)	603234	PSEUDOXANTHOMA ELASTICUM (AUTOSOMAL DOMINANT), PXE » GRONBLAD-STRANDBERG SYNDROME (AUTOSOMAL DOMINANT)	177850		Sequencing and Deletion-Duplication Testing	980
Molecular Tests	ABCC6 (MULTIDRUG RESISTANCE-ASSOCIATED PROTEIN 6, MRP6)	603234	PSEUDOXANTHOMA ELASTICUM (AUTOSOMAL DOMINANT), PXE » GRONBLAD-STRANDBERG SYNDROME (AUTOSOMAL DOMINANT)	177850		R1141X representing 30% of mutations	300
Molecular Tests	ABCC6 (MULTIDRUG RESISTANCE-ASSOCIATED PROTEIN 6, MRP6)	603234	PSEUDOXANTHOMA ELASTICUM (AUTOSOMAL DOMINANT), PXE » GRONBLAD-STRANDBERG SYNDROME (AUTOSOMAL DOMINANT)	177850		Deletion Exons 23 -29 representing 10% of mutations	500
Molecular Tests	ABCC6 (MULTIDRUG RESISTANCE-ASSOCIATED PROTEIN 6, MRP6)	603234	PSEUDOXANTHOMA ELASTICUM (AUTOSOMAL RECESSIVE), PXE » GRONBLAD-STRANDBERG SYNDROME (AUTOSOMAL RECESSIVE)	264800		Sequencing and Deletion-Duplication Testing	980
Molecular Tests	ABCC6 (MULTIDRUG RESISTANCE-ASSOCIATED PROTEIN 6, MRP6)	603234	PSEUDOXANTHOMA ELASTICUM (AUTOSOMAL RECESSIVE), PXE » GRONBLAD-STRANDBERG SYNDROME (AUTOSOMAL RECESSIVE)	264800		R1141X representing 30% of mutations	300
Molecular Tests	ABCC6 (MULTIDRUG RESISTANCE-ASSOCIATED PROTEIN 6, MRP6)	603234	PSEUDOXANTHOMA ELASTICUM (AUTOSOMAL RECESSIVE), PXE » GRONBLAD-STRANDBERG SYNDROME (AUTOSOMAL RECESSIVE)	264800		Deletion Exons 23 -29 representing 10% of mutations	500

Molecular Tests	ABCC8 (ATP-BINDING CASSETTE, SUBFAMILY C, MEMBER 8, SUR1)	600509	HYPERINSULINEMIC HYPOGLYCEMIA, FAMILIAL, TYPE 1 » PERSISTENT HYPERINSULINEMIC HYPOGLYCEMIA OF INFANCY » HYPOGLYCEMIA, HYPERINSULINEMIC, OF INFANCY » HYPERINSULINEMIC HYPOGLYCEMIA DUE TO FOCAL ADENOMATOUS HYPERPLASIA » NESIDIOBLASTOSIS OF PANCREAS » HYPERINSULINISM, FAMILIAL, WITH PANCREATIC NESIDIOBLASTOSIS	256450		Sequencing	3200
Molecular Tests	ABCC8 (ATP-BINDING CASSETTE, SUBFAMILY C, MEMBER 8, SUR1)	600509	HYPERINSULINEMIC HYPOGLYCEMIA, FAMILIAL, TYPE 1 » PERSISTENT HYPERINSULINEMIC HYPOGLYCEMIA OF INFANCY » HYPOGLYCEMIA, HYPERINSULINEMIC, OF INFANCY » HYPERINSULINEMIC HYPOGLYCEMIA DUE TO FOCAL ADENOMATOUS HYPERPLASIA » NESIDIOBLASTOSIS OF PANCREAS » HYPERINSULINISM, FAMILIAL, WITH PANCREATIC NESIDIOBLASTOSIS	256450		Deletion-Duplication Testing	350
Molecular Tests	ABCC8 (ATP-BINDING CASSETTE, SUBFAMILY C, MEMBER 8, SUR1)	600509	DIABETES MELLITUS, PERMANENT NEONATAL	606176		Sequencing	1300
Molecular Tests	ABCC9 (ATP-BINDING CASSETTE, SUBFAMILY C, MEMBER 9, SUR2)	601439	DIABETES MELLITUS, PERMANENT NEONATAL	606176		Deletion-Duplication Testing	350
Molecular Tests	ABCC9 (ATP-BINDING CASSETTE, SUBFAMILY C, MEMBER 9, SULFONYLUREA RECEPTOR 2, SUR2)	601439	CARDIOMYOPATHY, DILATED, TYPE 1O	608569		Sequencing	3360
Molecular Tests	ABCD1	300371	ADRENOLEUKODYSTROPHY, ALD » ADRENOMYELONEUROPATHY, AMN	300100		Sequencing	990
Molecular Tests	ACAD8 (ACYL-CoA DEHYDROGENASE FAMILY, MEMBER 8)	604773	ISOBUTYRYL-CoA DEHYDROGENASE DEFICIENCY » ISOBUTYRYL-CoA DEHYDROGENASE DEFICIENCY » ACADS DEFICIENCY	604773		Sequencing	850
Molecular Tests	ACAD9 (ACYL-CoA DEHYDROGENASE FAMILY, MEMBER 9)	611103	ACADV DEFICIENCY » ACYL-CoA DEHYDROGENASE FAMILY, MEMBER 9, DEFICIENCY OF	611126		Sequencing	2540
Molecular Tests	ACADM	607008	MEDIUM CHAIN ACYL-COA DEHYDROGENASE DEFICIENCY, MCAD » MEDIUM CHAIN ACYL-COA DEHYDROGENASE DEFICIENCY, MCAD	201450		Sequencing	900
Molecular Tests	ACADM	607008	MEDIUM CHAIN ACYL-COA DEHYDROGENASE DEFICIENCY, MCAD » MEDIUM CHAIN ACYL-COA DEHYDROGENASE DEFICIENCY, MCAD	201450		1 Mutation: K329E	250
Molecular Tests	ACADS (ACYL-CoA DEHYDROGENASE, SHORT-CHAIN)	606885	SCAD DEFICIENCY » SHORT-CHAIN ACYL-CoA DEHYDROGENASE DEFICIENCY	201470		Sequencing	610
Molecular Tests	ACADSB (ACYL-CoA DEHYDROGENASE, SHORT/BRANCHED CHAIN)	600301	2-ALPHA-METHYLBUTYRYLGLYCINURIA » 2-METHYLBUTYRYL-COA DEHYDROGENASE DEFICIENCY	600301		Sequencing	900
Molecular Tests	ACADVL (ACYL-CoA DEHYDROGENASE, VERY LONG-CHAIN)	609575	ACYL-CoA DEHYDROGENASE, VERY LONG-CHAIN, DEFICIENCY OF	201475		Sequencing	780
Molecular Tests	ACAT1 (ACETYL-CoA ACETYLTRANSFERASE 1; ACETOACETYL-CoA THIOLASE, MITOCHONDRIAL)	607809	ALPHA-METHYLACETOACETIC ACIDURIA » BETA-KETOTHIOLASE DEFICIENCY » MITOCHONDRIAL ACETOACETYL-CoA THIOLASE DEFICIENCY » 2-METHYL-3-HYDROXYBUTYRYL-COENZYME A DEHYDROGENASE	204750		Sequencing	1620
Molecular Tests	ACE (ANGIOTENSIN I-CONVERTING ENZYME;DIPEPTIDYL CARBOXYPEPTIDASE 1, KININASE 2)	106180	RENAL TUBULAR DYSGENESIS » RENAL TUBULAR DYSGENESIS, WITH CHOANAL ATRESIA AND ATHELIA	267430		Sequencing	1800
Molecular Tests	ACOX1 (ACYL-CoA OXIDASE 1, PALMITOYL; ACYL-CoA OXIDASE, PALMITOYL, PEROXISOMAL; PALMITOYL-CoA OXIDASE, ACYL-CoA OXIDASE, STRAIGHT-CHAIN; SCOX)	609751	PEROXISOMAL ACYL-CoA OXIDASE DEFICIENCY » STRAIGHT-CHAIN ACYL-CoA OXIDASE DEFICIENCY » PEROXISOMAL ACYL-COENZYME A OXIDASE DEFICIENCY	264470		Sequencing	1000
Molecular Tests	ACTA1 (ACTIN)	102610	ACTIN MYOPATHY	161600		Sequencing	500
Molecular Tests	ACTA1 (ACTIN)	102610	NEMALINE MYOPATHY 3; NEM3	161800		Sequencing	500
Molecular Tests	ACTA1 (ACTIN)	102610	MYOPATHY, CONGENITAL, WITH FIBER-TYPE DISPROPORTION, FETD	255310		Sequencing	500
Molecular Tests	ACTA1 (ACTIN)	102610	NEMALINE MYOPATHY 2; NEM2	256060		Sequencing	500
Molecular Tests	ACTA2 (ACTIN, ALPHA 2, SMOOTH MUSCLE, AORTA)	102620	AORTIC ANEURYSM, FAMILIAL THORACIC, TYPE 6	611788		Sequencing	550
Molecular Tests	ACTA2 (ACTIN, ALPHA, CARDIAC MUSCLE, SMOOTH MUSCLE)	102540	THE ACTIVITY ARRHYTHMOPATHY			Sequencing	550
Molecular Tests	ACTC1 (ACTIN, ALPHA, CARDIAC MUSCLE, SMOOTH MUSCLE ACTIN)	102540	HYPERTROPHIC CARDIOMYOPATHY, FAMILIAL, 1, CMH1 » VENTRICULAR HYPERTROPHY, HEREDITARY » ASYMMETRIC SEPTAL HYPERTROPHY » CARDIOMYOPATHY, FAMILIAL HYPERTROPHIC	192600		Sequencing	550
Molecular Tests	ACTC1, MYL2, MYL3		HYPERTROPHIC CARDIOMYOPATHY, FAMILIAL » VENTRICULAR HYPERTROPHY, HEREDITARY » ASYMMETRIC SEPTAL HYPERTROPHY » CARDIOMYOPATHY, FAMILIAL HYPERTROPHIC			19 Exons	1350
Molecular Tests	ACTN2 (ACTININ, ALPHA 2)	102575	CARDIOMYOPATHY, DILATED, TYPE 1AA	613756		Sequencing	1680
Molecular Tests	ACTN4 (ACTININ, ALPHA 4)	604638	FOCAL SEGMENTAL GLOMERULOSCLEROSIS, TYPE 1	603278		Sequencing	1100

Molecular Tests	ACVR1 (ACTIVIN A RECEPTOR, TYPE 1; ACTIVIN RECEPTOR-LIKE KINASE 2; ALK2)	102576	FIBRODYSPLASIA OSSIFICANS PROGRESSIVA, FOP	135100		c.617G>A (Arg206His)	510
Molecular Tests	ADA (ADENOSINE DEAMINASE, ADENOSINE AMINOHYDROLASE)	608958	SEVERE COMBINED IMMUNODEFICIENCY, (AUTOSOMAL RECESSIVE), T CELL-NEGATIVE, B CELL-NEGATIVE, NK CELL-NEGATIVE, DUE TO ADENOSINE DEAMINASE DEFICIENCY, SCID	102700		Sequencing	1500
Molecular Tests	ADAM9 (A DISINTEGRIN AND METALLOPROTEINASE DOMAIN 9; MYELOMA CELL METALLOPROTEINASE; MCMP; METALLOPROTEINASE-LIKE, DISINTEGRIN-LIKE, AND CYSTEINE-RICH PROTEIN 9; MDC9)	602713	CONE-ROD DYSTROPHY, TYPE 9, CORD9	612775		Sequencing	820
Molecular Tests	ADAMTS10 (A DISINTEGRIN-LIKE AND METALLOPROTEINASE WITH THROMBOSPONDIN TYPE 1 MOTIF, 10)	608990	WEILL-MARCHESANI SYNDROME (AUTOSOMAL RECESSIVE) » SPHEROPHAKIA-BRACHYMORPHIA SYNDROME » MESODERMAL DYSMORPHODYSTROPHY, CONGENITAL	277600		Sequencing	1970
Molecular Tests	ADAMTS13 (VON WILLEBRAND FACTOR-CLEAVING PROTEASE)	604134	HEMOLYTIC-UREMIC SYNDROME » COMBINED DEFICIENCY OF FACTOR H AND FACTOR H-LIKE 1	235400		Sequencing	1300
Molecular Tests	ADAMTS13 (VON WILLEBRAND FACTOR-CLEAVING PROTEASE)	604134	THROMBOTIC THROMBOCYTOPENIC PURPURA, CONGENITAL, TTP » SCHULMAN-UPSHAW SYNDROME	274150		Sequencing	1300
Molecular Tests	ADAMTS18 (A DISINTEGRIN-LIKE AND METALLOPROTEINASE WITH THROMBOSPONDIN TYPE 1 MOTIF, 10)	607929	WEILL-MARCHESANI SYNDROME (AUTOSOMAL RECESSIVE) » SPHEROPHAKIA-BRACHYMORPHIA SYNDROME » MESODERMAL DYSMORPHODYSTROPHY, CONGENITAL	277600		Sequencing	1970
Molecular Tests	ADAMTS2 (A DISINTEGRIN-LIKE AND METALLOPROTEINASE WITH THROMBOSPONDIN TYPE 1 MOTIF, 2)	604539	EHLERS-DANLOS SYNDROME, TYPE 7 (AUTOSOMAL RECESSIVE), TYPE 7, EDS7C » EHLERS-DANLOS SYNDROME, DERMATOSPARAXIS TYPE	225410		Sequencing	600
Molecular Tests	ADAMTSL4 (ADAMTS-LIKE 4; THROMBOSPONDIN REPEAT-CONTAINING 1; TSRC1)	610113	ECTOPIA LENTIS, ISOLATED (AUTOSOMAL RECESSIVE)	225100		Sequencing	720
Molecular Tests	ADCK3 (AARF DOMAIN-CONTAINING KINASE 3; CABC1)	606980	COENZYME Q10 DEFICIENCY, PRIMARY, TYPE 4 » SPINOCEREBELLAR ATAXIA (AUTOSOMAL RECESSIVE), TYPE 9, SCAR9	612016		Sequencing	1480
Molecular Tests	ADSL (ADENYLOSUCCINATE LYASE)	103050	ADENYLOSUCCINASE DEFICIENCY » SUCCINYLPURINEMIC AUTISM	103050		Sequencing	1150
Molecular Tests	AGL (AMYLO-1,6-GLUCOSIDASE, 4-ALPHA-GLUCANOTRANSFER, GLYCOGEN DEBRANCHER ENZYME)	610860	GLYCOGEN STORAGE DISEASE, TYPE 3 » GSD TYPE 3 » FORBES DISEASE » CORI DISEASE » AMYLO-1,6-GLUCOSIDASE DEFICIENCY	232400		Sequencing	1730
Molecular Tests	AGPAT2 (1-@ACYLGLYCEROL-3-PHOSPHATE O-ACYLTRANSFERASE 2)	603100	LIPODYSTROPHY, CONGENITAL GENERALIZED, TYPE 1 » BERARDINELLI-SEIP CONGENITAL LIPODYSTROPHY, TYPE 1 » BRUNZELL SYNDROME, AGPAT2-RELATED	608594		Sequencing	550
Molecular Tests	AGPS (ALKYLGLYCERONE-PHOSPHATE SYNTHASE; ALKYLDIHYDROXYACETONEPHOSPHATE SYNTHASE; ADHAPS; ALKYL-DHAP SYNTHASE)	603051	RHIZOMELIC CHONDRODYSPLASIA PUNCTATA, TYPE 3, RCDP3 » ALKYLDIHYDROXYACETONEPHOSPHATE SYNTHASE DEFICIENCY	600121		Sequencing	745
Molecular Tests	AGT (ANGIOTENSINOGEN, SERPINA8, ANGIOTENSIN)	106150	RENAL TUBULAR DYSGENESIS » RENAL TUBULAR DYSGENESIS WITH CHOANAL ATRESIA AND ATHELIA	267430		Sequencing	700
Molecular Tests	AGTR1 (ANGIOTENSIN RECEPTOR 1)	106165	RENAL TUBULAR DYSGENESIS » RENAL TUBULAR DYSGENESIS WITH CHOANAL ATRESIA AND ATHELIA	267430		Sequencing	400
Molecular Tests	AGXT (ALANINE-GLYOXYLATE AMINOTRANSFERASE, AGT, SERINE-PYRUVATE AMINOTRANSFERASE, SPT)	604285	HYPEROXALURIA, PRIMARY, TYPE 1 » OXALOSIS 1 » GLYCOLIC ACIDURIA » ALANINE-GLYOXYLATE AMINOTRANSFERASE DEFICIENCY » HEPATIC AGT DEFICIENCY	259900		Sequencing	700
Molecular Tests	AGXT (ALANINE-GLYOXYLATE AMINOTRANSFERASE, AGT, SERINE-PYRUVATE AMINOTRANSFERASE, SPT)	604285	HYPEROXALURIA, PRIMARY, TYPE 1 » OXALOSIS 1 » GLYCOLIC ACIDURIA » ALANINE-GLYOXYLATE AMINOTRANSFERASE DEFICIENCY » HEPATIC AGT DEFICIENCY	259900		Deletion-Duplication Testing	350
Molecular Tests	AGXT (ALANINE-GLYOXYLATE AMINOTRANSFERASE, AGT, SERINE-PYRUVATE AMINOTRANSFERASE, SPT)	604285	HYPEROXALURIA, PRIMARY, TYPE 1 » OXALOSIS 1 » GLYCOLIC ACIDURIA » ALANINE-GLYOXYLATE AMINOTRANSFERASE DEFICIENCY » HEPATIC AGT DEFICIENCY	259900		3 Exons: 1, 4 and 7 (Including 33-34insC, 508A, 731C Mutations)	255
Molecular Tests	AHI1 (ABELSON HELPER INTEGRATION SITE 1, JOUBERIN)	608894	JOUBERT SYNDROME, TYPE 3	608629		Sequencing	2600
Molecular Tests	AHI1 (ABELSON HELPER INTEGRATION SITE 1, JOUBERIN)	608894	JOUBERT SYNDROME, TYPE 3	608629		Deletion-Duplication Testing	580
Molecular Tests	AICARDI-GOUTIERES PANEL		AICARDI-GOUTIERES, TYPE 1-5			5 Genes: AGS1-5	1830

Molecular Tests	AICDA (ACTIVATION-INDUCED CYTIDINE DEAMINASE, AID)	605257	IMMUNODEFICIENCY WITH HYPER-IgM, TYPE 2 » HYPER-IgM SYNDROME 2	605258		Sequencing	1300
Molecular Tests	AIP (ARYL HYDROCARBON RECEPTOR-INTERACTING PROTEIN; HEPATITIS B VIRUS X-ASSOCIATED PROTEIN 2; XAP2)	605555	PITUITARY ADENOMA, GROWTH HORMONE-SECRETING » SOMATOTROPINOMA, FAMILIAL ISOLATED » ACROMEGALY DUE TO PITUITARY ADENOMA	102200		Sequencing	660
Molecular Tests	AIP (ARYL HYDROCARBON RECEPTOR-INTERACTING PROTEIN; HEPATITIS B VIRUS X-ASSOCIATED PROTEIN 2; XAP2)	605555	PITUITARY ADENOMA, ACTH-SECRETING » CUSHING DISEASE, PITUITARY	219090		Sequencing	660
Molecular Tests	AIP (ARYL HYDROCARBON RECEPTOR-INTERACTING PROTEIN; HEPATITIS B VIRUS X-ASSOCIATED PROTEIN 2; XAP2)	605555	PITUITARY ADENOMA, PROLACTIN-SECRETING » PROLACTINOMA, FAMILIAL	600634		Sequencing	660
Molecular Tests	AIPL1 (ARYLHYDROCARBON-INTERACTING RECEPTOR PROTEIN-LIKE 1)	604392	LEBER CONGENITAL AMAUROSIS, TYPE 4, LCA4 » RETINITIS PIGMENTOSA, JUVENILE, AIPL1-RELATED » CONE-ROD DYSTROPHY, AIPL1-RELATED	604393		Sequencing	530
Molecular Tests	AIRE (AUTOIMMUNE REGULATOR)	607358	AUTOIMMUNE POLYENDOCRINOPATHY SYNDROME, TYPE 1 » AUTOIMMUNE POLYENDOCRINOPATHY-CANDIDIASIS-ECTODERMAL DYSTROPHY, APECED » AUTOIMMUNE POLYGLANDULAR SYNDROME, TYPE 1 » HYPOADRENOCORTICISM WITH HYPOPARATHYROIDISM AND SUPERFICIAL MONILIASIS	240300		Sequencing	1620
Molecular Tests	ALAD (DELTA-AMINOLEVULINATE DEHYDRATASE; ALADH; PORPHOBILINOGEN SYNTHASE; PBGS)	125270	PORPHYRIA, ACUTE HEPATIC » DELTA-AMINOLEVULINATE DEHYDRATASE DEFICIENCY » PORPHOBILINOGEN SYNTHASE DEFICIENCY » LEAD POISONING, SUSCEPTIBILITY TO	612740		Sequencing	600
Molecular Tests	ALAS2 (DELTA-AMINOLEVULINATE SYNTHASE 2; ALASE)	301300	ANEMIA, SIDEROBLASTIC (X-LINKED) ANEMIA, HYPOCHROMIC HEREDITARY IRON-LOADING ANEMIA	300751		Sequencing	320
Molecular Tests	ALAS2 (DELTA-AMINOLEVULINATE SYNTHASE 2; ALASE)	301300	PROTOPORPHYRIA, ERYTHROPOIETIC (X-LINKED DOMINANT)	300752		Sequencing	320
Molecular Tests	ALBINISM PANEL : TYR (OCA1), P (OCA2), TYRP1 (OCA3), MATP (OCA4), GPR143 (OA1)		ALBINISM			Sequencing of 5 Genes	1960
Molecular Tests	ALDH18A1 (ALDEHYDE DEHYDROGENASE 18 FAMILY, MEMBER A1; 1-PYRROLINE-5-CARBOXYLATE SYNTHETASE; GLUTAMATE GAMMA-SEMIALDEHYDE SYNTHETASE)	138250	CUTIS LAXA, TYPE 3A (AUTOSOMAL RECESSIVE)	219150		Sequencing	600
Molecular Tests	ALDH18A1 (ALDEHYDE DEHYDROGENASE 18 FAMILY, MEMBER A1; 1-PYRROLINE-5-CARBOXYLATE SYNTHETASE; GLUTAMATE GAMMA-SEMIALDEHYDE SYNTHETASE)	138250	SPASTIC PARAPLEGIA 9A (AUTOSOMAL DOMINANT), SPG9A	601162		Sequencing	600
Molecular Tests	ALDH18A1 (ALDEHYDE DEHYDROGENASE 18 FAMILY, MEMBER A1; 1-PYRROLINE-5-CARBOXYLATE SYNTHETASE; GLUTAMATE GAMMA-SEMIALDEHYDE SYNTHETASE)	138250	SPASTIC PARAPLEGIA 9B (AUTOSOMAL RECESSIVE), SPG9B	616586		Sequencing	600
Molecular Tests	ALDH18A1 (ALDEHYDE DEHYDROGENASE 18 FAMILY, MEMBER A1; 1-PYRROLINE-5-CARBOXYLATE SYNTHETASE; GLUTAMATE GAMMA-SEMIALDEHYDE SYNTHETASE)	138250	CUTIS LAXA, TYPE 3 (AUTOSOMAL DOMINANT)	616603		Sequencing	600
Molecular Tests	ALDH3A2 (FALDH, ALDH10)	609523	SJOGREN-LARSSON SYNDROME » FATTY ALDEHYDE DEHYDROGENASE DEFICIENCY	270200		Sequencing	1560
Molecular Tests	ALDH7A1 (ATQ1; ALDEHYDE DEHYDROGENASE 7 FAMILY, MEMBER A1; ANTIQUITIN; ALPHA AMINO-ADIPIC SEMIALDEHYDE DEHYDROGENASE)	107323	EPILEPSY, PYRIDOXINE-DEPENDENT, EPD » AASA DEHYDROGENASE DEFICIENCY	266100		Sequencing	810
Molecular Tests	ALDOB (ALDOLASE B)	229600	FRUCTOSE INTOLERANCE » FRUCTOSEMIA » FRUCTOSE-1-PHOSPHATE » ALDOLASE B DEFICIENCY	229600		Sequencing	650
Molecular Tests	ALDOB (ALDOLASE B)	229600	FRUCTOSE INTOLERANCE » FRUCTOSEMIA » FRUCTOSE-1-PHOSPHATE » ALDOLASE B DEFICIENCY	229600		3 Common Mutations: A149P, A174D, N334K	250
Molecular Tests	ALG12 (ALG12, S. CEREVISIAE, HOMOLOG OF; ASPARAGINE-LINKED GLYCOSYLATION 12, HOMOLOG OF; DOLICHYL-P-MANNOSE:MAN-7-GlcNAc-2-PP-DOLICHYL-ALPHA-6-MANNOSYLTRANSFERASE)	607144	CONGENITAL DISORDER OF GLYCOSYLATION, TYPE 1G, CDG1G	607143		Sequencing	890
Molecular Tests	ALG6 (ALG6, S. CEREVISIAE, HOMOLOG OF)	604566	CONGENITAL DISORDER OF GLYCOSYLATION, TYPE 1C, CDG1C » CARBOHYDRATE-DEFICIENT GLYCOPROTEIN SYNDROME, TYPE I, WITH DEFICIENT GLYCOSYLATION OF DOLICHOL-LINKED OLIGOSACCHARIDE, FORMERLY	603147		Sequencing	1320
Molecular Tests	ALG8 (ALG8, S. CEREVISIAE, HOMOLOG OF)	608103	CONGENITAL DISORDER OF GLYCOSYLATION, TYPE 1H, CDG1H	608104		Sequencing	1240
Molecular Tests	ALG9 (ALG9, S. CEREVISIAE, HOMOLOG OF)	606941	CONGENITAL DISORDER OF GLYCOSYLATION, TYPE 1L, CDG1L	608776		Sequencing	1320
Molecular Tests	ALK1 (ACTIVIN A RECEPTOR, TYPE II-LIKE 1, ACVRL1)	601284	TELANGIECTASIA, HEREDITARY HEMORRHAGIC, OF RENDU, OSLER, AND WEBER, TYPE 2 » OSLER-RENDU-WEBER DISEASE » ORW DISEASE	600376		Sequencing	960
Molecular Tests	ALMS1	606844	ALSTROM SYNDROME, ALMS	203800		Sequencing	1100

Molecular Tests	ALOX12B (ARACHIDONATE 12-LIPOXYGENASE, R TYPE, 12R-@LIPOXYGENASE)	603741	ICHTHYOSIFORM ERYTHRODERMA, NONBULLOUS CONGENITAL	242100		Sequencing	1760
Molecular Tests	ALOXE3 (ARACHIDONATE LIPOXYGENASE 3, LIPOXYGENASE TYPE 3)	607206	ICHTHYOSIFORM ERYTHRODERMA, NONBULLOUS CONGENITAL	242100		Sequencing	1740
Molecular Tests	ALPL (ALKALINE PHOSPHATASE, LIVER; ALKALINE PHOSPHATASE, LIVER/BONE/KIDNEY TYPE)	171760	HYPOPHOSPHATASIA, ADULT » ODONTOHYPOPHOSPHATASIA	146300		Sequencing	600
Molecular Tests	ALPL (ALKALINE PHOSPHATASE, LIVER; ALKALINE PHOSPHATASE, LIVER/BONE/KIDNEY TYPE)	171760	HYPOPHOSPHATASIA, INFANTILE » HYPOPHOSPHATASIA, PERINATAL LETHAL	241500		Sequencing	600
Molecular Tests	ALX4 (ARISTALESS-LIKE 4, MOUSE, HOMOLOG OF)	605420	PARIETAL FORAMINA, TYPE 2 » FORAMINA PARIETALIA PERMAGNA » CATLIN MARKS	609597		Sequencing	900
Molecular Tests	AMACR (ALPHA-METHYLACYL-CoA RACEMASE)	604489	BILE ACID SYNTHESIS DEFECT, CONGENITAL, TYPE 4 » CHOLESTASIS, INTRAHEPATIC, WITH DEFECTIVE CONVERSION OF TRIHYDROXYCOPROSTANIC ACID TO CHOLIC ACID	214950		Sequencing	600
Molecular Tests	AMACR (ALPHA-METHYLACYL-CoA RACEMASE)	604489	ALPHA-METHYLACYL-CoA RACEMASE DEFICIENCY	614307		Sequencing	600
Molecular Tests	AMPD1 (AMP DEAMINASE)	102770	MYOADENYLATE DEAMINASE DEFICIENCY	102770		Sequencing	1770
Molecular Tests	AMPD1 (AMP DEAMINASE)	102770	MYOADENYLATE DEAMINASE DEFICIENCY	102770		2 Mutations: Q12X, P48L	250
Molecular Tests	AMT (AMINOMETHYLTRANSFERASE, GLYCINE CLEAVAGE SYSTEM T PROTEIN)	238310	NONKETOTIC HYPERGLYCINEMIA » GLYCINE ENCEPHALOPATHY	605899		Sequencing	850
Molecular Tests	ANG (ANGIOGENIN, RNASE5)	105850	AMYOTROPHIC LATERAL SCLEROSIS, TYPE 1, ALS1	105400		Sequencing	450
Molecular Tests	AP1S2 (ADAPTOR-RELATED PROTEIN COMPLEX 1, SIGMA-2 SUBUNIT; CLATHRIN-ASSOCIATED/ASSEMBLY/ADAPTOR PROTEIN, SMALL 1-LIKE)	300629	MENTAL RETARDATION, NONSPECIFIC (X-LINKED), TYPE 59, MRX59	300630		Sequencing	610
Molecular Tests	AP3B1 (ADAPTOR-RELATED PROTEIN COMPLEX 3, BETA-1 SUBUNIT; ADAPTIN, BETA-3A; ADTB3A; HPS2)	603401	HERMANSKY-PUDLAK SYNDROME, TYPE 2, HPS2	608233		Sequencing	870
Molecular Tests	APC	175100	POLYPOSIS COLI, ADENOMATOUS » FAMILIAL ADENOMATOUS POLYPOSIS, FAP » ATTENUATED POLYPOSIS COLI	175100		Sequencing	790
Molecular Tests	APC	175100	POLYPOSIS COLI, ADENOMATOUS » FAMILIAL ADENOMATOUS POLYPOSIS, FAP » ATTENUATED POLYPOSIS COLI	175100		Deletion-Duplication Testing	680
Molecular Tests	APOA1 (APOLIPOPROTEIN A-1)	107680	HYPO-ALPHALIPOPROTEINEMIA	604091		L178P Mutation	300
Molecular Tests	APOB (APOLIPOPROTEIN B, APOB100, APOB48)	107730	HYPOBETALIPOPROTEINEMIA, FAMILIAL » ABETALIPOPROTEINEMIA, NORMOTRIGLYCERIDEMIC, STEINBERG TYPE » HYPOBETALIPOPROTEINEMIA, FAMILIAL » ACANTHOCYTOSIS WITH HYPOBETALIPOPROTEINEMIA	107730		Sequencing	3950
Molecular Tests	APOB (APOLIPOPROTEIN B, APOB100, APOB48)	107730	HYPERCHOLESTEROLEMIA	143890		3 mutations: R3500Q, R3500W	250
Molecular Tests	APOB (APOLIPOPROTEIN B, APOB100, APOB48)	107730	HYPERCHOLESTEROLEMIA (AUTOSOMAL DOMINANT), TYPE B » APOLIPOPROTEIN B-100, FAMILIAL LIGAND - DEFECTIVE	144010		2 Mutations: R3500Q and R3531C	250
Molecular Tests	APOC2 (APOLIPOPROTEIN C2)	207750	HYPERCHYLOMICRONEMIA » APOLIPOPROTEIN C2 DEFICIENCY	207750		Sequencing	350
Molecular Tests	APOE (APOLIPOPROTEIN E)	107741	DYSBETALIPOPROTEINEMIA DUE TO DEFECT IN APOLIPOPROTEIN E » APOLIPOPROTEIN E DEFICIENCY » HYPERLIPOPROTEINEMIA, TYPE 3 » FAMILIAL HYPERBETA- AND PREBETALIPOPROTEINEMIA	107741		Sequencing	300
Molecular Tests	APP	104760	ALZHEIMER DEMENTIA, EARLY-ONSET, TYPE 1, AD1	104300		Sequencing	700
Molecular Tests	APP	104760	ALZHEIMER DEMENTIA, EARLY-ONSET, TYPE 1, AD1	104300		Exons 16 and 17 harbouring the majority of mutations	599
Molecular Tests	APP	104760	ALZHEIMER DEMENTIA, EARLY-ONSET, TYPE 1, AD1	104300		Deletion-Duplication Testing	600
Molecular Tests	APP	104760	CEREBRAL AMYLOID ANGIOPATHY » AMYLOIDOSIS, CEREBROARTERIAL » AMYLOIDOSIS, TYPE 6 » HEREDITARY CEREBRAL HEMORRHAGE WITH AMYLOIDOSIS, HCHWA	105150		Sequencing	700
Molecular Tests	APP	104760	CEREBRAL AMYLOID ANGIOPATHY » AMYLOIDOSIS, CEREBROARTERIAL » AMYLOIDOSIS, TYPE 6 » HEREDITARY CEREBRAL HEMORRHAGE WITH AMYLOIDOSIS, HCHWA	105150		Exons 16 and 17 harbouring the majority of mutations	599

Molecular Tests	APP	104760	CEREBRAL AMYLOID ANGIOPATHY » AMYLOIDOSIS, CEREBROARTERIAL » AMYLOIDOSIS, TYPE 6 » HEREDITARY CEREBRAL HEMORRHAGE WITH AMYLOIDOSIS, HCHWA	105150		Deletion-Duplication Testing	600
Molecular Tests	APRT (ADENINE PHOSPHORIBOSYLTRANSFERASE)	102600	APRT DEFICIENCY	102600		Sequencing	Upon Request
Molecular Tests	APTX (APRATAXIN)	606350	ATAXIA, EARLY-ONSET, WITH OCULOMOTOR APRAXIA AND HYPOALBUMINEMIA » ATAXIA-OCULOMOTOR APRAXIA SYNDROME » ATAXIA-OCULOMOTOR APRAXIA 1 » ATAXIA-TELANGIECTASIA-LIKE SYNDROME » CEREBELLAR ATAXIA, EARLY-ONSET, WITH HYPOALBUMINEMIA	208920		Sequencing	650
Molecular Tests	AQP2 (AQUAPORIN 2)	107777	DIABETES INSIPIDUS, NEPHROGENIC (AUTOSOMAL DOMINANT)	125800		Sequencing	320
Molecular Tests	AQP2 (AQUAPORIN 2)	107777	DIABETES INSIPIDUS, NEPHROGENIC (AUTOSOMAL RECESSIVE)	222000		Sequencing	320
Molecular Tests	AR (ANDROGEN RECEPTOR)	313700	HYPOSPADIAS (X-LINKED)			Sequencing	695
Molecular Tests	AR (ANDROGEN RECEPTOR)	313700	ANDROGEN INSENSITIVITY SYNDROME, AIS	300068		Sequencing	695
Molecular Tests	AR (ANDROGEN RECEPTOR)	313700	REIFENSTEIN SYNDROME	312300		Sequencing	695
Molecular Tests	AR (ANDROGEN RECEPTOR)	313700	KENNEDY DISEASE » SPINAL AND BULBAR MUSCULAR ATROPHY, SBMA	313200		Repeat	450
Molecular Tests	ARG1 (ARGINASE, LIVER)	608313	ARGININEMIA » ARGINASE DEFICIENCY » HYPERARGININEMIA	612714		Sequencing	940
Molecular Tests	ARH	605747	HYPERCHOLESTEROLEMIA (AUTOSOMAL RECESSIVE), ARH	603813		Sequencing	700
Molecular Tests	ARHGAP31 (RHO GTPase-ACTIVATING PROTEIN 31;CDC42 GTPase-ACTIVATING PROTEIN)	610911	ADAMS-OLIVER SYNDROME, TYPE 1 » APLASIA CUTIS CONGENITA WITH TERMINAL TRANSVERSE LIMB DEFECTS	100300		Exon 12 (all variants identified so far)	550
Molecular Tests	ARHGEF9 (RHO GUANINE NUCLEOTIDE EXCHANGE FACTOR 9; COLLYBISTIN)	300429	HYPEREKPLEXIA AND EPILEPSY	300607		Sequencing	650
Molecular Tests	ARIX (ARISTALESS HOMEOBOX, DROSOPHILA, HOMOLOG, PHOX2A)	602753	FIBROSIS OF EXTRAOCULAR MUSCLES, CONGENITAL, TYPE2, FEOM2, CFEOM2	602078		Sequencing	250
Molecular Tests	ARL13B (ADP-RIBOSYLATION FACTOR-LIKE 13B, ARL2-LIKE PROTEIN 1)	608922	JOUBERT SYNDROME, TYPE 8	612291		Sequencing	1300
Molecular Tests	ARL6 (ADP-RIBOSYLATION FACTOR-LIKE 6; BBS3 GENE)	608845	BARDET-BIEDL SYNDROME, BBS	209900		Sequencing	600
Molecular Tests	ARL6 (ADP-RIBOSYLATION FACTOR-LIKE 6; BBS3 GENE)	608845	RETINITIS PIGMENTOSA, TYPE 55, RP55	613575		Sequencing	600
Molecular Tests	ARSA (ARYLSULFATASE A, CEREBROSIDE-SULFATASE)	607574	METACHROMATIC LEUKODYSTROPHY » SULFATIDE LIPIDOSIS » ARYLSULFATASE A DEFICIENCY	250100		Sequencing	850
Molecular Tests	ARSA (ARYLSULFATASE A, CEREBROSIDE-SULFATASE)	607574	PSEUDOARYLSULFATASE A DEFICIENCY	250100		Sequencing	850
Molecular Tests	ARSA (ARYLSULFATASE A, CEREBROSIDE-SULFATASE)	607574	METACHROMATIC LEUKODYSTROPHY » SULFATIDE LIPIDOSIS » ARYLSULFATASE A DEFICIENCY	250100		Deletion-Duplication Testing	600
Molecular Tests	ARSA (ARYLSULFATASE A, CEREBROSIDE-SULFATASE)	607574	PSEUDOARYLSULFATASE A DEFICIENCY	250100		Deletion-Duplication Testing	600
Molecular Tests	ARSB (ARYLSULFATASE B, N-ACETYLGALACTOSAMINE-4-SULFATASE)	253200	MUCOPOLYSACCHARIDOSIS TYPE 6, MPS6 » MAROTEAUX-LAMY SYNDROME » ARYLSULFATASE B DEFICIENCY » N-ACETYLGALACTOSAMINE-4-SULFATASE DEFICIENCY	253200		Sequencing	850
Molecular Tests	ARSE (ARYLSULFATASE E)	300180	CHONDRODYSPLASIA PUNCTATA (X-LINKED RECESSIVE) » CHONDRODYSPLASIA PUNCTATA, BRACHYTELEPHALANGIC	302950		Sequencing	745
Molecular Tests	ARX	300382	LISSENCEPHALY WITH AMBIGUOUS GENITALIA (X-LINKED)	300215		Sequencing	450
Molecular Tests	ARX	300382	MENTAL RETARDATION, NONSPECIFIC (X-LINKED), TYPE54, MRX54	300412		Sequencing	450
Molecular Tests	ARX	300382	MYOCLONIC EPILEPSY WITH MENTAL RETARDATION AND SPASTICITY (X-LINKED)	300432		Sequencing	450
Molecular Tests	ARX	300382	INFANTILE SPASMS (X-LINKED), ISS X » WEST SYNDROME » EPILEPSY, INFANTILE SPASMS (X-LINKED)	308350		Sequencing	450
Molecular Tests	ARX	300382	PARTINGTON SYNDROME, MRXS1 » MENTAL RETARDATION, PARTINGTON SYNDROME, MRXS1	309510		Sequencing	450
Molecular Tests	ASCC3L1 (SMALL NUCLEAR RIBONUCLEOPROTEIN, 200-KD; SNRNP200; ACTIVATING SIGNAL COINTEGRATOR I COMPLEX SUBUNIT 3-LIKE 1; ASCC3L1; U5 snRNP-SPECIFIC PROTEIN, 200-KD)	601664	RETINITIS PIGMENTOSA, TYPE 33, RP33	610359		Sequencing	1060
Molecular Tests	ASL (ARGININOSUCCINATE LYASE, ARGININOSUCCINASE)	608310	ARGININOSUCCINIC ACIDURIA » ARGININOSUCCINASE DEFICIENCY » ARGININOSUCCINATE LYASE DEFICIENCY » ASL DEFICIENCY	207900		Sequencing	1390

Molecular Tests	ASPA (ASPARTOACYLASE)	608034	CANAVAN DISEASE » CANAVAN-VAN BOGAERT-BERTRAND DISEASE » ASPARTOACYLASE DEFICIENCY » SPONGIFORM ENCEPHALOPATHY, CANAVAN-VAN BOGAERT-BERTRAND TYPE	271900		Sequencing and Deletions-Duplications	930
Molecular Tests	ASPM (ABNORMAL SPINDLE-LIKE MICROCEPHALY-ASSOCIATED) MCPH5	605481	MICROCEPHALY, PRIMARY, TYPE 5 (AUTOSOMAL RECESSIVE) MCPH5	608716		Sequencing	2150
Molecular Tests	ASS (ARGININOSUCCINATE SYNTHETASE)	603470	CITRULLINEMIA, CLASSIC » CITRULLINEMIA, TYPE I » CITRULLINURIA » ARGININOSUCCINATE SYNTHETASE DEFICIENCY	215700		Sequencing	900
Molecular Tests	AT3 (SERPINC1)	107300	ANTITHROMBIN 3 DEFICIENCY » THROMBOPHILIA, HEREDITARY, DUE TO DEFICIENCY OF AT3	107300		Sequencing	460
Molecular Tests	AT3 (SERPINC1)	107300	ANTITHROMBIN 1 DEFICIENCY » THROMBOPHILIA, HEREDITARY, DUE TO DEFICIENCY OF AT3	107300		Deletion-Duplication Testing	Upon Request
Molecular Tests	ATM	607585	ATAXIA-TELANGIECTASIA, AT » LOUIS-BAR SYNDROME	208900		Sequencing	1620
Molecular Tests	ATP1A2	182340	FAMILIAL PARAPLEGIC MIGRAINE TYPE 2	602481		2 Mutations: L764P and W887R	300
Molecular Tests	ATP1A3	182350	FAMILIAL PARAPLEGIC MIGRAINE TYPE 3	602481		Sequencing	3100
Molecular Tests	ATP1A3 (ATPase, Na+/K+ TRANSPORTING, ALPHA-3 POLYPEPTIDE; SODIUM-POTASSIUM ATPase)	182350	DYSTONIA, TYPE 12, DYT12	128235		Sequencing	1700
Molecular Tests	ATP1A3 (ATPase, Na+/K+ TRANSPORTING, ALPHA-3 POLYPEPTIDE; SODIUM-POTASSIUM ATPase)	182350	DYSTONIA, TYPE 12, DYT12	128235		6 Exons	600
Molecular Tests	ATP2A2 (ATP2B, SERCA2)	108740	ACROKERATOSIS VERRUCIFORMIS » HAILEY-HAILEY	101900		Sequencing	2500
Molecular Tests	ATP2A2 (ATP2B, SERCA2)	108740	DARIER-WHITE DISEASE » KERATOSIS FOLLICULARIS » DARIER DISEASE	124200		Sequencing	2500
Molecular Tests	ATP2C1	604584	HAILEY-HAILEY DISEASE » BENIGNIC FAMILIAL PEMPHIGUS	169600		Sequencing	2830
Molecular Tests	ATPSF (ATP SYNTHASE, H+ TRANSPORTING, MITOCHONDRIAL F1 COMPLEX, F(O) SUBUNIT)	606153	MITOCHONDRIAL COMPLEX V (ATP SYNTHASE) DEFICIENCY, NUCLEAR TYPE 3	614053		Sequencing	690
Molecular Tests	ATP6V0A2 (ATPase, H+ TRANSPORTING, LYSOSOMAL, V0 SUBUNIT A2)	611716	CUTIS LAXA, TYPE 2A (AUTOSOMAL RECESSIVE) » CUTIS LAXA, DEBRE TYPE	219200		Sequencing	600
Molecular Tests	ATP6V0A2 (ATPase, H+ TRANSPORTING, LYSOSOMAL, V0 SUBUNIT A2)	611716	WORMIAN SKULL SYNDROME	278250		Sequencing	600
Molecular Tests	ATP6V0A4 (ATPase, H+ TRANSPORTING, LYSOSOMAL, V0 SUBUNIT A4)	605239	RENAL TUBULAR ACIDOSIS, DISTAL (AUTOSOMAL RECESSIVE) » RENAL TUBULAR ACIDOSIS (AUTOSOMAL RECESSIVE) WITH PRESERVED HEARING	602722		Sequencing	1140
Molecular Tests	ATP6V1B1 (ATP6B1 VACUOLAR ATP SYNTHASE SUBUNIT B, KIDNEY)	192132	RENAL TUBULAR ACIDOSIS, DISTAL, WITH PROGRESSIVE NERVE DEAFNESS	267300		Sequencing	800
Molecular Tests	ATP7A	300011	NEONATAL CUTIS LAXA » OCCIPITAL HORN SYNDROME	304150		Sequencing	600
Molecular Tests	ATP7A	300011	NEONATAL CUTIS LAXA » OCCIPITAL HORN SYNDROME	304150		Deletion-Duplication Testing	580
Molecular Tests	ATP7A	300011	MENKES DISEASE » KINKY HAIR DISEASE	309400		Sequencing	600
Molecular Tests	ATP7A	300011	MENKES DISEASE » KINKY HAIR DISEASE	309400		Deletion-Duplication Testing	580
Molecular Tests	ATP7B	606882	WILSON DISEASE » HEPATOLENTICULAR DEGENERATION	277900		Sequencing	1200
Molecular Tests	ATP7B	606882	WILSON DISEASE » HEPATOLENTICULAR DEGENERATION	277900		Deletion-Duplication Testing	300
Molecular Tests	ATP8B1 (FIC1)	602397	CHOLESTASIS, PROGRESSIVE FAMILIAL INTRAHEPATIC 1, PFIC1 » BYLER DISEASE	211600		Sequencing	1900
Molecular Tests	ATP8B1 (FIC1)	602397	CHOLESTASIS, BENIGN RECURRENT INTRAHEPATIC, BRIC » SUMMERSKILL SYNDROME	243300		Sequencing	1900
Molecular Tests	ATPAF2 (ATP SYNTHASE, MITOCHONDRIAL F1 COMPLEX, ASSEMBLY FACTOR 2; ATP12, S. CEREVISIAE, HOMOLOG OF; ATP12 S	608918	MITOCHONDRIAL COMPLEX 5 (ATP SYNTHASE) DEFICIENCY, NUCLEAR TYPE 1			Sequencing	970
Molecular Tests	ATRX (XNP)	300032	ALPHA-THALASSEMIA / MENTAL RETARDATION SYNDROME, ATRX (X-LINKED) » ATR-X SYNDROME » XL-MR-HYPOTONIC FACE SYNDROME » MENTAL RETARDATION, X-LINKED, HYPOTONIC FACE SYNDROME	301040	Blood in RNA PAX tubes	Sequencing	1540

Molecular Tests	ATRX (XNP)	300032	SMITH-FINEMAN-MYERS MENTAL RETARDATION SYNDROME » MENTAL RETARDATION, SMITH-FINEMAN-MYERS SYNDROME	309580	Blood in RNA PAX tubes	Sequencing	1540
Molecular Tests	ATRX (XNP)	300032	JUBERG-MARSIDI SYNDROME » MENTAL RETARDATION WITH GROWTH RETARDATION, DEAFNESS, AND MICROGENITALISM (X-LINKED)	309590	Blood in RNA PAX tubes	Sequencing	1540
Molecular Tests	ATXN1 (ATAXIN 1)	601556	SPINOCEREBELLAR ATAXIA 1, SCA1	164400	See SCA1, SCA2, SCA3, SCA6	Sequencing	
Molecular Tests	ATXN10 (ATAXIN 10)	603516	SPINOCEREBELLAR ATAXIA 10, SCA10	603516	See SCA8, SCA10, SCA12, SCA17	Repeat	250
Molecular Tests	ATXN2 (ATAXIN 2)	601517	SPINOCEREBELLAR ATAXIA 2, SCA2	183090	See SCA1, SCA2, SCA3, SCA6	Sequencing	
Molecular Tests	ATXN3 (ATAXIN 3)	607047	SPINOCEREBELLAR ATAXIA 3, SCA3 » MACHADO-JOSEPH DISEASE	109150	See SCA1, SCA2, SCA3, SCA6	Sequencing	
Molecular Tests	ATXN8OS (ATAXIN 8 OPPOSITE STRAND, SCA8)	603680	SPINOCEREBELLAR ATAXIA 8, SCA8	608768	See SCA8, SCA10, SCA12, SCA17	Repeat	250
Molecular Tests	AUH (AU-SPECIFIC RNA-BINDING PROTEIN, 3-ALPHA-METHYLGLUTACONYL -CoA HYDRATASE)	600529	3-ALPHA-METHYLGLUTACONICACIDURIA, TYPE 1 » 3-ALPHA-METHYLGLUTACONYL-CoA HYDRATASE DEFICIENCY	250950		Sequencing	790
Molecular Tests	AVP (ARGININE VASOPRESSIN, VASOPRESSIN-NEUROPHYSIN 2, ANTIDIURETIC HORMONE, ADH)	192340	DIABETES INSIPIDUS, NEUROHYPOPHYSEAL » DIABETES INSIPIDUS, PRIMARY CENTRAL	125700		Sequencing	250
Molecular Tests	AVPR2 (VASOPRESSIN RECEPTOR 2, ANTIDIURETIC HORMONE RECEPTOR)	304800	DIABETES INSIPIDUS, NEPHROGENIC (X-LINKED)	304800		Sequencing	350
Molecular Tests	AVPR2 (VASOPRESSIN RECEPTOR 2, ANTIDIURETIC HORMONE RECEPTOR)	304800	DIABETES INSIPIDUS, NEPHROGENIC (X-LINKED)	304800		Deletion-Duplication Testing	680
Molecular Tests	AXIN2 (AXIS INHIBITOR 2, CONDUCTIN)	604433	COLORECTAL CANCER, SOMATIC	114500		Sequencing	Upon Request
Molecular Tests	AXIN2 (AXIS INHIBITOR 2, CONDUCTIN)	604433	OLIGODONTIA-COLORECTAL CANCER SYNDROME » TOOTH AGENESIS-COLORECTAL CANCER SYNDROME	608615		Sequencing	Upon Request
Molecular Tests	AZFa, AZFb and AZFc (including DAZ)	415000	AZOSPERMIA-OLIGOSPERMIA » SERTOLI-CELL-ONLY SYNDROME	415000		Deletions	250
Molecular Tests	B3GALTL (UDP-GAL:BETA-GlcNAc BETA-1,3-GALACTOSYLTRANSFERASE-LIKE, BETA-1,3-GLUCOSYLTRANSFERASE)	610308	PETERS-PLUS SYNDROME » KRAUSE-KIVLIN SYNDROME	261540		Sequencing	1200
Molecular Tests	B4GALT1 (UDP-GAL:BETA-GlcNAc BETA-1,4-GALACTOSYLTRANSFERASE, POLYPEPTIDE 1; BETA-1,4-GALACTOSYLTRANSFERASE 1)	137060	CONGENITAL DISORDER OF GLYCOSYLATION, TYPE 2D, CDG2D	607091		Sequencing	890
Molecular Tests	BANF1 (BARRIER-TO-AUTOINTEGRATION FACTOR 1)	603811	NESTOR-GUILLERMO PROGERIA SYNDROME » PROGERIA SYNDROME, CHILDHOOD-ONSET, WITH OSTEOLYSIS	614008		Sequencing	250
Molecular Tests	BBS1	209901	BARDET-BIEDL SYNDROME TYPE 1, BBS1	209900		Sequencing	1100
Molecular Tests	BBS10	610148	BARDET-BIEDL SYNDROME TYPE 10, BBS10	209900		Sequencing	400
Molecular Tests	BBS12	610683	BARDET-BIEDL SYNDROME TYPE 12, BBS12	209900		Sequencing	350
Molecular Tests	BBS2	606151	BARDET-BIEDL SYNDROME TYPE 2, BBS2	209900		Sequencing	900
Molecular Tests	BBS6 (MKKS, MKS)	604896	BARDET-BIEDL SYNDROME TYPE 6, BBS6	209900		Sequencing	700
Molecular Tests	BBS6 (MKKS, MKS)	604896	MCKUSICK-KAUFMAN SYNDROME » HYDROMETROCOLPOS SYNDROME » HYDROMETROCOLPOS, POSTAXIAL POLYDACTYLY, AND CONGENITAL HEART MALFORMATION	236700		Sequencing	700
Molecular Tests	BCKDHA (BRANCHED-CHAIN KETO ACID DEHYDROGENASE E1, ALPHA POLYPEPTIDE)	608348	MAPLE SYRUP URINE DISEASE » BRANCHED-CHAIN KETOACIDURIA » BRANCHED-CHAIN ALPHA-KETO ACID DEHYDROGENASE DEFICIENCY » KETO ACID DECARBOXYLASE DEFICIENCY	248600		Sequencing	960
Molecular Tests	BCKDHA, BCKDHB, DBT		MAPLE SYRUP URINE DISEASE » BRANCHED-CHAIN KETOACIDURIA » BRANCHED-CHAIN ALPHA-KETO ACID DEHYDROGENASE DEFICIENCY » KETO ACID DECARBOXYLASE DEFICIENCY	248600		Sequencing	2330
Molecular Tests	BCKDHB (BRANCHED-CHAIN KETO ACID DEHYDROGENASE E1, BETA POLYPEPTIDE)	248611	MAPLE SYRUP URINE DISEASE » BRANCHED-CHAIN KETOACIDURIA » BRANCHED-CHAIN ALPHA-KETO ACID DEHYDROGENASE DEFICIENCY » KETO ACID DECARBOXYLASE DEFICIENCY	248600		Sequencing	1060
Molecular Tests	BCS1L (BCS1, S. CEREVISIAE, HOMOLOG-LIKE)	603647	CYTOCHROME c OXIDASE DEFICIENCY » COX DEFICIENCY » COMPLEX 3, MITOCHONDRIAL RESPIRATORY CHAIN, DEFICIENCY OF	124000		Sequencing	760
Molecular Tests	BCS1L (BCS1, S. CEREVISIAE, HOMOLOG-LIKE)	603647	LEIGH SYNDROME	256000		Sequencing	760

Molecular Tests	BCS1L (BCS1, S. CEREVISIAE, HOMOLOG-LIKE)	606647	GRACILE SYNDROME » GROWTH RETARDATION, AMINO ACIDURIA, CHOLESTASIS, IRON OVERLOAD, LACTIC ACIDOSIS, AND EARLY DEATH » FINNISH LETHAL NEONATAL METABOLIC SYNDROME » LACTIC ACIDOSIS, FINNISH, WITH HEPATIC HEMOSIDEROSIS	603358		Sequencing	760
Molecular Tests	BEST1 (VMD2, VITELLIFORM MACULAR DYSTROPHY GENE 2; BESTROPHIN)	607854	MACULAR DYSTROPHY, VITELLIFORM, VMD » MYOPATHY, CENTRONUCLEAR (AUTOSOMAL RECESSIVE)	153700		Sequencing	580
Molecular Tests	BEST1 (VMD2, VITELLIFORM MACULAR DYSTROPHY GENE 2; BESTROPHIN)	607854	MACULAR DYSTROPHY, CONCENTRIC, ANNULAR » BEST-LIKE MACULAR DYSTROPHY	153700		Sequencing	580
Molecular Tests	BEST1 (VMD2, VITELLIFORM MACULAR DYSTROPHY GENE 2; BESTROPHIN)	607854	VITELLIFORM MACULAR DYSTROPHY, ADULT-ONSET » FOVEOMACULAR VITREOPATHY, ADULT-ONSET, WITH CHOROIDAL NEOVASCULARIZATION	608161		Sequencing	580
Molecular Tests	BIN1 (BRIDGING INTEGRATOR 1; AMPHIPHYSIN 2; BOX-DEPENDENT MYC-INTERACTING PROTEIN 1; AMPHIPHYSIN-LIKE, FORMERLY)	601248	MYOPATHY, CENTRONUCLEAR, TYPE 2 » MYOPATHY, CENTRONUCLEAR (AUTOSOMAL RECESSIVE) » MYOPATHY, CENTRONUCLEAR (AUTOSOMAL DOMINANT)	255200		Sequencing	1990
Molecular Tests	BIRC4 (BACULOVIRAL IAP REPEAT-CONTAINING PROTEIN 4; APOPTOSIS INHIBITOR 3; API3 INHIBITOR OF APOPTOSIS, X-LINKED; XIAP)	300079	LYMPHOPROLIFERATIVE SYNDROME, TYPE 2 (X-LINKED), XLP2 » XIAP DEFICIENCY	300635		Sequencing	1430
Molecular Tests	BLOC1S3 (BIOGENESIS OF LYSOSOME-RELATED ORGANELLES COMPLEX 1, SUBUNIT 3; BLOS3; HPS8; REDUCED PIGMENTATION, MOUSE, HOMOLOG OF)	609762	HERMANSKY-PUDLAK SYNDROME, TYPE 8; HPS8	614077		Sequencing	440
Molecular Tests	BMPR1A (BONE MORPHOGENETIC PROTEIN RECEPTOR, TYPE 1A; ACTIVIN A RECEPTOR, TYPE II-LIKE KINASE 3; ACVRL6)	601299	COWDEN DISEASE » LHERMITTE-DUCLOS DISEASE	158350		Sequencing	720
Molecular Tests	BMPR1A (BONE MORPHOGENETIC PROTEIN RECEPTOR, TYPE 1A; ACTIVIN A RECEPTOR, TYPE II-LIKE KINASE 3;	601299	JUVENILE POLYPOSIS SYNDROME	174900		Sequencing	720
Molecular Tests	BMPR1A (BONE MORPHOGENETIC PROTEIN RECEPTOR, TYPE 1A; ACTIVIN A RECEPTOR, TYPE II-LIKE KINASE 3;	601299	POLYPOSIS SYNDROME, HEREDITARY MIXED, TYPE 2	610069		Sequencing	720
Molecular Tests	BMPR1B (BONE MORPHOGENETIC PROTEIN RECEPTOR, TYPE IB; ACTIVIN RECEPTOR-LIKE KINASE 4)	603248	BRACHYDACTYLY TYPE A2, BDA2 » BRACHYMESOPHALANGY 2 » SYNDR., BRIEST TYPE BRACHYDACTYLY	112600		Sequencing	450
Molecular Tests	BMPR2	600799	PRIMARY PULMONARY HYPERTENSION, PPH1	178600		Sequencing	1025
Molecular Tests	BRAF (V-RAF MURINE SARCOMA VIRAL ONCOGENE HOMOLOG B1, RAFB1)	164757	LYMPHOMA, NON-HODGKIN			Sequencing	710
Molecular Tests	BRAF (V-RAF MURINE SARCOMA VIRAL ONCOGENE HOMOLOG B1, RAFB1)	164757	MENINGIOMA, CELLULAR, CANCER, SOMATIC			Sequencing	710
Molecular Tests	BRAF (V-RAF MURINE SARCOMA VIRAL ONCOGENE HOMOLOG B1, RAFB1)	164757	ADENOCARCINOMA OF LUNG, SOMATIC			Sequencing	710
Molecular Tests	BRAF (V-RAF MURINE SARCOMA VIRAL ONCOGENE HOMOLOG B1, RAFB1)	164757	THYROTROPIC ADENOMA, SOMATIC			Sequencing	710
Molecular Tests	BRAF (V-RAF MURINE SARCOMA VIRAL ONCOGENE HOMOLOG B1, RAFB1)	164757	MELANOMA, MALIGNANT, SOMATIC			Sequencing	710
Molecular Tests	BRAF (V-RAF MURINE SARCOMA VIRAL ONCOGENE HOMOLOG B1, RAFB1)	164757	COLORECTAL CANCER, SOMATIC	114500		Sequencing	710
Molecular Tests	BRAF (V-RAF MURINE SARCOMA VIRAL ONCOGENE HOMOLOG B1, RAFB1)	164757	CARDIOFACIOCUTANEOUS SYNDROME, CFC	115150		Sequencing	710
Molecular Tests	BRCA1 and BRCA2		BREAST AND OVARIAN CANCER			Sequencing and Deletion-Duplication Testing	900
Molecular Tests	BRCA1 and BRCA2		BREAST AND OVARIAN CANCER			Deletion-Duplication Testing	570
Molecular Tests	BRCA1 and BRCA2		PROSTATE CANCER	176807		Sequencing and Deletion-Duplication Testing	900
Molecular Tests	BRCA1 and BRCA2		PROSTATE CANCER	176807		Deletion-Duplication Testing	570
Molecular Tests	BSCL2 (SEIPIN)	606158	FAMILIAL SPASTIC PARAPLEGIA 17, SPG17 » SPASTIC PARAPLEGIA WITH AMYOTROPHY OF HANDS AND FEET » SILVER SYNDROME » SILVER SPASTIC PARAPLEGIA SYNDROME	270685		Sequencing	740
Molecular Tests	BSND (BARTTIN)	606412	BARTTER SYNDROME, TYPE 4 » BARTTER SYNDROME, INFANTILE, WITH SENSORINEURAL DEAFNESS	602522		Sequencing	500
Molecular Tests	BTD (BIOTINIDASE)	609019	BIOTINIDASE DEFICIENCY » MULTIPLE CARBOXYLASE DEFICIENCY, LATE-ONSET » MULTIPLE CARBOXYLASE DEFICIENCY, JUVENILE-ONSET	253260		Sequencing	510
Molecular Tests	BTD (BIOTINIDASE)	609019	BIOTINIDASE DEFICIENCY » MULTIPLE CARBOXYLASE DEFICIENCY, LATE-ONSET » MULTIPLE CARBOXYLASE DEFICIENCY, JUVENILE-ONSET » PROFOUND	253260		8 Mutations: D444H, A171T, F403V, G98_7-BP DEL/5-BP INS, Q456H, R157H, R538C, D252G, Detecting 60%	320
Molecular Tests	BTK (BRUTON TYROSINE KINASE, ATK, BPK)	300300	AGAMMAGLOBULINEMIA (X-LINKED), XLA » BRUTON AGAMMAGLOBULINEMIA » HYPOGAMMAGLOBULINEMIA, X-LINKED	300755		Sequencing	750

Molecular Tests	C10ORF2 (CHROMOSOME 10 OPEN READING FRAME 2, T7 GENE 4-LIKE PROTEIN WITH INTRAMITOCHONDRIAL NUCLEOID LOCALIZATION, TWINKLE)	606075	PROGRESSIVE EXTERNAL OPHTHALMOPLEGIA WITH MITOCHONDRIAL DNA DELETIONS, DIGENIC	157640		Sequencing	880
Molecular Tests	C10ORF2 (CHROMOSOME 10 OPEN READING FRAME 2, T7 GENE 4-LIKE PROTEIN WITH INTRAMITOCHONDRIAL NUCLEOID LOCALIZATION, TWINKLE)	606075	SENSORY ATAXIC NEUROPATHY, DYSARTHRIA, AND OPHTHALMOPARESIS, SANDO » SPINOCEREBELLAR ATAXIA WITH EPILEPSY	607459		Sequencing	880
Molecular Tests	C10ORF2 (CHROMOSOME 10 OPEN READING FRAME 2, T7 GENE 4-LIKE PROTEIN WITH INTRAMITOCHONDRIAL NUCLEOID LOCALIZATION, TWINKLE)	606075	PROGRESSIVE EXTERNAL OPHTHALMOPLEGIA WITH MITOCHONDRIAL DNA DELETIONS (AUTOSOMAL DOMINANT), TYPE 3	609286		Sequencing	880
Molecular Tests	C1NH (C1 ESTERASE INHIBITOR, SERPING 1)	606860	ANGIOEDEMA » ANGIONEUROTIC EDEMA » QUINCKE EDEMA	106100		Sequencing and Deletion-Duplication Testing	1590
Molecular Tests	C20ORF7 (CHROMOSOME 20 OPEN READING FRAME 7; C20ORF7 ; NDUFAF5)	612360	CYTOCHROME c OXIDASE DEFICIENCY » COX DEFICIENCY » COMPLEX 1, MITOCHONDRIAL RESPIRATORY CHAIN, DEFICIENCY OF	252010		Sequencing	1280
Molecular Tests	C2ORF71 (CHROMOSOME 2 OPEN READING FRAME 71)	613425	RETINITIS PIGMENTOSA, TYPE 54, RP54	613428		Sequencing	680
Molecular Tests	C3 (COMPLEMENT COMPONENT 3)	120700	COMPLEMENT COMPONENT 3 DEFICIENCY (AUTOSOMAL RECESSIVE) » C3 DEFICIENCY (AUTOSOMAL RECESSIVE)	120700		Sequencing	1300
Molecular Tests	C3 (COMPLEMENT COMPONENT 3)	120700	MACULAR DEGENERATION, AGE-RELATED, TYPE 9, SUSCEPTIBILITY TO	611378		Sequencing	1300
Molecular Tests	C3 (COMPLEMENT COMPONENT 3)	120700	HEMOLYTIC UREMIC SYNDROME, ATYPICAL, SUSCEPTIBILITY TO, TYPE 5	612925		Sequencing	1300
Molecular Tests	C7ORF10 (CHROMOSOME 7 OPEN READING FRAME 10)	609187	GLUTARIC ACIDURIA, TYPE 3 » GLUTARYL-CoA OXIDASE DEFICIENCY	231690		Sequencing	1090
Molecular Tests	C9ORF72 (CHROMOSOME 9 OPEN READING FRAME 72)	614260	FRONTOTEMPORAL DEMENTIA AND/OR AMYOTROPHIC LATERAL SCLEROSIS » FRONTOTEMPORAL DEMENTIA AND/OR MOTOR NEURON DISEASE	105550		Hexanucleotide Repeat Expansion	400
Molecular Tests	CA2 (CARBONIC ANHYDRASE 2)	259730	OSTEOPETROSIS WITH RENAL TUBULAR ACIDOSIS » GUIBAUD - VAINSEL SYNDROME » CARBONIC ANHYDRASE 2 DEFICIENCY » MARBLE BRAIN DISEASE	259730		Sequencing	500
Molecular Tests	CA4 (CARBONIC ANHYDRASE 4)	114760	RETINITIS PIGMENTOSA, TYPE 17, RP17	600852		Sequencing	530
Molecular Tests	CABP4 (CALCIUM-BINDING PROTEIN 4)	608965	NIGHT BLINDNESS, CONGENITAL STATIONARY, TYPE 2B, CSNB2B » NIGHT BLINDNESS, CONGENITAL STATIONARY, INCOMPLETE (AUTOSOMAL RECESSIVE)	610427		Sequencing	530
Molecular Tests	CACNA1A (CALCIUM CHANNEL, VOLTAGE-DEPENDENT, P/Q TYPE, ALPHA-1A SUBUNIT, CALCIUM CHANNEL, L TYPE, ALPHA-1 POLYPEPTIDE, ISOFORM 4, CACNL1A4)	601011	EPISODIC ATAXIA, TYPE 2, EA2 » ATAXIA, EPISODIC, WITH NYSTAGMUS » EPISODIC ATAXIA, NYSTAGMUS-ASSOCIATED » CEREBELLOPATHY, HEREDITARY PAROXYSMAL ATAXIA » FAMILIAL PAROXYSMAL ACETAZOLAMIDE-RESPONSIVE » HEREDITARY PAROXYSMAL CEREBELLAR ATAXIA	108500		Sequencing	1300
Molecular Tests	CACNA1A (CALCIUM CHANNEL, VOLTAGE-DEPENDENT, P/Q TYPE, ALPHA-1A SUBUNIT, CALCIUM CHANNEL, L TYPE, ALPHA-1 POLYPEPTIDE, ISOFORM 4, CACNL1A4)	601011	EPISODIC ATAXIA, TYPE 2, EA2 » ATAXIA, EPISODIC, WITH NYSTAGMUS » EPISODIC ATAXIA, NYSTAGMUS-ASSOCIATED » CEREBELLOPATHY, HEREDITARY PAROXYSMAL ATAXIA » FAMILIAL PAROXYSMAL ACETAZOLAMIDE-RESPONSIVE » HEREDITARY PAROXYSMAL CEREBELLAR ATAXIA	108500		Deletion-Duplication Testing	850
Molecular Tests	CACNA1A (CALCIUM CHANNEL, VOLTAGE-DEPENDENT, P/Q TYPE, ALPHA-1A SUBUNIT, CALCIUM CHANNEL, L TYPE, ALPHA-1 POLYPEPTIDE, ISOFORM 4, CACNL1A4)	601011	MIGRAINE, FAMILIAL HEMIPLEGIC, 1 » MIGRAINE, SPORADIC HEMIPLEGIC	141500		Sequencing	1300
Molecular Tests	CACNA1A (CALCIUM CHANNEL, VOLTAGE-DEPENDENT, P/Q TYPE, ALPHA-1A SUBUNIT, CALCIUM CHANNEL, L TYPE, ALPHA-1 POLYPEPTIDE, ISOFORM 4, CACNL1A4)	601011	MIGRAINE, FAMILIAL HEMIPLEGIC, 1 » MIGRAINE, SPORADIC HEMIPLEGIC	141500		Deletion-Duplication Testing	850
Molecular Tests	CACNA1A (CALCIUM CHANNEL, VOLTAGE-DEPENDENT, P/Q TYPE, ALPHA-1A SUBUNIT, CALCIUM CHANNEL, L TYPE, ALPHA-1 POLYPEPTIDE, ISOFORM 4, CACNL1A4)	601011	SPINOCEREBELLAR ATAXIA 6, SCA6	183086	See SCA1, SCA2, SCA3, SCA6	Repeat	500
Molecular Tests	CACNA1C (CALCIUM CHANNEL, VOLTAGE-DEPENDENT, L TYPE, ALPHA-1C SUBUNIT; CACNL1A1; CCHL1A1; CaV1.2)	114205	TIMOTHY SYNDROME » LONG QT SYNDROME WITH SYNDACTYLY	601005		Exons 8 and 8a	390
Molecular Tests	CACNA1C (CALCIUM CHANNEL, VOLTAGE-DEPENDENT, L TYPE, ALPHA-1C SUBUNIT; CACNL1A1; CCHL1A1; CaV1.2)	114205	BRUGADA SYNDROME, TYPE 3	611875		Exons 8 and 8a	390
Molecular Tests	CACNA1F (CALCIUM CHANNEL, VOLTAGE-DEPENDENT, ALPHA-1F SUBUNIT)	300110	NIGHT BLINDNESS, CONGENITAL STATIONARY, TYPE 2A, CSNB2A » CSNB, INCOMPLETE (X-LINKED)	300071		Sequencing	1060
Molecular Tests	CACNA1F (CALCIUM CHANNEL, VOLTAGE-DEPENDENT, ALPHA-1F SUBUNIT)	300110	CONE-ROD DYSTROPHY (X-LINKED), TYPE 3, CORDX3	300476		Sequencing	1060

Molecular Tests	CACNA1F (CALCIUM CHANNEL, VOLTAGE-DEPENDENT, ALPHA-1F SUBUNIT)	300110	ALAND ISLAND EYE DISEASE » FORSIUS-ERIKSSON TYPE OCULAR ALBINISM	300600		Sequencing	1060
Molecular Tests	CACNA1S (CACNL1A3)	114208	HYPOKALEMIC PERIODIC PARALYSIS, HOKPP » HYPERKALEMIC PERIODIC PARALYSIS	170400		Exons 11 and 30, including the R528H, R1239H and R1239G Mutations	900
Molecular Tests	CACNA1S (CACNL1A3)	114208	THYROTOXIC PERIODIC PARALYSIS » THYROTOXIC PERIODIC PARALYSIS » HASHITOXIC PERIODIC PARALYSIS	188580		Exons 11 and 30, including the R528H, R1239H and R1239G Mutations	900
Molecular Tests	CACNA1S (CACNL1A3)	114208	MALIGNANT HYPERTHERMIA SUSCEPTIBILITY 5, MHS5	601887		Exons 11 and 30, including the R528H, R1239H and R1239G Mutations	900
Molecular Tests	CACNA2D4 (CALCIUM CHANNEL, VOLTAGE-DEPENDENT, ALPHA-2/DELTA SUBUNIT 4)	608171	RETINAL CONE DYSTROPHY, TYPE 4, RCD4	610478		Sequencing	960
Molecular Tests	CACNB4 (CALCIUM CHANNEL, VOLTAGE-DEPENDENT, BETA-4 SUBUNIT)	601949	EPILEPSY, IDIOPATHIC GENERALIZED » EPISODIC ATAXIA, TYPE 5, EA5	600669		Sequencing	1000
Molecular Tests	CACNB4 (CALCIUM CHANNEL, VOLTAGE-DEPENDENT, BETA-4 SUBUNIT)	601949	EPILEPSY, JUVENILE MYOCLONIC, JME » JANZ SYNDROME	606904		Sequencing	1000
Molecular Tests	CAPN3 (CALPAIN 3)	114240	MUSCULAR DYSTROPHY, LIMB GIRDLE, TYPE 2A, LGMD2A	253600		Sequencing	950
Molecular Tests	CASK (CALCIUM/CALMODULIN-DEPENDENT SERINE PROTEIN KINASE; VERTEBRATE LIN2 HOMOLOG; CAMGUK, DROSOPHILA, HOMOLOG OF)	300172	FG SYNDROME, TYPE 4			Sequencing	2140
Molecular Tests	CASK (CALCIUM/CALMODULIN-DEPENDENT SERINE PROTEIN KINASE; VERTEBRATE LIN2 HOMOLOG; CAMGUK, DROSOPHILA, HOMOLOG OF)	300172	MENTAL RETARDATION AND MICROCEPHALY WITH PONTINE AND CEREBELLAR HYPOPLASIA » MICPCH SYNDROME	300749		Sequencing	2140
Molecular Tests	CASP10 (CASPASE 10, MCH4, CASP10B, FLICE2)	601762	AUTOIMMUNE LYMPHOPROLIFERATIVE SYNDROME, TYPE 2, ALPS2	603909		Sequencing	890
Molecular Tests	CASP8 (CASPASE 8, FLICE, MCH5)	601763	AUTOIMMUNE LYMPHOPROLIFERATIVE SYNDROME, TYPE 1, ALPS, ALPS1A, ALPS1B	601859		Sequencing	1290
Molecular Tests	CASP8 (CASPASE 8, FLICE, MCH5)	601763	CASPASE 8 DEFICIENCY	607271		Sequencing	1290
Molecular Tests	CASQ2 (CALSEQUESTRIN 2)	114251	VENTRICULAR TACHYCARDIA, CATECHOLAMINERGIC POLYMORPHIC » VENTRICULAR TACHYCARDIA, STRESS-INDUCED POLYMORPHIC	604772		Sequencing	910
Molecular Tests	CASR (CALCIUM-SENSING RECEPTOR, PCAR1)	601199	HYPOCALCIURIC HYPERCALCEMIA, FAMILIAL, TYPE 1 » FAMILIAL BENIGN HYPERCALCEMIA 1	145980		Sequencing	550
Molecular Tests	CASR (CALCIUM-SENSING RECEPTOR, PCAR1)	601199	HYPOCALCIURIC HYPERCALCEMIA, FAMILIAL, TYPE 1 » FAMILIAL BENIGN HYPERCALCEMIA 1	145980		Deletion-Duplication Testing	350
Molecular Tests	CASR (CALCIUM-SENSING RECEPTOR, PCAR1)	601199	HYPOPARATHYROIDISM, FAMILIAL ISOLATED » HYPOPARATHYROIDISM (AUTOSOMAL DOMINANT) » HYPOCALCEMIA (AUTOSOMAL DOMINANT) » HYPERCALCIURIC HYPOCALCEMIA, FAMILIAL	146200		Sequencing	550
Molecular Tests	CASR (CALCIUM-SENSING RECEPTOR, PCAR1)	601199	HYPOPARATHYROIDISM, FAMILIAL ISOLATED » HYPOPARATHYROIDISM (AUTOSOMAL DOMINANT) » HYPOCALCEMIA (AUTOSOMAL DOMINANT) » HYPERCALCIURIC HYPOCALCEMIA, FAMILIAL	146200		Deletion-Duplication Testing	350
Molecular Tests	CASR (CALCIUM-SENSING RECEPTOR, PCAR1)	601199	NEONATAL SEVERE PRIMARY HYPERPARATHYROIDISM	239200		Sequencing	550
Molecular Tests	CASR (CALCIUM-SENSING RECEPTOR, PCAR1)	601199	NEONATAL SEVERE PRIMARY HYPERPARATHYROIDISM	239200		Deletion-Duplication Testing	350
Molecular Tests	CASR (CALCIUM-SENSING RECEPTOR, PCAR1)	601199	HYPOCALCEMIA (AUTOSOMAL DOMINANT)	601198		Sequencing	550
Molecular Tests	CASR (CALCIUM-SENSING RECEPTOR, PCAR1)	601199	HYPOCALCEMIA (AUTOSOMAL DOMINANT)	601198		Deletion-Duplication Testing	350
Molecular Tests	CAT (CATALASE)	115500	ACATALASEMIA » ACATALASIA » CATALASE DEFICIENCY	115500		Sequencing	1500
Molecular Tests	CAV3 (CAVEOLIN 3)	601253	RIPPLING MUSCLE DISEASE 2, RMD	606072		Sequencing	400
Molecular Tests	CAV3 (CAVEOLIN 3)	601253	MUSCULAR DYSTROPHY, LIMB-GIRDLE, TYPE 1C, LGMD1C	607801		Sequencing	400
Molecular Tests	CBS (CYSTATHIONINE BETA-SYNTHASE)	236200	HOMOCYSTINURIA » CYSTATHIONINE BETA-SYNTHASE DEFICIENCY	236200		Sequencing	1200
Molecular Tests	CBS (CYSTATHIONINE BETA-SYNTHASE)	236200	HOMOCYSTINURIA » CYSTATHIONINE BETA-SYNTHASE DEFICIENCY	236200		Exons 4 and 8 (Including GLY307SER and ILE278THR)	400
Molecular Tests	CCM2 (MALCAVERNIN)	607929	CEREBRAL CAVERNOUS MALFORMATIONS, TYPE 2 » CAVERNOUS ANGIOMA, FAMILIAL, TYPE 2 » HYPERKERATOTIC CUTANEOUS CAPILLARY-VENOUS MALFORMATIONS ASSOCIATED WITH CEREBRAL CAPILLARY MALFORMATIONS, TYPE 2	603284		Sequencing	2200
Molecular Tests	CCR5	601373	RESISTANCE TO HIV (HUMAN IMMUNODEFICIENCY VIRUS TYPE 1)	609423		1 Mutation: DEL32 BP	230
Molecular Tests	CD2AP (CD2-ASSOCIATED PROTEIN)	604241	FOCAL SEGMENTAL GLOMERULOSCLEROSIS, TYPE 3	607832		Sequencing	1100
Molecular Tests	CD40 (CD40 ANTIGEN, B CELL-ASSOCIATED MOLECULE CD40 TUMOR NECROSIS FACTOR RECEPTOR SUPERFAMILY, MEMBER 5, TNFRSF5)	109535	IMMUNODEFICIENCY WITH HYPER-IgM, TYPE 3 » HYPER-IgM SYNDROME 3	606843		Sequencing	1350

Molecular Tests	CD40LG (CD40 LIGAND; TNFSF5; TUMOR NECROSIS FACTOR LIGAND SUPERFAMILY, MEMBER 5; TRAP; GP39)	300386	IMMUNODEFICIENCY WITH HYPER-IgM, TYPE 1, HIGM1 » HYPER-IgM IMMUNODEFICIENCY (X-LINKED) » HYPER-IgM SYNDROME 1	308230		Sequencing	560
Molecular Tests	CDAN1 (CODANIN 1, DISCS LOST, DROSOPHILA, HOMOLOG OF)	607465	ANEMIA, DYSERYTHROPOIETIC CONGENITAL, TYPE 1 » DYSERYTHROPOIETIC ANEMIA, CONGENITAL, TYPE 1	224120		Sequencing	2150
Molecular Tests	CDC73 (CELL DIVISION CYCLE PROTEIN 73, S. CEREVISIAE, HOMOLOG OF; PARAFIBROMIN; HYRAX, DROSOPHILA, HOMOLOG OF; HRPT2)	145000	HYPERPARATHYROIDISM 1, HRPT1 » PARATHYROID ADENOMA, FAMILIAL	145000		Sequencing	660
Molecular Tests	CDC73 (CELL DIVISION CYCLE PROTEIN 73, S. CEREVISIAE, HOMOLOG OF; PARAFIBROMIN; HYRAX, DROSOPHILA, HOMOLOG OF; HRPT2)	145000	HYPERPARATHYROIDISM 2, HRPT2 » HYPERPARATHYROIDISM, FAMILIAL PRIMARY, WITH MULTIPLE OSSIFYING JAW FIBROMAS » HYPERPARATHYROIDISM-JAW TUMOR SYNDROME, HEREDITARY » PARATHYROID ADENOMATOSIS, FAMILIAL CYSTIC	145001		Sequencing	660
Molecular Tests	CDC73 (CELL DIVISION CYCLE PROTEIN 73, S. CEREVISIAE, HOMOLOG OF; PARAFIBROMIN; HYRAX, DROSOPHILA, HOMOLOG OF; HRPT2)	145000	PARATHYROID CARCINOMA	608266		Sequencing	660
Molecular Tests	CDH1 (CADHERIN 1, UVOMORULIN)	192090	CANCER (SOMATIC MUTATIOS)			Sequencing	1000
Molecular Tests	CDH1 (CADHERIN 1, UVOMORULIN)	192090	CANCER (SOMATIC MUTATIOS)			Deletion-Duplication Testing	600
Molecular Tests	CDH1 (CADHERIN 1, UVOMORULIN)	192090	GASTRIC CANCER, FAMILIAL DIFFUSE	137215		Sequencing	1000
Molecular Tests	CDH1 (CADHERIN 1, UVOMORULIN)	192090	GASTRIC CANCER, FAMILIAL DIFFUSE	137215		Deletion-Duplication Testing	600
Molecular Tests	CDH1 (CADHERIN 1, UVOMORULIN)	192090	CLEFT LIP WITH OR WITHOUT CLEFT PALATE, WITH GASTRIC CANCER, FAMILIAL DIFFUSE	192090		Sequencing	1000
Molecular Tests	CDH1 (CADHERIN 1, UVOMORULIN)	192090	CLEFT LIP WITH OR WITHOUT CLEFT PALATE, WITH GASTRIC CANCER, FAMILIAL DIFFUSE	192090		Deletion-Duplication Testing	600
Molecular Tests	CDH23 (CADHERIN 23; OTOCADHERIN; CADHERIN-RELATED FAMILY, MEMBER 23)	605516	USHER SYNDROME, TYPE 1D, USH1D	601067		Sequencing	1100
Molecular Tests	CDH23 (CADHERIN 23; OTOCADHERIN; CADHERIN-RELATED FAMILY, MEMBER 23)	605516	DEAFNESS, DFNB12 » DEAFNESS (AUTOSOMAL RECESSIVE) 12	601386		Sequencing	1100
Molecular Tests	CDHR1 (CADHERIN-RELATED FAMILY, MEMBER 1; PROTOCADHERIN 21; PCDH21; PHOTORECEPTOR CADHERIN; PRCAD)	609502	CONE-ROD DYSTROPHY, TYPE 15, CORD15	613660		Sequencing	960
Molecular Tests	CDK4	123829	MALIGNANT MELANOMA, CMM3	609048		Sequencing	800
Molecular Tests	CDK4	123829	MALIGNANT MELANOMA, CMM3	609048		Exon 2	310
Molecular Tests	CDKL5 (CYCLIN-DEPENDENT KINASE-LIKE 5, STK9)		ANGELMAN SYNDROME, ATYPICAL	105830		Sequencing	1400
Molecular Tests	CDKL5 (CYCLIN-DEPENDENT KINASE-LIKE 5, STK9)		INFANTILE SPASMS (X-LINKED), ISS X » WEST SYNDROME » EPILEPSY, INFANTILE SPASMS (X-LINKED)	308350		Sequencing	1400
Molecular Tests	CDKL5 (CYCLIN-DEPENDENT KINASE-LIKE 5, STK9)		RETT SYNDROME, ATYPICAL	312750		Sequencing	1400
Molecular Tests	CDKN1B (CYCLIN-DEPENDENT KINASE INHIBITOR 1B; p27-KIP1)	600778	MULTIPLE ENDOCRINE NEOPLASIA, TYPE 4, MEN4	610755		Sequencing	250
Molecular Tests	CDKN1C (CYCLIN-DEPENDENT KINASE INHIBITOR 1C, p57-KIP2)	600856	BECKWITH-WIEDEMANN SYNDROME » EXOMPHALOS-MACROGLOSSIA-GIGANTISM SYNDROME	130650		Sequencing	910
Molecular Tests	CDKN2A (CYCLIN-DEPENDENT KINASE INHIBITOR 2A; P16; CDKN2)	600160	CUTANEOUS MALIGNANT MELANOMA 2, CMM2	155601		Sequencing	490
Molecular Tests	CDKN2A (CYCLIN-DEPENDENT KINASE INHIBITOR 2A; P16; CDKN2)	600160	MALIGNANT MELANOMA WITH NEURAL CELL TUMORS » MELANOMA-ASTROCYTOMA SYNDROME	155755		Sequencing	490
Molecular Tests	CEBPA (CCAAT/ENHANCER-BINDING PROTEIN, ALPHA; C/EBP-ALPHA)	116897	LEUKEMIA, ACUTE MYELOID, AML	601626		Sequencing	450
Molecular Tests	CEL (CARBOXYL-ESTER LIPASE; LYSOPHOSPHOLIPASE; BILE SALT-STIMULATED LIPASE)	114840	MATURITY-ONSET DIABETES OF THE YOUNG, TYPE 8, WITH EXOCRINE DYSFUNCTION, MODY8 » DIABETES-PANCREATIC EXOCRINE DYSFUNCTION SYNDROME	609812		Sequencing	700
Molecular Tests	CEP290 (CENTROSOMAL PROTEIN, 290-KD, NEPHROCYSTIN 6; NPHP6)	610142	LEBER CONGENITAL AMAUROSIS, TYPE 1, LCA1 » RETINAL BLINDNESS, CONGENITAL	204000		Sequencing	820
Molecular Tests	CEP290 (CENTROSOMAL PROTEIN, 290-KD, NEPHROCYSTIN 6; NPHP6)	610142	JOUBERT SYNDROME, TYPE 5	610188		Sequencing	820
Molecular Tests	CEP290 (CENTROSOMAL PROTEIN, 290-KD, NEPHROCYSTIN 6; NPHP6)	610142	SENIOR-LOKEN SYNDROME TYPE 6, SLSN6	610189		Sequencing	820
Molecular Tests	CEP290 (CENTROSOMAL PROTEIN, 290-KD, NEPHROCYSTIN 6; NPHP6)	610142	MECKEL SYNDROME, TYPE 4	611134		Sequencing	820
Molecular Tests	CERKL (CERAMIDE KINASE-LIKE)	608381	RETINITIS PIGMENTOSA, TYPE 26, RP26	608380		Sequencing	680
Molecular Tests	CETP (CHOLESTERYL ESTER TRANSFER PROTEIN, LIPID TRANSFER PROTEIN I)	118470	CHOLESTERYL ESTER TRANSFER PROTEIN DEFICIENCY » CETP DEFICIENCY	607322		Sequencing	800
Molecular Tests	CFB (COMPLEMENT FACTOR B;PROPERDIN FACTOR B; FACTOR B; C3 PROACTIVATOR; GLYCINE-RICH BETA-	138470	MACULAR DEGENERATION, AGE-RELATED, REDUCED RISK OF	173470		Sequencing	1020
Molecular Tests	CFC1 (CRYPTIC PROTEIN)	605194	CONOTRUNCAL HEART MALFORMATIONS » TRUNCUS ARTERIOSUS COMMUNIS » DOUBLE OUTLET RIGHT VENTRICLE	217095		Sequencing	810
Molecular Tests	CFC1 (CRYPTIC PROTEIN)	605194	HETEROTAXY, VISCERAL, TYPE 2 (AUTOSOMAL DOMINANT)	605376		Sequencing	810
Molecular Tests	CFC1 (CRYPTIC PROTEIN)	605194	TRANSPOSITION OF THE GREAT ARTERIES, DEXTRO-LOOPED 2	613853		Sequencing	810
Molecular Tests	CFH (HF1, COMPLEMENT FACTOR H)	134370	HEMOLYTIC-UREMIC SYNDROME » COMBINED DEFICIENCY OF FACTOR H AND FACTOR H-LIKE 1	235400		Sequencing	1200
Molecular Tests	CFH (HF1, COMPLEMENT FACTOR H)	134370	HEMOLYTIC-UREMIC SYNDROME » COMBINED DEFICIENCY OF FACTOR H AND FACTOR H-LIKE 1	235400		Deletion-Duplication Testing	350
Molecular Tests	CFHR5 (COMPLEMENT FACTOR H-RELATED 5)	608593	HEMOLYTIC UREMIC SYNDROME, ATYPICAL, SUSCEPTIBILITY TO, TYPE 6	612926		Sequencing	680

Molecular Tests	CFI (I FACTOR, COMPLEMENT COMPONENT 3 INACTIVATOR)	217030	COMPLEMENT FACTOR I DEFICIENCY	217030		Sequencing	750
Molecular Tests	CFI (I FACTOR, COMPLEMENT COMPONENT 3 INACTIVATOR)	217030	COMPLEMENT FACTOR I DEFICIENCY	217030		Deletion/Duplication Tastes	Does Not exist
Molecular Tests	CFL2 (COFILIN 2, (MUSCLE))	601443	NEMALINE MYOPATHY 7, NEM7	610687		Sequencing	550
Molecular Tests	CFTR	602421	CYSTIC FIBROSIS, CF	219700		Sequencing	550
Molecular Tests	CFTR	602421	CYSTIC FIBROSIS, CF	219700		54 Mutations Kit	350
Molecular Tests	CFTR	602421	CYSTIC FIBROSIS, CF	219700		200 Mutations Kit (54 mutation kit and sequencing of exons 7, 10 and 19)	350
Molecular Tests	CFTR	602421	CYSTIC FIBROSIS, CF	219700		M.L.P.A.	250
Molecular Tests	CFTR	602421	CONGENITAL BILATERAL ABSENCE OF VAS DEFERENS, CBAVD	277180		Sequencing	550
Molecular Tests	CFTR	602421	CONGENITAL BILATERAL ABSENCE OF VAS DEFERENS, CBAVD	277180		54 Mutations Kit	350
Molecular Tests	CFTR	602421	CONGENITAL BILATERAL ABSENCE OF VAS DEFERENS, CBAVD	277180		200 Mutations Kit (54 mutation kit and sequencing of exons 7, 10 and 19)	550
Molecular Tests	CFTR	602421	CONGENITAL BILATERAL ABSENCE OF VAS DEFERENS, CBAVD	277180		M.L.P.A.	250
Molecular Tests	CGI58 (COMPARATIVE GENE IDENTIFICATION 58, ABHD5)	604780	CHANARIN-DORFMAN DISEASE » ICHTHYOTIC NEUTRAL LIPID STORAGE DISEASE » NEUTRAL LIPID STORAGE DISEASE » ICHTHYOSIFORM ERYTHRODERMA WITH LEUKOCYTE VACUOLATION	275630		Sequencing	1080
Molecular Tests	CHAT (CHOLINE ACETYLTRANSFERASE)	118490	MYASTHENIC SYNDROME, CONGENITAL, ASSOCIATED WITH EPISODIC APNEA » CONGENITAL MYASTHENIC SYNDROME, TYPE 5A » MYASTHENIA GRAVIS, FAMILIAL INFANTILE, TYPE 2	254210		Sequencing	1050
Molecular Tests	CHD1 (CHROMODOMAIN HELICASE DNA BINDING PROTEIN 1)	608997	CHARGE SYNDROME	214800		Sequencing	650
Molecular Tests	CHD1 (CHROMODOMAIN HELICASE DNA BINDING PROTEIN 1)	608997	CHARGE SYNDROME	214800		Deletion/Duplication Tastes	550
Molecular Tests	CHEK2 (CHECKPOINT KINASE 2)	604373	BREAST CANCER, FAMILIAL	114480		Sequencing	900
Molecular Tests	CHEK2 (CHECKPOINT KINASE 2)	604373	BREAST CANCER, FAMILIAL	114480		11 Mutation	440
Molecular Tests	CHEK2 (CHECKPOINT KINASE 2)	604373	LI-FRAUMENI SYNDROME 2	609265		Sequencing	900
Molecular Tests	CHM (REP1, RAB ESCORT PROTEIN 1, RAB GERANYLGERANYL TRANSFERASE)	300390	CHOROIDEREMIA » TAPETOCHOROIDAL DYSTROPHY, PROGRESSIVE	303100		Sequencing	1930
Molecular Tests	CHMP2B (CHMP FAMILY, MEMBER 2B; CHROMATIN-MODIFYING PROTEIN 2B; CHARGED MULTIVESICULAR BODY PROTEIN 2B; VACUOLAR PROTEIN SORTING 2 YEAST HOMOLOG OF, B, VPS2B)	609512	FRONTOTEMPORAL DEMENTIA, CHROMOSOME 3-LINKED, FTD3	600795		Sequencing	600
Molecular Tests	CHN1 (CHIMERIN 1; MERIN, ALPHA-1 GTPase-ACTIVATING PROTEIN; RHO, 2; ARHGAP2; MERIN, ALPHA-1 GTPase-ACTIVATING PROTEIN, RHO, 2; ARHGAP2)	118423	DUANE RETRACTION SYNDROME, TYPE 2	604356		Sequencing	740
Molecular Tests	CHRNA1 (CHOLINERGIC RECEPTOR, NICOTINIC, ALPHA POLYPEPTIDE 1; ACETYLCHOLINE RECEPTOR, MUSCLE, ALPHA SUBUNIT)	100690	MULTIPLE PTERYGIUM SYNDROME, LETHAL TYPE » PTERYGIUM SYNDROME, MULTIPLE, LETHAL TYPE	253290		Sequencing	650
Molecular Tests	CHRNA1 (CHOLINERGIC RECEPTOR, NICOTINIC, ALPHA POLYPEPTIDE 1; ACETYLCHOLINE RECEPTOR, MUSCLE, ALPHA SUBUNIT)	100690	MYASTHENIC SYNDROME, CONGENITAL, SLOW-CHANNEL » MYASTHENIC SYNDROME, CONGENITAL, FAST-CHANNEL	601462		Sequencing	650
Molecular Tests	CHRNA1 (CHOLINERGIC RECEPTOR, NICOTINIC, ALPHA POLYPEPTIDE 1; ACETYLCHOLINE RECEPTOR, MUSCLE, ALPHA SUBUNIT)	100690	MYASTHENIC SYNDROME, CONGENITAL, FAST-CHANNEL	608930		Sequencing	650
Molecular Tests	CHRNA2 (CHOLINERGIC RECEPTOR, NEURONAL NICOTINIC, ALPHA POLYPEPTIDE 2; ACETYLCHOLINE RECEPTOR, NEURONAL NICOTINIC, ALPHA-2 SUBUNIT)	118507	EPILEPSY, NOCTURNAL FRONTAL LOBE, TYPE 4 » EPILEPSY, FAMILIAL, WITH NOCTURNAL WANDERING AND/OR FEAR	610555		Sequencing	850
Molecular Tests	CHRNA4 (CHOLINERGIC RECEPTOR, NEURONAL NICOTINIC, ALPHA POLYPEPTIDE 4; ACETYLCHOLINE RECEPTOR, NEURONAL NICOTINIC, ALPHA-4 SUBUNIT)	118504	TOBACCO ADDICTION, SUSCEPTIBILITY TO » NICOTINE DEPENDENCY, SUSCEPTIBILITY TO	188890		Sequencing	700
Molecular Tests	CHRNA4 (CHOLINERGIC RECEPTOR, NEURONAL NICOTINIC, ALPHA POLYPEPTIDE 4; ACETYLCHOLINE RECEPTOR, NEURONAL NICOTINIC, ALPHA-4 SUBUNIT)	118504	EPILEPSY, NOCTURNAL FRONTAL LOBE, TYPE 1	600513		Sequencing	700
Molecular Tests	CHRNB1 (CHOLINERGIC RECEPTOR, NICOTINIC, BETA POLYPEPTIDE 1;ACETYLCHOLINE RECEPTOR, MUSCLE, BETA SUBUNIT)	100710	MYASTHENIC SYNDROME, CONGENITAL, SLOW-CHANNEL » MYASTHENIC SYNDROME, CONGENITAL, TYPE 3A	601462		Sequencing	650
Molecular Tests	CHRNB1 (CHOLINERGIC RECEPTOR, NICOTINIC, BETA POLYPEPTIDE 1;ACETYLCHOLINE RECEPTOR, MUSCLE, BETA SUBUNIT)	100710	MYASTHENIC SYNDROME, CONGENITAL, ASSOCIATED WITH ACETYLCHOLINE RECEPTOR DEFICIENCY » MYASTHENIC SYNDROME, CONGENITAL, TYPE 1d » MYASTHENIC SYNDROME, CONGENITAL, ASSOCIATED WITH FACIAL DYSMORPHISM	608931		Sequencing	650
Molecular Tests	CHRNB2 (CHOLINERGIC RECEPTOR, NEURONAL NICOTINIC, BETA POLYPEPTIDE 2)	118507	EPILEPSY, NOCTURNAL FRONTAL LOBE, TYPE 3	605375		Sequencing	700
Molecular Tests	CHRND (CHOLINERGIC RECEPTOR, NICOTINIC, DELTA POLYPEPTIDE; ACETYLCHOLINE RECEPTOR, MUSCLE, DELTA SUBUNIT)	100720	MYASTHENIC SYNDROME, CONGENITAL, SLOW-CHANNEL » MYASTHENIC SYNDROME, CONGENITAL, TYPE 3A	601462		Sequencing	750
Molecular Tests	CHRND (CHOLINERGIC RECEPTOR, NICOTINIC, DELTA POLYPEPTIDE; ACETYLCHOLINE RECEPTOR, MUSCLE, DELTA SUBUNIT)	100720	MYASTHENIC SYNDROME, CONGENITAL, FAST-CHANNEL	608930		Sequencing	750
Molecular Tests	CHRNE (CHOLINERGIC RECEPTOR, NICOTINIC, EPSILON POLYPEPTIDE; ACETYLCHOLINE RECEPTOR, MUSCLE, EPSILON SUBUNIT)	100725	MYASTHENIC SYNDROME, CONGENITAL, SLOW-CHANNEL » MYASTHENIC SYNDROME, CONGENITAL, TYPE 3A	601462		Sequencing	650

Molecular Tests	CHRNE (CHOLINERGIC RECEPTOR, NICOTINIC, EPSILON POLYPEPTIDE; ACETYLCHOLINE RECEPTOR, MUSCLE, EPSILON SUBUNIT)	100725	MYASTHENIC SYNDROME, CONGENITAL, FAST-CHANNEL	608930		Sequencing	650
Molecular Tests	CHRNE (CHOLINERGIC RECEPTOR, NICOTINIC, EPSILON POLYPEPTIDE; ACETYLCHOLINE RECEPTOR, MUSCLE, EPSILON SUBUNIT)	100725	MYASTHENIC SYNDROME, CONGENITAL, ASSOCIATED WITH ACETYLCHOLINE RECEPTOR DEFICIENCY » MYASTHENIC SYNDROME, CONGENITAL, TYPE 1d » MYASTHENIC SYNDROME, CONGENITAL, ASSOCIATED WITH FACIAL DYSMORPHISM	608931		Sequencing	650
Molecular Tests	CHRNG (CHOLINERGIC RECEPTOR, NICOTINIC, GAMMA POLYPEPTIDE)	100730	MYASTHENIA GRAVIS, NEONATAL TRANSIENT	100740		Sequencing	990
Molecular Tests	CHRNG (CHOLINERGIC RECEPTOR, NICOTINIC, GAMMA POLYPEPTIDE)	100730	MULTIPLE PTERYGIUM SYNDROME, LETHAL TYPE » PTERYGIUM MULTIPLEX, MULTIPLE, LETHAL TYPE	253290		Sequencing	990
Molecular Tests	CHRNG (CHOLINERGIC RECEPTOR, NICOTINIC, GAMMA POLYPEPTIDE)	100730	ESCOBAR SYNDROME » MULTIPLE PTERYGIUM SYNDROME, ESCOBAR VARIANT » MULTIPLE PTERYGIUM SYNDROME, NONLETHAL TYPE » PTERYGIUM COLLI SYNDROME	265000		Sequencing	990
Molecular Tests	CHST14 (CARBOHYDRATE SULFOTRANSFERASE 14;DERMATAN-4-SULFOTRANSFERASE 1; D4ST1; N-ACETYLGALACTOSAMINE 4-0 SULFOTRANSFERASE)	608429	EHLERS-DANLOS SYNDROME, MUSCULOCONTRACTURAL TYPE » ADDUCTED THUMB, CLUBFOOT, AND PROGRESSIVE JOINT AND SKIN LAXITY SYNDROME » ADDUCTED THUMB-CLUBFOOT SYNDROME » DERMATAN SULFATE DEFICIENCY	601776		Sequencing	600
Molecular Tests	NLRP3 (NLR FAMILY, PYRIN DOMAIN-CONTAINING 3; CIAS1 ; CRYOPYRIN)	606416	COLD URTICARIA, FCU » FAMILIAL COLD AUTOINFLAMMATORY SYNDROME	120100		Sequencing	580
Molecular Tests	NLRP3 (NLR FAMILY, PYRIN DOMAIN-CONTAINING 3; CIAS1 ; CRYOPYRIN)	606416	MUCKLE-WELLS SYNDROME	191900		Sequencing	580
Molecular Tests	NLRP3 (NLR FAMILY, PYRIN DOMAIN-CONTAINING 3; CIAS1 ; CRYOPYRIN)	606416	CINCA SYNDROME » MULTISYSTEM INFLAMMATORY DISEASE, NEONATAL ONSET, SEVERE	607115		Sequencing	580
Molecular Tests	NLRP3 (NLR FAMILY, PYRIN DOMAIN-CONTAINING 3; CIAS1 ; CRYOPYRIN)	606416	DEAFNESS, DFNA34 » DEAFNESS, AUTOSOMAL DOMINANT, WITH OR WITHOUT INFLAMMATION	617772		Sequencing	580
Molecular Tests	NLRP3 (NLR FAMILY, PYRIN DOMAIN-CONTAINING 3; CIAS1 ; CRYOPYRIN)	606416	KERATOENDOTHELITIS FUGAX HEREDITARIA	148190		Sequencing	580
Molecular Tests	OTOF (OTOFERLIN)	603454	DEAFNESS, DFNB9	609428		Sequencing	820
Molecular Tests	CISD2 (CDGSH IRON SULFUR DOMAIN PROTEIN 2; ZINC FINGER PROTEIN ZCD2; ENDOPLASMIC RETICULUM INTERMEMBRANE SMALL PROTEIN	611507	WOLFRAM SYNDROME, TYPE 2, WFS2	604928		Sequencing	250
Molecular Tests	CLCN1	118425	MYOTONIA LEVIOR			Sequencing	900
Molecular Tests	CLCN1	118425	MYOTONIA CONGENITA (AUTOSOMAL DOMINANT) » THOMSEN MYOTONIA	160800		Sequencing	900
Molecular Tests	CLCN1	118425	MYOTONIA CONGENITA (AUTOSOMAL RECESSIVE) » BECKER MYOTONIA	255700		Sequencing	900
Molecular Tests	CLCN2	600570	EPILEPSY, JUVENILE MYOCLONIC, IME » JME SYNDROME	606904		Sequencing	1350
Molecular Tests	CLCN2	600570	EPILEPSY WITH GRAND MAL SEIZURES ON AWAKENING	607628		Sequencing	1350
Molecular Tests	CLCN2	600570	EPILEPSY, IDIOPATHIC GENERALIZED	607631		Sequencing	1350
Molecular Tests	CLCN5	300008	DENT NEPHROLITHIASIS, TYPE 1	300009		Sequencing of both OCRL1 and CLCN5	690
Molecular Tests	CLCN5	300008	DENT NEPHROCALCINOSIS	300009		Deletion-Duplication Testing	650
Molecular Tests	CLCN5	300008	DENT'S DISEASE WITH HYPERCALCIURIA	310468		Sequencing of both OCRL1 and CLCN5	690
Molecular Tests	CLCN5	300008	NEPHROLITHIASIS/OSTEOPOROSIS, RENAL	310468		Deletion-Duplication Testing	650
Molecular Tests	CLCN7	602727	OSTEOPETROSIS, TYPE 2 (AUTOSOMAL DOMINANT), OPTA2 » ALBERS-SCHONBERG DISEASE (AUTOSOMAL DOMINANT) » MARBLE BONES (AUTOSOMAL DOMINANT) » OSTEOPETROSIS, AUTOSOMAL DOMINANT, TYPE II	166600		Sequencing	1970
Molecular Tests	CLCN7	602727	OSTEOPETROSIS, TYPE 4 (AUTOSOMAL RECESSIVE), OPTB4 » OSTEOPETROSIS, INFANTILE MALIGNANT, TYPE 3	611490		Sequencing	1970
Molecular Tests	CLCNKA (CLCK1, CHLORIDE CHANNEL, KIDNEY, A)	602024	BARTTER SYNDROME, TYPE 4 » BARTTER SYNDROME, INFANTILE, WITH SENSORINEURAL DEAFNESS	602522		Sequencing	1250
Molecular Tests	CLCNKB (CLCNB, CHLORIDE CHANNEL, KIDNEY, B)	602023	BARTTER SYNDROME, TYPE 4 » BARTTER SYNDROME, INFANTILE, WITH SENSORINEURAL DEAFNESS	602522		Sequencing	1100
Molecular Tests	CLCNKB (CLCNB, CHLORIDE CHANNEL, KIDNEY, B)	602023	BARTTER SYNDROME, TYPE 4 » BARTTER SYNDROME, INFANTILE, WITH SENSORINEURAL DEAFNESS	602522		Deletion-Duplication Testing	Upon Request
Molecular Tests	CLCNKB (CLCNB, CHLORIDE CHANNEL, KIDNEY, B)	602023	BARTTER SYNDROME, TYPE 3 » BARTTER SYNDROME, CLASSIC » BARTTER SYNDROME, TYPE 3, WITH HYPOCALCIURIA	607364		Sequencing	1100

Molecular Tests	CLCNKB (CLCNB, CHLORIDE CHANNEL, KIDNEY, B)	602023	BARTTER SYNDROME, TYPE 3 » BARTTER SYNDROME, CLASSIC » BARTTER SYNDROME, TYPE 3, WITH HYPOCALCIURIA	607364		Deletion-Duplication Testing	Upon Request
Molecular Tests	CLDN16 (CLAUDIN 16, PARACELLIN 1)	603959	HYPOMAGNESEMIA, PRIMARY » MAGNESIUM DEFECT IN RENAL TUBULAR TRANSPORT OF	248250		Sequencing	400
Molecular Tests	CLDN19 (CLAUDIN 19)	610036	HYPOMAGNESEMIA, RENAL, WITH OCULAR INVOLVEMENT » MACULAR COLOBOMA, BILATERAL, WITH HYPERCALCIURIA	248190		Sequencing	375
Molecular Tests	CLN PANEL: CLN1, CLN2, CLN3, CLN5, CLN6, CLN7, CLN8, CLN10		CEROID LIPOFUSCINOSIS, CLN			Sequencing	3490
Molecular Tests	CLN2 (TRIPEPTIDYL PEPTIDASE I; TPP1)	204500	CEROID LIPOFUSCINOSIS, CLN2 » JANSKY-BIELSCHOWSKY DISEASE	204500		Sequencing	950
Molecular Tests	CLN2 (TRIPEPTIDYL PEPTIDASE I; TPP1)	204500	CEROID LIPOFUSCINOSIS, CLN2 » JANSKY-BIELSCHOWSKY DISEASE	204500		2 Common Mutations: R208X and IVS5-1G>C	460
Molecular Tests	CLN3	607042	CEROID LIPOFUSCINOSIS, CLN3 » VOGT-SPIELMEYER DISEASE » BATTEN DISEASE	204200		Sequencing and Common 1kb Deletion	950
Molecular Tests	CLN5	608102	CEROID LIPOFUSCINOSIS, CLN5 » NEURONAL CEROID LIPOFUSCINOSIS, LATE INFANTILE, FINNISH VARIANT	256731		Sequencing	910
Molecular Tests	CLN6	606725	CEROID LIPOFUSCINOSIS, CLN6 » CEROID LIPOFUSCINOSIS, NEURONAL, LATE-INFANTILE	601780		Sequencing	720
Molecular Tests	CLN8	607837	CEROID LIPOFUSCINOSIS, CLN8 » CEROID LIPOFUSCINOSIS, NEURONAL, 8, NORTHERN EPILEPSY VARIANT » NORTHERN EPILEPSY » EPILEPSY, PROGRESSIVE, WITH MENTAL RETARDATION	610003		Sequencing	910
Molecular Tests	CLRN1 (CLARIN 1; USH3A; USH3)	606397	USHER SYNDROME, TYPE 3, USH3	276902		Sequencing	480
Molecular Tests	CLRN1 (CLARIN 1; USH3A; USH3)	606397	RETINITIS PIGMENTOSA, TYPE 61, RP61	614180		Sequencing	480
Molecular Tests	CNGA1 (CYCLIC NUCLEOTIDE-GATED CHANNEL, ALPHA-1; RETINAL ROD cGMP-GATED CHANNEL, ALPHA SUBUNIT)	123825	RETINITIS PIGMENTOSA, TYPE 49, RP49	613756		Sequencing	580
Molecular Tests	CNGA3 (CYCLIC NUCLEOTIDE-GATED CHANNEL, ALPHA-3; CONE PHOTORECEPTOR cGMP-GATED CHANNEL; CYCLIC NUCLEOTIDE-GATED CHANNEL, OLFACTORY, 3; CNG3)	600053	ACHROMATOPSIA, TYPE 2 » COLORBLINDNESS, TOTAL » ROD MONOCHROMATISM, TYPE 2	216900		Sequencing	680
Molecular Tests	CNGB1 (CYCLIC NUCLEOTIDE-GATED CHANNEL, BETA-1; CYCLIC NUCLEOTIDE-GATED CHANNEL, PHOTORECEPTOR, cGMP-GATED, 2; GLUTAMIC ACID-RICH PROTEIN 1; RETINAL ROD cGMP-GATED CHANNEL, BETA SUBUNIT)	600724	RETINITIS PIGMENTOSA, TYPE 45, RP45	613767		Sequencing	870
Molecular Tests	CNGB3 (CYCLIC NUCLEOTIDE-GATED CHANNEL, BETA-3)	605080	STARGARDT DISEASE, TYPE 1 » MACULAR DEGENERATION, JUVENILE » FUNDUS FLAVIMACULATUS » MACULAR DYSTROPHY WITH FLECKS, TYPE 1	248200		Sequencing	890
Molecular Tests	CNGB3 (CYCLIC NUCLEOTIDE-GATED CHANNEL, BETA-3)	605080	ACHROMATOPSIA, TYPE 3	262300		Sequencing	890
Molecular Tests	CNTNAP2 (CONTACTIN-ASSOCIATED PROTEIN-LIKE 2; CASPR2; NEUREXIN IV, DROSOPHILA, HOMOLOG OF; NRXN4)	604569	PITT-HOPKINS-LIKE SYNDROME, TYPE 1 » CORTICAL DYSPLASIA-FOCAL EPILEPSY SYNDROME	610042		Sequencing	Upon Request
Molecular Tests	COCH (COCHLIN)	603196	DEAFNESS, DFNA9 » DEAFNESS, (AUTOSOMAL DOMINANT) NONSYNDROMIC SENSORINEURAL 9	603196		Sequencing	970
Molecular Tests	COG7 (COMPONENT OF OLIGOMERIC GOLGI COMPLEX 7)	606978	CONGENITAL DISORDER OF GLYCOSYLATION, TYPE 2E, CDG2E	608779		Sequencing	1700
Molecular Tests	COG8 (COMPONENT OF OLIGOMERIC GOLGI COMPLEX 8)	606979	CONGENITAL DISORDER OF GLYCOSYLATION, TYPE 2H, CDG2H	611182		Sequencing	730
Molecular Tests	COH1	607817	COHEN SYNDROME, COH1	216550		Sequencing	5110
Molecular Tests	COH1	607817	COHEN SYNDROME, COH1	216550		Exon 23, including the "Finnish mutation" (c.3348-3349delCT)	480
Molecular Tests	COL10A1 (COLLAGEN, TYPE X, ALPHA1)	120110	METAPHYSEAL CHONDRODYSPLASIA, SCHMID TYPE	156500		Sequencing	830
Molecular Tests	COL11A1 (COLLAGEN, TYPE 11, ALPHA-1)	120280	MARSHALL SYNDROME	154780	Turn-around-time: 30 Weeks	Sequencing	3000
Molecular Tests	COL11A1 (COLLAGEN, TYPE 11, ALPHA-1)	120280	STICKLER SYNDROME, TYPE 2	604841	Turn-around-time: 30 Weeks	Sequencing	3000
Molecular Tests	COL11A2 (COLLAGEN, TYPE 11, ALPHA-2)	120290	STICKLER SYNDROME, TYPE 3	184840	Turn-around-time: 30 Weeks	Sequencing	3300
Molecular Tests	COL11A2 (COLLAGEN, TYPE 11, ALPHA-2)	120290	OTOSPONDYLOMEGAEPIPHYSEAL DYSPLASIA, OSMED	215150	Turn-around-time: 30 Weeks	Sequencing	3300
Molecular Tests	COL11A2 (COLLAGEN, TYPE 11, ALPHA-2)	120290	WEISSENBACHER-ZWEYMULLER SYNDROME » PIERRE ROBIN SYNDROME WITH FETAL CHONDRODYSPLASIA	277610	Turn-around-time: 30 Weeks	Sequencing	3300
Molecular Tests	COL11A2 (COLLAGEN, TYPE 11, ALPHA-2)	120290	DEAFNESS, DFNA13 » DEAFNESS, (AUTOSOMAL DOMINANT) NONSYNDROMIC SENSORINEURAL 13	601868	Turn-around-time: 30 Weeks	Sequencing	3300
Molecular Tests	COL11A2 (COLLAGEN, TYPE 11, ALPHA-2)	120290	DEAFNESS, DFNA53 » DEAFNESS, (AUTOSOMAL DOMINANT) NONSYNDROMIC SENSORINEURAL 53	609706	Turn-around-time: 30 Weeks	Sequencing	3300

Molecular Tests	COL17A1 (COLLAGEN, TYPE 17, ALPHA-1)	113811	EPIDERMOLYSIS BULLOSA, GENERALIZED ATROPHIC BENIGN » EPIDERMOLYSIS BULLOSA JUNCTIONALIS, PROGRESSIVE » EPIDERMOLYSIS BULLOSA JUNCTIONALIS, SEVERE NONLETHAL » EPIDERMOLYSIS BULLOSA JUNCTIONALIS, DISENTIS TYPE	226650		Sequencing	2500
Molecular Tests	COL1A1	120150	EHLERS-DANLOS TYPE 1, EDS1	130000	Only on skin fibroblast cultures from proband (See note)	Null Allele Detection	650
Molecular Tests	COL1A1	120150	EHLERS-DANLOS TYPE 1, EDS1	130000		Whole Gene Analysis	980
Molecular Tests	COL1A1	120150	OSTEOGENESIS IMPERFECTA TYPE 1, OI1	166200	Only on skin fibroblast cultures from proband (See note)	Null Allele Detection	650
Molecular Tests	COL1A1	120150	OSTEOGENESIS IMPERFECTA TYPE 1, OI1	166200		Whole Gene Analysis	980
Molecular Tests	COL1A1	120150	OSTEOGENESIS IMPERFECTA TYPE 2, OI2	166210	Only on skin fibroblast cultures from proband (See note)	Null Allele Detection	650
Molecular Tests	COL1A1	120150	OSTEOGENESIS IMPERFECTA TYPE 2, OI2	166210		Whole Gene Analysis	980
Molecular Tests	COL1A1	120150	OSTEOGENESIS IMPERFECTA TYPE 4, OI4	166220	Only on skin fibroblast cultures from proband (See note)	Null Allele Detection	650
Molecular Tests	COL1A1	120150	OSTEOGENESIS IMPERFECTA TYPE 4, OI4	166220		Whole Gene Analysis	980
Molecular Tests	COL1A1	120150	OSTEOGENESIS IMPERFECTA TYPE 3, OI3	259420	Only on skin fibroblast cultures from proband (See note)	Null Allele Detection	650
Molecular Tests	COL1A1	120150	OSTEOGENESIS IMPERFECTA TYPE 3, OI3	259420		Whole Gene Analysis	980
Molecular Tests	COL1A1 and COL1A2	120150	EHLERS-DANLOS TYPE 1, EDS1	130000	Only on skin fibroblast cultures from proband (See note)	Biochemical Test	650
Molecular Tests	COL1A1 and COL1A2	120150	EHLERS-DANLOS TYPE 7, DOMINANT, EDS7	130060		Splice Site Mutations Exon 6	400
Molecular Tests	COL1A1 and COL1A2	120150	OSTEOGENESIS IMPERFECTA TYPE 1, OI1	166200	Only on skin fibroblast cultures from proband (See note)	Biochemical Test	650
Molecular Tests	COL1A1 and COL1A2	120150	OSTEOGENESIS IMPERFECTA TYPE 2, OI2	166210	Only on skin fibroblast cultures from proband (See note)	Biochemical Test	650
Molecular Tests	COL1A1 and COL1A2	120150	OSTEOGENESIS IMPERFECTA TYPE 4, OI4	166220	Only on skin fibroblast cultures from proband (See note)	Biochemical Test	650
Molecular Tests	COL1A1 and COL1A2	120150	OSTEOGENESIS IMPERFECTA TYPE 3, OI3	259420	Only on skin fibroblast cultures from proband (See note)	Biochemical Test	650
Molecular Tests	COL1A2	120160	EHLERS-DANLOS TYPE 1, EDS1	130000		Whole Gene Analysis	980
Molecular Tests	COL1A2	120160	OSTEOGENESIS IMPERFECTA TYPE 1, OI1	166200		Whole Gene Analysis	980
Molecular Tests	COL1A2	120160	OSTEOGENESIS IMPERFECTA TYPE 2, OI2	166210		Whole Gene Analysis	980
Molecular Tests	COL1A2	120160	OSTEOGENESIS IMPERFECTA TYPE 4, OI4	166220		Whole Gene Analysis	980
Molecular Tests	COL1A2	120160	OSTEOGENESIS IMPERFECTA TYPE 3, OI3	259420		Whole Gene Analysis	980
Molecular Tests	COL2A1	120140	HYPOCHONDROGENESIS			Sequencing	1250
Molecular Tests	COL2A1	120140	STICKLER SYNDROME TYPE 1	108300		Sequencing	1250
Molecular Tests	COL2A1	120140	KNIEST DYSPLASIA	156550		Sequencing	1250
Molecular Tests	COL2A1	120140	SPONDYLOEPIPHYSEAL DYSPLASIA (SED CONGENITA)	183900		Sequencing	1250
Molecular Tests	COL2A1	120140	ACHONDROGENESIS TYPE 2	200610		Sequencing	1250
Molecular Tests	COL3A1	120180	EHLERS-DANLOS TYPE 4, EDS4	130050	Only on skin fibroblast cultures from proband (See note)	Biochemical Test	600
Molecular Tests	COL3A1	120180	EHLERS-DANLOS TYPE 4, EDS4	130050	Only on skin fibroblast cultures from proband (See note)	Null Allele Detection	650
Molecular Tests	COL3A1	120180	EHLERS-DANLOS TYPE 4, EDS4	130050	Only on skin fibroblast cultures from proband (See note)	Whole Gene Analysis	980
Molecular Tests	COL3A1	120180	FIBROMUSCULAR DYSPLASIA	135580	Only on skin fibroblast cultures from proband (See note)	Biochemical Test	600
Molecular Tests	COL3A1	120180	FIBROMUSCULAR DYSPLASIA	135580	Only on skin fibroblast cultures from proband (See note)	Null Allele Detection	650
Molecular Tests	COL3A1	120180	FIBROMUSCULAR DYSPLASIA	135580	Only on skin fibroblast cultures from proband (See note)	Whole Gene Analysis	980
Molecular Tests	COL4A1	120130	PORENCEPHALY, FAMILIAL » HEMIPLEGIA, INFANTILE, WITH PORENCEPHALY	175780		Sequencing	1595
Molecular Tests	COL4A1	120130	PORENCEPHALY, FAMILIAL » HEMIPLEGIA, INFANTILE, WITH PORENCEPHALY	175780		Deletion-Duplication Testing	800

Molecular Tests	COL4A1	120130	BRAIN SMALL VESSEL DISEASE WITH HEMORRHAGE » LEUKOENCEPHALOPATHY WITH AXENFELD-RIEGER ANOMALY » RETINAL ARTERIOLAR TORTUOSITY, INFANTILE HEMIPARESIS, AND LEUKOENCEPHALOPATHY	607595		Sequencing	1595
Molecular Tests	COL4A1	120130	BRAIN SMALL VESSEL DISEASE WITH HEMORRHAGE » LEUKOENCEPHALOPATHY WITH AXENFELD-RIEGER ANOMALY » RETINAL ARTERIOLAR TORTUOSITY, INFANTILE HEMIPARESIS, AND LEUKOENCEPHALOPATHY	607595		Deletion-Duplication Testing	800
Molecular Tests	COL4A1	120130	ANGIOPATHY, HEREDITARY, WITH NEPHROPATHY, ANEURYSMS, AND MUSCLE CRAMPS, HANAC	611773		Sequencing	1595
Molecular Tests	COL4A1	120130	ANGIOPATHY, HEREDITARY, WITH NEPHROPATHY, ANEURYSMS, AND MUSCLE CRAMPS, HANAC	611773		Deletion-Duplication Testing	800
Molecular Tests	COL4A3	120070	HEMATURIA, BENIGN FAMILIAL » THIN-BASEMENT-MEMBRANE NEPHROPATHY	141200		Sequencing	1300
Molecular Tests	COL4A3	120070	ALPORT SYNDROME (AUTOSOMAL RECESSIVE)	203780		Sequencing	1300
Molecular Tests	COL4A4	120131	HEMATURIA, BENIGN FAMILIAL » THIN-BASEMENT-MEMBRANE NEPHROPATHY	141200		Sequencing	1300
Molecular Tests	COL4A4	120131	ALPORT SYNDROME (AUTOSOMAL RECESSIVE)	203780		Sequencing	1300
Molecular Tests	COL4A5	303630	ALPORT SYNDROME (X-LINKED) » ALPORT SYNDROME-LIKE HEREDITARY NEPHRITIS	301050		Whole Gene Sequencing	1300
Molecular Tests	COL4A5	303630	ALPORT SYNDROME (X-LINKED) » ALPORT SYNDROME-LIKE HEREDITARY NEPHRITIS	301050		Deletion-Duplication Testing	500
Molecular Tests	COL5A1	120215	EHLERS-DANLOS TYPE 1, EDS1	130000	Only on skin fibroblast cultures from proband (See note)	Null Allele Detection	650
Molecular Tests	COL5A1	120215	EHLERS-DANLOS TYPE 1, EDS1	130000		Whole Gene Analysis	980
Molecular Tests	COL5A1	120215	EHLERS-DANLOS TYPE 2, EDS2	130010	Only on skin fibroblast cultures from proband (See note)	Null Allele Detection	650
Molecular Tests	COL5A1	120215	EHLERS-DANLOS TYPE 2, EDS2	130010		Whole Gene Analysis	980
Molecular Tests	COL5A1 and COL5A2	120215, 120190	EHLERS-DANLOS TYPE 1, EDS1	130000	Only on skin fibroblast cultures from proband (See note)	Biochemical Test	650
Molecular Tests	COL5A1 and COL5A2	120215, 120190	EHLERS-DANLOS TYPE 2, EDS2	130010	Only on skin fibroblast cultures from proband (See note)	Biochemical Test	650
Molecular Tests	COL5A2	120190	EHLERS-DANLOS TYPE 1, EDS1	130000		Whole Gene Analysis	980
Molecular Tests	COL5A2	120190	EHLERS-DANLOS TYPE 2, EDS2	130010		Whole Gene Analysis	980
Molecular Tests	COL7A1 (COLLAGEN, TYPE 7, ALPHA-1)	120120	TRANSIENT BULLOUS DERMOLYSIS OF THE NEWBORN » EPIDERMOLYSIS BULLOSA DYSTROPHICA, NEONATAL (AUTOSOMAL DOMINANT) » TOENAIL DYSTROPHY, ISOLATED	131705		Sequencing	3200
Molecular Tests	COL7A1 (COLLAGEN, TYPE 7, ALPHA-1)	120120	EPIDERMOLYSIS BULLOSA DYSTROPHICA (AUTOSOMAL DOMINANT) » EPIDERMOLYSIS BULLOSA DYSTROPHICA, PASINI TYPE » ALBOPAPULOID DOMINANT DYSTROPHIC EPIDERMOLYSIS BULLOSA » EPIDERMOLYSIS BULLOSA, PRETIBIAL, WITH LICHENOID FEATURES	131750		Sequencing	3200
Molecular Tests	COL7A1 (COLLAGEN, TYPE 7, ALPHA-1)	120120	EPIDERMOLYSIS BULLOSA, PRETIBIAL	131850		Sequencing	3200
Molecular Tests	COL7A1 (COLLAGEN, TYPE 7, ALPHA-1)	120120	EPIDERMOLYSIS BULLOSA WITH CONGENITAL LOCALIZED ABSENCE OF SKIN AND DEFORMITY OF NAILS » EPIDERMOLYSIS BULLOSA DYSTROPHICA, BART TYPE	132000		Sequencing	3200
Molecular Tests	COL7A1 (COLLAGEN, TYPE 7, ALPHA-1)	120120	EPIDERMOLYSIS BULLOSA DYSTROPHICA (AUTOSOMAL RECESSIVE) » EPIDERMOLYSIS BULLOSA DYSTROPHICA, TYPE HALLOPEAU-SIEMENS » EPIDERMOLYSIS BULLOSA DYSTROPHICA, LOCALISATA VARIANT (AUTOSOMAL RECESSIVE)	226600		Sequencing	3200
Molecular Tests	COL7A1 (COLLAGEN, TYPE 7, ALPHA-1)	120120	EPIDERMOLYSIS BULLOSA PRURIGINOSA (AUTOSOMAL DOMINANT AND RECESSIVE)	604129		Sequencing	3200
Molecular Tests	COL7A1 (COLLAGEN, TYPE 7, ALPHA-1)	120120	EPIDERMOLYSIS BULLOSA DYSTROPHICA WITH SUBCORNEAL CLEAVAGE » EPIDERMOLYSIS BULLOSA SIMPLEX SUPERFICIALIS » EPIDERMOLYSIS BULLOSA DYSTROPHICA, COCKAYNE-TOURAINE TYPE	607600		Sequencing	3200
Molecular Tests	COL9A1 (COLLAGEN, TYPE 9, ALPHA-1; CARTILAGE-SPECIFIC SHORT COLLAGEN)	120210	STICKLER SYNDROME, TYPE 1	108300		Sequencing	2300
Molecular Tests	COL9A1 (COLLAGEN, TYPE 9, ALPHA-1; CARTILAGE-SPECIFIC SHORT COLLAGEN)	120210	EPIPHYSEAL DYSPLASIA, MULTIPLE, TYPE 6, EDM6	120210		Sequencing	2300
Molecular Tests	COL9A2 (COLLAGEN, TYPE 9, ALPHA-2)	120260	EPIPHYSEAL DYSPLASIA, MULTIPLE, TYPE 2, EDM2	600204		Sequencing	2000
Molecular Tests	COLQ (COLLAGENIC TAIL OF ENDPLATE ACETYLCHOLINESTERASE; ACETYLCHOLINESTERASE-ASSOCIATED COLLAGEN)	603033	ENDPLATE ACETYLCHOLINESTERASE DEFICIENCY » ENGEL CONGENITAL MYASTHENIC SYNDROME » CONGENITAL MYASTHENIC SYNDROME, TYPE 1C	603034		Sequencing	1050
Molecular Tests	COMP	600310	MULTIPLE EPIPHYSEAL DYSPLASIA 1, EDM1	132400		Sequencing	1500
Molecular Tests	COMP	600310	PSEUDOACHONDROPLASIA	177170		Sequencing	1500
Molecular Tests	COQ2 (COQ2, S. CEREVISIAE, HOMOLOG OF; PARAHYDROXYBENZOATE-POLYPRENYLTRANSFERASE, MITOCHONDRIAL)	609825	COENZYME Q10 DEFICIENCY	607426		Sequencing	650
Molecular Tests	COQ9 (COQ9, S. CEREVISIAE, HOMOLOG OF)	612837	COENZYME Q10 DEFICIENCY, PRIMARY, TYPE 5	614654		Sequencing	990

Molecular Tests	COX10 (CYTOCHROME c OXIDASE ASSEMBLY PROTEIN COX10, HEME A:FARNESYLTRANSFERASE)	602125	CYTOCHROME c OXIDASE DEFICIENCY » COX DEFICIENCY » COMPLEX IV MITOCHONDRIAL RESPIRATORY CHAIN DEFICIENCY	220110		Sequencing	880
Molecular Tests	COX10 (CYTOCHROME c OXIDASE ASSEMBLY PROTEIN COX10, HEME A:FARNESYLTRANSFERASE)	602125	LEIGH SYNDROME	256000		Sequencing	880
Molecular Tests	COX15 (CYTOCHROME c OXIDASE ASSEMBLY PROTEIN COX15)	603646	CYTOCHROME c OXIDASE DEFICIENCY » COX DEFICIENCY » COMPLEX IV MITOCHONDRIAL RESPIRATORY CHAIN DEFICIENCY	220110		Sequencing	1020
Molecular Tests	COX15 (CYTOCHROME c OXIDASE ASSEMBLY PROTEIN COX15)	603646	LEIGH SYNDROME	256000		Sequencing	1020
Molecular Tests	COX4I2 (CYTOCHROME c OXIDASE, SUBUNIT IV, ISOFORM 2)	607976	EXOCRINE PANCREATIC INSUFFICIENCY, DYSERYTHROPOIETIC ANEMIA, AND CALVARIAL HYPEROSTOSIS	617714		Sequencing	580
Molecular Tests	COX6B1 (CYTOCHROME c OXIDASE, SUBUNIT VIb, POLYPEPTIDE 1)	124089	CYTOCHROME c OXIDASE DEFICIENCY » COX DEFICIENCY » COMPLEX IV MITOCHONDRIAL RESPIRATORY CHAIN DEFICIENCY	220110		Sequencing	500
Molecular Tests	CP (CERULOPLASMIN; FERROXIDASE)	117700	ACERULOPLASMINEMIA » HYPOCERULOPLASMINEMIA » CERULOPLASMIN DEFICIENCY » NEURODEGENERATION DUE TO CERULOPLASMIN	604290		Sequencing	1200
Molecular Tests	CPOX (CPO, COPROPORPHYRINOGEN OXIDASE)	123500	COPROPORPHYRIA » COPROPORPHYRINOGEN OXIDASE DEFICIENCY » HARDEROPORPHYRIA	121300		Sequencing	740
Molecular Tests	CPS1 (CARBAMOYL PHOSPHATE SYNTHETASE 1)	608307	CARBAMOYL PHOSPHATE SYNTHETASE 1 DEFICIENCY, HYPERAMMONEMIA DUE TO » CPS1 DEFICIENCY	237300		Sequencing	2430
Molecular Tests	CPT1A (CARNITINE PALMITOYLTRANSFERASE 1, LIVER, CPT1)	600528	CARNITINE PALMITOYLTRANSFERASE 1A DEFICIENCY » CPT1 DEFICIENCY	255120		Sequencing	850
Molecular Tests	CPT1B (CARNITINE PALMITOYLTRANSFERASE 1, MUSCLE)	601987	NO DISEASE			Sequencing	2020
Molecular Tests	CPT2 (CARNITINE PALMITOYLTRANSFERASE 2, LIVER, CPT2)	600650	MYOPATHY, VARIANT F			Sequencing	880
Molecular Tests	CPT2 (CARNITINE PALMITOYLTRANSFERASE 2, LIVER, CPT2)	600650	CARNITINE PALMITOYLTRANSFERASE 2 DEFICIENCY, LATE-ONSET » CARNITINE PALMITOYLTRANSFERASE 2 (CPT2) DEFICIENCY, MYOPATHIC » CARNITINE PALMITOYLTRANSFERASE 2 (CPT2) DEFICIENCY, ADULT-ONSET » CPT2 DEFICIENCY, MYOPATHIC	255110		Sequencing	880
Molecular Tests	CPT2 (CARNITINE PALMITOYLTRANSFERASE 2, LIVER, CPT2)	600650	CARNITINE PALMITOYLTRANSFERASE 2 DEFICIENCY, INFANTILE » CARNITINE PALMITOYLTRANSFERASE 2 DEFICIENCY WITH HYPOKETOTIC HYPOGLYCEMIA » CARNITINE PALMITOYLTRANSFERASE 2 DEFICIENCY, HEPATOCARDIOMUSCULAR » CPT2 DEFICIENCY, INFANTILE	600649		Sequencing	880
Molecular Tests	CPT2 (CARNITINE PALMITOYLTRANSFERASE 2, LIVER, CPT2)	600650	CARNITINE PALMITOYLTRANSFERASE 2 DEFICIENCY, LETHAL NEONATAL » CARNITINE PALMITOYLTRANSFERASE 2 DEFICIENCY, NEONATAL » CPT2 DEFICIENCY, LETHAL NEONATAL	608836		Sequencing	880
Molecular Tests	CRB1 (CRUMBS, DROSOPHILA, HOMOLOG OF, 1)	604210	PIGMENTED PARAVENOUS CHORIORETINAL ATROPHY	172870		Sequencing	770
Molecular Tests	CRB1 (CRUMBS, DROSOPHILA, HOMOLOG OF, 1)	604210	RETINITIS PIGMENTOSA, TYPE 12, RP12 » RP12 WITH COATS-LIKE EXUDATIVE VASCULOPATHY » RETINITIS PIGMENTOSA 12 WITH PARAFOVEOLAR PRESERVATION OR DEGENERATION OF RETINAL PIGMENT EPITHELIUM	600105		Sequencing	770
Molecular Tests	CRB1 (CRUMBS, DROSOPHILA, HOMOLOG OF, 1)	604210	FIBROCONGENITAL AMAUROSIS DUE TO DEFECT IN CRB1	204870		Sequencing	770
Molecular Tests	CREBBP (CREB-BINDING PROTEIN, CBP)	600140	RUBINSTEIN-TAYBI SYNDROME » BROAD THUMBS AND GREAT TOES, CHARACTERISTIC FACIES, AND MENTAL RETARDATION » BROAD THUMB-HALLUX SYNDROME	180849		Sequencing	910
Molecular Tests	CREBBP (CREB-BINDING PROTEIN, CBP)	600140	RUBINSTEIN-TAYBI SYNDROME » BROAD THUMBS AND GREAT TOES, CHARACTERISTIC FACIES, AND MENTAL RETARDATION » BROAD THUMB-HALLUX SYNDROME	180849		Deletion-Duplication Testing	550
Molecular Tests	CRTAP (CARTILAGE-ASSOCIATED PROTEIN)	605497	OSTEOGENESIS IMPERFECTA, TYPE 7, OI7	610682		Sequencing	550
Molecular Tests	CRX (CONE-ROD HOMEOBOX-CONTAINING GENE)	602225	CONE-ROD DYSTROPHY, TYPE 2, CORD2	120970		Sequencing	680
Molecular Tests	CRX (CONE-ROD HOMEOBOX-CONTAINING GENE)	602225	LEBER CONGENITAL AMAUROSIS, TYPE 7, LCA7	613829		Sequencing	680
Molecular Tests	CSRP3 (CYSTEINE- AND GLYCINE-RICH PROTEIN 3, LIM DOMAIN PROTEIN, CARDIAC)	600824	CARDIOMYOPATHY, DILATED, TYPE 1M	607482		Sequencing	1060
Molecular Tests	CSRP3 (CYSTEINE- AND GLYCINE-RICH PROTEIN 3, LIM DOMAIN PROTEIN, CARDIAC)	600824	CARDIOMYOPATHY, FAMILIAL HYPERTROPHIC, TYPE 12	612124		Sequencing	1060
Molecular Tests	CSTB (CYSTATIN B, STEFIN B)	601145	EPILEPSY, PROGRESSIVE MYOCLONUS » UNVERRICHT-LUNDBORG DISEASE	254800		Repeat	820
Molecular Tests	CTDP1 (C-TERMINAL DOMAIN OF RNA POLYMERASE II SUBUNIT A, PHOSPHATASE OF, SUBUNIT 1; TRANSCRIPTION FACTOR IIF-ASSOCIATING CYTOPHOSPHATASE 1; FCP1)	604927	CONGENITAL CATARACTS, FACIAL DYSMORPHISM, AND NEUROPATHY, CCFDN	604168		1 Mutation: IVS6, C-T, +389	250
Molecular Tests	CTF1 (CARDIOTROPHIN 1)	600435	CARDIOMYOPATHY, DILATED			Sequencing	960

Molecular Tests	CTNS (CYSTINOSIN)	606272	CYSTINOSIS, ADULT NONNEPHROPATHIC	219750		Sequencing	850
Molecular Tests	CTNS (CYSTINOSIN)	606272	CYSTINOSIS, ADULT NONNEPHROPATHIC	219750		Common 56 kb Deletion	250
Molecular Tests	CTNS (CYSTINOSIN)	606272	CYSTINOSIS, NEPHROPATHIC	219800		Sequencing	850
Molecular Tests	CTNS (CYSTINOSIN)	606272	CYSTINOSIS, NEPHROPATHIC	219800		Common 56 kb Deletion	250
Molecular Tests	CTNS (CYSTINOSIN)	606272	CYSTINOSIS, LATE-ONSET JUVENILE OR ADOLESCENT, NEPHROPATHIC TYPE » CYSTINOSIS, INTERMEDIATE	219900		Sequencing	850
Molecular Tests	CTNS (CYSTINOSIN)	606272	CYSTINOSIS, LATE-ONSET JUVENILE OR ADOLESCENT, NEPHROPATHIC TYPE » CYSTINOSIS, INTERMEDIATE	219900		Common 56 kb Deletion	250
Molecular Tests	CTRC (CHYMOTRYPSIN C, CALDECRIN)	601405	HEREDITARY PANCREATITIS	167800		Sequencing	960
Molecular Tests	CTSK (CATHEPSIN K)	601105	PYCNODYSOSTOSIS	265800			600
Molecular Tests	CYBA (p22 PHOX)	233690	CHRONIC GRANULOMATOUS DISEASE (AUTOSOMAL RECESSIVE) CYTOCHROME-b-NEGATIVE FORM	233690		Sequencing	940
Molecular Tests	CYBB (p91 PHOX)	306400	CHRONIC GRANULOMATOUS DISEASE (X-LINKED)	306400		Sequencing	750
Molecular Tests	CYP11A1 (CYTOCHROME P450, SUBFAMILY XIA, POLYPEPTIDE 1;CHOLESTEROL SIDE-CHAIN CLEAVAGE ENZYME;CYTOCHROME P450SCC	118485	ADRENAL INSUFFICIENCY, CONGENITAL, WITH 46,XY SEX REVERSAL	118485		Sequencing	1100
Molecular Tests	CYP11A1 (CYTOCHROME P450, SUBFAMILY XIA, POLYPEPTIDE 1;CHOLESTEROL SIDE-CHAIN CLEAVAGE ENZYME;CYTOCHROME P450SCC	118485	LIPOID CONGENITAL ADRENAL HYPERPLASIA	201710		Sequencing	1100
Molecular Tests	CYP11B1 (CYTOCHROME P450, SUBFAMILY 11B, POLYPEPTIDE 1, STEROID 11-BETA-HYDROXYLASE, P450C11)	610613	ADRENAL HYPERPLASIA, CONGENITAL, DUE TO STEROID 11-BETA-HYDROXYLASE DEFICIENCY » ADRENAL HYPERPLASIA 4 STEROID 11-BETA-HYDROXYLASE DEFICIENCY » 11-@BETA-HYDROXYLASE DEFICIENCY » ADRENAL HYPERPLASIA, HYPERTENSIVE FORM	202010		Sequencing	Upon Request
Molecular Tests	CYP11B1 (CYTOCHROME P450, SUBFAMILY 11B, POLYPEPTIDE 1, STEROID 11-BETA-HYDROXYLASE, P450C11)	610613	ADRENAL HYPERPLASIA, CONGENITAL, DUE TO STEROID 11-BETA-HYDROXYLASE DEFICIENCY » ADRENAL HYPERPLASIA 4 STEROID 11-BETA-HYDROXYLASE DEFICIENCY » 11-@BETA-HYDROXYLASE DEFICIENCY » ADRENAL HYPERPLASIA, HYPERTENSIVE FORM	202010		CYP11B1/CYP11B2 fusion	300
Molecular Tests	CYP11B2 (CYTOCHROME P450, SUBFAMILY XIB, POLYPEPTIDE 2, STEROID 11/18-BETA-HYDROXYLASE, STEROID 18-OXIDASE, ALDOSTERONE SYNTHASE, CORTICOSTERONE METHYLOXIDASE)	124080	CORTICOSTERONE METHYLOXIDASE TYPE 1 DEFICIENCY » ALDOSTERONE DEFICIENCY 1 » HYPERRENINEMIC HYPOALDOSTERONISM, FAMILIAL, 1 » ALDOSTERONE DEFICIENCY DUE TO DEFECT IN STEROID 18-HYDROXYLASE » 18-@HYDROXYLASE DEFICIENCY	203400		Sequencing	700
Molecular Tests	CYP17A1 (CYTOCHROME P450, FAMILY 17, SUBFAMILY A, POLYPEPTIDE 1; STEROID 17-ALPHA-MONOOXYGENASE)	609300	ADRENAL HYPERPLASIA, TYPE 5 » 17-@ALPHA-HYDROXYLASE/17,20-LYASE DEFICIENCY » 17-@ALPHA-HYDROXYLASE DEFICIENCY	202110		Sequencing	700
Molecular Tests	CYP19A1 (AROMATASE)	107910	AROMATASE DEFICIENCY » PSEUDOHERMAPHRODITISM, FEMALE, DUE TO PLACENTAL AROMATASE DEFICIENCY	107910		Sequencing	650
Molecular Tests	CYP1B1 (CYTOCHROME P450, SUBFAMILY 1, POLYPEPTIDE 1)	#WAARDE!	GLAUCOMA 3, PRIMARY INFANTILE A, GLC3A » BUPHTHALMOS	231300		Sequencing	450
Molecular Tests	CYP1B1 (CYTOCHROME P450, SUBFAMILY 1, POLYPEPTIDE 1)	#WAARDE!	PETERS ANOMALY	604229		Sequencing	450
Molecular Tests	CYP21A2	201910	HYPERANDROGENISM » 21-ALPHA-HYDROXYLASE DEFICIENCY » CYP21 DEFICIENCY			Sequencing and Deletions-Duplications	900
Molecular Tests	CYP21A2	201910	ADRENAL HYPERPLASIA, CONGENITAL DUE TO 21-HYDROXYLASE DEFICIENCY, CAH1 » 21-ALPHA-HYDROXYLASE DEFICIENCY » CYP21 DEFICIENCY	201910		Sequencing and Deletions-Duplications	900
Molecular Tests	CYP24A1 (CYTOCHROME P450, FAMILY 24, SUBFAMILY A, POLYPEPTIDE 1; VITAMIN D 24-HYDROXYLASE)	126065	HYPERCALCEMIA, INFANTILE » TRIHYDROXYCOPROSTANIC ACID IN BILE	143880		Sequencing	990
Molecular Tests	CYP27A1	606530	CEREBROTENDINOUS XANTHOMATOSIS, CTX » CEREBRAL CHOLESTERINOSIS	213700		Sequencing	530
Molecular Tests	CYP27B1	264700	PSEUDO-VITAMIN D DEFICIENCY RICKETS (AUTOSOMAL RECESSIVE), PDDR » RICKETS, PSEUDO-VITAMIN D DEFICIENCY RICKETS (AUTOSOMAL RECESSIVE)	264700		Sequencing	620
Molecular Tests	DARS2 (ASPARTYL-tRNA SYNTHETASE 2)	610956	LEUKOENCEPHALOPATHY WITH BRAINSTEM AND SPINAL CORD INVOLVEMENT AND LACTATE ELEVATION » MITOCHONDRIAL ASPARTYL-tRNA SYNTHETASE DEFICIENCY	611105		Sequencing	1400
Molecular Tests	DAX1 (NROB1)	300200	ADDISON DISEASE (X-LINKED) » ADRENAL HYPOPLASIA, CONGENITAL » CONGENITAL ADRENOCORTICAL HYPOPLASIA WITH HYPOGONADOTROPIC HYPOGONADISM	300200		Sequencing	710

Molecular Tests	DBH (DOPAMINE BETA-HYDROXYLASE, PLASMA; DOPAMINE BETA-MONOOXYGENASE)	609312	DOPAMINE BETA-HYDROXYLASE DEFICIENCY, CONGENITAL » NOREPINEPHRINE DEFICIENCY » ORTHOSTATIC HYPOTENSION	223360		Sequencing	1870
Molecular Tests	DBT (DIHYDROLIPOAMIDE BRANCHED-CHAIN TRANSACYLASE; BRANCHED-CHAIN ACYLTRANSFERASE, E2 COMPONENT)	248610	MAPLE SYRUP URINE DISEASE » BRANCHED-CHAIN KETOACIDURIA » BRANCHED-CHAIN ALPHA-KETO ACID DEHYDROGENASE DEFICIENCY » KETO ACID DECARBOXYLASE DEFICIENCY	248600		Sequencing	1250
Molecular Tests	DBT (DIHYDROLIPOAMIDE BRANCHED-CHAIN TRANSACYLASE; BRANCHED-CHAIN ACYLTRANSFERASE, E2 COMPONENT)	248610	MAPLE SYRUP URINE DISEASE » BRANCHED-CHAIN KETOACIDURIA » BRANCHED-CHAIN ALPHA-KETO ACID DEHYDROGENASE DEFICIENCY » KETO ACID DECARBOXYLASE DEFICIENCY	248600		Deletion-Duplication Testing	600
Molecular Tests	DCAF17 (DDB1- AND CUL4-ASSOCIATED FACTOR 17; CHROMOSOME 2 OPEN READING FRAME 57; C2ORF57)	612515	WOODHOUSE-SAKATI SYNDROME » HYPOGONADISM, ALOPECIA, DIABETES MELLITUS, MENTAL RETARDATION, DEAFNESS, AND EXTRAPYRAMIDAL SYNDROME	241080		Sequencing	2140
Molecular Tests	DCLRE1C (DNA CROSS-LINK REPAIR PROTEIN 1C, ARTEMIS)	605988	SEVERE COMBINED IMMUNODEFICIENCY WITH SENSITIVITY TO IONIZING RADIATION, SCID » SEVERE COMBINED IMMUNODEFICIENCY (AUTOSOMAL RECESSIVE), T CELL-NEGATIVE, B CELL-NEGATIVE, NK CELL-POSITIVE, WITH SENSITIVITY TO IONIZING RADIATION	602450		Sequencing and Deletion-Duplication Testing	2260
Molecular Tests	DCLRE1C (DNA CROSS-LINK REPAIR PROTEIN 1C, ARTEMIS)	605988	OMENN SYNDROME » RETICULOENDOTHELIOSIS, FAMILIAL, WITH EOSINOPHILIA	603554		Sequencing and Deletion-Duplication Testing	2260
Molecular Tests	DCTN1 (DYNACTIN 1)	601143	AMYOTROPHIC LATERAL SCLEROSIS, TYPE 1, AT 55	105400		Sequencing	2300
Molecular Tests	DCTN1 (DYNACTIN 1)	601143	NEUROMUSCULAR SYSTEM, HEREDITARY MOTOR, TYPE IIB	607641		Sequencing	2300
Molecular Tests	DCX (DOUBLECORTIN)	300121	DOUBLE CORTEX SYNDROME » LISSENCEPHALY (X-LINKED)	300067		Sequencing	750
Molecular Tests	DCX (DOUBLECORTIN)	300121	DOUBLE CORTEX SYNDROME » LISSENCEPHALY (X-LINKED)	300067		Deletion-Duplication Testing	580
Molecular Tests	DDC (DOPA DECARBOXYLASE; AROMATIC L-AMINO ACID DECARBOXYLASE; AADC)	107930	AROMATIC L-AMINO ACID DECARBOXYLASE DEFICIENCY » AADC DEFICIENCY	608643		Sequencing	850
Molecular Tests	DES (DESMIN)	125660	MYOPATHY, MYOFIBRILLAR, DESMIN-RELATED » DESMINOPATHY, PRIMARY	601419		Sequencing	770
Molecular Tests	DES (DESMIN)	125660	CARDIOMYOPATHY, DILATED, 1I; CMD1I	604765		Sequencing	770
Molecular Tests	DGUOK (DEOXYGUANOSINE KINASE; MITOCHONDRIAL DGK)	601465	MITOCHONDRIAL DNA DEPLETION SYNDROME, HEPATOCEREBRAL FORM	251880		Sequencing	690
Molecular Tests	DHCR7 (DEHYDROCHOLESTEROL REDUCTASE)	602858	SMITH-LEMLI-OPITZ SYNDROME, SLO	270400		Sequencing	1550
Molecular Tests	DHDDS (DEHYDRODOLICHYL DIPHOSPHATE SYNTHASE; DEDOL-PP SYNTHASE)	608172	RETINITIS PIGMENTOSA, TYPE 59, RP59	613861		Sequencing	590
Molecular Tests	DHH (DESERT HEDGEHOG)	605423	GONADAL DYSGENESIS, 46XY, PARTIAL	233400		Sequencing	1340
Molecular Tests	DHH (DESERT HEDGEHOG)	605423	GONADAL DYSGENESIS, 46XY, PARTIAL, WITH MINIFASCICULAR NEUROPATHY	607080		Sequencing	740
Molecular Tests	DIA1 (CYTOCHROME b5 REDUCTASE, CYB5R)	250800	METHEMOGLOBINEMIA DUE TO DEFICIENCY OF METHEMOGLOBIN REDUCTASE	250800		Sequencing	890
Molecular Tests	DIO1 (DEIODINASE, IODOTHYRONINE, TYPE 1; THYROXINE DEIODINASE, TYPE 1)	147892	HYPERTHYROXINEMIA DUE TO DECREASED PERIPHERAL CONVERSION OF T4	147892		Sequencing	850
Molecular Tests	DIO3 (DEIODINASE, IODOTHYRONINE, TYPE 3; THYROXINE DEIODINASE, TYPE 3)	601413	UNKNOWN DISEASE			Sequencing	800
Molecular Tests	DIO3 (DEIODINASE, IODOTHYRONINE, TYPE 3; THYROXINE DEIODINASE, TYPE 3)	601038	UNKNOWN DISEASE			Sequencing	500
Molecular Tests	DJ1	602533	PARKINSON DISEASE, TYPE 7, DJ1-RELATED	606324		Sequencing	630
Molecular Tests	DJ1	602533	PARKINSON DISEASE, TYPE 7, DJ1-RELATED	606324		Deletion-Duplication Testing	630
Molecular Tests	DKC1 (DYSKERIN)	300126	HOYERAAL-HREIDARSSON SYNDROME	300240		Sequencing	1840
Molecular Tests	DKC1 (DYSKERIN)	300126	DYSKERATOSIS CONGENITA (X-LINKED), DKC	305000		Sequencing	1840
Molecular Tests	DLAT (DIHYDROLIPOAMIDE S-ACETYLTRANSFERASE; PYRUVATE DEHYDROGENASE COMPLEX, E2 SUBUNIT; PDCE2; MG	608770	PYRUVATE DEHYDROGENASE E2 DEFICIENCY » LACTIC ACIDEMIA DUE TO DEFECT OF E2 (DIHYDROLIPOAMIDE ACETYLTRANSFERASE) OF THE PYRUVATE DEHYDROGENASE COMPLEX	245348		Sequencing	1450

Molecular Tests	DLD (DIHYDROLIPOAMIDE DEHYDROGENASE; BRANCHED CHAIN ALPHA-KETO ACID DEHYDROGENASE COMPLEX, E3 COMPONENT; PYRUVATE DEHYDROGENASE COMPONENT E3; GLYCINE CLEAVAGE SYSTEM L PROTEIN)	238331	MAPLE SYRUP URINE DISEASE » BRANCHED-CHAIN KETOACIDURIA » BRANCHED-CHAIN ALPHA-KETO ACID DEHYDROGENASE DEFICIENCY » KETO ACID DECARBOXYLASE DEFICIENCY	248600		Sequencing	1530
Molecular Tests	DLG3	300189	MENTAL RETARDATION, NONSPECIFIC (X-LINKED)	300189		Sequencing	1100
Molecular Tests	DLL3	602768	SPONDYLOCOSTAL DYSOSTOSIS, AUTOSOMAL RECESSIVE 1, SCDO1	277300		Sequencing	650
Molecular Tests	DMD (DYSTROPHIN)	300377	BECKER MUSCULAR DYSTROPHY, BMD » MUSCULAR DYSTROPHY, BECKER MUSCULAR DYSTROPHY	300376		Deletions-Duplications (MLPA)	500
Molecular Tests	DMD (DYSTROPHIN)	300377	BECKER MUSCULAR DYSTROPHY, BMD » MUSCULAR DYSTROPHY, BECKER MUSCULAR DYSTROPHY	300376		Sequencing	1400
Molecular Tests	DMD (DYSTROPHIN)	300377	DILATED CARDIOMYOPATHY (X-LINKED)	302045		Deletions-Duplications (MLPA)	500
Molecular Tests	DMD (DYSTROPHIN)	300377	DILATED CARDIOMYOPATHY (X-LINKED)	302045		Sequencing	1400
Molecular Tests	DMD (DYSTROPHIN)	300377	DUCHENNE MUSCULAR DYSTROPHY, DMD » MUSCULAR DYSTROPHY, DUCHENNE MUSCULAR DYSTROPHY	310200		Deletions-Duplications (MLPA)	500
Molecular Tests	DMD (DYSTROPHIN)	300377	DUCHENNE MUSCULAR DYSTROPHY, DMD » MUSCULAR DYSTROPHY, DUCHENNE MUSCULAR DYSTROPHY	310200		Sequencing	1400
Molecular Tests	DMP1 (DENTIN MATRIX ACIDIC PHOSPHOPROTEIN 1)	600980	HYPOPHOSPHATEMIC RICKETS (AUTOSOMAL RECESSIVE) » HYPOPHOSPHATEMIA (AUTOSOMAL RECESSIVE)	241520		Sequencing	595
Molecular Tests	DMPK (DM KINASE, MYOTONIN)	605377	MYOTONIC DYSTROPHY, TYPE 1 » STEINERT DISEASE	160900		Repeat analysed by PCR and TP-PCR	500
Molecular Tests	DMPK (DM KINASE, MYOTONIN)	605377	MYOTONIC DYSTROPHY, TYPE 1 » STEINERT DISEASE	160900	At least 20 microgram DNA of high molecular weight	Repeat analysed by Southern Blot	500
Molecular Tests	DNASE1 (DEOXYRIBONUCLEASE 1; DNase 1, LYSOSOMAL)	125505	SYSTEMIC LUPUS ERYTHEMATOSUS	152700		Sequencing	1600
Molecular Tests	DNM2 (DYNAMIN 2, DYN2)	602378	MYOPATHY, CENTRONUCLEAR (AUTOSOMAL DOMINANT)	160150		Sequencing	1550
Molecular Tests	DNM2 (DYNAMIN 2, DYN2)	602378	CHARCOT-MARIE-TOOTH DISEASE, INTERMEDIATE B, CMTDIB, (AUTOSOMAL DOMINANT)	606482		Sequencing	1550
Molecular Tests	DOK7 (DOWNSTREAM OF TYROSINE KINASE 7)	610285	MYASTHENIA, LIMB-GIRDLE, FAMILIAL » CONGENITAL MYASTHENIC SYNDROME, TYPE 1B	254300		Sequencing	750
Molecular Tests	DPAGT1 (DOLICHYL-PHOSPHATE N-ACETYLGLUCOSAMINEPHOSPHOTRANSFERASE; UDP-GlcNAc:DOLICHYL-PHOSPHATE N-ACETYLGLUCOSAMINE-PHOSPHOTRANSFERASE)	191350	CONGENITAL DISORDER OF GLYCOSYLATION, TYPE 1J, CDG1J	608093		Sequencing	1110
Molecular Tests	DPM1 (DOLICHYL-PHOSPHATE MANNOSYLTRANSFERASE 1, CATALYTIC SUBUNIT; DOLICHOL-PHOSPHATE MANNOSYLTRANSFERASE)	603503	CONGENITAL DISORDER OF GLYCOSYLATION, TYPE 1E, CDG1E	608799		Sequencing	960
Molecular Tests	DRPLA (ATROPHIN 1)	125370	DENTATORUBRAL-PALLIDOLUYSIAN ATROPHY, DRPLA	125370		Repeat	250
Molecular Tests	DSC2 (DESMOCOLLIN 2, DESMOSOMAL GLYCOPROTEIN 2/3)	125645	ARRHYTHMOGENIC RIGHT VENTRICULAR DYSPLASIA, FAMILIAL, TYPE 11, ARVD11 » ARRHYTHMOGENIC RIGHT VENTRICULAR CARDIOMYOPATHY, TYPE 10, ARVC10	610476		Sequencing	1060
Molecular Tests	DSG2 (DESMOGLEIN 2)	125671	ARRHYTHMOGENIC RIGHT VENTRICULAR DYSPLASIA, FAMILIAL, TYPE 10, ARVD10 » ARRHYTHMOGENIC RIGHT VENTRICULAR CARDIOMYOPATHY, TYPE 10, ARVC10	610193		Sequencing	1010
Molecular Tests	DSP (DESMOPLAKIN)	125647	KERATOSIS PALMOPLANTARIS STRIATA, TYPE 2	125647		Sequencing	700
Molecular Tests	DSP (DESMOPLAKIN)	125647	CARDIOMYOPATHY, DILATED, WITH WOOLLY HAIR AND KERATODERMA » PALMOPLANTAR KERATODERMA WITH LEFT VENTRICULAR CARDIOMYOPATHY AND WOOLLY HAIR » CARVAJAL SYNDROME	605676		Sequencing	700
Molecular Tests	DSP (DESMOPLAKIN)	125647	ARRHYTHMOGENIC RIGHT VENTRICULAR DYSPLASIA, FAMILIAL, TYPE 8, ARVD8 » ARRHYTHMOGENIC RIGHT VENTRICULAR CARDIOMYOPATHY, TYPE 8, ARVC8	607450		Sequencing	700
Molecular Tests	DSP (DESMOPLAKIN)	125647	SKIN FRAGILITY-WOOLLY HAIR SYNDROME	607655		Sequencing	700
Molecular Tests	DSP (DESMOPLAKIN)	125647	EPIDERMOLYSIS BULLOSA, LETHAL ACANTHOLYTIC	609638		Sequencing	700
Molecular Tests	DTNBP1 (HPS7; DYSTROBREVIN-BINDING PROTEIN 1; DYSBINDIN; SANDY, MOUSE, HOMOLOG OF; SDY)	607145	HERMANSKY-PUDLAK SYNDROME, TYPE 7, HPS7	614076		Sequencing	580
Molecular Tests	DYM (DYMECLIN)	607461	DYGGVE-MELCHIOR-CLAUSEN DISEASE	223800		Sequencing	1800
Molecular Tests	DYM (DYMECLIN)	607461	SMITH-MCCORT DYSPLASIA	607326		Sequencing	1800
Molecular Tests	DYNC2H1 (DYNEIN, CYTOPLASMIC 2, HEAVY CHAIN 1; DNCH2; DHC2)	603297	SHORT RIB-POLYDACTYLY SYNDROME, TYPE 3 » VERMA-NAUMOFF SYNDROME » POLYDACTYLY WITH NEONATAL CHONDRODYSTROPHY, TYPE 3	263510		Sequencing	2860
Molecular Tests	DYNC2H1 (DYNEIN, CYTOPLASMIC 2, HEAVY CHAIN 1; DNCH2; DHC2)	603297	SHORT RIB-POLYDACTYLY SYNDROME, TYPE 2 » MAJEWSKI SYNDROME » POLYDACTYLY WITH NEONATAL CHONDRODYSTROPHY, TYPE 2	263520		Sequencing	2860
Molecular Tests	DYNC2H1 (DYNEIN, CYTOPLASMIC 2, HEAVY CHAIN 1; DNCH2; DHC2)	603297	ASPHYXIATING THORACIC DYSTROPHY, TYPE 3	613091		Sequencing	2860
Molecular Tests	DYSF (DYSFERLIN)	603009	MUSCULAR DYSTROPHY, LIMB-GIRDLE, TYPE 2B, LGMD2B	253601		Sequencing	2010
Molecular Tests	DYSF (DYSFERLIN)	603009	MIYOSHI MYOPATHY » MUSCULAR DYSTROPHY, DISTAL, LATE-ONSET (AUTOSOMAL RECESSIVE)	254130		Sequencing	2010

Molecular Tests	DYSF (DYSFERLIN)	603009	MYOPATHY, DISTAL, WITH ANTERIOR TIBIAL ONSET	606768			Sequencing	3010
Molecular Tests	DYT1 (TORSA, TORSIN A)	605204	DYSTONIA MUSCULORUM DEFORMANS » TORSION DYSTONIA, EARLY ONSET, DYT1	128100			Sequencing	550
Molecular Tests	DYT1 (TORSA, TORSIN A)	605204	DYSTONIA MUSCULORUM DEFORMANS » TORSION DYSTONIA, EARLY ONSET, DYT1	128100			GAG deletion	250
Molecular Tests	EBP (EMOPAMIL-BINDING PROTEIN, 3-@BETA-HYDROXYSTEROID-DELTA-8, DELTA-7 ISOMERASE)	300205	CHONDRODYSPLASIA PUNCTATA 2 (X-LINKED DOMINANT), CDPX2 » CONRADI-HUNERMANN SYNDROME » HAJDU-CHEINEY SYNDROME	302960			Sequencing	250
Molecular Tests	ECM1	602201	LIPOID PROTEINOSIS OF URBACH AND WIETHE » MEMBRANOPROLIFERATIVE GLOMERULONEPHRITIS, TYPE II	247100			Sequencing	510
Molecular Tests	ED1 (ECTODYSPLASIN A, EDA)	300451	ECTODERMAL DYSPLASIA, HYPOHIDROTIC/X-LINKED, ED1	305100			Sequencing	750
Molecular Tests	EDA1 (ECTODYSPLASIN 1)	604095	ANHIDROTIC ECTODERMAL DYSPLASIA 3, ED3 » HYPOHIDROTIC ECTODERMAL DYSPLASIA (AUTOSOMAL DOMINANT), EDA3 » ECTODERMAL DYSPLASIA, HYPOHIDROTIC, TYPE 2	129490			Sequencing	750
Molecular Tests	EDA1 (ECTODYSPLASIN 1)	604095	HYPOHIDROTIC ECTODERMAL DYSPLASIA (AUTOSOMAL RECESSIVE) » ECTODERMAL DYSPLASIA, HYPOHIDROTIC (AUTOSOMAL RECESSIVE)	224900			Sequencing	750
Molecular Tests	EDARADD (EDAR-ASSOCIATED DEATH DOMAIN)	606605	ANHIDROTIC ECTODERMAL DYSPLASIA 3, ED3 » HYPOHIDROTIC ECTODERMAL DYSPLASIA (AUTOSOMAL DOMINANT), EDA3 » ECTODERMAL DYSPLASIA, HYPOHIDROTIC, TYPE 3	129490			Sequencing	750
Molecular Tests	EDARADD (EDAR-ASSOCIATED DEATH DOMAIN)	606605	HYPOHIDROTIC ECTODERMAL DYSPLASIA (AUTOSOMAL RECESSIVE) » ECTODERMAL DYSPLASIA, HYPOHIDROTIC (AUTOSOMAL RECESSIVE)	224900			Sequencing	750
Molecular Tests	EDN3 (ENDOTHELIN 3)	131242	CENTRAL HYPOVENTILATION SYNDROME, CONGENITAL » ONDINE-HIRSCHSPRUNG DISEASE » HAJDAD SYNDROME » CHRONIC IDIOPATHIC CONSTIPATION, SUSCEPTIBILITY TO	209880			Sequencing	480
Molecular Tests	EDN3 (ENDOTHELIN 3)	131242	WAARDENBURG SYNDROME, TYPE 4B, W54B » WAARDENBURG SYNDROME, TYPE 4B, WITH HIRSCHSPRUNG DISEASE	613265			Sequencing	480
Molecular Tests	EDNRA (ENDOTHELIN RECEPTOR, TYPE A)	131243	HIRSCHSPRUNG DISEASE, SUSCEPTIBILITY TO, TYPE 4, MCKBA	613711			Sequencing	480
Molecular Tests	EDNRB (ENDOTHELIN RECEPTOR, TYPE B)	131244	WAARDENBURG-SHAH SYNDROME » WAARDENBURG SYNDROME, TYPE 4 » WAARDENBURG-HIRSCHSPRUNG DISEASE	277580			Sequencing	550
Molecular Tests	EDNRB (ENDOTHELIN RECEPTOR, TYPE B)	131244	HIRSCHSPRUNG DISEASE, SUSCEPTIBILITY TO, TYPE 2	600155			Sequencing	550
Molecular Tests	EDNRB (ENDOTHELIN RECEPTOR, TYPE B)	131244	ABCD SYNDROME » ALBINISM-BLACK LOCK (CELL MIGRATION) DISORDER OF THE MELANOCYTES OF THE GUT AND DEAFNESS	600501			Sequencing	550
Molecular Tests	EFEMP1 (EGF-CONTAINING FIBULIN-LIKE EXTRACELLULAR MATRIX PROTEIN 1, FIBULIN 3, FIBRILLIN-LIKE)	601548	DOYNE HONEYCOMB RETINAL DYSTROPHY » MALATTIA LEVENTINESE » DRUSEN, RADIAL, AUTOSOMAL DOMINANT	126600			Sequencing	850
Molecular Tests	EFHC1 (EF-HAND DOMAIN (C-TERMINAL)-CONTAINING PROTEIN 1; MYOCLONIN 1)	608815	EPILEPSY, MYOCLONIC JUVENILE » JANZ SYNDROME » PETIT MAL, IDIOPATHIC	254770			Sequencing	800
Molecular Tests	EFHC1 (EF-HAND DOMAIN (C-TERMINAL)-CONTAINING PROTEIN 1; MYOCLONIN 1)	608815	EPILEPSY, JUVENILE ABSENCE, SUSCEPTIBILITY TO, TYPE 1	607631			Sequencing	800
Molecular Tests	EFNB1 (EPHRIN B1, EPLG2, LERK2, EFL5)	300035	CRANIOFRONTONASAL SYNDROME, CFNS » CRANIOFRONTONASAL DYSOSTOSIS	304110			Sequencing	850
Molecular Tests	EFNB1 (EPHRIN B1, EPLG2, LERK2, EFL5)	300035	CRANIOFRONTONASAL DYSOSTOSIS » CRANIOFRONTONASAL SYNDROME, CFNS	304110			Deletion-Duplication Testing	570
Molecular Tests	EGF (EPIDERMAL GROWTH FACTOR, UROGASTRONE)	131530	HYPOMAGNESEMIA, RENAL, NORMOCALCIURIC » HYPOMAGNESEMIA, RENAL, NORMOCALCIURIC	611718			Sequencing	1300
Molecular Tests	EGR2 (EARLY GROWTH RESPONSE 2)	129010	CHARCOT-MARIE-TOOTH DISEASE, DEMYELINATING, TYPE 4F, CMT4F » DEJERINE-SOTTAS HYPERTROPHIC NEUROPATHY » CHARCOT-MARIE-TOOTH AND SENSORY NEUROPATHY, TYPE 3, DSMN3	145900			Sequencing	550
Molecular Tests	EGR2 (EARLY GROWTH RESPONSE 2)	129010	CHARCOT-MARIE-TOOTH DISEASE, DEMYELINATING, TYPE 4E, CMT4E » CONGENITAL HYPOMYELINATING NEUROPATHY	605253			Sequencing	550
Molecular Tests	EGR3 (EARLY GROWTH RESPONSE 3)	128991	CHARCOT-MARIE-TOOTH DISEASE, AXONAL, TYPE 1D, CMT1D	607678			Sequencing	550
Molecular Tests	EHMT1 (EUCHROMATIC HISTONE METHYLTRANSFERASE 1; EUHMTASE1)	607001	KLEEFSTRA SYNDROME » CHROMOSOME 9q34.3 DELETION SYNDROME	610253			Sequencing	2180

Molecular Tests	EIF2AK3 (EUKARYOTIC TRANSLATION INITIATION FACTOR 2-ALPHA KINASE 3)	604032	EPIPHYSEAL DYSPLASIA, MULTIPLE, WITH EARLY-ONSET DIABETES MELLITUS » MED-IDDM SYNDROME » IDDM-MED SYNDROME	226980		Sequencing	745
Molecular Tests	EIF2B1 (EUKARYOTIC TRANSLATION INITIATION FACTOR 2B, SUBUNIT 1)	606686	LEUKOENCEPHALOPATHY WITH VANISHING WHITE MATTER » CHILDHOOD ATAXIA WITH CENTRAL NERVOUS SYSTEM HYPOMYELINIZATION » CREE LEUKOENCEPHALOPATHY	603896		Sequencing	850
Molecular Tests	EIF2B2 (EUKARYOTIC TRANSLATION INITIATION FACTOR 2B, SUBUNIT 2)	606454	LEUKOENCEPHALOPATHY WITH VANISHING WHITE MATTER » CHILDHOOD ATAXIA WITH CENTRAL NERVOUS SYSTEM HYPOMYELINIZATION » CREE LEUKOENCEPHALOPATHY	603896		Sequencing	850
Molecular Tests	EIF2B3 (EUKARYOTIC TRANSLATION INITIATION FACTOR 2B, SUBUNIT 3)	606273	LEUKOENCEPHALOPATHY WITH VANISHING WHITE MATTER » CHILDHOOD ATAXIA WITH CENTRAL NERVOUS SYSTEM HYPOMYELINIZATION » CREE LEUKOENCEPHALOPATHY	603896		Sequencing	850
Molecular Tests	EIF2B4 (EUKARYOTIC TRANSLATION INITIATION FACTOR 2B, SUBUNIT 4)	606687	LEUKOENCEPHALOPATHY WITH VANISHING WHITE MATTER » CHILDHOOD ATAXIA WITH CENTRAL NERVOUS SYSTEM HYPOMYELINIZATION » CREE LEUKOENCEPHALOPATHY	603896		Sequencing	850
Molecular Tests	EIF2B5 (EUKARYOTIC TRANSLATION INITIATION FACTOR 2B, SUBUNIT 5)	603945	LEUKOENCEPHALOPATHY WITH VANISHING WHITE MATTER » CHILDHOOD ATAXIA WITH CENTRAL NERVOUS SYSTEM HYPOMYELINIZATION » CREE LEUKOENCEPHALOPATHY	603896		Sequencing	850
Molecular Tests	ELA2 (ELASTASE 2)	130130	CYCLIC HEMATOPOIESIS » CYCLIC NEUTROPENIA	162800		Sequencing	850
Molecular Tests	ELA2 (ELASTASE 2)	130130	NEUTROPENIA, SEVERE CONGENITAL, SCN » CONGENITAL NEUTROPENIA » INFANTILE AGRANULOCYTOSIS	202700		Sequencing	850
Molecular Tests	ELN (ELASTIN)	130160	CUTIS LAXA (AUTOSOMAL DOMINANT)	123700		Sequencing	980
Molecular Tests	ELN (ELASTIN)	130160	CUTIS LAXA (AUTOSOMAL DOMINANT)	123700		MLPA	400
Molecular Tests	ELN (ELASTIN)	130160	SUPRAVALVAR AORTIC STENOSIS, EISENBERG TYPE	185500		Sequencing	980
Molecular Tests	ELN (ELASTIN)	130160	SUPRAVALVAR AORTIC STENOSIS, EISENBERG TYPE	185500		MLPA	400
Molecular Tests	ELOVL4 (ELONGATION OF VERY LONG CHAIN FATTY ACIDS-LIKE 4)	605512	STARGARDT DISEASE, TYPE 3 » MACULAR DYSTROPHY WITH FLECKS, TYPE 3	600110		Sequencing	530
Molecular Tests	EMERIN (EMD)	300384	EMERY-DREYFUSS MUSCULAR DYSTROPHY (X-LINKED), EDMD » MUSCULAR DYSTROPHY, EMERY-DREYFUSS MUSCULAR DYSTROPHY (X-LINKED), EDMD	310300		Sequencing	580
Molecular Tests	EMX2 (EMPTY SPIRACLES, DROSOPHILA, 2, HOMOLOG OF)	600035	SCHIZENCEPHALY	269160		Sequencing	600
Molecular Tests	ENG (ENDOGLIN)	131195	TELANGIECTASIA, HEREDITARY HEMORRHAGIC, OF RENDU, OSLER, AND WEBER, TYPE 1 » OSLER-RENDU-WEBER DISEASE	187300		Sequencing	1100
Molecular Tests	ENG (ENDOGLIN)	131195	TELANGIECTASIA, HEREDITARY HEMORRHAGIC, OF RENDU, OSLER, AND WEBER, TYPE 1 » OSLER-RENDU-WEBER DISEASE	187300		Deletion-Duplication Testing	730
Molecular Tests	ENG and ALK1		TELANGIECTASIA, HEREDITARY HEMORRHAGIC, OF RENDU, OSLER, AND WEBER » OSLER-RENDU-WEBER DISEASE			Sequencing and MLPA for 2 Genes	2300
Molecular Tests	ENG and ALK1		TELANGIECTASIA, HEREDITARY HEMORRHAGIC, OF RENDU, OSLER, AND WEBER » OSLER-RENDU-WEBER DISEASE			MLPA for 2 Genes	820
Molecular Tests	ENPP1 (ECTONUCLEOTIDE PYROPHOSPHATASE / PHOSPHODIESTERASE 1;PHOSPHODIESTERASE I / NUCLEOTIDE PYROPHOSPHATASE 1; PDNP1; PLASMA CELL MEMBRANE GLYCOPROTEIN PC-1; PC1; NUCLEOTIDE	173335	ARTERIAL CALCIFICATION, GENERALIZED, OF INFANCY » CORONARY SCLEROSIS, MEDIAL, OF INFANCY	208000		Sequencing	1275
Molecular Tests	ENPP1 (ECTONUCLEOTIDE PYROPHOSPHATASE / PHOSPHODIESTERASE 1;PHOSPHODIESTERASE I / NUCLEOTIDE PYROPHOSPHATASE 1; PDNP1; PLASMA CELL MEMBRANE GLYCOPROTEIN PC-1; PC1; NUCLEOTIDE	173335	HYPOPHOSPHATEMIC RICKETS (AUTOSOMAL RECESSIVE), TYPE 2	613312		Sequencing	1275
Molecular Tests	EP300 (E1A-BINDING PROTEIN, 300-KD)	602700	RUBINSTEIN-TAYBI SYNDROME » BROAD THUMBS AND GREAT TOES, CHARACTERISTIC FACIES, AND MENTAL RETARDATION	180849		Sequencing	950

Molecular Tests	EPM2A (LAFORIN)	607566	MYOCLONIC EPILEPSY OF LAFORA » LAFORA DISEASE » EPILEPSY, PROGRESSIVE MYOCLONIC TYPE 2	254780		Sequencing	920
Molecular Tests	ERCC4 (EXCISION-REPAIR, COMPLEMENTING DEFECTIVE, IN CHINESE HAMSTER, 4; XPF)	133520	XERODERMA PIGMENTOSUM, COMPLEMENTATION GROUP F, XPF » XERODERMA PIGMENTOSUM, TYPE 6, XP6	278760		Sequencing	1000
Molecular Tests	ERCC4 (EXCISION-REPAIR, COMPLEMENTING DEFECTIVE, IN CHINESE HAMSTER, 4; XPF)	133520	XFE PROGEROID SYNDROME » XPF-ERCC1 PROGEROID SYNDROME	610965		Sequencing	1000
Molecular Tests	ERCC5 (EXCISION-REPAIR, COMPLEMENTING DEFECTIVE, IN CHINESE HAMSTER, 5; ERCM2, XPG, XPGC, UVDR)	133530	XERODERMA PIGMENTOSUM, COMPLEMENTATION GROUP G, XPG » XERODERMA PIGMENTOSUM, TYPE 7, XP7 » XERODERMA PIGMENTOSUM, TYPE G/COCKAYNE SYNDROME, INCLUDED	278780		Sequencing	1100
Molecular Tests	ERCC6(EXCISION-REPAIR CROSS-COMPLEMENTING, GROUP 6; CSB)	133540	COCKAYNE SYNDROME, TYPE 2 » COCKAYNE SYNDROME, TYPE B	133540		Sequencing	1250
Molecular Tests	ERCC8 (EXCISION-REPAIR CROSS-COMPLEMENTING, GROUP 8; CSA)	609412	COCKAYNE SYNDROME, TYPE 1 » COCKAYNE SYNDROME, TYPE A	216400		Sequencing	850
Molecular Tests	ESCO2 (ESTABLISHMENT OF COHESION 1, S. CEREVISIAE, HOMOLOG OF, 2)	609353	ROBERTS SYNDROME			Sequencing	1540
Molecular Tests	ESCO2 (ESTABLISHMENT OF COHESION 1, S. CEREVISIAE, HOMOLOG OF, 2)	609353	SC PHOCOMELIA SYNDROME » SC PSEUDOTHALIDOMIDE SYNDROME	269000		Sequencing	1540
Molecular Tests	ETFA (ELECTRON TRANSFER FLAVOPROTEIN, ALPHA POLYPEPTIDE)	608053	MULTIPLE ACYL-CoA DEHYDROGENASE DEFICIENCY, MADD » GLUTARIC ACIDURIA, TYPE 2 » ETHYLMALONIC-ADIPICACIDURIA » ETFA DEFICIENCY	231680		Sequencing	890
Molecular Tests	ETFB (ELECTRON TRANSFER FLAVOPROTEIN, BETA POLYPEPTIDE)	130410	MULTIPLE ACYL-CoA DEHYDROGENASE DEFICIENCY, MADD » GLUTARIC ACIDURIA, TYPE 2 » ETHYLMALONIC-ADIPICACIDURIA	231680		Sequencing	600
Molecular Tests	ETFDH (ELECTRON TRANSFER FLAVOPROTEIN DEHYDROGENASE; ELECTRON TRANSFER FLAVOPROTEIN:UBIQUINONE OXIDOREDUCTASE)	231675	MULTIPLE ACYL-CoA DEHYDROGENASE DEFICIENCY, MADD » GLUTARIC ACIDURIA, TYPE 2 » ETHYLMALONIC-ADIPICACIDURIA	231680		Sequencing	1010
Molecular Tests	ETHE1	608451	ENCEPHALOPATHY, ETHYLMALONIC	602473		Sequencing	820
Molecular Tests	EVC	604831	ELLIS-VAN CREVELD SYNDROME	225500		Sequencing	1280
Molecular Tests	EVC2	607261	ELLIS-VAN CREVELD SYNDROME	225500		Sequencing	1710
Molecular Tests	EXT1 (EXOSTOSIN 1)	133700	MULTIPLE EXOSTOSES, TYPE 1, EXT1, HME	133700		Sequencing	900
Molecular Tests	EXT2 (EXOSTOSIN 2)	133701	MULTIPLE EXOSTOSES, TYPE 2, EXT2, HME	133701		Sequencing	900
Molecular Tests	EYA1	601653	BRANCHIO-OTO-RENAL SYNDROME, TYPE1, BOR1	113650		Sequencing	600
Molecular Tests	EYA1	601653	BRANCHIO-OTO-RENAL SYNDROME, TYPE1, BOR1	113650		Deletion-Duplication Testing	Upon Request
Molecular Tests	EYS (EYES SHUT, DROSOPHILA, HOMOLOG OF; SPACEMAKER; SPAM)	612424	RETINITIS PIGMENTOSA, TYPE 25, RP25	602772		Sequencing	1100
Molecular Tests	F10 (COAGULATION FACTOR 10)	227600	FACTOR X DEFICIENCY » STUART-PROWER FACTOR DEFICIENCY	227600		Sequencing	Upon Request
Molecular Tests	F11 (FACTOR 11, COAGULATION FACTOR 11)	264900	PTA DEFICIENCY » COAGULATION FACTOR 11 DEFICIENCY » ROSENTHAL SYNDROME	264900		Sequencing	370
Molecular Tests	F2 (FACTOR 2; COAGULATION FACTOR 2; THROMBIN; PROTHROMBIN)	176930	HYPOPROTHROMBINEMIA » DYSPROTHROMBINEMIA » HYPERPROTHROMBINEMIA	176930		Sequencing	1250
Molecular Tests	F5 (COAGULATION FACTOR 5; FACTOR 5 LEIDEN; APC COFACTOR)	612309	FACTOR V DEFICIENCY » OWREN PARAHEMOPHILIA » LABILE FACTOR DEFICIENCY	227400		Sequencing	1350
Molecular Tests	F7 (FACTOR 7, COAGULATION FACTOR 7)	227500	FACTOR VII DEFICIENCY » HYPOPROCONVERTINEMIA	227500		Sequencing	1250
Molecular Tests	F8 (FACTOR 8)	306700	HAEMOPHILIA A	306700		Sequencing	710
Molecular Tests	F8 (FACTOR 8)	306700	HAEMOPHILIA A	306700		Intron 22 Inversion	390
Molecular Tests	F8 (FACTOR 8)	306700	HAEMOPHILIA A	306700		Intron 1 Inversion	390
Molecular Tests	F9 (FACTOR 9)	306900	HAEMOPHILIA B	306900		Sequencing	710
Molecular Tests	FACL4 (FATTY ACID COA LIGASE, LONG CHAIN 4)	300157	MENTAL RETARDATION, NONSPECIFIC (X-LINKED), TYPE 63, MRX63	300387		Sequencing	850
Molecular Tests	FACL4 (FATTY ACID COA LIGASE, LONG CHAIN 4)	300157	MENTAL RETARDATION, NONSPECIFIC (X-LINKED), TYPE 68, MRX68	300387		Sequencing	850

Molecular Tests	FAH (FUMARYLACETOACETATE HYDROLASE, FUMARYLACETOACETASE)	276700	TYROSINEMIA, TYPE 1 » HEPATORENAL TYROSINEMIA » FUMARYLACETOACETASE DEFICIENCY » FAH DEFICIENCY	276700		Sequencing	980
Molecular Tests	FAH (FUMARYLACETOACETATE HYDROLASE, FUMARYLACETOACETASE)	276700	TYROSINEMIA, TYPE 1 » HEPATORENAL TYROSINEMIA » FUMARYLACETOACETASE DEFICIENCY » FAH DEFICIENCY	276700		5 Exons (Most Common Mutations)	710
Molecular Tests	FAM126A (FAMILY WITH SEQUENCE SIMILARITY 126, MEMBER A; DOWNREGULATED BY CTNNB1, PROTEIN A; DRCTNNB1A; HYCCIN)	610531	LEUKODYSTROPHY, HYPOMYELINATING, TYPE 5 » HYPOMYELINATION AND CONGENITAL CATARACT	610532		Sequencing	1360
Molecular Tests	FAM161A (FAMILY WITH SEQUENCE SIMILARITY 161, MEMBER A)	613596	RETINITIS PIGMENTOSA, TYPE 28, RP28	606068		Sequencing	580
Molecular Tests	FAM58A (FAMILY WITH SEQUENCE SIMILARITY 58, MEMBER A)	300708	TOE SYNDACTYLY, TELECANTHUS, AND ANOGENITAL AND RENAL MALFORMATIONS » STAR SYNDROME » SYNDACTYLY WITH RENAL AND ANOGENITAL MALFORMATIONS	300707		Sequencing	1250
Molecular Tests	FAM58A (FAMILY WITH SEQUENCE SIMILARITY 58, MEMBER A)	300708	TOE SYNDACTYLY, TELECANTHUS, AND ANOGENITAL AND RENAL MALFORMATIONS » STAR SYNDROME » SYNDACTYLY WITH RENAL AND ANOGENITAL MALFORMATIONS	300707		Deletion-Duplication Testing	600
Molecular Tests	FANCA	607139	FANCONI ANEMIA » FANCONI PANCYTOPENIA	227650		Sequencing	5000
Molecular Tests	FANCA	607139	FANCONI ANEMIA » FANCONI PANCYTOPENIA	227650		8 Exons: Exons 13, 27, 29, 34-38, representing 67% of reported	1000
Molecular Tests	FANCC	227645	FANCONI ANEMIA, COMPLEMENTATION GROUP C » FANCONI PANCYTOPENIA, TYPE 3	227645		Sequencing	1190
Molecular Tests	FANCC	227645	FANCONI ANEMIA, COMPLEMENTATION GROUP C » FANCONI PANCYTOPENIA, TYPE 3	227645		1 Mutation: IVS4+4A-G	230
Molecular Tests	FANCG (X-RAY REPAIR, COMPLEMENTING DEFECTIVE, IN CHINESE HAMSTER, 9; XRCC9)	602956	FANCONI ANEMIA, COMPLEMENTATION GROUP G	614082		Sequencing	1190
Molecular Tests	FASTKD2 (FAST KINASE DOMAIN-CONTAINING PROTEIN 2)	612322	CYTOCHROME c OXIDASE DEFICIENCY » COX DEFICIENCY » COMPLEX 4, MITOCHONDRIAL RESPIRATORY CHAIN, DEFICIENCY OF	220110		Sequencing	1300
Molecular Tests	FBLN4 (FIBULIN 4, EGF-CONTAINING FIBULIN-LIKE EXTRACELLULAR MATRIX PROTEIN 2, EFEMP2)	607844	CUTIS LAXA (AUTOSOMAL RECESSIVE)	219100		Sequencing	600
Molecular Tests	FBLN5 (FIBULIN 5)	604580	CUTIS LAXA (AUTOSOMAL DOMINANT)	123700		Sequencing	600
Molecular Tests	FBLN5 (FIBULIN 5)	604580	CUTIS LAXA (AUTOSOMAL RECESSIVE)	219100		Sequencing	600
Molecular Tests	FBLX4 (F-BOX AND LEUCINE-RICH REPEAT PROTEIN 4)	605654	MITOCHONDRIAL DNA DEPLETION SYNDROME 13 (ENCEPHALOMYOPATHIC TYPE)	615471		Sequencing	Upon Request
Molecular Tests	FBN1 (FIBRILLIN 1)	134797	FAMILIAL AORTIC ANEURYSM			Sequencing	950
Molecular Tests	FBN1 (FIBRILLIN 1)	134797	FAMILIAL AORTIC ANEURYSM			Deletion-Duplication Testing	400
Molecular Tests	FBN1 (FIBRILLIN 1)	134797	ECTOPIA LENTIS	129600		Sequencing	950
Molecular Tests	FBN1 (FIBRILLIN 1)	134797	ECTOPIA LENTIS	129600		Deletion-Duplication Testing	400
Molecular Tests	FBN1 (FIBRILLIN 1)	134797	MARFAN SYNDROME, TYPE 1, MFS1	154700		Sequencing	950
Molecular Tests	FBN1 (FIBRILLIN 1)	134797	MARFAN SYNDROME, TYPE 1, MFS1	154700		Deletion-Duplication Testing	400
Molecular Tests	FBN1 (FIBRILLIN 1)	134797	SHPHRINTZEN–GOLDBERG CRANIOSYNOSTOSIS SYNDROME	182212		Sequencing	950
Molecular Tests	FBN1 (FIBRILLIN 1)	134797	SHPHRINTZEN–GOLDBERG CRANIOSYNOSTOSIS SYNDROME	182212		Deletion-Duplication Testing	400
Molecular Tests	FBN1 (FIBRILLIN 1)	134797	MASS PHENOTYPE » MASS SYNDROME	604308		Sequencing	950
Molecular Tests	FBN1 (FIBRILLIN 1)	134797	MASS PHENOTYPE » MASS SYNDROME	604308		Deletion-Duplication Testing	400
Molecular Tests	FBN2 (FIBRILLIN 2)	121050	CONTRACTURAL CONGENITAL ARACHNODACTYLY, CCA » BEALS SYNDROME	121050		Sequencing	950
Molecular Tests	FBP1 (FRUCTOSE-1,6-BISPHOSPHATASE 1)	611570	FRUCTOSE-1,6-BISPHOSPHATASE DEFICIENCY	229700		Sequencing	700
Molecular Tests	FBXO7 (F-BOX ONLY PROTEIN 7; FBX7; FBX)	605648	PARKINSON DISEASE, TYPE 15, PARK15 (AUTOSOMAL RECESSIVE)	260300		Sequencing	650
Molecular Tests	FECH (FERROCHELATASE)	177000	PROTOPORPHYRIA, ERYTHROPOIETIC » HEME SYNTHETASE DEFICIENCY » FERROCHELATASE DEFICIENCY	177000		Sequencing	900
Molecular Tests	FERMT1 (FERMITIN FAMILY (DROSOPHILA) HOMOLOG 1; KIND1; KINDLIN 1)	607900	KINDLER SYNDROME » POIKILODERMA, HEREDITARY ACROKERATOTIC » BULLOUS ACROKERATOTIC POIKILODERMA OF KINDLER AND WEARY	173650		Sequencing	1500

Molecular Tests	FGA (FIBRINOGEN ALPHA)	134820	AMYLOIDOSIS, FAMILIAL VISCERAL » AMYLOIDOSIS, TYPE 8 » OSTERTAG TYPE AMYLOIDOSIS » GERMAN TYPE AMYLOIDOSIS » AMYLOIDOSIS, FAMILIAL RENAL	105200		Sequencing	550
Molecular Tests	FGA (FIBRINOGEN ALPHA)	134820	VENOUS THROMBOEMBOLISM	188050		Sequencing	550
Molecular Tests	FGA (FIBRINOGEN ALPHA)	134820	AFIBRINOGENEMIA » DYSFIBRINOGENEMIA » HYPODYSFIBRINOGENEMIA, CONGENITAL	202400		Sequencing	550
Molecular Tests	FGB (FIBRINOGEN BETA)	134830	VENOUS THROMBOEMBOLISM	188050		Sequencing	650
Molecular Tests	FGB (FIBRINOGEN BETA)	134830	AFIBRINOGENEMIA » DYSFIBRINOGENEMIA » HYPODYSFIBRINOGENEMIA, CONGENITAL	202400		Sequencing	650
Molecular Tests	FGD1	305400	AARSKOG SYNDROME » FACIODIGITOGENITAL SYNDROME	305400		Sequencing	1275
Molecular Tests	FGF14 (FIBROBLAST GROWTH FACTOR 14)	601515	SPINOCEREBELLAR ATAXIA 27, SCA27 » CEREBELLAR ATAXIA (AUTOSOMAL DOMINANT), FGF14-RELATED	609307		Sequencing	380
Molecular Tests	FGF23 (FIBROBLAST GROWTH FACTOR 23)	605380	HYPOPHOSPHATEMIC RICKETS (AUTOSOMAL DOMINANT) » RICKETS, HYPOPHOSPHATEMIC RICKETS (AUTOSOMAL DOMINANT)	193100		Sequencing	300
Molecular Tests	FGF23 (FIBROBLAST GROWTH FACTOR 23)	605380	HYPOPHOSPHATEMIC RICKETS (AUTOSOMAL DOMINANT) » RICKETS, HYPOPHOSPHATEMIC RICKETS (AUTOSOMAL DOMINANT)	193100		Deletion-Duplication Testing	Upon Request
Molecular Tests	FGF3 (FIBROBLAST GROWTH FACTOR 3; ONCOGENE INT2; INT2; V-INT2 MURINE MAMMARY TUMOR VIRUS INTEGRATION SITE ONCOGENE HOMOLOG)	164950	DEAFNESS, CONGENITAL, WITH INNER EAR AGENESIS, MICROTIA, AND MICRODONTIA » DEAFNESS, CONGENITAL, WITH LABYRINTHINE APLASIA, MICROTIA, AND MICRODONTIA » DEAFNESS WITH LAMM	610706		Sequencing	1100
Molecular Tests	FGFR1	136350	PFEIFFER SYNDROME » CRANIOSYNOSTOSIS, PFEIFFER SYNDROME	101600		Selected exons. Also includes testing of selected exons of FGFR2	510
Molecular Tests	FGFR1	136350	JACKSON-WEISS SYNDROME » CRANIOSYNOSTOSIS, JACKSON-WEISS SYNDROME	123150		Sequencing	510
Molecular Tests	FGFR1	136350	KALLMANN SYNDROME TYPE 2, KAL2 » HYPOGONADOTROPIC HYPOGONADISM AND ANOSMIA, TYPE 2	147950		Sequencing	1390
Molecular Tests	FGFR2	176943	DIFFERENT DISEASES			Sequencing	1390
Molecular Tests	FGFR2	176943	APERT SYNDROME » CRANIOSYNOSTOSIS, APERT SYNDROME	101200		2 Mutations: S252W, P253R	350
Molecular Tests	FGFR2	176943	PFEIFFER SYNDROME » CRANIOSYNOSTOSIS, PFEIFFER SYNDROME	101600		Selected exons. Also includes testing of selected exons of FGFR1	510
Molecular Tests	FGFR2	176943	JACKSON-WEISS SYNDROME » CRANIOSYNOSTOSIS, JACKSON-WEISS SYNDROME	123150		Sequencing	510
Molecular Tests	FGFR2	176943	CROUZON SYNDROME » CRANIOSYNOSTOSIS, CROUZON SYNDROME	123500		Sequencing	510
Molecular Tests	FGFR2	176943	BEARE-STEVENSON CUTIS GYRATA » CRANIOSYNOSTOSIS, BEARE-STEVENSON CUTIS GYRATA	123790		Sequencing	510
Molecular Tests	FGFR3	134934	DIFFERENT DISEASES			Sequencing	1100
Molecular Tests	FGFR3	134934	SADDAN DYSPLASIA » CRANIOSYNOSTOSIS, SADDAN DYSPLASIA			Sequencing	510
Molecular Tests	FGFR3	134934	CROUZON CRANIOSYNOSTOSIS WITH ACANTHOSIS NIGRICANS » CROUZONO DERMO SKELETAL SYNDROME » CRANIOSYNOSTOSIS, CROUZON CRANIOSYNOSTOSIS WITH ACANTHOSIS NIGRICANS			Sequencing	510
Molecular Tests	FGFR3	134934	ACHONDROPLASIA	100800		2 Mutations: G380R, G375C	350
Molecular Tests	FGFR3	134934	SAETHRE-CHOTZEN SYNDROME » CRANIOSYNOSTOSIS, SAETHRE-CHOTZEN SYNDROME	101400		Sequencing	510
Molecular Tests	FGFR3	134934	HYPOCHONDROPLASIA	146000		7 Mutations: I538V, N540T, N540S, N540K, K650N, K650M, K650Q	400
Molecular Tests	FGFR3	134934	HYPOCHONDROPLASIA	146000		7 Mutations: S84L, R200C, N262H, G268C, Y278C, S279C, V381E	400
Molecular Tests	FGFR3	134934	THANATOPHORIC DYSPLASIA, TYPE 1	187600		Sequencing	510
Molecular Tests	FGFR3	134934	THANATOPHORIC DYSPLASIA, TYPE 2	187601		Sequencing	510

Molecular Tests	FGFR3	134934	NONSYNDROMIC CORONAL CRANIOSYNOSTOSIS (MUENKE) » CRANIOSYNOSTOSIS, NONSYNDROMIC CORONAL CRANIOSYNOSTOSIS	602849	P250R mutation	Sequencing	360
Molecular Tests	FGG (FIBRINOGEN GAMMA)	134850	AFIBRINOGENEMIA » DYSFIBRINOGENEMIA » HYPOFIBRINOGENEMIA, CONGENITAL	202400		Sequencing	800
Molecular Tests	FH (FUMARATE HYDRATASE, FUMARASE)	136850	MULTIPLE CUTANEOUS AND UTERINE LEIOMYOMATA 1, MCUL1	150800		Sequencing	1150
Molecular Tests	FH (FUMARATE HYDRATASE, FUMARASE)	136850	HEREDITARY LEIOMYOMATOSIS AND RENAL CELL CANCER SYNDROME	605839		Sequencing	1150
Molecular Tests	FH (FUMARATE HYDRATASE, FUMARASE)	136850	FUMARASE DEFICIENCY » FUMARIC ACIDURIA	606812		Sequencing	1150
Molecular Tests	FHL1 (FOUR-AND-A-HALF LIM DOMAINS 1)	300163	SCAPULOPERONEAL MYOPATHY (X-LINKED DOMINANT)	300695		Sequencing	425
Molecular Tests	FHL1 (FOUR-AND-A-HALF LIM DOMAINS 1)	300163	MYOPATHY, WITH POSTURAL MUSCLE ATROPHY (X-LINKED) » EMERY-DREIFUSS MUSCULAR DYSTROPHY, TYPE 6 (X-LINKED)	300696		Sequencing	425
Molecular Tests	FHL1 (FOUR-AND-A-HALF LIM DOMAINS 1)	300163	MYOPATHY, REDUCING BODY, EARLY-ONSET, SEVERE (X-LINKED)	300717		Sequencing	425
Molecular Tests	FHL1 (FOUR-AND-A-HALF LIM DOMAINS 1)	300163	MYOPATHY, REDUCING BODY, CHILDHOOD-ONSET (X-LINKED)	300718		Sequencing	425
Molecular Tests	FKBP10 (FK506-BINDING PROTEIN 10; FKBP65)	607063	OSTEOGENESIS IMPERFECTA TYPE 6, OI6 » OSTEOGENESIS IMPERFECTA, TYPE IX, WITH OR WITHOUT JOINT CONTRACTURES	610968		Sequencing	550
Molecular Tests	FKBP (FUKUTIN-RELATED PROTEIN)	606596	MUSCLE AB INITIO DYSTROPHY, CONGENITAL, TYPE 5F, MDC5F	613153		Sequencing	720
Molecular Tests	FKTN (FUKUTIN; FCMD)	607440	WALKER-WARBURG SYNDROME » HYDROCEPHALUS, AGYRIA, AND RETINAL DYSPLASIA » HARD SYNDROME » MUSCULAR DYSTROPHY	236670		Sequencing	890
Molecular Tests	FKTN (FUKUTIN; FCMD)	607440	FUKUYAMA CONGENITAL MUSCULAR DYSTROPHY » MUSCULAR DYSTROPHY, CONGENITAL PROGRESSIVE, WITH MENTAL RETARDATION » MICROPOLYGYRIA WITH MUSCULAR DYSTROPHY » CEREBROOCULAR DYSPLASIA-MUSCULAR DYSTROPHY SYNDROME	253800		Sequencing	890
Molecular Tests	FKTN (FUKUTIN; FCMD)	607440	MUSCULAR DYSTROPHY, LIMB-GIRDLE, TYPE 2M, LGMD2M	611588		Sequencing	890
Molecular Tests	FKTN (FUKUTIN; FCMD)	607440	CARDIOMYOPATHY, DILATED, TYPE 1X	611615		Sequencing	890
Molecular Tests	FLCN (FOLLICULIN, FLCL)	607273	BIRT-HOGG-DUBE SYNDROME, BHD » FIBROFOLLICULOMAS WITH TRICHODISCOMAS AND ACROCHORDONS	135150		Sequencing	1120
Molecular Tests	FLCN (FOLLICULIN, FLCL)	607273	PNEUMOTHORAX, PRIMARY SPONTANEOUS	173600		Sequencing	1120
Molecular Tests	FLG (FILAGGRIN, PROFILAGGRIN)	135940	ATOPIC DERMATITIS			7 Mutations: p.R501X, c.2282del4, c.3702delG, p.E2422X, c.7267delCA, c.8534TG and c.8535TA	800
Molecular Tests	FLG (FILAGGRIN, PROFILAGGRIN)	135940	ASTHMA			7 Mutations: p.R501X, c.2282del4, c.3702delG, p.E2422X, c.7267delCA, c.8534TG and c.8535TA	800
Molecular Tests	FLG (FILAGGRIN, PROFILAGGRIN)	135940	ICHTHYOSIS VULGARIS » ICHTHYOSIS SIMPLEX	146700		7 Mutations: p.R501X, c.2282del4, c.3702delG, p.E2422X, c.7267delCA, c.8534TG and c.8535TA	800
Molecular Tests	FLNA (FILAMIN A)	300017	HETEROTOPIA, PERIVENTRICULAR (X-LINKED DOMINANT) » PERIVENTRICULAR NODULAR HETEROTOPIA 1 » NODULAR HETEROTOPIA » BILATERAL PERIVENTRICULAR HETEROTOPIA	300049		Sequencing	1100
Molecular Tests	FLNA (FILAMIN A)	300017	OTOPALATODIGITAL SYNDROME, TYPE 2, OPD2 » CRANIOFRONTODIGITAL SYNDROME » OTOPALATODIGITAL SYNDROME, TYPE 1	304120		Exons 3, 5, 11, 22, 29, 45	1200
Molecular Tests	FLNB (FILAMIN B)	603381	ATELOSTEOGENESIS, TYPE 1 » GIANT CELL CHONDRODYSPLASIA	108720		Sequencing	1900
Molecular Tests	FLNB (FILAMIN B)	603381	ATELOSTEOGENESIS, TYPE 3	108721		Sequencing	1900
Molecular Tests	FLNB (FILAMIN B)	603381	BOOMERANG DYSPLASIA	112310		Sequencing	1900
Molecular Tests	FLNB (FILAMIN B)	603381	LARSEN SYNDROME (AUTOSOMAL DOMINANT)	150250		Sequencing	1900
Molecular Tests	FLNB (FILAMIN B)	603381	SPONDYLOCARPOTARSAL SYNOSTOSIS SYNDROME » SYNDROMIC IOA, CONGENITAL	272460		Sequencing	1900

Molecular Tests	FLT4 (FMS-LIKE TYROSINE KINASE 4, VASCULAR ENDOTHELIAL GROWTH FACTOR RECEPTOR 3, VEGFR3)	136355	LYMPHEDEMA, HEREDITARY, TYPE 1 » NONNE-MILROY LYMPHEDEMA » MILROY DISEASE » [illegible]	153100		10 Exons: Exons 17-26, representing 90% of reported mutations	842
Molecular Tests	FLVCR2 (FELINE LEUKEMIA VIRUS SUBGROUP C RECEPTOR 2, CALCIUM CHELATE TRANSPORTER, FLVCR-LIKE ON CHROMOSOME 14q4	609865	PROLIFERATIVE VASCULOPATHY AND HYDRANENCEPHALY-HYDROCEPHALY SYNDROME » ENCEPHALOCLASTIC PROLIFERATIVE VASCULOPATHY	225790		Sequencing	700
Molecular Tests	FMO3 (FLAVIN - CONTAINING MONOOXYGENASE 3)	136132	FISH-ODOR SYNDROME » TRIMETHYLAMINURIA	602079		Sequencing	690
Molecular Tests	FMR1	309550	FRAGILE X SYNDROME, FRAXA	300624		Sequencing	1050
Molecular Tests	FMR1	309550	FRAGILE X SYNDROME, FRAXA	309550	At least 20 microgram DNA with a concentration higher than 300 nanogram	Repeat	600
Molecular Tests	FMR1	309550	FRAGILE X SYNDROME, FRAXE	309548		Repeat	250
Molecular Tests	FOXC1 (FORKHEAD BOX C1, FORKHEAD, DROSOPHILA, HOMOLOG-LIKE 7, FKHL7 FORKHEAD-RELATED ACTIVATOR 3, BREAC3)	601090	ANTERIOR SEGMENT MESENCHYMAL DYSGENESIS			Sequencing	710
Molecular Tests	FOXC1 (FORKHEAD BOX C1, FORKHEAD, DROSOPHILA, HOMOLOG-LIKE 7, FKHL7 FORKHEAD-RELATED ACTIVATOR 3, BREAC3)	601090	ANTERIOR SEGMENT MESENCHYMAL DYSGENESIS			Deletion-Duplication Testing	680
Molecular Tests	FOXC1 (FORKHEAD BOX C1, FORKHEAD, DROSOPHILA, HOMOLOG-LIKE 7, FKHL7 FORKHEAD-RELATED ACTIVATOR 3, BREAC3)	601090	IRIDOGONIODYSGENESIS SYNDROME, TYPE 2, IRID2 » IRIS HYPOPLASIA WITH FAR VISION OF GLAUCOMA (AUTOSOMAL DOMINANT), IHGA	137600		Sequencing	710
Molecular Tests	FOXC1 (FORKHEAD BOX C1, FORKHEAD, DROSOPHILA, HOMOLOG-LIKE 7, FKHL7 FORKHEAD-RELATED ACTIVATOR 3, BREAC3)	601090	IRIDOGONIODYSGENESIS SYNDROME, TYPE 2, IRID2 » IRIS HYPOPLASIA WITH FAR VISION OF GLAUCOMA (AUTOSOMAL DOMINANT), IHGA	137600		Deletion-Duplication Testing	680
Molecular Tests	FOXC1 (FORKHEAD BOX C1, FORKHEAD, DROSOPHILA, HOMOLOG-LIKE 7, FKHL7 FORKHEAD-RELATED ACTIVATOR 3, BREAC3)	601090	RIEGER SYNDROME, TYPE 1, RIEG1	180500		Sequencing	710
Molecular Tests	FOXC1 (FORKHEAD BOX C1, FORKHEAD, DROSOPHILA, HOMOLOG-LIKE 7, FKHL7 FORKHEAD-RELATED ACTIVATOR 3, BREAC3)	601090	RIEGER SYNDROME, TYPE 1, RIEG1	180500		Deletion-Duplication Testing	680
Molecular Tests	FOXC1 (FORKHEAD BOX C1, FORKHEAD, DROSOPHILA, HOMOLOG-LIKE 7, FKHL7 FORKHEAD-RELATED ACTIVATOR 3, BREAC3)	601090	PETERS ANOMALY	604229		Sequencing	710
Molecular Tests	FOXC1 (FORKHEAD BOX C1, FORKHEAD, DROSOPHILA, HOMOLOG-LIKE 7, FKHL7 FORKHEAD-RELATED ACTIVATOR 3, BREAC3)	601090	PETERS ANOMALY	604229		Deletion-Duplication Testing	680
Molecular Tests	FOXC2 (FORKHEAD BOX C2)	601403	LYMPHEDEMA, HEREDITARY, TYPE 2 » MEIGE LYMPHEDEMA	153400		Sequencing	570
Molecular Tests	FOXE1 (FORKHEAD BOX E1, THYROID TRANSCRIPTION FACTOR 2, TTF2, TITF2)	602617	BAMFORTH-LAZARUS SYNDROME » HYPOTHYROIDISM, ATHYROIDAL, WITH SPIKY HAIR AND CLEFT PALATE	241850		Sequencing	760
Molecular Tests	FOXG1 (FORKHEAD BOX G1, BRAIN FACTOR 1)	164874	RETT SYNDROME, CONGENITAL VARIANT	164874		Sequencing	490
Molecular Tests	FOXI3	605197	BLEPHAROPHIMOSIS, PTOSIS, AND EPICANTHUS INVERSUS, BPES	110100		Sequencing and MLPA	650
Molecular Tests	FOXL2	605597	BLEPHAROPHIMOSIS, PTOSIS, AND EPICANTHUS INVERSUS, BPES	110100		Sequencing	550
Molecular Tests	FOXP2 (FORKHEAD BOX P2, TRINUCLEOTIDE REPEAT -CONTAINING GENE 10)	605317	SPEECH-LANGUAGE DISORDER, TYPE 1 » DEVELOPMENTAL VERBAL DYSPRAXIA » [illegible]	602081		Sequencing	1700
Molecular Tests	FOXP3 (FORKHEAD BOX P3, SCURFIN)	300292	IMMUNODYSREGULATION, POLYENDOCRINOPATHY, AND ENTEROPATHY (X-LINKED), IPEX » X-LINKED AUTOIMMUNITY-ALLERGIC DYSREGULATION SYNDROME » IDDM--SECRETORY DIARRHEA SYNDROME » AUTOIMMUNITY-IMMUNODEFICIENCY SYNDROME » DIARRHEA, POLYENDOCRINOPATHY, FATAL INFECTION SYNDROME » ENTEROPATHY, AUTOIMMUNE, WITH HEMOLYTIC ANEMIA AND POLYENDOCRINOPATHY » POLYENDOCRINOPATHY, IMMUNE DYSFUNCTION, AND DIARRHEA » DIABETES MELLITUS, CONGENITAL INSULIN-DEPENDENT, WITH FATAL SECRETORY DIARRHEA	304790		Sequencing	745
Molecular Tests	FOXRED1 (FAD-DEPENDENT OXIDOREDUCTASE DOMAIN-CONTAINING PROTEIN 1)	613622	CYTOCHROME c OXIDASE DEFICIENCY » COX DEFICIENCY » [illegible]	252010		Sequencing	1060
Molecular Tests	FOXRED1 (FAD-DEPENDENT OXIDOREDUCTASE DOMAIN-CONTAINING PROTEIN 1)	613622	LEIGH SYNDROME DUE TO MITOCHONDRIAL COMPLEX 1 DEFICIENCY	256000		Sequencing	1060
Molecular Tests	FRAXF (FRAGILE SITE F)	300031	FRAGILE F SYNDROME	300031		Sequencing	180
Molecular Tests	FRDA (FRATAXIN1)	606829	FRIEDREICH ATAXIA, FRDA	229300		Sequencing	450
Molecular Tests	FRDA (FRATAXIN1)	606829	FRIEDREICH ATAXIA, FRDA	229300		Repeat	150

Molecular Tests	FREM2 (FRAS1-RELATED EXTRACELLULAR MATRIX PROTEIN 2)	608945	FRASER SYNDROME » CRYPTOPHTHALMOS WITH OTHER MALFORMATIONS	219000		Exon 6	250
Molecular Tests	FSCN2 (FASCIN, SEA URCHIN, HOMOLOG OF, 2; FASCIN, RETINAL; RFSN)	607643	RETINITIS PIGMENTOSA, TYPE 30, RP30	607921		Sequencing	530
Molecular Tests	FSHD	158900	FACIOSCAPULOHUMERAL MUSCULAR DYSTROPHY, FSHD » LANDOUZY-DEJERINE MUSCULAR DYSTROPHY » MUSCULAR DYSTROPHY, FACIOSCAPULOHUMERAL MUSCULAR DYSTROPHY, FSHD	158900	At least 20ml EDTA Blood	Repeat	950
Molecular Tests	FSHR (FOLLICLE-STIMULATING HORMONE RECEPTOR; FSH RECEPTOR; OVARIAN HYPERSTIMULATION SYNDROME, MODERATOR OF SEVERITY OF)	136435	OVARIAN DYSGENESIS TYPE 1, ODG1 » GONADAL DYSGENESIS, XX TYPE	233300		Sequencing	800
Molecular Tests	FTCD (FORMIMINOTRANSFERASE CYCLODEAMINASE)	606806	FORMIMINOTRANSFERASE DEFICIENCY » GLUTAMATE FORMIMINOTRANSFERASE DEFICIENCY » FORMIMINOGLUTAMIC ACIDURIA	229100		Sequencing	1390
Molecular Tests	FTL (FERRITIN LIGHT CHAIN)	134790	HYPERFERRITINEMIA-CATARACT SYNDROME	600886		Sequencing	650
Molecular Tests	FTL (FERRITIN LIGHT CHAIN)	134790	HYPERFERRITINEMIA-CATARACT SYNDROME	600886		Iron-responsive Element (IRE)	350
Molecular Tests	FTL (FERRITIN LIGHT CHAIN)	134790	BASAL GANGLIA DISEASE, ADULT-ONSET » NEUROFERRITINOPATHY	606159		Sequencing	650
Molecular Tests	FTL (FERRITIN LIGHT CHAIN)	134790	BASAL GANGLIA DISEASE, ADULT-ONSET » NEUROFERRITINOPATHY	606159		Iron-responsive Element (IRE)	350
Molecular Tests	FTSJ1 (FTSJ HOMOLOG 1)	300499	MENTAL RETARDATION, NONSPECIFIC (X-LINKED), TYPE 44, MRX44	300501		Sequencing	750
Molecular Tests	FTSJ1 (FTSJ HOMOLOG 1)	300499	MENTAL RETARDATION, NONSPECIFIC (X-LINKED), TYPE 9, MRX9	309549		Sequencing	750
Molecular Tests	FUCA1 (ALPHA-L-FUCOSIDASE)	230000	FUCOSIDOSIS » ALPHA-L-FUCOSIDASE DEFICIENCY	230000		Sequencing	850
Molecular Tests	FUS (FUSION, DERIVED FROM 12-16 TRANSLOCATION, MALIGNANT LIPOSARCOMA	137070	AMYOTROPHIC LATERAL SCLEROSIS, TYPE 6, ALS6	608030		Sequencing	1000
Molecular Tests	FXYD2 (FXYD DOMAIN-CONTAINING ION TRANSPORT REGULATOR 2, SODIUM-POTASSIUM-ATPase, GAMMA-1 POLYPEPTIDE)	601814	HYPOMAGNESEMIA, RENAL, TYPE 2	154020		Sequencing	375
Molecular Tests	FY (DUFFY BLOOD GROUP SYSTEM)	110700	HEMOLYTIC DISEASE, NEWBORN			1 Mutation: G44D	160
Molecular Tests	FZD4 (FRIZZLED, DROSOPHILA, HOMOLOG OF, 4)	604579	EXUDATIVE VITREORETINOPATHY, TYPE 1 » CRISWICK-SCHEPENS SYNDROME	133780		Sequencing	500
Molecular Tests	G6PC (GLUCOSE-6-PHOSPHATASE)	232200	GLYCOGEN STORAGE DISEASE, TYPE 1A » GSD TYPE 1A	232200		Sequencing	700
Molecular Tests	G6PD (GLUCOSE-6-PHOSPHATE DEHYDROGENASE)	305900	G6PD DEFICIENCY » GLUCOSE-6-PHOSPHATE DEHYDROGENASE DEFICIENCY	305900		Sequencing	690
Molecular Tests	GAA (GLUCOSIDASE, ALPHA, ACID, ACID MALTASE)	606800	GLYCOGEN STORAGE DISEASE, TYPE 2 » GSD TYPE 2 » ACID ALPHA-GLUCOSIDASE DEFICIENCY » POMPE DISEASE » GLYCOGENOSIS, GENERALIZED, CARDIAC FORM » CARDIOMEGALIA GLYCOGENICA DIFFUSA » ACID MALTASE DEFICIENCY	232300		Sequencing	1050
Molecular Tests	GABRA1 (GAMMA-AMINOBUTYRIC ACID RECEPTOR, ALPHA-1; GABA-A RECEPTOR, ALPHA-1 POLYPEPTIDE)	137160	EPILEPSY, JUVENILE MYOCLONIC, JME » JANZ SYNDROME	606904		Sequencing	1000
Molecular Tests	GABRA1 (GAMMA-AMINOBUTYRIC ACID RECEPTOR, ALPHA-1; GABA-A RECEPTOR, ALPHA-1 POLYPEPTIDE)	137160	EPILEPSY, CHILDHOOD ABSENCE, TYPE 4, ECA4	611136		Sequencing	1000
Molecular Tests	GABRD (GAMMA-AMINOBUTYRIC ACID RECEPTOR, DELTA;GABA-A RECEPTOR, DELTA POLYPEPTIDE)	137163	GENERALIZED EPILEPSY WITH FEBRILE SEIZURES PLUS, GEFS+ » GENERALIZED EPILEPSY WITH FEBRILE SEIZURES PLUS, TYPE 2, GEFS+, TYPE 2	604233		Sequencing	800
Molecular Tests	GABRG2 (GAMMA-AMINOBUTYRIC ACID RECEPTOR, GAMMA-2)	137164	GENERALIZED EPILEPSY WITH FEBRILE SEIZURES PLUS, GEFS+ » GENERALIZED EPILEPSY WITH FEBRILE SEIZURES PLUS, TYPE 2, GEFS+, TYPE 2	604233		Sequencing	800
Molecular Tests	GABRG2 (GAMMA-AMINOBUTYRIC ACID RECEPTOR, GAMMA-2)	137164	EPILEPSY, CHILDHOOD ABSENCE, TYPE 2, ECA2	607681		Sequencing	800
Molecular Tests	GAD1 (GLUTAMATE DECARBOXYLASE 1)	605363	CEREBRAL PALSY, SPASTIC QUADRIPLEGIC, TYPE 1	603513		Sequencing	1100

Molecular Tests	GALC (GALACTOSYL CERAMIDASE, GALACTOCEREBROSIDASE)	606890	KRABBE DISEASE » GLOBOID CELL LEUKODYSTROPHY » GALACTOSYLCERAMIDE BETA-GALACTOSIDASE DEFICIENCY	245200		Sequencing	790
Molecular Tests	GALE (UDP-GALACTOSE-4-EPIMERASE; GALACTOSE EPIMERASE)	606953	GALACTOSE EPIMERASE DEFICIENCY » GALE DEFICIENCY » GALACTOSEMIA, TYPE 3	230350		Sequencing	1040
Molecular Tests	GALK1 (GALACTOKINASE 1)	604313	GALACTOKINASE DEFICIENCY » GALK DEFICIENCY » GALACTOSEMIA TYPE 2	230200		Sequencing	650
Molecular Tests	GALNS (GALACTOSAMINE-6-SULFATE SULFATASE; N-ACETYLGALACTOSAMINE-SULFATE SULFATASE)	612222	MUCOPOLYSACCHARIDOSIS, TYPE 4A, MPS4A » MORQUIO SYNDROME A	253000		Sequencing	1200
Molecular Tests	GALNS (GALACTOSAMINE-6-SULFATE SULFATASE; N-ACETYLGALACTOSAMINE-SULFATE SULFATASE)	612222	MUCOPOLYSACCHARIDOSIS, TYPE 4A, MPS4A » MORQUIO SYNDROME A	253000		Deletion-Duplication Testing	600
Molecular Tests	GALNT3 (UDP-N-ACETYL-ALPHA-D-GALACTOSAMINE:POLYPEPTIDE N-ACETYLGALACTOSAMINYLTRANSFERASE 3; GalNAc TRANSFERASE 3; POLYPEPTIDE N-ACETYLGALACTOSAMINYLTRANSFERASE 3)	601756	TUMORAL CALCINOSIS, HYPERPHOSPHATEMIC, FAMILIAL » LIPOCALCINOGRANULOMATOSIS » TEUTSCHLAENDER DISEASE, FAMILIAL	211900		Sequencing	745
Molecular Tests	GALT (GALACTOSE-1-PHOSPHATE URIDYLYLTRANSFERASE)	606999	GALACTOSEMIA TYPE 1 » GALACTOSE-1-PHOSPHATE URIDYLYLTRANSFERASE DEFICIENCY	230400		Sequencing	1240
Molecular Tests	GAMT (GUANIDINOACETATE METHYLTRANSFERASE)	601240	GUANIDINOACETATE METHYLTRANSFERASE DEFICIENCY » GAMT DEFICIENCY » CREATINE DEFICIENCY SYNDROME DUE TO GAMT DEFICIENCY	601240		Sequencing	890
Molecular Tests	GAN (GAN GENE, GIGAXONIN)	605379	GIANT AXONAL NEUROPATHY 1 » NEUROPATHY, GIANT AXONAL (AUTOSOMAL RECESSIVE)	256850		Sequencing	850
Molecular Tests	GARS (GLYCYL T RNA SYNTHETASE)	600287	SPINAL MUSCULAR ATROPHY, DISTAL, TYPE 5, DSMA5	600794		Sequencing	1500
Molecular Tests	GARS (GLYCYL T RNA SYNTHETASE)	600287	CHARCOT-MARIE-TOOTH DISEASE, AXONAL, TYPE 2D, CMT2D	601472		Sequencing	1500
Molecular Tests	GATA1 (GATA-BINDING PROTEIN 1; ERYTHROID TRANSCRIPTION FACTOR 1, GLOBIN TRANSCRIPTION FACTOR 1)	305371	DYSERYTHROPOIETIC ANEMIA WITH THROMBOCYTOPENIA » MACROTHROMBOCYTOPENIA (X-LINKED)	300637		Sequencing	1420
Molecular Tests	GATA1 (GATA-BINDING PROTEIN 1; ERYTHROID TRANSCRIPTION FACTOR 1, GLOBIN TRANSCRIPTION FACTOR 1)	305371	THROMBOCYTOPENIA, PLATELET DYSFUNCTION, HEMOLYSIS, AND IMBALANCED GLOBIN SYNTHESIS » THROMBOCYTOPENIA WITH BETA-THALASSEMIA (X-LINKED)	314050		Sequencing	1420
Molecular Tests	GATA3 (GATA-BINDING PROTEIN 3)	131320	BARAKAT SYNDROME » HYPOPARATHYROIDISM, SENSORINEURAL DEAFNESS, AND RENAL DISEASE » HDR SYNDROME	146255		Sequencing	450
Molecular Tests	GATA4 (GATA-BINDING PROTEIN 4)	600576	ATRIAL SEPTAL DEFECT, TYPE 2	607941		Sequencing	1250
Molecular Tests	GATM (L-ARGININE:GLYCINE AMIDINOTRANSFERASE)	602360	ARGININE:GLYCINE AMIDINOTRANSFERASE DEFICIENCY » CREATINE DEFICIENCY SYNDROME DUE TO AGAT DEFICIENCY » AGAT DEFICIENCY	612718		Sequencing	1020
Molecular Tests	GBA (GLUCOSIDASE, GLUCOCEREBROSIDASE)	606463	GAUCHER DISEASE, TYPE 1	230800		Sequencing	1090
Molecular Tests	GBA (GLUCOSIDASE, GLUCOCEREBROSIDASE)	606463	GAUCHER DISEASE, TYPE 1	230800		6 Mutations : 84GG, IVS2+1, N370S, 1297T, L444P, V394I	150
Molecular Tests	GBA (GLUCOSIDASE, GLUCOCEREBROSIDASE)	606463	GAUCHER DISEASE, TYPE 2	230900		Sequencing	1090
Molecular Tests	GBA (GLUCOSIDASE, GLUCOCEREBROSIDASE)	606463	GAUCHER DISEASE, TYPE 2	230900		6 Mutations : 84GG, IVS2+1, N370S, 1297T, L444P, V394I	150
Molecular Tests	GBA (GLUCOSIDASE, GLUCOCEREBROSIDASE)	606463	GAUCHER DISEASE, TYPE 3	231000		Sequencing	1090
Molecular Tests	GBA (GLUCOSIDASE, GLUCOCEREBROSIDASE)	606463	GAUCHER DISEASE, TYPE 3	231000		6 Mutations : 84GG, IVS2+1, N370S, 1297T, L444P, V394I	150

Molecular Tests	GBE1 (GLYCOGEN BRANCHING ENZYME)	607839	GLYCOGEN STORAGE DISEASE, TYPE 4 » GSD TYPE 4 » GLYCOGEN BRANCHING ENZYME DEFICIENCY » GBE1 DEFICIENCY » ANDERSEN DISEASE » BRANCHER DEFICIENCY » GLYCOGENOSIS 4	232500		Whole Gene	1070
Molecular Tests	GBE1 (GLYCOGEN BRANCHING ENZYME)	607839	GLYCOGEN STORAGE DISEASE, TYPE 4 » GSD TYPE 4 » GLYCOGEN BRANCHING ENZYME DEFICIENCY » GBE1 DEFICIENCY » ANDERSEN DISEASE » BRANCHER DEFICIENCY » GLYCOGENOSIS 4	232500		Deletion-Duplication Testing	600
Molecular Tests	GCDH (GLUTARYL COA DEHYDROGENASE)	231670	GLUTARIC ACIDURIA, TYPE 1	231670		Sequencing	690
Molecular Tests	GCH1 (GTP CYCLOHYDROLASE 1)	600225	DYSTONIA, PROGRESSIVE, WITH DIURNAL VARIATION » DYSTONIA-PARKINSONISM WITH DIURNAL FLUCTUATION » DYSTONIA 5, DYT5 » SEGAWA SYNDROME (AUTOSOMAL DOMINANT)	128230		Sequencing	590
Molecular Tests	GCH1 (GTP CYCLOHYDROLASE 1)	600225	HYOCLONIC DYSTONIA » MYOCLONUS-DYSTONIA SYNDROME » DYSTONIA, ALCOHOL-RESPONSIVE	159900		Sequencing	590
Molecular Tests	GCH1 (GTP CYCLOHYDROLASE 1)	600225	GTP CYCLOHYDROLASE 1 DEFICIENCY » GCH DEFICIENCY » HYPERPHENYLALANINEMIA WITH NEOPTERIN DEFICIENCY	233910		Sequencing	590
Molecular Tests	GCK (GLUCOKINASE)	138079	MATURITY-ONSET DIABETES OF THE YOUNG, TYPE 2, MODY2	125851		Sequencing	650
Molecular Tests	GCK (GLUCOKINASE)	138079	MATURITY-ONSET DIABETES OF THE YOUNG, TYPE 2, MODY2	125851		Deletion-Duplication Testing	Upon Request
Molecular Tests	GCK (GLUCOKINASE)	138079	HYPERINSULINEMIC HYPOGLYCEMIA, FAMILIAL, TYPE 3	602485		Sequencing	650
Molecular Tests	GCK (GLUCOKINASE)	138079	HYPERINSULINEMIC HYPOGLYCEMIA, FAMILIAL, TYPE 3	602485		Deletion-Duplication Testing	Upon Request
Molecular Tests	GCMD (GLIAL CELLS MISSING, DROSOPHILA, HOMOLOG OF, 2; GCMB)	603756	HYPOPARATHYROIDISM, FAMILIAL ISOLATED » HYPOPARATHYROIDISM (AUTOSOMAL DOMINANT) » HYPOCALCEMIA (AUTOSOMAL DOMINANT)	146300		Sequencing	680
Molecular Tests	GCS1 (GLUCOSIDASE 1)	606336	CONGENITAL DISORDER OF GLYCOSYLATION, TYPE 2B, CDG2B » GLUCOSIDASE 1 DEFICIENCY	606056		Sequencing	810
Molecular Tests	GCSH (GLYCINE CLEAVAGE SYSTEM H PROTEIN)	238330	NONKETOTIC HYPERGLYCINEMIA » GLYCINE ENCEPHALOPATHY	605899		Sequencing	470
Molecular Tests	GDAP1 (GANGLIOSIDE-INDUCED DIFFERENTIATION-ASSOCIATED PROTEIN 1)	606598	CHARCOT-MARIE-TOOTH DISEASE, DEMYELINATING, TYPE 4A, CMT4A (AUTOSOMAL RECESSIVE)	214400		Sequencing	800
Molecular Tests	GDAP1 (GANGLIOSIDE-INDUCED DIFFERENTIATION-ASSOCIATED PROTEIN 1)	606598	CHARCOT-MARIE-TOOTH DISEASE, AXONAL, WITH VOCAL CORD PARESIS (AUTOSOMAL RECESSIVE)	607706		Sequencing	800
Molecular Tests	GDAP1 (GANGLIOSIDE-INDUCED DIFFERENTIATION-ASSOCIATED PROTEIN 1)	606598	CHARCOT-MARIE-TOOTH DISEASE, AXONAL, TYPE 2K, CMT2K	607831		Sequencing	800
Molecular Tests	GDAP1 (GANGLIOSIDE-INDUCED DIFFERENTIATION-ASSOCIATED PROTEIN 1)	606598	CHARCOT-MARIE-TOOTH DISEASE, INTERMEDIATE A, CMTIA (AUTOSOMAL RECESSIVE)	608340		Sequencing	800
Molecular Tests	GDF5 (GROWTH / DIFFERENTIATION FACTOR 5, CDMP1, LAP4)	601146	BRACHYDACTYLY, TYPE C	113100		Sequencing	900
Molecular Tests	GDF5 (GROWTH / DIFFERENTIATION FACTOR 5, CDMP1, LAP4)	601146	CHONDRODYSPLASIA, GREBE TYPE » ACHONDROGENESIS, BRAZILIAN TYPE	200700		Sequencing	900
Molecular Tests	GDF5 (GROWTH / DIFFERENTIATION FACTOR 5, CDMP1, LAP4)	601146	ACROMESOMELIC DYSPLASIA, HUNTER-THOMPSON TYPE	201250		Sequencing	900
Molecular Tests	GDF5 (GROWTH / DIFFERENTIATION FACTOR 5, CDMP1, LAP4)	601146	FIBULAR HYPOPLASIA AND COMPLEX BRACHYDACTYLY » TIBIAN SYNDROME	228900		Sequencing	900
Molecular Tests	GFAP (GLIAL FIBRILLARY ACIDIC PROTEIN)	137780	ALEXANDER DISEASE	203450		Sequencing	650
Molecular Tests	GGCX	137167	VITAMIN K-DEPENDENT CLOTTING FACTORS, COMBINED DEFICIENCY OF, 1	277450		Sequencing	550

Molecular Tests	GH1 (GROWTH HORMONE 1, GH)	139250	PITUITARY DWARFISM DUE TO ISOLATED GROWTH HORMONE DEFICIENCY (AUTOSOMAL DOMINANT) » GROWTH HORMONE DEFICIENCY, ISOLATED (AUTOSOMAL DOMINANT) » ISOLATED GROWTH HORMONE DEFICIENCY, TYPE 2	173100		Sequencing	400
Molecular Tests	GH1 (GROWTH HORMONE 1, GH)	139250	PITUITARY DWARFISM DUE TO ISOLATED GROWTH HORMONE DEFICIENCY (AUTOSOMAL DOMINANT) » GROWTH HORMONE DEFICIENCY, ISOLATED (AUTOSOMAL DOMINANT) » ISOLATED GROWTH HORMONE DEFICIENCY, TYPE 2	173100		Deletion-Duplication Testing	350
Molecular Tests	GH1 (GROWTH HORMONE 1, GH)	139250	PITUITARY DWARFISM 1 » PRIMORDIAL DWARFISM » SEXUAL ATELEIOTIC DWARFISM » GROWTH HORMONE DEFICIENCY, ISOLATED » AUTOSOMAL RECESSIVE ISOLATED GROWTH HORMONE DEFICIENCY, TYPE 1A / 1B	262400		Sequencing	400
Molecular Tests	GH1 (GROWTH HORMONE 1, GH)	139250	PITUITARY DWARFISM 1 » PRIMORDIAL DWARFISM » SEXUAL ATELEIOTIC DWARFISM » GROWTH HORMONE DEFICIENCY, ISOLATED » AUTOSOMAL RECESSIVE ISOLATED GROWTH HORMONE DEFICIENCY, TYPE 1A / 1B	262400		Deletion-Duplication Testing	350
Molecular Tests	GH1 (GROWTH HORMONE 1, GH)	139250	PITUITARY DWARFISM 4 » PITUITARY DWARFISM WITH NORMAL IMMUNOREACTIVE GROWTH HORMONE AND LOW SOMATOMEDIN » BIODEFECTIVE GROWTH HORMONE » KOWARSKI SYNDROME	262650		Sequencing	400
Molecular Tests	GH1 (GROWTH HORMONE 1, GH)	139250	PITUITARY DWARFISM 4 » PITUITARY DWARFISM WITH NORMAL IMMUNOREACTIVE GROWTH HORMONE AND LOW SOMATOMEDIN » BIODEFECTIVE GROWTH HORMONE » KOWARSKI SYNDROME	262650		Deletion-Duplication Testing	350
Molecular Tests	GH1 (GROWTH HORMONE 1, GH)	139250	SHORT STATURE, IDIOPATHIC (AUTOSOMAL)	604271		Sequencing	400
Molecular Tests	GH1 (GROWTH HORMONE 1, GH)	139250	SHORT STATURE, IDIOPATHIC (AUTOSOMAL)	604271		Deletion-Duplication Testing	350
Molecular Tests	GHR (GROWTH HORMONE RECEPTOR, GROWTH HORMONE-BINDING PROTEIN, GHBP)	600946	PITUITARY DWARFISM 2 » GROWTH HORMONE INSENSITIVITY SYNDROME » LARON SYNDROME » GROWTH HORMONE RECEPTOR DEFICIENCY	262500		Sequencing	1230
Molecular Tests	GHR (GROWTH HORMONE RECEPTOR, GROWTH HORMONE-BINDING PROTEIN, GHBP)	600946	SHORT STATURE, IDIOPATHIC (AUTOSOMAL)	604271		Sequencing	1230
Molecular Tests	GJA1 (GAP JUNCTION PROTEIN, ALPHA-1, CONNEXIN 43, CX43)	121014	OCULODENTODIGITAL DYSPLASIA » OCULODENTOOSSEOUS DYSPLASIA	164200		Sequencing	450
Molecular Tests	GJA1 (GAP JUNCTION PROTEIN, ALPHA-1, CONNEXIN 43, CX43)	121014	SYNDACTYLY, TYPE 3 » SYNDACTYLY OF FINGERS 4 AND 5	186100		Sequencing	450
Molecular Tests	GJA1 (GAP JUNCTION PROTEIN, ALPHA-1, CONNEXIN 43, CX43)	121014	HYPOPLASTIC LEFT HEART SYNDROME	241550		Sequencing	450
Molecular Tests	GJB1 (CONNEXIN 32, CX32)	304040	CHARCOT-MARIE-TOOTH PERONEAL MUSCULAR ATROPHY (X-LINKED), CMTX » HEREDITARY MOTOR AND SENSORY NEUROPATHY (X-LINKED)	302800		Sequencing	250
Molecular Tests	GJB1 (CONNEXIN 32, CX32)	304040	CHARCOT-MARIE-TOOTH PERONEAL MUSCULAR ATROPHY (X-LINKED), CMTX » HEREDITARY MOTOR AND SENSORY NEUROPATHY (X-LINKED)	302800		Deletion-Duplication Testing	350
Molecular Tests	GJB2 (CONNEXIN 26, CX26)	121011	VOHWINKEL SYNDROME	124500		Sequencing	250
Molecular Tests	GJB2 (CONNEXIN 26, CX26)	121011	KERATITIS-ICHTHYOSIS-DEAFNESS	148210		Sequencing	250
Molecular Tests	GJB2 (CONNEXIN 26, CX26)	121011	PALMOPLANTAR KERATODERMA WITH DEAFNESS » HYPERKERATOSIS, PALMOPLANTAR KERATODERMA WITH DEAFNESS	148350		Sequencing	250
Molecular Tests	GJB2 (CONNEXIN 26, CX26)	121011	DEAFNESS, DFNB1 » DEAFNESS (AUTOSOMAL RECESSIVE), NEUROSENSORY 1	220290		Sequencing	250
Molecular Tests	GJB2 (CONNEXIN 26, CX26)	121011	DEAFNESS, DFNA3 » DEAFNESS (AUTOSOMAL DOMINANT), NONSYNDROMIC SENSORINEURAL 3	601544		Sequencing	250
Molecular Tests	GJB2, GJB3, GJB6, POU3F4 and WFS1		DEAFNESS			Deletion-Duplication Testing GJB2, GJB3, GJB6, POU3F4 and WFS1 gene: also detects the GJB2 mutations c.35delG, c.313del14, c.235delC, c.167delT, IVS1+1G> A and c.101T> C	400
Molecular Tests	GJB3 (CONNEXIN 31, CX31)	603324	ERYTHROKERATODERMIA VARIABILIS, EKV	133200	Sequencing GJB3 and GJB4	Sequencing	940

Molecular Tests	GJB3 (CONNEXIN 31, CX31)	603324	DEAFNESS, DFNA2 » DEAFNESS (AUTOSOMAL DOMINANT) NONSYNDROMIC SENSORINEURAL 2	600101	Sequencing GJB3 and GJB4	Sequencing	940
Molecular Tests	GJB4 (CONNEXIN 30.3, CX30.3)	605425	ERYTHROKERATODERMIA VARIABILIS, EKV	133200	Sequencing GJB3 and GJB4	Sequencing	940
Molecular Tests	GJB6 (CONNEXIN 30, CX30)	604418	CLOUSTON SYNDROME » HYDROTIC ECTODERMAL DYSPLASIA, ED2 » ECTODERMAL DYSPLASIA, CLOUSTON SYNDROME	129500		Sequencing	250
Molecular Tests	GJB6 (CONNEXIN 30, CX30)	604418	DEAFNESS, DFNB1 » DEAFNESS (AUTOSOMAL RECESSIVE) NEUROSENSORY 1	220290		Sequencing	250
Molecular Tests	GJB6 (CONNEXIN 30, CX30)	604418	DEAFNESS, DFNA3 » DEAFNESS (AUTOSOMAL DOMINANT) NONSYNDROMIC SENSORINEURAL 3	601544		Sequencing	250
Molecular Tests	GJC2 (GAP JUNCTION PROTEIN, GAMMA-2; GJA12; CONNEXIN 47; CONNEXIN 46.6)	608803	PELIZAEUS-MERZBACHER-LIKE DISEASE, TYPE 1 (AUTOSOMAL RECESSIVE)	608804		Sequencing	840
Molecular Tests	GLA (GALACTOSIDASE)	301500	FABRY DISEASE	301500		Sequencing	450
Molecular Tests	GLA (GALACTOSIDASE)	301500	FABRY DISEASE	301500		Deletion-Duplication Testing	Upon Request
Molecular Tests	GLB1 (GALACTOSIDASE, BETA-1; ELASTIN-BINDING PROTEIN, ELASTIN RECEPTOR 1)	611458	GM1-GANGLIOSIDOSIS, TYPE 1 » GANGLIOSIDOSIS, GENERALIZED GM1, TYPE 1	230500		Sequencing	850
Molecular Tests	GLB1 (GALACTOSIDASE, BETA-1; ELASTIN-BINDING PROTEIN, ELASTIN RECEPTOR 1)	611458	GM1-GANGLIOSIDOSIS, TYPE 2 » GANGLIOSIDOSIS, GENERALIZED GM1, TYPE 2	230600		Sequencing	850
Molecular Tests	GLB1 (GALACTOSIDASE, BETA-1; ELASTIN-BINDING PROTEIN, ELASTIN RECEPTOR 1)	611458	GM1-GANGLIOSIDOSIS, TYPE 3 » GANGLIOSIDOSIS, GENERALIZED GM1, TYPE 3	230650		Sequencing	850
Molecular Tests	GLB1 (GALACTOSIDASE, BETA-1; ELASTIN-BINDING PROTEIN, ELASTIN RECEPTOR 1)	611458	MUCOPOLYSACCHARIDOSIS, TYPE 4B, MPS4B » MORQUIO SYNDROME B	253010		Sequencing	850
Molecular Tests	GLDC (GLYCINE DECARBOXYLASE, GLYCINE CLEAVAGE SYSTEM P PROTEIN)	238300	NONKETOTIC HYPERGLYCINEMIA » GLYCINE ENCEPHALOPATHY	605899		Sequencing	1550
Molecular Tests	GLE1 (GLE1, S. CEREVISIAE, HOMOLOG-LIKE)	603371	LETHAL ARTHROGRYPOSIS WITH ANTERIOR HORN CELL DISEASE	611890		Sequencing	1000
Molecular Tests	GLI3 (GLI-KRUPPEL FAMILY MEMBER 3)	165240	PALLISTER-HALL SYNDROME » HYPOTHALAMIC HAMARTOBLASTOMA, HYPOPITUITARISM, IMPERFORATE ANUS, AND POSTAXIAL POLYDACTYLY	146510		Sequencing	1700
Molecular Tests	GLI3 (GLI-KRUPPEL FAMILY MEMBER 3)	165240	PALLISTER-HALL SYNDROME » HYPOTHALAMIC HAMARTOBLASTOMA, HYPOPITUITARISM, IMPERFORATE ANUS, AND POSTAXIAL POLYDACTYLY	146510		Deletions	350
Molecular Tests	GLI3 (GLI-KRUPPEL FAMILY MEMBER 3)	165240	POLYDACTYLY, POSTAXIAL, TYPE A1	174200		Sequencing	1700
Molecular Tests	GLI3 (GLI-KRUPPEL FAMILY MEMBER 3)	165240	POLYDACTYLY, POSTAXIAL, TYPE A1	174200		Deletions	350
Molecular Tests	GLI3 (GLI-KRUPPEL FAMILY MEMBER 3)	165240	POLYDACTYLY, PREAXIAL 4 » POLYSYNDACTYLY, UNCOMPLICATED CROSSED » POLYDACTYLY, TYPE 4	174700		Sequencing	1700
Molecular Tests	GLI3 (GLI-KRUPPEL FAMILY MEMBER 3)	165240	POLYDACTYLY, PREAXIAL 4 » POLYSYNDACTYLY, UNCOMPLICATED CROSSED » POLYDACTYLY, TYPE 4	174700		Deletions	350
Molecular Tests	GLI3 (GLI-KRUPPEL FAMILY MEMBER 3)	165240	GREIG CEPHALOPOLYSYNDACTYLY SYNDROME » POLYSYNDACTYLY WITH PECULIAR SKULL SHAPE	175700		Sequencing	1700
Molecular Tests	GLI3 (GLI-KRUPPEL FAMILY MEMBER 3)	165240	GREIG CEPHALOPOLYSYNDACTYLY SYNDROME » POLYSYNDACTYLY WITH PECULIAR SKULL SHAPE	175700		Deletions	350
Molecular Tests	GLI3 (GLI-KRUPPEL FAMILY MEMBER 3)	165240	ACROCALLOSAL SYNDROME » SCHINZEL ACROCALLOSAL SYNDROME	200990		Sequencing	1700
Molecular Tests	GLI3 (GLI-KRUPPEL FAMILY MEMBER 3)	165240	ACROCALLOSAL SYNDROME » SCHINZEL ACROCALLOSAL SYNDROME	200990		Deletions	350
Molecular Tests	GLIS3 (GLIS FAMILY ZINC FINGER PROTEIN 3; GLI-SIMILAR PROTEIN 3; ZINC FINGER PROTEIN 515; ZNF515)	610192	DIABETES MELLITUS, NEONATAL, WITH CONGENITAL HYPOTHYROIDISM » NDH SYNDROME	610199		Sequencing	1190
Molecular Tests	GLRA1 (GLYCINE RECEPTOR, ALPHA-1 SUBUNIT)	138491	HYPEREKPLEXIA » STARTLE DISEASE » KOK DISEASE » STIFF BABY SYNDROME	149400		Sequencing of 7 Exons of GLRA1 and Deletion-Duplication Testing of GLRA1, GLRB en SLC6A5	950
Molecular Tests	GLRB (GLYCINE RECEPTOR, BETA SUBUNIT)	138492	HYPEREKPLEXIA » STARTLE DISEASE » KOK DISEASE » STIFF BABY SYNDROME	149400		Sequencing	650

Molecular Tests	GLUD1 (GLUTAMATE DEHYDROGENASE 1)	138130	HYPERINSULINEMIC HYPOGLYCEMIA, FAMILIAL, TYPE 6 » HYPERINSULINISM-HYPERAMMONEMIA SYNDROME » PERSISTENT HYPERINSULINEMIC HYPOGLYCEMIA OF INFANCY » HYPOGLYCEMIA, HYPERINSULINEMIC, OF INFANCY » HYPERINSULINEMIC HYPOGLYCEMIA DUE TO FOCAL ADENOMATOUS HYPERPLASIA » NESIDIOBLASTOSIS OF PANCREAS » HYPERINSULINISM, FAMILIAL, WITH PANCREATIC NESIDIOBLASTOSIS	606762		Sequencing	800
Molecular Tests	GLUD1 (GLUTAMATE DEHYDROGENASE 1)	138130	HYPERINSULINEMIC HYPOGLYCEMIA, FAMILIAL, TYPE 6 » HYPERINSULINISM-HYPERAMMONEMIA SYNDROME » PERSISTENT HYPERINSULINEMIC HYPOGLYCEMIA OF INFANCY » HYPOGLYCEMIA, HYPERINSULINEMIC, OF INFANCY » HYPERINSULINEMIC HYPOGLYCEMIA DUE TO FOCAL ADENOMATOUS HYPERPLASIA » NESIDIOBLASTOSIS OF PANCREAS » HYPERINSULINISM, FAMILIAL, WITH PANCREATIC NESIDIOBLASTOSIS	606762		Exons 6, 7, 10, 11 and 12	510
Molecular Tests	GM2A (GM2-ACTIVATOR)	272750	TAY-SACHS DISEASE, AB VARIANT » HEXOSAMINIDASE ACTIVATOR DEFICIENCY » GM2-GANGLIOSIDOSIS, TYPE AB » [illegible]	272750		Sequencing	350
Molecular Tests	GNAS (GNAS1, ALPHA SUBUNIT OF Gs, ALPHA SUBUNIT OF ADENYLATE CYCLASE STIMULATORY PROTEIN)	139320	PITUITARY TUMOR, SOMATOTROPHINOMA			Sequencing	790
Molecular Tests	GNAS (GNAS1, ALPHA SUBUNIT OF Gs, ALPHA SUBUNIT OF ADENYLATE CYCLASE STIMULATORY PROTEIN)	139320	ALBRIGHT HEREDITARY OSTEODYSTROPHY » PSEUDOHYPOPARATHYROIDISM, TYPE 1A » PSEUDOHYPOPARATHYROIDISM, TYPE 1C	103580		Sequencing	790
Molecular Tests	GNAS (GNAS1, ALPHA SUBUNIT OF Gs, ALPHA SUBUNIT OF ADENYLATE CYCLASE STIMULATORY PROTEIN)	139320	OSSEOUS HETEROPLASIA, PROGRESSIVE » ECTOPIC OSSIFICATION, FAMILIAL » [illegible]	166350		Sequencing	790
Molecular Tests	GNAS (GNAS1, ALPHA SUBUNIT OF Gs, ALPHA SUBUNIT OF ADENYLATE CYCLASE STIMULATORY PROTEIN)	139320	MCCUNE-ALBRIGHT SYNDROME » ALBRIGHT SYNDROME » [illegible]	174800		Sequencing	790
Molecular Tests	GNAS (GNAS1, ALPHA SUBUNIT OF Gs, ALPHA SUBUNIT OF ADENYLATE CYCLASE STIMULATORY PROTEIN)	139320	MCCUNE-ALBRIGHT SYNDROME » ALBRIGHT SYNDROME » [illegible]	174800		2 Common Mutations: R201H and R201C	250
Molecular Tests	GNAS (GNAS1, ALPHA SUBUNIT OF Gs, ALPHA SUBUNIT OF ADENYLATE CYCLASE STIMULATORY PROTEIN)	139320	PSEUDOHYPOPARATHYROIDISM, TYPE 1B	603233		Methylation	250
Molecular Tests	GNAT1 (GUANINE NUCLEOTIDE-BINDING PROTEIN, ALPHA-TRANSDUCING ACTIVITY POLYPEPTIDE 1;G PROTEIN, ALPHA TRANSDUCIN 1; TRANSDUCIN, ROD-SPECIFIC, ALPHA POLYPEPTIDE)	139330	NIGHT BLINDNESS, CONGENITAL STATIONARY (AUTOSOMAL DOMINANT), TYPE 1 » NIGHT BLINDNESS, CONGENITAL STATIONARY, INCOMPLETE TYPE	610444		Sequencing	550
Molecular Tests	GNAT2 (GUANINE NUCLEOTIDE-BINDING PROTEIN, ALPHA-TRANSDUCING ACTIVITY POLYPEPTIDE 2; G PROTEIN, ALPHA TRANSDUCIN 2; TRANSDUCIN, CONE-SPECIFIC, ALPHA POLYPEPTIDE)	139340	ACHROMATOPSIA, TYPE 4	613856		Sequencing	680
Molecular Tests	GNE (GLCNE, UDP-N-ACETYLGLUCOSAMINE 2-EPIMERASE/N-ACETYLMANNOSAMINE KINASE)	603824	SIALURIA	269921		Sequencing	750
Molecular Tests	GNE (GLCNE, UDP-N-ACETYLGLUCOSAMINE 2-EPIMERASE/N-ACETYLMANNOSAMINE KINASE)	603824	INCLUSION BODY MYOPATHY 2 (AUTOSOMAL RECESSIVE), IBM2	600737		Sequencing	750
Molecular Tests	GNE (GLCNE, UDP-N-ACETYLGLUCOSAMINE 2-EPIMERASE/N-ACETYLMANNOSAMINE KINASE)	603824	NONAKA DISTAL MYOPATHY » MYOPATHY, DISTAL, WITH RIMMED VACUOLES	605820		Sequencing	750
Molecular Tests	GNPAT (GLYCERONEPHOSPHATE O-ACYLTRANSFERASE; DIHYDROXYACETONEPHOSPHATE ACYLTRANSFERASE; DHAPAT; DIHYDROXYACETONEPHOSPHATE ACYLTRANSFERASE; DHAPAT)	222765	RHIZOMELIC CHONDRODYSPLASIA PUNCTATA, TYPE 2, RCDP2 » DIHYDROXYACETONEPHOSPHATE ACYLTRANSFERASE DEFICIENCY » DHAPAT DEFICIENCY » [illegible]	602744		Sequencing	1100
Molecular Tests	GNPTAB (N-ACETYLGLUCOSAMINE-1-PHOSPHOTRANSFERASE, ALPHA/BETA SUBUNITS)	607840	MUCOLIPIDOSIS, TYPE 2, ML2 » I-CELL DISEASE	252500		Sequencing	2020
Molecular Tests	GNPTAB (N-ACETYLGLUCOSAMINE-1-PHOSPHOTRANSFERASE, ALPHA/BETA SUBUNITS)	607840	MUCOLIPIDOSIS, TYPE 3, ML3 » PSEUDO-HURLER POLYDYSTROPHY	252600		Sequencing	2020
Molecular Tests	GNPTG (N-ACETYLGLUCOSAMINE-1-PHOSPHOTRANSFERASE, GAMMA SUBUNIT; GNPTAG)	607838	MUCOLIPIDOSIS, TYPE 3C, ML3C » MUCOLIPIDOSIS, TYPE 3, IRANIAN VARIANT » MUCOLIPIDOSIS, TYPE 3, GAMMA	252605		Sequencing	800
Molecular Tests	GNS (N-ACETYLGLUCOSAMINE-6-SULFATASE; GLUCOSAMINE-6-SULFATASE)	607664	MUCOPOLYSACCHARIDOSIS, TYPE IID, MPSIID » SANFILIPPO SYNDROME D » [illegible]	252940		Sequencing	1380

Molecular Tests	GORAB (GOLGIN, RAB6-INTERACTING; SCYL1-BINDING PROTEIN 1; SCYLBP1; NTKL-BINDING PROTEIN 1; NTKLBP1)	607983	GERODERMA OSTEODYSPLASTICUM » WALT DISNEY DWARFISM » CERODERMA OSTEODYSPLASTICA	231070		Sequencing	600
Molecular Tests	GP1BA (GLYCOPROTEIN Ib, PLATELET, ALPHA POLYPEPTIDE, GLYCOCALICIN)	606672	PSEUDO-VON WILLEBRAND DISEASE » VON WILLEBRAND DISEASE, PLATELET-TYPE	177820		Sequencing	550
Molecular Tests	GP1BA (GLYCOPROTEIN Ib, PLATELET, ALPHA POLYPEPTIDE, GLYCOCALICIN)	606672	BERNARD-SOULIER SYNDROME » GIANT PLATELET SYNDROME » VON WILLEBRAND FACTOR RECEPTOR DEFICIENCY	231200		Sequencing	550
Molecular Tests	GP1BB (GLYCOPROTEIN Ib, PLATELET, BETA POLYPEPTIDE)	138720	» BERNARD-SOULIER SYNDROME » GIANT PLATELET SYNDROME » PLATELET GLYCOPROTEIN Ib DEFICIENCY » GLYCOPROTEIN Ib, PLATELET, DEFICIENCY OF » VON WILLEBRAND FACTOR RECEPTOR DEFICIENCY	231200		Sequencing	400
Molecular Tests	GP9 (GLYCOPROTEIN IX, PLATELET)	173515	BERNARD-SOULIER SYNDROME » GIANT PLATELET SYNDROME » VON WILLEBRAND FACTOR RECEPTOR DEFICIENCY	231200		Sequencing	250
Molecular Tests	GPC1 (GLYPICAN 1)	600017	SIMPSON-GOLABI-BEHMEL SYNDROME, SGBS	312870		Deletion analysis	600
Molecular Tests	GPC1 (GLYPICAN 1)	600017	SIMPSON-GOLABI-BEHMEL SYNDROME, SGBS	312870		Deletion and whole gene analysis	980
Molecular Tests	GPHN (GEPHYRIN)	252150	MOLYBDENUM COFACTOR DEFICIENCY » GEPHYRIN (MOLYBDENUM COFACTOR BIOSYNTHESIS PROTEIN C)	603930		Sequencing	850
Molecular Tests	GPR143 (OA1)	300500	ALBINISM, OCULAR, TYPE 1, OA1 » NYSTAGMUS 1, X-LINKED, INCLUDED	300600		Sequencing	550
Molecular Tests	GPR56 (G PROTEIN-COUPLED RECEPTOR 56, TM7XN1)	604110	POLYMICROGYRIA, BILATERAL FRONTOPARIETAL » CEREBELLAR ATAXIA WITH NEUROPSYCHOLOGICAL MIGRATION DEFECT	606854		Sequencing	1540
Molecular Tests	GPR98 (G PROTEIN-COUPLED RECEPTOR 98; MONOGENIC AUDIOGENIC SEIZURE SUSCEPTIBILITY 1, MOUSE, HOMOLOG OF; MASS1; VERY LARGE G PROTEIN-COUPLED RECEPTOR 1; VLGR1)	602851	FEBRILE SEIZURES, FAMILIAL, TYPE4	604352		Sequencing	1300
Molecular Tests	GPR98 (G PROTEIN-COUPLED RECEPTOR 98; MONOGENIC AUDIOGENIC SEIZURE SUSCEPTIBILITY 1, MOUSE, HOMOLOG OF; MASS1; VERY LARGE G PROTEIN-COUPLED RECEPTOR 1; VLGR1)	602851	USHER SYNDROME, TYPE 2C, USH2C » USHER SYNDROME, TYPE 2B	605472		Sequencing	1300
Molecular Tests	GRHPR (GLYOXYLATE REDUCTASE / HYDROXYPYRUVATE REDUCTASE, GLXR)	604296	HYPEROXALURIA, PRIMARY, TYPE 2 » OXALOSIS 2 » GLYCERIC ACIDURIA » GLYOXYLATE REDUCTASE/HYDROXYPYRUVATE REDUCTASE DEFICIENCY	260000		Sequencing	650
Molecular Tests	GRHPR (GLYOXYLATE REDUCTASE / HYDROXYPYRUVATE REDUCTASE, GLXR)	604296	HYPEROXALURIA, PRIMARY, TYPE 2 » OXALOSIS 2 » GLYCERIC ACIDURIA » GLYOXYLATE REDUCTASE/HYDROXYPYRUVATE REDUCTASE DEFICIENCY	260000		2 Mutations: 103delG and c.403_405+2del4AGT	250
Molecular Tests	GRK1 (G PROTEIN-DEPENDENT RECEPTOR KINASE 1; RHODOPSIN KINASE, RHOK)	180381	OGUCHI DISEASE, TYPE 2 » NIGHT BLINDNESS, CONGENITAL STATIONARY, OGUCHI TYPE 2	613411		Sequencing	530
Molecular Tests	GRM6 (GLUTAMATE RECEPTOR, METABOTROPIC 6; MGLUR6)	604096	NIGHT BLINDNESS, CONGENITAL STATIONARY, TYPE 1B, CSNB1B	257270		Sequencing	530
Molecular Tests	GRN (GRANULIN, PROGRANULIN, EPITHELIN)	138945	FRONTOTEMPORAL DEMENTIA, UBIQUITIN-POSITIVE » FRONTAL HEMIATROPHY, PROGRESSIVE, INCLUDED	607485		Sequencing	800
Molecular Tests	GRN (GRANULIN, PROGRANULIN, EPITHELIN)	138945	FRONTOTEMPORAL DEMENTIA, UBIQUITIN-POSITIVE » FRONTAL HEMIATROPHY, PROGRESSIVE, INCLUDED	607485		Deletions	400
Molecular Tests	GUCA1A (GUANYLATE CYCLASE ACTIVATOR 1A; GUANYLIN 1; RETINA-GUCA1)	600364	CONE DYSTROPHY, TYPE 3, CODS	602093		Sequencing	680
Molecular Tests	GUCA1B (GUANYLATE CYCLASE ACTIVATOR 1B; GUCA1B; GUANYLIN 2; RETINA; GUCA2)	602275	RETINITIS PIGMENTOSA, TYPE 48, RP48	613827		Sequencing	680
Molecular Tests	GUCY2D (GUANYLATE CYCLASE 2D, MEMBRANE)	600179	LEBER CONGENITAL AMAUROSIS, TYPE 1, LCA1 » RETINAL DYSTROPHY, CONGENITAL	204000		Sequencing	870
Molecular Tests	GUCY2D (GUANYLATE CYCLASE 2D, MEMBRANE)	600179	CONE-ROD DYSTROPHY, TYPE 6, CORD6	601777		Sequencing	870
Molecular Tests	GUSB (BETA-GLUCURONIDASE)	611499	MUCOPOLYSACCHARIDOSIS TYPE 7, MPS7 » SLY SYNDROME » BETA-GLUCURONIDASE DEFICIENCY	253220		Sequencing	1100
Molecular Tests	GYS1 (GLYCOGEN SYNTHASE 1)	138570	GLYCOGEN STORAGE DISEASE, TYPE 0, MUSCLE » MUSCLE GLYCOGEN SYNTHASE DEFICIENCY	611556		Sequencing	1650

Molecular Tests	GYS2 (GLYCOGEN SYNTHASE 2)	138571	GLYCOGEN STORAGE DISEASE, TYPE 0 » GSD TYPE 0 » LIVER GLYCOGEN SYNTHASE DEFICIENCY	240600		Sequencing	1350
Molecular Tests	HADH (3-@HYDROXYACYL-CoA DEHYDROGENASE, HADSC, SCHAD)	601609	3-@HYDROXYACYL-CoA DEHYDROGENASE DEFICIENCY » HADH DEFICIENCY » SCHAD DEFICIENCY	231530		Sequencing	Upon Request
Molecular Tests	HADH (3-@HYDROXYACYL-CoA DEHYDROGENASE, HADSC, SCHAD)	601609	HYPERINSULINEMIC HYPOGLYCEMIA, FAMILIAL, TYPE 4	609975		Sequencing	Upon Request
Molecular Tests	HADH2 (HYDROXYACYL-CoA DEHYDROGENASE TYPE 2, AMYLOID BETA-BINDING POLYPEPTIDE, ERAB, 2-ALPHA-METHYL-3-HYDROXYBUTYRYL-CoA DEHYDROGENASE)	300256	2-ALPHA-METHYL-3-HYDROXYBUTYRYL-CoA DEHYDROGENASE DEFICIENCY	300438		Sequencing	600
Molecular Tests	HADHA (HYDROXYACYL-CoA DEHYDROGENASE/3-KETOACYL-CoA THIOLASE/ENOYL-CoA HYDRATASE, ALPHA SUBUNIT, TRIFUNCTIONAL PROTEIN, ALPHA SUBUNIT MITOCHONDRIAL TRIFUNCTIONAL PROTEIN, ALPHA SUBUNIT, LONG-CHAIN HYDROXYACYL-CoA DEHYDROGENASE, LCHAD)	600890	NEUROPATHY-MYOPATHY, PROGRESSIVE, DUE TO TRIFUNCTIONAL PROTEIN DEFICIENCY			Sequencing	1780
Molecular Tests	HADHA (HYDROXYACYL-CoA DEHYDROGENASE/3-KETOACYL-CoA THIOLASE/ENOYL-CoA HYDRATASE, ALPHA SUBUNIT, TRIFUNCTIONAL PROTEIN, ALPHA SUBUNIT MITOCHONDRIAL TRIFUNCTIONAL PROTEIN, ALPHA SUBUNIT, LONG-CHAIN HYDROXYACYL-CoA DEHYDROGENASE, LCHAD)	600890	NEUROPATHY-MYOPATHY, PROGRESSIVE, DUE TO TRIFUNCTIONAL PROTEIN DEFICIENCY			2 Common Mutations: (1528G>C and 1132C>T)	380
Molecular Tests	HADHA (HYDROXYACYL-CoA DEHYDROGENASE/3-KETOACYL-CoA THIOLASE/ENOYL-CoA HYDRATASE, ALPHA SUBUNIT, TRIFUNCTIONAL PROTEIN, ALPHA SUBUNIT MITOCHONDRIAL TRIFUNCTIONAL PROTEIN, ALPHA SUBUNIT, LONG-CHAIN HYDROXYACYL-CoA DEHYDROGENASE, LCHAD)	600890	TRIFUNCTIONAL PROTEIN DEFICIENCY	609015		Sequencing	1780
Molecular Tests	HADHA (HYDROXYACYL-CoA DEHYDROGENASE/3-KETOACYL-CoA THIOLASE/ENOYL-CoA HYDRATASE, ALPHA SUBUNIT, TRIFUNCTIONAL PROTEIN, ALPHA SUBUNIT MITOCHONDRIAL TRIFUNCTIONAL PROTEIN, ALPHA SUBUNIT, LONG-CHAIN HYDROXYACYL-CoA DEHYDROGENASE, LCHAD)	600890	TRIFUNCTIONAL PROTEIN DEFICIENCY	609015		2 Common Mutations: (1528G>C and 1132C>T)	380
Molecular Tests	HADHA (HYDROXYACYL-CoA DEHYDROGENASE/3-KETOACYL-CoA THIOLASE/ENOYL-CoA HYDRATASE, ALPHA SUBUNIT, TRIFUNCTIONAL PROTEIN, ALPHA SUBUNIT MITOCHONDRIAL TRIFUNCTIONAL PROTEIN, ALPHA SUBUNIT, LONG-CHAIN HYDROXYACYL-CoA DEHYDROGENASE, LCHAD)	600890	LONG-CHAIN 3-HYDROXYACYL-CoA DEHYDROGENASE DEFICIENCY, LCHAD DEFICIENCY	609016		Whole Gene	1780
Molecular Tests	HADHA (HYDROXYACYL-CoA DEHYDROGENASE/3-KETOACYL-CoA THIOLASE/ENOYL-CoA HYDRATASE, ALPHA SUBUNIT, TRIFUNCTIONAL PROTEIN, ALPHA SUBUNIT MITOCHONDRIAL TRIFUNCTIONAL PROTEIN, ALPHA SUBUNIT, LONG-CHAIN HYDROXYACYL-CoA DEHYDROGENASE, LCHAD)	600890	LONG-CHAIN 3-HYDROXYACYL-CoA DEHYDROGENASE DEFICIENCY, LCHAD DEFICIENCY	609016		2 Common Mutations: (1528G>C and 1132C>T)	380
Molecular Tests	HADHB (HYDROXYACYL-CoA DEHYDROGENASE/3-KETOACYL-CoA THIOLASE/ENOYL-CoA HYDRATASE, BETA SUBUNIT, TRIFUNCTIONAL PROTEIN, BETA SUBUNIT MITOCHONDRIAL TRIFUNCTIONAL PROTEIN, BETA SUBUNIT HYDROXYACYL-CoA DEHYDROGENASE, HADH)	143450	TRIFUNCTIONAL PROTEIN DEFICIENCY	609015		Sequencing	850
Molecular Tests	HAMP (HEPCIDIN ANTIMICROBIAL PEPTIDE)	606464	» HEMOCHROMATOSIS, TYPE 2, HFE2 HEMOCHROMATOSIS, JUVENILE » HEMOCHROMATOSIS DUE TO DEFECT IN HEMOJUVELIN » HEMOCHROMATOSIS DUE TO DEFECT IN HEPCIDIN ANTIMICROBIAL PEPTIDE	602390		Sequencing	300
Molecular Tests	HAX1 (HCLS1-ASSOCIATED PROTEIN X1)	605998	NEUTROPENIA, SEVERE CONGENITAL, TYPE 3 (AUTOSOMAL RECESSIVE) » KOSTMANN DISEASE » AGRANULOCYTOSIS, INFANTILE	610738		Sequencing	940
Molecular Tests	HBA 1 and HBA 2 (ALPHA GLOBIN)	141800	ALPHA HAEMOGLOBINOPATHIA » ALPHA THALASSEMIA	141800		Sequencing and Deletion Analysis (both HBA1 and HBA2)	650
Molecular Tests	HBB (BETA GLOBIN)	141900	BETA HAEMOGLOBINOPATHIA » BETA THALASSEMIA	141900		Sequencing	390
Molecular Tests	HBB (BETA GLOBIN)	141900	SICKLE CELL ANEMIA	603903		Sequencing	390
Molecular Tests	HD (HUNTINGTIN)	143100	HUNTINGTON CHOREA	143100		Repeat	490
Molecular Tests	HEPACAM (HEPATOCYTE CELL ADHESION MOLECULE; GLIAL CELL ADHESION MOLECULE; GLIALCAM; HEPATOCYTE AND GLIAL CELL ADHESION MOLECULE)	611642	MEGALENCEPHALIC LEUKOENCEPHALOPATHY WITH SUBCORTICAL CYSTS, TYPE 2A	613925		Sequencing	770
Molecular Tests	HEPACAM (HEPATOCYTE CELL ADHESION MOLECULE; GLIAL CELL ADHESION MOLECULE; GLIALCAM; HEPATOCYTE AND GLIAL CELL ADHESION MOLECULE)	611642	MEGALENCEPHALIC LEUKOENCEPHALOPATHY WITH SUBCORTICAL CYSTS, TYPE 2B, REMITTING, WITH OR WITHOUT MENTAL RETARDATION	613926		Sequencing	770
Molecular Tests	HERG	152427	LONG QT SYNDROME 2, LQT2 » ROMANO-WARD SYNDROME	152427		Sequencing	1400
Molecular Tests	HES7 (HAIRY/ENHANCER OF SPLIT, DROSOPHILA, HOMOLOG OF, 7)	608059	SPONDYLOCOSTAL DYSOSTOSIS, TYPE 4 (AUTOSOMAL RECESSIVE)	613686		Sequencing	370
Molecular Tests	HESX1 (HOMEOBOX GENE EXPRESSED IN ES CELLS)	601802	PITUITARY DWARFISM 3 » PANHYPOPITUITARISM » ATELIOTIC DWARFISM WITH HYPOGONADISM » HANHART DWARFISM	262600		Sequencing	440
Molecular Tests	HEXA (HEXOSAMINIDASE A)	606869	TAY-SACHS DISEASE » GANGLIOSIDOSIS, GENERALIZED GM2, TYPE 1 » HEXOSAMINIDASE A DEFICIENCY	272800		Sequencing	900

Molecular Tests	HEXA (HEXOSAMINIDASE A)	606869	TAY-SACHS DISEASE » GANGLIOSIDOSIS, GENERALIZED GM2, TYPE 1 » HEXOSAMINIDASE A DEFICIENCY	272800		1277insTATC, IVS12+1G-C, G269S	250
Molecular Tests	HEXB (HEXOSAMINIDASE B)	606873	SANDHOFF DISEASE » GM2-GANGLIOSIDOSIS, TYPE 2 » HEXOSAMINIDASES A AND B DEFICIENCY	268800		Sequencing	900
Molecular Tests	HFE	235200	HEMOCHROMATOSIS, TYPE 1, HFE1	235200		Sequencing	650
Molecular Tests	HFE	235200	HEMOCHROMATOSIS, TYPE 1, HFE1	235200		4 Mutations (C282Y, H63D, S65C, E168X)	280
Molecular Tests	HGD (HOMOGENTISATE 1,2-DIOXYGENASE)	607474	ALKAPTONURIA » HOMOGENTISIC ACID OXIDASE DEFICIENCY	203500		Sequencing	800
Molecular Tests	HGSNAT (HEPARAN-ALPHA-GLUCOSAMINIDE N-ACETYLTRANSFERASE; TRANSMEMBRANE PROTEIN 76; TMEM76)	610453	MUCOPOLYSACCHARIDOSIS, TYPE 3C, MPS3C » SANFILIPPO SYNDROME C » ACETYL-CoA:ALPHA-GLUCOSAMINIDE N-ACETYLTRANSFERASE DEFICIENCY	252930		Sequencing	1450
Molecular Tests	HGSNAT (HEPARAN-ALPHA-GLUCOSAMINIDE N-ACETYLTRANSFERASE; TRANSMEMBRANE PROTEIN 76; TMEM76)	610453	MUCOPOLYSACCHARIDOSIS, TYPE 3C, MPS3C » SANFILIPPO SYNDROME C » ACETYL-CoA:ALPHA-GLUCOSAMINIDE N-ACETYLTRANSFERASE DEFICIENCY	252930		Deletion-Duplication Testing	600
Molecular Tests	HJV (HEMOJUVELIN)	608374	HEMOCHROMATOSIS, TYPE 2, HFE2 » HEMOCHROMATOSIS, JUVENILE » HEMOCHROMATOSIS DUE TO DEFECT IN HEPCIDIN ANTIMICROBIAL PEPTIDE	602390		Sequencing	700
Molecular Tests	HLCS (HOLOCARBOXYLASE SYNTHETASE; HCS)	609018	HOLOCARBOXYLASE SYNTHETASE DEFICIENCY » MULTIPLE CARBOXYLASE DEFICIENCY, EARLY ONSET » HLCS DEFICIENCY	253270		Sequencing	890
Molecular Tests	HMBS (HYDROXYMETHYLBILANE SYNTHASE)	609806	PORPHYRIA, ACUTE INTERMITTENT » PORPHOBILINOGEN DEAMINASE DEFICIENCY » PBGD DEFICIENCY » UROPORPHYRINOGEN SYNTHASE DEFICIENCY	176000		Sequencing	880
Molecular Tests	HMGCL (HMG - COA SYNTHETASE)	246450	HMG-COA LYASE DEFICIENCY » HMG-COA SYNTHETASE DEFICIENCY	246450		Sequencing	1290
Molecular Tests	HNF1A (TCF1)	142410	HEPATIC ADENOMA	142330		Sequencing	650
Molecular Tests	HNF1A (TCF1)	142410	HEPATIC ADENOMA	142330		Deletion-Duplication Testing	350
Molecular Tests	HNF1A (TCF1)	142410	MATURITY-ONSET DIABETES OF THE YOUNG, TYPE 3, MODY3	600496		Sequencing	650
Molecular Tests	HNF1A (TCF1)	142410	MATURITY-ONSET DIABETES OF THE YOUNG, TYPE 3, MODY3	600496		Deletion-Duplication Testing	350
Molecular Tests	HNF1B (HNF2, TCF2)	189907	HYPOPLASTIC GLOMERULOCYSTIC KIDNEY DISEASE	137920		Sequencing	600
Molecular Tests	HNF1B (HNF2, TCF2)	189907	HYPOPLASTIC GLOMERULOCYSTIC KIDNEY DISEASE	137920		Deletion-Duplication Testing	350
Molecular Tests	HNF1B (HNF2, TCF2)	189907	MATURITY-ONSET DIABETES OF THE YOUNG, TYPE 5, MODY5	604284		Sequencing	600
Molecular Tests	HNF1B (HNF2, TCF2)	189907	MATURITY-ONSET DIABETES OF THE YOUNG, TYPE 5, MODY5	604284		Deletion-Duplication Testing	350
Molecular Tests	HNF4A	600281	MATURITY-ONSET DIABETES OF THE YOUNG, TYPE 1, MODY1	125850		Sequencing	745
Molecular Tests	HNF4A	600281	MATURITY-ONSET DIABETES OF THE YOUNG, TYPE 1, MODY1	125850		Sequencing of HNF1A and HNF4A with Deletion-Duplication Testing of HNF1A and HNF4A	1170
Molecular Tests	HOXA 13	142959	GUTTMACHER SYNDROME			Sequencing	510
Molecular Tests	HOXA 13	142959	HAND-FOOT-UTERUS SYNDROME » HAND-FOOT-GENITAL SYNDROME	140000		Sequencing	510
Molecular Tests	HOXA1 (HOMEOBOX A1; HOMEOBOX 1F; HOX1F)	142955	ATHABASKAN BRAINSTEM DYSGENESIS SYNDROME » BOSLEY-SALIH-ALORAINY SYNDROME	601536		Exon 1	250
Molecular Tests	HOXD 13 (HOMEOBOX D13)	142989	BRACHYDACTYLY, TYPE D, BDD	113200		Sequencing	510
Molecular Tests	HOXD 13 (HOMEOBOX D13)	142989	BRACHYDACTYLY, TYPE E, BDE	113300		Sequencing	510
Molecular Tests	HOXD 13 (HOMEOBOX D13)	142989	SYNDACTYLY, TYPE 2 » SYNPOLYDACTYLY	186000			510
Molecular Tests	HPRP3 (PRECURSOR mRNA-PROCESSING FACTOR 3, S. CEREVISIAE, HOMOLOG OF)	607301	RETINITIS PIGMENTOSA, TYPE 18, RP18	601414		Sequencing	1150
Molecular Tests	HPRT1 (HYPOXANTHINE GUANINE PHOSPHORIBOSYL TRANSFERASE 1, HGPRT)	308000	LESCH - NYHAN SYNDROME, LNS » HYPOXANTHINE GUANINE PHOSPHORIBOSYLTRANSFERASE 1 DEFICIENCY » HPRT1 DEFICIENCY » HPRT DEFICIENCY	300322		Sequencing	790
Molecular Tests	HPRT1 (HYPOXANTHINE GUANINE PHOSPHORIBOSYL TRANSFERASE 1, HGPRT)	308000	GOUT, HPRT-RELATED » KELLEY-SEEGMILLER SYNDROME	300323		Sequencing	790
Molecular Tests	HPS PANEL		HERMANSKY-PUDLAK SYNDROME, TYPE 1-8, HPS1-8			8 Genes: HPS1-8	2630

Molecular Tests	HPS1	604982	HERMANSKY-PUDLAK SYNDROME, TYPE 1, HPS1	203300		Sequencing	680
Molecular Tests	HPS1 and HPS3		HERMANSKY-PUDLAK SYNDROME	203300		3 Mutations: 16bp Duplication, 3.9kb Deletion and IVS5+1G>A	570
Molecular Tests	HPS3	606118	HERMANSKY-PUDLAK SYNDROME, TYPE 3, HPS3	203300		Sequencing	720
Molecular Tests	HPS4	606682	HERMANSKY-PUDLAK SYNDROME, TYPE 4, HPS4	203300		Sequencing	680
Molecular Tests	HPS5 (RUBY-EYE 2, MOUSE, HOMOLOG OF; RU2; ALPHA-INTEGRIN-BINDING PROTEIN 63; AIBP63)	607521	HERMANSKY-PUDLAK SYNDROME, TYPE 5, HPS5	614074		Sequencing	680
Molecular Tests	HPS6 (RUBY-EYE, MOUSE, HOMOLOG OF; RU)	607522	HERMANSKY-PUDLAK SYNDROME, TYPE 6, HPS6	614075		Sequencing	770
Molecular Tests	HR (HAIRLESS, MOUSE, HOMOLOG OF)	602302	ALOPECIA UNIVERSALIS CONGENITA » ATRICHIA, GENERALIZED	203655		Sequencing	1800
Molecular Tests	HR (HAIRLESS, MOUSE, HOMOLOG OF)	602302	ATRICHIA WITH PAPULAR LESIONS » PAPULAR ATRICHIA	209500		Sequencing	1800
Molecular Tests	HRAS (V-HA-RAS HARVEY RAT SARCOMA VIRAL ONCOGENE HOMOLOG)	190020	COSTELLO SYNDROME » FACIOCUTANEOSKELETAL SYNDROME	218040		Sequencing	580
Molecular Tests	HSD11B2	218030	CORTISOL 11-BETA-KETOREDUCTASE DEFICIENCY » APPARENT MINERALOCORTICOID EXCESS » 11-@BETA-HYDROXYSTEROID DEHYDROGENASE, TYPE 2	218030		Sequencing	400
Molecular Tests	HSD17B3 (17-@BETA HYDROXYSTEROID DEHYDROGENASE 3, ESTRADIOL 17-BETA-DEHYDROGENASE,EDH17B3)	605573	17-@BETA HYDROXYSTEROID DEHYDROGENASE 3 DEFICIENCY » PSEUDOHERMAPHRODITISM, MALE, WITH GYNECOMASTIA » POLYCYSTIC OVARIAN DISEASE DUE TO 17-KETOSTEROID REDUCTASE DEFICIENCY » 17-@KETOSTEROID REDUCTASE DEFICIENCY OF TESTIS	264300		Sequencing	1310
Molecular Tests	HSD3B2 (3-@BETA-HYDROXYSTEROID DEHYDROGENASE/DELTA-ISOMERASE, TYPE 2)	201810	ADRENAL HYPERPLASIA, TYPE 2 » 3-@BETA-HYDROXYSTEROID DEHYDROGENASE, DEFICIENCY OF, TYPE 2 » 3-@BETA-HSD DEFICIENCY	201810		Sequencing	500
Molecular Tests	HSN2	608620	NEUROPATHY, HEREDITARY SENSORY AND AUTONOMIC, TYPE 2, HSAN2 » ACROOSTEOLYSIS, NEUROGENIC » MORVAN DISEASE » NEUROPATHY, PROGRESSIVE SENSORY, OF CHILDREN	201300		Sequencing	480
Molecular Tests	HSPB8 (HEAT-SHOCK 22-KD PROTEIN 8)	608014	NEURONOPATHY, DISTAL HEREDITARY MOTOR, TYPE 2A » SPINAL MUSCULAR ATROPHY, DISTAL, ADULT (AUTOSOMAL DOMINANT), TYPE 2A » CHARCOT-MARIE-TOOTH DISEASE, SPINAL, TYPE 2A	158590		Sequencing	550
Molecular Tests	HSPB8 (HEAT-SHOCK 22-KD PROTEIN 8)	608014	CHARCOT-MARIE-TOOTH DISEASE, AXONAL, TYPE 2L, CMT2L	608673		Sequencing	550
Molecular Tests	HSPD1 (HEAT-SHOCK 60-KD PROTEIN 1, SPG13)	118190	SPASTIC PARAPLEGIA 13 (AUTOSOMAL DOMINANT)	118190		Sequencing	850
Molecular Tests	HSPG2 (PERLECAN, HEPARAN SULFATE PROTEOGLYCAN OF BASEMENT MEMBRANE)	142461	DYSSEGMENTAL DYSPLASIA, SILVERMAN-HANDMAKER TYPE » ANISOSPONDYLIC CAMPTOMICROMELIC DWARFISM, SILVERMAN-HANDMAKER TYPE	224410		Sequencing	4400
Molecular Tests	HSPG2 (PERLECAN, HEPARAN SULFATE PROTEOGLYCAN OF BASEMENT MEMBRANE)	142461	SCHWARTZ-JAMPEL SYNDROME, TYPE 1 » SJA SYNDROME » MYOTONIC MYOPATHY, DWARFISM, CHONDRODYSTROPHY, AND OCULAR AND FACIAL ABNORMALITIES » CHONDRODYSTROPHIC MYOTONIA	255800		Sequencing	4400
Molecular Tests	HYAL1 (HYALURONOGLUCOSAMINIDASE 1; HYALURONIDASE 1)	607071	MUCOPOLYSACCHARIDOSIS TYPE 9, MPS9 » HYALURONIDASE DEFICIENCY	601492		Sequencing	600
Molecular Tests	ICHTHYIN (ICHYN)	609383	ICHTHYOSIS, CONGENITAL (AUTOSOMAL RECESSIVE), ICHTHYIN-RELATED	612281		Sequencing	1070
Molecular Tests	ICOS (INDUCIBLE T-CELL COSTIMULATOR; ACTIVATION-INDUCIBLE LYMPHOCYTE IMMUNOMEDIATORY MOLECULE; AILIM)	604558	IMMUNODEFICIENCY, COMMON VARIABLE, TYPE 1 » ANTIBODY DEFICIENCY DUE TO ICOS DEFECT	607594		Sequencing	1150
Molecular Tests	ICR1	608897	SILVER-RUSSELL SYNDROME » RUSSELL-SILVER SYNDROME	180860		Combined testing of epimutation at ICR1 on chromosome 11p15 and UPD of chromosome 7	500
Molecular Tests	IDH3B (ISOCITRATE DEHYDROGENASE 3, BETA SUBUNIT; ISOCITRATE DEHYDROGENASE, NAD(+)-SPECIFIC, MITOCHONDRIAL, BETA SUBUNIT)	604526	RETINITIS PIGMENTOSA, TYPE 46, RP46	612572		Sequencing	530
Molecular Tests	IDS (IDURONATE SULFATASE)	309900	MUCOPOLYSACCHARIDOSIS, TYPE 2 » HUNTER SYNDROME	309900		Sequencing	930
Molecular Tests	IDUA (ALPHA-L-IDURONIDASE)	252800	MUCOPOLYSACCHARIDOSIS, TYPE IH » HURLER SYNDROME	607014		Sequencing	1150
Molecular Tests	IDUA (ALPHA-L-IDURONIDASE)	252800	MUCOPOLYSACCHARIDOSIS, TYPE IH/S » HURLER-SCHEIE SYNDROME	607015		Sequencing	1150
Molecular Tests	IDUA (ALPHA-L-IDURONIDASE)	252800	MUCOPOLYSACCHARIDOSIS, TYPE 5, MPS5 » SCHEIE SYNDROME	607016		Sequencing	1150
Molecular Tests	IFT80 (INTRAFLAGELLAR TRANSPORT 80, CHLAMYDOMONAS, HOMOLOG OF; WD REPEAT-CONTAINING PROTEIN 56)	611177	ASPHYXIATING THORACIC DYSTROPHY, TYPE 2 » JEUNE SYNDROME	611263		Sequencing	990

Molecular Tests	IGHM (IMMUNOGLOBULIN MU)	147020	AGAMMAGLOBULINEMIA, NON-BRUTON TYPE (AUTOSOMAL RECESSIVE)	601495		Sequencing	990
Molecular Tests	IGHMBP2 (IMMUNOGLOBULIN MU BINDING PROTEIN 2, CARDIAC TRANSCRIPTION FACTOR 1, CATF1)	600502	SPINAL MUSCULAR ATROPHY WITH RESPIRATORY DISTRESS 1, SMARD1 » NEURONOPATHY, DISTAL HEREDITARY MOTOR, TYPE 6 » SEVERE INFANTILE AXONAL NEUROPATHY WITH RESPIRATORY FAILURE	604320		Sequencing	1900
Molecular Tests	IHH (INDIAN HEDGEHOG)	600726	BRACHYDACTYLY TYPE A1, BDA1 » FARABEE TYPE BRACHYDACTYLY	112500		Sequencing	450
Molecular Tests	IHH (INDIAN HEDGEHOG)	600726	ACROCAPITOFEMORAL DYSPLASIA, ACFD	607778		Sequencing	450
Molecular Tests	IKBKAP (IKK COMPLEX-ASSOCIATED PROTEIN, IKAP)	603722	DYSAUTONOMIA, FAMILIAL, DYS » RILEY-DAY SYNDROME » HEREDITARY SENSORY AND AUTONOMIC NEUROPATHY, TYPE 3, HSAN3	223900		Sequencing	2400
Molecular Tests	IKBKAP (IKK COMPLEX-ASSOCIATED PROTEIN, IKAP)	603722	DYSAUTONOMIA, FAMILIAL, DYS » RILEY-DAY SYNDROME » HEREDITARY SENSORY AND AUTONOMIC NEUROPATHY, TYPE 3, HSAN3	223900		1 Mutation : 2507+6T>C	250
Molecular Tests	IKBKG (NEMO)	300248	HYPOHIDROTIC ECTODERMAL DYSPLASIA WITH IMMUNE DEFICIENCY » ECTODERMAL DYSPLASIA, HYPOHIDROTIC, WITH IMMUNE DEFICIENCY	300291		Whole Gene Sequencing	1000
Molecular Tests	IKBKG (NEMO)	300248	HYPOHIDROTIC ECTODERMAL DYSPLASIA WITH IMMUNE DEFICIENCY » ECTODERMAL DYSPLASIA, HYPOHIDROTIC, WITH IMMUNE DEFICIENCY	300291		1 Mutation: Exon 4-10 Deletion	450
Molecular Tests	IKBKG (NEMO)	300248	BLOCH-SULZBERGER DISEASE » INCONTINENTIA PIGMENTI	308300		Whole Gene Sequencing	1000
Molecular Tests	IKBKG (NEMO)	300248	BLOCH-SULZBERGER DISEASE » INCONTINENTIA PIGMENTI	308300		1 Mutation: Exon 4-10 Deletion	450
Molecular Tests	IL1RAPL1 (INTERLEUKIN 1 RECEPTOR ACCESSORY PROTEIN-LIKE 1; INTERLEUKIN 1 RECEPTOR 8)	300206	MENTAL RETARDATION, NONSPECIFIC (X-LINKED), TYPE 21, MRX21 » MENTAL RETARDATION, NONSPECIFIC (X-LINKED), TYPE 34, MRX34	300143		Sequencing	1040
Molecular Tests	IL2RG (INTERLEUKIN 2 RECEPTOR, GAMMA)	308380	SEVERE COMBINED IMMUNODEFICIENCY (X-LINKED), T CELL-NEGATIVE, B CELL-POSITIVE, NK CELL-NEGATIVE, SCIDX1	300400		Sequencing	850
Molecular Tests	IL7R (INTERLEUKIN 7 RECEPTOR)	146661	SEVERE COMBINED IMMUNODEFICIENCY (AUTOSOMAL RECESSIVE), T CELL-NEGATIVE, B CELL-POSITIVE, NK CELL-POSITIVE, SCID	608971		Sequencing	1120
Molecular Tests	IMPDH1 (IMP DEHYDROGENASE 1;INOSINE-5-PRIME-MONOPHOSPHATE DEHYDROGENASE, TYPE 1)	146690	LEBER CONGENITAL AMAUROSIS, TYPE 11, LCA11	146690		Sequencing	720
Molecular Tests	IMPDH1 (IMP DEHYDROGENASE 1;INOSINE-5-PRIME-MONOPHOSPHATE DEHYDROGENASE, TYPE 1)	146690	RETINITIS PIGMENTOSA, TYPE 10, RP10	180105		Sequencing	720
Molecular Tests	IMPG2 (INTERPHOTORECEPTOR MATRIX PROTEOGLYCAN 2; SPACRCAN)	607056	RETINITIS PIGMENTOSA, TYPE 56, RP56 » MACULOPATHY, IMPG2-RELATED	613581		Sequencing	820
Molecular Tests	INF2 (INVERTED FORMIN 2)	610982	FOCAL SEGMENTAL GLOMERULOSCLEROSIS, TYPE 5	613237		Sequencing	1150
Molecular Tests	INS (INSULIN, PROINSULIN)	176730	HYPERPROINSULINEMIA » HYPERINSULINEMIA	176730		Sequencing	440
Molecular Tests	INSR (INSULIN RECEPTOR)	147670	INSULIN RESISTANCE			Sequencing	1250
Molecular Tests	INSR (INSULIN RECEPTOR)	147670	DIABETES MELLITUS, INSULIN-RESISTANT, WITH ACANTHOSIS NIGRICANS	147670		Sequencing	1250
Molecular Tests	INSR (INSULIN RECEPTOR)	147670	LEPRECHAUNISM » DONOHUE SYNDROME	246200		Sequencing	1250
Molecular Tests	INSR (INSULIN RECEPTOR)	147670	RABSON-MENDENHALL SYNDROME » PINEAL HYPERPLASIA, INSULIN-RESISTANT DIABETES MELLITUS, AND SOMATIC ABNORMALITIES	262190		Sequencing	1250
Molecular Tests	INSR (INSULIN RECEPTOR)	147670	HYPERINSULINEMIC HYPOGLYCEMIA, FAMILIAL, TYPE 5	609968		Sequencing	1250
Molecular Tests	IQCB1 (IQ MOTIF-CONTAINING PROTEIN B1; NEPHROCYSTIN 5; NPHP5)	609237	SENIOR-LOKEN SYNDROME, TYPE 5	609254		Sequencing	680
Molecular Tests	IRAK4 (INTERLEUKIN 1 RECEPTOR-ASSOCIATED KINASE 4	606883	IRAK4 DEFICIENCY	607676		Sequencing	1510
Molecular Tests	IRAK4 (INTERLEUKIN 1 RECEPTOR-ASSOCIATED KINASE 4	606883	INVASIVE PNEUMOCOCCAL DISEASE, RECURRENT ISOLATED, TYPE 1, IPD1 » INVASIVE PNEUMOCOCCAL DISEASE, PROTECTION AGAINST	610799		Sequencing	1510
Molecular Tests	IRF6	607199	VAN DER WOUDE SYNDROME	119300		Sequencing	1030
Molecular Tests	IRF6	607199	VAN DER WOUDE SYNDROME	119300		Deletion-Duplication Testing	680
Molecular Tests	IRF6	607199	POPLITEAL PTERYGIUM SYNDROME	119500		Sequencing	1030
Molecular Tests	IRF6	607199	POPLITEAL PTERYGIUM SYNDROME	119500		Deletion-Duplication Testing	680
Molecular Tests	ISCU (IRON-SULFUR CLUSTER SCAFFOLD, E. COLI, HOMOLOG OF)	611911	MYOPATHY WITH LACTIC ACIDOSIS, HEREDITARY » MYOPATHY WITH EXERCISE INTOLERANCE, SWEDISH TYPE » MYOPATHY WITH DEFICIENCY OF SUCCINATE DEHYDROGENASE AND ACONITASE	255125		Sequencing	590
Molecular Tests	ITGA2B (INTEGRIN, ALPHA-2B; PLATELET GLYCOPROTEIN IIb; GP2B; PLATELET FIBRINOGEN RECEPTOR, ALPHA SUBUNIT; PLATELET-SPECIFIC ANTIGEN BAK)	607759	THROMBASTHENIA OF GLANZMANN AND NAEGELI » PLATELET GLYCOPROTEIN IIb-IIIa DEFICIENCY » PLATELET FIBRINOGEN RECEPTOR, DEFICIENCY OF	273800		Sequencing	1300
Molecular Tests	ITGA6 (INTEGRIN, ALPHA-6)	147556	EPIDERMOLYSIS BULLOSA WITH PYLORIC ATRESIA » APLASIA CUTIS CONGENITA WITH GASTROINTESTINAL ATRESIA » CARMI SYNDROME	226730		Sequencing	1500

Molecular Tests	ITGA7 (INTEGRIN, ALPHA-7)	600536	MUSCULAR DYSTROPHY, CONGENITAL, DUE TO INTEGRIN ALPHA-7 DEFICIENCY » MYOPATHY, CONGENITAL, DUE TO INTEGRIN ALPHA-7 DEFICIENCY	613204		Sequencing	3150
Molecular Tests	ITGB2 (INTEGRIN, BETA-2)	600065	LEUKOCYTE ADHESION DEFICIENCY, TYPE 1, LAD » LEAD IMMUNODEFICIENCY	116920		Sequencing	1950
Molecular Tests	ITGB3 (INTEGRIN, BETA-3)	173470	THROMBOCYTOPENIA, NEONATAL, ALLOIMMUNE » POSTTRANSFUSION PURPURA	173470		Sequencing	950
Molecular Tests	ITGB3 (INTEGRIN, BETA-3)	173470	THROMBASTHENIA OF GLANZMANN AND NAEGELI » PLATELET GLYCOPROTEIN IIb-IIIa DEFICIENCY » PLATELET FIBRINOGEN RECEPTOR, DEFICIENCY OF	273800		Sequencing	950
Molecular Tests	ITGB4 (INTEGRIN, BETA-4)	147557	EPIDERMOLYSIS BULLOSA OF HANDS AND FEET » WEBER-COCKAYNE TYPE EPIDERMOLYSIS BULLOSA SIMPLEX » COCKAYNE-TOURAINE TYPE EPIDERMOLYSIS BULLOSA » EPIDERMOLYSIS BULLOSA SIMPLEX, GENERALIZED, OTHER	131800		Sequencing	2500
Molecular Tests	ITGB4 (INTEGRIN, BETA-4)	147557	EPIDERMOLYSIS BULLOSA, GENERALIZED ATROPHIC BENIGN » EPIDERMOLYSIS BULLOSA JUNCTIONALIS, PROGRESSIVE » EPIDERMOLYSIS BULLOSA JUNCTIONALIS, SEVERE NONLETHAL » EPIDERMOLYSIS BULLOSA JUNCTIONALIS, DISENTIS TYPE » CARAL SYNDROME	226650		Sequencing	2500
Molecular Tests	ITGB4 (INTEGRIN, BETA-4)	147557	EPIDERMOLYSIS BULLOSA WITH PYLORIC ATRESIA » APLASIA CUTIS CONGENITA WITH GASTROINTESTINAL ATRESIA » CARAL SYNDROME	226730		Sequencing	2500
Molecular Tests	ITPR1 (INOSITOL 1,4,5-TRIPHOSPHATE RECEPTOR, TYPE 1; IP3R; IP3R1)	147265	SPINOCEREBELLAR ATAXIA, TYPE 15, SCA15 » SPINOCEREBELLAR ATAXIA, TYPE 16, SCA16	606658		Sequencing	1900
Molecular Tests	IVD (ISOVALERYL-CoA DEHYDROGENASE)	607036	ISOVALERIC ACIDEMIA » ISOVALERIC ACID-CoA DEHYDROGENASE DEFICIENCY	243500		Sequencing	710
Molecular Tests	JAG1 (JAGGED1)	601920	ALAGILLE SYNDROME	118450		Sequencing and MLPA	1480
Molecular Tests	JAG1 (JAGGED1)	601920	TETRALOGY OF FALLOT	187500		Sequencing and MLPA	1480
Molecular Tests	JAK2 (JANUS KINASE 2)	147796	THROMBOCYTHEMIA, ESSENTIAL » THROMBOCYTOSIS, AUTOSOMAL DOMINANT	187950		Exon 12, including V617F Mutation	450
Molecular Tests	JAK2 (JANUS KINASE 2)	147796	MYELOFIBROSIS WITH MYELOID METAPLASIA, INCLUDED	254450		Exon 12, including V617F Mutation	450
Molecular Tests	JAK2 (JANUS KINASE 2)	147796	POLYCYTHEMIA VERA	263300		Exon 12, including V617F Mutation	450
Molecular Tests	JAK2 (JANUS KINASE 2)	147796	BUDD-CHIARI SYNDROME » HEPATIC VEIN OBSTRUCTION OF INFERIOR VENA CAVA	600880		Exon 12, including V617F Mutation	450
Molecular Tests	JAK3 (JANUS KINASE 3)	600173	SEVERE COMBINED IMMUNODEFICIENCY (AUTOSOMAL RECESSIVE), T CELL-NEGATIVE, B CELL-POSITIVE, NK CELL-NEGATIVE, SCID	600802		Sequencing	2540
Molecular Tests	JAK3 (JANUS KINASE 3)	600173	HUNTINGTON DISEASE-LIKE 2, HDL2	606438		Partial	3150
Molecular Tests	KAL1 (ADHESION MOLECULE-LIKE, X-LINKED, ANOSMIN 1)	308700	KALLMANN SYNDROME, TYPE 1, KAL1 » HYPOGONADOTROPIC HYPOGONADISM AND ANOSMIA, TYPE 1 » DYSPLASIA OLFACTOGENITALIS OF DE MORSIER	308700		Sequencing	1670
Molecular Tests	KBTBD13 (KELCH REPEAT AND BTB (POZ) DOMAIN-CONTAINING PROTEIN 13)	615737	NEMALINE MYOPATHY 6, NEM6	609273		Sequencing	1365
Molecular Tests	KCNA1 (POTASSIUM CHANNEL, VOLTAGE-GATED, SHAKER-RELATED SUBFAMILY, MEMBER 1)	176260	EPISODIC ATAXIA, TYPE 1, EA1 » EPISODIC ATAXIA WITH MYOKYMIA » ATAXIA, EPISODIC, WITH MYOKYMIA » PARAMYOMAL ATAXIA WITH NEUROMYOTONIA, HEREDITARY » MYOKYMIA WITH PERIODIC ATAXIA » MYOKYMIA » CONTINUOUS MUSCLE FIBER ACTIVITY, HEREDITARY	160120		Sequencing	250
Molecular Tests	KCNC3 (POTASSIUM CHANNEL, VOLTAGE-GATED, SHAW-RELATED SUBFAMILY, MEMBER 3)	176264	SPINOCEREBELLAR ATAXIA 13, SCA13	605259		Sequencing	995
Molecular Tests	KCNE1 (MINK, ISK)	176261	LONG QT SYNDROME 5, LQT5 » ROMANO-WARD SYNDROME	176261		Sequencing	250
Molecular Tests	KCNE1 (MINK, ISK)	176261	JERVELL AND LANGE-NIELSEN SYNDROME, JLNS1	220400		Sequencing	250
Molecular Tests	KCNE2	603796	LONG QT SYNDROME 6, LQT6 » ROMANO-WARD SYNDROME	603796		Sequencing	250
Molecular Tests	KCNE3 (POTASSIUM CHANNEL, VOLTAGE-GATED, ISK-RELATED SUBFAMILY, MEMBER 3)	604433	HYPOKALEMIC PERIODIC PARALYSIS, HOKPP » HYPERKALEMIC PERIODIC PARALYSIS	170400		Sequencing	350

Molecular Tests	KCNJ1 (POTASSIUM CHANNEL, INWARDLY RECTIFYING, SUBFAMILY J, MEMBER 1, KIR1.1, ROMK1)	600359	ANTENATAL BARTTER SYNDROME, TYPE 2 » HYPOKALEMIC ALKALOSIS WITH HYPERCALCIURIA, ANTENATAL, TYPE 2	241200		Sequencing	375
Molecular Tests	KCNJ11	600937	NESIDIOBLASTOSIS » HYPERINSULINISM (AUTOSOMAL RECESSIVE) » HYPERINSULINISM, FAMILIAL, WITH PANCREATIC NESIDIOBLASTOSIS » PERSISTENT HYPERINSULINEMIC HYPOGLYCEMIA OF INFANCY	256450		Sequencing	400
Molecular Tests	KCNJ11	600937	DIABETES MELLITUS, PERMANENT NEONATAL	606176		Sequencing	400
Molecular Tests	KCNJ13 (POTASSIUM CHANNEL, INWARDLY RECTIFYING, SUBFAMILY J, MEMBER 13; INWARDLY RECTIFYING POTASSIUM CHANNEL Kir7.1)	603208	VITREORETINAL DEGENERATION, SNOWFLAKE TYPE	193230		Sequencing	480
Molecular Tests	KCNJ13 (POTASSIUM CHANNEL, INWARDLY RECTIFYING, SUBFAMILY J, MEMBER 13; INWARDLY RECTIFYING POTASSIUM CHANNEL Kir7.1)	603208	LEBER CONGENITAL AMAUROSIS, TYPE 16, LCA16	614186		Sequencing	480
Molecular Tests	KCNJ2 (POTASSIUM CHANNEL, INWARDLY RECTIFYING, SUBFAMILY J, MEMBER 2, KIR2.1)	600681	ANDERSEN CARDIODYSRHYTHMIC PERIODIC PARALYSIS » ANDERSEN SYNDROME LONG QT SYNDROME 7, LQT7 » PERIODIC PARALYSIS, POTASSIUM-SENSITIVE CARDIODYSRHYTHMIC TYPE	170390		Sequencing	800
Molecular Tests	KCNJ2 (POTASSIUM CHANNEL, INWARDLY RECTIFYING, SUBFAMILY J, MEMBER 2, KIR2.1)	600681	SHORT QT SYNDROME 3, SQT3	609622		Sequencing	800
Molecular Tests	KCNQ1 (KVLQT1)	192500	LONG QT SYNDROME 1, LQT1 » ROMANO-WARD SYNDROME	192500		Sequencing	1580
Molecular Tests	KCNQ1 (KVLQT1)	192500	JERVELL AND LANGE-NIELSEN SYNDROME, JLNS1	220400		Sequencing	1580
Molecular Tests	KCNQ1OT1 and H19	604115 and 103280	BECKWITH-WIEDEMANN SYNDROME, BWS » EXOMPHALOS-MACROGLOSSIA-GIGANTISM SYNDROME	130650		KCNQ1OT1 and H19 Imprinting	350
Molecular Tests	KCNQ2 (POTASSIUM CHANNEL, VOLTAGE-GATED, KQT-LIKE SUBFAMILY, MEMBER 2)	602235	EPILEPSY, BENIGN NEONATAL, TYPE 1 » CONVULSIONS, BENIGN FAMILIAL NEONATAL, TYPE 1	121200		Sequencing and MLPA	1050
Molecular Tests	KCNQ2 (POTASSIUM CHANNEL, VOLTAGE-GATED, KQT-LIKE SUBFAMILY, MEMBER 2)	602235	MYOKYMIA WITH NEONATAL EPILEPSY » EPILEPSY, BENIGN NEONATAL, WITH MYOKYMIA » CONVULSIONS, BENIGN FAMILIAL NEONATAL, WITH MYOKYMIA	606437		Sequencing and MLPA	1050
Molecular Tests	KCNQ3 (POTASSIUM CHANNEL, VOLTAGE-GATED, KQT-LIKE SUBFAMILY, MEMBER 3)	602232	EPILEPSY, BENIGN NEONATAL, TYPE 2 » CONVULSIONS, BENIGN FAMILIAL NEONATAL, TYPE 2	121201		Sequencing	1250
Molecular Tests	KCNQ4 (POTASSIUM CHANNEL, VOLTAGE-GATED, KQT-LIKE SUBFAMILY, MEMBER 4)	603537	DEAFNESS, DFNA2 » DEAFNESS (AUTOSOMAL DOMINANT), NONSYNDROMIC SENSORINEURAL 2	600101		Sequencing	990
Molecular Tests	KCNV2 (POTASSIUM CHANNEL, VOLTAGE-GATED, SUBFAMILY 5, MEMBER 2)	607604	RETINAL CONE DYSTROPHY, TYPE 3B » CONE DYSTROPHY WITH NIGHT BLINDNESS AND SUPERNORMAL ROD RESPONSES, KCNV2-RELATED	610356		Sequencing	1000
Molecular Tests	KDM6A (LYSINE-SPECIFIC DEMETHYLASE 6A; UBIQUITOUSLY TRANSCRIBED TETRATRICOPEPTIDE REPEAT GENE ON X CHROMOSOME; UTX)	300128	KABUKI SYNDROME, TYPE 2	300867		Sequencing	980
Molecular Tests	KIAA0196 (STRUMPELLIN)	610657	FAMILIAL SPASTIC PARAPLEGIA 8 (AUTOSOMAL DOMINANT), SPG8	603563		Sequencing	2200
Molecular Tests	KIAA1840 (SPATACSIN)	610844	FAMILIAL SPASTIC PARAPLEGIA 11 (AUTOSOMAL RECESSIVE), SPG11	604360		Sequencing	2400
Molecular Tests	KIF1B (KINESIN FAMILY MEMBER 1B)	605995	CHARCOT-MARIE-TOOTH DISEASE, AXONAL, TYPE 2A, CMT2A	118210		Sequencing	2900
Molecular Tests	KIF21A (KINESIN FAMILY MEMBER 21A)	608283	FIBROSIS OF EXTRAOCULAR MUSCLES, CONGENITAL, TYPE 1, FEOM1, CFEOM3 » OPHTHALMOPLEGIA, CONGENITAL	135700		Hot Spots (Exons 2, 8, 20, 21)	250
Molecular Tests	KIF21A (KINESIN FAMILY MEMBER 21A)	608283	FIBROSIS OF EXTRAOCULAR MUSCLES, CONGENITAL, TYPE 3, FEOM3, CFEOM3	600638		Hot Spots (Exons 2, 8, 20, 21)	250
Molecular Tests	KIF5A (KINESIN FAMILY MEMBER 5A)	602821	FAMILIAL SPASTIC PARAPLEGIA 10 (AUTOSOMAL DOMINANT), SPG10	604187		Sequencing	1000
Molecular Tests	KIT (V-KIT HARDY-ZUCKERMAN 4 FELINE SARCOMA VIRAL ONCOGENE HOMOLOG, MAST CELL GROWTH FACTOR RECEPTOR, STEM CELL FACTOR RECEPTOR)	164920	PIEBALD TRAIT » PIEBALDISM	172800		Sequencing	820
Molecular Tests	KL (KLOTHO)	604824	TUMORAL CALCINOSIS, HYPERPHOSPHATEMIC, FAMILIAL » CALCINOSIS, TUMORAL, WITH HYPERPHOSPHATEMIA » LIPOCALCINOGRANULOMATOSIS » TEUTSCHLAENDER DISEASE, FAMILIAL	211900		Sequencing	745
Molecular Tests	KLF11 (KRUPPEL-LIKE FACTOR 11;TRANSFORMING GROWTH FACTOR-BETA-INDUCIBLE EARLY GROWTH RESPONSE 2; TIEG2)	603301	MATURITY-ONSET DIABETES OF THE YOUNG, TYPE 7, MODY7	610508		Sequencing	400
Molecular Tests	KLHL7 (KELCH-LIKE 7)	611119	RETINITIS PIGMENTOSA, TYPE 42, RP42	612943		Sequencing	580
Molecular Tests	KRAS (V-KI-RAS2 KIRSTEN RAT SARCOMA 2 VIRAL ONCOGENE HOMOLOG, KRAS2, KRAS1)	190070	LUNG CANCER, SQUAMOUS CELL			Sequencing	530
Molecular Tests	KRAS (V-KI-RAS2 KIRSTEN RAT SARCOMA 2 VIRAL ONCOGENE HOMOLOG, KRAS2, KRAS1)	190070	LEUKEMIA, ACUTE MYELOGENOUS			Sequencing	530
Molecular Tests	KRAS (V-KI-RAS2 KIRSTEN RAT SARCOMA 2 VIRAL ONCOGENE HOMOLOG, KRAS2, KRAS1)	190070	STOMACH CANCER			Sequencing	530

Molecular Tests	KRAS (V-KI-RAS2 KIRSTEN RAT SARCOMA 2 VIRAL ONCOGENE HOMOLOG, KRAS2, KRAS1)	190070	BLADDER CANCER, TRANSITIONAL CELL			Sequencing		150
Molecular Tests	KRAS (V-KI-RAS2 KIRSTEN RAT SARCOMA 2 VIRAL ONCOGENE HOMOLOG, KRAS2, KRAS1)	190070	DANDRUFF, SUSCEPTIBILITY TO			Sequencing		150
Molecular Tests	KRAS (V-KI-RAS2 KIRSTEN RAT SARCOMA 2 VIRAL ONCOGENE HOMOLOG, KRAS2, KRAS1)	190070	BREAST CANCER, FAMILIAL	114480		Sequencing		150
Molecular Tests	KRAS (V-KI-RAS2 KIRSTEN RAT SARCOMA 2 VIRAL ONCOGENE HOMOLOG, KRAS2, KRAS1)	190070	CARDIOFACIOCUTANEOUS SYNDROME, CFCV	115150		Sequencing		150
Molecular Tests	KRAS (V-KI-RAS2 KIRSTEN RAT SARCOMA 2 VIRAL ONCOGENE HOMOLOG, KRAS2, KRAS1)	190070	NOONAN SYNDROME, TYPE X	609942		Sequencing		150
Molecular Tests	KRIT1 (KREV INTERACTION TRAPPED 1; CCM1)	604214	CEREBRAL CAVERNOUS MALFORMATIONS, TYPE 1 » CAVERNOUS ANGIOMA, FAMILIAL, TYPE 1 » HEMORRHAGIC CUTANEOUS CAPILLARY-VENOUS MALFORMATIONS, ASSOCIATED WITH CEREBRAL CAPILLARY MALFORMATIONS, TYPE 1	116860		Sequencing		1600
Molecular Tests	KRT1 and KRT10 (KERATIN 1 and KERATIN 10)		ICHTHYOSIFORM ERYTHRODERMA, BULLOUS CONGENITAL » BROCQ SYNDROME » EPIDERMOLYTIC HYPERKERATOSIS » HYPERKERATOSIS, ICHTHYOSIFORM ERYTHRODERMA, BULLOUS CONGENITAL	113800		Hotspots		940
Molecular Tests	KRT1 and KRT10 (KERATIN 1 and KERATIN 10)		ICHTHYOSIS, BULLOSA OF SIEMENS	146800		Hotspots		940
Molecular Tests	KRT1 (KERATIN 1)	139350	CYCLIC ICHTHYOSIS WITH EPIDERMOLYTIC HYPERKERATOSIS » HYPERKERATOSIS, CYCLIC ICHTHYOSIS WITH EPIDERMOLYTIC HYPERKERATOSIS			Sequencing		1160
Molecular Tests	KRT1 (KERATIN 1)	139350	CYCLIC ICHTHYOSIS WITH EPIDERMOLYTIC HYPERKERATOSIS » HYPERKERATOSIS, CYCLIC ICHTHYOSIS WITH EPIDERMOLYTIC HYPERKERATOSIS			Hotspots		465
Molecular Tests	KRT1 (KERATIN 1)	139350	ICHTHYOSIFORM ERYTHRODERMA, BULLOUS CONGENITAL » BROCQ SYNDROME » EPIDERMOLYTIC HYPERKERATOSIS » HYPERKERATOSIS, ICHTHYOSIFORM ERYTHRODERMA, BULLOUS CONGENITAL	113800		Sequencing		1160
Molecular Tests	KRT1 (KERATIN 1)	139350	ICHTHYOSIFORM ERYTHRODERMA, BULLOUS CONGENITAL » BROCQ SYNDROME » EPIDERMOLYTIC HYPERKERATOSIS » HYPERKERATOSIS, ICHTHYOSIFORM ERYTHRODERMA, BULLOUS CONGENITAL	113800		Hotspots		465
Molecular Tests	KRT1 (KERATIN 1)	139350	ICHTHYOSIS, BULLOSA OF SIEMENS	146800		Sequencing		1160
Molecular Tests	KRT1 (KERATIN 1)	139350	ICHTHYOSIS, BULLOSA OF SIEMENS	146800		Hotspots		465
Molecular Tests	KRT1 (KERATIN 1)	139350	NONEPIDERMOLYTIC PALMOPLANTAR KERATODERMA » UNNA-THOST DISEASE » HYPERKERATOSIS, NONEPIDERMOLYTIC PALMOPLANTAR KERATODERMA	600962		Sequencing		1160
Molecular Tests	KRT1 (KERATIN 1)	139350	NONEPIDERMOLYTIC PALMOPLANTAR KERATODERMA » UNNA-THOST DISEASE » HYPERKERATOSIS, NONEPIDERMOLYTIC PALMOPLANTAR KERATODERMA	600962		Hotspots		465
Molecular Tests	KRT10 (KERATIN 10)	148080	ICHTHYOSIFORM ERYTHRODERMA, BULLOUS CONGENITAL » BROCQ SYNDROME » EPIDERMOLYTIC HYPERKERATOSIS » HYPERKERATOSIS, ICHTHYOSIFORM ERYTHRODERMA, BULLOUS CONGENITAL	113800		Sequencing		1120
Molecular Tests	KRT10 (KERATIN 10)	148080	ICHTHYOSIFORM ERYTHRODERMA, BULLOUS CONGENITAL » BROCQ SYNDROME » EPIDERMOLYTIC HYPERKERATOSIS » HYPERKERATOSIS, ICHTHYOSIFORM ERYTHRODERMA, BULLOUS CONGENITAL	113800		Hotspots		520
Molecular Tests	KRT10 (KERATIN 10)	148080	ICHTHYOSIS, BULLOSA OF SIEMENS	146800		Sequencing		1120
Molecular Tests	KRT10 (KERATIN 10)	148080	ICHTHYOSIS, BULLOSA OF SIEMENS	146800		Hotspots		520
Molecular Tests	KRT13 (KERATIN 13)	148065	WHITE SPONGE NEVUS	193900		Sequencing		1120
Molecular Tests	KRT14 (KERATIN 14)	148066	EPIDERMOLYSIS BULLOSA SIMPLEX (DOWLING-MEARA TYPE)	131760		Sequencing		1120
Molecular Tests	KRT14 (KERATIN 14)	148066	EPIDERMOLYSIS BULLOSA SIMPLEX (KOEBNER TYPE)	131900		Sequencing		1120
Molecular Tests	KRT14 (KERATIN 14)	148066	EPIDERMOLYSIS BULLOSA SIMPLEX (KOEBNER TYPE)	131900		Sequencing		1120
Molecular Tests	KRT16 and KRT17 (KERATIN 16 and KERATIN 17)		NONEPIDERMOLYTIC PALMOPLANTAR KERATODERMA » UNNA-THOST DISEASE » HYPERKERATOSIS, NONEPIDERMOLYTIC PALMOPLANTAR KERATODERMA	600962		Hotspots		890
Molecular Tests	KRT16 and KRT6A (KERATIN 16 and KERATIN 6A)		JADASSOHN-LEWANDOWSKY SYNDROME » PACHYONYCHIA CONGENITA, TYPE 1 » JADASSOHN-LEWANDOWSKY SYNDROME	167200		Hotspots		940

Molecular Tests	KRT16 (KERATIN 16)	148067	JADASSOHN-LEWANDOWSKY SYNDROME » PACHYONYCHIA CONGENITA, TYPE 1	167200		Sequencing	1120
Molecular Tests	KRT16 (KERATIN 16)	148067	NONEPIDERMOLYTIC PALMOPLANTAR KERATODERMA » UNNA-THOST DISEASE » HYPERKERATOSIS, NONEPIDERMOLYTIC PALMOPLANTAR KERATODERMA	600962		Sequencing	1120
Molecular Tests	KRT17 (KERATIN 17)	148069	JACKSON-LAWLER DISEASE » PACHYONYCHIA CONGENITA, TYPE 2	167210		Sequencing	1120
Molecular Tests	KRT17 (KERATIN 17)	148069	STEATOCYSTOMA MULTIPLEX	184500		Sequencing	1120
Molecular Tests	KRT17 (KERATIN 17)	148069	NONEPIDERMOLYTIC PALMOPLANTAR KERATODERMA » UNNA-THOST DISEASE » HYPERKERATOSIS, NONEPIDERMOLYTIC PALMOPLANTAR KERATODERMA	600962		Sequencing	1120
Molecular Tests	KRT2E (KERATIN 2E)	600194	ICHTHYOSIFORM ERYTHRODERMA, BULLOUS CONGENITAL » BROCQ SYNDROME » EPIDERMOLYTIC HYPERKERATOSIS » HYPERKERATOSIS, ICHTHYOSIFORM ERYTHRODERMA, BULLOUS CONGENITAL	113800		Sequencing	1120
Molecular Tests	KRT2E (KERATIN 2E)	600194	ICHTHYOSIS BULLOSA (SIEMENS)	146800		Sequencing	1120
Molecular Tests	KRT4 and KRT13 (KERATIN 4 and KERATIN 13)		WHITE SPONGE NEVUS	193900		Hotspots	940
Molecular Tests	KRT4 (KERATIN 4)	123940	WHITE SPONGE NEVUS	193900		Sequencing	1120
Molecular Tests	KRT5 (KERATIN 5)	148040	EPIDERMOLYSIS BULLOSA SIMPLEX (DOWLING-MEARA TYPE)	131760		Sequencing	1120
Molecular Tests	KRT5 (KERATIN 5)	148040	EPIDERMOLYSIS BULLOSA SIMPLEX (WEBER-COCKAYNE TYPE)	131800		Sequencing	1120
Molecular Tests	KRT5 (KERATIN 5)	148040	EPIDERMOLYSIS BULLOSA SIMPLEX (KOEBNER TYPE)	131900		Sequencing	1120
Molecular Tests	KRT6A (KERATIN 6A)	148041	JADASSOHN-LEWANDOWSKY SYNDROME » PACHYONYCHIA CONGENITA, TYPE 1	167200		Sequencing	1120
Molecular Tests	KRT6B (KERATIN 6B)	148042	JACKSON-LAWLER DISEASE » PACHYONYCHIA CONGENITA, TYPE 2	167210		Sequencing	1120
Molecular Tests	KRT9 (KERATIN 9)	144200	EPIDERMOLYTIC PALMOPLANTAR KERATODERMA » VORNER DISEASE » HYPERKERATOSIS, EPIDERMOLYTIC PALMOPLANTAR KERATODERMA	144200		Sequencing	1120
Molecular Tests	KRT9 (KERATIN 9)	144200	EPIDERMOLYTIC PALMOPLANTAR KERATODERMA » VORNER DISEASE » HYPERKERATOSIS, EPIDERMOLYTIC PALMOPLANTAR KERATODERMA	144200		Hotspots	520
Molecular Tests	L1 (L1CAM)	308840	CRASH SYNDROME » MENTAL RETARDATION, APHASIA, SHUFFLING GATE, AND ADDUCTED THUMBS SYNDROME, MASA	303350		Sequencing	Upon Request
Molecular Tests	L1 (L1CAM)	308840	CORPUS CALLOSUM AGENESIS (X-LINKED), ACC	304100		Sequencing	Upon Request
Molecular Tests	L1 (L1CAM)	308840	HYDROCEPHALUS DUE TO CONGENITAL STENOSIS OF AQUEDUCT OF SYLVIUS, HSAS » AQUEDUCTAL STENOSIS (X-LINKED)	307000		Sequencing	Upon Request
Molecular Tests	L1 (L1CAM)	308840	FAMILIAL SPASTIC PARAPLEGIA 1 (X-LINKED), SPG1	312900		Sequencing	Upon Request
Molecular Tests	LAMA2 (ALPHA-2 LAMININ, HEAVY CHAIN LAMININ 2, MEROSIN)	156225	MUSCULAR DYSTROPHY, CONGENITAL MEROSIN-DEFICIENT, TYPE 1A, MDC1A	607855		Sequencing	1200
Molecular Tests	LAMA2 (ALPHA-2 LAMININ, HEAVY CHAIN LAMININ 2, MEROSIN)	156225	MUSCULAR DYSTROPHY, CONGENITAL MEROSIN-DEFICIENT, TYPE 1A, MDC1A	607855		Deletion-Duplication Testing	650
Molecular Tests	LAMA3 (LAMININ, ALPHA-3)	600805	EPIDERMOLYSIS BULLOSA, GENERALIZED ATROPHIC BENIGN » EPIDERMOLYSIS BULLOSA JUNCTIONALIS, PROGRESSIVE » EPIDERMOLYSIS BULLOSA JUNCTIONALIS, SEVERE NONLETHAL » EPIDERMOLYSIS BULLOSA JUNCTIONALIS, DISENTIS TYPE	226650		Sequencing	2500
Molecular Tests	LAMA3 (LAMININ, ALPHA-3)	600805	EPIDERMOLYSIS BULLOSA LETALIS » EPIDERMOLYSIS BULLOSA, JUNCTIONAL, HERLITZ TYPE » HERLITZ-PEARSON TYPE EPIDERMOLYSIS BULLOSA	226700		Sequencing	2500
Molecular Tests	LAMA3, LAMB3 and LAMC2	150292	EPIDERMOLYSIS BULLOSA, GENERALIZED ATROPHIC BENIGN » EPIDERMOLYSIS BULLOSA JUNCTIONALIS, PROGRESSIVE » EPIDERMOLYSIS BULLOSA JUNCTIONALIS, SEVERE NONLETHAL » EPIDERMOLYSIS BULLOSA JUNCTIONALIS, DISENTIS TYPE	226650		6 Common Mutations in 3 Genes: LAMA3: R650X - LAMB3: R42X, Q243X, R635X, 77 BP Deletion - LAMC2: R95X	800
Molecular Tests	LAMA3, LAMB3 and LAMC2	150292	EPIDERMOLYSIS BULLOSA LETALIS » EPIDERMOLYSIS BULLOSA, JUNCTIONAL, HERLITZ TYPE » HERLITZ-PEARSON TYPE EPIDERMOLYSIS BULLOSA	226700		6 Common Mutations in 3 Genes: LAMA3: R650X - LAMB3: R42X, Q243X, R635X, 77 BP Deletion - LAMC2: R95X	800

Molecular Tests	LAMB2 (LAMININ, BETA-2)	150325	PIERSON SYNDROME » MICROCORIA-CONGENITAL NEPHROTIC SYNDROME » NEPHROTIC SYNDROME, CONGENITAL, WITH OR WITHOUT OCULAR ABNORMALITIES INCLUDED	609049		Sequencing	1560
Molecular Tests	LAMB3 (LAMININ, BETA-3)	150310	EPIDERMOLYSIS BULLOSA, GENERALIZED ATROPHIC BENIGN » EPIDERMOLYSIS BULLOSA JUNCTIONALIS, PROGRESSIVE » EPIDERMOLYSIS BULLOSA JUNCTIONALIS, SEVERE NONLETHAL » EPIDERMOLYSIS BULLOSA JUNCTIONALIS, DISENTIS TYPE	226650		Sequencing	1500
Molecular Tests	LAMB3 (LAMININ, BETA-3)	150310	EPIDERMOLYSIS BULLOSA LETALIS » EPIDERMOLYSIS BULLOSA, JUNCTIONAL, HERLITZ TYPE » HEMIDESMOSOMAL EPIDERMOLYSIS BULLOSA	226700		Sequencing	1500
Molecular Tests	LAMC2 (LAMININ, GAMMA-2)	150292	EPIDERMOLYSIS BULLOSA, GENERALIZED ATROPHIC BENIGN » EPIDERMOLYSIS BULLOSA JUNCTIONALIS, PROGRESSIVE » EPIDERMOLYSIS BULLOSA JUNCTIONALIS, SEVERE NONLETHAL » EPIDERMOLYSIS BULLOSA JUNCTIONALIS, DISENTIS TYPE	226650		Sequencing	1500
Molecular Tests	LAMC2 (LAMININ, GAMMA-2)	150292	EPIDERMOLYSIS BULLOSA LETALIS » EPIDERMOLYSIS BULLOSA, JUNCTIONAL, HERLITZ TYPE » HEMIDESMOSOMAL EPIDERMOLYSIS BULLOSA	226700		Sequencing	1500
Molecular Tests	LAMP2 (LYSOSOME-ASSOCIATED MEMBRANE PROTEIN 2)	309060	GLYCOGEN STORAGE DISEASE, TYPE 2B » GSD TYPE 2B » VACUOLAR CARDIOMYOPATHY AND MYOPATHY (X-LINKED) » LYSOSOMAL GLYCOGEN STORAGE DISEASE WITHOUT ACID MALTASE DEFICIENCY » GLYCOGEN STORAGE CARDIOMYOPATHY » PSEUDOGLYCOGENOSIS 2 » GLYCOGEN STORAGE DISEASE LIMITED TO THE HEART » ANTOPOL DISEASE	300257		Sequencing	770
Molecular Tests	LARGE (ACETYLGLUCOSAMINYLTRANSFERASE-LIKE PROTEIN LARGE; LARGE-GLYCOSYLTRANSFERASE)	603590	WALKER-WARBURG SYNDROME » HYDROCEPHALUS, AGYRIA, AND RETINAL DYSPLASIA » HARD SYNDROME	236670		Sequencing	1320
Molecular Tests	LARGE (ACETYLGLUCOSAMINYLTRANSFERASE-LIKE PROTEIN LARGE; LARGE-GLYCOSYLTRANSFERASE)	603590	MUSCULAR DYSTROPHY, CONGENITAL, TYPE 1D	608840		Sequencing	1320
Molecular Tests	LCA5 (LEBERCILIN)	611408	LEBER CONGENITAL AMAUROSIS, TYPE 5, LCA5	604537		Sequencing	520
Molecular Tests	LCAT (LECITHINCHOLESTEROL ACYLTRANSFERASE)	606967	FISH-EYE DISEASE	136120		Sequencing	450
Molecular Tests	LCAT (LECITHINCHOLESTEROL ACYLTRANSFERASE)	606967	LECITHINCHOLESTEROL ACYLTRANSFERASE DEFICIENCY » LCAT DEFICIENCY » NORUM DISEASE	245900		Sequencing	450
Molecular Tests	LCT (LACTASE)	603202	LACTOSE INTOLERANCE » HYPOLACTASIA » DISACCHARIDE INTOLERANCE » ADULT LACTASE DEFICIENCY	223100		1 Mutation T13910C	260
Molecular Tests	LDB3 (LIM DOMAIN-BINDING 3, ZASP, CYPHER)	605906	CARDIOMYOPATHY, DILATED, TYPE 1C » CARDIOMYOPATHY, DILATED, WITH LEFT VENTRICULAR NONCOMPACTION	601493		Sequencing	1100
Molecular Tests	LDB3 (LIM DOMAIN-BINDING 3, ZASP, CYPHER)	605906	MYOPATHY, MYOFIBRILLAR, ZASP-RELATED	609452		Sequencing	1100
Molecular Tests	LDHA (LACTATE DEHYDROGENASE A)	150000	GLYCOGEN STORAGE DISEASE, TYPE 11 » LACTATE DEHYDROGENASE A DEFICIENCY	612933		Sequencing	800
Molecular Tests	LDLR (LDL RECEPTOR)	606945	HYPERCHOLESTEROLEMIA (AUTOSOMAL DOMINANT)	143890		Whole LDLR Gene + 5 Mutations in APOB	1000
Molecular Tests	LDLR (LDL RECEPTOR)	606945	HYPERCHOLESTEROLEMIA (AUTOSOMAL DOMINANT)	143890		Deletion/Duplication Testing	550
Molecular Tests	LDLRAP1 (LOW DENSITY LIPOPROTEIN RECEPTOR ADAPTOR PROTEIN 1; LDLR ADAPTOR PROTEIN 1; ARH)	605747	HYPERCHOLESTEROLEMIA (AUTOSOMAL RECESSIVE), ARH	107811		Sequencing	500
Molecular Tests	LEMD3 (LEM DOMAIN-CONTAINING 3; MAN1)	607844	MELORHEOSTOSIS » MELORHEOSTOSIS WITH OSTEOPOIKILOSIS	155950		Sequencing	850

Molecular Tests	LEMD3 (LEM DOMAIN-CONTAINING 3, MAN1)	607844	BUSCHKE-OLLENDORFF SYNDROME » DERMATOOSTEOPOIKILOSIS » DERMATOFIBROSIS, DISSEMINATED, WITH OSTEOPOIKILOSIS » DERMATOFIBROSIS LENTICULARIS DISSEMINATA WITH OSTEOPOIKILOSIS » OSTEOPATHIA CONDENSANS DISSEMINATA	166700		Sequencing	850
Molecular Tests	LEP (LEPTIN)	164160	OBESITY, SEVERE, DUE TO LEPTIN DEFICIENCY » OBESITY, MORBID, WITH HYPOGONADISM	601007		Sequencing	450
Molecular Tests	LEPR (LEPTIN RECEPTOR)	601007	OBESITY, SEVERE, DUE TO LEPTIN DEFICIENCY » OBESITY, MORBID, WITH HYPOGONADISM	601007		Sequencing	1450
Molecular Tests	LEPRE1 (LEUCINE- AND PROLINE-ENRICHED PROTEOGLYCAN 1, LEPRECAN, PROLYL 3-HYDROXYLASE 1)	610339	OSTEOGENESIS IMPERFECTA, TYPE 8, OI8	610915		Sequencing	550
Molecular Tests	LFNG (LUNATIC FRINGE; FRINGE, DROSOPHILA, HOMOLOG OF, LUNATIC)	602576	SPONDYLOCOSTAL DYSOSTOSIS, TYPE 3 (AUTOSOMAL RECESSIVE)	609813		Sequencing	670
Molecular Tests	LGI1 (LEUCINE-RICH GENE, GLIOMA-INACTIVATED, 1; EPITEMPIN)	604619	EPILEPSY, LATERAL TEMPORAL LOBE (AUTOSOMAL DOMINANT), ADLTE » EPILEPSY, PARTIAL, WITH AUDITORY FEATURES, ADPEAF	600512		Sequencing	750
Molecular Tests	LHCGR (LUTEINIZING HORMONE / CHORIOGONADOTROPIN RECEPTOR, LUTROPIN-CHORIOGONADOTROPIN RECEPTOR	152790	LEYDIG CELL HYPOPLASIA WITH MALE PSEUDOHERMAPHRODITISM			Sequencing	1670
Molecular Tests	LHCGR (LUTEINIZING HORMONE / CHORIOGONADOTROPIN RECEPTOR, LUTROPIN-CHORIOGONADOTROPIN RECEPTOR	152790	HYPERGONADOTROPIC HYPOGONADISM, FEMALE			Sequencing	1670
Molecular Tests	LHCGR (LUTEINIZING HORMONE / CHORIOGONADOTROPIN RECEPTOR, LUTROPIN-CHORIOGONADOTROPIN RECEPTOR	152790	MICROPENIS			Sequencing	1670
Molecular Tests	LHCGR (LUTEINIZING HORMONE / CHORIOGONADOTROPIN RECEPTOR, LUTROPIN-CHORIOGONADOTROPIN RECEPTOR)	152790	PRECOCIOUS PUBERTY, MALE-LIMITED » SEXUAL PRECOCITY, FAMILIAL, GONADOTROPIN-INDEPENDENT » TESTOTOXICOSIS, FAMILIAL	176410		Sequencing	1670
Molecular Tests	LHX3 (LIM HOMEOBOX GENE 3, LIM3)	600577	PITUITARY DWARFISM 3 » PANHYPOPITUITARISM » ATELIOTIC DWARFISM WITH HYPOGONADISM » HANHART DWARFISM » PITUITARY HORMONE DEFICIENCY, COMBINED	262600		Sequencing	540
Molecular Tests	LHX4 (LIM HOMEOBOX GENE 4, LHX4, LHX4/IGHG1 FUSION GENE)	602146	SHORT STATURE, PITUITARY AND CEREBELLAR DEFECTS, AND SMALL SELLA TURCICA	606606		Sequencing	840
Molecular Tests	LIFR (LEUKEMIA INHIBITORY FACTOR RECEPTOR)	151443	STUVE-WIEDEMANN SYNDROME » SCHWARTZ-JAMPEL SYNDROME, TYPE 2 » SCHWARTZ-JAMPEL SYNDROME, NEONATAL	601559		Sequencing	2400
Molecular Tests	LIFR (LEUKEMIA INHIBITORY FACTOR RECEPTOR)	151443	STUVE-WIEDEMANN SYNDROME » SCHWARTZ-JAMPEL SYNDROME, TYPE 2 » SCHWARTZ-JAMPEL SYNDROME, NEONATAL	601559		653-654insT	400
Molecular Tests	LIPA (LIPASE A, LYSOSOMAL ACID;CHOLESTEROL ESTER HYDROLASE)	278000	WOLMAN DISEASE » LYSOSOMAL ACID LIPASE DEFICIENCY » LIPA DEFICIENCY » CHOLESTERYL ESTER STORAGE DISEASE	278000		Sequencing	900
Molecular Tests	LIPC (HEPATIC LIPASE, LIPH, HEPATIC TRIGLYCERIDE LIPASE, HTGL)	151670	HEPATIC LIPASE DEFICIENCY	151670		Sequencing	850
Molecular Tests	LITAF (LIPOPOLYSACCHARIDE-INDUCED TUMOR NECROSIS FACTOR-ALPHA FACTOR)	603795	CHARCOT-MARIE-TOOTH DISEASE, DEMYELINATING, TYPE 1C, CMT1C, HMSN1C	601098		Sequencing	400
Molecular Tests	LMBRD1 (LMBR1 DOMAIN-CONTAINING PROTEIN 1; LMBD1; NES-INTERACTING PROTEIN; NESI)	612625	METHYLMALONIC ACIDURIA AND HOMOCYSTINURIA, cblF TYPE » VITAMIN B12 LYSOSOMAL RELEASE DEFECT	277380		Sequencing	1790
Molecular Tests	LMNA (LAMIN A/C)	150330	DILATED CARDIOMYOPATHY WITH CARDIAC CONDUCTION DEFECTS, CMD1A	115200		Sequencing	700
Molecular Tests	LMNA (LAMIN A/C)	150330	LIPODYSTROPHY, FAMILIAL PARTIAL, TYPE 2, FPLD2 » LIPODYSTROPHY, DUNNIGAN TYPE	151660		Sequencing	700
Molecular Tests	LMNA (LAMIN A/C)	150330	MUSCULAR DYSTROPHY, LIMB-GIRDLE, TYPE 1B, LGMD1B	159001		Sequencing	700
Molecular Tests	LMNA (LAMIN A/C)	150330	HUTCHINSON-GILFORD PROGERIA SYNDROME	176670		Sequencing	700
Molecular Tests	LMNA (LAMIN A/C)	150330	EMERY–DREYFUSS MUSCULAR DYSTROPHY (AUTOSOMAL DOMINANT), EDMD2	181350		Sequencing	700
Molecular Tests	LMNA (LAMIN A/C)	150330	MANDIBULOACRAL DYSPLASIA WITH TYPE A LIPODYSTROPHY, MADA » CRANIOMANDIBULAR DERMATODYSOSTOSIS	248370		Sequencing	700
Molecular Tests	LMNA (LAMIN A/C)	150330	WERNER SYNDROME	277700		Sequencing	700
Molecular Tests	LMNA (LAMIN A/C)	150330	EMERY–DREYFUSS MUSCULAR DYSTROPHY (AUTOSOMAL RECESSIVE), EDMD3	604949		Sequencing	700
Molecular Tests	LMNA (LAMIN A/C)	150330	CHARCOT-MARIE-TOOTH DISEASE, AXONAL, TYPE 2B1, CMT2B1	605588		Sequencing	700
Molecular Tests	LMNA (LAMIN A/C)	150330	LIPOATROPHY WITH DIABETES, HEPATIC STEATOSIS, HYPERTROPHIC CARDIOMYOPATHY AND LEUKOMELANODERMIC PAPULES, LDHCP	608056		Sequencing	700
Molecular Tests	LMNB1 (LAMIN B1)	150340	LEUKODYSTROPHY, DEMYELINATING, ADULT-ONSET (AUTOSOMAL DOMINANT), ADLD » PELIZAEUS-MERZBACHER DISEASE (AUTOSOMAL DOMINANT) OR LATE-ONSET TYPE, FORMERLY	169500		Sequencing	460
Molecular Tests	LMX1B	602575	NAIL-PATELLA SYNDROME, NPS	161200		Sequencing	550
Molecular Tests	LMX1B	602575	NAIL-PATELLA SYNDROME, NPS	161200		Deletion-Duplication Testing	Upon Request
Molecular Tests	LONG QT PANEL 1: KCNQ1, HERG		LONG QT SYNDROME			9 Exons with Mutation Hotspots	850

Molecular Tests	LPIN1 (LIPIN 1; PHOSPHATIDIC ACID PHOSPHOHYDROLASE 1; PAP1)	605518	MYOGLOBINURIA, ACUTE RECURRENT, (AUTOSOMAL RECESSIVE) » MYOGLOBINURIA, FAMILIAL PAROXYSMAL PARALYTIC » RHABDOMYOLYSIS, ACUTE RECURRENT	268200		Sequencing	1130
Molecular Tests	LPL (LIPOPROTEIN LIPASE)	238600	HYPERLIPOPROTEINEMIA TYPE 1 » LIPOPROTEIN LIPASE DEFICIENCY » HYPERCHYLOMICRONEMIA	238600		Sequencing	650
Molecular Tests	LPL (LIPOPROTEIN LIPASE)	238600	HYPERLIPOPROTEINEMIA TYPE 1 » LIPOPROTEIN LIPASE DEFICIENCY » HYPERCHYLOMICRONEMIA	238600		Deletion-Duplication Testing	350
Molecular Tests	LRAT (LECITHIN RETINOL ACYLTRANSFERASE)	604863	RETINAL DYSTROPHY, EARLY-ONSET SEVERE	604863		Sequencing	440
Molecular Tests	LRP2 (LOW DENSITY LIPOPROTEIN RECEPTOR-RELATED PROTEIN 2; GLYCOPROTEIN 330; MEGALIN)	600073	DONNAI-BARROW SYNDROME » FACIOOCULOACOUSTICORENAL SYNDROME » DBS/FOAR SYNDROME » DIAPHRAGMATIC HERNIA, EXOMPHALOS, ABSENT CORPUS CALLOSUM, HYPERTELORISM, MYOPIA, SENSORINEURAL DEAFNESS, AND PROTEINURIA	222448		Sequencing	2400
Molecular Tests	LRPPRC (LEUCINE-RICH PPR MOTIF-CONTAINING PROTEIN)	607544	LEIGH SYNDROME, FRENCH CANADIAN TYPE » CYTOCHROME c OXIDASE DEFICIENCY, FRENCH CANADIAN TYPE » LEIGH SYNDROME, SAGUENAY-LAC-SAINT-JEAN TYPE	220111		Sequencing	1900
Molecular Tests	LRRK2 (LEUCINE-RICH REPEAT KINASE)	609007	PARKINSON DISEASE, TYPE 8, PARK8	607060		Exon 41	350
Molecular Tests	LRRK2 (LEUCINE-RICH REPEAT KINASE)	609007	PARKINSON DISEASE, TYPE 8, PARK8	607060		Sequencing	2500
Molecular Tests	LTBP4 (LATENT TRANSFORMING GROWTH FACTOR-BETA-BINDING PROTEIN 4)	604710	CUTIS LAXA WITH SEVERE PULMONARY, GASTROINTESTINAL, AND URINARY ABNORMALITIES » URBAN-RIFKIN-DAVIS SYNDROME	613177		Sequencing	850
Molecular Tests	LYST (LYSOSOMAL TRAFFICKING REGULATOR; CHS1)	606897	CHEDIAK-HIGASHI SYNDROME	214500		Sequencing	1100
Molecular Tests	MAK (MALE GERM CELL-ASSOCIATED KINASE)	154235	RETINITIS PIGMENTOSA, TYPE 62, RP62	614181		Sequencing	680
Molecular Tests	MAN2B1 (MANNOSIDASE, ALPHA, CLASS 2B, MEMBER 1; MANB)	609458	ALPHA-MANNOSIDOSIS	248500		Sequencing	2190
Molecular Tests	MANBA (MANNOSIDASE, BETA A, LYSOSOMAL)	609489	MANNOSIDOSIS, BETA A, LYSOSOMAL » BETA-MANNOSIDASE DEFICIENCY	248510		Sequencing	1630
Molecular Tests	MAP2K1 (MITOGEN-ACTIVATED PROTEIN KINASE KINASE 1, MEK1)	176872	CARDIOFACIOCUTANEOUS SYNDROME, CFC » CFC SYNDROME	115150		Sequencing	680
Molecular Tests	MAP2K2 (MITOGEN-ACTIVATED PROTEIN KINASE KINASE 2, MEK2)	601263	CARDIOFACIOCUTANEOUS SYNDROME, CFC » CFC SYNDROME	115150		Sequencing	680
Molecular Tests	MAPT (MICROTUBULE-ASSOCIATED PROTEIN TAU)	157140	17q21.31 MICRODELETION SYNDROME			Deletions	350
Molecular Tests	MAPT (MICROTUBULE-ASSOCIATED PROTEIN TAU)	157140	PICK DISEASE OF BRAIN	172700		Sequencing	1500
Molecular Tests	MAPT (MICROTUBULE-ASSOCIATED PROTEIN TAU)	157140	FRONTOTEMPORAL DEMENTIA » MULTIPLE SYSTEM TAUOPATHY WITH PRESENILE DEMENTIA » DISINHIBITION-DEMENTIA-PARKINSONISM-AMYOTROPHY COMPLEX » WILHELMSEN-LYNCH DISEASE » FRONTOTEMPORAL DEMENTIA-AMYOTROPHIC LATERAL SCLEROSIS	600274		Sequencing	1500
Molecular Tests	MAPT (MICROTUBULE-ASSOCIATED PROTEIN TAU)	157140	SUPRANUCLEAR PALSY, PROGRESSIVE, 1 » STEELE-RICHARDSON-OLSZEWSKI SYNDROME	601104		Sequencing	1500
Molecular Tests	MAT1A (METHIONINE ADENOSYLTRANSFERASE I, ALPHA; S-ADENOSYLMETHIONINE SYNTHETASE 1, SAMS1)	610550	METHIONINE ADENOSYLTRANSFERASE DEFICIENCY » MAT DEFICIENCY » HYPERMETHIONINEMIA, ISOLATED PERSISTENT	250850		Sequencing	600
Molecular Tests	MATN3 (MATRILIN 3)	602109	EPIPHYSEAL DYSPLASIA, MULTIPLE, TYPE 5, EDM5 » MICROEPIPHYSEAL DYSPLASIA, BILATERAL, HEREDITARY	607078		Sequencing	Upon Request
Molecular Tests	MATN3 (MATRILIN 3)	602109	SPONDYLOEPIMETAPHYSEAL DYSPLASIA, MATRILIN-3 RELATED	608728		Sequencing	Upon Request
Molecular Tests	MBL2 (LECTIN, MANNOSE-BINDING, SOLUBLE, 2, MANNAN-BINDING PROTEIN, COLLECTIN 1)	154545	SUSCEPTIBILITY TO INFECTION			Sequencing	500
Molecular Tests	MBL2 (LECTIN, MANNOSE-BINDING, SOLUBLE, 2, MANNAN-BINDING PROTEIN, COLLECTIN 1)	154545	SUSCEPTIBILITY TO INFECTION			5 Mutations: ARG52CYS, GLY54ASP, GLY57GLU, -550G>C and -221G>C	250
Molecular Tests	MBL2 (LECTIN, MANNOSE-BINDING, SOLUBLE, 2, MANNAN-BINDING PROTEIN, COLLECTIN 1)	154545	MANNOSE-BINDING PROTEIN DEFICIENCY			Sequencing	500
Molecular Tests	MBL2 (LECTIN, MANNOSE-BINDING, SOLUBLE, 2, MANNAN-BINDING PROTEIN, COLLECTIN 1)	154545	MANNOSE-BINDING PROTEIN DEFICIENCY			5 Mutations: ARG52CYS, GLY54ASP, GLY57GLU, -550G>C and -221G>C	250
Molecular Tests	MBTPS2 (MEMBRANE-BOUND TRANSCRIPTION FACTOR PROTEASE, SITE 2; SITE-2 PROTEASE; S2P)	300293	IFAP SYNDROME WITH OR WITHOUT BRESHECK SYNDROME	308205		Sequencing	750
Molecular Tests	MBTPS2 (MEMBRANE-BOUND TRANSCRIPTION FACTOR PROTEASE, SITE 2; SITE-2 PROTEASE; S2P)	300293	KERATOSIS FOLLICULARIS SPINULOSA DECALVANS, X-LINKED » KERATOSIS FOLLICULARIS SPINULOSA DECALVANS CUM OPHIASIS	308800		Sequencing	750
Molecular Tests	MC2R (MELANOCORTIN 2 RECEPTOR; ACTH RECEPTOR)	607397	GLUCOCORTICOID DEFICIENCY 1 » ADRENAL UNRESPONSIVENESS TO ACTH » ACTH RESISTANCE	202200		Sequencing	540

Molecular Tests	MC4R (MELANOCORTIN 4 RECEPTOR)	155541	OBESITY	601665		Sequencing	300
Molecular Tests	MCCC1 (3-@METHYLCROTONYL-CoA CARBOXYLASE 1; MCCA)	609014	3-@METHYLCROTONYL-CoA CARBOXYLASE 1 DEFICIENCY » METHYLCROTONYLGLYCINURIA TYPE 1	210200		MCCC1 and MCCC2	1460
Molecular Tests	MCCC1 (3-@METHYLCROTONYL-CoA CARBOXYLASE 1; MCCA)	609014	3-@METHYLCROTONYL-CoA CARBOXYLASE 1 DEFICIENCY » METHYLCROTONYLGLYCINURIA TYPE 1	210200		Sequencing	930
Molecular Tests	MCCC2 (3-@METHYLCROTONYL-CoA CARBOXYLASE 2; MCCB)	605195	3-@METHYLCROTONYL-CoA CARBOXYLASE 2 DEFICIENCY » METHYLCROTONYLGLYCINURIA TYPE 2	210210		MCCC1 and MCCC2	1460
Molecular Tests	MCCC2 (3-@METHYLCROTONYL-CoA CARBOXYLASE 2; MCCB)	605195	3-@METHYLCROTONYL-CoA CARBOXYLASE 2 DEFICIENCY » METHYLCROTONYLGLYCINURIA TYPE 2	210210		Sequencing	850
Molecular Tests	MCEE (METHYLMALONYL-CoA EPIMERASE; METHYLMALONYL-CoA RACEMASE)	608419	METHYLMALONYL-CoA EPIMERASE DEFICIENCY » METHYLMALONIC ACIDURIA, TYPE 3	251120		Sequencing	410
Molecular Tests	MCOLN1 (ML4, MUCOLYPIN)	605248	MUCOLIPIDOSIS, TYPE 4, ML4	252650		2 Common Askhenazi Mutations: IVS3-2A>G and 6.4 kb Deletion	570
Molecular Tests	MCP (MEMBRANE COFACTOR PROTEIN, MEASLES VIRUS RECEPTOR, CD46, MIC10, TLX, TRA2.10)	120920	HEMOLYTIC-UREMIC SYNDROME, HUS » COMBINED DEFICIENCY OF FACTOR H AND FACTOR H-LIKE 1	235400		Sequencing	820
Molecular Tests	MCP (MEMBRANE COFACTOR PROTEIN, MEASLES VIRUS RECEPTOR, CD46, MIC10, TLX, TRA2.10)	120920	HEMOLYTIC-UREMIC SYNDROME, HUS » COMBINED DEFICIENCY OF FACTOR H AND FACTOR H-LIKE 1	235400		Deletion-Duplication Testing	Upon Request
Molecular Tests	MECP2	300005	NONSPECIFIC MENTAL RETARDATION (X-LINKED), MRX16 » MENTAL RETARDATION, NONSPECIFIC (X-LINKED), TYPE16, MRX16			Sequencing	690
Molecular Tests	MECP2	300005	NONSPECIFIC MENTAL RETARDATION (X-LINKED), MRX16 » MENTAL RETARDATION, NONSPECIFIC (X-LINKED), TYPE16, MRX16			Deletion Analysis (MLPA)	510
Molecular Tests	MECP2	300005	MENTAL RETARDATION WITH PSYCHOSIS, PYRAMIDAL SIGNS, AND MACROORCHIDISM	300055		Sequencing	690
Molecular Tests	MECP2	300005	MENTAL RETARDATION WITH PSYCHOSIS, PYRAMIDAL SIGNS, AND MACROORCHIDISM	300055		Deletion Analysis (MLPA)	510
Molecular Tests	MECP2	300005	RETT SYNDROME	312750		Sequencing	690
Molecular Tests	MECP2	300005	RETT SYNDROME	312750		Deletion Analysis (MLPA)	510
Molecular Tests	MED12 (MEDIATOR OF RNA POLYMERASE II TRANSCRIPTION, SUBUNIT 12, S. CEREVISIAE, HOMOLOG OF; TRINUCLEOTIDE REPEAT-CONTAINING GENE 11; THYROID HORMONE RECEPTOR-ASSOCIATED PROTEIN, 230-KD SUBUNIT)	300188	OPITZ-KAVEGGIA SYNDROME » FG SYNDROME, TYPE 1 » MENTAL RETARDATION, LARGE HEAD, IMPERFORATE ANUS, CONGENITAL HYPOTONIA, AND PARTIAL AGENESIS OF CORPUS CALLOSUM	305450		Exon 21 and 22, including the R961W Mutation	900
Molecular Tests	MEF2C (MADS BOX TRANSCRIPTION ENHANCER FACTOR 2, POLYPEPTIDE C)	600662	MENTAL RETARDATION, STEREOTYPIC MOVEMENTS, EPILEPSY, AND/OR CEREBRAL MALFORMATIONS	613443		Sequencing	1540
Molecular Tests	MEFV	249100	FAMILIAL MEDITERRANEAN FEVER, FMF	249100		Sequencing	750
Molecular Tests	MEFV	249100	FAMILIAL MEDITERRANEAN FEVER, FMF	249100		Exons 2, 3, 5, 10	350
Molecular Tests	MEN1 (MENIN)	131100	MULTIPLE ENDOCRINE NEOPLASIA, TYPE 1, MEN1	131100		Sequencing	590
Molecular Tests	MERTK (MER TYROSINE KINASE PROTOONCOGENE)	604705	RETINITIS PIGMENTOSA, MERTK-RELATED	604705		Sequencing	750
Molecular Tests	MESP2 (MESODERM POSTERIOR 2)	605195	SPONDYLOCOSTAL DYSOSTOSIS, TYPE 2 (AUTOSOMAL RECESSIVE)	608681		Sequencing	300
Molecular Tests	MEST (PEG1)	601029	SILVER-RUSSELL SYNDROME	180860		UPD Study by Methylation Analysis	195
Molecular Tests	MET (MET PROTOONCOGENE; HEPATOCYTE GROWTH FACTOR RECEPTOR)	164860	HEPATOCELLULAR CARCINOMA, CHILDHOOD TYPE			Sequencing	2150
Molecular Tests	MET (MET PROTOONCOGENE; HEPATOCYTE GROWTH FACTOR RECEPTOR)	164860	RENAL CELL CARCINOMA, PAPILLARY	605074		Sequencing	2150
Molecular Tests	MFN2 (MITOFUSIN 2)	608507	CHARCOT-MARIE-TOOTH DISEASE, AXONAL, TYPE 2A, CMT2A	118210		Sequencing	990
Molecular Tests	MGAT2 (ALPHA-1,6-@MANNOSYL-GLYCOPROTEIN BETA-1,2-N-ACETYLGLUCOSAMINYLTRANSFERASE)	602616	CONGENITAL DISORDER OF GLYCOSYLATION, TYPE 2A, CDG2A	212066		Sequencing	380
Molecular Tests	MICROCEPHALY PANEL: MCPH1, CENPJ, STIL, CDK5RAP2		MICROCEPHALY			Sequencing and Deletion-Duplication Testing	4130
Molecular Tests	MID1 (MIDLINE 1, MIDIN, MIDLINE 1 RING FINGER GENE)	300000	OPITZ SYNDROME » OPITZ G/BBB SYNDROME (X-LINKED) » OPITZ SYNDROME (X-LINKED) » OPITZ G SYNDROME	300000		Sequencing	850
Molecular Tests	MITF	156845	WAARDENBURG SYNDROME, TYPE 2, WS2	193510		Sequencing	680
Molecular Tests	MITF	156845	WAARDENBURG SYNDROME, TYPE 2, WS2	193510		Deletion Analysis (MLPA)	500
Molecular Tests	MITOCHONDRIAL MUTATIONS				Listed Separately: See Mitochondrial Molecular Tests Table	Sequencing	
Molecular Tests	MKS1	609883	MECKEL SYNDROME, TYPE 1 » DYSENCEPHALIA SPLANCHNOCYSTICA » GRUBER SYNDROME » MECKEL-GRUBER SYNDROME	249000		Sequencing	1200
Molecular Tests	MLC1	605908	MEGALENCEPHALIC LEUKOENCEPHALOPATHY WITH SUBCORTICAL CYSTS	605908		Sequencing and Deletion-Duplication Testing	910
Molecular Tests	MLH1	120436	COLORECTAL CANCER, HEREDITARY NONPOLYPOSIS, HNPCC, TYPE 2 » LYNCH CANCER FAMILY SYNDROME, TYPE 2	114400		Sequencing	970

Molecular Tests	MLH1	120436	COLORECTAL CANCER, HEREDITARY NONPOLYPOSIS, HNPCC, TYPE 1 » LYNCH CANCER FAMILY SYNDROME, TYPE 1	114500		Sequencing	970
Molecular Tests	MLL2 (MYELOID/LYMPHOID OR MIXED LINEAGE LEUKEMIA 2; ALR)	602113	KABUKI SYNDROME » NIIKAWA-KUROKI SYNDROME	147920		Sequencing	2910
Molecular Tests	MLL2 (MYELOID/LYMPHOID OR MIXED LINEAGE LEUKEMIA 2; ALR)	602113	KABUKI SYNDROME » NIIKAWA-KUROKI SYNDROME	147920		Deletion-Duplication Testing	570
Molecular Tests	MLYCD (MALONYL-CoA DECARBOXYLASE)	606761	MALONYL-CoA DECARBOXYLASE DEFICIENCY	248360		Sequencing	810
Molecular Tests	MMAA	607481	METHYLMALONIC ACIDURIA, cblA TYPE » METHYLMALONIC ACIDURIA, VITAMIN B12-RESPONSIVE, DUE TO DEFECT IN SYNTHESIS OF ADENOSYLCOBALAMIN, cblA TYPE	251100		Sequencing	540
Molecular Tests	MMAB (COBALAMIN ADENOSYLTRANSFERASE)	607568	METHYLMALONIC ACIDURIA, cblB TYPE » METHYLMALONIC ACIDURIA, VITAMIN B12-RESPONSIVE, DUE TO DEFECT IN SYNTHESIS OF ADENOSYLCOBALAMIN, cblB TYPE	251110		Sequencing	720
Molecular Tests	MMACHC	609831	METHYLMALONIC ACIDURIA AND HOMOCYSTINURIA, cblC TYPE » VITAMIN B12 METABOLIC DEFECT WITH COMBINED DEFICIENCY OF METHYLMALONYL-CoA MUTASE AND HOMOCYSTEINE:METHYLTETRAHYDROFOLATE METHYLTRANSFERASE	277400		Sequencing	410
Molecular Tests	MMADHC	611935	METHYLMALONIC ACIDURIA AND HOMOCYSTINURIA, cblD TYPE » HOMOCYSTINURIA, cblD TYPE	277410		Sequencing	820
Molecular Tests	MNX1 (MOTOR NEURON AND PANCREAS HOMEOBOX 1; HLXB9; HOMEOBOX GENE HB9)	142994	CURRARINO TRIAD » SACRAL AGENESIS SYNDROME » SACRAL AGENESIS, HEREDITARY, WITH PRESACRAL MASS, ANTERIOR MENINGOCELE, AND/OR TERATOMA, AND ANORECTAL MALFORMATION	176450		Sequencing	650
Molecular Tests	MOCS1 (MOLYBDENUM COFACTOR SYNTHESIS GENE 1)	603707	MOLYBDENUM COFACTOR DEFICIENCY » SULFITE OXIDASE, XANTHINE DEHYDROGENASE, AND ALDEHYDE OXIDASE, COMBINED DEFICIENCY OF	252150		Sequencing	1140
Molecular Tests	MOCS2 (MOLYBDENUM COFACTOR SYNTHESIS GENE 2)	603708	MOLYBDENUM COFACTOR DEFICIENCY » SULFITE OXIDASE, XANTHINE DEHYDROGENASE, AND ALDEHYDE OXIDASE, COMBINED DEFICIENCY OF	252150		Sequencing	890
Molecular Tests	MPDU1 (MANNOSE-P-DOLICHOL UTILIZATION DEFECT 1)	609458	CONGENITAL DISORDER OF GLYCOSYLATION, TYPE 1F, CDG1F	609180		Sequencing	730
Molecular Tests	MPI (MANNOSEPHOSPHATE ISOMERASE; PHOSPHOMANNOSE ISOMERASE 1)	604041	CONGENITAL DISORDER OF GLYCOSYLATION, TYPE 1B, CDG1B » SAGUENAY-LAC SAINT-JEAN SYNDROME, SLSJ SYNDROME » MANNOSEPHOSPHATE ISOMERASE DEFICIENCY » PROTEIN-LOSING ENTEROPATHY, HEPATIC FIBROSIS SYNDROME	602579		Sequencing	580
Molecular Tests	MPL (MYELOPROLIFERATIVE LEUKEMIA VIRUS ONCOGENE, TPOR)	159530	THROMBOCYTHEMIA, ESSENTIAL » THROMBOCYTOSIS (AUTOSOMAL DOMINANT)	187950		Exon 10, including W515L Mutation	450
Molecular Tests	MPL (MYELOPROLIFERATIVE LEUKEMIA VIRUS ONCOGENE, TPOR)	159530	MYELOFIBROSIS WITH MYELOID METAPLASIA, INCLUDED	254450		Exon 10, including W515L Mutation	450
Molecular Tests	MPV17 (MPV17, MOUSE, HOMOLOG OF)	137960	MITOCHONDRIAL DNA DEPLETION SYNDROME, HEPATOCEREBRAL FORM	251880		Sequencing	450
Molecular Tests	MPZ (MYELIN PROTEN ZERO, P0)	159440	CHARCOT-MARIE-TOOTH DISEASE, DEMYELINATING, TYPE 1B, CMT1B » HEREDITARY MOTOR AND SENSORY NEUROPATHY 1B, HMSN1B	118200		Sequencing	500
Molecular Tests	MPZ (MYELIN PROTEN ZERO, P0)	159440	CHARCOT-MARIE-TOOTH DISEASE, DEMYELINATING, TYPE 4F, CMT4F » DEJERINE-SOTTAS HYPERTROPHIC NEUROPATHY	145900		Sequencing	500
Molecular Tests	MPZ (MYELIN PROTEN ZERO, P0)	159440	ROUSSY-LEVY HEREDITARY AREFLEXIC DYSTASIA	180800		Sequencing	500
Molecular Tests	MPZ (MYELIN PROTEN ZERO, P0)	159440	NEUROPATHY, CONGENITAL HYPOMYELINATING	605253		Sequencing	500
Molecular Tests	MPZ (MYELIN PROTEN ZERO, P0)	159440	CHARCOT-MARIE-TOOTH DISEASE, AXONAL, TYPE 2I, CMT2I	607677		Sequencing	500
Molecular Tests	MPZ (MYELIN PROTEN ZERO, P0)	159440	CHARCOT-MARIE-TOOTH DISEASE, AXONAL, TYPE 2J, CMT2J	607736		Sequencing	500
Molecular Tests	MPZ (MYELIN PROTEN ZERO, P0)	159440	CHARCOT-MARIE-TOOTH DISEASE, DOMINANT INTERMEDIATE D, CMTDID	607791		Sequencing	500
Molecular Tests	MR1 (MYOFIBRILLOGENESIS REGULATOR 1)	609023	PAROXYSMAL NONKINESIGENIC DYSKINESIA » PAROXYSMAL DYSTONIC CHOREOATHETOSIS » MOUNT-REBACK SYNDROME » CHOREOATHETOSIS, NONKINESIGENIC	118800		Sequencing	850
Molecular Tests	MRAP (MELANOCORTIN 2 RECEPTOR ACCESSORY PROTEIN)	609196	GLUCOCORTICOID DEFICIENCY 2	607398		Sequencing	750
Molecular Tests	MRE11A (MEIOTIC RECOMBINATION 11, S. CEREVISIAE, HOMOLOG OF, A; MRE11)	600814	ATAXIA-TELANGIECTASIA-LIKE DISORDER	604391		Sequencing	1200
Molecular Tests	MRPS16 (MITOCHONDRIAL RIBOSOMAL PROTEIN S16)	609204	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY, TYPE 2 » CORPUS CALLOSUM, AGENESIS OF, WITH DYSMORPHISM AND FATAL LACTIC ACIDOSIS	610498		Sequencing	500
Molecular Tests	MSH2	120435	COLORECTAL CANCER, HEREDITARY NONPOLYPOSIS, HNPCC, TYPE 2 » LYNCH CANCER FAMILY SYNDROME, TYPE 2	114400		Sequencing	950
Molecular Tests	MSH2	120435	COLORECTAL CANCER, HEREDITARY NONPOLYPOSIS, HNPCC, TYPE 1 » LYNCH CANCER FAMILY SYNDROME, TYPE 1	114500		Sequencing	950
Molecular Tests	MSH6	600678	COLORECTAL CANCER, HEREDITARY NONPOLYPOSIS, HNPCC, TYPE 2 » LYNCH CANCER FAMILY SYNDROME, TYPE 2	114400		Sequencing	870
Molecular Tests	MSH6	600678	COLORECTAL CANCER, HEREDITARY NONPOLYPOSIS, HNPCC, TYPE 1 » LYNCH CANCER FAMILY SYNDROME, TYPE 1	114500		Sequencing	870

Molecular Tests	MSX1 (MUSCLE SEGMENT HOMEOBOX, DROSOPHILA, HOMOLOG OF, 1)	142983	OROFACIAL CLEFT, TYPE 5 » CLEFT LIP WITH OR WITHOUT CLEFT PALATE, NONSYNDROMIC, TYPE 5	106600		Sequencing	550
Molecular Tests	MSX1 (MUSCLE SEGMENT HOMEOBOX, DROSOPHILA, HOMOLOG OF, 1)	142983	TOOTH AGENESIS, SELECTIVE, TYPE 1 » HYPODONTIA/OLIGODONTIA, TYPE 1	189500		Sequencing	550
Molecular Tests	MSX1 (MUSCLE SEGMENT HOMEOBOX, DROSOPHILA, HOMOLOG OF, 1)	142983	WITKOP SYNDROME » NAIL DYSPLASIA WITH HYPODONTIA » TOOTH AND NAIL SYNDROME	608874		Sequencing	550
Molecular Tests	MSX2 (MUSCLE SEGMENT HOMEOBOX, DROSOPHILA, HOMOLOG OF, 2)	123101	PARIETAL FORAMINA, TYPE 1 » FORAMINA PARIETALIA PERMAGNA » CATLIN MARKS	168500		Sequencing	900
Molecular Tests	MSX2 (MUSCLE SEGMENT HOMEOBOX, DROSOPHILA, HOMOLOG OF, 2)	123101	PARIETAL FORAMINA WITH CLEIDOCRANIAL DYSPLASIA	168550		Sequencing	900
Molecular Tests	MSX2 (MUSCLE SEGMENT HOMEOBOX, DROSOPHILA, HOMOLOG OF, 2)	123101	CRANIOSYNOSTOSIS, TYPE 2 » CRANIOSYNOSTOSIS, BOSTON-TYPE	604757		Sequencing	900
Molecular Tests	MTHFR (5,10-@METHYLENETETRAHYDROFOLATE REDUCTASE)	607093	MTHFR DEFICIENCY » HOMOCYSTINURIA DUE TO DEFICIENCY OF N(5,10)-METHYLENETETRAHYDROFOLATE REDUCTASE ACTIVITY	236250		Sequencing	600
Molecular Tests	MTM1 (MYOTUBULARIN)	300415	MYOTUBULAR MYOPATHY (X-LINKED)	310400		Sequencing	1560
Molecular Tests	MTMR2 (MYOTUBULARIN-RELATED PROTEIN 2)	603557	CHARCOT-MARIE-TOOTH DISEASE, TYPE 4B1 » CHARCOT-MARIE-TOOTH DISEASE WITH FOCALLY FOLDED MYELIN SHEATHS, TYPE 4B1 (AUTOSOMAL RECESSIVE)	601382		Sequencing	1250
Molecular Tests	MTP	157147	ABETALIPOPROTEINEMIA » ACANTHOCYTOSIS » BASSEN-KORNZWEIG SYNDROME » APOLIPOPROTEIN B DEFICIENCY » MICROSOMAL TRIGLYCERIDE TRANSFER PROTEIN DEFICIENCY	200100		Sequencing	1670
Molecular Tests	MTR (5-@METHYLTETRAHYDROFOLATE-HOMOCYSTEINE S-METHYLTRANSFERASE; TETRAHYDROPTEROYLGLUTAMATE METHYLTRANSFERASE)	156570	METHYLCOBALAMIN DEFICIENCY, cblG TYPE » HOMOCYSTINURIA-MEGALOBLASTIC ANEMIA DUE TO DEFECT IN COBALAMIN METABOLISM, cblG COMPLEMENTATION TYPE	250940		Sequencing	2300
Molecular Tests	MTRR (METHIONINE SYNTHASE REDUCTASE)	602568	HOMOCYSTINURIA-MEGALOBLASTIC ANEMIA DUE TO DEFECT IN COBALAMIN METABOLISM, TYPE cblE » VITAMIN B12-RESPONSIVE HOMOCYSTINURIA, cblE TYPE » METHYLCOBALAMIN DEFICIENCY, cblE TYPE	236270		Sequencing	2390
Molecular Tests	MTRR (METHIONINE SYNTHASE REDUCTASE)	602568	NEURAL TUBE DEFECTS, FOLATE-SENSITIVE	601634		Sequencing	2390
Molecular Tests	MUT (METHYLMALONYL CoA MUTASE, MCM)	251000	METHYLMALONIC ACIDURIA DUE TO MCM DEFICIENCY » MMA DUE TO MCM DEFICIENCY » MCM DEFICIENCY	251000		Sequencing	980
Molecular Tests	MVK (MEVALONATE KINASE)	251170	HYPER-IgD SYNDROME » PERIODIC FEVER, DUTCH TYPE	260920		Exons 8-10	610
Molecular Tests	MVK (MEVALONATE KINASE)	251170	HYPER-IgD SYNDROME » PERIODIC FEVER, DUTCH TYPE	260920		Remaining Exons	840
Molecular Tests	MYBPC3 (MYOSIN-BINDING PROTEIN C, CARDIAC)	600958	HYPERTROPHIC CARDIOMYOPATHY, FAMILIAL, 4A, CMD4A » VENTRICULAR HYPERTROPHY, HEREDITARY » ASYMMETRIC SEPTAL HYPERTROPHY » HYPERTROPHIC SUBAORTIC STENOSIS, IDIOPATHIC	115197		Sequencing	
Molecular Tests	MYCN (V-MYC AVIAN MYELOCYTOMATOSIS VIRAL-RELATED ONCOGENE, NEUROBLASTOMA-DERIVED)	164840	FEINGOLD SYNDROME » OCULODIGITOESOPHAGODUODENAL SYNDROME » ODED SYNDROME » MICROCEPHALY-OCULO-DIGITO-ESOPHAGEAL-DUODENAL SYNDROME » DIGITAL ANOMALIES WITH SHORT PALPEBRAL FISSURES AND ATRESIA OF ESOPHAGUSOR DUODENUM » MICROCEPHALY, MENTAL RETARDATION, AND TRACHEOESOPHAGEAL FISTULA SYNDROME	164280		Sequencing	800
Molecular Tests	MYCN (V-MYC AVIAN MYELOCYTOMATOSIS VIRAL-RELATED ONCOGENE, NEUROBLASTOMA-DERIVED)	164840	FEINGOLD SYNDROME » OCULODIGITOESOPHAGODUODENAL SYNDROME » ODED SYNDROME » MICROCEPHALY-OCULO-DIGITO-ESOPHAGEAL-DUODENAL SYNDROME » DIGITAL ANOMALIES WITH SHORT PALPEBRAL FISSURES AND ATRESIA OF ESOPHAGUSOR DUODENUM » MICROCEPHALY, MENTAL RETARDATION, AND TRACHEOESOPHAGEAL FISTULA SYNDROME	164280		Deletions	350
Molecular Tests	MYH (MUTYH, MUTY, E. COLI, HOMOLOG OF)	604933	COLORECTAL ADENOMATOUS POLYPOSIS (AUTOSOMAL RECESSIVE)	608456		Sequencing	740
Molecular Tests	MYH11 (MYOSIN, HEAVY CHAIN 11, SMOOTH MUSCLE, SMOOTH MUSCLE MYOSIN HEAVY CHAIN)	160745	AORTIC ANEURYSM, FAMILIAL THORACIC, TYPE 4 » AORTIC ANEURYSM/AORTIC DISSECTION AND PATENT DUCTUS ARTERIOSUS	132900		Sequencing	950

Molecular Tests	MYH11 (MYOSIN, HEAVY CHAIN 11, SMOOTH MUSCLE, SMOOTH MUSCLE MYOSIN HEAVY CHAIN)	160745	AORTIC ANEURYSM, FAMILIAL THORACIC, TYPE 4 » AORTIC ANEURYSM, AORTIC DISSECTION AND PATENT DUCTUS ARTERIOSUS	132900		Deletion-Duplication Testing	500
Molecular Tests	MYH3 (MYOSIN, HEAVY CHAIN 3, SKELETAL MUSCLE, EMBRYONIC)	160720	ARTHROGRYPOSIS, DISTAL, TYPE 2A » FREEMAN-SHELDON SYNDROME » WHISTLING FACE-WINDMILL VANE HAND SYNDROME	193700		Sequencing	2600
Molecular Tests	MYH3 (MYOSIN, HEAVY CHAIN 3, SKELETAL MUSCLE, EMBRYONIC)	160720	ARTHROGRYPOSIS, DISTAL, TYPE 2A » FREEMAN-SHELDON SYNDROME » WHISTLING FACE-WINDMILL VANE HAND SYNDROME	193700		Exons 6, 9, 10, 12, 15, 16, 18, 21, 22, 34	1000
Molecular Tests	MYH3 (MYOSIN, HEAVY CHAIN 3, SKELETAL MUSCLE, EMBRYONIC)	160720	ARTHROGRYPOSIS, DISTAL, TYPE 2B » ARTHROGRYPOSIS MULTIPLEX CONGENITA, DISTAL, TYPE 2B » SHELDON-HALL SYNDROME » FREEMAN-SHELDON SYNDROME VARIANT	601680		Sequencing	2600
Molecular Tests	MYH3 (MYOSIN, HEAVY CHAIN 3, SKELETAL MUSCLE, EMBRYONIC)	160720	ARTHROGRYPOSIS, DISTAL, TYPE 2B » ARTHROGRYPOSIS MULTIPLEX CONGENITA, DISTAL, TYPE 2B » SHELDON-HALL SYNDROME » FREEMAN-SHELDON SYNDROME VARIANT	601680		Exons 6, 9, 10, 12, 15, 16, 18, 21, 22, 34	1000
Molecular Tests	MYH6 (MYOSIN, HEAVY CHAIN 6, CARDIAC MUSCLE, ALPHA-MYOSIN, CARDIAC, HEAVY CHAIN, ALPHA)	160710	CARDIOMYOPATHY, FAMILIAL, HYPERTROPHIC, TYPE 14	613251		Sequencing	1850
Molecular Tests	MYH6 (MYOSIN, HEAVY CHAIN 6, CARDIAC MUSCLE, ALPHA-MYOSIN, CARDIAC, HEAVY CHAIN, ALPHA)	160710	CARDIOMYOPATHY, DILATED, TYPE 1EE	613252		Sequencing	1850
Molecular Tests	MYH6 (MYOSIN, HEAVY CHAIN 6, CARDIAC MUSCLE, ALPHA-MYOSIN, CARDIAC, HEAVY CHAIN, ALPHA)	160710	ATRIAL SEPTAL DEFECT, TYPE 5, ADVS	614089		Sequencing	1850
Molecular Tests	MYH6 (MYOSIN, HEAVY CHAIN 6, CARDIAC MUSCLE, ALPHA-MYOSIN, CARDIAC, HEAVY CHAIN, ALPHA)	160710	SICK SINUS SYNDROME, TYPE 3, SUSCEPTIBILITY TO	614990		Sequencing	1850
Molecular Tests	MYH7 (MYOSIN, HEAVY CHAIN 7, CARDIAC MUSCLE, BETA)	160760	MYOPATHY, DISTAL 1, MPD1 » LAING DISTAL MYOPATHY » MYOPATHY, DISTAL, EARLY ONSET, AUTOSOMAL DOMINANT	160500		Sequencing	1140
Molecular Tests	MYH7 (MYOSIN, HEAVY CHAIN 7, CARDIAC MUSCLE, BETA)	160760	CARDIOMYOPATHY, DILATED, TYPE 1S	613426		Sequencing	1140
Molecular Tests	MYH7 (MYOSIN, HEAVY CHAIN 7, CARDIAC MUSCLE, BETA)	160760	HYPERTROPHIC CARDIOMYOPATHY, FAMILIAL, 1, CMH1 » VENTRICULAR HYPERTROPHY, HEREDITARY » ASYMMETRIC SEPTAL HYPERTROPHY	192600		Sequencing	1140
Molecular Tests	MYH7 (MYOSIN, HEAVY CHAIN 7, CARDIAC MUSCLE, BETA)	160760	MYOPATHY, MYOSIN STORAGE » MYOPATHY, HYALINE BODY, AUTOSOMAL DOMINANT	608758		Sequencing	1140
Molecular Tests	MYH8 (MYOSIN, HEAVY CHAIN 8, SKELETAL MUSCLE, PERINATAL)	160741	TRISMUS-PSEUDOCAMPTODACTYLY SYNDROME » CARNEY COMPLEX VARIANT ASSOCIATED WITH DISTAL ARTHROGRYPOSIS	598857		Sequencing	2350
Molecular Tests	MYH9 (MYOSIN, HEAVY CHAIN 9)	160775	FECHTNER SYNDROME » MACROTHROMBOCYTOPATHY, NEPHRITIS, DEAFNESS, AND LEUKOCYTE INCLUSIONS » ALPORT SYNDROME WITH LEUKOCYTE INCLUSIONS	153640		Sequencing	1250
Molecular Tests	MYH9 (MYOSIN, HEAVY CHAIN 9)	160775	EPSTEIN SYNDROME » MACROTHROMBOCYTOPATHY, NEPHRITIS, AND DEAFNESS » ALPORT SYNDROME WITH MACROTHROMBOCYTOPENIA	153650		Sequencing	1250
Molecular Tests	MYH9 (MYOSIN, HEAVY CHAIN 9)	160775	MAY-HEGGLIN ANOMALY » DOHLE LEUKOCYTE INCLUSIONS WITH GIANT PLATELETS » MACROTHROMBOCYTOPENIA WITH LEUKOCYTE INCLUSIONS	155100		Sequencing	1250
Molecular Tests	MYH9 (MYOSIN, HEAVY CHAIN 9)	160775	DEAFNESS, DFNA17 » DEAFNESS, AUTOSOMAL DOMINANT, NONSYNDROMIC SENSORINEURAL 17	601652		Sequencing	1250
Molecular Tests	MYH9 (MYOSIN, HEAVY CHAIN 9)	160775	SEBASTIAN SYNDROME » SEBASTIAN PLATELET SYNDROME	605249		Sequencing	1250
Molecular Tests	MYL2 (MYOSIN, LIGHT CHAIN 2, REGULATORY, CARDIAC, SLOW)	160781	HYPERTROPHIC CARDIOMYOPATHY, FAMILIAL, 10, CMH10 » VENTRICULAR HYPERTROPHY, HEREDITARY » ASYMMETRIC SEPTAL HYPERTROPHY » HYPERTROPHIC SUBAORTIC STENOSIS, IDIOPATHIC	608758		Sequencing	655

Molecular Tests	MYL3 (MYOSIN, LIGHT CHAIN 3, ALKALI, VENTRICULAR, SKELETAL, SLOW, ESSENTIAL LIGHT CHAIN OF MYOSIN)	160790	HYPERTROPHIC CARDIOMYOPATHY, FAMILIAL, 8, CMH8 » VENTRICULAR HYPERTROPHY, HEREDITARY » ASYMMETRIC SEPTAL HYPERTROPHY » HYPERTROPHIC SUBAORTIC STENOSIS, IDIOPATHIC	608751		Sequencing	550
Molecular Tests	MYO6 (MYOSIN 6)	600970	DEAFNESS, DFNA22 » DEAFNESS (AUTOSOMAL DOMINANT) NONSYNDROMIC SENSORINEURAL 22	606346		Sequencing	1300
Molecular Tests	MYO6 (MYOSIN 6)	600970	DEAFNESS, DFNB37 » DEAFNESS (AUTOSOMAL RECESSIVE) NONSYNDROMIC SENSORINEURAL 37	607821		Sequencing	1300
Molecular Tests	MYO7A (MYOSIN 7A)	276903	USHER SYNDROME, TYPE 1B, USH1B » USHER SYNDROME, TYPE 1, NON-ACADIAN VARIETY	276903		Sequencing	1060
Molecular Tests	MYOC (MYOCILIN)	601652	GLAUCOMA, PRIMARY OPEN ANGLE, JUVENILE-ONSET, 1	137750		Sequencing	600
Molecular Tests	NAGLU (N-ACETYLGLUCOSAMINIDASE, ALPHA)	609701	MUCOPOLYSACCHARIDOSIS TYPE 3B, MPS3B » SANFILIPPO SYNDROME B » N-ACETYL-ALPHA-D-GLUCOSAMINIDASE DEFICIENCY	252920		Sequencing	700
Molecular Tests	NAGS (N-ACETYLGLUTAMATE SYNTHASE)	608300	N-ACETYLGLUTAMATE SYNTHASE DEFICIENCY » HYPERAMMONEMIA DUE TO N-ACETYLGLUTAMATE SYNTHETASE DEFICIENCY	237310		Sequencing	780
Molecular Tests	NBS1 (NIBRIN)	602667	NIJMEGEN BREAKAGE SYNDROME, NBS » SEEMANOVA SYNDROME, TYPE 2 » ATAXIA-TELANGIECTASIA VARIANT » MICROCEPHALY WITH NORMAL INTELLIGENCE, IMMUNODEFICIENCY, AND LYMPHORETICULAR MALIGNANCIES	251260		Sequencing	1600
Molecular Tests	NBS1 (NIBRIN)	602667	NIJMEGEN BREAKAGE SYNDROME, NBS » SEEMANOVA SYNDROME, TYPE 2 » ATAXIA-TELANGIECTASIA VARIANT » MICROCEPHALY WITH NORMAL INTELLIGENCE, IMMUNODEFICIENCY, AND LYMPHORETICULAR MALIGNANCIES	251260		1 Mutation: 5bp Deletion (90% of mutations)	250
Molecular Tests	NCF1 (p47 PHOX)	233700	CHRONIC GRANULOMATOUS DISEASE (AUTOSOMAL RECESSIVE) CYTOCHROME-b-POSITIVE FORM TYPE 1	233700		Sequencing	470
Molecular Tests	NCF2 (p67 PHOX)	233710	CHRONIC GRANULOMATOUS DISEASE (AUTOSOMAL RECESSIVE) CYTOCHROME-b-POSITIVE FORM TYPE 2	233710		Sequencing	1910
Molecular Tests	NDP (NORRIN)	310600	COATS DISEASE	300216		Sequencing	530
Molecular Tests	NDP (NORRIN)	310600	COATS DISEASE	300216		Deletion-Duplication Testing	500
Molecular Tests	NDP (NORRIN)	310600	NORRIE DISEASE » ATROPHIA BULBORUM HEREDITARIA » PSEUDOGLIOMA » EPISKOPI BLINDNESS	310600		Sequencing	530
Molecular Tests	NDP (NORRIN)	310600	NORRIE DISEASE » ATROPHIA BULBORUM HEREDITARIA » PSEUDOGLIOMA » EPISKOPI BLINDNESS	310600		Deletion-Duplication Testing	500
Molecular Tests	NDRG1 (NMYC DOWNSTREAM-REGULATED GENE 1; PROTEIN REGULATED BY OXYGEN 1; PROXY1)	605262	CHARCOT-MARIE-TOOTH DISEASE, TYPE 4D, CMT4D » NEUROPATHY, HEREDITARY MOTOR AND SENSORY, LOM TYPE (HMSNL)	601455		Sequencing	550
Molecular Tests	NDUFA1 (NADH-UBIQUINONE OXIDOREDUCTASE 1 ALPHA SUBCOMPLEX, 1)	300078	CYTOCHROME c OXIDASE DEFICIENCY » COX DEFICIENCY » COMPLEX 1, MITOCHONDRIAL RESPIRATORY CHAIN, DEFICIENCY OF	252010		Sequencing	610
Molecular Tests	NDUFA10 (NADH-UBIQUINONE OXIDOREDUCTASE 1 ALPHA SUBCOMPLEX, 10)	603835	LEIGH SYNDROME DUE TO MITOCHONDRIAL COMPLEX 1 DEFICIENCY	256000		Sequencing	970
Molecular Tests	NDUFA11 (NADH DEHYDROGENASE 1 ALPHA SUBCOMPLEX, 11)	612638	CYTOCHROME c OXIDASE DEFICIENCY » COX DEFICIENCY » COMPLEX 1, MITOCHONDRIAL RESPIRATORY CHAIN, DEFICIENCY OF	252010		Sequencing	560
Molecular Tests	NDUFA13 (NADH-UBIQUINONE OXIDOREDUCTASE 1 ALPHA SUBCOMPLEX, 13)	609435	THYROID CARCINOMA, HURTHLE CELL			Sequencing	580
Molecular Tests	NDUFA2 (NADH-UBIQUINONE OXIDOREDUCTASE 1 ALPHA SUBCOMPLEX, 2; B8 PROTEIN)	602137	LEIGH SYNDROME DUE TO MITOCHONDRIAL COMPLEX 1 DEFICIENCY	256000		Sequencing	530
Molecular Tests	NDUFAF1 (NADH DEHYDROGENASE 1 ALPHA SUBCOMPLEX, ASSEMBLY FACTOR 1; NADH-UBIQUINONE OXIDOREDUCTASE 1 ALPHA SUBCOMPLEX, ASSEMBLY FACTOR 1; COMPLEX I INTERMEDIATE-ASSOCIATED PROTEIN 30; CIA30)	606934	CYTOCHROME c OXIDASE DEFICIENCY » COX DEFICIENCY » COMPLEX 1, MITOCHONDRIAL RESPIRATORY CHAIN, DEFICIENCY OF	252010		Sequencing	490

Molecular Tests	NDUFAF2 (NADH DEHYDROGENASE 1 ALPHA SUBCOMPLEX, ASSEMBLY FACTOR 2; MYC-INDUCED MITOCHONDRIAL PROTEIN; MIMITIN; MMTN)	606934	CYTOCHROME c OXIDASE DEFICIENCY » COX DEFICIENCY » COMPLEX I, MITOCHONDRIAL RESPIRATORY CHAIN, DEFICIENCY OF	252010		Sequencing	750
Molecular Tests	NDUFAF2 (NADH DEHYDROGENASE 1 ALPHA SUBCOMPLEX, ASSEMBLY FACTOR 2; MYC-INDUCED MITOCHONDRIAL PROTEIN; MIMITIN; MMTN)	606934	LEIGH SYNDROME DUE TO MITOCHONDRIAL COMPLEX 1 DEFICIENCY	256000		Sequencing	750
Molecular Tests	NDUFAF3 (NADH DEHYDROGENASE 1 ALPHA SUBCOMPLEX, ASSEMBLY FACTOR 3)	612911	CYTOCHROME c OXIDASE DEFICIENCY » COX DEFICIENCY » COMPLEX I, MITOCHONDRIAL RESPIRATORY CHAIN, DEFICIENCY OF	252010		Sequencing	580
Molecular Tests	NDUFAF4 (NADH DEHYDROGENASE 1 ALPHA SUBCOMPLEX, ASSEMBLY FACTOR 4; HORMONE-REGULATED PROLIFERATION-ASSOCIATED PROTEIN, 20-KD; HRPAP20; C6ORF66)	611776	CYTOCHROME c OXIDASE DEFICIENCY » COX DEFICIENCY » COMPLEX I, MITOCHONDRIAL RESPIRATORY CHAIN, DEFICIENCY OF	252010		Sequencing	530
Molecular Tests	NDUFS1 (NADH-UBIQUINONE OXIDOREDUCTASE Fe-S PROTEIN 1, COMPLEX 1, MITOCHONDRIAL RESPIRATORY CHAIN, 75-KD SUBUNIT)	157655	CYTOCHROME c OXIDASE DEFICIENCY » COX DEFICIENCY » COMPLEX I, MITOCHONDRIAL RESPIRATORY CHAIN, DEFICIENCY OF	252010		Sequencing	1560
Molecular Tests	NDUFS2 (NADH-UBIQUINONE OXIDOREDUCTASE Fe-S PROTEIN 2, COMPLEX 1, MITOCHONDRIAL RESPIRATORY CHAIN, 49-KD SUBUNIT)	602985	CYTOCHROME c OXIDASE DEFICIENCY » COX DEFICIENCY » COMPLEX I, MITOCHONDRIAL RESPIRATORY CHAIN, DEFICIENCY OF	252010		Sequencing	710
Molecular Tests	NDUFS3 (NADH-UBIQUINONE OXIDOREDUCTASE Fe-S PROTEIN 3, NADH-COENZYME Q REDUCTASE, 30-KD COMPLEX 1, MITOCHONDRIAL RESPIRATORY CHAIN, 30-KD SUBUNIT)	603846	CYTOCHROME c OXIDASE DEFICIENCY » COX DEFICIENCY » COMPLEX I, MITOCHONDRIAL RESPIRATORY CHAIN, DEFICIENCY OF	220110		Sequencing	820
Molecular Tests	NDUFS3 (NADH-UBIQUINONE OXIDOREDUCTASE Fe-S PROTEIN 3, NADH-COENZYME Q REDUCTASE, 30-KD COMPLEX 1, MITOCHONDRIAL RESPIRATORY CHAIN, 30-KD SUBUNIT)	603846	LEIGH SYNDROME	256000		Sequencing	820
Molecular Tests	NDUFS4 (NADH-UBIQUINONE OXIDOREDUCTASE Fe-S PROTEIN 4, NADH-COENZYME Q REDUCTASE, 18-KD COMPLEX 1, MITOCHONDRIAL RESPIRATORY CHAIN, 18-KD SUBUNIT, AQDQ)	602694	CYTOCHROME c OXIDASE DEFICIENCY » COX DEFICIENCY » COMPLEX I, MITOCHONDRIAL RESPIRATORY CHAIN, DEFICIENCY OF	220110		Sequencing	750
Molecular Tests	NDUFS4 (NADH-UBIQUINONE OXIDOREDUCTASE Fe-S PROTEIN 4, NADH-COENZYME Q REDUCTASE, 18-KD COMPLEX 1, MITOCHONDRIAL RESPIRATORY CHAIN, 18-KD SUBUNIT, AQDQ)	602694	LEIGH SYNDROME	256000		Sequencing	750
Molecular Tests	NDUFS6 (NADH-UBIQUINONE OXIDOREDUCTASE Fe-S PROTEIN 6, COMPLEX I, MITOCHONDRIAL RESPIRATORY CHAIN, 13-KD SUBUNIT)	603848	CYTOCHROME c OXIDASE DEFICIENCY » COX DEFICIENCY » COMPLEX I, MITOCHONDRIAL RESPIRATORY CHAIN, DEFICIENCY OF	252010		Sequencing	530
Molecular Tests	NDUFS7 (NADH-UBIQUINONE OXIDOREDUCTASE Fe-S PROTEIN 7, NADH-COENZYME Q REDUCTASE, 20-KD COMPLEX 1, MITOCHONDRIAL RESPIRATORY CHAIN, 20-KD SUBUNIT, PSST)	601825	CYTOCHROME c OXIDASE DEFICIENCY » COX DEFICIENCY » COMPLEX I, MITOCHONDRIAL RESPIRATORY CHAIN, DEFICIENCY OF	220110		Sequencing	900
Molecular Tests	NDUFS7 (NADH-UBIQUINONE OXIDOREDUCTASE Fe-S PROTEIN 7, NADH-COENZYME Q REDUCTASE, 20-KD COMPLEX 1, MITOCHONDRIAL RESPIRATORY CHAIN, 20-KD SUBUNIT, PSST)	601825	LEIGH SYNDROME	256000		Sequencing	900
Molecular Tests	NDUFS8 (NADH-UBIQUINONE OXIDOREDUCTASE Fe-S PROTEIN 8, COMPLEX 1, MITOCHONDRIAL RESPIRATORY CHAIN, 23-KD SUBUNIT, TYKY)	602141	CYTOCHROME c OXIDASE DEFICIENCY » COX DEFICIENCY » COMPLEX I, MITOCHONDRIAL RESPIRATORY CHAIN, DEFICIENCY OF	220110		Sequencing	590
Molecular Tests	NDUFS8 (NADH-UBIQUINONE OXIDOREDUCTASE Fe-S PROTEIN 8, COMPLEX 1, MITOCHONDRIAL RESPIRATORY CHAIN, 23-KD SUBUNIT, TYKY)	602141	LEIGH SYNDROME	256000		Sequencing	590
Molecular Tests	NDUFV1 (NADH-UBIQUINONE OXIDOREDUCTASE FLAVOPROTEIN 1, COMPLEX 1, MITOCHONDRIAL RESPIRATORY CHAIN, 51-KD SUBUNIT, UQOR1)	161015	CYTOCHROME c OXIDASE DEFICIENCY » COX DEFICIENCY » COMPLEX I, MITOCHONDRIAL RESPIRATORY CHAIN, DEFICIENCY OF	220110		Sequencing	900
Molecular Tests	NDUFV1 (NADH-UBIQUINONE OXIDOREDUCTASE FLAVOPROTEIN 1, COMPLEX 1, MITOCHONDRIAL RESPIRATORY CHAIN, 51-KD SUBUNIT, UQOR1)	161015	LEIGH SYNDROME	256000		Sequencing	900
Molecular Tests	NEB (NEBULIN)	161650	NEMALINE MYOPATHY 2, NEM2	256030		1 Mutation: Common Deletion of Exon 55	450
Molecular Tests	NEFL (NEUROFILAMENT PROTEIN, LIGHT POLYPEPTIDE)	162280	CHARCOT-MARIE-TOOTH DISEASE, AXONAL, TYPE 2E, CMT2E	607684		Sequencing	600
Molecular Tests	NEFL (NEUROFILAMENT PROTEIN, LIGHT POLYPEPTIDE)	162280	CHARCOT-MARIE-TOOTH DISEASE, DEMYELINATING, TYPE 1F, CMT1F	607754		Sequencing	600
Molecular Tests	NEU1 (NEURAMINIDASE 1, SIALIDASE)	608272	NEURAMINIDASE DEFICIENCY » SIALIDOSIS, TYPE 2 » SIALIDOSIS, TYPE 1 » MUCOLIPIDOSIS, TYPE 1, ML1 » LIPOMUCOPOLYSACCHARIDOSIS » SIALIDASE DEFICIENCY » NEU1 DEFICIENCY » CHERRY RED SPOT--MYOCLONUS SYNDROME	256550		Sequencing	650

Molecular Tests	NEUROD1 (BETA2)	601724	DIABETES MELLITUS, NONINSULIN-DEPENDENT, NIDDM » DIABETES MELLITUS, TYPE 2 » NONINSULIN-DEPENDENT DIABETES MELLITUS » MATURITY-ONSET DIABETES	125853		Sequencing	250
Molecular Tests	NEUROD1 (BETA2)	601724	MATURITY-ONSET DIABETES OF THE YOUNG, TYPE 6, MODY6	606394		Sequencing	250
Molecular Tests	NF1 (NEUROFIBROMIN)	162200	NEUROFIBROMATOSIS, TYPE 1, NF1 » VON RECKLINGHAUSEN DISEASE	162200		Sequencing and Deletion-Duplication Testing	1100
Molecular Tests	NF1 (NEUROFIBROMIN)	162200	NEUROFIBROMATOSIS, TYPE 1, NF1 » VON RECKLINGHAUSEN DISEASE	162200		Deletion-Duplication Testing	450
Molecular Tests	NF2 (NEUROFIBROMIN 2, MERLIN, SCHWANNOMIN)	607379	NEUROFIBROMATOSIS TYPE 2, NF2 » NEUROFIBROMATOSIS, CENTRAL TYPE » ACOUSTIC SCHWANNOMAS, BILATERAL » BILATERAL ACOUSTIC NEUROFIBROMATOSIS	101000		Sequencing and Deletion-Duplication Testing	980
Molecular Tests	NGFB (NERVE GROWTH FACTOR, BETA SUBUNIT)	162030	NEUROPATHY, HEREDITARY SENSORY AND AUTONOMIC, TYPE 5, HSAN5 » INSENSITIVITY TO PAIN, CONGENITAL	608654		Sequencing	300
Molecular Tests	NHLRC1 (NHL REPEAT-CONTAINING 1 GENE, EPM2B, MALIN)	608072	MYOCLONIC EPILEPSY OF LAFORA » LAFORA DISEASE » EPILEPSY, PROGRESSIVE MYOCLONIC TYPE 2	254780		Sequencing	300
Molecular Tests	NHS	300457	NANCE-HORAN SYNDROME » CATARACT-DENTAL SYNDROME » CATARACT, X-LINKED, WITH HUTCHINSONIAN TEETH » MESIODENS-CATARACT SYNDROME	302350		Sequencing	1000
Molecular Tests	NIPA1 (NONIMPRINTED GENE IN PRADER-WILLI SYNDROME/ANGELMAN SYNDROME CHROMOSOME REGION 1)	608145	FAMILIAL SPASTIC PARAPLEGIA 6 (AUTOSOMAL DOMINANT), SPG6	600363		Sequencing	1080
Molecular Tests	NIPBL	608667	CORNELIA DE LANGE SYNDROME » BRACHMANN-DE LANGE SYNDROME	122470		Sequencing	1350
Molecular Tests	NIPBL	608667	CORNELIA DE LANGE SYNDROME » BRACHMANN-DE LANGE SYNDROME	122470		Deletion-Duplication Testing	350
Molecular Tests	NKX2E (NK2, DROSOPHILA, HOMOLOG OF, E, NKX2.5, CSX)	600584	ATRIOVENTRICULAR BLOCK, IDIOPATHIC SECOND-DEGREE			Sequencing	770
Molecular Tests	NKX2E (NK2, DROSOPHILA, HOMOLOG OF, E, NKX2.5, CSX)	600584	VARIOUS HEART MALFORMATIONS			Sequencing	770
Molecular Tests	NKX2E (NK2, DROSOPHILA, HOMOLOG OF, E, NKX2.5, CSX)	600584	ATRIAL SEPTAL DEFECT WITH ATRIOVENTRICULAR CONDUCTION DEFECTS	108900		Sequencing	770
Molecular Tests	NKX2E (NK2, DROSOPHILA, HOMOLOG OF, E, NKX2.5, CSX)	600584	TETRALOGY OF FALLOT	187500		Sequencing	770
Molecular Tests	NLGN3 (NEUROLIGIN 3)	300336	AUTISM (X-LINKED)	300425		Sequencing	1040
Molecular Tests	NLGN3 (NEUROLIGIN 3)	300336	ASPERGER SYNDROME (X-LINKED)	300494		Sequencing	1040
Molecular Tests	NLGN3 AND NLGN4	300336	AUTISM (X-LINKED)	300425		Sequencing	1810
Molecular Tests	NLGN3 AND NLGN4	300336	ASPERGER SYNDROME (X-LINKED)	300494		Sequencing	1810
Molecular Tests	NLGN4 (NEUROLIGIN 4)	300336	AUTISM (X-LINKED)	300425		Sequencing	1110
Molecular Tests	NLGN4 (NEUROLIGIN 4)	300336	ASPERGER SYNDROME (X-LINKED)	300494		Sequencing	1110
Molecular Tests	NOG (NOGGIN)	602991	STAPES ANKYLOSIS » TEUNISSEN-CREMERS SYNDROME	184460		Sequencing	300
Molecular Tests	NOG (NOGGIN)	602991	PROXIMAL SYMPHALANGISM	185800		Sequencing	300
Molecular Tests	NOG (NOGGIN)	602991	MULTIPLE SYNOSTOSIS SYNDROME 1	186500		Sequencing	300
Molecular Tests	NOG (NOGGIN)	602991	TARSAL-CARPAL COALITION SYNDROME	186570		Sequencing	300
Molecular Tests	NOTCH1 (NOTCH, DROSOPHILA, HOMOLOG OF, 1)	190198	AORTIC VALVE DISEASE » BICUSPID AORTIC VALVE » LVOT	109730		Sequencing	1275
Molecular Tests	NOTCH2 (NOTCH, DROSOPHILA, HOMOLOG OF, 2)	600275	HAJDU-CHENEY SYNDROME » ACROOSTEOLYSIS WITH OSTEOPOROSIS AND CHANGES IN SKULL AND MANDIBLE » ARTHRODENTOOSTEODYSPLASIA	102500		Sequencing	1275
Molecular Tests	NOTCH2 (NOTCH, DROSOPHILA, HOMOLOG OF, 2)	600275	ALAGILLE SYNDROME, TYPE 2	610205		Sequencing	1275
Molecular Tests	NOTCH3	600276	CEREBRAL AUTOSOMAL DOMINANT ARTERIOPATHY WITH SUBCORTICAL INFARCTS AND LEUKOENCEPHALOPATHY, CADASIL	125310		Exons 2, 3, 4, 5, 6 and 11 harboring the majority of mutations	500
Molecular Tests	NOTCH3	600276	CEREBRAL AUTOSOMAL DOMINANT ARTERIOPATHY WITH SUBCORTICAL INFARCTS AND LEUKOENCEPHALOPATHY, CADASIL	125310		Exons 3-24 harboring all known mutations	1275
Molecular Tests	NPC1	607623	NIEMANN-PICK DISEASE, TYPE C1, NPC1 » NIEMANN-PICK DISEASE, TYPE D » NIEMANN-PICK DISEASE, NOVA SCOTIAN TYPE	257220		Sequencing	1500

Molecular Tests	NPC2	601015	NIEMANN-PICK DISEASE, TYPE C2	607625		Sequencing	400
Molecular Tests	NPHP1 (NEPHROCYSTIN 1)	607100	NEPHRONOPHTHISIS, TYPE 1	256100		Sequencing	770
Molecular Tests	NPHP1 (NEPHROCYSTIN 1)	607100	SENIOR-LOKEN SYNDROME » RENAL - RETINAL SYNDROME » JUVENILE NEPHRONOPHTHISIS WITH LEBER AMAUROSIS	266900		Sequencing	770
Molecular Tests	NPHP1 (NEPHROCYSTIN 1)	607100	JOUBERT SYNDROME, TYPE 4	609583		Sequencing	770
Molecular Tests	NPHP2 (NEPHROCYSTIN 2, INVS, INVERSIN)	243305	NEPHRONOPHTHISIS, TYPE 2 » NEPHRONOPHTHISIS, INFANTILE	602088		Sequencing	950
Molecular Tests	NPHP3 (NEPHROCYSTIN 3)	608002	NEPHRONOPHTHISIS, TYPE 3 » NEPHRONOPHTHISIS, ADOLESCENT	604387		Sequencing	1250
Molecular Tests	NPHP4 (NEPHROCYSTIN 4, NEPHRORETININ)	607215	SENIOR-LOKEN SYNDROME 4			Sequencing	820
Molecular Tests	NPHP4 (NEPHROCYSTIN 4, NEPHRORETININ)	607215	NEPHRONOPHTHISIS, TYPE 4 » NEPHRONOPHTHISIS, JUVENILE	606966		Sequencing	820
Molecular Tests	NPHS1 (NEPHRIN)	602716	NEPHROSIS 1, CONGENITAL, FINNISH TYPE, NPHS1 » FINNISH CONGENITAL NEPHROSIS	256300		Sequencing	1150
Molecular Tests	NPHS1 (NEPHRIN)	602716	NEPHROTIC SYNDROME, STEROID-RESISTANT, (AUTOSOMAL RECESSIVE), SRN1 » FAMILIAL FOCAL SEGMENTAL GLOMERULOSCLEROSIS (AUTOSOMAL RECESSIVE)	600995		Sequencing	1150
Molecular Tests	NPHS2 (PODOCIN)	604766	NEPHROSIS 1, CONGENITAL, FINNISH TYPE, NPHS1 » FINNISH CONGENITAL NEPHROSIS	256300		Sequencing	550
Molecular Tests	NPHS2 (PODOCIN)	604766	NEPHROTIC SYNDROME, STEROID-RESISTANT, (AUTOSOMAL RECESSIVE), SRN1 » FAMILIAL FOCAL SEGMENTAL GLOMERULOSCLEROSIS (AUTOSOMAL RECESSIVE)	600995		Sequencing	550
Molecular Tests	NPR2 (NATRIURETIC PEPTIDE RECEPTOR B/GUANYLATE CYCLASE B, ATRIAL NATRIURETIC PEPTIDE RECEPTOR, TYPE B, ANPRB)	108961	ACROMESOMELIC DYSPLASIA, MAROTEAUX TYPE » ST. HELENA DYSPLASIA	602875		Sequencing	670
Molecular Tests	NR2E3 (NUCLEAR RECEPTOR SUBFAMILY 2, GROUP E, MEMBER 3; PHOTORECEPTOR-SPECIFIC NUCLEAR RECEPTOR; PNR)	604485	ENHANCED S-CONE SYNDROME » GOLDMANN-FAVRE SYNDROME » RETINOSCHISIS WITH EARLY HEMERALOPIA	268100		Sequencing	530
Molecular Tests	NR2E3 (NUCLEAR RECEPTOR SUBFAMILY 2, GROUP E, MEMBER 3; PHOTORECEPTOR-SPECIFIC NUCLEAR RECEPTOR;	604485	RETINITIS PIGMENTOSA, TYPE 37, RP37	611131		Sequencing	530
Molecular Tests	NR3C1	138040	GLUCOCORTICOID RECEPTOR DEFICIENCY » GLUCOCORTICOID RESISTANCE » CORTISOL RESISTANCE FROM GLUCOCORTICOID RECEPTOR DEFECT	138040		Sequencing	770
Molecular Tests	NR3C2 (MINERALOCORTICOID RECEPTOR, MLR, MCR, MR, ALDOSTERONE RECEPTOR)	600983	PSEUDOHYPOALDOSTERONISM, TYPE 1 (AUTOSOMAL DOMINANT)	177735		Sequencing	600
Molecular Tests	NR3C2 (MINERALOCORTICOID RECEPTOR, MLR, MCR, MR, ALDOSTERONE RECEPTOR)	600983	HYPERTENSION, EARLY-ONSET (AUTOSOMAL DOMINANT)	605115		Sequencing	600
Molecular Tests	NR5A1 (NUCLEAR RECEPTOR SUBFAMILY 5, GROUP A, MEMBER 1; STEROIDOGENIC FACTOR 1; SF1)	184757	ADRENOCORTICAL INSUFFICIENCY			Sequencing	700
Molecular Tests	NR5A1 (NUCLEAR RECEPTOR SUBFAMILY 5, GROUP A, MEMBER 1; STEROIDOGENIC FACTOR 1; SF1)	184757	PREMATURE OVARIAN FAILURE 7, POF7	612964		Sequencing	700
Molecular Tests	NR5A1 (NUCLEAR RECEPTOR SUBFAMILY 5, GROUP A, MEMBER 1; STEROIDOGENIC FACTOR 1; SF1)	184757	46,XY GONADAL DYSGENESIS, COMPLETE OR PARTIAL, WITH OR WITHOUT ADRENAL FAILURE	612965		Sequencing	700
Molecular Tests	NRAS (NEUROBLASTOMA RAS VIRAL ONCOGENE HOMOLOG)	164790	AUTOIMMUNE LYMPHOPROLIFERATIVE SYNDROME, TYPE 4, ALPS4	164790		Sequencing	980
Molecular Tests	NRAS (NEUROBLASTOMA RAS VIRAL ONCOGENE HOMOLOG)	164790	NOONAN SYNDROME, TYPE 6	613224		Sequencing	980
Molecular Tests	NRL (NEURAL RETINA LEUCINE ZIPPER; NEURAL RETINA-SPECIFIC GENE)	162080	RETINAL DEGENERATION (AUTOSOMAL RECESSIVE), CLUMPED PIGMENT TYPE	162080		Sequencing	440
Molecular Tests	NRL (NEURAL RETINA LEUCINE ZIPPER; NEURAL RETINA-SPECIFIC GENE)	162080	RETINITIS PIGMENTOSA, TYPE 27, RP27	613750		Sequencing	440
Molecular Tests	NRXN1 (NEUREXIN 1)	600565	PITT-HOPKINS-LIKE SYNDROME, TYPE 2	600565		Sequencing	1470
Molecular Tests	NSD1	606681	CEREBRAL GIGANTISM » SOTOS SYNDROME	117550	At least 20 microgram DNA is needed	Sequencing	950
Molecular Tests	NSDHL (NAD(P)H STEROID DEHYDROGENASE-LIKE PROTEIN)	300275	CHILD SYNDROME » CONGENITAL HEMIDYSPLASIA WITH ICHTHYOSIFORM ERYTHRODERMA AND LIMB DEFECTS » ICHTHYOSIFORM ERYTHRODERMA, UNILATERAL, WITH IPSILATERAL MALFORMATIONS, ESPECIALLY ABSENCE DEFORMITY OF LIMBS	308050		Sequencing	1390
Molecular Tests	NTRK1	191315	INSENSITIVITY TO PAIN, CONGENITAL, WITH ANHIDROSIS, CIPA » NEUROPATHY, CONGENITAL SENSORY, WITH ANHIDROSIS » HEREDITARY SENSORY AND AUTONOMIC NEUROPATHY 4, HSAN4	256800		Sequencing	1000
Molecular Tests	NUBPL (NUCLEOTIDE-BINDING PROTEIN-LIKE PROTEIN; IRON-SULFUR PROTEIN REQUIRED FOR NADH DEHYDROGENASE; IND1)	613621	CYTOCHROME c OXIDASE DEFICIENCY » COX DEFICIENCY » COMPLEX 1, MITOCHONDRIAL RESPIRATORY CHAIN, DEFICIENCY OF	252010		Sequencing	1060

Molecular Tests	NYX (NYCTALOPIN)	300278	NIGHT BLINDNESS, CONGENITAL STATIONARY, TYPE 1A » HEMERALOPIA-MYOPIA » MYOPIA-NIGHT BLINDNESS	310500		Sequencing	530
Molecular Tests	OCA2 (P gene)	203200	OCULOCUTANEOUS ALBINISM, TYPE 2, OCA2 (TYROSINASE - POSITIVE) » PINK-EYED DILUTION	203200		Sequencing	820
Molecular Tests	OCA2 (P gene)	203200	OCULOCUTANEOUS ALBINISM, TYPE 2, OCA2 (TYROSINASE - POSITIVE) » PINK-EYED DILUTION	203200		Common Deletion	430
Molecular Tests	OCRL1	309000	DENT NEPHROCALCINOSIS	300009		Sequencing of both OCRL1 and CLCN5	890
Molecular Tests	OCRL1	309000	LOWE OCULOCEREBRORENAL SYNDROME, OCRL	309000		Sequencing of both OCRL1 and CLCN5	890
Molecular Tests	OCRL1	309000	DENT NEPHROCALCINOSIS	300009		Deletion-Duplication Testing	690
Molecular Tests	OCRL1	309000	LOWE OCULOCEREBRORENAL SYNDROME, OCRL	309000		Deletion-Duplication Testing	690
Molecular Tests	OFD1 (CHROMOSOME X OPEN READING FRAME 5; CXORF5)	300170	SIMPSON-GOLABI-BEHMEL SYNDROME, TYPE 2	300209		Sequencing	1990
Molecular Tests	OFD1 (CHROMOSOME X OPEN READING FRAME 5; CXORF5)	300170	OROFACIODIGITAL SYNDROME, TYPE 1 » OFD SYNDROME, TYPE 1	311200		Sequencing	1990
Molecular Tests	OPA1	605290	OPTIC ATROPHY 1 » OPTIC ATROPHY, JUVENILE » KJER TYPE OPTIC ATROPHY	165500		Sequencing	850
Molecular Tests	OPA3	606580	OPTIC ATROPHY, TYPE 3, OPA3 (AUTOSOMAL DOMINANT)	165300		Sequencing	610
Molecular Tests	OPA3	606580	OPTIC ATROPHY, TYPE 3, OPA3 (AUTOSOMAL RECESSIVE) OPTIC ATROPHY PLUS SYNDROME COSTEFF SYNDROME	258501		Sequencing	610
Molecular Tests	OPHN1 (OLIGOPHRENIN 1, OPN1)	300127	MENTAL RETARDATION WITH CEREBELLAR HYPOPLASIA AND DISTINCTIVE FACIAL APPEARANCE (X-LINKED) » MENTAL RETARDATION (X-LINKED), TYPE 60, MRX60	300486		Sequencing	1990
Molecular Tests	OPTN (OPTINEURIN)	602432	GLAUCOMA, PRIMARY OPEN ANGLE, ADULT-ONSET, POAG	137760		Sequencing	690
Molecular Tests	OPTN (OPTINEURIN)	602432	AMYOTROPHIC LATERAL SCLEROSIS, TYPE 12, ALS12	613435		Sequencing	690
Molecular Tests	OTC (ORNITHINE CARBAMOYL TRANSFERASE GENE)	311250	ORNITHINE CARBAMOYLTRANSFERASE DEFICIENCY	311250		Sequencing	810
Molecular Tests	OTC (ORNITHINE CARBAMOYL TRANSFERASE GENE)	311250	ORNITHINE CARBAMOYLTRANSFERASE DEFICIENCY	311250		Deletion-Duplication Testing	680
Molecular Tests	OTOF (OTOFERLIN)	603681	DEAFNESS, DFNB9 » DEAFNESS, (AUTOSOMAL RECESSIVE), NONSYNDROMIC SENSORINEURAL 9	601071		Sequencing	1680
Molecular Tests	OTX2 (ORTHODENTICLE, DROSOPHILA, HOMOLOG OF, 2)	600037	MICROPHTHALMIA, SYNDROMIC, TYPE 5, MCOPS5 » RETINAL DYSTROPHY, EARLY-ONSET, AND PITUITARY DYSFUNCTION	610125		Sequencing	480
Molecular Tests	OTX2 (ORTHODENTICLE, DROSOPHILA, HOMOLOG OF, 2)	600037	PITUITARY HORMONE DEFICIENCY, COMBINED, TYPE 6, CPHD6	613986		Sequencing	480
Molecular Tests	P53 (TP53)	191170	BREAST CANCER, FAMILIAL	114480		Sequencing	550
Molecular Tests	P53 (TP53)	191170	BREAST CANCER, FAMILIAL	114480		Exons 5-8	450
Molecular Tests	P53 (TP53)	191170	LI-FRAUMENI SYNDROME 1	151623		Sequencing	550
Molecular Tests	P53 (TP53)	191170	LI-FRAUMENI SYNDROME 1	151623		Exons 5-8	450
Molecular Tests	P53 (TP53)	191170	PEDIATRIC ADRENOCORTICAL CARCINOMA	202300		Sequencing	550
Molecular Tests	P53 (TP53)	191170	PEDIATRIC ADRENOCORTICAL CARCINOMA	202300		Exons 5-8	450
Molecular Tests	P53 (TP53)	191170	OSTEOSARCOMA	259500		Sequencing	550
Molecular Tests	P53 (TP53)	191170	OSTEOSARCOMA	259500		Exons 5-8	450
Molecular Tests	PABPN1 (PAB2, POLYA BINDING PROTEIN)	602279	OCULOPHARYNGEAL MUSCULAR DYSTROPHY (AUTOSOMAL DOMINANT) » MUSCULAR DYSTROPHY, OCULOPHARYNGEAL MUSCULAR DYSTROPHY (AUTOSOMAL DOMINANT)	164300		GCN Repeat	250
Molecular Tests	PABPN1 (PAB2, POLYA BINDING PROTEIN)	602279	OCULOPHARYNGEAL MUSCULAR DYSTROPHY (AUTOSOMAL RECESSIVE) » MUSCULAR DYSTROPHY, OCULOPHARYNGEAL MUSCULAR DYSTROPHY (AUTOSOMAL RECESSIVE)	257950		GCN Repeat	250
Molecular Tests	PAFAH1B1 (LIS1)	601545	MILLER-DIEKER LISSENCEPHALY SYNDROME, MDLS	247200		Sequencing	1100
Molecular Tests	PAFAH1B1 (LIS1)	601545	MILLER-DIEKER LISSENCEPHALY SYNDROME, MDLS	247200		Deletions	450
Molecular Tests	PAFAH1B1 (LIS1)	601545	LISSENCEPHALY 1, LIS1 » SUBCORTICAL LAMINAR HETEROTOPIA	607432		Sequencing	1100

Molecular Tests	PAFAH1B1 (LIS1)	601545	LISSENCEPHALY 1, LIS1 » SUBCORTICAL LAMINAR HETEROTOPIA » SUBCORTICAL BAND HETEROTOPIA	607432		Deletions	450
Molecular Tests	PAH (PHENYLALANINE HYDROXYLASE)	261600	PHENYLKETONURIA, PKU » HYPERPHENYLALANINEMIA » FOLLING DISEASE	261600		Sequencing	950
Molecular Tests	PALB2 (PARTNER AND LOCALIZER OF BRCA2; FANCN)	610355	BREAST CANCER			Sequencing	880
Molecular Tests	PALB2 (PARTNER AND LOCALIZER OF BRCA2; FANCN)	610355	FANCONI ANEMIA, COMPLEMENTATION GROUP N, FANCN	610832		Sequencing	880
Molecular Tests	PALB2 (PARTNER AND LOCALIZER OF BRCA2; FANCN)	610355	PANCREATIC CANCER, SUSCEPTIBILITY TO, TYPE 3	613348		Sequencing	880
Molecular Tests	PANK2 (PANTOTHENATE KINASE 2)	606157	HALLERVORDEN-SPATZ DISEASE » PANTOTHENATE KINASE-ASSOCIATED NEURODEGENERATION, PKAN » NEUROAXONAL DYSTROPHY, JUVENILE ONSET	234200		Sequencing	500
Molecular Tests	PANK2 (PANTOTHENATE KINASE 2)	606157	HARP SYNDROME (HYPOPREBETALIPOPROTEINEMIA, ACANTHOCYTOSIS, RETINITIS PIGMENTOSA, AND PALLIDAL DEGENERATION)	607236		Sequencing	500
Molecular Tests	PARKIN (PARK2)	602544	PARKINSON DISEASE, TYPE 2, PARK2 (AUTOSOMAL RECESSIVE)	600116		Sequencing	840
Molecular Tests	PARKIN (PARK2)	602544	PARKINSON DISEASE, TYPE 2, PARK2 (AUTOSOMAL RECESSIVE)	600116		Deletions	840
Molecular Tests	PAX2 (PAIRED BOX GENE 2)	167409	RENAL-COLOBOMA SYNDROME » PAPILLORENAL SYNDROME	120330		Exons 2-5, 7 en 9	1380
Molecular Tests	PAX2 (PAIRED BOX GENE 2)	167409	RENAL HYPOPLASIA	191830		Exons 2-5, 7 en 9	1380
Molecular Tests	PAX3 (PAIRED BOX GENE 3)	606597	WAARDENBURG SYNDROME, TYPE 3, WS3 » KLEIN-WAARDENBURG SYNDROME	148820		Sequencing	730
Molecular Tests	PAX3 (PAIRED BOX GENE 3)	606597	WAARDENBURG SYNDROME, TYPE 1, WS1	193500		Sequencing	730
Molecular Tests	PAX3 (PAIRED BOX GENE 3)	606597	WAARDENBURG SYNDROME, TYPE 1, WS1	193500		Deletion Analysis (MLPA)	550
Molecular Tests	PAX4 (PAIRED BOX GENE 4)	167413	MATURITY-ONSET DIABETES OF THE YOUNG, TYPE 9, MODY9	612225		Sequencing	550
Molecular Tests	PAX4 (PAIRED BOX GENE 4)	167413	DIABETES MELLITUS, KETOSIS-PRONE	612227		Sequencing	550
Molecular Tests	PAX6 (PAIRED BOX GENE 6)	607108	CATARACT, CONGENITAL, WITH LATE-ONSET CORNEAL DYSTROPHY			Deletion Analysis (MLPA)	350
Molecular Tests	PAX6 (PAIRED BOX GENE 6)	607108	CATARACT, CONGENITAL, WITH LATE-ONSET CORNEAL DYSTROPHY			Sequencing	580
Molecular Tests	PAX6 (PAIRED BOX GENE 6)	607108	ANIRIDIA, TYPE 2, AN2	106210		Deletion Analysis (MLPA)	350
Molecular Tests	PAX6 (PAIRED BOX GENE 6)	607108	ANIRIDIA, TYPE 2, AN2	106210		Sequencing	580
Molecular Tests	PAX6 (PAIRED BOX GENE 6)	607108	ECTOPIA PUPILLAE	129750		Deletion Analysis (MLPA)	350
Molecular Tests	PAX6 (PAIRED BOX GENE 6)	607108	ECTOPIA PUPILLAE	129750		Sequencing	580
Molecular Tests	PAX6 (PAIRED BOX GENE 6)	607108	FOVEAL HYPOPLASIA AND PRESENILE CATARACT SYNDROME	136520		Deletion Analysis (MLPA)	350
Molecular Tests	PAX6 (PAIRED BOX GENE 6)	607108	FOVEAL HYPOPLASIA AND PRESENILE CATARACT SYNDROME	136520		Sequencing	580
Molecular Tests	PAX6 (PAIRED BOX GENE 6)	607108	KERATITIS	148190		Deletion Analysis (MLPA)	350
Molecular Tests	PAX6 (PAIRED BOX GENE 6)	607108	KERATITIS	148190		Sequencing	580
Molecular Tests	PAX6 (PAIRED BOX GENE 6)	607108	PETERS ANOMALY	604229		Deletion Analysis (MLPA)	350
Molecular Tests	PAX6 (PAIRED BOX GENE 6)	607108	PETERS ANOMALY	604229		Sequencing	580
Molecular Tests	PAX8 (PAIRED BOX GENE 8)	167415	HYPOTHYROIDISM, CONGENITAL, NONGOITROUS, TYPE 2 » THYROID DYSGENESIS » THYROID AGENESIS » THYROID HYPOPLASIA » THYROID, ECTOPIC » HYPOTHYROIDISM, CONGENITAL, DUE TO THYROID DYSGENESIS » HYPOTHYROIDISM, ATHYREOTIC	218700		Sequencing	1310
Molecular Tests	PAX9 (PAIRED BOX GENE 9)	167416	HYPODONTIA/OLIGODONTIA, TYPE 3 » TOOTH AGENESIS, SELECTIVE, TYPE 3	604625		Sequencing	400
Molecular Tests	PC (PYRUVATE CARBOXYLASE)	608786	PYRUVATE CARBOXYLASE DEFICIENCY » LEIGH NECROTIZING ENCEPHALOPATHY DUE TO PYRUVATE CARBOXYLASE DEFICIENCY » ATAXIA WITH LACTIC ACIDOSIS, TYPE 2	266150		Sequencing	1650
Molecular Tests	PCCA (PROPIONYL-CoA CARBOXYLASE, ALPHA SUBUNIT)	232000	PROPIONIC ACIDEMIA » PROPIONYL-CoA CARBOXYLASE DEFICIENCY » GLYCINEMIA, KETOTIC » HYPERGLYCINEMIA WITH KETOACIDOSIS AND LEUKOPENIA	606054		Sequencing	1300

Molecular Tests	PCCB (PROPIONYL-CoA CARBOXYLASE, BETA SUBUNIT)	232050	PROPIONIC ACIDEMIA » PROPIONYL-CoA CARBOXYLASE DEFICIENCY » GLYCINEMIA, KETOTIC » HYPERGLYCINEMIA WITH KETOACIDOSIS AND LEUKOPENIA	606054		Sequencing	870
Molecular Tests	PCDH19 (PROTOCADHERIN 19; KIAA1313)	300460	EPILEPTIC ENCEPHALOPATHY, EARLY INFANTILE, TYPE 9, EIEE9 » JUBERG-HELLMAN SYNDROME » EPILEPSY, FEMALE-RESTRICTED, WITH MENTAL RETARDATION	300088		Sequencing	1540
Molecular Tests	PCNT (PERICENTRIN; KENDRIN)	605925	MICROCEPHALIC OSTEODYSPLASTIC PRIMORDIAL DWARFISM, TYPE 2, MOPD2	210720		Sequencing	2350
Molecular Tests	PCNT (PERICENTRIN; KENDRIN)	605925	MICROCEPHALIC OSTEODYSPLASTIC PRIMORDIAL DWARFISM, TYPE 2, MOPD2	210720		Selected Exons	770
Molecular Tests	PCSK9 (PROPROTEIN CONVERTASE, SUBTILISIN/KEXIN-TYPE, 9; NEURAL APOPTOSIS-REGULATED CONVERTASE 1; NARC1)	607786	HYPERCHOLESTEROLEMIA, AUTOSOMAL DOMINANT, TYPE 3, HCHOLA3 » LOW DENSITY LIPOPROTEIN CHOLESTEROL LEVEL QUANTITATIVE TRAIT LOCUS 1	603776		Sequencing	700
Molecular Tests	PDCD10 (PROGRAMMED CELL DEATH 10)	609118	CEREBRAL CAVERNOUS MALFORMATIONS, TYPE 3 » CAVERNOUS ANGIOMA, FAMILIAL, TYPE 3 » HYPERKERATOTIC CUTANEOUS CAPILLARY-VENOUS MALFORMATIONS ASSOCIATED WITH CEREBRAL CAPILLARY MALFORMATIONS, TYPE 3	603285		Sequencing	1700
Molecular Tests	PDE6A (PHOSPHODIESTERASE 6A, cGMP-SPECIFIC, ROD, ALPHA; RETINAL ROD PHOTORECEPTOR cGMP PHOSPHODIESTERASE, ALPHA SUBUNIT)	180071	RETINITIS PIGMENTOSA, TYPE 43, RP43	613810		Sequencing	770
Molecular Tests	PDE6B (PHOSPHODIESTERASE 6B, cGMP-SPECIFIC, ROD, BETA; RETINAL ROD PHOTORECEPTOR cGMP PHOSPHODIESTERASE, BETA SUBUNIT; RD, MOUSE, HOMOLOG OF)	180072	NIGHT BLINDNESS, CONGENITAL STATIONARY, TYPE 2 (AUTOSOMAL DOMINANT) » NIGHT BLINDNESS, CONGENITAL STATIONARY, RAMBUSCH TYPE	163500		Sequencing	820
Molecular Tests	PDE6B (PHOSPHODIESTERASE 6B, cGMP-SPECIFIC, ROD, BETA; RETINAL ROD PHOTORECEPTOR cGMP PHOSPHODIESTERASE, BETA SUBUNIT; RD, MOUSE, HOMOLOG OF)	180072	RETINITIS PIGMENTOSA, TYPE 40, RP40	613801		Sequencing	820
Molecular Tests	PDE6C (PHOSPHODIESTERASE 6C, cGMP-SPECIFIC, CONE, ALPHA-PRIME; PDEA2)	600827	ACHROMATOPSIA, TYPE 5 » CONE DYSTROPHY, TYPE 4	613093		Sequencing	890
Molecular Tests	PDE6G (PHOSPHODIESTERASE 6G, cGMP-SPECIFIC, ROD, GAMMA; RETINAL ROD PHOTORECEPTOR cGMP PHOSPHODIESTERASE, GAMMA SUBUNIT)	180073	RETINITIS PIGMENTOSA, TYPE 57, RP57	613582		Sequencing	440
Molecular Tests	PDHA1 (PYRUVATE DEHYDROGENASE COMPLEX, E1-ALPHA POLYPEPTIDE 1)	300502	CYTOCHROME c OXIDASE DEFICIENCY » COX DEFICIENCY » COMPLEX 4, MITOCHONDRIAL RESPIRATORY CHAIN, DEFICIENCY OF	220110		Sequencing	1340
Molecular Tests	PDHA1 (PYRUVATE DEHYDROGENASE COMPLEX, E1-ALPHA POLYPEPTIDE 1)	300502	LEIGH SYNDROME	256000		Sequencing	1340
Molecular Tests	PDHA1 (PYRUVATE DEHYDROGENASE COMPLEX, E1-ALPHA POLYPEPTIDE 1)	300502	PYRUVATE DECARBOXYLASE DEFICIENCY » ATAXIA, INTERMITTENT, WITH ABNORMAL PYRUVATE METABOLISM » PYRUVATE DEHYDROGENASE DEFICIENCY » PDH DEFICIENCY » ATAXIA, INTERMITTENT, WITH PYRUVATE DEHYDROGENASE, OR DECARBOXYLASE, DEFICIENCY » ATAXIA WITH LACTIC ACIDOSIS I LACTIC ACIDEMIA, THIAMINE-RESPONSIVE » THIAMINE-RESPONSIVE LACTIC ACIDEMIA	312170		Sequencing	1340
Molecular Tests	PDHB (PYRUVATE DEHYDROGENASE, BETA POLYPEPTIDE)	179060	PYRUVATE DEHYDROGENASE E1-BETA DEFICIENCY	614111		Sequencing	1060
Molecular Tests	PDHX (PYRUVATE DEHYDROGENASE COMPLEX, COMPONENT X; PYRUVATE DEHYDROGENASE COMPLEX, E3-BINDING PROTEIN SUBUNIT; E3BP; PDX1)	608769	PYRUVATE DEHYDROGENASE E3-BINDING PROTEIN DEFICIENCY » LACTIC ACIDEMIA DUE TO DEFECT IN LIPOYL-CONTAINING COMPONENT X OF THE PYRUVATE DEHYDROGENASE COMPLEX	245349		Sequencing	2390
Molecular Tests	PDP1 (PYRUVATE DEHYDROGENASE PHOSPHATASE CATALYTIC SUBUNIT 1; PROTEIN PHOSPHATASE, MAGNESIUM-DEPENDENT)	605993	PYRUVATE DEHYDROGENASE PHOSPHATASE DEFICIENCY » LACTIC ACIDEMIA WITH PYRUVATE DEHYDROGENASE PHOSPHATASE DEFICIENCY	608782		Sequencing	580
Molecular Tests	PDSS1 (PRENYL DIPHOSPHATE SYNTHASE, SUBUNIT 1)	607429	COENZYME Q10 DEFICIENCY	607426		Sequencing	800
Molecular Tests	PDSS2 (PRENYL DIPHOSPHATE SYNTHASE, SUBUNIT 2)	610564	COENZYME Q10 DEFICIENCY	607426		Sequencing	650
Molecular Tests	PDX1 (PANCREAS / DUODENUM HOMEOBOX PROTEIN 1; INSULIN PROMOTER FACTOR 1; IPF1; HOMEODOMAIN TRANSCRIPTION FACTOR IPF1; SOMATOSTATIN TRANSCRIPTION FACTOR 1; STF1)	600733	PANCREATIC AGENESIS, CONGENITAL » PANCREATIC HYPOPLASIA, CONGENITAL	260370		Sequencing	250
Molecular Tests	PDX1 (PANCREAS / DUODENUM HOMEOBOX PROTEIN 1; INSULIN PROMOTER FACTOR 1; IPF1; HOMEODOMAIN TRANSCRIPTION FACTOR IPF1; SOMATOSTATIN TRANSCRIPTION FACTOR 1; STF1)	600733	MATURITY-ONSET DIABETES OF THE YOUNG, TYPE 4, MODY4	606392		Sequencing	250
Molecular Tests	PEX1 (PEROXISOME BIOGENESIS FACTOR 1)	602136	ADRENOLEUKODYSTROPH, AUTOSOMAL, NEONATAL FORM, NALD	202370		Sequencing	1100
Molecular Tests	PEX1 (PEROXISOME BIOGENESIS FACTOR 1)	602136	ADRENOLEUKODYSTROPH, AUTOSOMAL, NEONATAL FORM, NALD	202370		Exons 13, 15, 18, 19	600
Molecular Tests	PEX1 (PEROXISOME BIOGENESIS FACTOR 1)	602136	ZELLWEGER SYNDROME » CEREBROHEPATORENAL SYNDROME	214100		Sequencing	1100
Molecular Tests	PEX1 (PEROXISOME BIOGENESIS FACTOR 1)	602136	ZELLWEGER SYNDROME » CEREBROHEPATORENAL SYNDROME	214100		Exons 13, 15, 18, 19	600
Molecular Tests	PEX1 (PEROXISOME BIOGENESIS FACTOR 1)	602136	REFSUM DISEASE, INFANTILE FORM » INFANTILE PHYTANIC ACID STORAGE DISEASE	266510		Sequencing	1100
Molecular Tests	PEX1 (PEROXISOME BIOGENESIS FACTOR 1)	602136	REFSUM DISEASE, INFANTILE FORM » INFANTILE PHYTANIC ACID STORAGE DISEASE	266510		Exons 13, 15, 18, 19	600

Molecular Tests	PEX1, PEX6, PEX10, PEX12 and PEX26		ADRENOLEUKODYSTROPH, AUTOSOMAL NEONATAL FORM_NALD	202370		PEX1 (exons 13, 15, 18), PEX6 (Exon 5), PEX10 (Exons 4 and 5), PEX12 (Exons 2 and 3) and PEX26 (Exons 3 and 4)	700
Molecular Tests	PEX1, PEX6, PEX10, PEX12 and PEX26		ZELLWEGER SYNDROME + CEREBROHEPATORENAL SYNDROME	214100		PEX1 (exons 13, 15, 18), PEX6 (Exon 5), PEX10 (Exons 4 and 5), PEX12 (Exons 2 and 3) and PEX26 (Exons 3 and 4)	700
Molecular Tests	PEX1, PEX6, PEX10, PEX12 and PEX26		REFSUM DISEASE, INFANTILE FORM + INFANTILE PHYTANIC ACID STORAGE DISEASE	266510		PEX1 (exons 13, 15, 18), PEX6 (Exon 5), PEX10 (Exons 4 and 5), PEX12 (Exons 2 and 3) and PEX26 (Exons 3 and 4)	700
Molecular Tests	PEX2, PEX10, PEX12 and PEX26		ADRENOLEUKODYSTROPH, AUTOSOMAL NEONATAL FORM_NALD	202370		PEX2 (Exon 4), PEX10 (Exons 4 and 5), PEX12 (Exons 2 and 3) and PEX26 (Exons 3 and 4)	1440
Molecular Tests	PEX2, PEX10, PEX12 and PEX26		ZELLWEGER SYNDROME + CEREBROHEPATORENAL SYNDROME	214100		PEX2 (Exon 4), PEX10 (Exons 4 and 5), PEX12 (Exons 2 and 3) and PEX26 (Exons 3 and 4)	1440
Molecular Tests	PEX2, PEX10, PEX12 and PEX26		REFSUM DISEASE, INFANTILE FORM + INFANTILE PHYTANIC ACID STORAGE DISEASE	266510		PEX2 (Exon 4), PEX10 (Exons 4 and 5), PEX12 (Exons 2 and 3) and PEX26 (Exons 3 and 4)	1440
Molecular Tests	PEX6, PEX10, PEX12 and PEX26		ADRENOLEUKODYSTROPH, AUTOSOMAL NEONATAL FORM_NALD	202370		Sequencing	1900
Molecular Tests	PEX6, PEX10, PEX12 and PEX26		ZELLWEGER SYNDROME + CEREBROHEPATORENAL SYNDROME	214100		Sequencing	1900
Molecular Tests	PEX6, PEX10, PEX12 and PEX26		REFSUM DISEASE, INFANTILE FORM + INFANTILE PHYTANIC ACID STORAGE DISEASE	266510		Sequencing	1900
Molecular Tests	PEX7 (PEROXISOME BIOGENESIS FACTOR 7)	601757	RHIZOMELIC CHONDRODYSPLASIA PUNCTATA, TYPE 1 + CHONDRODYSTROPHIA CALCIFICANS PUNCTATA	215100		Sequencing	745
Molecular Tests	PEX7 (PEROXISOME BIOGENESIS FACTOR 7)	601757	REFSUM DISEASE + PHYTANIC ACID OXIDASE DEFICIENCY + HEREDITARY MOTOR AND SENSORY NEUROPATHY, TYPE 4 (HMSN4	266500		Sequencing	745
Molecular Tests	PFKM (PHOSPHOFRUCTOKINASE, MUSCLE TYPE)	606800	GLYCOGEN STORAGE DISEASE, TYPE 7 + GSD TYPE 7 + TARUI DISEASE	232800		Sequencing	1150
Molecular Tests	PGAM2 (PHOSPHOGLYCERATE MUTASE 2)	612931	GLYCOGEN STORAGE DISEASE, TYPE 10 + PHOSPHOGLYCERATE MUTASE, MUSCLE, DEFICIENCY OF + MYOPATHY, GLYCOGENIC	261670		Sequencing	500
Molecular Tests	PHEX	307800	HYPOPHOSPHATEMIC RICKETS (X-LINKED) + VITAMIN D-RESISTANT RICKETS (X-LINKED) + RICKETS, HYPOPHOSPHATEMIC, RICKETS (X-LINKED)	307800		Sequencing	900
Molecular Tests	PHEX	307800	HYPOPHOSPHATEMIC RICKETS (X-LINKED) + VITAMIN D-RESISTANT RICKETS (X-LINKED) + RICKETS, HYPOPHOSPHATEMIC, RICKETS (X-LINKED)	307800		Deletion-Duplication	550
Molecular Tests	PHEX (PHOSPHATE REGULATING ENDOPEPTIDASE HOMOLOG, X-LINKED)	300550	HYPOPHOSPHATEMIC RICKETS (X-LINKED) + VITAMIN D-RESISTANT RICKETS (X-LINKED) + RICKETS, HYPOPHOSPHATEMIC, RICKETS (X-LINKED)	307800		Sequencing	900
Molecular Tests	PHKA1 (PHOSPHORYLASE KINASE, MUSCLE, ALPHA-1 SUBUNIT)	311870	GLYCOGEN STORAGE DISEASE, TYPE 9D + GSD 9D + MUSCLE PHOSPHORYLASE KINASE DEFICIENCY + GLYCOGEN STORAGE DISEASE, TYPE 9	300559		Sequencing	1690
Molecular Tests	PHKA2 (PHOSPHORYLASE KINASE, MUSCLE, ALPHA-2 SUBUNIT)	306000	GLYCOGEN STORAGE DISEASE, TYPE 9A + GSD TYPE 9A + LIVER PHOSPHORYLASE KINASE DEFICIENCY + LIVER GLYCOGENOSIS (X-LINKED) + GLYCOGEN STORAGE DISEASE, TYPE 8	306000		Sequencing	1690
Molecular Tests	PHKB (PHOSPHORYLASE KINASE, BETA SUBUNIT)	172490	GLYCOGEN STORAGE DISEASE, TYPE 9B + PHOSPHORYLASE KINASE DEFICIENCY OF LIVER AND MUSCLE (AUTOSOMAL RECESSIVE)	261750		Sequencing	1690
Molecular Tests	PHKG1 (PHOSPHORYLASE KINASE, MUSCLE, GAMMA-1)	172470				Sequencing	Upon Request
Molecular Tests	PHKG2 (PHOSPHORYLASE KINASE, TESTIS/LIVER, GAMMA-2)	172471	GLYCOGEN STORAGE DISEASE, TYPE 9C + GSD TYPE 9C + GLYCOGENOSE, HEPATIC (AUTOSOMAL RECESSIVE)			Sequencing	580

Molecular Tests	PHKG2 (PHOSPHORYLASE KINASE, TESTIS/LIVER, GAMMA-2)	172471	CIRRHOSIS DUE TO LIVER PHOSPHORYLASE KINASE DEFICIENCY	172471		Sequencing	580
Molecular Tests	PHOX2B	603851	HIRSCHSPRUNG DISEASE WITH NEUROBLASTOMA			Sequencing	500
Molecular Tests	PHOX2B	603851	CONGENITAL CENTRAL HYPOVENTILATION SYNDROME » ONDINE CURSE » ONDINE-HIRSCHSPRUNG DISEASE	209880		Sequencing	500
Molecular Tests	PHYH (PHYTANOYL-CoA HYDROXYLASE)	602026	REFSUM DISEASE » PHYTANIC ACID OXIDASE DEFICIENCY » HEREDITARY MOTOR AND SENSORY NEUROPATHY TYPE 4, HMSN4	266500		Sequencing	850
Molecular Tests	PIGG (PHOSPHATIDYLINOSITOL GLYCAN ANCHOR BIOSYNTHESIS CLASS G PROTEIN; GP17)	616918	MENTAL RETARDATION (AUTOSOMAL RECESSIVE), TYPE 53, MRT53 » GLYCOSYLPHOSPHATIDYLINOSITOL BIOSYNTHESIS DEFECT, TYPE 13	616917		Sequencing	1150
Molecular Tests	PIGG (PHOSPHATIDYLINOSITOL GLYCAN ANCHOR BIOSYNTHESIS CLASS G PROTEIN; GP17)	616918	MENTAL RETARDATION (AUTOSOMAL RECESSIVE), TYPE 53, MRT53 » GLYCOSYLPHOSPHATIDYLINOSITOL BIOSYNTHESIS DEFECT, TYPE 13	616917		Sequencing and Deletion-Duplication Testing	1350
Molecular Tests	PIGL (PHOSPHATIDYLINOSITOL GLYCAN, CLASS L)	605947	CHIME SYNDROME » ZUNICH NEUROECTODERMAL SYNDROME » COLOBOMA, CONGENITAL HEART DISEASE, ICHTHYOSIFORM DERMATOSIS, MENTAL RETARDATION, AND EAR ANOMALIES SYNDROME	280000		Sequencing	1320
Molecular Tests	PINK1	608309	PARKINSON DISEASE, TYPE 6, PARK6 (AUTOSOMAL RECESSIVE) PARKINSON DISEASE, TYPE 6, PARK6 (AUTOSOMAL RECESSIVE)	605909		Sequencing	720
Molecular Tests	PINK1	608309	PARKINSON DISEASE, TYPE 6, PARK6 (AUTOSOMAL RECESSIVE) PARKINSON DISEASE, TYPE 6, PARK6 (AUTOSOMAL RECESSIVE)	605909		Deletions	720
Molecular Tests	PITPNM3 (PHOSPHATIDYLINOSITOL TRANSFER PROTEIN, MEMBRANE-ASSOCIATED, 3; PYK2 N-TERMINAL DOMAIN-INTERACTING RECEPTOR 1; NIR1)	608921	CONE-ROD DYSTROPHY, TYPE 5, CORD5	600977		Sequencing	770
Molecular Tests	PITX2 (PAIRED-LIKE HOMEODOMAIN TRANSCRIPTION FACTOR 2, PTX2)	#WAARDE!	IRIDOGONIODYSGENESIS SYNDROME, TYPE 2, IRID2 » IRIS HYPOPLASIA WITH EARLY-ONSET GLAUCOMA (AUTOSOMAL DOMINANT), IHGA	137600		Sequencing	840
Molecular Tests	PITX2 (PAIRED-LIKE HOMEODOMAIN TRANSCRIPTION FACTOR 2, PTX2)	#WAARDE!	IRIDOGONIODYSGENESIS SYNDROME, TYPE 2, IRID2 » IRIS HYPOPLASIA WITH EARLY-ONSET GLAUCOMA (AUTOSOMAL DOMINANT), IHGA	137600		Deletion-Duplication Testing	680
Molecular Tests	PITX2 (PAIRED-LIKE HOMEODOMAIN TRANSCRIPTION FACTOR 2, PTX2)	#WAARDE!	RIEGER SYNDROME, TYPE 1, RIEG1 » AXENFELD-RIEGER SYNDROME	180500		Sequencing	840
Molecular Tests	PITX2 (PAIRED-LIKE HOMEODOMAIN TRANSCRIPTION FACTOR 2, PTX2)	#WAARDE!	RIEGER SYNDROME, TYPE 1, RIEG1 » AXENFELD-RIEGER SYNDROME	180500		Deletion-Duplication Testing	680
Molecular Tests	PITX2 (PAIRED-LIKE HOMEODOMAIN TRANSCRIPTION FACTOR 2, PTX2)	#WAARDE!	PETERS ANOMALY	604229		Sequencing	840
Molecular Tests	PITX2 (PAIRED-LIKE HOMEODOMAIN TRANSCRIPTION FACTOR 2, PTX2)	#WAARDE!	PETERS ANOMALY	604229		Deletion-Duplication Testing	680
Molecular Tests	PKD1 (POLYCYSTIN 1) and PKD2 (POLYCYSTIN 2)	601313 and 173910	POLYCYSTIC KIDNEY DISEASE (AUTOSOMAL DOMINANT), PKD, ADPKD » ADULT POLYCYSTIC KIDNEYS	173900		Sequencing (2 Genes)	4590
Molecular Tests	PKD1 (POLYCYSTIN 1) and PKD2 (POLYCYSTIN 2)	601313 and 173910	POLYCYSTIC KIDNEY DISEASE (AUTOSOMAL DOMINANT), PKD, ADPKD » ADULT POLYCYSTIC KIDNEYS	173900		Deletion-Duplication Testing (2 Genes)	890
Molecular Tests	PKHD1 (FIBROCYSTIN, POLYDUCTIN)	606702	POLYCYSTIC KIDNEY DISEASE (AUTOSOMAL RECESSIVE), ARPKD » POLYCYSTIC KIDNEY DISEASE, INFANTILE, TYPE 1 » POLYCYSTIC KIDNEY AND HEPATIC DISEASE » CAROLI DISEASE	263200		Deletion-Duplication Testing	450
Molecular Tests	PKHD1 (FIBROCYSTIN, POLYDUCTIN)	606702	POLYCYSTIC KIDNEY DISEASE (AUTOSOMAL RECESSIVE), ARPKD » POLYCYSTIC KIDNEY DISEASE, INFANTILE, TYPE 1 » POLYCYSTIC KIDNEY AND HEPATIC DISEASE » CAROLI DISEASE	263200		Sequencing	1550
Molecular Tests	PKP2 (PLAKOPHILIN 2)	602861	ARRHYTHMOGENIC RIGHT VENTRICULAR DYSPLASIA, FAMILIAL, TYPE 9, ARVD9 » ARRHYTHMOGENIC RIGHT VENTRICULAR CARDIOMYOPATHY, TYPE 9, ARVC9	609040		Sequencing	1380
Molecular Tests	PKP2, DSP, DSG2, DSC2 PANEL		ARRHYTHMOGENIC RIGHT VENTRICULAR DYSPLASIA, FAMILIAL			Sequencing	1870
Molecular Tests	PLA2G6 (PHOSPHOLIPASE A2, GROUP 6)	603604	HALLERVORDEN-SPATZ DISEASE » PANTOTHENATE KINASE-ASSOCIATED NEURODEGENERATION, PKAN » NEUROAXONAL DYSTROPHY, JUVENILE-ONSET	234200		Sequencing	1600
Molecular Tests	PLA2G6 (PHOSPHOLIPASE A2, GROUP 6)	603604	NEUROAXONAL DYSTROPHY, INFANTILE (INAD)	256600		Sequencing	1600
Molecular Tests	PLA2G6 (PHOSPHOLIPASE A2, GROUP 6)	603604	KARAK SYNDROME	608395		Sequencing	1600
Molecular Tests	PLA2G6 (PHOSPHOLIPASE A2, GROUP 6)	603604	NEURODEGENERATION WITH BRAIN IRON ACCUMULATION, PLA2G6-RELATED	610217		Sequencing	1600
Molecular Tests	PLCE1 (PHOSPHOLIPASE C, EPSILON-1)	608414	NEPHROTIC SYNDROME, TYPE 3	610725		Sequencing	1200
Molecular Tests	PLEC1 (PLECTIN 1)	601282	EPIDERMOLYSIS BULLOSA SIMPLEX, OGNA TYPE » EPIDERMOLYSIS BULLOSA SIMPLEX 1, EBS1	131950		Sequencing	2500

Molecular Tests	PLEC1 (PLECTIN 1)	601282	MUSCULAR DYSTROPHY, LIMB-GIRDLE, WITH EPIDERMOLYSIS BULLOSA SIMPLEX	226670		Sequencing	2500
Molecular Tests	PLEKHG4 (PLECKSTRIN HOMOLOGY DOMAIN-CONTAINING PROTEIN, FAMILY G, MEMBER 4; PURATROPHIN 1)	609526	SPINOCEREBELLAR ATAXIA, 16q22-LINKED	117210		Sequencing	1700
Molecular Tests	PLEKHG4 (PLECKSTRIN HOMOLOGY DOMAIN-CONTAINING PROTEIN, FAMILY G, MEMBER 4; PURATROPHIN 1)	609526	SPINOCEREBELLAR ATAXIA, 16q22-LINKED	117210		Exon 1: (c.-16C>T)	250
Molecular Tests	PLN (PHOSPHOLAMBAN)	172405	CARDIOMYOPATHY, DILATED, TYPE 1P	609909		Sequencing	390
Molecular Tests	PLOD1 (PROCOLLAGEN-LYSINE, 2-OXOGLUTARATE 5-DIOXYGENASE; LYSYL HYDROXYLASE)	153454	EHLERS-DANLOS TYPE 6, EDS6 » EHLERS-DANLOS SYNDROME, KYPHOSCOLIOTIC TYPE	225400		Sequencing	600
Molecular Tests	PLOD1 (PROCOLLAGEN-LYSINE, 2-OXOGLUTARATE 5-DIOXYGENASE; LYSYL HYDROXYLASE)	153454	EHLERS-DANLOS TYPE 6, EDS6 » EHLERS-DANLOS SYNDROME, KYPHOSCOLIOTIC TYPE	225400		Duplication Exons 10-16	350
Molecular Tests	PLOD2 (PROCOLLAGEN-LYSINE, 2-OXOGLUTARATE 5-DIOXYGENASE 2; LYSINE HYDROXYLASE 2)	601865	BRUCK SYNDROME, TYPE 2	609220		Sequencing	1200
Molecular Tests	PLP1 (PROTEOLIPID PROTEIN 1, PLP)	300401	PELIZAEUS-MERZBACHER DISEASE, PMD	312080		Sequencing	710
Molecular Tests	PLP1 (PROTEOLIPID PROTEIN 1, PLP)	300401	PELIZAEUS-MERZBACHER DISEASE, PMD	312080		Duplication	460
Molecular Tests	PLP1 (PROTEOLIPID PROTEIN 1, PLP)	300401	FAMILIAL SPASTIC PARAPLEGIA 2 (X-LINKED), SPG2	312920		Sequencing	710
Molecular Tests	PLP1 (PROTEOLIPID PROTEIN 1, PLP)	300401	FAMILIAL SPASTIC PARAPLEGIA 2 (X-LINKED), SPG2	312920		Duplication	460
Molecular Tests	PMM2 (PHOSPHOMANNOMUTASE 2)	601785	CONGENITAL DISORDER OF GLYCOSYLATION, TYPE 1A, CDG1A » JAEKEN SYNDROME » CARBOHYDRATE-DEFICIENT GLYCOPROTEIN SYNDROME, TYPE 1A » PHOSPHOMANNOMUTASE 2 DEFICIENCY	212065		Sequencing	570
Molecular Tests	PMP22 (PERIPHERAL MYELIN PROTEIN)	601097	CHARCOT-MARIE-TOOTH DISEASE, DEMYELINATING, TYPE 1A, CMT1A » HEREDITARY MOTOR AND SENSORY NEUROPATHY 1A, HMSN1A	118220		Sequencing	600
Molecular Tests	PMP22 (PERIPHERAL MYELIN PROTEIN)	601097	CHARCOT-MARIE-TOOTH DISEASE, DEMYELINATING, TYPE 1A, CMT1A » HEREDITARY MOTOR AND SENSORY NEUROPATHY 1A, HMSN1A	118220		Duplication	450
Molecular Tests	PMP22 (PERIPHERAL MYELIN PROTEIN)	601097	CHARCOT-MARIE-TOOTH DISEASE AND DEAFNESS	118300		Sequencing	600
Molecular Tests	PMP22 (PERIPHERAL MYELIN PROTEIN)	601097	CHARCOT-MARIE-TOOTH DISEASE AND DEAFNESS	118300		Duplication	450
Molecular Tests	PMP22 (PERIPHERAL MYELIN PROTEIN)	601097	CHARCOT-MARIE-TOOTH DISEASE, DEMYELINATING, TYPE 4F, CMT4F » DEJERINE-SOTTAS HYPERTROPHIC NEUROPATHY	145900		Sequencing	600
Molecular Tests	PMP22 (PERIPHERAL MYELIN PROTEIN)	601097	HEREDITARY NEUROPATHY WITH LIABILITY TO PRESSURE PALSIES, HNPP » TOMACULOUS NEUROPATHY	162500		Sequencing	600
Molecular Tests	PMP22 (PERIPHERAL MYELIN PROTEIN)	601097	HEREDITARY NEUROPATHY WITH LIABILITY TO PRESSURE PALSIES, HNPP » TOMACULOUS NEUROPATHY	162500		Deletion	450
Molecular Tests	PMS2	600259	COLORECTAL CANCER, HEREDITARY NONPOLYPOSIS, HNPCC, TYPE 2 » LYNCH CANCER FAMILY SYNDROME, TYPE 2	114400		Sequencing	960
Molecular Tests	PMS2	600259	COLORECTAL CANCER, HEREDITARY NONPOLYPOSIS, HNPCC, TYPE 1 » LYNCH CANCER FAMILY SYNDROME, TYPE 1	114500		Sequencing	960
Molecular Tests	PMS2	600259	MISMATCH REPAIR CANCER SYNDROME » TURCOT SYNDROME BRAIN TUMOR » POLYPOSIS SYNDROME 1 » MMR DEFICIENCY	276300		Sequencing	960
Molecular Tests	PNKP (POLYNUCLEOTIDE KINASE 3-PRIME PHOSPHATASE; DNA KINASE)	605610	EPILEPTIC ENCEPHALOPATHY, EARLY INFANTILE, TYPE 10, EIEE10	613402		Sequencing	1690
Molecular Tests	PNPO (PYRIDOXAMINE 5-PRIME-PHOSPHATE OXIDASE, PYRIDOXAMINE-PHOSPHATE OXIDASE)	603287	PYRIDOXAMINE 5-PRIME-PHOSPHATE OXIDASE DEFICIENCY » PNPO DEFICIENCY » EPILEPTIC ENCEPHALOPATHY, NEONATAL, PNPO-RELATED	610090		Sequencing	620
Molecular Tests	POLG (POLYMERASE, DNA, GAMMA)	174763	PROGRESSIVE EXTERNAL OPHTHALMOPLEGIA WITH MITOCHONDRIAL DNA DELETIONS (AUTOSOMAL DOMINANT), TYPE 1 » PROGRESSIVE EXTERNAL OPHTHALMOPLEGIA (AUTOSOMAL DOMINANT), 1 » PROGRESSIVE EXTERNAL OPHTHALMOPLEGIA WITH HYPOGONADISM » OCULAR MYOPATHY WITH HYPOGONADISM	157640		Sequencing	850
Molecular Tests	POLG (POLYMERASE, DNA, GAMMA)	174763	ALPERS DIFFUSE DEGENERATION OF CEREBRAL GRAY MATTER WITH HEPATIC CIRRHOSIS » ALPERS PROGRESSIVE INFANTILE POLIODYSTROPHY » ALPERS SYNDROME » ALPERS-HUTTENLOCHER SYNDROME	203700		Sequencing	850
Molecular Tests	POLG (POLYMERASE, DNA, GAMMA)	174763	PROGRESSIVE EXTERNAL OPHTHALMOPLEGIA WITH MITOCHONDRIAL DNA DELETIONS (AUTOSOMAL RECESSIVE) » SPINOCEREBELLAR ATAXIA WITH EPILEPSY » ALPERS SYNDROME	258450		Sequencing	850
Molecular Tests	POLG (POLYMERASE, DNA, GAMMA)	174763	SENSORY ATAXIC NEUROPATHY, DYSARTHRIA, AND OPHTHALMOPARESIS, SANDO » SPINOCEREBELLAR ATAXIA WITH EPILEPSY	607459		Sequencing	850
Molecular Tests	POLG2 (POLYMERASE, DNA, GAMMA-2; POLGB)	604983	PROGRESSIVE EXTERNAL OPHTHALMOPLEGIA WITH MITOCHONDRIAL DNA DELETIONS (AUTOSOMAL DOMINANT), TYPE 4	610131		Sequencing	650

Molecular Tests	POLR3A (POLYMERASE III, RNA, SUBUNIT A; RNA POLYMERASE III, SUBUNIT C1)	614258	LEUKODYSTROPHY, HYPOMYELINATING, TYPE 7, WITH OR WITHOUT OLIGODONTIA AND/OR HYPOGONADOTROPIC HYPOGONADISM » ATAXIA, DELAYED DENTITION, AND HYPOMYELINATION	607694		Sequencing	1870
Molecular Tests	POLR3B (POLYMERASE III, RNA, SUBUNIT B)	614366	LEUKODYSTROPHY, HYPOMYELINATING, TYPE 8, WITH OR WITHOUT OLIGODONTIA AND/OR HYPOGONADOTROPIC HYPOGONADISM	614381		Sequencing	1820
Molecular Tests	POMC (PROOPIOMELANOCORTIN)	176830	PROOPIOMELANOCORTIN DEFICIENCY » OBESITY, EARLY-ONSET, ADRENAL INSUFFICIENCY, AND RED HAIR	609734		Sequencing	400
Molecular Tests	POMGNT1 (PROTEIN O-LINKED MANNOSE BETA-1,2-N-ACETYLGLUCOSAMINYLTRANSFERASE)	606822	MUSCULAR DYSTROPHY-DYSTROGLYCANOPATHY (LIMB-GIRDLE), TYPE 2N	613151		Sequencing	1430
Molecular Tests	POMT1 (PROTEIN O-MANNOSYLTRANSFERASE 1)	607423	WALKER-WARBURG SYNDROME » HYDROCEPHALUS, AGYRIA, AND RETINAL DYSPLASIA » HARD SYNDROME	236670		Sequencing	1490
Molecular Tests	POMT2 (PROTEIN O-MANNOSYLTRANSFERASE 2)	607439	WALKER-WARBURG SYNDROME » HYDROCEPHALUS, AGYRIA, AND RETINAL DYSPLASIA » HARD SYNDROME	236670		Sequencing	1490
Molecular Tests	PORCN (PORCUPINE, DROSOPHILA, HOMOLOG OF)	300651	FOCAL DERMAL HYPOPLASIA » GOLTZ SYNDROME	305600		Sequencing	1460
Molecular Tests	PITUITARY HOMEOBOX 2 (POU CLASS 1, TRANSCRIPTION FACTOR 1, GROWTH HORMONE FACTOR 1, PIT1)	173110	PITUITARY HORMONE DEFICIENCY, COMBINED	173110		Sequencing	650
Molecular Tests	POU3F4 (POU DOMAIN, CLASS 3, TRANSCRIPTION FACTOR 4)	300039	DEAFNESS, CONDUCTIVE, WITH STAPES FIXATION, DFN3 » PERILYMPHATIC GUSHER DEAFNESS SYNDROME			Sequencing	500
Molecular Tests	PPARG (PEROXISOME PROLIFERATOR-ACTIVATED RECEPTOR-GAMMA, PPARG1, PPARG2, PPARG3, PAX8/PPARG FUSION GENE)	601487	DIABETES MELLITUS, NONINSULIN-DEPENDENT, NIDDM » DIABETES MELLITUS, TYPE 2 » NONINSULIN-DEPENDENT DIABETES MELLITUS » MATURITY-ONSET DIABETES	125853		Sequencing	500
Molecular Tests	PPARG (PEROXISOME PROLIFERATOR-ACTIVATED RECEPTOR-GAMMA, PPARG1, PPARG2, PPARG3, PAX8/PPARG FUSION GENE)	601487	DIABETES MELLITUS, NONINSULIN-DEPENDENT, NIDDM » DIABETES MELLITUS, TYPE 2 » NONINSULIN-DEPENDENT DIABETES MELLITUS » MATURITY-ONSET DIABETES	125853		2 Mutations: P12A and P115Q	Upon Request
Molecular Tests	PPARG (PEROXISOME PROLIFERATOR-ACTIVATED RECEPTOR-GAMMA, PPARG1, PPARG2, PPARG3, PAX8/PPARG FUSION GENE)	601487	OBESITY	601665		Sequencing	500
Molecular Tests	PPARG (PEROXISOME PROLIFERATOR-ACTIVATED RECEPTOR-GAMMA, PPARG1, PPARG2, PPARG3, PAX8/PPARG FUSION GENE)	601487	OBESITY	601665		2 Mutations: P12A and P115Q	Upon Request
Molecular Tests	PPIB (PEPTIDYL-PROLYL ISOMERASE B; CYCLOPHILIN B; CYPB)	123841	OSTEOGENESIS IMPERFECTA TYPE 9, OI9 » OSTEOGENESIS IMPERFECTA, SILENCE TYPE II-III, WITHOUT ABNORMALITY OF TYPE I COLLAGEN	259440		Sequencing	550
Molecular Tests	PPOX (PROTOPORPHYRINOGEN OXIDASE)	600923	PORPHYRIA VARIEGATA » PROTOPORPHYRINOGEN OXIDASE DEFICIENCY » HEREDITARY COPROPORPHYRIA	176200		Sequencing	810
Molecular Tests	PPP2R2B (PROTEIN PHOSPHATASE 2, REGULATORY SUBUNIT B, BETA)	604325	SPINOCEREBELLAR ATAXIA 12, SCA12	604326	See SCA8, SCA10, SCA12, SCA17	Sequencing	150
Molecular Tests	PPT1 (PALMITOYL-PROTEIN THIOESTERASE)	600722	CEROID LIPOFUSCINOSIS, CLN1 » SANTAVUORI DISEASE	256730		Sequencing	1090
Molecular Tests	PQBP1 (POLYGLUTAMINE-BINDING PROTEIN 1)	300463	MENTAL RETARDATION, SYNDROMIC (X-LINKED), TYPE 1, MRXS1 » SUTHERLAND-HAAN MENTAL RETARDATION SYNDROME (X-LINKED) » MENTAL RETARDATION, NONSPECIFIC (X-LINKED), TYPE 1, MRX51	309470		Sequencing	500
Molecular Tests	PRKAG2 (PROTEIN KINASE, AMP-ACTIVATED, NONCATALYTIC, GAMMA-2, AMP-ACTIVATED PROTEIN KINASE, NONCATALYTIC, GAMMA-2 AMP-GAMMA-2)	602743	GLYCOGEN STORAGE DISEASE OF HEART, LETHAL CONGENITAL » PHOSPHORYLASE KINASE DEFICIENCY OF HEART » GLYCOGEN STORAGE DISEASE OF HEART	261740		Sequencing	760
Molecular Tests	PRKAG2 (PROTEIN KINASE, AMP-ACTIVATED, NONCATALYTIC, GAMMA-2, AMP-ACTIVATED PROTEIN KINASE, NONCATALYTIC, GAMMA-2 AMP-GAMMA-2)	602743	CARDIOMYOPATHY, FAMILIAL HYPERTROPHIC, WITH WOLFF-PARKINSON-WHITE SYNDROME	600858		Sequencing	760
Molecular Tests	PRKAR1A	188830	CARNEY COMPLEX, TYPE 1 » PRIMARY PIGMENTED NODULAR ADRENOCORTICAL DISEASE	160980		Sequencing	250
Molecular Tests	PRKAR1A	188830	CARNEY MYXOMA	255960		Sequencing	250

Molecular Tests	PRKCG (PROTEIN KINASE C, GAMMA, PKCC)	176980	SPINOCEREBELLAR ATAXIA 14, SCA14	605361		Sequencing	1200
Molecular Tests	PRKCSH (PROTEIN KINASE C SUBSTRATE, 80-KD, HEAVY CHAIN; GLUCOSIDASE II, BETA SUBUNIT; HEPATOCYSTIN)	177060	POLYCYSTIC LIVER DISEASE	174050		Sequencing	1390
Molecular Tests	PRNP (PRION PROTEIN)	176640	CREUTZFELDT-JAKOB DISEASE, CJD	123400		Sequencing	300
Molecular Tests	PRNP (PRION PROTEIN)	176640	GERSTMANN-STRAUSSLER DISEASE, GSD	137440		Sequencing	300
Molecular Tests	PRNP (PRION PROTEIN)	176640	INSOMNIA-DYSAUTONOMIA » FAMILIAL FATAL INSOMNIA	600072		Sequencing	300
Molecular Tests	PRNP (PRION PROTEIN)	176640	HUNTINGTON DISEASE-LIKE 1, HDL1	603218		Sequencing	300
Molecular Tests	PROC (PROTEIN C)	176860	THROMBOEMBOLISM	176860		Sequencing	Upon Request
Molecular Tests	PROC (PROTEIN C)	176860	THROMBOEMBOLISM	176860		Deletion-Duplication Testing	Upon Request
Molecular Tests	PROK2 (PROKINETICIN 2)	607002	KALLMANN SYNDROME, TYPE 4, KAL4 » HYPOGONADOTROPIC HYPOGONADISM AND ANOSMIA, TYPE 4	610628		Sequencing	600
Molecular Tests	PROKR2 (PROKINETICIN RECEPTOR 2; G PROTEIN-COUPLED RECEPTOR 73-LIKE 1)	607123	KALLMANN SYNDROME, TYPE 3, KAL3 » HYPOGONADOTROPIC HYPOGONADISM AND ANOSMIA, TYPE 3	244200		Sequencing	400
Molecular Tests	PROM1 (PROMININ 1; PROMININ, MOUSE, HOMOLOG-LIKE 1; PROML1)	604365	STARGARDT DISEASE, TYPE 4	603786		Sequencing	770
Molecular Tests	PROM1 (PROMININ 1; PROMININ, MOUSE, HOMOLOG-LIKE 1; PROML1)	604365	MACULAR DYSTROPHY, RETINAL, TYPE 2	608051		Sequencing	770
Molecular Tests	PROM1 (PROMININ 1; PROMININ, MOUSE, HOMOLOG-LIKE 1; PROML1)	604365	RETINITIS PIGMENTOSA, TYPE 41, RP41	612095		Sequencing	770
Molecular Tests	PROM1 (PROMININ 1; PROMININ, MOUSE, HOMOLOG-LIKE 1; PROML1)	604365	CONE-ROD DYSTROPHY, TYPE 12, CORD12	612657		Sequencing	770
Molecular Tests	PROP1 (PROPHET OF PIT1, PAIRED-LIKE HOMEODOMAIN TRANSCRIPTION FACTOR)	601538	PITUITARY DWARFISM 3 » PANHYPOPITUITARISM » ATELIOTIC DWARFISM WITH HYPOGONADISM » HANHART DWARFISM PITUITARY HORMONE DEFICIENCY, COMBINED » PITUITARY HORMONE DEFICIENCY, COMBINED WITH RIGID CERVICAL SPINE	262600		Sequencing	540
Molecular Tests	PROS1 (PROTEIN S)	176880	THROMBOEMBOLISM	176880		Sequencing	600
Molecular Tests	PROS1 (PROTEIN S)	176880	THROMBOEMBOLISM	176880		Deletion-Duplication Testing	350
Molecular Tests	PRPF3 (PRECURSOR mRNA-PROCESSING FACTOR 3, S. CEREVISIAE, HOMOLOG OF; PRP3)	607301	RETINITIS PIGMENTOSA, TYPE 18, RP18	601414		Sequencing	680
Molecular Tests	PRPF31 (PRP31, PRECURSOR mRNA-PROCESSING FACTOR 31, S. CEREVISIAE, HOMOLOG OF)	606419	RETINITIS PIGMENTOSA, TYPE 11, RP11	600138		Sequencing	680
Molecular Tests	PRPF6 (PRECURSOR mRNA-PROCESSING FACTOR 6, S. CEREVISIAE, HOMOLOG OF; PRP6; ANDROGEN RECEPTOR N-TERMINAL DOMAIN-TRANSACTIVATING PROTEIN 1; ANT1)	613979	RETINITIS PIGMENTOSA, TYPE 60, RP60	613983		Sequencing	770
Molecular Tests	PRPF8 (PRECURSOR mRNA-PROCESSING FACTOR 8, S. CEREVISIAE, HOMOLOG OF)	607300	RETINITIS PIGMENTOSA, TYPE 13, RP13	600059		Sequencing	1010
Molecular Tests	PRPH2 (PERIPHERIN 2, MOUSE, HOMOLOG OF, RDS)	179605	MACULAR DYSTROPHY			Sequencing	350
Molecular Tests	PRPH2 (PERIPHERIN 2, MOUSE, HOMOLOG OF, RDS)	179605	FUNDUS ALBIPUNCTATUS » RETINITIS PUNCTATA ALBESCENS	136880		Sequencing	350
Molecular Tests	PRPH2 (PERIPHERIN 2, MOUSE, HOMOLOG OF, RDS)	179605	PATTERNED DYSTROPHY OF RETINAL PIGMENT EPITHELIUM » MACULAR DYSTROPHY, BUTTERFLY-SHAPED PIGMENTARY » BUTTERFLY DYSTROPHY OF RETINAL PIGMENT EPITHELIUM	169150		Sequencing	350
Molecular Tests	PRPH2 (PERIPHERIN 2, MOUSE, HOMOLOG OF, RDS)	179605	STARGARDT DISEASE, TYPE 1 » MACULAR DEGENERATION, JUVENILE » FUNDUS FLAVIMACULATUS » MACULAR DYSTROPHY WITH FLECKS, TYPE 1	248200		Sequencing	350
Molecular Tests	PRPH2 (PERIPHERIN 2, MOUSE, HOMOLOG OF, RDS)	179605	RETINITIS PIGMENTOSA, TYPE 7, RP7	608133		Sequencing	350
Molecular Tests	PRPH2 (PERIPHERIN 2, MOUSE, HOMOLOG OF, RDS)	179605	MACULAR DYSTROPHY, VITELLIFORM, ADULT-ONSET » FOVEOMACULAR DYSTROPHY, ADULT-ONSET	608161		Sequencing	350
Molecular Tests	PRRT2 (PROLINE-RICH TRANSMEMBRANE PROTEIN 2)	614386	EPISODIC KINESIGENIC DYSKINESIA 1 » PAROXYSMAL KINESIGENIC CHOREOATHETOSIS » DYSTONIA TYPE 10, DYT10	128200		Sequencing	450
Molecular Tests	PRSS1 (PROTEASE, SERINE 1)	276000	PANCREATITIS, HEREDITARY, PCTT	167800		Exons 1-3 of PRSS1 and exon 3 of SPINK1	300
Molecular Tests	PRX (PERIAXIN)	605725	CHARCOT-MARIE-TOOTH DISEASE, DEMYELINATING, TYPE 4F, CMT4F » DEJERINE-SOTTAS HYPERTROPHIC NEUROPATHY » HEREDITARY MOTOR AND SENSORY NEUROPATHY TYPE 3, HMSN3	145900		Sequencing	1100
Molecular Tests	PSAP (PROSAPOSIN)	176801	METACHROMATIC LEUKODYSTROPHY DUE TO SAP1 DEFICIENCY » GAUCHER DISEASE DUE TO SAP2 DEFICIENCY » PROSAPOSIN DEFICIENCY	249900		Sequencing	1200
Molecular Tests	PSEN1 (PRESENILIN 1, PS1)	104311	ALZHEIMER DEMENTIA, EARLY-ONSET, TYPE 3, AD3	607822		Sequencing	590
Molecular Tests	PSEN2 (PRESENILIN 2, PS2)	600759	ALZHEIMER DEMENTIA, EARLY-ONSET, TYPE 4, AD4	606889		Sequencing	590

Molecular Tests	PSTPIP1 (PROLINE/ SERINE/ THREONINE PHOSPHATASE-INTERACTING PROTEIN 1)	606347	PYOGENIC STERILE ARTHRITIS, PYODERMA GANGRENOSUM, AND ACNE » PAPA SYNDROME » FAMILIAL RECURRENT ARTHRITIS	604416		Sequencing	1700
Molecular Tests	PTCH1 (PTCH, PATCHED, PTC)	601309	BASAL CELL NEVUS SYNDROME » GORLIN SYNDROME	109400		Whole Gene Sequencing and Deletion-Duplication Testing	1800
Molecular Tests	PTCH1 (PTCH, PATCHED, PTC)	601309	HOLOPROSENCEPHALY 7, HPE7	601309		Whole Gene Sequencing and Deletion-Duplication Testing	1800
Molecular Tests	PTEN	601728	PTEN HAMARTOMA TUMOR SYNDROME			Sequencing	765
Molecular Tests	PTEN	601728	PTEN HAMARTOMA TUMOR SYNDROME			Deletion-Duplication Testing	530
Molecular Tests	PTEN	601728	OLIGODENDROGLIOMA	137800		Sequencing	780
Molecular Tests	PTEN	601728	OLIGODENDROGLIOMA	137800		Deletion-Duplication Testing	530
Molecular Tests	PTEN	601728	BANNAYAN-MYHRE-RILEY-RUVALCAVA-SMITH-ZONANA SYNDROME	153480		Sequencing	780
Molecular Tests	PTEN	601728	BANNAYAN-MYHRE-RILEY-RUVALCAVA-SMITH-ZONANA SYNDROME	153480		Deletion-Duplication Testing	530
Molecular Tests	PTEN	601728	COWDEN DISEASE » LHERMITTE-DUCLOS DISEASE	158350		Sequencing	780
Molecular Tests	PTEN	601728	COWDEN DISEASE » LHERMITTE-DUCLOS DISEASE	158350		Deletion-Duplication Testing	530
Molecular Tests	PTEN	601728	PROTEUS SYNDROME » GIGANTISM, PARTIAL, OF HANDS AND FEET, NEVI, HEMIHYPERTROPHY, AND MACROCEPHALY » ELATTOPROTEUS SYNDROME » CONGENITAL HEMIHYPERTROPHY, LIPOMATOSIS, CAPILLARY AND	176920		Sequencing	780
Molecular Tests	PTEN	601728	PROTEUS SYNDROME » GIGANTISM, PARTIAL, OF HANDS AND FEET, NEVI, HEMIHYPERTROPHY, AND MACROCEPHALY » ELATTOPROTEUS SYNDROME » CONGENITAL HEMIHYPERTROPHY, LIPOMATOSIS, CAPILLARY AND	176920		Deletion-Duplication Testing	530
Molecular Tests	PTEN	601728	VATER-HYDROCEPHALUS SYNDROME	276950		Sequencing	765
Molecular Tests	PTEN	601728	VATER-HYDROCEPHALUS SYNDROME	276950		Deletion-Duplication Testing	530
Molecular Tests	PTPN	601738	MACROCEPHALY/AUTISM SYNDROMIC	605309		Sequencing	780
Molecular Tests	PTPN	601738	MACROCEPHALY/AUTISM SYNDROMIC	605309		Deletion-Duplication Testing	530
Molecular Tests	PTF1A (PANCREAS TRANSCRIPTION FACTOR 1, ALPHA SUBUNIT)	607194	PANCREATIC AND CEREBELLAR AGENESIS » DIABETES MELLITUS, PERMANENT NEONATAL, WITH CEREBELLAR AGENESIS	609069		Sequencing	990
Molecular Tests	PTPN11	176876	LEOPARD SYNDROME, TYPE 1	151100		Sequencing	700
Molecular Tests	PTPN11	176876	NOONAN SYNDROME, TYPE 1	163950		Sequencing	700
Molecular Tests	PTPN11	176876	JUVENILE MYELOMONOCYTIC LEUKEMIA, JMML	607785		Exons 3 and 13	600
Molecular Tests	PUS1 (PSEUDOURIDINE SYNTHASE 1)	608109	MYOPATHY, LACTIC ACIDOSIS, AND SIDEROBLASTIC ANEMIA 1	600462		Sequencing	720
Molecular Tests	PWS	176270	PRADER-WILLI SYNDROME	176270		Methylation	590
Molecular Tests	PWS	176270	PRADER-WILLI SYNDROME	176270		Deletion of Imprinting Centre	570
Molecular Tests	PYCR1 (PYRROLINE-5-CARBOXYLATE REDUCTASE 1)	179035	CUTIS LAXA, TYPE 2B (AUTOSOMAL RECESSIVE)	612940		Sequencing	550
Molecular Tests	PYGL (GLYCOGEN PHOSPHORYLASE, LIVER)	232700	GLYCOGEN STORAGE DISEASE, TYPE 6 » GSD TYPE 6 » HERS DISEASE » GLYCOGENOSIS, TYPE VI (HERS DISEASE)	232700		Sequencing	1230
Molecular Tests	PYGM (GLYCOGEN PHOSPHORYLASE, MUSCLE, MYOPHOSPHORYLASE)	608455	GLYCOGEN STORAGE DISEASE, TYPE 5 » GSD TYPE 5 » MCARDLE DISEASE » GLYCOGENOSIS, TYPE V (MCARDLE DISEASE)	232600		Sequencing	1110
Molecular Tests	RAB3GAP1, RAB3GAP2, RAB18	602536 609270, 607961	WARBURG MICRO SYNDROME			Sequencing	5510
Molecular Tests	RAB7 (RAS-ASSOCIATED PROTEIN RAB7)	602298	CHARCOT-MARIE-TOOTH DISEASE, AXONAL, TYPE 2B, CMT2B	600882		Sequencing	600
Molecular Tests	RAF1 (V-RAF-1 MURINE LEUKEMIA VIRAL ONCOGENE HOMOLOG 1)	164760	NOONAN SYNDROME, TYPE 5	611553		Sequencing	1510
Molecular Tests	RAF1 (V-RAF-1 MURINE LEUKEMIA VIRAL ONCOGENE HOMOLOG 1)	164760	NOONAN SYNDROME, TYPE 5	611553		Sequencing of Exons 7, 14, 17	590
Molecular Tests	RAF1 (V-RAF-1 MURINE LEUKEMIA VIRAL ONCOGENE HOMOLOG 1)	164760	LEOPARD SYNDROME, TYPE 1	611554		Sequencing	1510
Molecular Tests	RAF1 (V-RAF-1 MURINE LEUKEMIA VIRAL ONCOGENE HOMOLOG 1)	164760	LEOPARD SYNDROME, TYPE 1	611554		Sequencing of Exons 7, 14, 17	590
Molecular Tests	RAG1 (RECOMBINATION-ACTIVATING GENE 1)	179615	SEVERE COMBINED IMMUNODEFICIENCY (AUTOSOMAL RECESSIVE), T CELL-NEGATIVE, B CELL-NEGATIVE, NK CELL-POSITIVE » SCID, T CELL-NEGATIVE, B CELL-NEGATIVE, NK CELL-POSITIVE	601457		Testing of both RAG1 and RAG2	1930

Molecular Tests	RAG2 (RECOMBINATION-ACTIVATING GENE 2)	179616	OMENN SYNDROME x RETICULOENDOTHELIOSIS, FAMILIAL, WITH EOSINOPHILIA x SEVERE COMBINED IMMUNODEFICIENCY WITH HYPEREOSINOPHILIA	603554		Testing of both RAG1 and RAG2	1930
Molecular Tests	RAI1 (RETINOIC ACID-INDUCED GENE 1, RAI1)	607642	SMITH-MAGENIS SYNDROME	182290		Sequencing and Deletion-Duplication Testing	2090
Molecular Tests	RAPSN (RAPSYN, RECEPTOR-ASSOCIATED PROTEIN OF THE SYNAPSE, 43-KD)	601592	FETAL AKINESIA DEFORMATION SEQUENCE x PENA-SHOKEIR SYNDROME, TYPE 1 x MYASTHENIC SYNDROME, CONGENITAL, TYPE 1A	208150		Sequencing	750
Molecular Tests	RAPSN (RAPSYN, RECEPTOR-ASSOCIATED PROTEIN OF THE SYNAPSE, 43-KD)	601592	MYASTHENIC SYNDROME, CONGENITAL, ASSOCIATED WITH ACETYLCHOLINE RECEPTOR DEFICIENCY x MYASTHENIC SYNDROME, CONGENITAL, TYPE 1A x MYASTHENIC SYNDROME, CONGENITAL, ASSOCIATED WITH FACIAL DYSMORPHISM	608931		Sequencing	750
Molecular Tests	RARS2 (ARGINYL-tRNA SYNTHETASE 2; RARS1)	611524	PONTOCEREBELLAR HYPOPLASIA, TYPE 6 x ENCEPHALOPATHY, FATAL INFANTILE, WITH MITOCHONDRIAL RESPIRATORY CHAIN DEFECTS x COMBINED MITOCHONDRIAL RESPIRATORY CHAIN DEFICIENCY	611523		Sequencing	1960
Molecular Tests	RAX2 (RETINA AND ANTERIOR NEURAL FOLD HOMEOBOX-LIKE 1; OSG-TYPE RETINAL HOMEOBOX; ORX1	610567	CONE-ROD DYSTROPHY, TYPE 11, CORD11	610381		Sequencing	960
Molecular Tests	RAX2 (RETINA AND ANTERIOR NEURAL FOLD HOMEOBOX-LIKE 1; OSG-TYPE RETINAL HOMEOBOX; ORX1	610567	MACULAR DEGENERATION, AGE-RELATED, TYPE 6, ARMD6	613757		Sequencing	960
Molecular Tests	RB1	180200	RETINOBLASTOMA, RB1	180200		Sequencing	2160
Molecular Tests	RB1	180200	RETINOBLASTOMA, RB1	180200		Deletion-Duplication Testing	600
Molecular Tests	RBMBA	274300	THROMBOCYTOPENIA-ABSENT RADIUS SYNDROME x TAR SYNDROME x THROMBOCYTOPENIA, THROMBOCYTOPENIA SYNDROME	274000		Common Deletion	600
Molecular Tests	RBP3 (RETINOL-BINDING PROTEIN 3; INTERSTITIAL RETINOL-BINDING PROTEIN)	180290	RETINITIS PIGMENTOSA (AUTOSOMAL RECESSIVE)			Sequencing	680
Molecular Tests	RBPJ (RECOMBINATION SIGNAL-BINDING PROTEIN FOR IMMUNOGLOBULIN KAPPA J REGION; C PROMOTER-BINDING FACTOR 1; CBF1)	147183	ADAMS-OLIVER SYNDROME, TYPE 3	614814		Sequencing	850
Molecular Tests	RDH12 (RETINOL DEHYDROGENASE 12)	608830	LEBER CONGENITAL AMAUROSIS, TYPE 3, LCA3 x AMAUROTIC CONGENITA OF LEBER 3	604232		Sequencing	680
Molecular Tests	RDH5 (RETINOL DEHYDROGENASE 5)	601617	FUNDUS ALBIPUNCTATUS x RETINITIS PUNCTATA ALBESCENS	136880		Sequencing	550
Molecular Tests	RECQL2 (RECQ PROTEIN-LIKE 2)	604611	WERNER SYNDROME	277700		Sequencing	1400
Molecular Tests	RECQL4 (RECQ PROTEIN-LIKE 4)	604610	ROTHMUND-THOMSON SYNDROME	268400		Sequencing	1700
Molecular Tests	RECQL4 (RECQ PROTEIN-LIKE 4)	604610	BLOOM SYNDROME	210900		3 Mutations - 2385delA / ins7	250
Molecular Tests	RECQL4 (RECQ PROTEIN-LIKE 4, DNA HELICASE, RECQ-LIKE, TYPE 4)	603780	BALLER-GEROLD SYNDROME x CRANIOSYNOSTOSIS WITH RADIAL DEFECTS x CRANIOSYNOSTOSIS, RADIAL APLASIA SYNDROME	218600		Sequencing	1810
Molecular Tests	RECQL4 (RECQ PROTEIN-LIKE 4, DNA HELICASE, RECQ-LIKE, TYPE 4)	603780	RADIAL HYPOPLASIA SYNDROME	266780		Sequencing	1810
Molecular Tests	RECQL4 (RECQ PROTEIN-LIKE 4, DNA HELICASE, RECQ-LIKE, TYPE 4)	603780	ROTHMUND-THOMSON SYNDROME x POIKILODERMA ATROPHICANS AND CATARACT	268400		Sequencing	1810
Molecular Tests	REEP1 (RECEPTOR EXPRESSION-ENHANCING PROTEIN 1)	609139	SPASTIC PARAPLEGIA 31, SPG31 (AUTOSOMAL DOMINANT)	610250		Sequencing	500
Molecular Tests	REN (RENIN)	179820	HYPOPLASTIC RENAL DYSGENESIS	179830		Sequencing	650
Molecular Tests	REN (RENIN)	179820	RENAL TUBULAR DYSGENESIS x RENAL TUBULAR DYSGENESIS, WITH CHOANAL ATRESIA AND ATHELIA	267430		Sequencing	650
Molecular Tests	RET (RET KINASE)	164761	HIRSCHSPRUNG DISEASE, TYPE 1 x AGANGLIONIC MEGACOLON	142623		Sequencing	1275
Molecular Tests	RET (RET KINASE)	164761	MEDULLARY THYROID CARCINOMA, MTC	155240		Exons 10-11, 13-16	590
Molecular Tests	RET (RET KINASE)	164761	MULTIPLE ENDOCRINE NEOPLASIA, TYPE 2B, MEN2B	162300		Sequencing	1275
Molecular Tests	RET (RET KINASE)	164761	MULTIPLE ENDOCRINE NEOPLASIA, TYPE 2B, MEN2B	162300		Exons 10-11, 13-16	590
Molecular Tests	RET (RET KINASE)	164761	MULTIPLE ENDOCRINE NEOPLASIA, TYPE 2A, MEN2A	171400		Exons 10-11, 13-16	590
Molecular Tests	RET (RET KINASE)	164761	RENAL AGENESIS	191830		Sequencing	1275
Molecular Tests	RFT1 (RFT1, S. CEREVISIAE, HOMOLOG OF)	611908	CONGENITAL DISORDER OF GLYCOSYLATION, TYPE 1N, CDG1N	612015		Sequencing	1730
Molecular Tests	RGR (G PROTEIN-COUPLED RECEPTOR, RETINAL; RPE-RETINAL G PROTEIN-COUPLED RECEPTOR)	600342	RETINITIS PIGMENTOSA, TYPE 44, RP44	613359		Sequencing	530
Molecular Tests	RHO (RHODOPSIN, OPSIN 2, OPN2)	180380	RETINITIS PIGMENTOSA, TYPE 4, RP4 x NIGHT BLINDNESS, CONGENITAL STATIONARY	180380		Sequencing	480
Molecular Tests	RIMS1 (PROTEIN REGULATING SYNAPTIC MEMBRANE EXOCYTOSIS 1; BIM- RAB3A-INTERACTING MOLECULE 1)	606629	CONE-ROD DYSTROPHY, TYPE 7, CORD7	603649		Sequencing	860
Molecular Tests	RIN2 (RAS AND RAB INTERACTOR 2; RAB5-INTERACTING PROTEIN 2; RAS INHIBITOR IC265)	610222	MACROCEPHALY, ALOPECIA, CUTIS LAXA, AND SCOLIOSIS x MACS SYNDROME	613075		Sequencing	600
Molecular Tests	RLBP1 (RETINALDEHYDE-BINDING PROTEIN 1; CELLULAR RETINALDEHYDE-BINDING PROTEIN; CRALBP)	180090	FUNDUS ALBIPUNCTATUS x RETINITIS PUNCTATA ALBESCENS	136880		Sequencing	550

Molecular Tests	RLBP1 (RETINALDEHYDE-BINDING PROTEIN 1; CELLULAR RETINALDEHYDE-BINDING PROTEIN; CRALBP)	180090	BOTHNIA RETINAL DYSTROPHY » VASTERBOTTEN DYSTROPHY	607475		Sequencing	530
Molecular Tests	RLBP1 (RETINALDEHYDE-BINDING PROTEIN 1; CELLULAR RETINALDEHYDE-BINDING PROTEIN; CRALBP)	180090	NEWFOUNDLAND ROD-CONE DYSTROPHY	607476		Sequencing	530
Molecular Tests	RMRP (MITOCHONDRIAL RNA-PROCESSING ENDORIBONUCLEASE, RNA COMPONENT OF)	157660	CARTILAGE-HAIR HYPOPLASIA » METAPHYSEAL CHONDRODYSPLASIA, MCKUSICK TYPE	250250		Sequencing	450
Molecular Tests	RMRP (MITOCHONDRIAL RNA-PROCESSING ENDORIBONUCLEASE, RNA COMPONENT OF)	157660	METAPHYSEAL DYSPLASIA WITHOUT HYPOTRICHOSIS » CARTILAGE-HAIR HYPOPLASIA-LIKE SKELETAL DYSPLASIA WITHOUT HYPOTRICHOSIS OR IMMUNODEFICIENCY » CARTILAGE-HAIR HYPOPLASIA VARIANT	250460		Sequencing	450
Molecular Tests	RNASEH2A (RIBONUCLEASE H2, LARGE SUBUNIT)	606034	AICARDI-GOUTIERES SYNDROME 4	610333		Sequencing	680
Molecular Tests	RNASEH2B (RIBONUCLEASE H2, SUBUNIT B)	610326	AICARDI-GOUTIERES SYNDROME 2	610181		Sequencing	730
Molecular Tests	RNASEH2C (RIBONUCLEASE H2, SUBUNIT C)	610330	AICARDI-GOUTIERES SYNDROME 3	610329		Sequencing	550
Molecular Tests	RNU4ATAC (RNA, U4ATAC SMALL NUCLEAR; RNA, U4, SMALL NUCLEAR, AT-AC FORM; U4ATAC)	601428	MICROCEPHALIC OSTEODYSPLASTIC PRIMORDIAL DWARFISM, TYPE 1, MOPD1 » TAYBI-LINDER SYNDROME » BRACHYMELIC PRIMORDIAL DWARFISM » CEPHALOSKELETAL DYSPLASIA	210710		Sequencing	290
Molecular Tests	ROBO3 (ROUNDABOUT, DROSOPHILA, HOMOLOG OF, 3; RB-INHIBITING GENE 1; RBIG1; RIG1)	608630	GAZE PALSY, FAMILIAL HORIZONTAL, WITH PROGRESSIVE SCOLIOSIS » OPHTHALMOPLEGIA, PROGRESSIVE EXTERNAL, AND SCOLIOSIS	607313		Sequencing	1275
Molecular Tests	ROR2 (NTRKR2)	602337	BRACHYDACTYLY, TYPE B, BDB	113000		Exons 8 and 9	530
Molecular Tests	ROR2 (NTRKR2)	602337	ROBINOW SYNDROME (AUTOSOMAL RECESSIVE) » COVESDEM SYNDROME	268310		Sequencing	700
Molecular Tests	RP1 (RP1 GENE; OXYGEN-REGULATED PHOTORECEPTOR PROTEIN 1; ORP1)	603937	RETINITIS PIGMENTOSA, TYPE 1, RP1	180100		Sequencing	870
Molecular Tests	RP2	312600	RETINITIS PIGMENTOSA, TYPE 2, RP2, (X-LINKED)	312600		Sequencing	480
Molecular Tests	RP9 (RP9 GENE; PIM1-ASSOCIATED PROTEIN, MOUSE, HOMOLOG OF; PAP1)	607331	RETINITIS PIGMENTOSA, TYPE 9, RP9	180104		Sequencing	480
Molecular Tests	RPE65 (RETINAL PIGMENT EPITHELIUM-SPECIFIC PROTEIN, 65-KD)	180069	LEBER CONGENITAL AMAUROSIS, TYPE 2, LCA2	204100		Sequencing	680
Molecular Tests	RPE65 (RETINAL PIGMENT EPITHELIUM-SPECIFIC PROTEIN, 65-KD)	180069	RETINITIS PIGMENTOSA, TYPE 20, RP20	613794		Sequencing	680
Molecular Tests	RPGR (RETINITIS PIGMENTOSA GTPase REGULATOR)	312610	RETINITIS PIGMENTOSA, TYPE 15, RP15 » CONE-ROD DEGENERATION (X-LINKED)	300029		Exons 1-15 and ORF15	950
Molecular Tests	RPGR (RETINITIS PIGMENTOSA GTPase REGULATOR)	312610	RETINITIS PIGMENTOSA, TYPE 3, RP3 » CHOROIDORETINAL DEGENERATION WITH RETINAL REFLEX IN HETEROZYGOUS WOMEN	300389		Exons 1-15 and ORF15	950
Molecular Tests	RPGR (RETINITIS PIGMENTOSA GTPase REGULATOR)	312610	CONE-ROD DYSTROPHY, (X-LINKED) TYPE 1, CORDX1 » ACHROMATOPSIA, INCOMPLETE (X-LINKED)	304020		Exons 1-15 and ORF15	950
Molecular Tests	RPGRIP1 (RETINITIS PIGMENTOSA GTPase REGULATOR-INTERACTING PROTEIN, RPGR-INTERACTING PROTEIN)	605446	LEBER CONGENITAL AMAUROSIS, TYPE 1, LCA1	204000		Sequencing	870
Molecular Tests	RPGRIP1 (RETINITIS PIGMENTOSA GTPase REGULATOR-INTERACTING PROTEIN, RPGR-INTERACTING PROTEIN)	605446	CONE-ROD DYSTROPHY, TYPE 13, CORD13	608194		Sequencing	870
Molecular Tests	RPGRIP1L (RPGRIP1-LIKE)	610937	JOUBERT SYNDROME, TYPE 7	611560		Sequencing	2600
Molecular Tests	RPGRIP1L (RPGRIP1-LIKE)	610937	MECKEL SYNDROME, TYPE 5	611561		Sequencing	2600
Molecular Tests	RPL5 (RIBOSOMAL PROTEIN L5)	603634	DIAMOND-BLACKFAN ANEMIA, TYPE 6, DBA6	612561		Sequencing	1210
Molecular Tests	RPS19 (RIBOSOMAL PROTEIN S19)	603474	DIAMOND-BLACKFAN ANEMIA, TYPE 1, DBA1	105650		Sequencing	470
Molecular Tests	RPS19 (RIBOSOMAL PROTEIN S19)	603474	DIAMOND-BLACKFAN ANEMIA, TYPE 7, DBA7	612562		Sequencing	940
Molecular Tests	RPS6KA3 (RSK2)	300075	MENTAL RETARDATION, NONSPECIFIC (X-LINKED), TYPE19, MRX19	300075		Sequencing	1100
Molecular Tests	RPS6KA3 (RSK2)	300075	COFFIN-LOWRY SYNDROME, CLS	303600		Sequencing	1100
Molecular Tests	RRM2B (RIBONUCLEOTIDE REDUCTASE, M2 B; RIBONUCLEOTIDE REDUCTASE SMALL SUBUNIT 2-LIKE, p53-INDUCIBLE; P53R2)	604712	MITOCHONDRIAL DNA DEPLETION SYNDROME, ENCEPHALOMYOPATHIC FORM, WITH RENAL TUBULOPATHY	612075		Sequencing	700
Molecular Tests	RUNX2 (CBFA 1)	600211	CLEIDOCRANIAL DYSPLASIA, CCD	119600		Sequencing	900
Molecular Tests	RYR1 (RYANODINE RECEPTOR 1)	180901	MALIGNANT HYPERTHERMIA SUSCEPTIBILITY 1, MHS1 » KING SYNDROME	145600			1700
Molecular Tests	RYR2 (RYANODINE RECEPTOR 2)	180902	ARRHYTHMOGENIC RIGHT VENTRICULAR DYSPLASIA, FAMILIAL, TYPE 2, ARVD2 » ARRHYTHMOGENIC RIGHT VENTRICULAR CARDIOMYOPATHY, TYPE 2, ARVC2	600996		29 Hot Spot Exons (Exons 3, 8, 14, 15, 37, 42, 44, 45, 46, 47, 49, 50, 83, 87, 88, 89, 90, 91, 93, 94, 95, 97, 99, 100, 101, 102, 103, 104 and 105)	800
Molecular Tests	RYR2 (RYANODINE RECEPTOR 2)	180902	VENTRICULAR TACHYCARDIA, CATECHOLAMINERGIC POLYMORPHIC » VENTRICULAR TACHYCARDIA, STRESS-INDUCED POLYMORPHIC	604772		29 Hot Spot Exons (Exons 3, 8, 14, 15, 37, 42, 44, 45, 46, 47, 49, 50, 83, 87, 88, 89, 90, 91, 93, 94, 95, 97, 99, 100, 101, 102, 103, 104 and 105)	800
Molecular Tests	SACS (SACSIN)	604490	SPASTIC ATAXIA, CHARLEVOIX-SAGUENAY TYPE, SACS, ARSACS	270550		Sequencing	2500
Molecular Tests	SACS (SACSIN)	604490	SPASTIC ATAXIA, CHARLEVOIX-SAGUENAY TYPE, SACS, ARSACS	270550		2 French Canadian Mutations: 6594delT and 5254 C>T	350
Molecular Tests	SAG (S-ANTIGEN; ARRESTIN)	181031	OGUCHI DISEASE, TYPE 1 » NIGHT BLINDNESS, CONGENITAL STATIONARY, OGUCHI TYPE 1	258100		Sequencing	680

Molecular Tests	SAG (S-ANTIGEN; ARRESTIN)	181031	RETINITIS PIGMENTOSA, TYPE 47, RP47	613758		Sequencing	680
Molecular Tests	SALL1 (SAL-LIKE 1)	602218	TOWNES-BROCKS SYNDROME » RENAL-EAR-ANAL-RADIAL SYNDROME » REAR SYNDROME » ANUS, IMPERFORATE, WITH HAND, FOOT, AND EAR ANOMALIES » DEAFNESS, SENSORINEURAL, WITH IMPERFORATE ANUS AND THUMB ANOMALIES	107480		Sequencing	1100
Molecular Tests	SALL1 (SAL-LIKE 1)	602218	TOWNES-BROCKS SYNDROME » RENAL-EAR-ANAL-RADIAL SYNDROME » REAR SYNDROME » ANUS, IMPERFORATE, WITH HAND, FOOT, AND EAR ANOMALIES » DEAFNESS, SENSORINEURAL, WITH IMPERFORATE ANUS AND THUMB ANOMALIES	107480		Deletions	550
Molecular Tests	SALL4 (SAL-LIKE 4)	607343	DUANE-RADIAL RAY SYNDROME » OKIHIRO SYNDROME » DUANE ANOMALY WITH RADIAL RAY ABNORMALITIES AND DEAFNESS » DR SYNDROME	607323		Sequencing	670
Molecular Tests	SALL4 (SAL-LIKE 4)	607343	DUANE-RADIAL RAY SYNDROME » OKIHIRO SYNDROME » DUANE ANOMALY WITH RADIAL RAY ABNORMALITIES AND DEAFNESS » DR SYNDROME	607323		Deletions	550
Molecular Tests	SAMHD1 (SAM DOMAIN- AND HD DOMAIN-CONTAINING PROTEIN 1; DENDRITIC CELL-DERIVED IFNG-INDUCED PROTEIN; DCIP)	606754	AICARDI-GOUTIERES SYNDROME, TYPE 5, AGS5	612952		Sequencing	850
Molecular Tests	SAMHD1 (SAM DOMAIN- AND HD DOMAIN-CONTAINING PROTEIN 1; DENDRITIC CELL-DERIVED IFNG-INDUCED PROTEIN; DCIP)	606754	CHILBLAIN LUPUS, TYPE 2	614415		Sequencing	850
Molecular Tests	SBDS	607444	SHWACHMAN-DIAMOND SYNDROME, SDS » PANCREATIC INSUFFICIENCY AND BONE MARROW DYSFUNCTION » CONGENITAL HYPOPLASTIC PANCREAS	260400		Sequencing	850
Molecular Tests	SBF2 (SET-BINDING FACTOR 2; MYOTUBULARIN-RELATED 13; MTMR13)	607697	CHARCOT-MARIE-TOOTH DISEASE, TYPE 4B2, CMT4B2 » CHARCOT-MARIE-TOOTH DISEASE, TYPE 4B2, WITH EARLY-ONSET GLAUCOMA » CHARCOT-MARIE-TOOTH DISEASE, TYPE 4B2, WITH LOCALLY EXTENDED MYELIN SHEATHS, TYPE 4B2 (AUTOSOMAL RECESSIVE)	604563		Sequencing	2600
Molecular Tests	SBF2 (SET-BINDING FACTOR 2; MYOTUBULARIN-RELATED 13; MTMR13)	607697	CHARCOT-MARIE-TOOTH DISEASE, TYPE 4B2, CMT4B2 » CHARCOT-MARIE-TOOTH DISEASE, TYPE 4B2, WITH EARLY-ONSET GLAUCOMA » CHARCOT-MARIE-TOOTH DISEASE, TYPE 4B2, WITH LOCALLY EXTENDED MYELIN SHEATHS, TYPE 4B2 (AUTOSOMAL RECESSIVE)	604563		Exons 14, 23, 27, 32	450
Molecular Tests	SCA1, SCA2, SCA3, SCA6		SPINOCEREBELLAR ATAXIA » AUTOSOMAL DOMINANT CEREBELLAR ATAXIA, ADCA			Screening for Repeats in 4 SCA Genes	500
Molecular Tests	SCA8, SCA10, SCA12, SCA17		SPINOCEREBELLAR ATAXIA » AUTOSOMAL DOMINANT CEREBELLAR ATAXIA, ADCA			Screening for Repeats in 4 SCA Genes	440
Molecular Tests	SCN1A (SODIUM CHANNEL, NEURONAL TYPE 1, ALPHA SUBUNIT)	182389	INTRACTABLE CHILDHOOD EPILEPSY WITH GENERALISED TONIC-CLONIC SEIZURES (ICEGTC)			Sequencing and Deletion-Duplication Testing	1100
Molecular Tests	SCN1A (SODIUM CHANNEL, NEURONAL TYPE 1, ALPHA SUBUNIT)	182389	BORDERLINE SMEI (SMEIB)			Sequencing and Deletion-Duplication Testing	1100
Molecular Tests	SCN1A (SODIUM CHANNEL, NEURONAL TYPE 1, ALPHA SUBUNIT)	182389	GENERALIZED EPILEPSY WITH FEBRILE SEIZURES PLUS, GEFS+ » GENERALIZED EPILEPSY WITH FEBRILE SEIZURES PLUS, TYPE 2, GEFS+, TYPE 2 » FEBRILE SEIZURES, ASSOCIATED WITH SEVERE EPILEPSIES	604233		Sequencing and Deletion-Duplication Testing	1100
Molecular Tests	SCN1A (SODIUM CHANNEL, NEURONAL TYPE 1, ALPHA SUBUNIT)	182389	SEVERE MYOCLONIC EPILEPSY OF INFANCY » DRAVET SYNDROME	607208		Sequencing and Deletion-Duplication Testing	1100
Molecular Tests	SCN1B (SODIUM CHANNEL, VOLTAGE-GATED, TYPE 1, BETA SUBUNIT)	600235	GENERALIZED EPILEPSY WITH FEBRILE SEIZURES PLUS, GEFS+ » GENERALIZED EPILEPSY WITH FEBRILE SEIZURES PLUS, TYPE 2, GEFS+, TYPE 2 » FEBRILE SEIZURES, ASSOCIATED WITH SEVERE EPILEPSIES	604233		Sequencing	600
Molecular Tests	SCN2A (SODIUM CHANNEL, VOLTAGE-GATED, TYPE 2, ALPHA SUBUNIT)	182390	GENERALIZED EPILEPSY WITH FEBRILE SEIZURES PLUS, GEFS+ » GENERALIZED EPILEPSY WITH FEBRILE SEIZURES PLUS, TYPE 2, GEFS+, TYPE 2 » FEBRILE SEIZURES, ASSOCIATED WITH SEVERE EPILEPSIES	604233		Sequencing	2200
Molecular Tests	SCN2A (SODIUM CHANNEL, VOLTAGE-GATED, TYPE 2, ALPHA SUBUNIT)	182390	SEIZURES, BENIGN FAMILIAL NEONATAL-INFANTILE » EPILEPSY, BENIGN NEONATAL, INFANTILE	607745		Sequencing	2200

Molecular Tests	SCN4A and CACNA1S (CACNL1A3)	603967 and 114208	HYPOKALEMIC PERIODIC PARALYSIS, HOKPP » HYPERKALEMIC PERIODIC PARALYSIS	170400		Sequencing	900
Molecular Tests	SCN5A	600163	HEART BLOCK, FAMILIAL	113900		Sequencing	2800
Molecular Tests	SCN5A	600163	SUDDEN INFANT DEATH SYNDROME, SIDS	272120		Sequencing	2800
Molecular Tests	SCN5A	600163	BRUGADA SYNDROME	601144		Sequencing	2800
Molecular Tests	SCN5A	600163	IDIOPATHIC VENTRICULAR FIBRILLATION	603829		Sequencing	2800
Molecular Tests	SCN5A	600163	LONG QT SYNDROME 3, LQT3 » ROMANO-WARD SYNDROME	603830		Sequencing	2800
Molecular Tests	SCN9A (SODIUM CHANNEL, VOLTAGE-GATED, TYPE IX, ALPHA SUBUNIT)	603415	ERYTHERMALGIA, PRIMARY	133020		Sequencing	900
Molecular Tests	SCN9A (SODIUM CHANNEL, VOLTAGE-GATED, TYPE IX, ALPHA SUBUNIT)	603415	PAROXYSMAL EXTREME PAIN DISORDER » PAIN, SUBMANDIBULAR, OCULAR, AND RECTAL, WITH FLUSHING	167400		Sequencing	900
Molecular Tests	SCN9A (SODIUM CHANNEL, VOLTAGE-GATED, TYPE IX, ALPHA SUBUNIT)	603415	INDIFFERENCE TO PAIN, CONGENITAL (AUTOSOMAL RECESSIVE) » INSENSITIVITY TO PAIN, CHANNELOPATHY-ASSOCIATED » CONGENITAL ANALGESIA (AUTOSOMAL RECESSIVE)	243000		Sequencing	900
Molecular Tests	SCNN1A (SODIUM CHANNEL, NONVOLTAGE-GATED 1, ALPHA SUBUNIT, SCNN1, SCNEA)	600228	PSEUDOHYPOALDOSTERONISM, TYPE 1 (AUTOSOMAL RECESSIVE)	264350		Sequencing	750
Molecular Tests	SCNN1B	600760	LIDDLE SYNDROME » PSEUDOALDOSTERONISM	177200		Sequencing	750
Molecular Tests	SCNN1B	600760	PSEUDOHYPOALDOSTERONISM, TYPE 1 (AUTOSOMAL RECESSIVE)	264350		Sequencing	750
Molecular Tests	SCNN1G (SODIUM CHANNEL, NONVOLTAGE-GATED 1, GAMMA SUBUNIT, SCNEG)	600761	LIDDLE SYNDROME » PSEUDOALDOSTERONISM	177200		Sequencing	800
Molecular Tests	SCNN1G (SODIUM CHANNEL, NONVOLTAGE-GATED 1, GAMMA SUBUNIT, SCNEG)	600761	PSEUDOHYPOALDOSTERONISM, TYPE 1 (AUTOSOMAL RECESSIVE)	264350		Sequencing	800
Molecular Tests	SCO1 (S. CEREVISIAE, HOMOLOG OF, CYTOCHROME OXIDASE-DEFICIENT 1, S. CEREVISIAE, HOMOLOG OF)	603644	HEPATIC FAILURE, EARLY-ONSET, AND NEUROLOGIC DISORDER DUE TO CYTOCHROME c OXIDASE DEFICIENCY	220110		Sequencing	760
Molecular Tests	SCO2	604272	CYTOCHROME c OXIDASE DEFICIENCY » COX DEFICIENCY » COMPLEX 4, MITOCHONDRIAL RESPIRATORY CHAIN, DEFICIENCY OF	220110		Sequencing	290
Molecular Tests	SCO2	604272	LEIGH SYNDROME	256000		Sequencing	290
Molecular Tests	SCO2	604272	CARDIOENCEPHALOMYOPATHY, FATAL INFANTILE, DUE TO CYTOCHROME c OXIDASE DEFICIENCY » CYTOCHROME c OXIDASE DEFICIENCY, FATAL INFANTILE, WITH CARDIOENCEPHALOMYOPATHY	604377		Sequencing	290
Molecular Tests	SDHA (SUCCINATE DEHYDROGENASE COMPLEX, SUBUNIT A, FLAVOPROTEIN, SUCCINATE DEHYDROGENASE 2, FLAVOPROTEIN SUBUNIT; SDH2)	600857	CYTOCHROME c OXIDASE DEFICIENCY » COX DEFICIENCY » COMPLEX 4, MITOCHONDRIAL RESPIRATORY CHAIN, DEFICIENCY OF	220110		Sequencing	1340
Molecular Tests	SDHA (SUCCINATE DEHYDROGENASE COMPLEX, SUBUNIT A, FLAVOPROTEIN, SUCCINATE DEHYDROGENASE 2, FLAVOPROTEIN SUBUNIT; SDH2)	600857	LEIGH SYNDROME	256000		Sequencing	1340
Molecular Tests	SDHAF1 (SUCCINATE DEHYDROGENASE COMPLEX ASSEMBLY FACTOR 1)	612848	CYTOCHROME c OXIDASE DEFICIENCY » COX DEFICIENCY » COMPLEX 2, MITOCHONDRIAL RESPIRATORY CHAIN, DEFICIENCY OF	252011		Sequencing	500
Molecular Tests	SDHAF2 (SUCCINATE DEHYDROGENASE COMPLEX ASSEMBLY FACTOR 2; SUCCINATE DEHYDROGENASE 5; SDH5)	613019	PARAGANGLIOMAS, PGL2 » GLOMUS TUMORS, TYPE 2	601650		Sequencing	500
Molecular Tests	SDHB (SUCCINATE DEHYDROGENASE 1, SDH1)	185470	PARAGANGLIOMAS, PGL4 » GLOMUS TUMORS, TYPE 4	115310		Sequencing	750
Molecular Tests	SDHB, SDHC, SDHD, SDHAF2		PARAGANGLIOMAS, PGL » GLOMUS TUMORS			Deletion-Duplication Testing of 4 Genes	770
Molecular Tests	SDHB, SDHC, SDHD, SDHAF2, TMEM127		PARAGANGLIOMAS, PGL » GLOMUS TUMORS			Whole Gene Sequencing and Deletion-Duplication Testing of 5 Genes	4120
Molecular Tests	SDHC (SUCCINATE DEHYDROGENASE 3, SDH3)	602413	PARAGANGLIOMAS, PGL 3 » GLOMUS TUMORS TYPE 3	605373		Sequencing	740
Molecular Tests	SDHD (SUCCINATE DEHYDROGENASE 4, SDH4)	602690	PARAGANGLIOMAS, PGL 1 » GLOMUS TUMORS TYPE 1	168000		Sequencing	470
Molecular Tests	SEC63 (SEC63, S. CEREVISIAE, HOMOLOG OF)	608648	POLYCYSTIC LIVER DISEASE	174050		Sequencing	1700
Molecular Tests	SEDL (SEDLIN)	300202	SPONDYLOEPIPHYSEAL DYSPLASIA TARDA (X-LINKED), SEDT	313400		Sequencing	750
Molecular Tests	SEMA4A (SEMAPHORIN 4A; SEMA4A; SEMAPHORIN B; SEMAB; SEMB)	607292	RETINITIS PIGMENTOSA, TYPE 35, RP35	610282		Sequencing	680
Molecular Tests	SEMA4A (SEMAPHORIN 4A; SEMA4A; SEMAPHORIN B; SEMAB; SEMB)	607292	CONE-ROD DYSTROPHY, TYPE 10, CORD10	610283		Sequencing	680
Molecular Tests	SEPN1 (SELENOPROTEIN N, 1; SELN)	606210	RIGID SPINE MUSCULAR DYSTROPHY, TYPE 1 » MULTICORE MYOPATHY, SEVERE CLASSIC FORM » MINICORE MYOPATHY, SEVERE CLASSIC FORM » DESMIN-RELATED MYOPATHY WITH MALLORY BODIES	602771			1240

Molecular Tests	SEPT9 (SEPTIN 9)	604061	AMYOTROPHY, HEREDITARY NEURALGIC » BRACHIAL PLEXUS NEUROPATHY, HEREDITARY	162100		Sequencing and Deletion-Duplication Testing	1100
Molecular Tests	SEPT9 (SEPTIN 9)	604061	AMYOTROPHY, HEREDITARY NEURALGIC » BRACHIAL PLEXUS NEUROPATHY, HEREDITARY	162100		Sequencing Exons 1 and 2	450
Molecular Tests	SEPT9 (SEPTIN 9)	604061	AMYOTROPHY, HEREDITARY NEURALGIC » BRACHIAL PLEXUS NEUROPATHY, HEREDITARY	162100		Arg88Trp and Ser93Phe	300
Molecular Tests	SEPT9 (SEPTIN 9)	604061	SCHILBACH-ROTT SYNDROME » OCULAR HYPOTELORISM, SUBMUCOSAL CLEFT PALATE, AND HYPOSPADIAS » BLEPHAROFACIOSKELETAL SYNDROME	164220		Sequencing and Deletion-Duplication Testing	1100
Molecular Tests	SEPT9 (SEPTIN 9)	604061	SCHILBACH-ROTT SYNDROME » OCULAR HYPOTELORISM, SUBMUCOSAL CLEFT PALATE, AND HYPOSPADIAS » BLEPHAROFACIOSKELETAL SYNDROME	164220		Sequencing Exons 1 and 2	450
Molecular Tests	SEPT9 (SEPTIN 9)	604061	SCHILBACH-ROTT SYNDROME » OCULAR HYPOTELORISM, SUBMUCOSAL CLEFT PALATE, AND HYPOSPADIAS » BLEPHAROFACIOSKELETAL SYNDROME	164220		Arg88Trp and Ser93Phe	300
Molecular Tests	SERPINA1 (SERPIN PEPTIDASE INHIBITOR, CLADE A, MEMBER 1, PROTEASE INHIBITOR 1, PI1, ALPHA-1-ANTITRYPSIN	107400	ANTITRYPSINE DEFICIENCY, AAT	107400		Alleles M, S and Z	250
Molecular Tests	SERPINH1 (SERPIN PEPTIDASE INHIBITOR, CLADE H, MEMBER 1; COLLAGEN-BINDING PROTEIN 2; COLLIGIN 2; SERPINH2; HEAT-SHOCK PROTEIN 47; HSP47; RHEUMATOID ARTHRITIS ANTIGEN-A47; RA-A47)	604539	PRETERM PREMATURE RUPTURE OF THE MEMBRANES, PPROM	610504		Sequencing	550
Molecular Tests	SERPINH1 (SERPIN PEPTIDASE INHIBITOR, CLADE H, MEMBER 1; COLLAGEN-BINDING PROTEIN 2; COLLIGIN 2; SERPINH2; HEAT-SHOCK PROTEIN 47; HSP47; RHEUMATOID ARTHRITIS ANTIGEN-A47; RA-A47)	604539	OSTEOGENESIS IMPERFECTA TYPE 10, OI10	613848		Sequencing	550
Molecular Tests	SETX (SENATAXIN)	608465	AMYOTROPHIC LATERAL SCLEROSIS 4, JUVENILE, ALS4 » NEURONOPATHY, DISTAL HEREDITARY MOTOR, WITH PYRAMIDAL FEATURES	602433		Sequencing	1370
Molecular Tests	SETX (SENATAXIN)	608465	SPINOCEREBELLAR ATAXIA (AUTOSOMAL RECESSIVE), 1 » ATAXIA-OCULOMOTOR APRAXIA 2	606002		Sequencing	1370
Molecular Tests	SFTPB (SURFACTANT PULMONARY-ASSOCIATED PROTEIN B)	178640	SURFACTANT METABOLISM DYSFUNCTION, PULMONARY, TYPE 1, SMDP1 » PULMONARY ALVEOLAR PROTEINOSIS DUE TO SURFACTANT PROTEIN B DEFICIENCY	265120		Sequencing	1150
Molecular Tests	SFTPC (SURFACTANT PULMONARY-ASSOCIATED PROTEIN C)	178620	SURFACTANT METABOLISM DYSFUNCTION, PULMONARY, TYPE 2, SMDP2 » PULMONARY ALVEOLAR PROTEINOSIS DUE TO SURFACTANT PROTEIN C DEFICIENCY	610913		Sequencing	860
Molecular Tests	SGCA (SARCOGLYCAN, ALPHA, ADHALIN, DYSTROGLYCAN 2)	600119	MUSCULAR DYSTROPHY, LIMB-GIRDLE, TYPE 2D, LGMD2D » DUCHENNE - LIKE MUSCULAR DYSTROPHY, TYPE 2, AUTOSOMAL RECESSIVE, DMDA2 » ADHALINOPATHY, PRIMARY » MUSCULAR DYSTROPHY, DUCHENNE - LIKE MUSCULAR DYSTROPHY, TYPE 2	608099		Sequencing	600
Molecular Tests	SGCA, SGCB, SGCD, SGCG		MUSCULAR DYSTROPHY, LIMB-GIRDLE			Deletion-Duplication Testing	350
Molecular Tests	SGCB (SARCOGLYCAN, BETA)	600900	MUSCULAR DYSTROPHY, LIMB-GIRDLE, TYPE 2E, LGMD2E	604286		Sequencing	450
Molecular Tests	SGCD (SARCOGLYCAN DELTA)	601411	MUSCULAR DYSTROPHY, LIMB-GIRDLE, TYPE 2F, LGMD2F	601287		Sequencing	550
Molecular Tests	SGCD (SARCOGLYCAN DELTA)	601411	DILATED CARDIOMYOPATHY, 1L, CMD1L	606685		Sequencing	550
Molecular Tests	SGCE (SARCOGLYCAN, EPSILON)	604149	MYOCLONIC DYSTONIA » MYOCLONUS-DYSTONIA SYNDROME » DYSTONIA, ALCOHOL-RESPONSIVE » DYSTONIA 11, DYT11	159900		Sequencing	590
Molecular Tests	SGCG (SARCOGLYCAN GAMMA, ADHALIN)	608896	DUCHENNE-LIKE MUSCULAR DYSTROPHY, TYPE 1 » LIMB GIRDLE MUSCULAR DYSTROPHY 2C, LGMD2C » MUSCULAR DYSTROPHY, DUCHENNE-LIKE MUSCULAR DYSTROPHY, TYPE 1	253700		Sequencing	690
Molecular Tests	SGCZ (SARCOGLYCAN, ZETA; ZSG1)	608113	UNKNOWN DISEASE			Sequencing	500
Molecular Tests	SGSH (HEPARAN SULFATE SULFATASE, SULFAMIDASE, N-SULFOGLUCOSAMINE SULFOHYDROLASE)	605270	» MUCOPOLYSACCHARIDOSIS TYPE 3A, MPS3A SANFILIPPO SYNDROME A	252900		Sequencing	500
Molecular Tests	SH2D1A	308240	LYMPHOPROLIFERATIVE SYNDROME » LYMPHOPROLIFERATIVE DISEASE (X-LINKED)	308240		Sequencing	420
Molecular Tests	SH3BP2	602104	CHERUBISM	118400		Exon 9	300
Molecular Tests	SH3TC2 (SH3 DOMAIN AND TETRATRICOPEPTIDE REPEAT DOMAIN 2)	68206	CHARCOT-MARIE-TOOTH DISEASE, DEMYELINATING, TYPE 4C, CMT4C (AUTOSOMAL RECESSIVE)	601596		Sequencing	1700
Molecular Tests	SHH (SONIC HEDGEHOG)	600725	HOLOPROSENCEPHALY 3, HPE3	142945		Sequencing	680
Molecular Tests	SHH (SONIC HEDGEHOG)	600725	SOLITARY MEDIAN MAXILLARY CENTRAL INCISOR	147250		Sequencing	680
Molecular Tests	SHH, SIX3, TGIF and ZIC2		HOLOPROSENCEPHALY			Sequencing and Deletion-Duplication Testing	2350
Molecular Tests	SHOC2 (SUPPRESSOR OF CLEAR, C. ELEGANS, HOMOLOG OF; RAS-BINDING PROTEIN SUR8, C. ELEGANS, HOMOLOG OF; SUR8)	602775	NOONAN SYNDROME-LIKE DISORDER WITH LOOSE ANAGEN HAIR	607721		Sequencing	290

Molecular Tests	SHOX (SHORT STATURE HOMEOBOX)	312865	LERI-WEILL DYSCHONDROSTEOSIS » DYSCHONDROSTEOSIS » MADELUNG DEFORMITY	127300		Sequencing	500
Molecular Tests	SHOX (SHORT STATURE HOMEOBOX)	312865	LERI-WEILL DYSCHONDROSTEOSIS » DYSCHONDROSTEOSIS » MADELUNG DEFORMITY	127300		MLPA for Deletions-Duplications of SHOX and PAR1	500
Molecular Tests	SHOX (SHORT STATURE HOMEOBOX)	312865	LANGER MESOMELIC DYSPLASIA » DYSCHONDROSTEOSIS, HOMOZYGOUS » MESOMELIC DWARFISM OF THE HYPOPLASTIC ULNA, FIBULA, AND MANDIBLE TYPE	249700		Sequencing	500
Molecular Tests	SHOX (SHORT STATURE HOMEOBOX)	312865	LANGER MESOMELIC DYSPLASIA » DYSCHONDROSTEOSIS, HOMOZYGOUS » MESOMELIC DWARFISM OF THE HYPOPLASTIC ULNA, FIBULA, AND MANDIBLE TYPE	249700		MLPA for Deletions-Duplications of SHOX and PAR1	500
Molecular Tests	SHOX (SHORT STATURE HOMEOBOX)	312865	SHORT STATURE	604271		Sequencing	500
Molecular Tests	SHOX (SHORT STATURE HOMEOBOX)	312865	SHORT STATURE	604271		MLPA for Deletions-Duplications of SHOX and PAR1	500
Molecular Tests	SIL1 (SIL1, S. CEREVISIAE, HOMOLOG OF; BIP-ASSOCIATED PROTEIN, BAP)	608005	MARINESCO-SJOGREN SYNDROME	248800		Sequencing	1040
Molecular Tests	SIX1	601205	DEAFNESS, DFNA23 (AUTOSOMAL DOMINANT)	605192		Sequencing	500
Molecular Tests	SIX1	601205	BRANCHIOOTIC SYNDROME, TYPE 3 » BO SYNDROME, TYPE 3	608389		Sequencing	500
Molecular Tests	SIX3	603714	HOLOPROSENCEPHALY 2, HPE2	157170		Sequencing	650
Molecular Tests	SIX5	600963	BRANCHIO-OTO-RENAL SYNDROME, TYPE 2, BOR2	610896		Sequencing	650
Molecular Tests	SIX6	606326	MICROPHTHALMIA, ISOLATED, WITH CATARACT 2	212550		Deletion-Duplication Testing	680
Molecular Tests	SLC12A1 (SOLUTE CARRIER FAMILY 12 (SODIUM/POTASSIUM/CHLORIDE TRANSPORTER), MEMBER 1, SODIUM-POTASSIUM-CHLORIDE TRANSPORTER 2, NKCC2)	600839	BARTTER SYNDROME, ANTENATAL, TYPE 1 » HYPOKALEMIC ALKALOSIS WITH HYPERCALCIURIA, ANTENATAL, TYPE 1 » HYPERPROSTAGLANDIN E SYNDROME 1	601678		Sequencing	1250
Molecular Tests	SLC12A3 (SOLUTE CARRIER FAMILY 12, SODIUM/CHLORIDE TRANSPORTER, MEMBER 3, THIAZIDE-SENSITIVE NA-CL COTRANSPORTER)	600968	GITELMAN SYNDROME » HYPOMAGNESEMIA-HYPOKALEMIA, PRIMARY RENOTUBULAR, WITH HYPOCALCIURIA » POTASSIUM AND MAGNESIUM DEPLETION	263800		Sequencing	1100
Molecular Tests	SLC12A3 (SOLUTE CARRIER FAMILY 12, SODIUM/CHLORIDE TRANSPORTER, MEMBER 3, THIAZIDE-SENSITIVE NA-CL COTRANSPORTER)	600968	GITELMAN SYNDROME » HYPOMAGNESEMIA-HYPOKALEMIA, PRIMARY RENOTUBULAR, WITH HYPOCALCIURIA » POTASSIUM AND MAGNESIUM DEPLETION	263800		Deletion-Duplication Testing	350
Molecular Tests	SLC12A6	604878	CORPUS CALLOSUM, AGENESIS OF, WITH NEURONOPATHY » CHARLEVOIX DISEASE » ANDERMANN SYNDROME » POLYNEUROPATHY, SENSORIMOTOR, WITH OR WITHOUT AGENESIS OF THE CORPUS CALLOSUM	218000		Sequencing	1400
Molecular Tests	SLC16A2 (SOLUTE CARRIER FAMILY 16, MEMBER 2; MONOCARBOXYLATE TRANSPORTER 8; MCT8)	300095	ALLAN-HERNDON-DUDLEY SYNDROME » MONOCARBOXYLATE TRANSPORTER 8 DEFICIENCY » MENTAL RETARDATION WITH HYPOTONIA (X-LINKED)	300523		Sequencing	910
Molecular Tests	SLC17A5	604322	INFANTILE SIALIC ACID STORAGE DISORDER » SIALURIA, INFANTILE FORM » N-ACETYLNEURAMINIC ACID STORAGE DISEASE » NANA STORAGE DISEASE	269920		Sequencing	580
Molecular Tests	SLC17A5	604322	SIALURIA, FINNISH TYPE » SALLA DISEASE	604369		Sequencing	580
Molecular Tests	SLC19A2 (SOLUTE CARRIER FAMILY 19 (THIAMINE TRANSPORTER), MEMBER 2; THIAMINE TRANSPORTER PROTEIN 1)	603941	THIAMINE-RESPONSIVE MEGALOBLASTIC ANEMIA SYNDROME, TRMA » ROGERS SYNDROME » THIAMINE-RESPONSIVE MYELODYSPLASIA » MEGALOBLASTIC ANEMIA, THIAMINE-RESPONSIVE, WITH DIABETES MELLITUS AND SENSORINEURAL DEAFNESS	249270		Sequencing	500
Molecular Tests	SLC1A3 (SOLUTE CARRIER FAMILY 1 (GLIAL HIGH AFFINITY GLUTAMATE TRANSPORTER), MEMBER 3; EXCITATORY AMINO ACID TRANSPORTER 1; EAAT1; GLIAL HIGH AFFINITY GLUTAMATE TRANSPORTER; GLUTAMATE/ASPARTATE TRANSPORTER, HIGH AFFINITY, SODIUM-DEPENDENT; GLAST1)	600111	EPISODIC ATAXIA, TYPE 6, EA6	612656		Sequencing	650
Molecular Tests	SLC22A12 (SOLUTE CARRIER FAMILY 22, URATE TRANSPORTER, MEMBER 12, ORGANIC ANION TRANSPORTER 4-LIKE, OAT4L, URATE TRANSPORTER 1, URAT1	607096	HYPOURICEMIA, RENAL » DALMATIAN HYPOURICEMIA » RENAL HYPOURICEMIA » URIC ACID UROLITHIASIS	220150		Sequencing	500
Molecular Tests	SLC22A4 (SOLUTE CARRIER FAMILY 22 (ORGANIC CATION TRANSPORTER), MEMBER 4; OCTN1)	604190	RHEUMATOID ARTHRITIS, SUSCEPTIBILITY TO	180300		Sequencing	700

Molecular Tests	SLC22A5 (SOLUTE CARRIER FAMILY 22, ORGANIC CATION TRANSPORTER, MEMBER 5, OCTN2)	603377	CARNITINE DEFICIENCY, SYSTEMIC PRIMARY » SYSTEMIC CARNITINE DEFICIENCY » CARNITINE DEFICIENCY, SYSTEMIC, DUE TO DEFECT IN RENAL REABSORPTION OF CARNITINE » CARNITINE DEFICIENCY, PRIMARY » CARNITINE TRANSPORTER, PLASMA-MEMBRANE, DEFICIENCY OF	212140		Sequencing	700
Molecular Tests	SLC24A1 (SOLUTE CARRIER FAMILY 24 (SODIUM/POTASSIUM/CALCIUM EXCHANGER), MEMBER 1)	603617	NIGHT BLINDNESS, CONGENITAL STATIONARY, TYPE 1D » CSBN, COMPLETE (AUTOSOMAL RECESSIVE)	613830		Sequencing	720
Molecular Tests	SLC25A13 (SOLUTE CARRIER FAMILY 25, MEMBER 13; CITRIN)	603859	CITRULLINEMIA, TYPE 2, ADULT-ONSET	603471		Sequencing	1150
Molecular Tests	SLC25A13 (SOLUTE CARRIER FAMILY 25, MEMBER 13; CITRIN)	603859	CITRULLINEMIA, TYPE 2, NEONATAL-ONSET » CHOLESTASIS, NEONATAL INTRAHEPATIC, CAUSED BY CITRIN DEFICIENCY	605814		Sequencing	1150
Molecular Tests	SLC25A15 (SOLUTE CARRIER FAMILY 25 (MITOCHONDRIAL CARRIER, ORNITHINE TRANSPORTER), MEMBER 15;ORNITHINE TRANSPORTER, MITOCHONDRIAL, 1)	603861	HHH SYNDROME » HYPERORNITHINEMIA-HYPERAMMONEMIA-HOMOCITRULLINURIA SYNDROME	238970		Sequencing	500
Molecular Tests	SLC25A19 (SOLUTE CARRIER FAMILY 25 (MITOCHONDRIAL THIAMINE PYROPHOSPHATE CARRIER), MEMBER 19; MITOCHONDRIAL UNCOUPLING PROTEIN 1; MITOCHONDRIAL DEOXYNUCLEOTIDE CARRIER, FORMERLY)	606521	THIAMINE METABOLISM DYSFUNCTION SYNDROME, TYPE 3, MICROCEPHALY TYPE » MICROCEPHALY, AMISH TYPE	607196		Sequencing	710
Molecular Tests	SLC25A19 (SOLUTE CARRIER FAMILY 25 (MITOCHONDRIAL THIAMINE PYROPHOSPHATE CARRIER), MEMBER 19; MITOCHONDRIAL UNCOUPLING PROTEIN 1; MITOCHONDRIAL DEOXYNUCLEOTIDE CARRIER, FORMERLY)	606521	THIAMINE METABOLISM DYSFUNCTION SYNDROME, TYPE 4, BILATERAL STRIATAL DEGENERATION AND PROGRESSIVE POLYNEUROPATHY TYPE » BILATERAL STRIATAL DEGENERATION AND PROGRESSIVE POLYNEUROPATHY	613710		Sequencing	710
Molecular Tests	SLC25A20 (SOLUTE CARRIER FAMILY 25, MEMBER 20, CARNITINE-ACYLCARNITINE TRANSLOCASE, CARNITINE-ACYLCARNITINE CARRIER)	212138	CARNITINE-ACYLCARNITINE TRANSLOCASE DEFICIENCY » CACT DEFICIENCY	212138		Sequencing	1040
Molecular Tests	SLC25A22 (SOLUTE CARRIER FAMILY 25 (MITOCHONDRIAL CARRIER, GLUTAMATE), MEMBER 22; GLUTAMATE CARRIER 1; GC1)	609302	EPILEPTIC ENCEPHALOPATHY, EARLY INFANTILE, TYPE 3, EIEE3	609304		Sequencing	1060
Molecular Tests	SLC25A4 (SOLUTE CARRIER FAMILY 25 (MITOCHONDRIAL CARRIER), MEMBER 4, ADENINE NUCLEOTIDE TRANSLOCATOR 1, ANT1 ADP/ATP TRANSLOCATOR OF SKELETAL MUSCLE, ADP/ATP TRANSLOCASE 1)	103220	PROGRESSIVE EXTERNAL OPHTHALMOPLEGIA WITH MITOCHONDRIAL DNA DELETIONS, (AUTOSOMAL DOMINANT), TYPE 2 » PROGRESSIVE EXTERNAL OPHTHALMOPLEGIA (AUTOSOMAL DOMINANT), 2	609283		Sequencing	500
Molecular Tests	SLC26A2 (DTD SULFATE TRANSPORTER, DTDST)	606718	DIASTROPHIC DYSPLASIA, DTD	222600		Sequencing	790
Molecular Tests	SLC26A2 (DTD SULFATE TRANSPORTER, DTDST)	606718	EPIPHYSEAL DYSPLASIA, MULTIPLE, TYPE 4, EDM4 » MULTIPLE EPIPHYSEAL DYSPLASIA (AUTOSOMAL RECESSIVE)	226900		Sequencing	790
Molecular Tests	SLC26A2 (DTD SULFATE TRANSPORTER, DTDST)	606718	ATELOSTEOGENESIS TYPE 2 » AO TYPE 2 » DE LA CHAPELLE DYSPLASIA	256050		Sequencing	790
Molecular Tests	SLC26A2 (DTD SULFATE TRANSPORTER, DTDST)	606718	ACHONDROGENESIS TYPE 1B » ACHONDROGENESIS, FRACCARO TYPE	600972		Sequencing	790
Molecular Tests	SLC26A3 (SOLUTE CARRIER FAMILY 26, MEMBER 3)	126650	DIARRHEA 1, SECRETORY CHLORIDE, CONGENITAL » CHLORIDE DIARRHEA, CONGENITAL, FINNISH TYPE	214700		Sequencing	1100
Molecular Tests	SLC26A4 (PENDRIN)	605646	PENDRED SYNDROME, PDS	274600		Sequencing	900
Molecular Tests	SLC26A4 (PENDRIN)	605646	DEAFNESS, DFNB4 » DEAFNESS (AUTOSOMAL RECESSIVE), NEUROSENSORY 4	600791		Sequencing	900
Molecular Tests	SLC26A4 (PENDRIN)	605646	ENLARGED VESTIBULAR AQUEDUCT SYNDROME, EVA	603545		Sequencing	900
Molecular Tests	SLC29A3 (SOLUTE CARRIER FAMILY 29 (NUCLEOSIDE TRANSPORTER), MEMBER 3; EQUILIBRATIVE NUCLEOSIDE TRANSPORTER 3; ENT3)	612373	HISTIOCYTOSIS-LYMPHADENOPATHY PLUS SYNDROME » FAISALABAD HISTIOCYTOSIS » ROSAI-DORFMAN DISEASE, FAMILIAL » HISTIOCYTOSIS WITH JOINT CONTRACTURES AND SENSORINEURAL DEAFNESS » PIGMENTED HYPERTRICHOSIS WITH INSULIN-DEPENDENT DIABETES MELLITUS	602782		Sequencing	2140
Molecular Tests	SLC2A1 (SOLUTE CARRIER FAMILY 2 (FACILITATED GLUCOSE TRANSPORTER), MEMBER 1; GLUT1; ERYTHROCYTE/HEPATOMA GLUCOSE TRANSPORTER)	138140	GLUCOSE TRANSPORT DEFECT, BLOOD-BRAIN BARRIER » GLUT1 DEFICIENCY SYNDROME	606777		Sequencing	1060
Molecular Tests	SLC2A1 (SOLUTE CARRIER FAMILY 2 (FACILITATED GLUCOSE TRANSPORTER), MEMBER 1; GLUT1; ERYTHROCYTE/HEPATOMA GLUCOSE TRANSPORTER)	138140	GLUCOSE TRANSPORT DEFECT, BLOOD-BRAIN BARRIER » GLUT1 DEFICIENCY SYNDROME	606777		Deletion-Duplication Testing	600
Molecular Tests	SLC2A10 (SOLUTE CARRIER FAMILY 2 - FACILITATED GLUCOSE TRANSPORTER, MEMBER 10, GLUCOSE TRANSPORTER 10, GLUT10)	606145	ARTERIAL TORTUOSITY SYNDROME, ATS	208050		Sequencing	550
Molecular Tests	SLC2A2 (SOLUTE CARRIER FAMILY 2 (FACILITATED GLUCOSE TRANSPORTER), MEMBER 2)	138160	FANCONI-BICKEL SYNDROME » HEPATORENAL GLYCOGENOSIS WITH RENAL FANCONI SYNDROME » GLYCOGENOSIS, FANCONI TYPE	227810		Sequencing	650
Molecular Tests	SLC34A3 (SOLUTE CARRIER FAMILY 34 (SODIUM/PHOSPHATE COTRANSPORTER), MEMBER 3; SODIUM/INORGANIC PHOSPHATE COTRANSPORTER, TYPE IIC; NPTIIC)	609826	SODIUM/INORGANIC PHOSPHATE COTRANSPORTER, TYPE 2C, NPT2C » HYPERCALCIURIC RICKETS	241530		Sequencing	745

Molecular Tests	SLC35A1 (SOLUTE CARRIER FAMILY 35 (CMP-SIALIC ACID TRANSPORTER), MEMBER 1; CYTIDINE MONOPHOSPHATE-SIALIC ACID TRANSPORTER)	605634	CONGENITAL DISORDER OF GLYCOSYLATION, TYPE 2F, CDG2F	603585		Sequencing	1040
Molecular Tests	SLC35C1 (SOLUTE CARRIER FAMILY 35, MEMBER C1;GDP-FUCOSE TRANSPORTER 1)	605881	CONGENITAL DISORDER OF GLYCOSYLATION, TYPE 2C, CDG2C » LEUKOCYTE ADHESION DEFICIENCY, TYPE 2C » RAMBAM-HASHARON SYNDROME	266265		Sequencing	530
Molecular Tests	SLC37A4 (GLUCOSE-6-PHOSPHATE TRANSPORTER 1; G6PT1; GLUCOSE-6-PHOSPHATE TRANSLOCASE)	602671	GLYCOGEN STORAGE DISEASE, TYPE 1B » GSD TYPE 1B	232220		Sequencing	620
Molecular Tests	SLC3A1 (RBAT)	104614	CYSTINURIA TYPE 1	220100		Sequencing	650
Molecular Tests	SLC3A1 (RBAT) (analyzed together with SLC7A9)	104614	CYSTINURIA TYPE 1	220100		10 mutations in SLC3A1: Thr216Met, Ser217Arg, Arg365Trp, Arg365Leu, Arg362Cys, Met467Thr, Met467Lys, 1500+1G/T, Glu483X duplication of exons 5-9 includes 7 mutations in SLC7A9: Gly105Arg, Thr123Met, Phe140Ser, Ala182Thr, 747delG,	700
Molecular Tests	SLC40A1 (FERROPORTIN 1, IREG1, SLC11A3)	604653	HEMOCHROMATOSIS, TYPE 4, HFE4	606069		Sequencing	950
Molecular Tests	SLC45A2 (SOLUTE CARRIER FAMILY 45, MEMBER 2, MATP (MEMBRANE-ASSOCIATED TRANSPORTER PROTEIN)	606202	OCULOCUTANEOUS ALBINISM, TYPE 4, OCA4	606574		Sequencing	530
Molecular Tests	SLC4A1 (BAND 3 OF RED CELL MEMBRANE, ERYTHROID PROTEIN BAND 3, ANION EXCHANGE PROTEIN 1)	109270	HEMOLYTIC ANEMIA DUE TO BAND 3 MONTEFIORE SPHEROCYTOSIS, HEREDITARY, DUE TO BAND 3 (TUSCALOOSA, PRAGUE, CHUR, NOIRTERRE, LYON, GENAS, FUKUOKA, TOKYO, COIMBRA, CAPE TOWN, PRAGUE III) OVALOCYTOSIS (SOUTHEAST ASIAN , MALAYSIAN-MELANESIAN-FILIPINO TYPE) ACANTHOCYTOSIS ERYTHROCYTOSIS, STOMATOCYTIC HEREDITARY	109270		Sequencing	950
Molecular Tests	SLC4A1 (BAND 3 OF RED CELL MEMBRANE, ERYTHROID PROTEIN BAND 3, ANION EXCHANGE PROTEIN 1)	109270	DIEGO BLOOD GROUP ANTIGEN	110500		Sequencing	950
Molecular Tests	SLC4A1 (BAND 3 OF RED CELL MEMBRANE, ERYTHROID PROTEIN BAND 3, ANION EXCHANGE PROTEIN 1)	109270	WALDNER BLOOD GROUP ANTIGEN WD(A)	112010		Sequencing	950
Molecular Tests	SLC4A1 (BAND 3 OF RED CELL MEMBRANE, ERYTHROID PROTEIN BAND 3, ANION EXCHANGE PROTEIN 1)	109270	WRIGHT BLOOD GROUP ANTIGEN	112050		Sequencing	950
Molecular Tests	SLC4A1 (BAND 3 OF RED CELL MEMBRANE, ERYTHROID PROTEIN BAND 3, ANION EXCHANGE PROTEIN 1)	109270	RENAL TUBULAR ACIDOSIS, DISTAL (AUTOSOMAL DOMINANT) » RENAL TUBULAR ACIDOSIS 1	179800		Sequencing	950
Molecular Tests	SLC4A1 (BAND 3 OF RED CELL MEMBRANE, ERYTHROID PROTEIN BAND 3, ANION EXCHANGE PROTEIN 1)	109270	RENAL TUBULAR ACIDOSIS, DISTAL (AUTOSOMAL RECESSIVE) » RENAL TUBULAR ACIDOSIS (AUTOSOMAL RECESSIVE) WITH PRESERVED HEARING » RENAL TUBULAR ACIDOSIS, DISTAL, (AUTOSOMAL RECESSIVE) WITH LATE-ONSET SENSORINEURAL HEARING LOSS	602722		Sequencing	950
Molecular Tests	SLC4A4 (SODIUM BICARBONATE COTRANSPORTER 1, NBC1)	603345	RENAL TUBULAR ACIDOSIS, PROXIMAL, WITH OCULAR ABNORMALITIES » RENAL TUBULAR ACIDOSIS 2	604278		Sequencing	1100
Molecular Tests	SLC5A1 (SOLUTE CARRIER FAMILY 5 (SODIUM/GLUCOSE COTRANSPORTER), MEMBER 1;SOLUTE CARRIER FAMILY 5 (SODIUM/GLUCOSE COTRANSPORTER), MEMBER 1	182380	GLUCOSE/GALACTOSE MALABSORPTION » MONOSACCHARIDE MALABSORPTION	606824		Sequencing	745
Molecular Tests	SLC5A2	182381	RENAL GLUCOSURIA	233100		Sequencing	550
Molecular Tests	SLC5A5 (SODIUM-IODIDE SYMPORTER, NIS)	601843	THYROID HORMONOGENESIS	274400		Sequencing	1220
Molecular Tests	SLC6A3 (SOLUTE CARRIER FAMILY 6 (NEUROTRANSMITTER TRANSPORTER, DOPAMINE), MEMBER 3; DOPAMINE TRANSPORTER; DAT1)	126455	TOBACCO ADDICTION, SUSCEPTIBILITY TO » NICOTINE DEPENDENCE, SUSCEPTIBILITY TO	188890		Sequencing	850
Molecular Tests	SLC6A8 (CREATINE TRANSPORTER, CT1)	300036	CREATINE DEFICIENCY SYNDROME (X-LINKED)	300352		Sequencing	1450
Molecular Tests	SLC6A8 (CREATINE TRANSPORTER, CT1)	300036	MENTAL RETARDATION, WITH SEIZURES, SHORT STATURE AND MIDFACE HYPOPLASIA (X-LINKED)	300397		Sequencing	1450
Molecular Tests	SLC7A7 (SOLUTE CARRIER FAMILY 7, MEMBER 7)	603593	LYSINURIC PROTEIN INTOLERANCE » DIBASICAMINO ACIDURIA, TYPE 2	222700		Sequencing	600
Molecular Tests	SLC7A9	604144	CYSTINURIA TYPE 1	220100		Sequencing	650
Molecular Tests	SLC7A9 (analyzed together with SLC3A1)	604144	CYSTINURIA TYPE 1	220100		7 mutations in SLC7A9: Gly105Arg, Thr123Met, Phe140Ser, Ala182Thr, 747delG, 244delE, Arg333Trp includes 10 mutations in SLC3A1: Thr216Met, Ser217Arg, Arg365Trp, Arg365Leu, Arg362Cys, Met467Thr, Met467Lys, 1500+1G/T, Glu483X duplication of	700

Molecular Tests	SLC7A9 (analyzed together with SLC3A1)	604144	CYSTINURIA TYPE 1	220100		7 mutations in SLC7A9: Gly105Arg, Thr123Met, Phe140Ser, Ala182Thr, 747delG, 244delE, Arg333Trp includes 10 mutations in SLC3A1: Thr216Met, Ser217Arg, Arg365Trp, Arg365Leu, Arg362Cys, Met467Thr, Met467Lys, 1500+1G/T, Glu483X duplication of	700
Molecular Tests	SLC7A9 (analyzed together with SLC3A1)	604144	CYSTINURIA TYPE 1	220100		7 mutations in SLC7A9: Gly105Arg, Thr123Met, Phe140Ser, Ala182Thr, 747delG, 244delE, Arg333Trp includes 10 mutations in SLC3A1: Thr216Met, Ser217Arg, Arg365Trp, Arg365Leu, Arg362Cys, Met467Thr, Met467Lys, 1500+1G/T, Glu483X duplication of	700
Molecular Tests	SLC7A9 (analyzed together with SLC3A1)	604144	CYSTINURIA TYPE 1	220100		7 mutations in SLC7A9: Gly105Arg, Thr123Met, Phe140Ser, Ala182Thr, 747delG, 244delE, Arg333Trp includes 10 mutations in SLC3A1: Thr216Met, Ser217Arg, Arg365Trp, Arg365Leu, Arg362Cys, Met467Thr, Met467Lys, 1500+1G/T, Glu483X duplication of	700
Molecular Tests	SLC7A9 (analyzed together with SLC3A1)	604144	CYSTINURIA TYPE 1	220100		7 mutations in SLC7A9: Gly105Arg, Thr123Met, Phe140Ser, Ala182Thr, 747delG, 244delE, Arg333Trp includes 10 mutations in SLC3A1: Thr216Met, Ser217Arg, Arg365Trp, Arg365Leu, Arg362Cys, Met467Thr, Met467Lys, 1500+1G/T, Glu483X duplication of	700
Molecular Tests	SLC7A9 (analyzed together with SLC3A1)	604144	CYSTINURIA TYPE 1	220100		7 mutations in SLC7A9: Gly105Arg, Thr123Met, Phe140Ser, Ala182Thr, 747delG, 244delE, Arg333Trp includes 10 mutations in SLC3A1: Thr216Met, Ser217Arg, Arg365Trp, Arg365Leu, Arg362Cys, Met467Thr, Met467Lys, 1500+1G/T, Glu483X duplication of	700
Molecular Tests	SLC9A6 (SOLUTE CARRIER FAMILY 9, ISOFORM A6)	300231	MENTAL RETARDATION, SYNDROMIC (X-LINKED), CHRISTIANSON TYPE	300243		Sequencing	1650
Molecular Tests	SMAD3 (MOTHERS AGAINST DECAPENTAPLEGIC, DROSOPHILA, HOMOLOG OF,3)	603109	LOEYS-DIETZ SYNDROME, TYPE 1C » ANEURYSMS-OSTEOARTHRITIS SYNDROME, AOS » LOEYS-DIETZ SYNDROME WITH OSTEOARTHRITIS	613795		Sequencing	550
Molecular Tests	SMAD4 (DPC4)	600993	POLYPOSIS, JUVENILE INTESTINAL	174900		Sequencing	740
Molecular Tests	SMAD4 (DPC4)	600993	JUVENILE POLYPOSIS/HEREDITARY HEMORRHAGIC TELANGIECTASIA SYNDROME » POLYPOSIS, GENERALIZED JUVENILE, WITH PULMONARY ARTERIOVENOUS MALFORMATION	175050		Sequencing	740
Molecular Tests	SMARCAL1 (SWI/SNF-RELATED, MATRIX-ASSOCIATED, ACTIN-DEPENDENT REGULATOR OF CHROMATIN, SUBFAMILY A-LIKE PROTEIN 1)	606622	IMMUNOOSSEOUS DYSPLASIA, SCHIMKE TYPE	242900		Sequencing	1000
Molecular Tests	SMC1A (STRUCTURAL MAINTENANCE OF CHROMOSOMES 1A, SMC1L1, SMC1)	300040	CORNELIA DE LANGE SYNDROME (X-LINKED)	300590		Sequencing	850
Molecular Tests	SMC3 (STRUCTURAL MAINTENANCE OF CHROMOSOMES 3; CHONDROITIN SULFATE PROTEOGLYCAN 6; BAMACAN)	606062	CORNELIA DE LANGE SYNDROME, TYPE 3	610759		Sequencing	1200
Molecular Tests	SMCX (JARID1C)	314690	MENTAL RETARDATION, SYNDROMIC (X –LINKED)	300534		Sequencing	1150
Molecular Tests	SMN1 (SURVIVAL MOTOR NEURON PROTEIN)	600354	SPINAL MUSCULAR ATROPHY 1, SMA1	253300		Sequencing	1050
Molecular Tests	SMN1 (SURVIVAL MOTOR NEURON PROTEIN)	600354	SPINAL MUSCULAR ATROPHY 1, SMA1	253300		Common Deletion	400
Molecular Tests	SMN1 (SURVIVAL MOTOR NEURON PROTEIN)	600354	SPINAL MUSCULAR ATROPHY 3, SMA3	253400		Sequencing	1050
Molecular Tests	SMN1 (SURVIVAL MOTOR NEURON PROTEIN)	600354	SPINAL MUSCULAR ATROPHY 3, SMA3	253400		Common Deletion	400
Molecular Tests	SMN1 (SURVIVAL MOTOR NEURON PROTEIN)	600354	SPINAL MUSCULAR ATROPHY 2, SMA2	253550		Sequencing	1050
Molecular Tests	SMN1 (SURVIVAL MOTOR NEURON PROTEIN)	600354	SPINAL MUSCULAR ATROPHY 2, SMA2	253550		Common Deletion	400

Molecular Tests	SMPD1 (SPHINGOMYELINASE)	607608	NIEMANN-PICK DISEASE, TYPE A » SPHINGOMYELIN LIPIDOSIS » SPHINGOMYELINASE DEFICIENCY	257200		Sequencing	960
Molecular Tests	SMPD1 (SPHINGOMYELINASE)	607608	NIEMANN-PICK DISEASE, TYPE A » SPHINGOMYELIN LIPIDOSIS » SPHINGOMYELINASE DEFICIENCY	257200		3 Mutations: R496L, FSP330, L302P	250
Molecular Tests	SMPD1 (SPHINGOMYELINASE)	607608	NIEMANN-PICK DISEASE, TYPE A » SPHINGOMYELIN LIPIDOSIS » SPHINGOMYELINASE DEFICIENCY	257200		1 Mutation: DelR608	250
Molecular Tests	SMPD1 (SPHINGOMYELINASE)	607608	NIEMANN-PICK DISEASE, TYPE B	607616		Sequencing	960
Molecular Tests	SMPD1 (SPHINGOMYELINASE)	607608	NIEMANN-PICK DISEASE, TYPE B	607616		3 Mutations: R496L, FSP330, L302P	250
Molecular Tests	SMPD1 (SPHINGOMYELINASE)	607608	NIEMANN-PICK DISEASE, TYPE B	607616		1 Mutation: DelR608	250
Molecular Tests	SNCA (ALPHA SYNUCLEIN)	163890	LEWY BODY DEMENTIA LEWY BODY DEMENTIA	127750		Sequencing	450
Molecular Tests	SNCA (ALPHA SYNUCLEIN)	163890	LEWY BODY DEMENTIA LEWY BODY DEMENTIA	127750		Deletions	450
Molecular Tests	SNCA (ALPHA SYNUCLEIN)	163890	PARKINSON DISEASE, TYPE 1, PARK1 (AUTOSOMAL DOMINANT) » LEWY BODY PARKINSONISM	168601		Sequencing	450
Molecular Tests	SNCA (ALPHA SYNUCLEIN)	163890	PARKINSON DISEASE, TYPE 1, PARK1 (AUTOSOMAL DOMINANT) » LEWY BODY PARKINSONISM	168601		Deletions	450
Molecular Tests	SNCA (ALPHA SYNUCLEIN)	163890	PARKINSON DISEASE, TYPE 4, PARK4 (AUTOSOMAL DOMINANT)	665543		Sequencing	450
Molecular Tests	SNCA (ALPHA SYNUCLEIN)	163890	PARKINSON DISEASE, TYPE 4, PARK4 (AUTOSOMAL DOMINANT)	665543		Deletions	450
Molecular Tests	SOD1 (SUPEROXIDE DISMUTASE 1)	147450	AMYOTROPHIC LATERAL SCLEROSIS, ALS » LOU GEHRIG'S DISEASE	105400		Sequencing	550
Molecular Tests	SOD1, TARDBP, ANG, ALS6		AMYOTROPHIC LATERAL SCLEROSIS			Panel of 4 Genes	1290
Molecular Tests	SOS1 (SON OF SEVENLESS, DROSOPHILA, HOMOLOG 1)	308700	FIBROMATOSIS, GINGIVAL, TYPE 1	135300		Sequencing	1360
Molecular Tests	SOS1 (SON OF SEVENLESS, DROSOPHILA, HOMOLOG 1)	308700	NOONAN SYNDROME, TYPE 4	610733		Sequencing	1360
Molecular Tests	SOX10 (SRY-BOX 10)	602229	WAARDENBURG-SHAH SYNDROME » WAARDENBURG SYNDROME, TYPE 4 » WAARDENBURG-HIRSCHSPRUNG DISEASE	277580		Sequencing	470
Molecular Tests	SOX10 (SRY-BOX 10)	602229	YEMENITE DEAF-BLIND HYPOPIGMENTATION SYNDROME	601706		Sequencing	470
Molecular Tests	SOX10 (SRY-BOX 10)	602229	WAARDENBURG-SHAH SYNDROME, NEUROLOGIC VARIANT » PERIPHERAL DEMYELINATING NEUROPATHY, CENTRAL DYSMYELINATING LEUKODYSTROPHY, WAARDENBURG SYNDROME, AND HIRSCHSPRUNG DISEASE	609136		Sequencing	470
Molecular Tests	SOX10 (SRY-BOX 10)	602229	WAARDENBURG SYNDROME, TYPE 2E	611584		Sequencing	470
Molecular Tests	SOX2 (SRY-BOX 2)	184429	ANOPHTHALMIA, ANOP3	206900		Sequencing	710
Molecular Tests	SOX3 (SRY-BOX 3)	313430	MENTAL RETARDATION (X-LINKED), WITH ISOLATED GROWTH HORMONE DEFICIENCY, MRGH	300123		Sequencing	530
Molecular Tests	SOX9 (SRY-BOX 9)	608160	CAMPOMELIC DYSPLASIA » ACAMPOMELIC CAMPOMELIC DYSPLASIA	114290		Sequencing	500
Molecular Tests	SPATA7 (SPERMATOGENESIS-ASSOCIATED PROTEIN 7)	609868	LEBER CONGENITAL AMAUROSIS, TYPE 3, LCA3 » RETINITIS PIGMENTOSA, JUVENILE, SPATA7-RELATED » AMAUROSIS CONGENITA OF LEBER 3	604232		Sequencing	680
Molecular Tests	SPG20 (SPARTIN)	607111	FAMILIAL SPASTIC PARAPLEGIA 20 (AUTOSOMAL RECESSIVE), SPG20 » TROYER SYNDROME » SPASTIC PARAPARESIS, CHILDHOOD-ONSET, WITH DISTAL MUSCLE WASTING	275900		Sequencing	950
Molecular Tests	SPG3A (ATLASTIN)	606439	FAMILIAL SPASTIC PARAPLEGIA 3 (AUTOSOMAL DOMINANT), SPG3A, FSP1 » STRUMPELL DISEASE	182600		Sequencing	1000
Molecular Tests	SPG4 (SPASTIN, SPAST)	604277	FAMILIAL SPASTIC PARAPLEGIA 4 (AUTOSOMAL DOMINANT), SPG4, FSP2	182601		Sequencing	1000
Molecular Tests	SPG7 (PARAPLEGIN)	602783	SPASTIC PARAPLEGIA 7 (AUTOSOMAL RECESSIVE), SPG7, FSP7	607259		Sequencing	850
Molecular Tests	SPINK1 (PANCREATIC SECRETORY TRYPSIN INHIBITOR, PSTI, TUMOR-ASSOCIATED TRYPSIN INHIBITOR, TATI)	167790	PANCREATITIS, HEREDITARY, PCTT	167800		Exon 3 of SPINK1 and Exons 1-3 of PRSS1	300
Molecular Tests	SPINK5 (LEKTI)	605010	NETHERTON SYNDROME	256500		Sequencing	2150
Molecular Tests	SPR (SEPIAPTERIN REDUCTASE)	182125	DYSTONIA, DOPA-RESPONSIVE, DUE TO SEPIAPTERIN REDUCTASE DEFICIENCY » SEPIAPTERIN REDUCTASE DEFICIENCY	182125		Sequencing	470
Molecular Tests	SPRED1 (SPROUTY-RELATED EVH1 DOMAIN-CONTAINING PROTEIN 1)	609291	NEUROFIBROMATOSIS, TYPE 1-LIKE SYNDROME » LEGIUS SYNDROME	611431		Sequencing	650
Molecular Tests	SPTBN2 (SPECTRIN, BETA, NONERYTHROCYTIC, 2)	604985	SPINOCEREBELLAR ATAXIA 5, SCA5	600224		Sequencing	2600

Molecular Tests	SPTBN2 (SPECTRIN, BETA, NONERYTHROCYTIC, 2)	604985	SPINOCEREBELLAR ATAXIA 5, SCA5	600224		Exons 7 (c.733P), 12 (c.1592-1630del and 14 (c.1884-1909del)	350
Molecular Tests	SPTLC1 (SERINE PALMITOYLTRANSFERASE, LONG-CHAIN BASE SUBUNIT 1)	605712	NEUROPATHY, HEREDITARY SENSORY AND AUTONOMIC, TYPE 1, HSAN1 » NEUROPATHY, HEREDITARY SENSORY AUTONOMIC, TYPE I, WITH DEAFNESS	162400		Sequencing	1050
Molecular Tests	SRD5A2 (STEROID 5-ALPHA-REDUCTASE 2)	607306	PSEUDOVAGINAL PERINEOSCROTAL HYPOSPADIAS, PPSH » MALE PSEUDOHERMAPHRODITISM DUE TO 5-ALPHA-REDUCTASE DEFICIENCY » FAMILIAL INCOMPLETE MALE PSEUDOHERMAPHRODITISM, TYPE 3	264600		Sequencing	550
Molecular Tests	SRY (SEX-DETERMINING REGION Y, TESTIS-DETERMINING FACTOR, TDF)	480000	GONADAL DYSGENESIS, XY FEMALE TYPE, GDXY » SWYER SYNDROME » SEX REVERSAL	306100		Sequencing	390
Molecular Tests	SRY (SEX-DETERMINING REGION Y, TESTIS-DETERMINING FACTOR, TDF)	480000	GONADAL DYSGENESIS, XY FEMALE TYPE, GDXY » SWYER SYNDROME » SEX REVERSAL	306100		Positive / Negative	250
Molecular Tests	STAR (STEROIDOGENIC ACUTE REGULATORY PROTEIN; START DOMAIN-CONTAINING PROTEIN 1; STARD1)	600617	LIPOID CONGENITAL ADRENAL HYPERPLASIA » ADRENAL HYPERPLASIA, TYPE I	201710		Sequencing	650
Molecular Tests	STAT3 (SIGNAL TRANSDUCER AND ACTIVATOR OF TRANSCRIPTION 3; ACUTE-PHASE RESPONSE FACTOR)	102582	HYPERIMMUNOGLOBULIN E RECURRENT INFECTION SYNDROME (AUTOSOMAL DOMINANT) » JOB SYNDROME » HYPER-IGE SYNDROME (AUTOSOMAL DOMINANT)	147060		Sequencing	1350
Molecular Tests	STK11 (SERINE THREONINE KINASE)	602216	PEUTZ-JEGHERS SYNDROME, PJS » HAMARTOMATOUS INTESTINAL POLYPOSIS	175200		Sequencing and Deletion-Duplication Testing	740
Molecular Tests	STRA6 (STIMULATED BY RETINOIC ACID 6, MOUSE, HOMOLOG OF)	610745	MICROPHTHALMIA, SYNDROMIC, TYPE 9; MCOPS9 » SPEAR SYNDROME » MATTHEW-WOOD SYNDROME » PULMONARY AGENESIS, MICROPHTHALMIA, AND DIAPHRAGMATIC DEFECT, PMD » ANOPHTHALMIA / MICROPHTHALMIA AND PULMONARY HYPOPLASIA	601186		Sequencing	1500
Molecular Tests	STS (STEROID SULFATASE; ARYLSULFATASE C; ARSC)	300747	ICHTHYOSIS (X-LINKED) » STEROID SULFATASE DEFICIENCY	308100		Sequencing	1100
Molecular Tests	STS (STEROID SULFATASE; ARYLSULFATASE C; ARSC)	300747	ICHTHYOSIS (X-LINKED) » STEROID SULFATASE DEFICIENCY	308100		Deletion-Duplication Testing	400
Molecular Tests	STX11 (SYNTAXIN 11)	603914	HEMOPHAGOCYTIC LYMPHOHISTIOCYTOSIS, FAMILIAL, TYPE 4, FHL4	603552		Sequencing	500
Molecular Tests	STX14 (SYNTAXIN 14)	606666	POIKILODERMA PARANEOPLASTICUM, TYPE 1A	601933		Sequencing	600
Molecular Tests	STX16 (SYNTAXIN 16)	605666	POIKILODERMA PARANEOPLASTICUM, TYPE 1B	601933		Deletion-Duplication Testing	350
Molecular Tests	STXBP1 (SYNTAXIN-BINDING PROTEIN 1; MUNC18-1)	602926	EPILEPTIC ENCEPHALOPATHY, EARLY INFANTILE, TYPE 4, EIEE4	612164		Sequencing	1090
Molecular Tests	STXBP2 (SYNTAXIN-BINDING PROTEIN 2; UNC18-2, C. ELEGANS, HOMOLOG OF 2; UNC18B; MUNC18-2)	601717	HEMOPHAGOCYTIC LYMPHOHISTIOCYTOSIS, FAMILIAL, TYPE 5, FHL5	613101		Sequencing	800
Molecular Tests	SUCLA2 (SUCCINATE-CoA LIGASE, ADP-FORMING, BETA SUBUNIT; ATP-SPECIFIC SUCCINYL-CoA SYNTHETASE, BETA SUBUNIT)	603921	MITOCHONDRIAL DNA DEPLETION SYNDROME, ENCEPHALOMYOPATHIC FORM, WITH METHYLMALONIC ACIDURIA	612073		Sequencing	650
Molecular Tests	SUCLG (SUCCINATE-CoA LIGASE, ALPHA SUBUNIT; SUCCINATE-CoA LIGASE, ADP-FORMING, ALPHA SUBUNIT; SUCLA1)	611224	LACTIC ACIDOSIS, FATAL INFANTILE	245400		Sequencing	650
Molecular Tests	SUMF1 (SULFATASE-MODIFYING FACTOR 1)	607939	MULTIPLE SULFATASE DEFICIENCY » MUCOSULFATIDOSIS » SULFATIDOSIS, JUVENILE, VARIANT TYPE	272200		Sequencing	810
Molecular Tests	SURF1 (SURFEIT 1)	185620	CYTOCHROME c OXIDASE DEFICIENCY » COX DEFICIENCY » COMPLEX 4 MITOCHONDRIAL RESPIRATORY CHAIN DEFICIENCY OF	220110		Sequencing	600
Molecular Tests	SURF1 (SURFEIT 1)	185620	LEIGH SYNDROME	256000		Sequencing	600
Molecular Tests	SYCP3 (SYNAPTONEMAL COMPLEX PROTEIN 3)	604709	PREGNANCY LOSS, SUSCEPTIBILITY TO	614980		Sequencing	890
Molecular Tests	SYCP3 (SYNAPTONEMAL COMPLEX PROTEIN 3)	604759	AZOOSPERMIA DUE TO PERTURBATIONS OF MEIOSIS » AZOOSPERMIA WITH MATURATION ARREST	270960		Sequencing	890
Molecular Tests	TAB2 (TAK1-BINDING PROTEIN 2; MITOGEN-ACTIVATED PROTEIN KINASE KINASE KINASE 7-INTERACTING PROTEIN 2; MAP3K7IP2; TAK1 BETA ACTIVATING KINASE 2)	605101	LEFT VENTRICULAR OUTFLOW TRACT OBSTRUCTION			Sequencing	650
Molecular Tests	TACO1 (TRANSLATIONAL ACTIVATOR OF MITOCHONDRIALLY ENCODED CYTOCHROME c OXIDASE SUBUNIT I; TRANSLATIONAL ACTIVATOR OF COX I; COILED-COIL DOMAIN-CONTAINING PROTEIN 44)	612958	CYTOCHROME c OXIDASE DEFICIENCY » COX DEFICIENCY » COMPLEX 4 MITOCHONDRIAL RESPIRATORY CHAIN DEFICIENCY OF	220110		Sequencing	800
Molecular Tests	TARDBP (TAR DNA BINDING PROTEIN)	605078	AMYOTROPHIC LATERAL SCLEROSIS, TYPE 10, ALS10	612069		Sequencing	750
Molecular Tests	TAT (TYROSINE)	613018	RIETH SYNDROME	613018		Sequencing	550

Molecular Tests	TBCE (TUBULIN-SPECIFIC CHAPERONE E)	604934	HYPOPARATHYROIDISM-RETARDATION-DYSMORPHISM SYNDROME » HYPOPARATHYROIDISM WITH SHORT STATURE, MENTAL RETARDATION, AND SEIZURES » SANAD-SAKATI SYNDROME	241410		Sequencing	1150
Molecular Tests	TBCE (TUBULIN-SPECIFIC CHAPERONE E)	604934	HYPOPARATHYROIDISM-RETARDATION-DYSMORPHISM SYNDROME » HYPOPARATHYROIDISM WITH SHORT STATURE, MENTAL RETARDATION, AND SEIZURES » SANAD-SAKATI SYNDROME	241410		12 bp Deletion in Exon 2	540
Molecular Tests	TBCE (TUBULIN-SPECIFIC CHAPERONE E)	604934	KENNY-CAFFEY SYNDROME, TYPE 1, KCS1 (AUTOSOMAL RECESSIVE)	244460		Sequencing	1150
Molecular Tests	TBCE (TUBULIN-SPECIFIC CHAPERONE E)	604934	KENNY-CAFFEY SYNDROME, TYPE 1, KCS1 (AUTOSOMAL RECESSIVE)	244460		43 bp Deletion in Exon 2	540
Molecular Tests	TBP (TATA BOX-BINDING PROTEIN, SCA17)	600075	SPINOCEREBELLAR ATAXIA 17, SCA17	607136	See SCA8, SCA10, SCA12, SCA17	Repeat	250
Molecular Tests	TBX1 (T-BOX 1)	602054	DIGEORGE SYNDROME, DGS » CATCH22	188400		Sequencing	1250
Molecular Tests	TBX1 (T-BOX 1)	602054	DIGEORGE SYNDROME, DGS » CATCH22	188400		Deletion-Duplication Testing	400
Molecular Tests	TBX1 (T-BOX 1)	602054	VELOCARDIOFACIAL SYNDROME, VCFS » SHPRINTZEN SYNDROME	192430		Sequencing	1250
Molecular Tests	TBX1 (T-BOX 1)	602054	VELOCARDIOFACIAL SYNDROME, VCFS » SHPRINTZEN SYNDROME	192430		Deletion-Duplication Testing	400
Molecular Tests	TBX1 (T-BOX 1)	602054	CONOTRUNCAL ANOMALY FACE SYNDROME » DOUBLE-OUTLET RIGHT VENTRICLE	217095		Sequencing	1250
Molecular Tests	TBX1 (T-BOX 1)	602054	CONOTRUNCAL ANOMALY FACE SYNDROME » DOUBLE-OUTLET RIGHT VENTRICLE	217095		Deletion-Duplication Testing	400
Molecular Tests	TBX19 (T-BOX 19, T-BOX FACTOR, PITUITARY)	604614	ACTH DEFICIENCY	201400		Sequencing	1390
Molecular Tests	TBX3 (T-BOX 3)	601621	ULNAR - MAMMARY SYNDROME	181450		Sequencing	1250
Molecular Tests	TBX5 (T-BOX 5)	601620	HOLT-ORAM SYNDROME, HOS1	142900		Sequencing	650
Molecular Tests	TBX5 (T-BOX 5)	601620	HOLT-ORAM SYNDROME, HOS1	142900		Deletions	450
Molecular Tests	TCAP (TITIN-CAP)	604488	MUSCULAR DYSTROPHY, LIMB-GIRDLE, TYPE 2G, LGMD2G	601954		Sequencing	420
Molecular Tests	TCAP (TITIN-CAP)	604488	CARDIOMYOPATHY, DILATED, TYPE 1N	607487		Sequencing	420
Molecular Tests	TCF4 (TRANSCRIPTION FACTOR 4)	602272	PITT-HOPKINS SYNDROME » ENCEPHALOPATHY, SEVERE EPILEPTIC, WITH AUTONOMIC DYSFUNCTION	610954		Sequencing	500
Molecular Tests	TCIRG1 (T CELL IMMUNE REGULATOR 1, TIRC7, IOC116)	604592	OSTEOPETROSIS (AUTOSOMAL RECESSIVE) » MARBLE BONES (AUTOSOMAL RECESSIVE)	259700		Sequencing	1360
Molecular Tests	TCN2 (TRANSCOBALAMIN II; VITAMIN B12-BINDING PROTEIN 2)	613441	TRANSCOBALAMIN 2 DEFICIENCY	275350		Sequencing	990
Molecular Tests	TCOF1 (TREACLE)	606847	TREACHER COLLINS-FRANCESCHETTI SYNDROME (TCOF) » TREACHER COLLINS SYNDROME	154500		Sequencing	980
Molecular Tests	TCOF1 (TREACLE)	606847	TREACHER COLLINS-FRANCESCHETTI SYNDROME (TCOF) » TREACHER COLLINS SYNDROME	154500		Deletion-Duplication Testing	1350
Molecular Tests	TECTA (TECTORIN, ALPHA)	602574	DEAFNESS, DFNA12 » DEAFNESS, AUTOSOMAL DOMINANT, NONSYNDROMIC SENSORINEURAL 12	601543		Sequencing	1320
Molecular Tests	TECTA (TECTORIN, ALPHA)	602574	DEAFNESS, DFNB21 » DEAFNESS, AUTOSOMAL RECESSIVE, NONSYNDROMIC SENSORINEURAL 21	603629		Sequencing	1320
Molecular Tests	TERC (TELOMERASE RNA COMPONENT)	602322	APLASTIC ANEMIA			Sequencing	490
Molecular Tests	TERC (TELOMERASE RNA COMPONENT)	602322	DYSKERATOSIS CONGENITA (AUTOSOMAL DOMINANT) » DYSKERATOSIS CONGENITA, SCROGGINS TYPE	127550		Sequencing	490

Molecular Tests	TFR2 (TRANSFERRIN RECEPTOR 2)	604720	HEMOCHROMATOSIS, TYPE 3, HFE3 » HEMOCHROMATOSIS DUE TO DEFECT IN TRANSFERRIN RECEPTOR 2	604720		Sequencing	3100
Molecular Tests	TG (SIMPLE, THYROGLOBULIN)	188450	GOITER, FAMILIAL, WITH HYPOTHYROIDISM (AUTOSOMAL RECESSIVE) » GOITER, NONENDEMIC SIMPLE » GOITER, ADENOMATOUS » [illegible]	188450		Sequencing	450
Molecular Tests	TGFB1 (TGFB, TRANSFORMING GROWTH FACTOR, BETA-1)	190181	CAMURATI-ENGELMANN DISEASE » [illegible]	131300		Sequencing	800
Molecular Tests	TGFB1 (TGFB, TRANSFORMING GROWTH FACTOR, BETA-1)	190181	CAMURATI-ENGELMANN DISEASE » [illegible]	131300		Exon 24	400
Molecular Tests	TGFBI (TRANSFORMING GROWTH FACTOR, BETA-INDUCED, 68-KD; KERATOEPITHELIN; BETA-IG-H3; BIGH3)	601692	CORNEAL DYSTROPHY, EPITHELIAL BASEMENT MEMBRANE » COGAN CORNEAL DYSTROPHY » CORNEAL DYSTROPHY, MAP-DOT-FINGERPRINT TYPE » [illegible]	121820		Sequencing	770
Molecular Tests	TGFBI (TRANSFORMING GROWTH FACTOR, BETA-INDUCED, 68-KD; KERATOEPITHELIN; BETA-IG-H3; BIGH3)	601692	CORNEAL DYSTROPHY, GROENOUW TYPE 1 » GRANULAR CORNEAL DYSTROPHY, TYPE 1 » [illegible]	121900		Sequencing	770
Molecular Tests	TGFBI (TRANSFORMING GROWTH FACTOR, BETA-INDUCED, 68-KD; KERATOEPITHELIN; BETA-IG-H3; BIGH3)	601692	CORNEAL DYSTROPHY, LATTICE TYPE 1	122200		Sequencing	770
Molecular Tests	TGFBI (TRANSFORMING GROWTH FACTOR, BETA-INDUCED, 68-KD; KERATOEPITHELIN; BETA-IG-H3; BIGH3)	601692	CORNEAL DYSTROPHY OF BOWMAN LAYER, TYPE 2 » CORNEAL DYSTROPHY, THIEL-BEHNKE TYPE » CORNEAL DYSTROPHY, HONEYCOMB-SHAPED	607882		Sequencing	770
Molecular Tests	TGFBI (TRANSFORMING GROWTH FACTOR, BETA-INDUCED, 68-KD; KERATOEPITHELIN; BETA-IG-H3; BIGH3)	601692	CORNEAL DYSTROPHY, AVELLINO TYPE » COMBINED GRANULAR-LATTICE CORNEAL DYSTROPHY » [illegible]	607541		Sequencing	770
Molecular Tests	TGFBI (TRANSFORMING GROWTH FACTOR, BETA-INDUCED, 68-KD; KERATOEPITHELIN; BETA-IG-H3; BIGH3)	601692	CORNEAL DYSTROPHY OF BOWMAN LAYER, TYPE 1 » CORNEAL DYSTROPHY, REIS-BUCKLERS TYPE » CORNEAL DYSTROPHY, GEOGRAPHIC » [illegible]	608470		Sequencing	770
Molecular Tests	TGFBI (TRANSFORMING GROWTH FACTOR, BETA-INDUCED, 68-KD; KERATOEPITHELIN; BETA-IG-H3; BIGH3)	601692	CORNEAL DYSTROPHY, LATTICE TYPE 3A	608471		Sequencing	770
Molecular Tests	TGFBR1 (TRANSFORMING GROWTH FACTOR, BETA RECEPTOR, TYPE I, ALK5)	190181	LOEYS-DIETZ SYNDROME	609192		Sequencing	550
Molecular Tests	TGFBR1 and TGFBR2	190182	MARFAN SYNDROME, TYPE 2, MFS2 » MARFANLIKE CONNECTIVE TISSUE DISORDER	154705		Sequencing	820
Molecular Tests	TGFBR1 and TGFBR2	190182	LOEYS-DIETZ SYNDROME	609192			820
Molecular Tests	TGFBR2 (TRANSFORMING GROWTH FACTOR-BETA RECEPTOR, TYPE 2)	190182	MARFAN SYNDROME, TYPE 2, MFS2 » MARFANLIKE CONNECTIVE TISSUE DISORDER	154705		Sequencing	580
Molecular Tests	TGFBR2 (TRANSFORMING GROWTH FACTOR, BETA RECEPTOR, TYPE II)	190182	LOEYS-DIETZ SYNDROME	609192		Sequencing	580
Molecular Tests	TGIF	602630	HOLOPROSENCEPHALY 4	142946		Sequencing	1
Molecular Tests	TGM1 (TRANSGLUTAMINASE)	190195	ICHTHYOSIFORM ERYTHRODERMA, NONBULLOUS CONGENITAL	242300		Sequencing	1550
Molecular Tests	TGM1 (TRANSGLUTAMINASE)	190195	COLLODION FETUS	242300		Sequencing	1550
Molecular Tests	TH (TYROSINE HYDROXYLASE)	191290	LAMELLAR ICHTHYOSIS SEGAWA SYNDROME (AUTOSOMAL RECESSIVE) » TYROSINE HYDROXYLASE DEFICIENCY » DOPA-RESPONSIVE DYSTONIA (AUTOSOMAL RECESSIVE) » [illegible]	242300 605407		Sequencing	1550 850
Molecular Tests	THAP1 (THAP DOMAIN CONTAINING PROTEIN 1)	609739	DYSTONIA 6, DYT6	602629		Sequencing	550
Molecular Tests	THBD (THROMBOMODULIN)	188040	THROMBOPHILIA DUE TO THROMBOMODULIN DEFECT	188040		Sequencing	450
Molecular Tests	THBD (THROMBOMODULIN)	188040	BEHCET'S DISEASE, SUSCEPTIBILITY TO, TYPE 4	613076		Sequencing	450
Molecular Tests	THRB (THYROID HORMONE RECEPTOR, BETA, ERBA2)	190160	THYROID HORMONE RESISTANCE, SELECTIVE PITUITARY » HYPERTHYROIDISM, FAMILIAL, DUE TO INAPPROPRIATE THYROTROPIN SECRETION	145600		Sequencing	1230
Molecular Tests	THRB (THYROID HORMONE RECEPTOR, BETA, ERBA2)	190160	THYROID HORMONE RESISTANCE, GENERALIZED (AUTOSOMAL DOMINANT) » HYPERTHYROXINEMIA, FAMILIAL, EUTHYROID, SECONDARY TO PITUITARY AND PERIPHERAL RESISTANCE TO THYROID HORMONES	188570		Sequencing	1230
Molecular Tests	THRB (THYROID HORMONE RECEPTOR, BETA, ERBA2)	190160	THYROID HORMONE RESISTANCE, GENERALIZED (AUTOSOMAL RECESSIVE) » THYROID HORMONE UNRESPONSIVENESS » REFETOFF SYNDROME	274500		Sequencing	1230
Molecular Tests	TIMM8A (TRANSLOCASE OF INNER MITOCHONDRIAL MEMBRANE 8, YEAST, HOMOLOG OF, A; DEAFNESS/DYSTONIA PEPTIDE 1; DDP1)	300356	MOHR-TRANEBJAERG SYNDROME » DYSTONIA-DEAFNESS SYNDROME » DEAFNESS-DYSTONIA-OPTIC ATROPHY SYNDROME » [illegible]	304700		Sequencing	490

Molecular Tests	TIMM8A (TRANSLOCASE OF INNER MITOCHONDRIAL MEMBRANE 8, YEAST, HOMOLOG OF, A; DEAFNESS/DYSTONIA PEPTIDE 1; DDP1)	300356	JENSEN SYNDROME » OPTICOACOUSTIC NERVE ATROPHY WITH DEMENTIA	311150		Sequencing	490
Molecular Tests	TIMP (THYMIDINE PHOSPHORYLASE; ECGF1; ENDOTHELIAL CELL GROWTH FACTOR, PLATELET-DERIVED, GLIOSTATIN)	131222	MITOCHONDRIAL NEUROGASTROINTESTINAL ENCEPHALOPATHY SYNDROME, MNGIE » MYONEUROGASTROINTESTINAL ENCEPHALOPATHY SYNDROME » POLYNEUROPATHY, OPHTHALMOPLEGIA, LEUKOENCEPHALOPATHY, AND INTESTINAL PSEUDOOBSTRUCTION » POLIP SYNDROME	603041		Sequencing	1080
Molecular Tests	TIMP3 (TISSUE INHIBITOR OF METALLOPROTEINASE 3)	188826	FUNDUS DYSTROPHY, PSEUDOINFLAMMATORY, OF SORSBY » SORSBY SYNDROME	136900		Sequencing	700
Molecular Tests	TITF1 (THYROID NUCLEAR FACTOR, NKX2A)	600635	CHOREA, HEREDITARY BENIGN	118700		Sequencing	490
Molecular Tests	TK2 (THYMIDINE KINASE, MITOCHONDRIAL)	188250	MITOCHONDRIAL DNA DEPLETION SYNDROME, MYOPATHIC FORM » MITOCHONDRIAL DNA DEPLETION MYOPATHY	609560		Sequencing	770
Molecular Tests	TM4SF2 (TRANSMEMBRANE 4 SUPERFAMILY, MEMBER 2)	300096	MENTAL RETARDATION, NONSPECIFIC (X-LINKED), TYPE 58, MRX58	300210		Sequencing	750
Molecular Tests	TMC1 (TRANSMEMBRANE COCHLEAR-EXPRESSED GENE 1)	606706	DEAFNESS, DFNB7 » DEAFNESS, (AUTOSOMAL RECESSIVE), NONSYNDROMIC SENSORINEURAL 7 » DEAFNESS, DFNB11	600974		Sequencing	1300
Molecular Tests	TMC1 (TRANSMEMBRANE COCHLEAR-EXPRESSED GENE 1)	606706	DEAFNESS, DFNA36 » DEAFNESS, (AUTOSOMAL DOMINANT), NONSYNDROMIC SENSORINEURAL 36	606705		Sequencing	1300
Molecular Tests	TMEM126A (TRANSMEMBRANE PROTEIN 126A)	612988	OPTIC ATROPHY, TYPE 7, OPA7	612989		Sequencing	850
Molecular Tests	TMEM127 (TRANSMEMBRANE PROTEIN 127)	613403	PHEOCHROMOCYTOMA	171300		Sequencing	560
Molecular Tests	TMEM67 (MKS3, MECKELIN, TRANSMEMBRANE PROTEIN 67)	609884	MECKEL SYNDROME, TYPE 3 » DYSENCEPHALIA SPLANCHNOCYSTICA » GRUBER SYNDROME	607361		Sequencing	2600
Molecular Tests	TMEM67 (MKS3, MECKELIN, TRANSMEMBRANE PROTEIN 67)	609884	JOUBERT SYNDROME, TYPE 6	610688		Sequencing	2600
Molecular Tests	TMEM70 (TRANSMEMBRANE PROTEIN 70)	612418	MITOCHONDRIAL COMPLEX 5 (ATP SYNTHASE) DEFICIENCY, NUCLEAR TYPE 2 » ENCEPHALOCARDIOMYOPATHY, MITOCHONDRIAL, NEONATAL, DUE TO ATP SYNTHASE DEFICIENCY	614052		Sequencing	500
Molecular Tests	TMIE (TRANSMEMBRANE INNER EAR-EXPRESSED GENE)	607237	DEAFNESS, DFNB6 » DEAFNESS, (AUTOSOMAL RECESSIVE), NONSYNDROMIC SENSORINEURAL 6	600971		Sequencing	350
Molecular Tests	TNFRSF13B (TUMOR NECROSIS FACTOR RECEPTOR SUPERFAMILY, MEMBER 13B, TRANSMEMBRANE ACTIVATOR AND CAML INTERACTOR, TACI1)	604907	COMMON VARIABLE IMMUNODEFICIENCY » COMMON VARIABLE HYPOGAMMAGLOBULINEMIA » HYPOGAMMAGLOBULINEMIA, ACQUIRED	240500		Sequencing	450
Molecular Tests	TNFRSF1A (TNFR1)	191190	HIBERNIAN FEVER, FAMILIAL » FAMILIAL PERIODIC FEVER (AUTOSOMAL DOMINANT)	142680		Exons 2-4 (> 95% of mutations)	350
Molecular Tests	TNFRSF6 (TUMOR NECROSIS FACTOR RECEPTOR SUPERFAMILY, MEMBER 6, APT1, FAS)	134637	AUTOIMMUNE LYMPHOPROLIFERATIVE SYNDROME, TYPE 1, ALPS, ALPS1A, ALPS1B » CANALE-SMITH SYNDROME	601859		Sequencing	1380
Molecular Tests	TNNI2 (TROPONIN 1, FAST-TWITCH SKELETAL MUSCLE ISOFORM)	191043	ARTHROGRYPOSIS, DISTAL, TYPE 2B » ARTHROGRYPOSIS MULTIPLEX CONGENITA, DISTAL, TYPE 2B » SHELDON-HALL SYNDROME » FREEMAN-SHELDON SYNDROME VARIANT	601680		Sequencing	500
Molecular Tests	TNNI3 (TROPONIN I, CARDIAC)	191044	CARDIOMYOPATHY, FAMILIAL RESTRICTIVE, 1	115210		Sequencing	500
Molecular Tests	TNNI3 (TROPONIN I, CARDIAC)	191044	HYPERTROPHIC CARDIOMYOPATHY, FAMILIAL, 7, CMH7 » VENTRICULAR HYPERTROPHY, HEREDITARY » ASYMMETRIC SEPTAL HYPERTROPHY	191044		Sequencing	500
Molecular Tests	TNNT1 (TROPONIN T1, SKELETAL, SLOW; TROPONIN T)	191041	NEMALINE MYOPATHY 5, NEM5 » NEMALINE MYOPATHY, AMISH TYPE	605355		Sequencing	950
Molecular Tests	TNNT2 (TROPONIN T2, CARDIAC)	191045	HYPERTROPHIC CARDIOMYOPATHY, FAMILIAL, 2, CMH2 » VENTRICULAR HYPERTROPHY, HEREDITARY » ASYMMETRIC SEPTAL HYPERTROPHY	115195		Sequencing	600
Molecular Tests	TNNT2 (TROPONIN T2, CARDIAC)	191045	DILATED CARDIOMYOPATHY, 1D, CMD1D	601494		Sequencing	600

Molecular Tests	TNNT3 (TROPONIN T3, FAST SKELETAL)	600692	ARTHROGRYPOSIS, DISTAL, TYPE 2B » ARTHROGRYPOSIS MULTIPLEX CONGENITA, DISTAL, TYPE 2B » SHELDON-HALL SYNDROME » FREEMAN-SHELDON SYNDROME VARIANT	601680		Sequencing	1250
Molecular Tests	TOPORS (TOPOISOMERASE I-BINDING ARGININE/SERINE-RICH PROTEIN; p53-BINDING PROTEIN 3; P53BP3)	609507	RETINITIS PIGMENTOSA, TYPE 31, RP31	609923		Sequencing	680
Molecular Tests	TP73L (TUMOR PROTEIN p73-LIKE)	603273	ACRO-DERMATO-UNGUAL-LACRIMAL-TOOTH SYNDROME » ADULT SYNDROME	103285		Sequencing	1500
Molecular Tests	TP73L (TUMOR PROTEIN p73-LIKE)	603273	ACRO-DERMATO-UNGUAL-LACRIMAL-TOOTH SYNDROME » ADULT SYNDROME	103285		Deletion-Duplication Testing	600
Molecular Tests	TP73L (TUMOR PROTEIN p73-LIKE)	603273	ANKYLOBLEPHARON-ECTODERMAL DEFECTS WITH CLEFT LIP AND PALATE » HAY-WELLS SYNDROME	106260		Sequencing	1500
Molecular Tests	TP73L (TUMOR PROTEIN p73-LIKE)	603273	ANKYLOBLEPHARON-ECTODERMAL DEFECTS WITH CLEFT LIP AND PALATE » HAY-WELLS SYNDROME	106260		Deletion-Duplication Testing	600
Molecular Tests	TP73L (TUMOR PROTEIN p73-LIKE)	603273	ECTODERMAL DYSPLASIA, ANHIDROTIC, WITH CLEFT LIP/PALATE » RAPP-HODGKIN SYNDROME	129400		Sequencing	1500
Molecular Tests	TP73L (TUMOR PROTEIN p73-LIKE)	603273	ECTODERMAL DYSPLASIA, ANHIDROTIC, WITH CLEFT LIP/PALATE » RAPP-HODGKIN SYNDROME	129400		Deletion-Duplication Testing	600
Molecular Tests	TP73L (TUMOR PROTEIN p73-LIKE)	603273	ECTRODACTYLY-ECTODERMAL DYSPLASIA-CLEFTING, TYPE 1, EEC1 » ECTODERMAL DYSPLASIA, ECTRODACTYLY, CLEFTING, TYPE 1	129900		Sequencing	1500
Molecular Tests	TP73L (TUMOR PROTEIN p73-LIKE)	603273	ECTRODACTYLY-ECTODERMAL DYSPLASIA-CLEFTING, TYPE 1, EEC1 » ECTODERMAL DYSPLASIA, ECTRODACTYLY, CLEFTING, TYPE 1	129900		Deletion-Duplication Testing	600
Molecular Tests	TP73L (TUMOR PROTEIN p73-LIKE)	603273	LIMB-MAMMARY SYNDROME	603543		Sequencing	1500
Molecular Tests	TP73L (TUMOR PROTEIN p73-LIKE)	603273	LIMB-MAMMARY SYNDROME	603543		Deletion-Duplication Testing	600
Molecular Tests	TP73L (TUMOR PROTEIN p73-LIKE)	603273	ECTRODACTYLY-ECTODERMAL DYSPLASIA-CLEFTING, TYPE 3, EEC3 » ECTODERMAL DYSPLASIA, ECTRODACTYLY, CLEFTING, TYPE 3	604292		Sequencing	1500
Molecular Tests	TP73L (TUMOR PROTEIN p73-LIKE)	603273	ECTRODACTYLY-ECTODERMAL DYSPLASIA-CLEFTING, TYPE 3, EEC3 » ECTODERMAL DYSPLASIA, ECTRODACTYLY, CLEFTING, TYPE 3	604292		Deletion-Duplication Testing	600
Molecular Tests	TP73L (TUMOR PROTEIN p73-LIKE)	603273	SPLIT HAND - SPLIT FOOT, TYPE 4, SHFM4	605289		Sequencing	1500
Molecular Tests	TP73L (TUMOR PROTEIN p73-LIKE)	603273	SPLIT HAND - SPLIT FOOT, TYPE 4, SHFM4	605289		Deletion-Duplication Testing	600
Molecular Tests	TPM1 (TROPOMYOSIN 1)	191010	HYPERTROPHIC CARDIOMYOPATHY, FAMILIAL, 3, CMH3 » VENTRICULAR HYPERTROPHY, HEREDITARY » ASYMMETRIC SEPTAL HYPERTROPHY	115196		Sequencing	500
Molecular Tests	TPM2 (TROPOMYOSIN 2, beta TROPOMYOSIN)	190990	ARTHROGRYPOSIS, DISTAL, TYPE 1 » ARTHROGRYPOSIS MULTIPLEX CONGENITA, DISTAL, TYPE 1	108120		Sequencing	850
Molecular Tests	TPM2 (TROPOMYOSIN 2, beta TROPOMYOSIN)	190990	NEMALINE MYOPATHY 4, NEM4 » NEMALINE MYOPATHY CAUSED BY MUTATION IN THE TROPOMYOSIN 2 GENE	609285		Sequencing	850
Molecular Tests	TPM3 (TROPOMYOSIN 3; ALPHA-TROPOMYOSIN, SLOW SKELETAL; TRK ONCOGENE)	191030	MYOPATHY, CONGENITAL, WITH FIBER-TYPE DISPROPORTION, CFTD	255310		Sequencing	900
Molecular Tests	TPM3 (TROPOMYOSIN 3; ALPHA-TROPOMYOSIN, SLOW SKELETAL; TRK ONCOGENE)	191030	NEMALINE MYOPATHY 1, NEM1 » CAP MYOPATHY, TPM3-RELATED	609284		Sequencing	900
Molecular Tests	TREM2	605086	PRESENILE DEMENTIA WITH BONE CYSTS » POLYCYSTIC LIPOMEMBRANOUS OSTEODYSPLASIA WITH SCLEROSING LEUKOENCEPHALOPATHY » DEMENTIA, PROGRESSIVE, WITH LIPOMEMBRANOUS POLYCYSTIC OSTEODYSPLASIA » BRAIN-BONE-FAT DISEASE » DEMENTIA, PREFRONTAL, WITH BONE CYSTS	221770		Sequencing	350
Molecular Tests	TREX1 (3-PRIME @REPAIR EXONUCLEASE 1, ATRIP)	606605	AICARDI-GOUTIERES SYNDROME 1 » ENCEPHALOPATHY, FAMILIAL INFANTILE, WITH INTRACRANIAL CALCIFICATION AND CHRONIC CEREBROSPINAL FLUID LYMPHOCYTOSIS » CREE ENCEPHALITIS » PSEUDO-TORCH SYNDROME	225750		Sequencing	510
Molecular Tests	TRIM32 (TRIPARTITE MOTIF-CONTAINING PROTEIN 32)	602290	MUSCULAR DYSTROPHY, LIMB-GIRDLE, TYPE 2H, LGMD2H	254110		Sequencing	730
Molecular Tests	TRMU (tRNA 5-METHYLAMINOMETHYL-2-THIOURIDYLATE METHYLTRANSFERASE; TRNT1)	610230	LIVER FAILURE, INFANTILE, TRANSIENT	613070		Sequencing	1210
Molecular Tests	TRPC6 (TRANSIENT RECEPTOR POTENTIAL CATION CHANNEL, SUBFAMILY C, MEMBER 6)	603652	FOCAL SEGMENTAL GLOMERULOSCLEROSIS, TYPE 2	603965		Sequencing	750
Molecular Tests	TRPM6 (TRANSIENT RECEPTOR POTENTIAL CATION CHANNEL, SUBFAMILY M, MEMBER 6;CHANNEL KINASE 2; CHAK2 MELASTATIN-RELATED TRP CATION CHANNEL 6)	300095	HYPOMAGNESEMIA WITH SECONDARY HYPOCALCEMIA » HYPOMAGNESEMIA, INTESTINAL, WITH SECONDARY HYPOCALCEMIA	300523		Sequencing	1500

Molecular Tests	TRPM7 (TRANSIENT RECEPTOR POTENTIAL CATION CHANNEL, SUBFAMILY M, MEMBER 7;LONG TRANSIENT RECEPTOR POTENTIAL CHANNEL 7; LTRPC7 TRANSIENT RECEPTOR POTENTIAL-PHOSPHOLIPASE C-INTERACTING KINASE; TRP-PLIK; CHAK)	605692	GUAM DISEASE » AMYOTROPHIC LATERAL SCLEROSIS-PARKINSONISM/DEMENTIA COMPLEX 1	105500		Sequencing	Upon Request
Molecular Tests	TRPS1 (ZINC FINGER TRANSCRIPTION FACTOR TRPS1)	604386	TRICHORHINOPHALANGEAL SYNDROME, TYPE 1, TRPS1	190350		Sequencing	1070
Molecular Tests	TRPV4 (TRANSIENT RECEPTOR POTENTIAL CATION CHANNEL, SUBFAMILY V, MEMBER 4; VANILLOID RECEPTOR-RELATED OSMOTICALLY ACTIVATED CHANNEL; OSM9-LIKE TRANSIENT RECEPTOR POTENTIAL CHANNEL 4; TRANSIENT RECEPTOR POTENTIAL CHANNEL 12)	605427	BRACHYOLMIA, TYPE 3 » BRACHYOLMIA (AUTOSOMAL DOMINANT) » BRACHYRACHIA	113500		Sequencing	1580
Molecular Tests	TRPV4 (TRANSIENT RECEPTOR POTENTIAL CATION CHANNEL, SUBFAMILY V, MEMBER 4; VANILLOID RECEPTOR-RELATED OSMOTICALLY ACTIVATED CHANNEL; OSM9-LIKE TRANSIENT RECEPTOR POTENTIAL CHANNEL 4; TRANSIENT RECEPTOR POTENTIAL CHANNEL 12)	605427	METATROPIC DYSPLASIA » METATROPIC DWARFISM	156530		Sequencing	1580
Molecular Tests	TRPV4 (TRANSIENT RECEPTOR POTENTIAL CATION CHANNEL, SUBFAMILY V, MEMBER 4; VANILLOID RECEPTOR-RELATED OSMOTICALLY ACTIVATED CHANNEL; OSM9-LIKE TRANSIENT RECEPTOR POTENTIAL CHANNEL 4; TRANSIENT RECEPTOR POTENTIAL CHANNEL 12)	605427	SPONDYLOMETAPHYSEAL DYSPLASIA, KOZLOWSKI TYPE	184252		Sequencing	1580
Molecular Tests	TSC1 and TSC2		TUBEROUS SCLEROSIS	191100	At least 20 microgram DNA is needed	Sequencing of 2 Genes	1700
Molecular Tests	TSC1 (HAMARTIN)		TUBEROUS SCLEROSIS	191100	At least 20 microgram DNA is needed	Deletion Testing	620
Molecular Tests	TSC2 (TUBERIN)		TUBEROUS SCLEROSIS	191100	At least 20 microgram DNA is needed	Deletion Testing	610
Molecular Tests	TSEN54 (tRNA SPLICING ENDONUCLEASE 54, S. CEREVISIAE, HOMOLOG OF; SEN54)	608755	PONTOCEREBELLAR HYPOPLASIA, TYPE 4, PCH4 » ENCEPHALOPATHY, FATAL INFANTILE, WITH OLIVOPONTOCEREBELLAR HYPOPLASIA	225753		Sequencing	1680
Molecular Tests	TSEN54 (tRNA SPLICING ENDONUCLEASE 54, S. CEREVISIAE, HOMOLOG OF; SEN54)	608755	PONTOCEREBELLAR HYPOPLASIA, TYPE 2A, PCH2A » VOLENDAM NEURODEGENERATIVE DISEASE » PONTOCEREBELLAR HYPOPLASIA WITH PROGRESSIVE CEREBRAL ATROPHY	277470		Sequencing	1680
Molecular Tests	TSFM (Ts TRANSLATION ELONGATION FACTOR, MITOCHONDRIAL)	604723	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY 3, COXPD3 » ENCEPHALOMYOPATHY, RESPIRATORY FAILURE, AND LACTIC ACIDOSIS	610505		Sequencing	500
Molecular Tests	TSHR (THYROID-STIMULATING HORMONE RECEPTOR)	603372	THYROID ADENOMA, HYPERFUNCTIONING			Sequencing	690
Molecular Tests	TSHR (THYROID-STIMULATING HORMONE RECEPTOR)	603372	THYROTROPIN, UNRESPONSIVENESS TO			Sequencing	690
Molecular Tests	TSHR (THYROID-STIMULATING HORMONE RECEPTOR)	603372	HYPERTHYROIDISM			Sequencing	690
Molecular Tests	TSHR (THYROID-STIMULATING HORMONE RECEPTOR)	603372	HYPOTHYROIDISM			Sequencing	690
Molecular Tests	TSHR (THYROID-STIMULATING HORMONE RECEPTOR)	603372	THYROID CARCINOMA			Sequencing	690
Molecular Tests	TSHR (THYROID-STIMULATING HORMONE RECEPTOR)	603372	GRAVES DISEASE	275000		Sequencing	690
Molecular Tests	TTC19 (TETRATRICOPEPTIDE REPEAT DOMAIN 19)	613814	CYTOCHROME c OXIDASE DEFICIENCY » COX DEFICIENCY » COMPLEX 3, MITOCHONDRIAL RESPIRATORY CHAIN, DEFICIENCY OF	124000		Sequencing	1060
Molecular Tests	TTC8 (TETRATRICOPEPTIDE REPEAT DOMAIN 8; BBS8)	608132	BARDET-BIEDL SYNDROME TYPE 8, BBS8	209900		Sequencing	680
Molecular Tests	TTC8 (TETRATRICOPEPTIDE REPEAT DOMAIN 8; BBS8)	608132	RETINITIS PIGMENTOSA, TYPE 51, RP51	613464		Sequencing	680
Molecular Tests	TTID (TITIN IMMUNOGLOBULIN DOMAIN PROTEIN, MYOTILIN, MYOT)	604103	MUSCULAR DYSTROPHY, LIMB-GIRDLE, TYPE 1A, LGMD1A	159000		Sequencing	700
Molecular Tests	TTID (TITIN IMMUNOGLOBULIN DOMAIN PROTEIN, MYOTILIN, MYOT)	604103	MYOTILINOPATHY » MYOPATHY, MYOFIBRILLAR, MYOTILIN-RELATED	609200		Sequencing	700
Molecular Tests	TTN (TITIN, CONNECTIN)	188840	CARDIOMYOPATHY, FAMILIAL HYPERTROPHIC, TYPE 9	188840		Exons 312-313	250
Molecular Tests	TTN (TITIN, CONNECTIN)	188840	TIBIAL MUSCULAR DYSTROPHY, TARDIVE » UDD MYOPATHY	600334		Exons 312-313	250
Molecular Tests	TTN (TITIN, CONNECTIN)	188840	CARDIOMYOPATHY, DILATED, 1G, CMD1G	604145		Exons 312-313	250
Molecular Tests	TTPA (TOCOPHEROL TRANSFER PROTEIN, ALPHA, TTP1)	600415	ATAXIA AND RETINITIS PIGMENTOSA WITH ISOLATED VITAMIN E DEFICIENCY			Sequencing	550
Molecular Tests	TTPA (TOCOPHEROL TRANSFER PROTEIN, ALPHA, TTP1)	600415	VITAMIN E, FAMILIAL ISOLATED DEFICIENCY OF » ATAXIA, FRIEDREICH-LIKE, WITH SELECTIVE VITAMIN E DEFICIENCY » FRIEDREICH-LIKE ATAXIA	277460		Sequencing	550
Molecular Tests	TTR (TRANSTHYRETIN)	176300	AMYLOID POLYNEUROPATHY » AMYLOIDOSIS, TYPE 1	176300		Sequencing	580
Molecular Tests	TUBA1A (TUBULIN, ALPHA-1A)	602529	LISSENCEPHALY 3, LIS3	611603		Sequencing	970
Molecular Tests	TUBB2B (TUBULIN, BETA-2B)	612850	POLYMICROGYRIA, ASYMMETRIC	610031		Sequencing	1060
Molecular Tests	TUBB3 (TUBULIN, BETA-3; TUBB4)	602661	FIBROSIS OF EXTRAOCULAR MUSCLES, CONGENITAL, TYPE 3A, WITH OR WITHOUT EXTRAOCULAR INVOLVEMENT	600638		Sequencing	745
Molecular Tests	TUBB3 (TUBULIN, BETA-3; TUBB4)	602661	CORTICAL DYSPLASIA, COMPLEX, WITH OTHER BRAIN MALFORMATIONS	614039		Sequencing	745
Molecular Tests	TUFM (Tu TRANSLATION ELONGATION FACTOR, MITOCHONDRIAL; EF-Tu, MITOCHONDRIAL)	602389	COMBINED OXIDATIVE PHOSPHORYLATION DEFICIENCY, TYPE 4	610678		Sequencing	900
Molecular Tests	TULP1 (TUBBY-LIKE PROTEIN 1)	602280	RETINITIS PIGMENTOSA, TYPE 14, RP14 » RETINITIS PIGMENTOSA, JUVENILE, TULP1-RELATED	600132		Sequencing	680
Molecular Tests	TULP1 (TUBBY-LIKE PROTEIN 1)	602280	LEBER CONGENITAL AMAUROSIS, TYPE 15, LCA15	613843		Sequencing	680
Molecular Tests	TUSC3 (TUMOR SUPPRESSOR CANDIDATE 3)	601385	MENTAL RETARDATION (AUTOSOMAL RECESSIVE), TYPE 7, MRT7	611093		Sequencing	1110
Molecular Tests	TWIST	601622	SAETHRE-CHOTZEN SYNDROME » CRANIOSYNOSTOSIS, SAETHRE-CHOTZEN SYNDROME	101400		Sequencing	510

Molecular Tests	TWIST	601622	ROBINOW-SORAUF SYNDROME	180750		Sequencing	510
Molecular Tests	TYMP (THYMIDINE PHOSPHORYLASE; PLATELET-DERIVED ENDOTHELIAL CELL GROWTH FACTOR; GLIOSTATIN)	131222	MITOCHONDRIAL NEUROGASTROINTESTINAL ENCEPHALOPATHY SYNDROME, MNGIE » MYONEUROGASTROINTESTINAL ENCEPHALOPATHY SYNDROME » POLYNEUROPATHY, OPHTHALMOPLEGIA, LEUKOENCEPHALOPATHY, AND INTESTINAL PSEUDOOBSTRUCTION » POLIP SYNDROME	603041		Sequencing	990
Molecular Tests	TYR (OCA1, TYROSINASE)	606933	OCULOCUTANEOUS ALBINISM, TYPE 1, OCA1 (TYROSINASE-NEGATIVE)	203100		Sequencing	1010
Molecular Tests	TYRP1 (TYROSINASE-RELATED PROTEIN 1; CATALASE B)	115501	OCULOCUTANEOUS ALBINISM, TYPE 3, OCA3	203290		Sequencing	530
Molecular Tests	TYRP1 (TYROSINASE-RELATED PROTEIN 1; CATALASE B)	115501	ALBINISM, RUFOUS OCULOCUTANEOUS, ROCA	278400		Sequencing	530
Molecular Tests	UBE1 (UBIQUITIN-ACTIVATING ENZYME 1)	314370	SPINAL MUSCULAR ATROPHY (X-LINKED), TYPE 2, SMAX2 » SPINAL MUSCULAR ATROPHY (X-LINKED), LETHAL INFANTILE » ARTHROGRYPOSIS MULTIPLEX CONGENITA, DISTAL, (X-LINKED)	301830		Sequencing	2500
Molecular Tests	UBE3A	601623	ANGELMAN SYNDROME, AS	105830		Sequencing	990
Molecular Tests	UBE3A	601623	ANGELMAN SYNDROME, AS	105830		IC Deletion	570
Molecular Tests	UBE3A	601623	ANGELMAN SYNDROME, AS	105830		Methylation	390
Molecular Tests	UBR1 (UBIQUITIN-PROTEIN LIGASE E3 COMPONENT N-RECOGNIN 1)	605981	JOHANSON-BLIZZARD SYNDROME » NASAL ALAR HYPOPLASIA, HYPOTHYROIDISM, PANCREATIC ACHYLIA, AND CONGENITAL DEAFNESS	243800		Sequencing	1300
Molecular Tests	UGT1A1 (UDP-GLYCURONOSYL TRANSFERASE)	191740	GILBERT SYNDROME	143500		1 Mutation: TA insertion in promotor	390
Molecular Tests	UGT1A1 (UDP-GLYCURONOSYL TRANSFERASE)	191740	CRIGLER-NAJJAR SYNDROME TYPE 1	218800		Sequencing	730
Molecular Tests	UGT1A1 (UDP-GLYCURONOSYL TRANSFERASE)	191740	CRIGLER-NAJJAR SYNDROME TYPE 2	606785		Sequencing	730
Molecular Tests	UMOD (UROMODULIN)	191845	FAMILIAL JUVENILE HYPERURICEMIC NEPHROPATHY, HNFJ » GOUTY NEPHROPATHY	162000		Sequencing	700
Molecular Tests	UMOD (UROMODULIN)	191845	MEDULLARY CYSTIC KIDNEY DISEASE 2, MCKD2	603860		Sequencing	700
Molecular Tests	UNC119 (UNC119, C. ELEGANS, HOMOLOG OF; HUMAN RETINAL GENE 4; HRG4)	604011	CONE-ROD DYSTROPHY	604011		Sequencing	480
Molecular Tests	UNC13D (UNC13, C. ELEGANS, HOMOLOG OF, D; MUNC13-4)	608897	HEMOPHAGOCYTIC LYMPHOHISTIOCYTOSIS, FAMILIAL, TYPE 3, FHL3	608898		Sequencing	1600
Molecular Tests	UNG (URACIL-DNA GLYCOSYLASE)	191525	IMMUNODEFICIENCY WITH HYPER-IgM, TYPE 5 » HYPER-IgM SYNDROME 5	608106		Sequencing	1350
Molecular Tests	UPF3B (UPF3, YEAST, HOMOLOG OF, B; REGULATOR OF NONSENSE TRANSCRIPTS 3B)	300298	MENTAL RETARDATION SYNDROMIC (X-LINKED), TYPE 14, MRXS14	300676		Sequencing	1500
Molecular Tests	UPF3B (UPF3, YEAST, HOMOLOG OF, B; REGULATOR OF NONSENSE TRANSCRIPTS 3B)	300298	OPITZ-KAVEGGIA SYNDROME	305450		Sequencing	1500
Molecular Tests	UPF3B (UPF3, YEAST, HOMOLOG OF, B; REGULATOR OF NONSENSE TRANSCRIPTS 3B)	300298	LUJAN-FRYNS SYNDROME	309520		Sequencing	1500
Molecular Tests	UQCRB (UBIQUINOL-CYTOCHROME c REDUCTASE-BINDING PROTEIN; UBIQUINONE-BINDING PROTEIN; UQBC)	191330	CYTOCHROME c OXIDASE DEFICIENCY » COX DEFICIENCY » COMPLEX 3, MITOCHONDRIAL RESPIRATORY CHAIN, DEFICIENCY OF	124000		Sequencing	710
Molecular Tests	UQCRQ (UBIQUINOL-CYTOCHROME c REDUCTASE, COMPLEX III SUBUNIT VII, 9.5-KD)	612080	CYTOCHROME c OXIDASE DEFICIENCY » COX DEFICIENCY » COMPLEX 3, MITOCHONDRIAL RESPIRATORY CHAIN, DEFICIENCY OF	124000		Sequencing	490
Molecular Tests	UROD (UROPORPHYRINOGEN DECARBOXYLASE)	176100	PORPHYRIA CUTANEA TARDA » PORPHYRIA, HEPATOCUTANEOUS TYPE » UROPORPHYRINOGEN DECARBOXYLASE DEFICIENCY	176100		Sequencing	830
Molecular Tests	UROS (UROPORPHYRINOGEN III SYNTHASE)	606938	PORPHYRIA, CONGENITAL ERYTHROPOIETIC » UROPORPHYRINOGEN 3 SYNTHASE, DEFICIENCY OF	263700		Sequencing	810
Molecular Tests	USH1C (HARMONIN)	605242	USHER SYNDROME, TYPE 1C, USH1C	276904		Sequencing	870
Molecular Tests	USH1C (HARMONIN)	605242	DEAFNESS, DFNB18 » DEAFNESS, (AUTOSOMAL RECESSIVE), NONSYNDROMIC SENSORINEURAL 18	602092		Sequencing	870
Molecular Tests	USH2A	608400	USHER SYNDROME, TYPE 2A, USH2A	276901		Sequencing	820
Molecular Tests	USH2A	608400	RETINITIS PIGMENTOSA, TYPE 39, RP39	608400		Sequencing	820
Molecular Tests	VAPB (VESICLE-ASSOCIATED MEMBRANE PROTEIN-ASSOCIATED PROTEIN B; VAMP-ASSOCIATED PROTEIN)	605704	AMYOTROPHIC LATERAL SCLEROSIS TYPE 8, ALS8	608627		Sequencing	780
Molecular Tests	VCAN (VERSICAN; CHONDROITIN SULFATE PROTEOGLYCAN 2; CSPG2; CHONDROITIN SULFATE PROTEOGLYCAN CORE PROTEIN, CARTILAGE)	118661	WAGNER SYNDROME, TYPE 1 » WAGNER VITREORETINAL DEGENERATION » HYALOIDEORETINAL DEGENERATION OF WAGNER » EROSIVE VITREORETINOPATHY	143200		Sequencing	1900
Molecular Tests	VCL (VINCULIN, METAVINCULIN)	193065	CARDIOMYOPATHY, DILATED, TYPE 1W	611407		Sequencing	1680
Molecular Tests	VCP (VALOSIN-CONTAINING PROTEIN)	601023	INCLUSION BODY MYOPATHY WITH EARLY-ONSET PAGET DISEASE AND FRONTOTEMPORAL DEMENTIA, IBMPFD » MUSCULAR DYSTROPHY, LIMB-GIRDLE, WITH PAGET DISEASE OF BONE » PAGETOID AMYOTROPHIC LATERAL SCLEROSIS » PAGETOID NEUROSKELETAL SYNDROME	167320		Sequencing	1100
Molecular Tests	VCP (VALOSIN-CONTAINING PROTEIN)	601023	AMYOTROPHIC LATERAL SCLEROSIS 14, WITH OR WITHOUT FRONTOTEMPORAL DEMENTIA, ALS14	613954		Sequencing	1100

Molecular Tests	VDR (VITAMIN D HORMONE RECEPTOR)	601769	VITAMIN D-DEPENDENT RICKETS, TYPE 2A » RICKETS-ALOPECIA SYNDROME » HYPOCALCEMIC VITAMIN D-RESISTANT RICKETS » RICKETS, VITAMIN D-DEPENDENT, TYPE 2A	277440		Sequencing	550
Molecular Tests	VHL	193300	CEREBELLAR HEMANGIOMA			Sequencing and Deletion-Duplication Testing	500
Molecular Tests	VHL	193300	RENAL CELL CARCINOMA 1	144700		Sequencing and Deletion-Duplication Testing	500
Molecular Tests	VHL	193300	PHEOCHROMOCYTOMA	171300		Sequencing and Deletion-Duplication Testing	500
Molecular Tests	VHL	193300	VON HIPPEL-LINDAU SYNDROME, VHL	193300		Sequencing and Deletion-Duplication Testing	500
Molecular Tests	VHL	193300	CHUVASH POLYCYTHEMIA	263400		Sequencing and Deletion-Duplication Testing	500
Molecular Tests	VIPAR (VPS33B-INTERACTING PROTEIN, APICAL-BASOLATERAL POLARITY REGULATOR; SPE39, C. ELEGANS, HOMOLOG OF; SPE39; CHROMOSOME 14 OPEN READING FRAME 133; C14ORF133)	613401	ARTHROGRYPOSIS, RENAL DYSFUNCTION, AND CHOLESTASIS, TYPE 2, ARCS2	613404		Sequencing	1160
Molecular Tests	VKORC1	608547	VITAMIN K-DEPENDENT CLOTTING FACTORS, COMBINED DEFICIENCY OF, 2	607473		Sequencing	300
Molecular Tests	VLDLR (VERY LOW DENSITY LIPOPROTEIN RECEPTOR)	192977	CEREBELLAR ATAXIA, MENTAL RETARDATION, AND DYSEQUILIBRIUM SYNDROME 1 » DYSEQUILIBRIUM SYNDROME » CEREBELLAR HYPOPLASIA, VLDLR-ASSOCIATED » CEREBELLAR ATAXIA AND MENTAL RETARDATION WITH OR WITHOUT QUADRUPEDAL LOCOMOTION 1	224050		Sequencing	1400
Molecular Tests	VPS33B (VACUOLAR PROTEIN SORTING 33, YEAST, HOMOLOG OF, B)	608552	ARTHROGRYPOSIS, RENAL DYSFUNCTION, AND CHOLESTASIS 1, ARC SYNDROME	208085		Sequencing	1160
Molecular Tests	VSX1 (VISUAL SYSTEM HOMEOBOX GENE 1, ZEBRAFISH, HOMOLOG OF)		CRANIOFACIAL ANOMALIES, EMPTY SELLA TURCICA, CORNEAL ENDOTHELIAL CHANGES, AND ABNORMAL RETINAL AND AUDITORY BIPOLAR CELLS			Sequencing	850
Molecular Tests	VSX1 (VISUAL SYSTEM HOMEOBOX GENE 1, ZEBRAFISH, HOMOLOG OF)		CORNEAL DYSTROPHY, POSTERIOR POLYMORPHOUS, 1, PPCD1	122000		Sequencing	850
Molecular Tests	VSX1 (VISUAL SYSTEM HOMEOBOX GENE 1, ZEBRAFISH, HOMOLOG OF)		KERATOCONUS 1, KTCN1	148300		Sequencing	850
Molecular Tests	VSX2 (VISUAL SYSTEM HOMEOBOX GENE 2, ZEBRAFISH, HOMOLOG OF)		MICROPHTHALMIA			Sequencing	860
Molecular Tests	VWF (VON WILLEBRAND FACTOR)	193400	VON WILLEBRAND DISEASE, TYPE NORMANDY	193400		Sequencing	1800
Molecular Tests	VWF (VON WILLEBRAND FACTOR)	193400	VON WILLEBRAND DISEASE, TYPE NORMANDY	193400		Exon 28	390
Molecular Tests	WAS (WASP)	300392	NEUTROCYTOPENIA (X-LINKED)	300299		Sequencing	750
Molecular Tests	WAS (WASP)	300392	WISKOTT-ALDRICH SYNDROME, WAS	301000		Sequencing	750
Molecular Tests	WAS (WASP)	300392	THROMBOCYTOPENIA (X-LINKED)	313900		Sequencing	750
Molecular Tests	WDR36 (WD40-REPEAT 36)	300392	GLAUCOMA, PRIMARY OPEN ANGLE, ADULT-ONSET, POAG	137760		Sequencing	800
Molecular Tests	WDR62 (WD REPEAT-CONTAINING PROTEIN 62; C19ORF14)	613583	MICROCEPHALY, PRIMARY, TYPE 2, WITH OR WITHOUT CORTICAL MALFORMATIONS (AUTOSOMAL RECESSIVE), MCPH2	604317		Sequencing	2310
Molecular Tests	WFS1 (WOLFRAMIN)	606201	WOLFRAM SYNDROME » DIABETES INSIPIDUS AND MELLITUS WITH OPTIC ATROPHY AND DEAFNESS, DIDMOAD	222300		Sequencing	400
Molecular Tests	WFS1 (WOLFRAMIN)	606201	WOLFRAM SYNDROME » DIABETES INSIPIDUS AND MELLITUS WITH OPTIC ATROPHY AND DEAFNESS, DIDMOAD	222300		Deletion-Duplication Testing	350
Molecular Tests	WFS1 (WOLFRAMIN)	606201	DEAFNESS, DFNA6 » DEAFNESS (AUTOSOMAL DOMINANT), NONSYNDROMIC SENSORINEURAL 6	600965		Sequencing	400
Molecular Tests	WFS1 (WOLFRAMIN)	606201	DEAFNESS, DFNA6 » DEAFNESS (AUTOSOMAL DOMINANT), NONSYNDROMIC SENSORINEURAL 6	600965		Deletion-Duplication Testing	350
Molecular Tests	WISP3 (WNT1-INDUCIBLE SIGNALING PATHWAY PROTEIN 3)	603400	ARTHROPATHY, PROGRESSIVE PSEUDORHEUMATOID, OF CHILDHOOD » SPONDYLOEPIPHYSEAL DYSPLASIA TARDA WITH PROGRESSIVE ARTHROPATHY » PROGRESSIVE PSEUDORHEUMATOID DYSPLASIA	208230		Sequencing	400
Molecular Tests	WNK1 (PROTEIN KINASE, LYSINE-DEFICIENT 1)	605232	PSEUDOHYPOALDOSTERONISM, TYPE 2 » HYPERPOTASSEMIA AND HYPERTENSION, FAMILIAL » HYPERTENSIVE HYPERKALEMIA, FAMILIAL » GORDON HYPERKALEMIA-HYPERTENSION SYNDROME	145260		Sequencing	Upon Request
Molecular Tests	WNK4 (PROTEIN KINASE, LYSINE-DEFICIENT 4)	601844	PSEUDOHYPOALDOSTERONISM, TYPE 2 » HYPERPOTASSEMIA AND HYPERTENSION, FAMILIAL » HYPERTENSIVE HYPERKALEMIA, FAMILIAL » GORDON HYPERKALEMIA-HYPERTENSION SYNDROME	145260		Sequencing	1050
Molecular Tests	WNT10A (WINGLESS-TYPE MMTV INTEGRATION SITE FAMILY, MEMBER 10A)	606268	SCHOPF-SCHULZ-PASSARGE SYNDROME » KERATOSIS PALMOPLANTARIS WITH CYSTIC EYELIDS, HYPODONTIA, AND HYPOTRICHOSIS	224750		Sequencing	950
Molecular Tests	WNT10A (WINGLESS-TYPE MMTV INTEGRATION SITE FAMILY, MEMBER 10A)	606268	ODONTOONYCHODERMAL DYSPLASIA	257980		Sequencing	950
Molecular Tests	WNT3 (WINGLESS-TYPE MMTV INTEGRATION SITE FAMILY, MEMBER 3)	165330	TETRA-AMELIA (AUTOSOMAL RECESSIVE)	273395		Sequencing	700
Molecular Tests	WNT7A (WINGLESS-TYPE MMTV INTEGRATION SITE FAMILY, MEMBER 7A)	601570	FIBULAR APLASIA OR HYPOPLASIA, FEMORAL BOWING AND POLY-, SYN-, AND OLIGODACTYLY » FUHRMANN SYNDROME	228930		Sequencing	600

Molecular Tests	WNT7A (WINGLESS-TYPE MMTV INTEGRATION SITE FAMILY, MEMBER 7A)	601570	ULNA AND FIBULA, ABSENCE OF, WITH SEVERE LIMB DEFICIENCY » LIMB/PELVIS-HYPOPLASIA/APLASIA SYNDROME » AL-AWADI/RAAS-ROTHSCHILD SYNDROME	276820		Sequencing	600
Molecular Tests	WT1 (WILMS TUMOR 1 GENE)	607102	FRASIER SYNDROME	136680		Sequencing	600
Molecular Tests	WT1 (WILMS TUMOR 1 GENE)	607102	FRASIER SYNDROME	136680		Deletion-Duplication Testing	350
Molecular Tests	WT1 (WILMS TUMOR 1 GENE)	607102	WILMS TUMOR 1 » NEPHROBLASTOMA	194070		Sequencing	600
Molecular Tests	WT1 (WILMS TUMOR 1 GENE)	607102	WILMS TUMOR 1 » NEPHROBLASTOMA	194070		Deletion-Duplication Testing	350
Molecular Tests	WT1 (WILMS TUMOR 1 GENE)	607102	DENYS-DRASH SYNDROME » WILMS TUMOR AND PSEUDOHERMAPHRODITISM	194080		Sequencing	600
Molecular Tests	WT1 (WILMS TUMOR 1 GENE)	607102	DENYS-DRASH SYNDROME » WILMS TUMOR AND PSEUDOHERMAPHRODITISM	194080		Deletion-Duplication Testing	350
Molecular Tests	WT1 (WILMS TUMOR 1 GENE)	607102	NEPHROTIC SYNDROME, EARLY-ONSET, WITH DIFFUSE MESANGIAL SCLEROSIS » MESANGIAL SCLEROSIS, FAMILIAL	256370		Sequencing	600
Molecular Tests	WT1 (WILMS TUMOR 1 GENE)	607102	NEPHROTIC SYNDROME, EARLY-ONSET, WITH DIFFUSE MESANGIAL SCLEROSIS » MESANGIAL SCLEROSIS, FAMILIAL	256370		Deletion-Duplication Testing	350
Molecular Tests	X-INACTIVATION STUDIES		X-LINKED DISORDERS				500
Molecular Tests	XLRS1 (RS1, RETINOSCHISIN)	312700	RETINOSCHISIS 1 (X-LINKED), JUVENILE, RS1	312700		Sequencing	480
Molecular Tests	XPA	611153	XERODERMA PIGMENTOSUM, COMPLEMENTATION GROUP A, XPA	278700		Sequencing	1780
Molecular Tests	XPC	278720	XERODERMA PIGMENTOSUM, COMPLEMENTATION GROUP C, XPC	278720		Sequencing	2400
Molecular Tests	YARS2 (TYROSYL-tRNA SYNTHETASE 2)	610957	MYOPATHY, LACTIC ACIDOSIS, AND SIDEROBLASTIC ANEMIA, TYPE 2	613561		Sequencing	580
Molecular Tests	ZAP70 (ZETA-CHAIN-ASSOCIATED PROTEIN KINASE, SYK-RELATED TYROSINE KINASE)	176947	SELECTIVE T-CELL DEFECT, IMMUNODEFICIENCY DUE TO	176947		Sequencing	1250
Molecular Tests	ZEB2 (ZINC FINGER HOMEOBOX 1B; ZFHX1B, SMAD-INTERACTING PROTEIN 1, SMADIP1, SIP1)	605802	HIRSCHSPRUNG DISEASE-MENTAL RETARDATION SYNDROME, LATE INFANTILE			Sequencing and Deletion-Duplication Testing	550
Molecular Tests	ZEB2 (ZINC FINGER HOMEOBOX 1B; ZFHX1B, SMAD-INTERACTING PROTEIN 1, SMADIP1, SIP1)	605802	MOWAT-WILSON SYNDROME » MICROCEPHALY, MENTAL RETARDATION, AND DISTINCT FACIAL FEATURES, WITH OR WITHOUT HIRSCHSPRUNG DISEASE	235730		Sequencing and Deletion-Duplication Testing	550
Molecular Tests	ZFYVE26 (ZINC FINGER FYVE DOMAIN-CONTAINING PROTEIN 26; SPASTIZIN)	612012	FAMILIAL SPASTIC PARAPLEGIA 15 (AUTOSOMAL RECESSIVE), SPG15 » SPASTIC PARAPLEGIA AND RETINAL DEGENERATION	270700		Sequencing	2800
Molecular Tests	ZIC2 (ZINC FINGER PROTEIN OF CEREBELLUM, 2)					Sequencing	750
Molecular Tests	ZIC3	300265	TRANSPOSITION OF GREAT ARTERIES (X-LINKED)			Sequencing	550
Molecular Tests	ZIC3	300265	CONGENITAL HEART DISEASE (X-LINKED)			Sequencing	550
Molecular Tests	ZIC3	300265	HETEROTAXY, VISCERAL (X-LINKED) » SITUS INVERSUS (X-LINKED)	306955		Sequencing	550
Molecular Tests	ZMPSTE24 (ZINC METALLOPROTEINASE STE24)	606480	TIGHT SKIN CONTRACTURE SYNDROME, LETHAL » HYPERKERATOSIS-CONTRACTURE SYNDROME » RESTRICTIVE DERMOPATHY, LETHAL	275210		Sequencing	745
Molecular Tests	ZMPSTE24 (ZINC METALLOPROTEINASE STE24)	606480	MANDIBULOACRAL DYSPLASIA WITH TYPE B LIPODYSTROPHY » LIPODYSTROPHY, TYPE B, ASSOCIATED WITH MANDIBULOACRAL DYSPLASIA	608612		Sequencing	745
Molecular Tests	ZNF41 (ZINC FINGER PROTEIN 41)	314995	MENTAL RETARDATION, NONSPECIFIC (X-LINKED)	314995		Sequencing	850
Molecular Tests	ZNF513 (ZINC FINGER PROTEIN 513)	613598	RETINITIS PIGMENTOSA, TYPE 58, RP58	613617		Sequencing	530
Molecular Tests	ZNF9	116955	MYOTONIC DYSTROPHY, TYPE 2 » PROXIMAL MYOTONIC MYOPATHY, PROMM » MYOTONIC MYOPATHY, PROXIMAL	602668	At least 20 microgram DNA with a concentration higher than 300 nanogram per microliter is needed	Repeat Expansion (CCTG)	450