Athena Diagnostics Client Test Requisition

Client Services is available Monday through Friday from 8:30 AM to 9:00 PM EST at 1.800.394.4493, option 2
If you wish to have Athena Diagnostics bill the insurance company directly, please use the Insurance and Advance Pay Test Requisition.
Please note: Athena Diagnostics must bill hospitals directly for all Medicare hospital inpatient and outpatient testing.

Patient Information

Patient Name _____ Patient ID# (if available) _____
Date of Birth _____ Sex designated at birth: ☐ Male ☐ Female
Street address _____
City _____ State _____ Zip _____
Mobile phone #1 _____ Other Phone #2 _____
Patient email _____
Language spoken if other than English _____

Who Should Athena Diagnostics Contact with Questions About this Order?

Name _____ Phone _____
Fax _____ Email _____

Authorized Result Report Recipients Required Physician Information

NPI # _____ Name _____
First Last
Address _____
City _____ State _____ Zip _____
Phone Fax _____ Email _____

Laboratory Information

CLIA # _____ Lab Name _____
Address _____
City _____ State _____ Zip _____
Phone _____ Fax _____

Ordering Account Information - Hospital/Laboratory Billing Information

(Hospital billing is required for all Medicare patients – both inpatients and outpatients.)

Ordering physician name: _____
Address _____
City _____ State _____ Zip _____
Phone _____ Fax _____ Email _____
NPI# _____ Athena Account # (if assigned) _____ Reporting preference: ☐ Fax ☐ Email

Send additional report copies to:

Clinician/Facility _____
NPI# or CLIA _____
Address _____
City _____ State _____ Zip _____
Phone _____ Fax _____ Email _____
Athena Account # (if assigned) _____ CLIA # _____
Purchase Order # (if available) _____ Billing Contact _____
Email _____ Phone _____ Fax _____
Hospital/Lab Name _____ Address _____
City _____ State _____ Zip _____

Test Information

Consult test list for test code, name and acceptable specimen options. Specimen requirements are referenced at the top of the test list. Call Client Services at 1.800.394.4493, option 2 for additional details.

ICD-10 Codes (required for billing insurance): _____

Test Code	Test Name

Statement of Medical Necessity and Informed Consent:

In accordance with Massachusetts General Law Chapter 111, Section 70G, and New York Civil Rights Law Section 79-1 verification of patient informed consent is required for genetic testing. Additionally, testing laboratories located in Massachusetts require a signed acknowledgment from the ordering medical practitioner. The signed acknowledgment is required to complete the genetic testing ordered if you have not previously signed a blanket Physician Attestation of Informed Consent (PAIC) at any Quest lab.

Prior to ordering genetic testing on the patient listed above, I have obtained a signed, written consent form from the patient (or their authorized representative) as required by applicable state law and/or regulations, and I will maintain all written consent forms as part of the patient file and make them available to Athena Diagnostics upon reasonable request. Many payers (including Medicare and Medicaid) have medical necessity requirements consistent with local state regulatory requirements for the test ordered. I understand I should only order those tests which are medically necessary for the diagnosis and treatment of the patient consistent with local state regulatory requirements for the test ordered. I further confirm this test is medically necessary for the diagnosis or detection of disease, illness, impairment, symptom, syndrome, or disorder and the results will be used in the medical management and treatment decisions for the patient consistent with local state regulatory requirements for the test ordered. I confirm that the person listed in the Ordering Physician space above is authorized by law to order the test(s) requested herein consistent with local state regulatory requirements for the test ordered.

Please sign, date and include your credentialed (MD, DO, NP) to document your intent to order the testing. Please note that if the information is not provided, you may be required to provide medical records and/or progress notes to support intent to order on payor request.

Medical Practitioner Signature: _____ Date _____/_____/_____
Medical Practitioner Credentials: _____

Client Services is available Monday through Friday from 8:30 AM to 9:00 PM EST at 1.800.394.4493, option 2
If you wish to have Athena Diagnostics bill the insurance company directly, please use the Insurance Test Requisition.
Please note: Athena Diagnostics must bill hospitals directly for all Medicare hospital inpatient and outpatient testing.

Clinical Information

Clinical diagnosis: _____

Age at Initial Presentation: _____

Ancestral Background (check all that apply):

☐ African ☐ Asian: East ☐ Asian: Southeast ☐ Central/South American

☐ Hispanic ☐ Native American ☐ Ashkenazi Jewish ☐ Asian: Indian

☐ Caribbean ☐ European ☐ Middle Eastern ☐ Pacific Islander

Other: _____

Indications for genetic testing (please check one):

☐ Diagnostic (symptomatic) ☐ Predictive (asymptomatic) ☐ Prenatal (Contact Athena prior to sending)

☐ Carrier ☐ Family testing/single site

Relationship to Proband: _____

If performed at Athena, provide relative's accession #_____.

If performed at another lab, a copy of the relative's report is required.

Please attach detailed medical records and family history information.

Specimen Information

Specimen Type: Date sample obtained: _____/_____/_____

☐ Whole Blood ☐ Serum ☐ Cerebrospinal Fluid (CSF) ☐ CVS: Cultured

☐ Amniotic Fluid: Cultured ☐ Saliva (Not available for all tests)

☐ DNA* source: _____ Concentration _____ ug/ml

*DNA must be extracted at a CLIA-certified or a laboratory meeting equivalent requirements (as determined by CAP and/or CMS).

☐ Other** source (provide specimen type): _____

Contact Athena **prior to sending specimen types not listed above.

If not collected same day as shipped, how was sample stored? ☐ Room temp ☐ Refrigerated ☐ Frozen

History of ☐ blood transfusion or ☐ bone marrow transplant? ☐ Yes ☐ No

Date of most recent transfusion/transplant: _____/_____/_____

Important: Please be sure to write in the test code(s) and test name(s) within the Test Information section on page 1.

Reflexive testing is performed at an additional charge.

The Advance Pay Option is accepted for all Molecular Genetics test codes that do not have an Immunology or STAT component. These test codes will be noted as not qualifying for Advance Pay in the Additional Information (Genes, Antibodies, Comments) Columns below.

athena diagnostics

MOLECULAR GENETICS SPECIMEN REQUIREMENTS: Specimen Type = Blood, Volume = 8 mL, Tube Type = Lavender Top.
NOTE1: Saliva is acceptable for most genetic tests. Call Athena at 1-800-394-4493 to order Saliva kits and see the Additional Information Column for exceptions.
NOTE2: The pediatric minimum is 2 mL for Neurodevelopmental Disorders & Epilepsy.
NOTE3: Copy Number Variant (CNV) is equivalent to Deletion and Duplication.

IMMUNOLOGY SPECIMEN REQUIREMENTS: Specimen Type = Serum, Volume = 2 mL, Tube Type = Serum Separator Tube.

Please Refer to the Additional Information (Genes, Antibodies, Comments) Column below for specimen requirement exceptions.

NEUROLOGY GENETIC & IMMUNOLOGY TESTING

Test Code	Test Name	Additional Information (Genes, Antibodies, Comments)
Cerebrovascular Disease (Stroke): Molecular Genetics		
☐ 1175	Notch3 (CADASIL) Sequencing Test	
☐ 1149	HTRA1 (CARASIL) Sequencing Test	
☐ 1120	COL4A1 Sequencing Test (CSVD)	
☐ **1122**	**Complete CCM Sequencing and CNV Evaluation**	
Individual CCM single gene tests: Only order single gene tests when not ordering the panel. ☐ 1152 KRIT1 (CCM1) Seq. and CNV Evaluation ☐ 1106 CCM2 Seq. and CNV Evaluation ☐ 1179 PDCD10 (CCM3) Seq. and CNV Evaluation		
Dementia: Molecular Genetics		
☐ **178**	**ADmark® Alzheimer's Evaluation**	Does not qualify for the Advance Pay Option. Molecular Genetics Component(s): ApoE Immunology Component(s): AB42, Phospho-Tau & Total-Tau. **Specimen Requirements**: Cerebrospinal Fluid (CSF) 2 mL in Polypropylene Tube and must arrive on cold pack or frozen. Whole blood 8 mL (6 mL minimum) in Lavender top (EDTA) tube. Cannot be performed with Saliva sample type.
☐ 109	ADmark® ApoE Genotype Analysis & Interpretation (Symptomatic for Dementia)	
☐ **179**	**ADmark® Early-Onset Alzheimer's Evaluation**	PSEN1, APP Seq./Dup., PSEN2
Individual ADmark® Early-Onset Alzheimer's single gene tests: Only order single gene tests when not ordering the panel. ☐ 168 ADmark® APP DNA Sequencing Test and Duplication Test ☐ 167 ADmark® PSEN1 DNA Sequencing Test ☐ 169 ADmark® PSEN2 DNA Sequencing Test		
☐ **281**	**Frontotemporal Dementia (FTD) Evaluation**	MAPT, GRN, C9orf72
Individual FTD single gene tests: Only order single gene tests when not ordering the panel. ☐ 209 C9orf72 (FTD) DNA Test ☐ 204 GRN DNA Sequencing Test ☐ 205 MAPT DNA Sequencing Test		
Dementia: Immunology		
☐ 177	ADmark® Phospho-Tau/Total-Tau/Aß42 CSF	Analysis & Interpretation (Symptomatic) Specimen Type = Cerebrospinal Fluid (CSF) Volume = 2 mL **Tube Type = Polypropylene Tube** **Must arrive on cold pack or frozen.**
☐ **1711**	**Autoimmune Rapidly Progressive Dementia Evaluation with Recombx®**	
Individual Autoimmune Dementia single antibody tests: Only order single autoantibody tests when not ordering the panel. ☐ 1714 Recombx® Hu Autoantibody Test* ☐ 1716 Recombx® MaTa Autoantibody Test* ☐ 1717 Recombx® CV2 Autoantibody Test* ☐ 1718 Recombx® Amphiphysin Autoantibody Test* ☐ 1705 GAD65 Autoantibody Test ☐ 1706 NMDA Receptor Autoantibody Test* ☐ 1707 VGKC Autoantibody Test ☐ 1708 LGI1 Autoantibody Test* ☐ 1709 CASPR2 Autoantibody Test* * NOTE: Cerebrospinal Fluid (CSF) is an acceptable sample type for these tests.		
Epilepsy: Molecular Genetics		
☐ **6000**	**Epilepsy Advanced Sequencing and CNV Evaluation**	Test 6000 contains all genes included in the sub-panels. NOTE: Only select sub-panels if 6000 is not ordered. Please see website for the list of genes in each panel. .
☐ 6018	Developmental Brain Malformations	
☐ 6023	Epilepsy with Migraine	
☐ 6010	Epileptic Encephalopathy	
☐ 6008	Generalized, Absence, Focal, Febrile and Myoclonic Epilepsies	
☐ 6038	Infantile Spasms	
☐ 6019	Intellectual Disability	
☐ 6022	Neuronal Ceroid Lipofuscinosis	
☐ 6033	Syndromic Disorders	

Test Code	Test Name	Additional Information (Genes, Antibodies, Comments)
Epilepsy: Molecular Genetics (Continued)		
☐ **1131**	**Complete Tuberous Sclerosis Sequencing and CNV Evaluation**	Full Sequencing of TSC1 & TSC2
Individual Tuberous Sclerosis single gene tests: Only order single gene tests when not ordering the panel. ☐ 1236 TSC1 CNV Test ☐ 1254 TSC2 CNV Test ☐ 508 TSC1 Deletion Analysis (for NYS Only) ☐ 524 TSC2 DNA Deletion Test (for NYS Only) ☐ 1245 TSC1 Sequencing Test ☐ 1255 TSC2 Sequencing Test		
☐ 523	TSC Familial DNA Seq. Mutation Evaluation Proband Accession # _____ Relationship _____	
☐ **1129**	**SCN1A Seq. and CNV Evaluation**	
Individual SCN1A tests: ☐ 1191 SCN1A CNV Test ☐ 537 SCN1A Deletion Test		
☐ **1133**	**CSTB (EPM1) Sequencing and Repeat Expansion Evaluation**	Cannot be done on saliva.
☐ 410	EPM1 DNA Test	Repeat Expansion Testing Cannot be done on saliva.
☐ 1036	ARX Seq. and CNV Evaluation (Epilepsy)	
☐ 1115	CDKL5 Seq. and CNV Evaluation (Epilepsy)	
☐ 4411	SLC2A1 DNA Sequencing Test	
☐ 1003	GFAP (Alexander Disease) Seq. Test	
☐ 443	POLG DNA Seq. Test (Alpers Syndrome)	
Epilepsy: Immunology		
☐ **5120**	**Autoimmune Epilepsy Evaluation**	GAD65, VGKC, CASPR2, LGI1, NMDA
Individual Autoimmune Epilepsy single antibody tests: Only order single antibody tests when not ordering the panel. ☐ 5103 CASPR2 Autoantibody Test (Epilepsy) (Single) ☐ 5101 GAD65 Neurological Syndrome Autoantibody Test (Epilepsy) (Single) ☐ 5104 LGI1 Autoantibody Test (Epilepsy) (Single) ☐ 5105 NMDA Receptor Autoantibody Test (Epilepsy) (Single) ☐ 5102 VGKC Autoantibody Test (Epilepsy) (Single)		
Family Testing		
☐ 185	Familial DNA Sequence Evaluation	This test detects previously identified sequence variants in at-risk family members. For Familial TSC variants, please order Code 523. Proband Accession #_____ Relationship _____
Immunology: Anti-Drug Antibody		
☐ 1181	AAV9 Antibody Test	Does not qualify for the Advance Pay Option.
Leukodystrophy: Molecular Genetics		
☐ **6106**	**Leukoencephalopathy with Vanishing White Matter Evaluation**	EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5
Individual Leukoencephalopathy with Vanishing White Matter single gene tests: Only order single gene tests when not ordering the panel. ☐ 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		
☐ 1183	PLP1 Sequencing and CNV Evaluation	
☐ 6108	ABCD1 DNA Sequencing Test	
☐ 6107	ARSA DNA Sequencing Test	
☐ 6109	GJC2 DNA Sequencing Test	
☐ 1175	Notch3(CADASIL) Sequencing Test	

Important: Please be sure to write in the test code(s) and test name(s) within the Test Information section on page 1.

Reflexive testing is performed at an additional charge.

The Advance Pay Option is accepted for all Molecular Genetics test codes that do not have an Immunology or STAT component. These test codes will be noted as not qualifying for Advance Pay in the Additional Information (Genes, Antibodies, Comments) Columns below.

athena diagnostics

MOLECULAR GENETICS SPECIMEN REQUIREMENTS: Specimen Type = Blood, Volume = 8 mL, Tube Type = Lavender Top.
NOTE1: Saliva is acceptable for most genetic tests. Call Athena at 1-800-394-4493 to order Saliva kits and see the Additional Information Column for exceptions.
NOTE2: The pediatric minimum is 2 mL for Neurodevelopmental Disorders & Epilepsy.
NOTE3: Copy Number Variant (CNV) is equivalent to Deletion and Duplication.

IMMUNOLOGY SPECIMEN REQUIREMENTS: Specimen Type = Serum, Volume = 2 mL, Tube Type = Serum Separator Tube.
Please Refer to the Additional Information (Genes, Antibodies, Comments) Column below for specimen requirement exceptions.

Test Code	Test Name	Additional Information (Genes, Antibodies, Comments)
Migraine: Molecular Genetics		
☐ **1148**	**Hemiplegic Migraine Sequencing Evaluation**	CACNA1A, ATP1A2, SCN1A
	Individual Hemiplegic Migraine single gene tests: Only order single gene tests when not ordering the panel. ☐ 1101 ATP1A2 Sequencing Test ☐ 1103 CACNA1A Sequencing Test ☐ 1136 SCN1A Sequencing Test (FHM)	
Motor Neuron Diseases: Molecular Genetics		
☐ 6520	Amyotrophic Lateral Sclerosis Advanced Evaluation	Please see website for the complete list of genes.
☐ 6522	Nonprevalent Amyotrophic Lateral Sclerosis Advanced Sequencing Evaluation	
☐ 670	C9orf72 DNA Test	
☐ 620	SOD1 DNA Sequencing Test	
☐ **6630**	**HSP, Comprehensive Evaluation**	Please see website for the complete list of genes. Test 6630 contains all genes included in the sub-panels. NOTE: Only select sub-panels if 6630 is not ordered.
☐ 6601	HSP, Common Sporadic Evaluation	SPAST, SPG7
☐ 6602	HSP, Supplemental Sporadic Evaluation	Please see website for the complete list of genes.
☐ 6610	HSP, Complete Dominant Evaluation	
☐ 6611	HSP, Common Dominant Evaluation	SPAST, ATLN, REEP1, KIF5A
☐ 6612	HSP, Supplemental Dominant Evaluation	BSCL2, HSPD1, KIAA0196, NIPA1, RTN2, SLC33A1
☐ 6620	HSP, Complete Recessive Evaluation	Please see website for the complete list of genes.
☐ 6621	HSP, Common Recessive Evaluation	SPG11, ZFYVE26, SPG7
☐ 6622	HSP, Supplemental Recessive Evaluation	Please see website for the complete list of genes.
☐ 6631	HSP, X-Linked Evaluation	L1CAM, PLP1
☐ 6509	SPG4 Evaluation	SPAST
Movement Disorders: Molecular Genetics		
	Individual HSP DNA Tests: Only order single gene tests when not ordering the panel. ☐ 531 Atlastin ☐ 632 Paraplegin ☐ 633 Spatacsin ☐ 614 ZFYVE26 ☐ 117 Kennedy's Disease (SBMA) DNA Test	SPG3A SPG7 SPG11 SPG15
☐ **6930**	**Ataxia, Comprehensive Evaluation**	Please see website for the complete list of genes. Test 6930 contains all genes included in the sub-panels. NOTE: Only select sub-panels if 6930 is not ordered. Cannot be performed on saliva.
☐ 6900	Ataxia, Complete Dominant Evaluation	Please see website for the complete list of genes. Cannot be performed on saliva.
☐ 6901	Ataxia, Common Repeat Expansion Evaluation	
☐ 6903	Ataxia, Supplemental Dominant Evaluation	
☐ 6910	Ataxia, Complete Recessive Evaluation	
☐ 6911	Ataxia, Supplemental Recessive Evaluation	Please see website for the complete list of genes.
☐ 6912	Oculomotor Apraxia Ataxia Advanced Sequencing Evaluation	APTX, SETX
☐ 6920	Episodic Ataxia Evaluation	CACNB4, KCNA1, SLC1A3, CACNA1A
☐ 349	Ataxia, Friedreich (FXN) Evaluation	FRDA/FXN Seq., FRDA/FXN Expansion
☐ 353	Ataxia-Telangiectasia (ATM) Evaluation	ATM Seq., ATM Dup./Del.

Test Code	Test Name	Additional Information (Genes, Antibodies, Comments)
Movement Disorders: Molecular Genetics (Continued)		
	Individual Ataxia single gene DNA Tests: Only order single gene tests when not ordering the panel or sub-panels. ☐ 401 DRPLA ☐ 119 FRDA/FXN Expansion) ☐ 348 FRDA/FXN Seq. ☐ 383 POLG1 (MIRAS) ☐ 371 SCA1 (ATXN1) ☐ 672 SCA2 (ATXN2) ☐ 105 SCA3 (ATXN3) ☐ 373 SCA6 (CACNA1A) ☐ 677 SCA7 (ATXN7) ☐ 384 SCA8 (ATXN8OS) ☐ 387 SCA10 (ATXN10) ☐ 285 SCA12 (PPP2R2B) ☐ 388 SCA17 (TBP) ☐ 283 TTPA (AVED)	SCA8 and SCA10 test cannot be performed on saliva.
☐ **402**	**Chorea Differential Evaluation** (DRPLA, Huntington's Disease)	Cannot be performed on saliva.
☐ 116	Huntington Disease Repeat Expansion Test	Cannot be performed on saliva.
☐ **639**	**Isolated Dystonia Evaluation**	DYT1, THAP1
	Individual Isolated Dystonia single gene tests: Only order single gene tests when not ordering the panel. ☐ 626 Dystonia (DYT1) DNA Test ☐ 618 THAP1 DNA Sequencing Test	
☐ **629**	**Complete Dopa-Responsive Dystonia (DYT5) Evaluation**	GCH1 Seq., GCH1 Del., TH Seq.
	Individual Dopa-Responsive Dystonia single gene tests: Only order single gene tests when not ordering the panel. ☐ 637 GCH1 DNA Sequencing Test ☐ 638 GCH1 Deletion Analysis ☐ 634 TH DNA Sequencing Test	DYT5A DYT5A DYT5B
☐ 624	SGCE DNA Sequencing Test	DYT11
☐ 627	SGCE Deletion Analysis	DYT11
☐ 617	PNKD (MR-1) DNA Sequencing Test	
☐ **588**	**Complete Parkinsonism Evaluation**	LRRK2, PARK2, PINK1, PARK7, SNCA
	Individual Parkinsonism single gene tests: Only order single gene tests when not ordering the panel. ☐ 557 Alpha Synuclein (SNCA) DNA Seq. Test ☐ 059 Alpha Synuclein (SNCA) Dup./Del. Test ☐ 558 LRRK2 DNA Sequencing Test ☐ 559 PARK2 (Parkin) DNA Sequencing Test ☐ 040 PARK2 (Parkin) Duplication/Deletion Test ☐ 554 PARK7 (DJ1) DNA Sequencing Test ☐ 047 PARK7 (DJ1) Deletion Test ☐ 542 PINK1 DNA Sequencing Test ☐ 058 PINK1 Deletion Test	
☐ 1187	PRRT2 (Dyskinesia/IC) Seq. Test	
Multiple Sclerosis/Demylenating Diseases: Immunology		
☐ 1287	NMO Spectrum Evaluation	AQP4, CBA reflex to MOG, CBA
☐ 1282	Aquaporin-4 (AQP4) (NMO IgG) Antibody, CBA with Reflex to Titer	Cerebrospinal Fluid (CSF) is an acceptable sample type.
☐ 1523	Myelin Oligodendrocyte Glycoprotein (MOG) Antibody, CBA with Reflex to Titer	Cerebrospinal Fluid (CSF) is an acceptable sample type.
☐ 1284	NMO Spectrum Evaluation	AQP4, ELISA reflex to MOG, CBA
☐ 193	Aquaporin-4 (AQP4) Antibody (NMO-IgG), ELISA	
☐ 112	NAbFeron® (INFB-1) Neutralizing Antibody Test	
☐ 197	TYSABRI® (Natalizumab) Antibody Test	See website for collection notes
Myasthenia Gravis: Immunology		
☐ 1521	Myasthenia Gravis Panel 2 with Reflex to MuSK Antibody	
☐ 1514	Myasthenia Gravis Panel 2	Includes AChR Binding / Blocking / Modulating Antibody
☐ 1490	MuSK and LRP4	
☐ 1510	Acetylcholine Receptor Binding Antibody with Reflex to Musk Antibody	
☐ 1511	Acetylcholine Receptor Binding Antibody with Reflex to MuSK/LRP4 Antibodies	
	Individual Myasthenia Gravis single antibody tests: Only order single antibody tests when not ordering the corresponding panel option(s). ☐ 1513 Acetylcholine Receptor Binding Antibody ☐ 1483 LRP4 Autoantibody Test ☐ 1516 Acetylcholine Receptor Blocking Antibody ☐ 1481 RyR Autoantibody Test ☐ 1517 Acetylcholine Receptor Modulating Antibody ☐ 1480 Titin Autoantibody Test ☐ 482 MuSK Antibody Test	

Important: Please be sure to write in the test code(s) and test name(s) within the Test Information section on page 1.

Reflexive testing is performed at an additional charge.

The Advance Pay Option is accepted for all Molecular Genetics test codes that do not have an Immunology or STAT component. These test codes will be noted as not qualifying for Advance Pay in the Additional Information (Genes, Antibodies, Comments) Columns below.

athena diagnostics

MOLECULAR GENETICS SPECIMEN REQUIREMENTS: Specimen Type = Blood, Volume = 8 mL, Tube Type = Lavender Top.
NOTE1: Saliva is acceptable for most genetic tests. Call Athena at 1-800-394-4493 to order Saliva kits and see the Additional Information Column for exceptions.
NOTE2: The pediatric minimum is 2 mL for Neurodevelopmental Disorders & Epilepsy.
NOTE3: Copy Number Variant (CNV) is equivalent to Deletion and Duplication.

IMMUNOLOGY SPECIMEN REQUIREMENTS: Specimen Type = Serum, Volume = 2 mL, Tube Type = Serum Separator Tube.

Please Refer to the Additional Information (Genes, Antibodies, Comments) Column below for specimen requirement exceptions.

Test Code	Test Name	Additional Information (Genes, Antibodies, Comments)
Neurodevelopmental Disorders: Molecular Genetics		
☐ **1186**	**Primary Microcephaly Sequencing Evaluation**	ASPM, MCPH1, WDR62
	Individual Primary Microcephaly single gene tests: Only order single gene tests when not ordering the panel. ☐ 1092 ASPM Sequencing Test ☐ 1153 MCPH1 Sequencing Test ☐ 1257 WDR62 Sequencing Test	
☐ 1193	SHANK3 Sequencing Test	
☐ 1192	SHANK2 Sequencing Test	
☐ 1190	PTEN Sequencing Test	
☐ **795**	**Joubert Syndrome Evaluation**	
	Individual Joubert Syndrome single gene tests: Only order single gene tests when not ordering the panel. ☐ 790 AHI1 DNA Sequencing Test ☐ 794 CC2D2A DNA Sequencing Test ☐ 791 CEP290 DNA Sequencing Test ☐ 793 NPHP1 DNA Deletion Test ☐ 789 TMEM216 DNA Sequencing Test ☐ 792 TMEM67 DNA Sequencing Test	
☐ **1155**	**MECP2 Sequencing and CNV Evaluation**	
☐ 1114	CDKL5 Seq. and CNV Evaluation (Atypical Rett)	
☐ 148	Rett Syndrome (MECP2) Dup./Del. Test	
☐ 737	Smith-Lemli-Opitz Syndrome (DHCR7) DNA Sequencing Test	
☐ 1256	VPS13B (COH1) Sequencing Test	
☐ 1038	ARX Seq. and CNV Evaluation (Intellectual Disability)	
☐ 1194	SYNGAP1 Sequencing Test	
☐ 1166	MEF2C Sequencing and CNV Evaluation	
☐ 1142	FOXG1 Sequencing and CNV Evaluation	
Neuromuscular Disorders: Molecular Genetics		
☐ 5501	Muscular Dystrophy Advanced Evaluation	Please see website for the complete list of genes.
☐ 5502	Congenital Muscular Dystrophy Advanced Sequencing Evaluation	
☐ 5503	Congenital Myopathy Advanced Sequencing Evaluation	
☐ 5504	Distal Myopathy Advanced Sequencing Evaluation	
☐ 5505	Myofibrillar Myopathy Advanced Sequencing Evaluation	
☐ 5506	Myotonic Syndromes Advanced Evaluation	Please see website for the complete list of genes. Cannot be performed on saliva.
☐ 5507	Periodic Paralysis Advanced Sequencing Evaluation	Please see website for the complete list of genes.
☐ 5508	Malignant Hyperthermia Advanced Sequencing Evaluation	
☐ 5511	Congenital Myasthenic Syndrome Advanced Sequencing Evaluation	
☐ 5518	Emery-Dreifuss Muscular Dystrophy Advanced Sequencing Evaluation	
☐ 5519	Limb Girdle Muscular Dystrophy Advanced Evaluation	
	Individual Limb Girdle Muscular Dystrophy Tests: Only order single gene tests when not ordering the panel. ☐ 563 Calpain 3 DNA Sequencing Test ☐ 584 CAPN3 Duplication/Deletion Test ☐ 566 CAV3 DNA Sequencing Test ☐ 562 FKRP DNA Sequencing Test ☐ 565 LMNA DNA Sequencing Test ☐ 582 SGCA Duplication/Deletion Test ☐ 583 SGCG Duplication/Deletion Test	
☐ **5530**	**DMD Evaluation**	
	Individual DMD Evaluation single gene tests: Only order single gene tests when not ordering the panel. ☐ 183 DMD DNA Sequencing Test ☐ 5531 DMD Duplication/Deletion Test	
☐ **207**	**Early-Onset Myotonia Evaluation**	DM1, CLCN1, SCN4A Cannot be performed on saliva.
	Individual Early-Onset Myotonia single gene tests: Only order single gene tests when not ordering the panel. ☐ 128 CLCN1 DNA Sequencing Test ☐ 146 SCN4A (Myotonia) DNA Sequencing Test	
☐ 108	DMPK DNA Test (DM1)	Cannot be performed on saliva.
☐ 110	CNBP DNA Test (DM2) (DM2 testing is not recommended for patients with early onset myotonic dystrophy)	Cannot be performed on saliva.
☐ **585**	**CAPN3 Evaluation**	Includes CAPN3 Seq., CAPN3 Del.
☐ 571	Dysferlin DNA Sequencing Test	

Test Code	Test Name	Additional Information (Genes, Antibodies, Comments)
Neuromuscular Disorders: Molecular Genetics (Continued)		
☐ 405	FSHD1 Southern Blot Test	Specimen Type: Whole Blood Specimen Requirements: 10 mL (7 mL minimum) whole blood collected in two (lavender-top) EDTA tubes Sample must be received within 72 hours of collection and refrigerated. Ship sample M-Th only Cannot be performed on saliva or extracted DNA.
☐ 300	OPMD Repeat Expansion Test	Cannot be performed on saliva.
☐ 490	OPA1 DNA Sequencing Test (optic atrophy)	Related to optic atrophy.
Neuro-Oncology: Molecular Genetics		
☐ **648**	**Neurofibromatosis Type 1 (NF1) Evaluation**	NF1 Sequencing, NF1 Deletion
	Individual NF1 single gene tests: Only order single gene tests when not ordering the panel. ☐ 647 Neurofibromatosis Type 1 Deletion Test ☐ 646 Neurofibromatosis Type 1 DNA Sequencing Test	
☐ **645**	**Neurofibromatosis Type 2 (NF2) Evaluation**	NF2 Seq., NF2 Dup./Del.
	Individual NF2 single gene tests: Only order single gene tests when not ordering the panel. ☐ 635 Neurofibromatosis Type 2 DNA Sequencing Test ☐ 644 Neurofibromatosis Type 2 Duplication/Deletion Test	
Paraneoplastic & Other Antibody Disorders of the CNS: Immunology		
☐ 4711	Paraneoplastic Neurological Syndromes Evaluation with Recombx®, Initial Assessment	Cerebrospinal Fluid (CSF) is an acceptable sample type. Amphiphysin, CV2, Hu, MaTa, Ri, Yo
☐ 4620	NeoComplete Paraneoplastic Evaluation with Recombx®	* NOTE: Cerebrospinal Fluid (CSF) is an acceptable sample type Please see website for the complete list of antibodies.
☐ 4640	Paraneoplastic Autoantibody Evaluation with Recombx®, CSF *	
☐ 4724	NeoCerebellar Degeneration Paraneoplastic Profile with Recombx®	
☐ 4722	NeoEncephalitis Paraneoplastic Evaluation with Recombx®	
☐ 4725	NeoSensory Neuropathy Paraneoplastic Profile with Recombx®	Cerebrospinal Fluid (CSF) is an acceptable sample type. Amphiphysin, CV2, Hu
☐ 4727	Neuromyotonia Evaluation	CASPR2, VGKC
	Individual antibody Tests: Only order single antibody tests when not ordering the corresponding panel option(s). ☐ 419 NMDA Receptor Autoantibody Test* ☐ 4681 Recombx® CV2 Autoantibody Test * ☐ 422 GAD65 Neurological Syndrome Antibody Test ☐ 4682 Recombx® Hu Autoantibody Test * ☐ 428 Ganglionic AChR Antibody Test ☐ 4683 Recombx® MaTa Autoantibody Test * ☐ 449 LGI1 Antibody Test* ☐ 4684 Recombx® CAR (Anti-Recoverin) Autoantibody Test * ☐ 475 VGCC Type P/Q Autoantibody Test (LEMS) ☐ 485 VGKC Antibody Test ☐ 4685 Recombx® Ri Autoantibody Test * ☐ 499 CASPR2 Antibody Test* ☐ 4686 Recombx® Yo Autoantibody Test * ☐ 4674 Recombx® Amphiphysin Autoantibody Test * ☐ 4689 Recombx® Zic4 Autoantibody Test * * NOTE: Cerebrospinal Fluid (CSF) is an acceptable sample type for these tests.	
Peripheral Neuropathy (Hereditary): Molecular Genetics		
☐ **4001**	**CMT Advanced Evaluation Comprehensive (Reflexive)**	Testing is performed in this order: 1. PMP22 Dup./Del. If negative: 2. Cx32, PMP22, MFN2, MPZ, EGR2, LITAF, PRX, GDAP1, RAB7, GARS, NFL, HSPB1, LMNA, FIG4, SH3TC2, DNM2, YARS, FGD4, NDRG1, TRPV4, HSPB8, MTMR2, SBF2 DNA Seq.
☐ 4002	CMT Advanced Evaluation – Dominant, Demyelinating (Reflexive)	Testing is performed in this order: 1. PMP22 Dup./Del. If negative: 2. MPZ, PMP22 Seq., EGR2, LITAF, DNM2, YARS DNA Seq.
☐ 4003	CMT Advanced Evaluation – Dominant, Axonal	Please see website for the complete list of genes.
☐ 4004	CMT Advanced Evaluation – Recessive, Demyelinating	
☐ 4005	CMT Advanced Evaluation – Dominant (Reflexive)	Testing is performed in this order: 1. PMP22 Dup./Del. If negative: 2. MFN2, MPZ, PMP22 Seq., EGR2, LITAF, RAB7, GARS, NFL, HSPB1, DNM2, YARS, TRPV4, HSPB8 DNA Seq.
☐ 4006	CMT Advanced Evaluation – Recessive	Please see website for the complete list of genes.

Important: Please be sure to write in the test code(s) and test name(s) within the Test Information section on page 1.

Reflexive testing is performed at an additional charge.

The Advance Pay Option is accepted for all Molecular Genetics test codes that do not have an Immunology or STAT component. These test codes will be noted as not qualifying for Advance Pay in the Additional Information (Genes, Antibodies, Comments) Columns below.

athena diagnostics

MOLECULAR GENETICS SPECIMEN REQUIREMENTS: Specimen Type = Blood, Volume = 8 mL, Tube Type = Lavender Top.
NOTE1: Saliva is acceptable for most genetic tests. Call Athena at 1-800-394-4493 to order Saliva kits and see the Additional Information Column for exceptions.
NOTE2: The pediatric minimum is 2 mL for Neurodevelopmental Disorders & Epilepsy.
NOTE3: Copy Number Variant (CNV) is equivalent to Deletion and Duplication.

IMMUNOLOGY SPECIMEN REQUIREMENTS: Specimen Type = Serum, Volume = 2 mL, Tube Type = Serum Separator Tube.

Please Refer to the Additional Information (Genes, Antibodies, Comments) Column below for specimen requirement exceptions.

Test Code	Test Name	Additional Information (Genes, Antibodies, Comments)
Peripheral Neuropathy (Hereditary): Molecular Genetics (Continued)		
☐ 4007	CMT Advanced Evaluation – Demyelinating (Reflexive)	Testing is performed in this order: 1. PMP22 Dup./Del. If negative: 2. Cx32, MPZ, PMP22 Seq., EGR2, LITAF, PRX, GDAP1, DNM2, YARS, SH3TC2, MTMR2, NDRG1, FGD4, FIG4, SBF2 DNA Seq
☐ 4008	CMT Advanced Evaluation – Axonal	Please see website for the complete list of genes.
☐ 4010	CMT Advanced Evaluation – Initial Genetic Assessment	
☐ 4011	CMT Advanced Evaluation – Nonprevalent Axonal	
☐ 4012	CMT Advanced Evaluation – Nonprevalent Demyelinating	
☐ 4013	CMT Advanced Evaluation – Nonprevalent	
Individual CMT single gene tests: Only order single gene tests when not ordering the panel or sub-panels. ☐ 143 CX32 Seq./Del. (CMTX) ☐ 208 FGD4 ☐ 221 GDAP1 (CMT2K, 4A) ☐ 222 LITAF/SIMPLE (CMT1C) ☐ 134 MPZ (CMT1B, 2I, 2J) ☐ 249 NFL (CMT2E, 1F) ☐ 239 PRX (CMT4F) ☐ 224 SH3TC2 (CMT4C) ☐ 235 TTR DNA Sequencing Test ☐ 253 DNM2 ☐ 225 FIG4 (CMT4J) ☐ 229 HSPB1 (CMT2F) ☐ 226 LMNA (CMT2B1, 4C1) ☐ 354 MTMR2 ☐ 131 PMP22 Dup./Del. (CMT1A) ☐ 227 RAB7A (CMT2B) ☐ 144 TRPV4 ☐ 468 YARS ☐ 248 EGR2 (CMT1D) ☐ 228 GARS (CMT2D) ☐ 463 HSPB8 ☐ 223 MFN2 (CMT2A2) ☐ 394 NDRG1 ☐ 247 PMP22 Seq. ☐ 164 SBF2		
☐ **691**	**Early-Onset HSAN Evaluation**	NTRK1 and WNK1
☐ 243	Complete HNPP Evaluation	PMP22 Sequencing, PMP22 Dup./Del.
☐ 245	Congenital Hypomyelination Evaluation	MPZ, EGR2
☐ 296	Entrapment Neuropathy Evaluation	PMP22 Seq., PMP22 Dup./Del., TTR
Peripheral Neuropathy (Hereditary Sensory Autonomic Neuropathy): Molecular Genetics		
Individual Early-Onset HSAN single gene tests: Only order single gene tests when not ordering the panel. ☐ 659 NTRK1 (HSAN IV) DNA Sequencing Test ☐ 553 WNK1 (HSAN II) DNA Sequencing Test		
☐ **698**	**Late-Onset HSAN Evaluation**	SPTLC1 and SPTLC2
Individual Late-Onset HSAN single gene tests: Only order single gene tests when not ordering the panel. ☐ 551 SPTLC1 (HSAN I) DNA Sequencing Test ☐ 552 SPTLC2 (HSAN I) DNA Sequencing Test		
☐ 660	ATL1 (HSAN I) DNA Sequencing Test	
☐ 719	SEPT9 (HNA) DNA Sequencing Test	

Test Code	Test Name	Additional Information (Genes, Antibodies, Comments)
Peripheral Neuropathy (Autoimmune): Immunology		
☐ 3100	SensoriMotor Neuropathy Profile with Recombx® - Complete	GM1 Quattro®, MAG 'Dual Antigen'®, Hu, GALOPTM, Sulfatide
☐ 3148	Sensory Neuropathy Profile with Recombx®	(MAG 'Dual Antigen'®, Hu, GALOPTM, Sulfatide)
☐ 3163	Motor Neuropathy Profile - Complete	GM1 Quattro®, MAG 'Dual Antigen'®
☐ 289	Multifocal Motor Neuropathy Evaluation	Requires both Serum and whole blood. GM1 Quattro®, PMP22 Dup./Del.
☐ 3155	Co-GM1 Quattro® Autoantibody Test	(Asialo, GD1a, GD1b and GM1)
Individual Peripheral Neuropathy antibody tests: Only order the single antibody tests when not ordering the corresponding panel option(s). ☐ 3127 MAG 'Dual Antigen'® Autoantibody Test ☐ 261 GALOP™ Autoantibody Test ☐ 210 Sulfatide Autoantibody Test ☐ 160 GQ1b Autoantibody Test ☐ 278 GD1a Autoantibody Test ☐ 272 Asialo Autoantibody Test ☐ 273 GD1b Autoantibody Test ☐ 271 GM1 Autoantibody Test ☐ 4682 Recombx® Hu Autoantibody Test * * NOTE: Cerebrospinal Fluid (CSF) is an acceptable sample type for these tests.		
Spinal Muscular Atrophy (SMA): Molecular Genetics		
☐ 5056	SMA Carrier Screen (New York)	Does not qualify for the Advance Pay Option. Test Codes are for New York State Clients ordering SMA testing. 4 mL (2 mL minimum) whole blood collected in an EDTA (lavender-top) tube. Pediatric (0-3 years): 2 mL (1 mL minimum).
☐ 5026	SMA Diagnostic (New York)	
☐ 5070	SMA Plus (New York)	
☐ 214	SMA Plus (Reflexive)	Does not qualify for the Advance Pay Option. Test 214 includes 111 with reflex to 211.
☐ 111	Spinal Muscular Atrophy-Diagnostic	
☐ 444	Spinal Muscular Atrophy-Carrier	
☐ 211	Spinal Muscular Atrophy - SMN1 DNA Sequencing Test	
☐ 6521	Atypical SMA Advanced Sequencing Evaluation	

RENAL GENETIC TESTING

Test Code	Test Name	Additional Information (Genes, Antibodies, Comments)
Alport Syndrome: Molecular Genetics		
☐ **759**	**Complete Alport Syndrome Evaluation**	COL4A3,4,5 DNA Sequencing; COL4A5 Deletion Test
Individual Alport Syndrome single gene tests: Only order single gene tests when not ordering the panel. ☐ 757 COL4A3 DNA Sequencing Test ☐ 756 COL4A5 Deletion Analysis ☐ 758 COL4A4 DNA Sequencing Test ☐ 755 COL4A5 Sequencing and Deletion Analysis		
Amyloidosis: Molecular Genetics		
☐ 235	TTR DNA Sequencing Test	
Bardet-Biedl Syndrome: Molecular Genetics		
☐ **887**	**Bardet-Biedl Syndrome Evaluation**	BBS1, BBS2, BBS10
Individual Bardet-Biedl Syndrome single gene tests: Only order single gene tests when not ordering the panel. ☐ 871 BBS1 (BBS) DNA Sequencing Test ☐ 886 BBS10 (BBS) DNA Sequencing Test ☐ 872 BBS2 (BBS) DNA Sequencing Test		
Family Testing		
☐ **185**	**Familial DNA Sequence Evaluation** Proband Accession # _____ Relationship _____	This test detects previously identified sequence variants in at-risk family members. For Familial PKD1 and PKD2 variants, please order Code 728.

Test Code	Test Name	Additional Information (Genes, Antibodies, Comments)
Hereditary Renal Tubular Disorders: Molecular Genetics		
☐ **767**	**Hereditary Renal Tubular Disorders Evaluation**	SLC12A1, KCNJ1, CLCNKB, BSND, SLC12A3
Individual Hereditary Renal Tubular Disorder single gene tests: Only order single gene tests when not ordering the panel. ☐ 765 BSND DNA Sequencing Test ☐ 763 KCNJ1 DNA Sequencing Test ☐ 766 SLC12A3 DNA Sequencing Test ☐ 764 CLCNKB DNA Sequencing Test ☐ 762 SLC12A1 DNA Sequencing Test		
☐ 825	CASR DNA Sequencing Test	
Monogenic Hypertension: Molecular Genetics		
☐ **749**	**Monogenic Hypertension Evaluation**	SCNN1B, SCNN1G, CYP11B1, HSD11B2
☐ **747**	**Liddle's Syndrome Evaluation**	SCNN1B, SCNN1G
☐ **748**	**Pseudohypoaldosteronism Type 1 Evaluation**	SCNN1A, SCNN1B, SCNN1G
Individual Monogenic Hypertension single gene tests: Only order single gene tests when not ordering the panel. ☐ 779 CYP11B1/CYP11B2 Chimeric Gene Fusion Test ☐ 774 CYP11B1 DNA Sequencing Test ☐ 772 SCNN1A DNA Sequencing Test ☐ 746 SCNN1G DNA Sequencing Test ☐ 775 HSD11B2 DNA Sequencing Test ☐ 745 SCNN1B DNA Sequencing Test		

Important: Please be sure to write in the test code(s) and test name(s) within the Test Information section on page 1.

Reflexive testing is performed at an additional charge.

The Advance Pay Option is accepted for all Molecular Genetics test codes that do not have an Immunology or STAT component. These test codes will be noted as not qualifying for Advance Pay in the Additional Information (Genes, Antibodies, Comments) Columns below.

athena diagnostics

MOLECULAR GENETICS SPECIMEN REQUIREMENTS: Specimen Type = Blood, Volume = 8 mL, Tube Type = Lavender Top.
NOTE1: Saliva is acceptable for most genetic tests. Call Athena at 1-800-394-4493 to order Saliva kits and see the Additional Information Column for exceptions.
NOTE2: The pediatric minimum is 2 mL for Neurodevelopmental Disorders & Epilepsy.
NOTE3: Copy Number Variant (CNV) is equivalent to Deletion and Duplication.

IMMUNOLOGY SPECIMEN REQUIREMENTS: Specimen Type = Serum, Volume = 2 mL, Tube Type = Serum Separator Tube.
Please Refer to the Additional Information (Genes, Antibodies, Comments) Column below for specimen requirement exceptions.

Test Code	Test Name	Additional Information (Genes, Antibodies, Comments)
Nephrogenic Diabetes Insipidus: Molecular Genetics		
☐ **854**	**Nephrogenic Diabetes Insipidus Evaluation**	AVPR2, AQP2
	Individual Nephrogenic Diabetes Insipidus single gene tests: Only order single gene tests when not ordering the panel. ☐ 852 AQP2 DNA Sequencing Test ☐ 851 AVPR2 DNA Sequencing Test	
Nephronophthisis: Molecular Genetics		
☐ 750	NPHP1 Deletion Test (Familial Juvenile Nephronophthisis)	
Nephrotic Syndrome: Molecular Genetics		
☐ **722**	**Early Onset Nephrotic Syndrome Evaluation**	PLCE1, LAMB2, WT1, NPHS1, NPHS2
	Individual Early Onset Nephrotic Syndrome tests: Only order single gene tests when not ordering the panel. ☐ 711 ACTN4 DNA Sequencing Test ☐ 712 TRPC6 DNA Sequencing Test ☐ 718 PLCE1 DNA Sequencing Test ☐ 713 WT1 DNA Sequencing Test ☐ 714 LAMB2 DNA Sequencing Test ☐ 710 NPHS2 DNA Sequencing Test ☐ 730 NPHS1 DNA Sequencing Test	
☐ **717**	**Focal and Segmental Glomerulosclerosis (FSGS) Evaluation**	INF2, ACTN4, TRPC6, NPHS2
	Individual FSGS single gene tests: Only order single gene tests when not ordering the panel. ☐ 716 INF2 DNA Sequencing Test ☐ 710 NPHS2 DNA Sequencing Test	
Polycystic Kidney Disease: Molecular Genetics		
☐ 728	PKDx® Familial Mutation Evaluation Proband Accession # _____ Relationship _____	Does not qualify for the Advance Pay Option. PKD1 and PKD2 Variants
☐ **8100**	**Complete PKDx Evaluation**	Does not qualify for the Advance Pay Option.
	Individual PKDx single gene tests: Only order single gene tests when not ordering the panel. ☐ 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	Does not qualify for the Advance Pay Option.

Test Code	Test Name	Additional Information (Genes, Antibodies, Comments)
Other Cystic Diseases: Molecular Genetics		
☐ **1131**	**Complete Tuberous Sclerosis Sequencing and CNV Evaluation**	TSC1 & TSC2
	Individual Tuberous Sclerosis single gene tests: Only order single gene tests when not ordering the panel. ☐ 1236 TSC1 CNV Test ☐ 1254 TSC2 CNV Test ☐ 508 TSC1 Deletion Analysis (for NYS Only) ☐ 524 TSC2 DNA Deletion Test (for NYS Only) ☐ 1245 TSC1 Sequencing Test ☐ 1255 TSC2 Sequencing Test	
☐ 523	TSC Familial Mutation Evaluation Proband Accession # _____ Relationship _____	
☐ 770	Hereditary Interstitial Kidney Disease (UMOD) DNA Sequencing Test	
Renal Cancer: Molecular Genetics		
☐ **889**	**Pheochromocytoma Evaluation**	RET, VHL, SDHB
	Individual Pheochromocytoma single gene tests: Only order single gene tests when not ordering the panel. ☐ 813 MEN2 (RET) DNA Sequencing Test ☐ 888 SDHB DNA Sequencing Test ☐ 858 von Hippel-Lindau Syndrome (VHL) DNA Sequencing Test	
☐ 818	MEN1 DNA Sequencing Test	
Renal Cysts and Diabetes: Molecular Genetics		
☐ 776	HNF1ß DNA Sequencing and Deletion Evaluation (RCAD)	
Rickets: Molecular Genetics		
☐ **857**	**Hypophosphatemic Rickets Evaluation**	PHEX, FGF23
	Individual Hypophosphatemic Rickets single gene tests: Only order single gene tests when not ordering the panel. ☐ 856 FGF23 (Hypophosphatemic Rickets) DNA Sequencing Test ☐ 855 PHEX (Hypophosphatemic Rickets) DNA Sequencing Test	

ENDOCRINE GENETIC TESTING

Test Code	Test Name	Additional Information (Genes, Antibodies, Comments)
Adrenal Disorders: Molecular Genetics		
☐ **816**	**Primary Adrenal Insufficiency Evaluation**	ABCD1, NR0B1, AIRE
	Individual Primary Adrenal Insufficiency single gene tests: Only order single gene tests when not ordering the panel. ☐ 815 ABCD1 (Adrenoleukodystrophy) DNA Sequencing Test ☐ 812 Autoimmune Polyglandular Syndrome (AIRE) Evaluation ☐ 814 NR0B1 (Adrenal Hypoplasia Congenita) DNA Sequencing Test	
☐ **879**	**Congenital Adrenal Hyperplasia (CAH) Evaluation**	Includes CYP21A2 sequencing and deletion, CYP11B1 sequencing
	Individual CAH single gene tests: Only order single gene tests when not ordering the panel. ☐ 875 CYP11B1 (CAH) DNA Sequencing Test ☐ 880 CYP21A2 (CAH) Evaluation ☐ 1180 CYP21A2 Deletion Only Test	Required for tests 879, 880, 875: Indication for Study (check one or more below): ☐ Family history of CAH ☐ Virilization (ambiguous genitalia) ☐ Salt Wasting ☐ Parent/sibling of CAH patient ☐ 17-hydroxyprogesterone (17-OHP) elevated concentration in serum ☐ Other _____
☐ 877	CYP17A1 DNA Sequencing Test	
☐ 881	Endocrine Hypertension (HSD11B2) Evaluation	
☐ 878	HSD3B2 DNA Sequencing Test	
☐ 874	Lipoid CAH (STAR) DNA Sequencing Test	
Bone Diseases: Molecular Genetics		
☐ **860**	**Osteogenesis Imperfecta Evaluation**	COL1A1, COL1A2
	Individual Osteogenesis Imperfecta single gene tests: Only order single gene tests when not ordering the panel. ☐ 861 COL1A1 (OI) DNA Sequencing Test ☐ 862 COL1A2 (OI) DNA Sequencing Test	
☐ **857**	**Hypophosphatemic Rickets Evaluation**	PHEX, FGF23

Test Code	Test Name	Additional Information (Genes, Antibodies, Comments)
Bone Diseases: Molecular Genetics (Continued)		
	Individual Hypophosphatemic Rickets single gene tests: Only order single gene tests when not ordering the panel. ☐ 856 FGF23 (Hypophosphatemic Rickets) DNA Sequencing Test ☐ 855 PHEX (Hypophosphatemic Rickets) DNA Sequencing Test	
☐ 811	LRP5 (OPPG) DNA Sequencing Test	
☐ 821	LRP5 Idiopathic Osteoporosis (IOP) DNA Sequencing Test	
Congenital Hyperinsulinism: Molecular Genetics		
☐ **819**	**Congenital Hyperinsulinism Evaluation**	Does not qualify for the Advance Pay Option. GLUD1, GCK, KCNJ11, ABCC8 Indication for Study (check one or more below): ☐ Diazoxide Responsive ☐ Diazoxide Non-Responsive ☐ Hypoglycemic ☐ Large for Gestational Age (LGA) ☐ Other (describe) _____
	Individual Congenital Hyperinsulinism single gene tests: Only order single gene tests when not ordering the panel. **Single gene tests for the CH Panel, do not qualify for the Advance Pay Option.** ☐ 822 GLUD1 (CHI) DNA Sequencing Test ☐ 823 GCK (CHI) DNA Sequencing Test ☐ 826 KCNJ11 (CHI) DNA Sequencing Test ☐ 827 ABCC8 (CHI) DNA Sequencing Test	
☐ 42	CH Parental Testing – To augment child/proband diagnosis	Does not qualify for the Advance Pay Option. **For expedited diagnosis of proband, send parental testing samples as soon as possible and provide information below.** ☐ Mother ☐ Father Proband Name/Accession #_____

Important: Please be sure to write in the test code(s) and test name(s) within the Test Information section on page 1.

Reflexive testing is performed at an additional charge.

The Advance Pay Option is accepted for all Molecular Genetics test codes that do not have an Immunology or STAT component. These test codes will be noted as not qualifying for Advance Pay in the Additional Information (Genes, Antibodies, Comments) Columns below.

athena diagnostics

MOLECULAR GENETICS SPECIMEN REQUIREMENTS: Specimen Type = Blood, Volume = 8 mL, Tube Type = Lavender Top.
NOTE1: Saliva is acceptable for most genetic tests. Call Athena at 1-800-394-4493 to order Saliva kits and see the Additional Information Column for exceptions.
NOTE2: The pediatric minimum is 2 mL for Neurodevelopmental Disorders & Epilepsy.
NOTE3: Copy Number Variant (CNV) is equivalent to Deletion and Duplication.

IMMUNOLOGY SPECIMEN REQUIREMENTS: Specimen Type = Serum, Volume = 2 mL, Tube Type = Serum Separator Tube.
Please Refer to the Additional Information (Genes, Antibodies, Comments) Column below for specimen requirement exceptions.

Test Code	Test Name	Additional Information (Genes, Antibodies, Comments)
Diabetes: Molecular Genetics		
☐ 885	Monogenic Diabetes (MODY) Five-Gene Evaluation	HNF1A (TCF1), GCK, HNF4A, HNF1B (TCF2), IPF1
☐ 8800	Monogenic Diabetes (MODY) Four-Gene Evaluation	HNF1A (TCF1), GCK, HNF4A, HNF1B (TCF2)
☐ 8801	Monogenic Diabetes (MODY) Three-Gene Evaluation	HNF1A (TCF1), GCK, HNF1B (TCF2)
☐ 8802	Monogenic Diabetes (MODY) Two-Gene Evaluation	HNF1A (TCF1), GCK
☐ 803	GCK (MODY2) DNA Sequencing and Deletion Test	
☐ 802	HNF4A (MODY1) DNA Sequencing and Deletion Test	
☐ 834	IPF1 (MODY4) DNA Sequencing Test	
☐ 804	TCF1 (MODY3) DNA Sequencing and Deletion Test	HNF1A (TCF1)
☐ 805	TCF2 (MODY5) DNA Sequencing and Deletion Test	HNF1B (TCF2)
☐ 837	CEL (MODY8) Mutation Analysis	
☐ **882**	**Neonatal Diabetes Mellitus Evaluation**	IPF1, GCK, KCNJ11, INS, ABCC8
Individual Neonatal Diabetes Mellitus single gene tests: Only order single gene tests when not ordering the panel. ☐ 876 ABCC8 (NDM) DNA Sequencing Test ☐ 842 GCK (NDM) DNA Sequencing Test ☐ 853 INS (NDM) DNA Sequencing Test ☐ 841 IPF1 (NDM) DNA Sequencing Test ☐ 843 KCNJ11 (NDM) DNA Sequencing Test		
Nephrogenic Diabetes: Molecular Genetics		
☐ **854**	**Nephrogenic Diabetes Insipidus Evaluation**	AVPR2, AQP2
Individual Nephrogenic Diabetes Mellitus single gene tests: Only order single gene tests when not ordering the panel. ☐ 852 AQP2 (Nephrogenic Diabetes Insipidus) DNA Sequencing Test ☐ 851 Nephrogenic Diabetes Insipidus (AVPR2) DNA Sequencing Test		
Familial Cancer Syndromes: Molecular Genetics		
☐ 818	MEN1 DNA Sequencing Test	
☐ **889**	**Pheochromocytoma Evaluation**	RET, VHL, SDHB
Individual Pheochromocytoma single gene tests: Only order single gene tests when not ordering the panel. ☐ 813 MEN2 (RET) DNA Sequencing Test ☐ 888 SDHB DNA Sequencing Test ☐ 858 von Hippel-Lindau Syndrome (VHL) DNA Sequencing Test		
Familial Hypocalciuric Hypercalcemia: Molecular Genetics		
☐ 829	Familial Hypocalciuric Hypercalcemia (CASR) DNA Sequencing Test	
Family Testing		
☐ **185**	**Familial DNA Sequence Evaluation** Proband Accession # _____ Relationship _____	This test detects previously identified sequence variants in at-risk family members.

Test Code	Test Name	Additional Information (Genes, Antibodies, Comments)
Noonan Syndrome: Molecular Genetics		
☐ 846	Noonan Syndrome (PTPN11) DNA Sequencing Test	
☐ **658**	**KRAS/RAF1/SOS1 DNA Sequencing Evaluation**	SOS1, RAF1, KRAS
Individual KRAS/RAF1/SOS1 single gene tests: Only order single gene tests when not ordering the panel. ☐ 664 KRAS DNA Sequencing Test ☐ 663 RAF1 DNA Sequencing Test ☐ 662 SOS1 DNA Sequencing Test		
Obesity: Molecular Genetics		
☐ **884**	**Early Onset Obesity Evaluation**	LEPR, MC4R
Individual Early Onset Obesity single gene tests: Only order single gene tests when not ordering the panel. ☐ 640 Early Onset Obesity (MC4R) DNA Sequencing Test ☐ 883 Early Onset Obesity (LEPR) DNA Sequencing Test		
☐ **887**	**Bardet-Biedl Syndrome Evaluation**	BBS1, BBS2, BBS10
Individual Bardet-Biedl Syndrome single gene tests: Only order single gene tests when not ordering the panel. ☐ 871 BBS1 (BBS) DNA Sequencing Test ☐ 872 BBS2 (BBS) DNA Sequencing Test ☐ 886 BBS10 (BBS) DNA Sequencing Test		
Reproductive Disorders: Molecular Genetics		
☐ **679**	**Complete Kallmann/IHH Evaluation**	
Individual Kallmann/IHH single gene tests: Only order single gene tests when not ordering the panel. ☐ 461 CHD7 DNA Sequencing Test ☐ 195 FGF8 DNA Sequencing Test ☐ 196 FGFR1 DNA Sequencing Test ☐ 343 GnRH1 DNA Sequencing Test ☐ 279 GnRHR DNA Sequencing Test ☐ 173 KAL1 DNA Sequencing Test ☐ 364 KISS1R DNA Sequencing Test ☐ 175 PROK2 DNA Sequencing Test ☐ 180 PROKR2 DNA Sequencing Test ☐ 358 TACR3 DNA Sequencing Test		
☐ **462**	**Anosmic Kallmann/IHH Evaluation**	Please see website for the complete list of genes.
☐ **667**	**Normosmic Kallmann/IHH Evaluation**	
☐ 817	Male Precocious Puberty (LHCGR) DNA Sequencing Test	
Short Stature: Molecular Genetics		
☐ **865**	**Combined Pituitary Hormone Deficiency Evaluation**	PROP1, POU1F1
Individual Pituitary Hormone Deficiency single gene tests: Only order single gene tests when not ordering the panel. ☐ 864 POU1F1 (CPHD) DNA Sequencing Test ☐ 863 PROP1 (CPHD) DNA Sequencing Test		
☐ **848**	**Growth Hormone Deficiency Evaluation**	GH1 and GHRHR Seq.; SHOX Seq. and Del.
Individual Growth Hormone Deficiency single gene tests: Only order single gene tests when not ordering the panel. ☐ 866 GH1 (GHD) DNA Sequencing Test ☐ 868 GHRHR (GHD) DNA Sequencing Test ☐ 847 SHOX (GHD) DNA Sequencing and Deletion Test		
☐ 867	GHR DNA Sequencing Test	

For Specimen Collection Services, Please Fax this Test Requisition to 1.610.271.6085

For additional information on test-specific genes and requirements (preferred volume, specimen, and tube type), please visit AthenaDiagnostics.com or call 1.800.394.4493.

Reviewed December 2023