Quest Diagnostics™

Genetic Test Menu

Realize the full potential of genetic testing

Genetic testing is on the forefront of one of the most exciting areas in personalized medicine, with results that can identify risk, help confirm difficult diagnoses, and unlock treatment options. With over 40 years of experience in molecular and genomic testing, Quest Diagnostics can help you gain deeper insight for optimal patient care. Backed by the industry's broadest menu of over 700 advanced genetic tests and our experienced genetic counselors, we help translate complex genetic content into actionable insights.

Helping you every step of the way is a large network of technical, medical, and scientific experts from multiple centers of excellence, including our renowned Nichols Institute and our Lab of the Future in Massachusetts. Our Genetic Test Menu[1] ranges from prenatal testing to hereditary cancer screening, to advanced neurological testing. We continue to make significant investments in state-of-the-art, high-resolution chromosomal microarray and next-generation sequencing technologies that deliver the high-quality results you need to care for your patients.

Genetic specialists and counselors from Quest Diagnostics, along with its subsidiary Athena Diagnostics, are always here to assist you with test selection, interpretation, and clinical consultation at **1.866.GENE.INFO (1.866.436.3463)**.

Information is also available in our Test Center at **QuestDiagnostics.com/TestCenter** or by calling **1.866.MY.QUEST (1.866.697.8378)**.

[1]Please note that we are constantly adding new tests to our menu for your use. For the most updated list, please refer to the Quest Diagnostics Test Center (QuestDiagnostics.com/TestCenter) and the Athena Diagnostics Test Catalog (AthenaDiagnostics.com/view-full-catalog).

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Part 1. Tests Offered by Quest Diagnostics

Part 2. Tests Offered by Athena Diagnostics

Multiple test codes are available. Refer to the Quest Diagnostics Nichols Institute Directory of Services or the online Test Center (QuestDiagnostics.com/TestCenter) for test information.

Part 1. Tests Offered by Quest Diagnostics

Test Code	Test Name
Cardiology	
14531	Acylcarnitine, Plasma[1]
11210(X)	Angiotensin-Converting Enzyme (*ACE*) Polymorphism (Insertion/Deletion)[1]
11118(X)	Angiotensin II Type 1 Receptor (*AGTR1*) Gene 1166A>C Polymorphism[1]
90948	Cardio IQ® 4q25-AF Risk Genotype[1]
90648	Cardio IQ® 9p21 Genotype[1]
90649	Cardio IQ® *APOE* Genotype[1]
90668	Cardio IQ® *CYP2C19* Genotype
90645	Cardio IQ® *KIF6* Genotype[1]
90553	Cardio IQ® *LPA* Aspirin Genotype[1]
90655	Cardio IQ® *LPA* Intron-25 Genotype[1]
94877	Familial Hypercholesterolemia Panel[1]
94878	Familial Hypercholesterolemia Single-Site[1]
31789	Homocysteine
34879	Methylmalonic Acid[1]
91003	Methylmalonic Acid and Homocysteine[1]
11244	Long Chain Acyl-CoA Dehydrogenase (*LCHAD*) Mutation Analysis[1]
Dermatology	
93797	*APC* Sequencing and Deletion/Duplication[1]
93939	*CDKN2A* Sequencing and Deletion/Duplication[1]
219	Delta Aminolevulinic Acid, 24-Hour Urine
6301	Delta Aminolevulinic Acid, Random Urine
35819	Organic Acids, Comprehensive, Quantitative, Urine[1]
35820	Organic Acids, Limited, Quantitative, Urine[1]
726	Porphobilinogen, Quantitative, 24-Hour Urine[1]
6329	Porphobilinogen, Quantitative, Random Urine
5519	Porphyrins, Fractionated, Plasma[1]
17198	Porphyrins, Fractionated, Quantitative and Porphobilinogen, 24-Hour Urine[1]
729	Porphyrins, Fractionated, Quantitative, 24-Hour Urine[1]
36592	Porphyrins, Fractionated, Quantitative, Random Urine[1]
10290	Porphyrins, Total, Plasma[1]
92566	*PTEN* Sequencing and Deletion/Duplication[1]
93941	*NF1* Sequencing and Deletion/Duplication[1]
92565	*STK11* Sequencing and Deletion/Duplication[1]
Endocrinology *(See also Endocrinology tests offered by Athena Diagnostics in Part 2 of the Genetic Test Menu, pages 13-15)*	
14531	Acylcarnitine, Plasma[1]
91680	CAH (21-Hydroxylase Deficiency) Common Mutations, Fetal Cells[1]
14755(X)	CAH (21-Hydroxylase Deficiency) Common Mutations[1]
16072(X)	CAH (21-Hydroxylase Deficiency) Rare Mutations[1]
92045	Familial Hyperinsulinism[1]
14615(X)	FISH, Kallmann[1]
14606(X)	FISH, *SRY/X* Centromere[1]
93942	*MEN1* Sequencing and Deletion/Duplication[1]
11369	Prader-Willi/Angelman Syndrome, DNA Methylation Analysis[1]
16053(X)	Resistance to Thyroid Hormone (RTH) Mutation Analysis[1]
93796	*RET* Sequencing and Deletion/Duplication[1]
91566	*SHOX* (GHD) DNA Sequencing and Deletion[2]
90397	Steroid Panel, 21-Hydroxylase Deficiency/Stress[1] Includes 17-hydroxyprogesterone (17180), androstenedione (17182), and cortisol (11281).
90392	Steroid Panel, Comprehensive[1] Includes androstenedione (17182), corticosterone (6547X), cortisol (11281), cortisone (37098X), deoxycorticosterone (90973), 11-deoxycortisol (30543), DHEA (19894), 18-hydroxycorticosterone (94621), 17-hydroxypregnenolone (8352), 17-hydroxyprogesterone (17180), pregnenolone (31493X), progesterone (17183), and total testosterone (15983).

Test Code	Test Name
90398	Steroid Panel, Congenital Adrenal Hyperplasia (CAH)[1] Includes androstenedione (17182), cortisol (11281), deoxycorticosterone (90973), 11-deoxycortisol (30543), DHEA (19894), 17-hydroxypregnenolone (8352), 17-hydroxyprogesterone (17180), progesterone (17183), and total testosterone (15983).
90426	Steroid Panel, PCOS/CAH Differentiation[1] Includes 11-deoxycortisol (30543), 17-hydroxyprogesterone (17180), androstenedione (17182), DHEA (19894), and total and free testosterone (36170).
90559	Very Long Chain Fatty Acids[1]
93943	*VHL* Sequencing and Deletion/Duplication[1]
Gastroenterology *(See also tests listed under Oncology)*	
14531	Acylcarnitine, Plasma[1]
767(X)	Amino Acid Analysis, LC/MS, Plasma[1]
36183(X)	Amino Acid Analysis, LC/MS, Urine[1]
219	Delta Aminolevulinic Acid, 24-Hour Urine
6301	Delta Aminolevulinic Acid, Random Urine
93791	GIvantage® Hereditary Colorectal Cancer Panel[1] Includes detection of point mutations, duplications, and/or deletions in 13 genes: *APC, BMPR1A, CDH1, EPCAM, MLH1, MSH2, MSH6, MUTYH, PMS2, PTEN, SMAD4, STK11,* and *TP53*
94053	Juvenile Polyposis Panel (*BMPR1A* and *SMAD4*)[1] Includes detection of point mutations, deletions, and duplications in the *BMPR1A* and *SMAD4* genes.
91461	Lynch Syndrome Panel[1] Includes detection of point mutations, deletions, and duplications in *MLH1, MSH2, MSH6,* and *PMS2* genes and testing for 3′-*EPCAM* deletion; performed on blood specimen.
91332 91333	Lynch Syndrome Tumor Panel, IHC Includes MLH1, MSH2, MSH6, PMS2, and microsatellite instability.
35819	Organic Acids, Comprehensive, Quantitative, Urine[1]
35820	Organic Acids, Limited, Quantitative, Urine[1]
726	Porphobilinogen, Quantitative, 24-Hour Urine[1]
6329	Porphobilinogen, Quantitative, Random Urine
5519	Porphyrins, Fractionated, Plasma[1]

Test Code	Test Name
17198	Porphyrins, Fractionated, Quantitative and Porphobilinogen, 24-Hour Urine[1]
729	Porphyrins, Fractionated, Quantitative, 24-Hour Urine[1]
36592	Porphyrins, Fractionated, Quantitative, Random Urine[1]
10290	Porphyrins, Total, Plasma[1]
Hematology and Coagulation	
16533(X)	19911A>G Mutation Analysis[1]
11175	Alpha-Globin Common Mutation Analysis[1]
16124(X)	Alpha-Globin Gene Deletion or Duplication[1]
16116(X)	Alpha-Globin Gene Sequencing
16182(X)	Beta-fibrinogen -455G>A Mutation[1]
14974	Beta-Globin Complete[1]
16346	Beta-Globin Gene Dosage Analysis[1]
17902(X)	Factor V *HR2* Allele DNA Mutation Analysis[1]
17904	Factor V (Leiden) Mutation Analysis with Reflex to *HR2* Mutation Analysis[1]
17900	Factor V (Leiden) Mutation Analysis[1]
16023(X)	Factor XI Mutation Analysis (Ashkenazi Jewish)[1]
513(X)	Fetal Hemoglobin, Whole Blood
219	Delta Aminolevulinic Acid, 24-Hour Urine
6301	Delta Aminolevulinic Acid, Random Urine
500	Glucose-6-Phosphate Dehydrogenase (G-6-PD), Quantitative
511(X)	Hemoglobin A2, Quantitative
31852(X)	Hemoglobin S, Quantitative
514	Hemoglobin, Free, Plasma
35489	Hemoglobinopathy Evaluation Includes hemoglobin A, A2, F, and any variants (eg, C, E, S), RBC count, hemoglobin, hematocrit, MCV, MCH, and RDW.
90828	Hemophilia A (Factor VIII) Inversions[1]
35079	Hereditary Hemochromatosis DNA Mutation Analysis[1]
11368	Plasminogen Activator Inhibitor-1 (PAI-1) 4G/5G[1]
726	Porphobilinogen, Quantitative, 24-Hour Urine[1]

Test Code	Test Name
6329	Porphobilinogen, Quantitative, Random Urine
5519	Porphyrins, Fractionated, Plasma[1]
17198	Porphyrins, Fractionated, Quantitative and Porphobilinogen, 24-Hour Urine[1]
729	Porphyrins, Fractionated, Quantitative, 24-Hour Urine[1]
36592	Porphyrins, Fractionated, Quantitative, Random Urine[1]
10290	Porphyrins, Total, Plasma[1]
17909	Prothrombin (Factor II) 20210G>A Mutation Analysis[1]
17364(X)	RBC Fragility - Incubated[1]
825	Sickle Cell Screen
37679(X)	Sickle Cell Screen with Reflex to Hemoglobinopathy Evaluation
17907	Thrombophilia DNA Mutation Analysis[1] Includes factor V (Leiden) and prothrombin (factor II) 20210G>A mutation analyses.
11126	Thrombophilia Mutation Analysis with Reflex to *HR2* Mutation Analysis[1] Includes factor V (Leiden) and prothrombin (factor II) 20210G>A mutation analyses with reflex to factor V *HR2* mutation analysis.
11327	Thrombophilia Screen II, Inherited[1] Includes antithrombin III activity (216), factor V (Leiden) mutation with reflex to factor V *HR2* mutation (17904), protein C activity (1777), free protein S (10170), and prothrombin (factor II) 20210G>A mutation (17909).
19837(X)	von Willebrand Disease Mutation Analysis[1]
Inborn Errors of Metabolism	
14531	Acylcarnitine, Plasma[1]
15340(X)	Alpha-1 Antitrypsin (AAT) Mutation Analysis[1]
17307(X)	Alpha-1 Antitrypsin (AAT) Quantitation and Mutation Analysis[1]
39521(X)	Alpha-1-Antitrypsin (AAT) Quantitation and Phenotype
235	Alpha-1 Antitrypsin, Quantitative
19779(X)	Amino Acid Analysis for MSUD, LC/MS, Plasma[1]
29881	Amino Acid Analysis, LC/MS, CSF[1]
767(X)	Amino Acid Analysis, LC/MS, Plasma[1]

Test Code	Test Name
36183(X)	Amino Acid Analysis, LC/MS, Urine[1]
34694	Arylsulfatase A[1]
70132(X)	Biotinidase[1]
16537(X)	Biotinidase Activity with Reflex to Mutation Analysis[1]
16526(X)	Biotinidase Deficiency Mutation Analysis[1]
14755(X)	CAH (21-Hydroxylase Deficiency) Common Mutations[1]
16072(X)	CAH (21-Hydroxylase Deficiency) Rare Mutations[1]
70107(X)	Carnitine, LC/MS/MS[1]
15948(X)	Carnitine, LC/MS/MS and Acylcarnitine[1]
90905	Canavan Disease Mutation Analysis[1]
335	Cholinesterase, Plasma
338	Cholinesterase, RBC and Plasma
37965	Cholinesterase, Serum
39481	Cholinesterase, Serum, Plasma, RBC
7961	Cholinesterase, Serum, with Dibucaine Inhibition
94600	Creatine Biosynthesis Disorders Panel, Urine[1] Includes guanidinoacetate, creatine, creatinine, and creatine/creatinine (calculated).
10947(X)	Cystine, 24-Hour Urine[1]
401(X)	Cystine, Quantitative, Random Urine[1]
219	Delta Aminolevulinic Acid, 24-Hour Urine
6301	Delta Aminolevulinic Acid, Random Urine
15538(X)	Dihydropyrimidine Dehydrogenase (*DPD*) Gene Mutation Analysis[1]
17568	Fatty Acid Panel, Comprehensive (C8-C26), Serum[1]
11254	Fatty Acid Panel, Essential[1]
94823	Fatty Acid Panel, Mitochondrial (C8-C18), Serum[1]
16613(X)	Galactosemia Mutation Analysis[1]
90907	Gaucher Disease, DNA Mutation Analysis[1]
500	Glucose-6-Phosphate Dehydrogenase, Quantitative
90915	Glycogen Storage Disease Type Ia Mutation Analysis (Ashkenazi Jewish)[1]

Test Code	Test Name
16612	*HEXA* Mutation Analysis, Gene Sequencing[1] Includes sequencing of the entire coding region, the intron-exon splice sites, and the promoter region of the *HEXA* gene. Consider common mutation testing (Tay-Sachs Disease Mutation Analysis) prior to, or concurrently with, this test. Hexosaminidase testing should also be considered before or concurrently with this test.
31789	Homocysteine
523(X)	5-Hydroxyindoleacetic Acid (5-HIAA), 24-Hour Urine[1]
39625(X)	5-Hydroxyindoleacetic Acid (5-HIAA), 24-Hour Urine, with Creatinine[1]
1648(X)	5-Hydroxyindoleacetic Acid (5-HIAA), Random Urine[1]
11244	Long Chain Acyl-CoA Dehydrogenase (*LCHAD*) Mutation Analysis[1]
90909	Maple Syrup Urine Disease (MSUD) Mutation Analysis (Ashkenazi Jewish)[1]
91284	Medium Chain Acyl-CoA Dehydrogenase (MCAD) Gene Sequencing[1]
11176(X)	Medium Chain Acyl-CoA Dehydrogenase (MCAD) Mutation Analysis[1]
34879	Methylmalonic Acid[1]
91003	Methylmalonic Acid and Homocysteine[1]
91032	Methylmalonic Acid, GC/MS/MS, Urine[1]
90899	Mucolipidosis Type IV Mutation Analysis[1]
90893	Niemann-Pick Disease Mutation Analysis[1]
35819	Organic Acids, Comprehensive, Quantitative, Urine[1]
35820	Organic Acids, Limited, Quantitative, Urine[1]
37356	Phenylalanine[1]
26336	Phenylalanine and Tyrosine[1]
16152(X)	Phenylketonuria (PKU) Mutation Analysis[1]
726	Porphobilinogen, Quantitative, 24-Hour Urine[1]
6329	Porphobilinogen, Quantitative, Random Urine
5519	Porphyrins, Fractionated, Plasma[1]
17198	Porphyrins, Fractionated, Quantitative and Porphobilinogen, 24-Hour Urine[1]
729	Porphyrins, Fractionated, Quantitative, 24-Hour Urine[1]
36592	Porphyrins, Fractionated, Quantitative, Random Urine[1]
10290	Porphyrins, Total, Plasma[1]
90392	Steroid Panel, Comprehensive[1] Includes androstenedione (17182), corticosterone (6547X), cortisol (11281), cortisone (37098X), deoxycorticosterone (90973), 11-deoxycortisol (30543), DHEA (19894), 18-hydroxycorticosterone (94621), 17-hydroxypregnenolone (8352), 17-hydroxyprogesterone (17180), pregnenolone (31493X), progesterone (17183), and total testosterone (15983).
90398	Steroid Panel, Congenital Adrenal Hyperplasia (CAH)[1] Includes androstenedione (17182), cortisol (11281), deoxycorticosterone (90973), 11-deoxycortisol (30543), DHEA (19894), 17-hydroxypregnenolone (8352), 17-hydroxyprogesterone (17180), progesterone (17183), and total testosterone (15983).
90397	Steroid Panel, 21-Hydroxylase Deficiency/ Stress[1] Includes 17-hydroxyprogesterone (17180), androstenedione (17182), and cortisol (11281).
90426	Steroid Panel, PCOS/CAH Differentiation[1] Includes 11-deoxycortisol (30543), 17-hydroxyprogesterone (17180), androstenedione (17182), DHEA (19894), and total and free testosterone (36170).
90903	Tay-Sachs Disease Mutation Analysis[1] Includes *HEXA* gene analysis for 1278insTATC, delta7.6kb, G269S, IVS9+1G>A, IVS12+1G>C, and R178H mutations and the R247W pseudodeficiency allele. Consider hexosaminidase testing before or concurrently with this test.
959	Tryptophan, LC/MS[1]
902	Tyrosine[1]
90559	Very Long Chain Fatty Acids[1]
39517(X)	VMA (Vanillylmandelic Acid), 24-Hour Urine[1]
934(X)	VMA (Vanillylmandelic Acid), 24-Hour Urine without Creatinine[1]
1710	VMA (Vanillylmandelic Acid), Random Urine[1]
Intellectual Disability/Autism *(See also Neurology—Intellectual Disability/Autism tests offered by Athena Diagnostics in Part 2 of the Genetic Test Menu, page 18)*	
14531	Acylcarnitine, Plasma[1]
29881	Amino Acid Analysis, LC/MS, CSF[1]
767(X)	Amino Acid Analysis, LC/MS, Plasma[1]

Test Code	Test Name
36183(X)	Amino Acid Analysis, LC/MS, Urine[1]
70132(X)	Biotinidase[1]
16537(X)	Biotinidase Activity with Reflex to Mutation Analysis[1]
16526(X)	Biotinidase Deficiency Mutation Analysis[1]
16478	Chromosomal Microarray, Postnatal, ClariSure® Oligo-SNP
14596	Chromosome Analysis, Blood
18980	Chromosome Analysis, Blood with Reflex to Postnatal ClariSure® Oligo-SNP Array
10708(X)	Chromosome Analysis, Follow-up
14595	Chromosome Analysis, High Resolution
18983	Chromosome Analysis, High Resolution with reflex to Postnatal ClariSure® Oligo-SNP Array
14597(X)	Chromosome Analysis, Mosaicism
16843	Chromosome Analysis, Neonatal Blood
94600	Creatine Biosynthesis Disorders Panel, Urine[1] Includes guanidinoacetate, creatine, creatinine, and creatine/creatinine (calculated).
14608(X)	FISH, Angelman[1]
37343(X)	FISH, Chromosome-Specific Probe[1] Choose one of the following: chromosome-specific (1-22, X and Y) centromere or chromosome-specific (1-22, X and Y) painting.
40047	FISH, Chromosome-Specific Probe (x1), Follow-up Study[1]
14614(X)	FISH, Cri du chat[1]
14610(X)	FISH, DiGeorge, Velocardiofacial (VCFS)[1]
19045(X)	FISH, Duplication 15q11q13[1]
16672(X)	FISH, Duplication 22q11.2[1]
14612(X)	FISH, Miller-Dieker[1]
36053	FISH, Neonatal Screen[1] Includes chromosomes 13, 18, 21, X, and Y.
14605(X)	FISH, Prader Willi[1]
14611(X)	FISH, Smith-Magenis[1]
14609(X)	FISH, Williams[1]
14613(X)	FISH, Wolf-Hirschhorn[1]
16300	Fragile X DNA Analysis, Fetus[1]

Test Code	Test Name
16612	*HEXA* Mutation Analysis, Gene Sequencing[1] Includes sequencing of the entire coding region, the intron-exon splice sites, and the promoter region of the *HEXA* gene. Consider common mutation testing (Tay-Sachs Disease Mutation Analysis) prior to, or concurrently with, this test. Hexosaminidase testing should also be considered before or concurrently with this test.
90899	Mucolipidosis Type IV Mutation Analysis[1]
35819	Organic Acids, Comprehensive, Quantitative, Urine[1]
35820	Organic Acids, Limited, Quantitative, Urine[1]
37356	Phenylalanine[1]
26336	Phenylalanine and Tyrosine[1]
16152(X)	Phenylketonuria (PKU) Mutation Analysis[1] Phenylalanine testing should be considered before or concurrently with this test.
11369	Prader-Willi/Angelman Syndrome, DNA Methylation Analysis[1]
92566	*PTEN* Sequencing and Deletion/Duplication[1]
15088(X)	Rett Syndrome Mutation Analysis[1]
16662	Rett Syndrome Rearrangement (Deletion or Duplication)[1]
90559	Very Long Chain Fatty Acids[1]
16326	XSense®, Fragile X with Reflex and Chromosome Analysis, Blood[1] Includes a reflex to fragile X methylation analysis when PCR result is either not normal or gray zone.
16313	XSense®, Fragile X with Reflex[1] Includes a reflex to fragile X methylation analysis when PCR result is either not normal or gray zone.
Neurology *(See also Neurology tests offered by Athena Diagnostics in Part 2 of the Genetic Test Menu, pages 15-24)*	
14531	Acylcarnitine, Plasma[1]
10642	ADmark® *APOE* Genotype Analysis and Interpretation (Symptomatic)[2] Includes detection of *APOE2, E3*, and *E4* alleles.
29881	Amino Acid Analysis, LC/MS, CSF[1]
767(X)	Amino Acid Analysis, LC/MS, Plasma[1]
36183(X)	Amino Acid Analysis, LC/MS, Urine[1]
70132(X)	Biotinidase[1]
16537(X)	Biotinidase Activity with Reflex to Mutation Analysis[1]

Test Code	Test Name
16526(X)	Biotinidase Deficiency Mutation Analysis[1]
90905	Canavan Disease Mutation Analysis[1]
92435	Connexin 26 (GJB2) DNA Sequencing Test[2]
94009	Connexin 30 (GJB6) DNA Test[2]
90482	Connexin Related Deafness Evaluation[2]
94600	Creatine Biosynthesis Disorders Panel, Urine[1] Includes guanidinoacetate, creatine, creatinine, and creatine/creatinine (calculated).
14678	Dystonia (DYT1) DNA Test[1]
92843	*GCH1* DNA Sequencing Test (DYT5A)[1]
16612	*HEXA* Mutation Analysis, Gene Sequencing[1] Includes sequencing of the entire coding region, the intron-exon splice sites, and the promoter region of the *HEXA* gene. Consider common mutation testing (Tay-Sachs Disease Mutation Analysis) prior to, or concurrently with, this test. Hexosaminidase testing should also be considered before or concurrently with this test.
31789	Homocysteine
10247(X)	Huntington Disease Mutation Analysis[1]
92955	Isolated Dystonia Evaluation[1]
11244	Long Chain Acyl-CoA Dehydrogenase (*LCHAD*) Mutation Analysis[1]
91284	Medium Chain Acyl-CoA Dehydrogenase (MCAD) Gene Sequencing[1]
11176(X)	Medium Chain Acyl-CoA Dehydrogenase (MCAD) Mutation Analysis[1]
34879	Methylmalonic Acid[1]
91003	Methylmalonic Acid and Homocysteine[1]
91032	Methylmalonic Acid, GC/MS/MS, Urine[1]
93941	*NF1* Sequencing and Deletion/Duplication[1]
1175	*NOTCH3* (CADASIL) Sequencing Test[2]
35819	Organic Acids, Comprehensive, Quantitative, Urine[1]
35820	Organic Acids, Limited, Quantitative, Urine[1]
92369	*PMP22* DNA Sequencing Test[2]
16869	SMA Diagnostic Test
90903	Tay-Sachs Disease Mutation Analysis[1] Includes *HEXA* gene analysis for 1278insTATC, delta7.6kb, G269S, IVS9+1G>A, IVS12+1G>C, and R178H mutations and the R247W pseudodeficiency allele. Consider hexosaminidase testing before or concurrently with this test.

Test Code	Test Name
92915	*THAP1* DNA Sequencing Test (DYT6)[1]
90559	Very Long Chain Fatty Acids[1]
Oncology	
93797	*APC* Sequencing and Deletion/Duplication[1]
16767	*BRAF* Mutation Analysis[1]
91864	BRCAvantage®, Ashkenazi Jewish Screen[1] Includes detection of the 3 HBOC syndrome founder mutations (c.68_69delAG [185delAG, 187delAG], c.5266dupC [5382insC, 5385insC], and c.5946delT [6174delT]).
92140	BRCAvantage®, Ashkenazi Jewish Screen w/ Reflex BRCAvantage®, Comprehensive[1] Includes test code 91864; test code 91863 added with additional charge and CPT code when none of the 3 founder mutations are detected.
91863	BRCAvantage®, Comprehensive[1] Includes detection of point mutations, deletions, and duplications in the *BRCA1* and *BRCA2* genes.
91866	BRCAvantage®, Rearrangements[1] Includes detection of deletions and duplications in the *BRCA1* and *BRCA2* genes.
92568	*CDH1* Sequencing and Deletion/Duplication[1]
93939	*CDKN2A* Sequencing and Deletion/ Duplication[1]
93940	*CHEK2* Sequencing and Deletion/Duplication[1]
93791	GIvantage® Hereditary Colorectal Cancer Panel[1] Includes detection of point mutations, duplications, and/or deletions in 13 genes: *APC, BMPR1A, CDH1, EPCAM, MLH1, MSH2, MSH6, MUTYH, PMS2, PTEN, SMAD4, STK11,* and *TP53*.
94053	Juvenile Polyposis Panel (*BMPR1A* and *SMAD4*)[1] Includes detection of point mutations, deletions, and duplications in the *BMPR1A* and *SMAD4* genes.
91461	Lynch Syndrome Panel[1] Includes detection of point mutations, deletions, and duplications in *MLH1, MSH2, MSH6*, and *PMS2* genes and testing for *3′-EPCAM* deletion; performed on blood specimen.
91332 91333	Lynch Syndrome Tumor Panel, IHC Includes MLH1, MSH2, MSH6, PMS2, and microsatellite instability.
91460	Lynch Syndrome, *MLH1* Sequencing and Deletion/Duplication[1]
91471	Lynch Syndrome, *MSH2* Sequencing and Deletion/Duplication (Including *EPCAM*)[1]

Test Code	Test Name
91458	Lynch Syndrome, *MSH6* Sequencing and Deletion/Duplication[1]
91457	Lynch Syndrome, *PMS2* Sequencing and Deletion/Duplication[1]
93942	*MEN1* Sequencing and Deletion/Duplication[1]
14989(X)	Microsatellite Instability (MSI), HNPCC[1]
70196(X)	MLH1, IHC with Interpretation
16967	MLH1, IHC without Interpretation
70197(X)	MSH2, IHC with Interpretation
16971	MSH2, IHC without Interpretation
16938	MSH6, IHC with Interpretation
16252	MSH6, IHC without Interpretation
93944	*MUTYH* Sequencing and Deletion/Duplication[1]
93768	MyVantage® Hereditary Comprehensive Cancer Panel[1] Includes detection of point mutations, duplications, and/or deletions in 34 genes: *APC, ATM, BARD1, BMPR1A, BRCA1, BRCA2, BRIP1, CDH1, CDK4, CDKN2A, CHEK2, EPCAM, MEN1, MLH1, MSH2, MSH6, MUTYH, NBN, NF1, PALB2, PMS2, POLD1, POLE, PTEN, RAD51C, RAD51D, RET, SDHB, SDHC, SDHD, SMAD4, STK11, TP53,* and *VHL*.
93945	MyVantage®, Single Site[1] Includes detection of a single, familial mutation in any of the 34 genes included on the MyVantage® panel. Report of results for family member with known mutation must be provided.
93941	*NF1* Sequencing and Deletion/Duplication[1]
92571	*PALB2* Sequencing and Deletion/Duplication[1]
16997	PMS2, IHC with Interpretation
16254	PMS2, IHC without Interpretation
92566	*PTEN* Sequencing and Deletion/Duplication[1]
93796	*RET* Sequencing and Deletion/Duplication[1]
92565	*STK11* Sequencing and Deletion/Duplication[1]
92560	*TP53* Sequencing and Deletion/Duplication[1]
93943	*VHL* Sequencing and Deletion/Duplication[1]
Pharmacogenomics	
16924	AccuType® CP, Clopidogrel *CYP2C19* Genotype[1]
90251	AccuType® IL28B[1]
91416	AccuType® Ribavirin (ITPA)[1]
16160(X)	AccuType® Warfarin[1] Includes detection of variants in the *VKORC1* and *CYP2C9* genes.
16176(X)	Beta2-Adrenergic Receptor Mutations[1]
11294(X)	Cytochrome P450 2C9 Genotype[1]
10490	Cytochrome P450 2D6 Genotype[1]
15538(X)	Dihydropyrimidine Dehydrogenase (*DPD*) Gene Mutation Analysis[1]
19774	HLA-B*5701 Typing
93932	HLA-B*58:01 Typing[1]
91597	Pain Management, *CYP450 3A4/3A5* Genotype, Qualitative[1]
91617	Pain Management, *CYP450 3A4* Genotype, Qualitative[1]
91618	Pain Management, *CYP450 3A5* Genotype, Qualitative[1]
18946	Pain Management, *CYP450 2D6/2C19* Genotype, Qualitative[1]
37742(Z)	Thiopurine S-Methyltransferase (*TPMT*) Genotype[1]
17813(X)	*UGT1A1* Gene Polymorphism (TA Repeat)[1]
16959	*VEGF* Polymorphism Analysis[1]
Prenatal Diagnosis, Screening, and Reproductive Genetics *(Syndrome-specific FISH tests that may be appropriate for postnatal testing are listed in the Intellectual Disability/Autism section in Part 1 of the Genetic Test Menu, pages 6-7)*	
Fetal Aneuploidy Screening	
16145	First Trimester Screen, hCG[3] Includes pregnancy-associated plasma protein-A (PAPP-A), hCG, nuchal translucency (NT), Down syndrome risk, and interpretation.
16020	First Trimester Screen, Hyperglycosylated hCG (h-hCG)[3] Includes PAPP-A, h-hCG, NT, Down syndrome risk, and interpretation.
16148	Integrated Screen, Part 1[1] Includes PAPP-A and NT.
16150	Integrated Screen, Part 2[3] Includes AFP; hCG; unconjugated estriol (uE3); inhibin A; PAPP-A and NT from part 1; neural tube defect (NTD), Down syndrome, and trisomy 18 risk; and interpretation.

Test Code	Test Name
5059	Maternal Serum AFP Includes AFP, NTD risk, and interpretation.
15934	Penta Screen[1] Includes AFP; hCG; uE3; inhibin A; h-hCG; NTD, Down syndrome, and trisomy 18 risk; and interpretation.
92777	QNatal® Advanced[1] Includes cfDNA noninvasive prenatal screen for fetal chromosomal abnormalities: trisomy 21, 18, 13, as well as sex chromosome abnormalities; microdeletion panel (optional); and reporting of fetal sex (optional).
30294	Quad Screen Includes AFP; hCG; uE3; inhibin A; NTD, Down syndrome, and trisomy 18 risk; and interpretation.
16131	Sequential Integrated Screen, Part 1[3] Includes PAPP-A; hCG; NT; Down syndrome and trisomy 18 risk (if screen positive); and interpretation.
16133	Sequential Integrated Screen, Part 2[3] Includes AFP; hCG; uE3; inhibin A; PAPP-A and NT from part 1; NTD, Down syndrome, and trisomy 18 risk; and interpretation.
16165	Serum Integrated Screen, Part 1[1] Includes PAPP-A.
16167	Serum Integrated Screen, Part 2[3] Includes AFP; hCG; uE3; inhibin A; PAPP-A from part 1; NTD, Down syndrome, and trisomy 18 risk; and interpretation.
16463	Stepwise, Part 1[3] Includes PAPP-A; hCG; NT; Down syndrome and trisomy 18 risk; and interpretation.
16465	Stepwise, Part 2[3] Includes AFP; hCG; uE3; inhibin A; PAPP-A and NT from part 1; NTD, Down syndrome, and trisomy 18 risk; and interpretation.
7292	Triple Screen Includes AFP; hCG; uE3; NTD, Down syndrome, and trisomy 18 risk; and interpretation.
Other	
4929(X)	Acetylcholinesterase[1]
16061(X)	Achondroplasia Mutation Analysis[1]
91615	Alpha-Fetoprotein and Acetylcholinesterase, Amniotic Fluid with Reflex to HbF[1]
232(Z)	Alpha-Fetoprotein, Amniotic Fluid with Reflex to AchE and Fetal Hgb[1]
11175	Alpha-Globin Common Mutation Analysis[1]
91711	Alpha-Globin Common Mutation Analysis, Fetus[1]
16124(X)	Alpha-Globin Gene Deletion or Duplication[1]
16116(X)	Alpha-Globin Gene Sequencing
90891	Ashkenazi Jewish Panel (11 Tests)[1] Includes mutations associated with Bloom syndrome, Canavan disease, cystic fibrosis, Gaucher disease, glycogen storage disease, familial dysautonomia, Fanconi anemia, MSUD Jewish mutation, mucolipodosis type IV, Niemann-Pick disease, and Tay-Sachs disease.
90994	Ashkenazi Jewish Panel (4 Tests)[1] Includes mutations associated with Canavan disease, cystic fibrosis, familial dysautonomia, and Tay-Sachs disease.
14974	Beta-Globin Complete[1]
91709	Beta-Globin Complete™, Fetus
16346	Beta-Globin Gene Dosage Analysis[1]
90872	Bloom Syndrome DNA Mutation Analysis[1]
90905	Canavan Disease Mutation Analysis[1]
10225(X)	Cell Culture for Possible Additional Prenatal Studies
15053(X)	*CFTR* Intron 8 Poly-T Analysis[1]
92068	CFvantage® Cystic Fibrosis Expanded Screen[1]
90929	Chromosomal Microarray, POC, ClariSure® Oligo-SNP Includes detection of genomic alterations that may be associated with fetal miscarriage by assaying products of conception.
16478	Chromosomal Microarray, Postnatal, ClariSure® Oligo-SNP
90927	Chromosomal Microarray, Prenatal, ClariSure® Oligo-SNP
14590(X)	Chromosome Analysis, Amniotic Fluid
14591(Z)	Chromosome Analysis and AFP with Reflex to AchE, Fetal Hgb, Amniotic Fluid[1]
14596	Chromosome Analysis, Blood
14592(X)	Chromosome Analysis, Chorionic Villus Sample
17455(X)	Chromosome Analysis, Fanconi Anemia, Prenatal
10708(X)	Chromosome Analysis, Follow-up
14595	Chromosome Analysis, High Resolution
14597(X)	Chromosome Analysis, Mosaicism

Test Code	Test Name
16843	Chromosome Analysis, Neonatal Blood
14599(Z)	Chromosome Analysis, Sister Chromatid Exchange
14593(X)	Chromosome Analysis, Tissue
91126	Chromosome Analysis, Tissue w/ Reflex to Microarray, ClariSure® Oligo-SNP
14598(Z)	Chromosomes, DEB Assay for Fanconi Anemia
10917	Cystic Fibrosis Complete Rare Mutation Analysis, Entire Gene Sequence[1]
15335(X)	Cystic Fibrosis D1152H Mutation Analysis[1]
10226(X)	Cystic Fibrosis DNA Analysis, Fetus[1]
16080(X)	Cystic Fibrosis Gene Deletion or Duplication[1]
17726	Cystic Fibrosis Mutation Screen with Reflex to CF Complete™ (Clinics Only)[1]
10913(X)	Cystic Fibrosis Rare Mutation Analysis, One Exon[1]
10915(X)	Cystic Fibrosis Rare Mutation Analysis, Two Exon[1]
10458	Cystic Fibrosis Screen[1]
92046	Dihydrolipoamide Dehydrogenase (DLD) Deficiency[1]
90912	Familial Dysautonomia Mutation Analysis[1]
92045	Familial Hyperinsulinism[1]
16141(X)	Familial Mediterranean Fever Mutation Analysis[1]
90897	Fanconi Anemia DNA Mutation Analysis[1]
36208	Fetal Hemoglobin, Amniotic Fluid
14604(X)	FISH, Prenatal Screen[1] Includes enumeration of chromosomes 13, 18, 21, X, and Y.
14607(X)	FISH, X-Linked Ichthyosis Steroid Sulfatase Deficiency[1]
16300	Fragile X DNA Analysis, Fetus[1]
90907	Gaucher Disease, DNA Mutation Analysis[1]
90915	Glycogen Storage Disease Type Ia Mutation Analysis (Ashkenazi Jewish)[1]

Test Code	Test Name
16612	*HEXA* Mutation Analysis, Gene Sequencing[1] Includes sequencing of the entire coding region, the intron-exon splice sites, and the promoter region of the *HEXA* gene. Common mutation testing (Tay-Sachs Disease Mutation Analysis) must be performed prior to, or concurrently with, this test. Hexosaminidase enzyme carrier screening should also be considered before or concurrently with this test.
92050	Joubert Syndrome[1]
90909	Maple Syrup Disease (MSUD) Mutation Analysis (Ashkenazi Jewish)[1]
10262	Maternal Cell Contamination Study, STR Analysis[3]
90899	Mucolipidosis Type IV Mutation Analysis[1]
92055	Nemaline Myopathy[1]
90893	Niemann-Pick Disease Mutation Analysis[1]
16152(X)	Phenylketonuria (PKU) Mutation Analysis[1]
93349	Prenatal Carrier Panel (CFvantage®, Fragile X, SMA)[1]
90949	Prenatal Carrier Screen (CF, Fragile X, SMA)[1]
94372	QHerit® Expanded Carrier Screen Analyzes 24 genes associated with 22 diseases, including: alpha thalassemia, beta thalassemia (including sickle cell anemia), Bloom syndrome, Canavan disease, cystic fibrosis (CFvantage®), dihydrolipoamide dehydrogenase deficiency, familial dysautonomia, familial hyperinsulinism, Fanconi anemia type C, fragile X syndrome, Gaucher disease, glycogen storage disease type IA, Joubert syndrome 2, maple syrup urine disease, mucolipidosis type IV, nemaline myopathy, Niemann-Pick disease type A, spinal muscular atrophy, Tay-Sachs disease, Usher syndrome type IF, Usher syndrome type IIIA, and Walker-Warburg syndrome.
26382(X)	Sickle Cell Anemia, DNA Probe Analysis, Fetus[1]
18041	SMA Carrier Screen[1]
16869	SMA Diagnostic Test
90903	Tay-Sachs Disease Mutation Analysis[1] Includes *HEXA* gene analysis for 1278insTATC, delta7.6kb, G269S, IVS9+1G>A, IVS12+1G>C, and R178H mutations and the R247W pseudodeficiency allele. Consider hexosaminidase enzyme carrier screening before, or concurrently with, this test.
10556(X)	Twin Zygosity[3]
92047	Usher Syndrome Type IF[1]
92048	Usher Syndrome Type IIIA[1]

Test Code	Test Name
92051	Walker-Warburg Syndrome[1]
16326	XSense®, Fragile X with Reflex and Chromosome Analysis, Blood[1] Includes a reflex to fragile X methylation analysis when PCR result is either not normal or gray zone.
16313	XSense®, Fragile X with Reflex[1] Includes a reflex to fragile X methylation analysis when PCR result is either not normal or gray zone.
14679	Y Chromosome Microdeletion, DNA Analysis[3]

Test Code	Test Name
Other Genetic Tests	
15053(X)	*CFTR* Intron 8 Poly-T Analysis[1]
38956(X)	*CKR-5* Gene, DNA Mutation Analysis[1]
16155(X)	Macular Degeneration Mutation Analysis[1]
17911	Methylenetetrahydrofolate Reductase (*MTHFR*), DNA Mutation Analysis[1]

[1]This test was developed and its analytical performance characteristics have been determined by Quest Diagnostics. It has not been cleared or approved by the U.S. Food and Drug Administration. This assay has been validated pursuant to the CLIA regulations and is used for clinical purposes.

[2]This test was developed and its analytical performance characteristics have been determined by Athena Diagnostics. It has not been cleared or approved by the U.S. Food and Drug Administration. This assay has been validated pursuant to the CLIA regulations and is used for clinical purposes.

[3]This test was performed using a kit that has not been cleared or approved by the FDA. The analytical performance characteristics of this test have been determined by Quest Diagnostics. This test should not be used for diagnosis without confirmation by other medically established means.

Reflex tests are performed at an additional charge and are associated with an additional CPT code.

Panel components may be ordered separately.

Polymerase chain reaction (PCR) is performed pursuant to a license agreement with Roche Molecular Systems, Inc.

Multiple test codes are available. Refer to the Quest Diagnostics Directory of Services or the online Test Center (QuestDiagnostics.com/TestCenter) for test information.

Additional assistance in test selection is available from Quest Diagnostics Genomics Client Services by calling 1.866.GENE.INFO (1.866.436.3463).

Part 2. Tests Offered by Athena Diagnostics

Test Code	Test Name
Endocrinology	
827	*ABCC8* (CHI) DNA Sequencing Test
876	*ABCC8* (NDM) DNA Sequencing Test
815	*ABCD1* (Adrenoleukodystrophy) DNA Sequencing Test
6108	*ABCD1* (Neurology) DNA Sequencing Test
462	Anosmic Kallmann/IHH Evaluation Includes sequencing of the *FGF8*, *FGFR1*, *GNRHR*, *KAL1*, *KISS1R*, *PROK2*, and *PROKR2* genes.
852	*AQP2* (Nephrogenic Diabetes Insipidus) DNA Sequencing Test
812	Autoimmune Polyglandular Syndrome (*AIRE*) Evaluation
887	Bardet-Biedl Syndrome Evaluation Includes sequencing of the *BBS1*, *BBS2*, and *BBS10* genes.
871	*BBS1* (BBS) DNA Sequencing Test
886	*BBS10* (BBS) DNA Sequencing Test
872	*BBS2* (BBS) DNA Sequencing Test
837	*CEL (MODY8)* Mutation Analysis
461	*CHD7* (Kallmann/IHH) DNA Sequencing Test
861	*COL1A1* (OI) DNA Sequencing Test
862	*COL1A2* (OI) DNA Sequencing Test
865	Combined Pituitary Hormone Deficiency Evaluation Includes sequencing of the *POU1F1* and *PROP1* genes.
679	Complete Kallmann/IHH Evaluation Includes sequencing of the *CHD7*, *FGF8*, *FGFR1*, *GNRHR*, *GNRH1*, *KAL1*, *KISS1R*, *PROK2*, *PROKR2*, and *TACR3* genes.
879	Congenital Adrenal Hyperplasia (CAH) Evaluation Includes sequencing and deletion analysis of *CYP21A2* and sequencing of *CYP11B1*.
819	Congenital Hyperinsulinism Evaluation Includes sequencing of the *ABCC8*, *GCK*, *GLUD1*, and *KCNJ11* genes.
875	*CYP11B1* (CAH) DNA Sequencing Test Includes sequencing and analysis for the 30kb deletion.
774	*CYP11B1* DNA Sequencing Test Includes sequencing of the *CYP11B1* gene.
877	*CYP17A1* DNA Sequencing Test
880	*CYP21A2* (CAH) Evaluation Includes sequencing and analysis for the 30kb deletion.
1180	*CYP21A2* Deletion Test
1188	*CYP21A2* DNA Sequencing Test
883	Early Onset Obesity (*LEPR*) DNA Sequencing Test
640	Early Onset Obesity (*MC4R*) DNA Sequencing Test
884	Early-Onset Obesity Evaluation Includes sequencing of the *LEPR* and *MC4R* genes.
881	Endocrine Hypertension (*HSD11B2*) Evaluation Includes sequencing of the *HSD11B2* gene.
185	Familial DNA Sequence Evaluation Includes analysis for a familial mutation only.
829	Familial Hypocalciuric Hypercalcemia (*CASR*) DNA Sequencing Test
856	*FGF23* (Hypophosphatemic Rickets) DNA Sequencing Test
195	*FGF8* DNA Sequencing Test
196	*FGFR1* DNA Sequencing Test
823	*GCK* (CHI) DNA Sequencing Test
803	*GCK (MODY2)* DNA Sequencing and Deletion Test
842	*GCK* (NDM) DNA Sequencing Test
866	*GH1* (GHD) DNA Sequencing Test
867	*GHR* DNA Sequencing Test
868	*GHRHR* (GHD) DNA Sequencing Test
822	*GLUD1* (CHI) DNA Sequencing Test
343	*GNRH1* DNA Sequencing Test
279	*GNRHR* DNA Sequencing Test
848	Growth Hormone Deficiency Evaluation Includes sequencing of the *GH1*, *GHRHR*, and *SHOX* genes and detection of deletions in the *SHOX* gene.

Test Code	Test Name
802	*HNF4A (MODY1)* DNA Sequencing and Deletion Test
775	*HSD11B2* DNA Sequencing Test
878	*HSD3B2* DNA Sequencing Test
857	Hypophosphatemic Rickets Evaluation Includes sequencing of the *FGF23* and *PHEX* genes.
853	*INS* (NDM) DNA Sequencing Test
834	*IPF1* (*MODY4*) DNA Sequencing Test
841	*IPF1* (NDM) DNA Sequencing Test
173	*KAL1* DNA Sequencing Test
826	*KCNJ11* (CHI) DNA Sequencing Test
843	*KCNJ11* (NDM) DNA Sequencing Test
364	*KISS1R* DNA Sequencing Test
664	*KRAS* DNA Sequencing Test
658	*KRAS/RAF1/SOS1* DNA Sequencing Evaluation
747	Liddle's Syndrome Evaluation Includes sequencing of the *SCNN1B* and *SCNN1G* genes.
874	Lipoid CAH (*STAR*) Evaluation Includes sequencing of the *STAR* gene.
821	*LRP5* Idiopathic Osteoporosis (IOP) DNA Sequencing Test
811	*LRP5* (OPPG) DNA Sequencing
817	Male Precocious Puberty (LHCGR) DNA Sequencing Test
818	MEN1 DNA Sequencing Test
813	MEN2 (*RET*) DNA Sequencing Test
885	Monogenic Diabetes (MODY) Five Gene Evaluation *(GCK,HNF1A,HNF1B,HNF4A,IPF1)*
8800	Monogenic Diabetes (MODY) Four Gene Evaluation *(GCK,HNF1A, HNF1B, HNF4A)*
8801	Monogenic Diabetes (MODY) Three Gene Evaluation (*GCK,HNF1A, HNF1B*)
8802	Monogenic Diabetes (MODY) Two Gene Evaluation (*GCK,HNF1A*)
749	Monogenic Hypertension Evaluation Includes sequencing of the *CYP11B1, HSD11B2, SCNN1B,* and *SCNN1G* genes.
882	Neonatal Diabetes Mellitus Evaluation Includes sequencing of the *ABCC8, GCK, INS, IPF1,* and *KCNJ11* genes.
851	Nephrogenic Diabetes Insipidus (*AVPR2*) DNA Sequencing Test
854	Nephrogenic Diabetes Insipidus Evaluation Includes sequencing of the *AQP2* and *AVPR2* genes.
846	Noonan Syndrome (*PTPN11*) DNA Sequencing Test
667	Normosmic Kallmann/IHH Evaluation Includes sequencing of the *FGFR1, GNRHR, GNRH1, KISS1R, PROK2, PROKR2,* and *TACR3* genes.
814	*NR0B1* (Adrenal Hypoplasia Congenita) DNA Sequencing Test
860	Osteogenesis Imperfecta Evaluation Includes sequencing of the *COL1A1* and *COL1A2* genes.
889	Pheochromocytoma Evaluation Includes sequencing of the *RET, SDHB,* and *VHL* genes.
855	*PHEX* (Hypophosphatemic Rickets) DNA Sequencing Test
864	*POU1F1* (CPHD) DNA Sequencing Test
816	Primary Adrenal Insufficiency Evaluation Includes sequencing of the *ABCD1, AIRE*, and *NR0B1* genes.
175	*PROK2* DNA Sequencing Test
180	*PROKR2* DNA Sequencing Test
863	*PROP1* (CPHD) DNA Sequencing Test
748	Pseudohypoaldosteronism Type 1 Evaluation Includes sequencing of the *SCNN1A, SCNN1B*, and *SCNN1G* genes.
663	*RAF1* DNA Sequencing Test
772	*SCNN1A* DNA Sequencing Test
745	*SCNN1B* DNA Sequencing Test
746	*SCNN1G* DNA Sequencing Test
888	*SDHB* DNA Sequencing Test
847	*SHOX* (GHD) DNA Sequencing and Deletion Test
662	*SOS1* DNA Sequencing Test
358	*TACR3* DNA Sequencing Test
804	*TCF1 (MODY3)* DNA Sequencing and Deletion Test

Test Code	Test Name
805	*TCF2 (MODY5)* DNA Sequencing and Deletion Test
858	von Hippel-Lindau Syndrome (*VHL*) DNA Sequencing Test
Nephrology	
711	*ACTN4* DNA Sequencing Test
852	*AQP2* (Nephrogenic Diabetes Insipidus) DNA Sequencing Test
765	*BSND* DNA Sequencing Test
825	*CASR* DNA Sequencing Test
764	*CLCNKB* DNA Sequencing Test
757	*COL4A3* DNA Sequencing Test
758	*COL4A4* DNA Sequencing Test
756	*COL4A5* Deletion Analysis
755	*COL4A5* Sequencing and Deletion Analysis
759	Complete Alport Syndrome Evaluation Includes sequencing of the *COL4A3*, *COL4A4*, and *COL4A5* genes and detection of deletions in *COL4A5*.
8100	Complete PKDx® Evaluation Includes sequencing of the *PKD1* and *PKD2* genes and detection of deletions in both genes.
779	*CYP11B1/2* DNA Chimeric Gene Fusion Test
722	Early Onset Nephrotic Syndrome Evaluation Includes sequencing of the *LAMB2*, *NPHS1*, *NPHS2*, *PLCE1*, and *WT1* genes.
185	Familial DNA Sequence Evaluation Includes analysis for a familial mutation only.
717	Focal and Segmental Glomerulosclerosis (FSGS) Evaluation Includes sequencing of the *ACTN4*, *INF2*, *NPHS2*, and *TRPC6* genes.
770	Hereditary Interstitial Kidney Disease (UMOD) DNA Sequencing Test Includes sequencing of the *UMOD* gene.
767	Hereditary Renal Tubular Disorders Evaluation Includes sequencing of the *BSND*, *CLCNKB*, *KCNJ1*, *SLC12A1*, and *SLC12A3* genes.
776	*HNF1B* DNA Sequencing and Deletion Evaluation
775	*HSD11B2* DNA Sequencing Test
716	*INF2* DNA Sequencing Test
763	*KCNJ1* DNA Sequencing Test

Test Code	Test Name
714	*LAMB2* DNA Sequencing Test
747	Liddle's Syndrome Evaluation Includes sequencing of the *SCNN1B* and *SCNN1G* genes.
851	Nephrogenic Diabetes Insipidus (*AVPR2*) DNA Sequencing Test
854	Nephrogenic Diabetes Insipidus Evaluation Includes sequencing of the *AQP2* and *AVPR2* genes.
750	*NPHP1* Deletion Test (Familial Juvenile Nephronophthisis) Includes detection of homozygous deletions in the *NPHP1* gene.
730	*NPHS1* DNA Sequencing Test
710	*NPHS2* DNA Sequencing Test
8105	*PKD1* Deletion Test
8101	*PKD1* DNA Sequencing and Deletion Evaluation
8103	*PKD1* DNA Sequencing Test
8106	*PKD2* Deletion Test
8102	*PKD2* DNA Sequencing and Deletion Evaluation
8104	*PKD2* DNA Sequencing Test
728	PKDx® Familial Mutation Evaluation Includes detection of the familial polycystic kidney disease mutation only.
718	*PLCE1* DNA Sequencing Test
748	Pseudohypoaldosteronism Type 1 Evaluation Includes sequencing of the *SCNN1A*, *SCNN1B*, and *SCNN1G* genes.
772	*SCNN1A* DNA Sequencing Test
745	*SCNN1B* DNA Sequencing Test
746	*SCNN1G* DNA Sequencing Test
762	*SLC12A1* DNA Sequencing Test
766	*SLC12A3* DNA Sequencing Test
712	*TRPC6* DNA Sequencing Test
713	*WT1* DNA Sequencing Test
Neurology—Cerebrovascular Disorders	
1106	*CCM2* Sequencing and CNV Evaluation Includes sequencing and analysis of copy number variations (CNV) in the *CCM2* gene.
1120	*COL4A1* Sequencing Test (CSVD)

Test Code	Test Name
1122	Complete CCM Sequencing and CNV Evaluation Includes sequencing and analysis of copy number variations (CNV) in the *CCM2*, *KRIT1*, and *PDCD10* genes.
1149	*HTRA1* (CARASIL) Sequencing Test
1152	*KRIT1 (CCM1)* Sequencing and CNV Evaluation Includes sequencing and analysis of copy number variations (CNV) in the *KRIT1* gene.
1175	*NOTCH3* (CADASIL) Sequencing Test
1179	*PDCD10 (CCM3)* Sequencing and CNV Evaluation Includes sequencing and analysis of copy number variations (CNV) in the *PDCD10* gene.
Neurology—Dementia	
178	ADmark® Alzheimer's Evaluation Includes *APOE* genotyping and concentrations of phosphorylated tau protein, total tau protein, and β-amyloid 42.
109	ADmark® *APOE* Genotype Analysis and Interpretation (Symptomatic)
168	ADmark® *APP* DNA Sequencing Test and Duplication Test
179	ADmark® Early Onset Alzheimer's Evaluation Includes sequencing of the *APP*, *PS-1 (PSEN1)*, and *PS-2 (PSEN2)* genes and detection of duplications in the *APP* gene.
167	ADmark® *PSEN1* DNA Sequencing Test
169	ADmark® *PSEN2* DNA Sequencing Test
209	*C9orf72* (FTD) DNA Test
281	Frontotemporal Dementia Evaluation Includes sequencing of *GRN* and *MAPT* genes and detection of expansions in the *C9orf72* gene.
204	*GRN* DNA Sequencing Test
205	*MAPT* DNA Sequencing Test
Neurology—Epilepsy	
1036	*ARX* Sequencing and CNV Evaluation (Epilepsy) Includes sequencing and analysis of copy number variations (CNV) in the *ARX* gene.
1115	*CDKL5* Sequencing and CNV Evaluation (Epilepsy) Includes sequencing and analysis of copy number variations (CNV) in the *CDKL5* gene.

Test Code	Test Name
1131	Complete Tuberous Sclerosis Sequencing and CNV Evaluation Includes sequencing and analysis of copy number variations (CNV) in the *TSC1* and *TSC2* genes.
1133	*CSTB (EPM1)* Sequencing and Repeat Expansion Evaluation Includes sequencing and analysis of copy number variations (CNV) in the *CSTB (EPM1)* gene.
6000	Epilepsy Advanced Sequencing and CNV Evaluation Includes sequencing and analysis of copy number variations (CNV) in the *ABAT, ADGRV1, ADSL, ALDH7A1, ALG13, ALG9, ALPL, AMT, ANKRD11, ARFGEF2, ARHGEF9, ARX, ASAH1, ASPM, ATP13A2, ATP1A2, ATP1A3, ATP2A2, ATP6AP2, ATP6V0A2, ATRX, BCKDK, BRAT1, CACNA1A, CACNA1H, CACNA2D1, CACNA2D2, CACNB4, CASK, CASR, CDKL5, CENPJ, CHD2, CHRNA2, CHRNA4, CHRNA7, CHRNB2, CLN3, CLN5, CLN6, CLN8, CNTNAP2, COL4A1, CPA6, CPT2, CRH, CSTB, CTSD, CTSF, CUL4B, CYP27A1, DCX, DEAF1, DEPDC5, DNAJC5, DNM1, DOCK7, DPYD, DYNC1H1, DYRK1A, EEF1A2, EFHC1, EMX2, EPM2A, FGD1, FGFR3, FKRP, FKTN, FLNA, FOLR1, FOXG1, GABRA1, GABRB2, GABRB3, GABRD, GABRG2, GAMT, GATM, GFAP, GLDC, GNAO1, GOSR2, GPC3, GPR56, GRIA3, GRIN1, GRIN2A, GRIN2B, GRN, HCN1, HCN4, HNRNPU, HPRT1, HSD17B10, IQSEC2, KANSL1, KCNA1, KCNA2, KCNB1, KCNC1, KCNH2, KCNJ10, KCNJ11, KCNMA1, KCNQ2, KCNQ3, KCNT1, KCTD7, KDM5C, KIAA1279, KIAA2022, KMT2D, L2HGDH, LAMA2, LARGE1, LBR, LGI1, LIAS, LMNB2, MAGI2, MBD5, MCPH1, MECP2, MEF2C, MFSD8, NDE1, NDUFA1, NHLRC1, NIPBL, NOTCH3, NR2F1, NRXN1, OFD1, OPHN1, PAFAH1B1, PAK3, PANK2, PAX6, PCDH19, PEX7, PHF6, PHGDH, PIGA, PIGN, PIGO, PIGV, PLA2G6, PLCB1, PLP1, PNKP, PNPO, POLG, POMGNT1, POMT1, POMT2, PPT1, PQBP1, PRICKLE1, PRICKLE2, PRIMA1, PRRT2, PURA, QARS, RAB39B, RAB3GAP1, RAI1, RBFOX1, RELN, RNASEH2A, RNASEH2B, RNASEH2C, ROGDI, SAMHD1, SCARB2, SCN1A, SCN1B, SCN2A, SCN3A, SCN5A, SCN8A, SCN9A, SERPINI1, SETBP1, SETD2, SHH, SIK1, SIX3, SLC13A5, SLC19A3, SLC25A19, SLC25A22, SLC2A1, SLC35A2, SLC4A10, SLC6A1, SLC6A8, SLC9A6, SMC1A, SMC3, SMS, SNAP25, SPATA5, SPTAN1, SRPX2, ST3GAL3, ST3GAL5, STIL, STX1B, STXBP1, SUCLA2, SYN1, SYNGAP1, SYNJ1, SYP, SZT2, TBC1D24, TBL1XR1, TBX1, TCF4, TPP1, TREX1, TSC1, TSC2, TSEN54, TUBA1A, TUBA8, TUBB2B, UBE3A, VPS13A, VPS13B, WDR45, WDR62, WWOX,* and *ZEB2* genes.

Test Code	Test Name
6018	**Epilepsy Advanced Sequencing and CNV Evaluation–Developmental Brain Malformations** Includes sequencing and analysis of copy number variations (CNV) in the *ARFGEF2, ARX, ASPM, CENPJ, COL4A1, CPT2, DCX, DEAF1, DPYD, EMX2, FGFR3, FKRP, FKTN, FLNA, FOLR1, GPR56, LAMA2, LARGE1, MCPH1, NDE1, PAFAH1B1, PAX6, PEX7, POMGNT1, POMT1, POMT2, PQBP1, QARS, RAB3GAP1, RELN, SHH, SIX3, SLC25A19, SRPX2, STIL, TSEN54, TUBA1A, TUBA8, TUBB2B,* and *WDR62* genes.
6023	**Epilepsy Advanced Sequencing and CNV Evaluation–Epilepsy with Migraine** Includes sequencing and analysis of copy number variations (CNV) in the *ATP1A2, CACNA1A, NOTCH3, POLG, PRRT2, SCN1A,* and *SLC2A1* genes.
6010	**Epilepsy Advanced Sequencing and CNV Evaluation–Epileptic Encephalopathy** Includes sequencing and analysis of copy number variations (CNV) in the *ADSL, ALG13, ALG9, AMT, ARHGEF9, ARX, BRAT1, CACNA2D1, CACNA2D2, CDKL5, CHD2, CNTNAP2, DNM1, DOCK7, EEF1A2, FOXG1, GABRG2, GLDC, GNAO1, GRIN1, GRIN2A, GRIN2B, HCN1, HNRNPU, KCNA2, KCNB1 , KCNJ11, KCNT1, LIAS, MECP2, NRXN1, PCDH19, PIGA, PIGN, PLCB1, PNKP, PNPO, PURA, QARS, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, SCN1A, SCN1B, SCN2A, SCN8A, SCN9A, SIK1, SLC13A5, SLC19A3, SLC25A19, SLC25A22, SLC2A1, SLC35A2, SLC9A6, SPTAN1, ST3GAL5, STXBP1, SYNGAP1, SZT2, TCF4, TREX1, UBE3A, WDR45, WWOX,* and *ZEB2* genes.
6008	**Epilepsy Advanced Sequencing and CNV Evaluation–Generalized, Absence, Focal, Febrile and Myoclonic Epilepsies** Includes sequencing and analysis of copy number variations (CNV) in the *ABAT, ADSL, ALDH7A1, ALG13, ALG9, ALPL, AMT, ASAH1, ASPM, ATP1A3 , BCKDK, BRAT1, CACNA1A, CACNA1H, CACNB4, CASR, CHD2, CHRNA2, CHRNA4, CHRNA7 , CHRNB2, CPA6, CRH, CSTB, CYP27A1, DEPDC5, DYNC1H1, EFHC1, EPM2A, FOLR1, GABRA1, GABRB2 , GABRB3, GABRD, GABRG2, GAMT, GATM, GLDC, GOSR2, GRIN2A, GRIN2B, HCN1, HCN4, KCNC1, KCNH2, KCNMA1, KCNQ2, KCNQ3, KCNT1, KCTD7, L2HGDH, LGI1, LIAS, LMNB2, MBD5, NDUFA1, NHLRC1, PCDH19, PHGDH, PIGO , PNPO, PRICKLE1, PRICKLE2, PRIMA1, PRRT2, SCARB2, SCN1A, SCN1B, SCN2A, SCN3A, SCN5A, SCN9A, SLC19A3, SLC25A19, SLC2A1, SLC35A2, SLC4A10, SLC6A1, SLC6A8, ST3GAL5, STX1B, SUCLA2, SYNJ1,* and *TBC1D24* genes.
6038	**Epilepsy Advanced Sequencing and CNV Evaluation–Infantile Spasms** Includes sequencing and analysis of copy number variations (CNV) in the *ADSL, ARX, CACNA2D1, CDKL5, FOXG1, GABRB3, GRIN2A, MEF2C, NR2F1, SCN2A, SLC25A22, SLC35A2, SPTAN1, ST3GAL3, STXBP1,* and *TBL1XR1* genes.

Test Code	Test Name
6019	**Epilepsy Advanced Sequencing and CNV Evaluation–Intellectual Disability** Includes sequencing and analysis of copy number variations (CNV) in the *ABAT, ADSL, ALG13, ALG9, ARHGEF9, ARX, ATP6AP2, ATRX, BCKDK, CACNA2D1, CASK, CDKL5, CHRNA7 , CUL4B, DCX, DEAF1, DPYD, DYRK1A, EEF1A2, FGD1, FOLR1, GABRB2 , GAMT, GATM, GFAP, GPC3, GRIA3, GRIN2B, HNRNPU, HSD17B10, IQSEC2, KDM5C, KIAA2022, MECP2, OFD1, OPHN1, PAK3, PCDH19, PHF6, PLP1, PQBP1, PURA, RAB39B, RBFOX1, SETD2, SLC35A2, SLC6A8, SLC9A6, SMC1A, SMS, SNAP25, SPATA5, SRPX2, SYN1, SYP,* and *WDR45* genes.
6022	**Epilepsy Advanced Sequencing and CNV Evaluation–Neuronal Ceroid Lipofuscinosis** Includes sequencing and analysis of copy number variations (CNV) in *ATP13A2, CLN3, CLN5, CLN6, CLN8, CTSD, CTSF, DNAJC5, GRN, KCTD7, MFSD8, PPT1,* and *TPP1* genes.
6033	**Epilepsy Advanced Sequencing and CNV Evaluation–Syndromic Disorders** Includes sequencing and analysis of copy number variations (CNV) in the *ADGRV1, ANKRD11, ATP2A2, ATP6V0A2, GFAP, HPRT1, KANSL1, KCNA1, KCNJ10, KIAA1279, KMT2D, LBR, LGI1, MAGI2, NIPBL, PANK2, PIGA, PIGN, PIGV, PLA2G6, RAI1, ROGDI, SERPINI1, SETBP1, SMC3, SYNGAP1, TBX1, TSC1, TSC2, VPS13A,* and *VPS13B* genes.
410	*EPM1* DNA Test Includes detection of dodecamer expansions in the *CSTB* gene.
1003	*GFAP* (Alexander Disease) Sequencing Test
518	*MERRF* mtDNA Evaluation Includes detection of a point mutation in each of the *MT-TH, MT-TK, MT-TL1, MT-TS1* genes in mitochondrial DNA.
443	*POLG* DNA Sequencing Test (Alpers' Syndrome)
1191	*SCN1A* CNV Test
1129	*SCN1A* Sequencing and CNV Evaluation Includes sequencing and analysis of copy number variations (CNV) in the *SCN1A* gene.
537	*SCN1A* Deletion Test
4411	*SLC2A1* DNA Sequencing Test
523	*TSC* Familial Mutation Evaluation Includes detection of the familial tuberous sclerosis mutation only.
1236	*TSC1* CNV Test
508	*TSC1* Deletion Analysis
1245	*TSC1* Sequencing Test

Test Code	Test Name
1254	*TSC2* CNV Test
524	*TSC2* DNA Deletion Test
1255	*TSC2* Sequencing Test
Neurology—Hearing Loss	
321	Connexin 26 (*GJB2*) DNA Sequencing Test
319	Connexin 30 DNA Test Includes deletion analysis of the *GJB6* gene.
329	Connexin Related Deafness Evaluation Includes sequencing of the *GJB2* gene and deletion analysis of the *GJB6* gene.
3029	Hearing Loss Advanced Sequencing and CNV Analysis Includes analysis of DFNB1 Locus and the following genes: *ABHD12, ACTB, ACTG1, ADCY1, ADGRV1 (GPR98), AIFM1, ALMS1, ANKH, ATP2B2 (PMCA2), ATP6V1B1, ATP6V1B2, BCS1L, BDP1, BSND, CABP2, CACNA1D, CCDC50, CD151, CD164, CDC14A, CDH23, CEACAM16, CEMIP, CHD7, CHSY1, CIB2, CISD2, CLDN14, CLIC5, CLPP, CLRN1, COCH, COL9A3, COL11A1, COL11A2, COL2A1, COL4A3, COL4A4, COL4A5, COL4A6, COL9A1, COL9A2, CRYM, DCDC2, DFNA5, DFNB59 (PJVK), DIABLO, DIAPH1, DIAPH3, DLX5, DNMT1, DSPP, EDN3, EDNRA, EDNRB, ELMOD3, EPS8, EPS8L2, ERCC2, ERCC3, ESPN, ESRRB, EYA1, EYA4, FGF3, FGFR1, FGFR2, FGFR3, FOXI1, GATA3, GIPC3, GJB2, GJB3, GJB6**,GPSM2, GRHL2, GRXCR1, GRXCR2, HARS2, HGF, HOMER2, HOXB1, HSD17B4, ILDR1, KARS, KCNE1, KCNJ10, KCNQ1, KCNQ4, LARS2, LHFPL5, LOXHD1, LRTOMT, MANBA, MARVELD2, MCM2, MET, MIR96* (miRNA)*, MITF, MSRB3, MT-CO1*, MT-CO2*, MT-ND1*, MT-RNR1* (rRNA)*, MT-TH* (tRNA)*, MT-TI* (tRNA)*, MT-TK* (tRNA)*, MT-TL1* (tRNA)*, MT-TQ* (tRNA)*, MT-TS1* (tRNA)*, MT-TS2* (tRNA)*, MYH14, MYH9, MYO15A, MYO3A, MYO6, MYO7A, NARS2, NDP, NLRP3, NR2F1, OPA1, OSBPL2, OTOA, OTOF, OTOG, OTOGL, P2RX2, PAX3, PCDH15, PDZD7, PEX1, PEX6, PMP22, PNPT1, POLR1C, POLR1D, POU3F4, POU4F3, PRPS1, PTPRQ, RDX, RIPOR2 (FAM65B), ROR1, S1PR2, SALL1*, SEMA3E, SERPINB6, SIX1, SIX5, SLC12A1, SLC17A8, SLC19A2, SLC22A4, SLC26A4, SLC26A5, SLC4A11, SLITRK6, SMPX, SNAI2, SOX10, STRC, SYNE4, TBC1D24, TBX1, TCOF1, TECTA, TFAP2A, TIMM8A, TJP2, TMC1, TMEM132E, TMIE, TMPRSS3, TNC, TPRN, TRIOBP, TSPEAR, USH1C, USH1G, USH2A, WFS1,* and *WHRN*. *Sequencing only. **CNV analysis only.
Neurology—Intellectual Disability/Autism	
790	*AHI1* DNA Sequencing Test (Joubert Syndrome)
1038	*ARX* Sequencing and CNV Evaluation (Intellectual Disability) Includes sequencing and analysis of copy number variations (CNV) in the *ARX* gene.

Test Code	Test Name
1092	*ASPM* Sequencing Test
794	*CC2D2A* DNA Sequencing Test (Joubert Syndrome)
1114	*CDKL5* Sequencing and CNV Evaluation (Atypical Rett) Includes sequencing and analysis of copy number variations (CNV) in the *CDKL5* gene.
791	*CEP290* DNA Sequencing Test (Joubert Syndrome)
1142	*FOXG1* Sequencing and CNV Evaluation Includes sequencing and analysis of copy number variations (CNV) in the *FOXG1* gene.
795	Joubert Syndrome Evaluation Includes sequencing of the *AHI1, CC2D2A, CEP290, TMEM67*, and *TMEM216* genes and deletion analysis of the *NPHP1* gene.
1153	*MCPH1* Sequencing Test
1155	*MECP2* Sequencing and CNV Evaluation Includes sequencing and analysis of copy number variations (CNV) in the *MECP2* gene.
7540	*MEF2C* Evaluation Includes sequencing and detection of mutations and large deletions in the *MEF2C* gene.
1166	*MEF2C* Sequencing and CNV Evaluation Includes sequencing and analysis of copy number variations (CNV) in the *MEF2C* gene.
793	*NPHP1* Deletion Test (Joubert Syndrome)
1186	Primary Microcephaly Sequencing Evaluation Includes sequencing and analysis of copy number variations (CNV) in the *ASPM, MCPH1*, and *WDR62* genes.
1190	*PTEN* Sequencing Test
148	Rett Syndrome (*MECP2*) Duplication/Deletion Analysis
1192	*SHANK2* Sequencing Test
1193	*SHANK3* Sequencing Test
737	Smith-Lemli-Opitz Syndrome (*DHCR7*) DNA Sequencing Test
1194	*SYNGAP1* Sequencing Test
789	*TMEM216* DNA Sequencing (Joubert Syndrome)
792	*TMEM67* DNA Sequencing Test (Joubert Syndrome)
1256	*VPS13B (COH1)* Sequencing Test
1257	*WDR62* Sequencing Test

Test Code	Test Name
Neurology—Leukodystrophy	
815	*ABCD1* (Adrenoleukodystrophy) DNA Sequencing Test
6108	*ABCD1* (Neurology) DNA Sequencing Test
6107	*ARSA* DNA Sequencing Test
6101	*EIF2B1* DNA Sequencing Test
6102	*EIF2B2* DNA Sequencing Test
6103	*EIF2B3* DNA Sequencing Test
6104	*EIF2B4* DNA Sequencing Test
6105	*EIF2B5* DNA Sequencing Test
6109	*GJC2* DNA Sequencing Test
6106	Leukoencephalopathy with Vanishing White Matter Includes sequencing of the *EIF2B1*, *EIF2B2*, *EIF2B3*, *EIF2B4*, and *EIF2B5* genes.
1183	*PLP1* Sequencing and CNV Evaluation Includes sequencing and analysis of copy number variations (CNV) in the *PLP1* gene.
Neurology—Mitochondrial Disorders	
466	*ANT1 (SLC25A4)* DNA Sequencing Test (related to mtDNA depletion)
487	*DGUOK* DNA Sequencing Test (related to mtDNA depletion)
575	Initial Mitochondrial Evaluation Includes restriction fragment polymorphism analysis of the *MT-ATP6*, *MT-ND1*, *MT-ND5*, *MT-TH*, *MT-TK*, *MT-TL1*, *MT-TS1*, and *MT-TV* genes and sequencing of the *POLG* gene.
517	MELAS mtDNA Evaluation Includes restriction fragment polymorphism analysis of the *MT-ND1*, *MT-ND5*, *MT-TH*, *MT-TL1*, and *MT-TV* genes.
579	Mitochondrial Encephalomyopathic Evaluation Includes sequencing of the *POLG*, *RRM2B*, and *TK2* genes.
578	Mitochondrial Hepatoencephalopathic Evaluation Includes sequencing of the *DGUOK*, *MPV17*, *POLG*, TWINKLE *(PEO1/C10orf2)* genes.
577	Mitochondrial Neurogastrointestinal Encephalopathy Evaluation (MNGIE) Includes sequencing of the *RRM2B* and *TYMP* genes and restriction fragment polymorphism analysis of the *MT-ND1*, *MT-ND5*, *MT-TH*, *MT-TL1*, and *MT-TV* genes.
488	*MPV17* DNA Sequencing Test (related to mtDNA depletion)
469	*OPA1* DNA Sequencing Test (related to mtDNA depletion)
824	*PDHA1* DNA Sequencing Test
474	*POLG* DNA Sequencing Test (related to all allelic disorders)
576	Progressive External Ophthalmoplegia Evaluation Includes sequencing of the *ANT1 (SCL25A4)*, *OPA1*, *POLG*, TWINKLE *(PEO1/C10orf2)* genes and restriction fragment polymorphism analysis of the *MT-ND1*, *MT-ND5*, *MT-TH*, *MT-TL1*, and *MT-TV* genes.
486	*RRM2B* DNA Sequencing Test (related to mtDNA depletion)
489	*TK2* DNA Sequencing Test (related to mtDNA depletion)
479	TWINKLE (*PEO1/C10orf2*) DNA Sequencing Test (related to mtDNA depletion)
484	*TYMP* DNA Sequencing Test (related to mtDNA depletion)
Neurology—Motor Neuron Disorders	
6521	Atypical Spinal Muscular Atrophy Advanced Sequencing Evaluation
670	*C9orf72* DNA Test Includes analysis for repeat expansions in the *C9orf72* gene.
6611	HSP, Common Dominant Evaluation Includes sequencing 4 genes *(ATL1, KIF5A, REEP1, SPAST)* and a *SPAST* deletion test.
6621	HSP, Common Recessive Evaluation Includes sequencing 3 genes: *SPG7*, *SPG11*, and *ZFYVE26*.
6601	HSP, Common Sporadic Evaluation Includes sequencing 2 genes *(SPAST, SPG7)* and *SPAST* deletion test.
6610	HSP, Complete Dominant Evaluation Includes sequencing 10 genes *(ATL1, BSCL2, HSPD1, KIAA0196, KIF5A, NIPA1, REEP1, RTN2, SLC33A1, SPAST)* and *SPAST* deletion test.
6620	HSP, Complete Recessive Evaluation Includes sequencing 12 genes: *ALS2*, *AP5Z1*, *CYP7B1*, *FA2H*, *KIF1A*, *PNPLA6*, *SACS*, *SPG11*, *SPG20*, *SPG21*, *SPG7*, and *ZFYVE26*.

Test Code	Test Name
6630	HSP, Comprehensive Evaluation Includes sequencing 24 genes *(ALS2, AP5Z1, BSCL2, CYP7B1, FA2H, HSPD1, KIAA0196, KIF1A, KIF5A, L1CAM, NIPA1, PLP1, PNPLA6, REEP1, RTN2, SACS, SLC33A1, SPAST, SPG11, SPG20, SPG21, SPG3A, SPG7, and ZFYVE2)* and *SPAST* deletion test.
6612	HSP, Supplemental Dominant Evaluation Includes sequencing 6 genes: *BSCL2, HSPD1, KIAA0196, NIPA1, RTN2,* and *SLC33A1*.
6622	HSP, Supplemental Recessive Evaluation Includes sequencing 9 genes: *ALS2, AP5Z1, CYP7B1, FA2H, KIF1A, PNPLA6, SACS, SPG20* and *SPG21*.
6602	HSP, Supplemental Sporadic Evaluation Includes sequencing 22 genes: *ALS2, AP5Z1, ATL1, BSCL2, CYP7B1, FA2H, HSPD1, KIAA0196, KIF1A, KIF5A, L1CAM, NIPA1, PLP1, PNPLA6, REEP1, RTN2, SACS, SLC33A1, SPG11, SPG20, SPG21,* and *ZFYVE26*.
6631	HSP, X-Linked Evaluation Includes sequencing 2 genes: *L1CAM* and *PLP1*.
117	Kennedy's Disease (SBMA) DNA Includes detection of CAG triplet repeats in the *AR* gene.
444	SMA Carrier Screen Includes deletion analysis of the *SMN1* and *SMN2* genes.
214	SMA Plus Includes deletion analysis of the *SMN1* gene. If an *SMN1* deletion is detected on only 1 allele, sequencing of the *SMN1* and *SMN2* genes will be performed at an additional charge.
211	*SMN* DNA Sequencing Test Includes sequencing of the *SMN1* and *SMN2* genes.
620	*SOD1* DNA Sequencing Test
633	Spatacsin (*SPG11*) DNA Sequencing Test
531	*SPG3A* (Atlastin) DNA Sequencing Test
632	*SPG7* (Paraplegin) DNA Sequencing Test
614	*ZFYVE26 (SPG15)* DNA Sequencing Test
Neurology—Movement Disorders	
557	Alpha Synuclein (*SNCA*) DNA Sequencing Test
59	Alpha Synuclein (*SNCA*) Duplication/Deletion Test
283	Ataxia with Vitamin E Deficiency (AVED) *TTPA* DNA Sequencing Test

Test Code	Test Name
6901	Ataxia, Common Repeat Expansion Evaluation Includes repeat expansion detection for the 8 most common genetic causes of SCA (1, 2, 3, 6, 7, 8, 10, and 17) including the *ATXN1, ATXN2, ATXN3, ATXN7, ATXN8OS, ATXN10, CACNA1A* and *TBP* genes.
6900	Ataxia, Complete Dominant Evaluation Includes sequencing of 16 genes (*AFG3L2, CACNB4, EEF2, FGF14, ITPR1, KCNA1, KCNC3, KCND3, PDYN, PPP2R2B, PRKCG, SLC1A3, SPTBN2, TGM6, TTBK2, VAMP1*) and 10 repeat expansion tests (*ATN1, ATXN1, ATXN10, ATXN2, ATXN3, ATXN7, ATXN8OS, CACNA1A, PPP2R2B,* and *TBP*). Smaller panels of these components are also available.
6910	Ataxia, Complete Recessive Evaluation Includes sequencing of 18 genes (*ADCK3, AFG3L2, ANO10, APTX, ATM, FLVCR1, FXN, GRM1, MRE11A, MTPAP, POLG, SACS, SETX, SIL1, SYNE1, SYT14, TDP1, TTPA*), *FXN* repeat expansion test, and *ATM* deletion test. Smaller panels of these components are also available.
6930	Ataxia, Comprehensive Evaluation Includes sequencing of 33 genes *(ADCK3, AFG3L2, ANO10, APTX, ATM, CACNA1A, CACNB4, EEF2, FGF14, FLVCR1, FXN, GRM1, ITPR1, KCNA1, KCNC3, KCND3, MRE11A, MTPAP, PDYN, POLG, PRKCG, SACS, SETX, SIL1, SLC1A3, SPTBN2, SYNE1, SYT14, TDP1, TGM6, TTBK2, TTPA,* and *VAMP1),* 11 repeat expansion tests *(ATN1, ATXN1, ATXN10, ATXN2, ATXN3, ATXN7, ATXN8OS, CACNA1A, FXN, PPP2R2B,* and *TBP),* and *ATM* deletion test. Smaller panels of these components are also available.
349	Ataxia, Friedreich (FXN) Evaluation Includes detection of GAA triplet repeats and sequencing of the *FXN* gene.
6903	Ataxia, Supplemental Dominant Evaluation Includes sequencing 16 genes: *AFG3L2, CACNA1A, CACNB4, EEF2, FGF14, ITPR1, KCNA1, KCNC3, KCND3, PDYN, PRKCG, SLC1A3, SPTBN2, TGM6, TTBK2,* and *VAMP1*.
6911	Ataxia, Supplemental Recessive Evaluation Includes sequencing of 17 genes (*ADCK3, AFG3L2, ANO10, APTX, ATM, FLVCR1, GRM1, MRE11A, MTPAP, POLG, SACS, SETX, SIL1, SYNE1, SYT14, TDP1, TTPA*) and dosage *ATM* deletions.
353	Ataxia-Telangiectasia (ATM) Evaluation Includes sequencing and deletion analysis of the *ATM* gene.
402	Chorea Differential Evaluation Includes detection of CAG triplet repeats in the *HTT (IT15)* and *DRPLA* genes.
629	Complete Dopa-Responsive Dystonia (DYT5) Evaluation Includes sequencing of the *GCH1* and *TH* genes and deletion analysis of the *GCH1* gene.

Test Code	Test Name
588	Complete Parkinsonism Evaluation Includes sequencing and duplication/deletion analysis of the *PARK2, PARK7 (DJ1), PINK1,* and *SNCA* genes and sequencing of the *LRRK2* gene.
401	*DRPLA* (ATN1) Repeat Expansion Test Includes detection of CAG triplet repeats in the *DRPLA* gene.
626	Dystonia (*DYT1*) DNA Test Includes deletion analysis of the *TOR1A (DYT1)* gene.
6920	Episodic Ataxia Evaluation Includes sequencing of the *CACNA1A, SLC1A3, KCNA1,* and *CACNB4* genes.
348	Friedreich Ataxia (*FXN*) DNA Sequencing Test Includes sequencing of the *FXN* gene.
119	Friedreich Ataxia (*FXN*) Repeat Expansion Test Includes detection of GAA triplet repeats in the *FXN* gene.
638	*GCH1* Deletion Analysis (DYT5)
637	*GCH1* DNA Sequencing Test (DYT5A)
116	Huntington Disease Repeat Expansion Test Includes detection of CAG triplet repeats in the *HTT (IT15)* gene.
639	Isolated Dystonia Evaluation Includes deletion analysis of the *TOR1A (DYT1)* gene and sequencing of the *THAP1* gene.
558	*LRRK2* DNA Sequencing Test
543	*LRRK2* Targeted DNA Test Includes sequencing of 3 codons in the *LRRK2* gene: 1441 on exon 31, and 2019 and 2020 on exon 41.
383	MIRAS-Specific *POLG1* DNA Sequencing Test Includes sequencing of the 3 *PLOG1* gene exons that are associated with mitochondrial recessive ataxia syndrome.
6912	Oculomotor Apraxia Ataxia Advanced Sequencing Evaluation Includes sequencing 2 genes: *APTX* and *SETX*.
559	*PARK2* (Parkin) DNA Sequencing Test
40	*PARK2* (Parkin) Duplication/Deletion Test
47	*PARK7 (DJ1) Deletion Test*
554	*PARK7 (DJ1)* DNA Sequencing Test
58	*PINK1* Deletion Test
542	*PINK1* DNA Sequencing Test
617	*PNKD* (MR-1) DNA Sequencing Test
1187	*PRRT2* (Dyskinesia/IC) Sequencing Test

Test Code	Test Name
371	*SCA1 (ATXN1)* Repeat Expansion Test Includes detection of CAG triplet repeats in the *SCA1* gene.
387	*SCA10 (ATXN10)* Repeat Expansion Test Includes detection of ATTCT pentanucleotide repeats in the *SCA10* gene.
285	*SCA12 (PPP2R2B)* Repeat Expansion Test Includes detection of CAG triplet repeats in the SCA12 *(PPP2R2B)* gene.
388	*SCA17 (TBP)* Repeat Expansion Test Includes detection of CAG/CAA triplet repeats in the *TBP (SCA17)* gene.
672	*SCA2 (ATXN2)* Repeat Expansion Test Includes detection of CAG triple repeats in the *ATXN2 (SCA2)* gene.
105	*SCA3 (MJD/ATXN3)* Repeat Expansion Test Includes detection of CAG triplet repeats in the *ATXN3 (SCA3)* gene.
373	*SCA6 (CACNA1A)* Repeat Expansion Test Includes detection of CAG triplet repeats in the *CACNA1A (SCA6)* gene.
677	*SCA7 (ATXN7)* Repeat Expansion Test Includes detection of CAG triplet repeats in the *ATXN7 (SCA7)* gene.
384	*SCA8* (ATXN8OS) Repeat Expansion Test Includes detection of CTA/CTG triplet repeats in the *ATXN8OS (SCA8)* gene.
627	*SGCE* Deletion Analysis (DYT11)
624	*SGCE* DNA Sequencing Test (DYT11)
634	*TH* DNA Sequencing Test (DYT5B)
618	*THAP1* DNA Sequencing Test (DYT6)
Neurology—Neuromuscular Disorders	
584	*CAPN3* Duplication/Deletion Test
585	*CAPN3* Evaluation Includes sequencing and deletion analysis of the CAPN3 gene.
566	Caveolin 3 (*CAV3*) DNA Sequencing Test Includes sequencing of the *CAV3* gene.
128	*CLCN1* DNA Sequencing Test
110	*CNBP* DNA Test (*DM2*) Includes detection of CCTG repeats in the *CNBP (DM2, ZNF9)* genes.

Test Code	Test Name
5502	Congenital Muscular Dystrophy Advanced Sequencing Evaluation Includes sequencing and detection of sequence variations in 23 genes: *B3GALNT2, B3GNT1, CHKB, COL6A1, COL6A2, COL6A3, DNM2, DPM2, FHL1, FKRP, FKTN, ISPD, ITGA7, LAMA2, LARGE, LMNA, POMGNT1, POMGNT2, POMT1, POMT2, SEPN1, TCAP,* and *TMEM5*.
5511	Congenital Myasthenic Syndrome Advanced Sequencing Evaluation Includes sequencing of 13 genes: *AGRN, CHAT, CHRNA1, CHRNB1, CHRND, CHRNE, COLQ, DOK7, DPAGT1, GFPT1, MUSK, RAPSN,* and *SCN4A*.
5503	Congenital Myopathy Advanced Sequencing Evaluation Includes sequencing and detection of sequence variations in 21 genes: *ACTA1, BIN1, CCDC78, CFL2, CNTN1, DNM2, KBTBD13, KLHL40, MEGF10, MTM1, MYBPC3, MYH2, MYH7, NEB, RYR1, SEPN1, TNNT1, TPM2, TPM3, TRIM32,* and *TTN*.
5504	Distal Myopathy Advanced Sequencing Evaluation Includes sequencing and detection of sequence variations in 17 genes: *ANO5, CAV3, CRYAB, DES, DNM2, DYSF, FLNC, GNE, KLHL9, LDB3, MATR3, MYH7, MYOT, NEB, TIA1, TTN,* and *VCP*.
183	*DMD* DNA Sequencing Test
5531	*DMD* Duplication/Deletion Test
5530	*DMD* Evaluation Includes sequencing and detection of sequence variations, duplications, and deletions in the dystrophin (*DMD*) gene.
108	*DMPK* DNA Test (DM1) Includes detection of CTG triplet repeats in the *DMPK (DM1)* gene.
571	Dysferlin DNA Sequencing Test Includes sequencing of the *DYSF* gene.
207	Early Onset Myotonia Evaluation Includes sequencing of the *CLCN1* and *SCN4A* genes and detection of CTG triplet repeats in the *DMPK* gene.
5518	Emery-Dreifuss Muscular Dystrophy Advanced Sequencing Evaluation Includes sequencing and detection of sequence variations in 6 genes: *EMD, FHL1, LMNA, SYNE1, SYNE2,* and *TMEM43*.
562	*FKRP* DNA Sequencing Test
5905	FSHD Molecular Combing Test Includes detection of 4q35 deletions and haplotype associated with facioscalpulohumeral muscular dystrophy.

Test Code	Test Name
405	FSHD1 Southern Blot Test Includes 4q35 deletions associated with facioscalpulohumeral muscular dystrophy.
565	Lamin A/C (*LMNA*) DNA Sequencing Test Includes sequencing of the *LMNA* gene.
515	LHON mtDNA Evaluation Includes detection of point mutations in the *ND1* (3460G>A), *ND4* (11778G>A), and *ND6* (14484T>C) genes.
5519	Limb Girdle Muscular Dystrophy Evaluation Includes sequencing of the *CAPN3, CAV3, DYSF, FKRP, LMNA, MYOT, SGCA, SGCB, SGCD* and *SGCG* genes. Also includes deletion analysis in the *CAPN3, SGCA,* and *SGCG* genes.
5508	Malignant Hyperthermia Advanced Sequencing Evaluation Includes sequencing of 2 genes: *CACNA1S* and *RYR1*.
517	MELAS mtDNA Evaluation Includes restriction fragment polymorphism analysis of the *MT-ND1, MT-ND5, MT-TH, MT-TL1,* and *MT-TV* genes.
518	*MERRF* mtDNA Evaluation Includes detection of a point mutation in each of the *MT-TH, MT-TK, MT-TL1, MT-TS1* genes in mitochondrial DNA.
5501	Muscular Dystrophy Advanced Evaluation Includes sequencing and detection of sequence variations in 33 genes (*ANO5, CAPN3, CAV3, CCDC78, DAG1, DES, DMD, DNAJB6, DYSF, EMD, FHL1, FKRP, FKTN, ISPD, LMNA, MYOT, PLEC, POMGNT1, POMT1, POMT2, PTRF, SGCA, SGCB, SGCD, SGCG, SMCHD1, SYNE1, SYNE2, TCAP, TMEM43, TRAPPC11, TRIM32,* and *TTN*) and duplications/deletions in 4 of these genes.
5505	Myofibrillar Myopathy Advanced Sequencing Evaluation Includes sequencing and detection of sequence variations in 9 genes: *BAG3, CRYAB, DES, FHL1, FLNC, LDB3, MYOT, SEPN1,* and *TTN*.
5506	Myotonic Syndrome Advanced Evaluation Includes sequencing and detection of sequence variations in 5 genes (*ATP2A1, CAV3, CLCN1, CN4A,* and *HSPG2*) and repeat expansions in 2 genes (*CNBP, DMPK*).
516	NARP mtDNA Evaluation Includes detection of point mutations in the *MT-ATP6* gene in mitochondrial DNA.
6522	Nonprevalent Amyotrophic Lateral Sclerosis Advanced Sequencing Evaluation Includes sequencing of 15 genes: *ALS2, ANG, CHMP2B, DCTN1, FIG4, FUS, OPTN, PFN1, SETX, SIGMAR1, SQSTM1, TARDBP, UBQLN2, VAPB,* and *VCP*.

Test Code	Test Name
490	*OPA1* DNA Sequencing Test (optic atrophy)
300	OPMD Repeat Expansion Test Includes detection of GCG triplet repeats in the *PABP2* gene.
5507	Periodic Paralysis Advanced Sequencing Evaluation Includes sequencing of 3 genes: *CACNA1S*, *KCNJ2*, and *SCN4A*.
146	*SCN4A* (Myotonia) DNA Sequencing Test
582	*SGCA* Duplication/Deletion Test
583	*SGCG* Duplication/Deletion Test
Neurology—Peripheral Neuropathy	
660	*ATL1* (HSAN) DNA Sequencing Test Includes sequencing of the *ALT1* gene, targeting mutations associated with hereditary sensory and autonomic neuropathy type 1D.
224	Charcot-Marie-Tooth 4C (CMT4C) *SH3TC2* DNA Sequencing Test
4008	CMT Advanced Evaluation–Axonal Includes sequencing of the *DNM2, GARS, GDAP1, GJB1 (CX32), HSPB1, HSPB8, LMNA, MFN2, MPZ, NEFL, RAB7, TRPV4,* and *YARS* genes and deletion analysis of the *GJB1 (CX32)* and *GJB1* genes.
4001	CMT Advanced Evaluation–Comprehensive Includes sequencing of the *DNM2, EGR2, FGD4, FIG4, GARS, GDAP1, GJB1 (CX32), HSPB1, HSPB8, LITAF, LMNA, MFN2, MTMR2, MPZ, NDRG1, NEFL, PMP22, PRX, RAB7, SBF2, SH3TC2, TRPV4,* and *YARS* genes; deletion analysis of the *GJB1 (CX32)* gene; and duplicaton/deletion analysis of the *PMP22* gene.
4007	CMT Advanced Evaluation–Demyelinating Includes sequencing of the *DNM2, EGR2, FGD4, FIG4, GDAP1, GJB1 (CX32), LITAF, MTMR2, MPZ, NDRG1, PMP22, PRX, SBF2, SH3TC2,* and *YARS* genes; deletion analysis of the *GJB1 (CX32)* gene; and duplicaton/deletion analysis of the *PMP22* gene.
4005	CMT Advanced Evaluation–Dominant Includes sequencing of the *DNM2, EGR2, GARS, HSPB1, HSPB8, LITAF, MFN2, MPZ, NFL, PMP22, RAB7, TRPV4,* and *YARS* genes, and duplicaton/deletion analysis of the *PMP22* gene.
4003	CMT Advanced Evaluation–Dominant, Axonal Includes sequencing of the *DNM2, GARS, HSPB1, HSPB8, LMNA, MFN2, MPZ, NEFL, RAB7, TRPV4,* and *YARS* genes.
4002	CMT Advanced Evaluation–Dominant, Demyelinating Includes sequencing of the *DNM2, EGR2, LITAF, MPZ, PMP22,* and *YARS* genes and duplicaton/deletion analysis of the *PMP22* gene.

Test Code	Test Name
4010	CMT Advanced Evaluation–Initial Genetic Assessment Includes sequencing of the *MPZ, MFN2,* and *GJB1* genes, and duplications/deletion analysis of the *PMP22* gene.
4013	CMT Advanced Evaluation–Nonprevalent Includes sequencing of the *DNM2, EGR2, FGD4, FIG4, GARS, GDAP1, GJB1, HSPB1, HSPB8, LITAF, LMNA, MTMR2, NDRG1, NEFL, PMP22, PRX, RAB7A, SBF2, SH3TC2, TRPV4,* and *YARS* genes and deletion analysis of the *GJB1(CX32)* gene.
4011	CMT Advanced Evaluation–Nonprevalent Axonal Includes sequencing of the *DNM2, GARS, GDAP1, GJB1, HSPB1, HSPB8, LMNA, NEFL, RAB7A, TRPV4,* and *YARS* genes and deletion analysis of the *GJB1(CX32)* gene.
4012	CMT Advanced Evaluation–Nonprevalent Demyelinating Includes sequencing of the *DNM2, EGR2, FGD4, FIG4, GDAP1, GJB1, LITAF, MTMR2, NDRG1, PMP22, PRX, SBF2, SH3TC2,* and *YARS* genes and deletion analysis of the *GJB1(CX32)* gene.
4006	CMT Advanced Evaluation–Recessive Includes sequencing of the *FGD4, FIG4, GDAP1, LMNA, MTMR2, NDRG1, PRX, SBF2,* and *SH3TC2* genes.
4004	CMT Advanced Evaluation–Recessive, Demyelinating Includes sequencing of the *PRX, GDAP1, SBF2, SH3TC2, MTMR2, NDRG1, FGD4,* and *FIG4* genes.
243	Complete HNPP Evaluation Includes sequencing and duplication/deletion analysis of the *PMP22* gene.
245	Congenital Hypomyelination Evaluation Includes sequencing of the *EGR2* and *MPZ* genes.
253	*DNM2* DNA Sequencing Test
691	Early Onset Hereditary Sensory and Autonomic Neuropathy (HSAN) Evaluation Includes sequencing of the *NTRK1* and *WNK1* genes.
248	*EGR2* DNA Sequencing Test
296	Entrapment Neuropathy Evaluation Includes sequencing of the *PMP22* and *TTR* genes and duplication/deletion analysis of the *PMP22* gene.
208	*FGD4* DNA Sequencing Test
225	*FIG4* (CMT) DNA Sequencing Test
228	*GARS* DNA Sequencing Test
221	*GDAP1* DNA Sequencing Test

Test Code	Test Name
143	*GJB1* (CX32) Sequencing and Deletion Evaluation
229	*HSPB1* DNA Sequencing Test
463	*HSPB8* DNA Sequencing Test
698	Late Onset Hereditary Sensory and Autonomic Neuropathy (HSAN) Evaluation Includes sequencing of the *SPTLC1* and *SPTLC2* genes.
222	*LITAF/SIMPLE* DNA Sequencing Test
226	*LMNA* (CMT) DNA Sequencing Test
518	*MERRF* mtDNA Evaluation Includes detection of a point mutation in each of the *MT-TH, MT-TK, MT-TL1, MT-TS1* genes in mitochondrial DNA.
223	*MFN2* DNA Sequencing Test
134	*MPZ* DNA Sequencing Test
354	*MTMR2* DNA Sequencing Test
289	Multifocal Motor Neuropathy Evaluation Includes *PMP22* deletion analysis and ELISA detection of GM1, GD1a, Asialo GM1, and GD1b antibodies.
394	*NDRG1* DNA Sequencing Test
249	Neurofilament Light (*NEFL*) DNA Sequencing Test
659	*NTRK1* DNA Sequencing Test
247	*PMP22* DNA Sequencing Test
131	*PMP22* Duplication/Deletion Test
239	*PRX* DNA Sequencing Test
227	*RAB7A* DNA Sequencing Test
164	*SBF2* DNA Sequencing Test
719	*SEPT9* DNA Sequencing Test

Test Code	Test Name
551	*SPTLC1* DNA Sequencing Test
552	*SPTLC2* DNA Sequencing Test
144	*TRPV4* DNA Sequencing Test
235	*TTR* DNA Sequencing
553	*WNK1* DNA Sequencing Test
468	*YARS* DNA Sequencing Test
Neurology—Other	
1101	*ATP1A2* (FHM) Sequencing Test
1103	*CACNA1A* (FHM) Sequencing Test
185	Familial DNA Sequence Evaluation Includes analysis for a familial mutation only.
1148	Hemiplegic Migraine Sequencing Evaluation Includes sequencing of the *ATP1A2, CACNA1A,* and *SCN1A* genes.
647	Neurofibromatosis Type 1 Deletion Test Includes deletion analysis of the *NF1* gene.
646	Neurofibromatosis Type 1 DNA Sequencing Test Includes sequencing of the *NF1* gene.
644	Neurofibromatosis Type 2 (NF2) Duplication/ Deletion Test
645	Neurofibromatosis Type 2 (NF2) Evaluation Includes sequencing and duplication/deletion analysis of the *NF2* gene.
635	Neurofibromatosis Type 2 DNA Sequencing Includes sequencing of the *NF2* gene.
648	Neurofibromatosis Type 1 (NF1) Evaluation Includes sequencing and deletion analysis of the *NF1* gene.
1136	*SCN1A* Sequencing Test (FHM)

All tests were developed and their performance characteristics have been determined by Athena Diagnostics. Performance characteristics refer to the analytical performance of the test.