

Athena Diagnostics Client Test Requisition

Client Services is available Monday through Friday from 8:30 AM to 7:00 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. NOTE 1: Athena Diagnostics must bill hospitals directly for all Medicare hospital inpatient and outpatient testing.

NOTE 2: Please complete each section in full. Missing information will delay your patient's testing

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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, which is 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, which is the patient consistent with local state regulatory requirements for the test ordered. 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 r	equest.			
Medical Practitioner Signature:		_ Date	//	/
Medical Practitioner Credentials:		_	Reviewed De	ecember 2025

Client Services is available Monday through Friday from 8:30 AM to 7:00 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.

NOTE 1: Athena Diagnostics must bill hospitals directly for all Medicare hospital inpatient and outpatient testing.

NOTE 2: Please complete each section in full. Missing information will delay your patient's testing.

Clinical Informatio	n		
Clinical diagnosis:			
Age at Initial Presenta	tion:		
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 o	ne):	
☐ Diagnostic (symptor	matic) 🗆 Predictiv	ve (asymptomatic)	☐ Prenatal (Contact Athena prior to sending)
☐ Carrier	☐ Family t	esting/single site	
Relationship to Probar	nd:		
If performed at Athena	ı, provide relative's acce	ession #	
If performed at anothe	r lab, a copy of the relat	rive's report is required.	
Please attach detailed	I medical records and fa	amily history informatio	n.
Specimen Informa	tion		
Specimen Type: Date	sample obtained:	////	
☐ Whole Blood	☐ Serum	☐ Cerebrospinal Fluid	(CSF) CVS: Cultured
☐ Amniotic Fluid: Cult	ured	☐ Saliva (Not available	e for all tests)
☐ DNA* source:		Concentration	nug/ml
*DNA must be extracted	d at a CLIA-certified or a l	aboratory meeting equiv	alent requirements (as determined by CAP and/or CMS).
· ·	vide specimen type): <u>r to sending</u> specimen t		
If not collected same of	lay as shipped, how was	s sample stored? 🔲 Ro	oom temp 🔲 Refrigerated 🔲 Frozen
History of □ blood tra	nsfusion or 🗌 bone ma	arrow transplant? 🗌 Ye	s 🗆 No
Date of most recent tra	ansfusion/transplant:_	//	

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.



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)		Test Name	Additional Information (Genes, Antibodies, Comments)
Cerebro	vascular Disease (Stroke): Molecular Gener	tics	Epilepsy	r: Molecular Genetics (Continued)	
□ 1175	Notch3 (CADASIL) Sequencing Test		□ 603	38 Infantile Spasms	Test 6000 contains all genes included in the sub-panels.
□ 1149	HTRA1 (CARASIL) Sequencing Test		☐ 60°	19 Intellectual Disability	NOTE: Only select sub-panels if 6000 is
□ 1120	COL4A1 Sequencing Test (CSVD)		□ 602	22 Neuronal Ceroid Lipofuscinosis	not ordered. Please see website for the list of genes
□ 1122	Complete CCM Sequencing and CNV			33 Syndromic Disorders	in each panel
	Evaluation			Complete Tuberous Sclerosis Sequencing and CNV Evaluation	Full Sequencing of TSC1 & TSC2
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		Individual Tuberous Sclerosis single gene tests: Only order single gene tests when not ordering the panel. ☐ 1236 TSC1 CNV Test ☐ 508 TSC1 Deletion Analysis (for NYS Only) ☐ 524 TSC2 DNA Deletion Test (for NYS Only)			
	a: Molecular Genetics				255 TSC2 Sequencing Test
	9 ADmark® ApoE Genotype Analysis & Interpretation (Symptomatic for Dementia)			TSC Familial DNA Seq. Mutation Evaluation Proband Accession # Relationship	
□ 179	ADmark® Early-Onset Alzheimer's Evaluation	PSEN1, APP Seq./Dup., PSEN2		SCN1A Seg. and CNV Evaluation	
	Il ADmark® Early-Onset Alzheimer's single gen	ne tests:	Individua	I SCN1A tests:	
	er single gene tests when not ordering the panel. 8 ADmark® APP DNA Sequencing Test and I	Dunlication Test			37 SCN1A Deletion Test
□ 16	7 ADmark® PSEN1 DNA Sequencing Test	Suplication rest	□ 1036	ARX Seq. and CNV Evaluation (Epilepsy)	
□ 16	9 ADmark® PSEN2 DNA Sequencing Test		□ 1115	CDKL5 Seq. and CNV Evaluation (Epilepsy)	
□ 281	Frontotemporal Dementia (FTD)	MAPT, GRN, C9orf72	□ 4411	SLC2A1 DNA Sequencing Test	
Table 18 Table	Evaluation		□ 1003	GFAP (Alexander Disease) Seq. Test	
Individual FTD single gene tests: Only order single gene tests when not ordering the panel. 209 C9orf72 (FTD) DNA Test		□ 443	POLG DNA Seq. Test (Alpers Syndrome)		
		Epilepsy	r: Immunology		
	5 MAPT DNA Sequencing Test		□ 5120	Autoimmune Epilepsy Evaluation	GAD65, VGKC, CASPR2, LGI1, NMDA
Dementia: Immunology □ 5209 ADmark® Alzheimer's Evaluation, CSF (FDA Cleared) Collection Instructions: Perform lumbar puncture (LP) using gravity drip collection method prior to 12 PM. Avoid the use of syringes or tubings. Do not use the first 2 mL of CSF for AD Biomarker measurement. Specimen Requirements: 1 mL (0.7 mL minimum) of CSF directly into		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			
		the CSF tube 63.614.625 (Sarstedt). <u>Transport Requirements:</u> Ship on cold packs or frozen; Keep sample at 2-8° C during transport and storage up to the time of measurement.	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 #
	Autoimmune Rapidly Progressive				Relationship
	Dementia Evaluation with Recombx®		Immunol	logy: Anti-Drug Antibody	
	Il Autoimmune Dementia single antibody tests: er single autoantibody tests when not ordering the			AAV9 Antibody Test	Does not qualify for the Advance Pay Option.
17	14 Recombx® Hu Autoantibody Test* 1716	Recombx [®] MaTa Autoantibody Test*		strophy: Molecular Genetics	FIF2D4 FIF2D2 FIF2D2 FIF2D4
	17 Recombx [®] CV2 Autoantibody Test* ☐ 1718 05 GAD65 Autoantibody Test ☐ 1708	Recombx® Amphiphysin Autoantibody Test* NMDA Receptor Autoantibody Test*		Leukoencephalopathy with Vanishing White Matter Evaluation	EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5
		B LGI1 Autoantibody Test*	Individua	I Leukoencephalopathy with Vanishing White I	Matter single gene tests:
☐ 1709 CASPR2 Autoantibody Test*			er single gene tests when not ordering the panel.		
* NOTE: Cerebrospinal Fluid (CSF) is an acceptable sample type for these tests.				6102 EIF2B2 DNA Sequencing Test	
Epilepsy: Molecular Genetics			03 EIF2B3 DNA Sequencing Test 05 EIF2B5 DNA Sequencing Test] 6104 EIF2B4 DNA Sequencing Test	
6000	Epilepsy Advanced Sequencing and CNV Evaluation	Test 6000 contains all genes included in the sub-panels.		PLP1 Sequencing and CNV Evaluation	
□ 60	18 Developmental Brain Malformations	·		ABCD1 DNA Sequencing Test	
□ 60	23 Epilepsy with Migraine	NOTE: Only select sub-panels if 6000 is not ordered.		ARSA DNA Sequencing Test	
□ 60	10 Epileptic Encephalopathy	Please see website for the list of genes		GJC2 DNA Sequencing Test	
□ 60	08 Generalized, Absence, Focal, Febrile and Myoclonic Epilepsies	in each panel.		Notch3(CADASIL) Sequencing Test	

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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 Test Code Name	Additional Information (Genes, Antibodies, Comments)	Test Test Code Name	Additional Information (Genes, Antibodies, Comments)
Migraine: Molecular Genetics		Movement Disorders: Molecular Genetics (Continu	ed)
☐ 1148 Hemiplegic Migraine Sequencing Evaluation	CACNA1A, ATP1A2, SCN1A	Individual Ataxia single gene DNA Tests: Only order single gene tests when not ordering the panel	
Individual Hemiplegic Migraine single gene tests:		or sub-panels. ☐ 401 DRPLA ☐ 119 FRDA/FXN Expansion)	
Only order single gene tests when not ordering the panel.		☐ 348 FRDA/FXN Seq. ☐ 383 POLG1 (MIRAS)	SCA8 and SCA10 test cannot be
☐ 1101 ATP1A2 Sequencing Test	1103 CACNA1A Sequencing Test	☐ 371 SCA1 (ATXN1) ☐ 672 SCA2 (ATXN2)	performed on saliva.
☐ 1136 SCN1A Sequencing Test (FHM)	and a second second second	☐ 105 SCA3 (ATXN3) ☐ 373 SCA6 (CACNA1A)	
, , ,		☐ 677 SCA7 (ATXN7) ☐ 384 SCA8 (ATXN8OS)	
Motor Neuron Diseases: Molecular Genetics G520 Amyotrophic Lateral Sclerosis Advanced		☐ 387 SCA10 (ATXN10) ☐ 285 SCA12 (PPP2R2B)	
Evaluation	Please see website for the complete list	☐ 388 SCA17 (TBP) ☐ 283 TTPA (AVED) ☐ 402 Chorea Differential Evaluation (DRPLA,	Cannot be performed on saliva.
☐ 6522 Nonprevalent Amyotrophic Lateral Sclerosis Advanced Sequencing Evaluation	of genes.	Huntington's Disease) 116 Huntington Disease Repeat Expansion Test	Cannot be performed on saliva.
☐ 670 C9orf72 DNA Test		☐ 639 Isolated Dystonia Evaluation	DYT1, THAP1
☐ 620 SOD1 DNA Sequencing Test		Individual Isolated Dystonia single gene tests:	
☐ 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	Only order single gene tests when not ordering the panel. G26 Dystonia (DYT1) DNA Test HAP1 DNA Sequencing Test	
	not ordered.	☐ 629 Complete Dopa-Responsive Dystonia	00140 00145 1 7110
☐ 6601 HSP, Common Sporadic Evaluation	SPAST, SPG7	(DYT5) Evaluation	GCH1 Seq., GCH1 Del., TH Seq.
☐ 6602 HSP, Supplemental Sporadic Evaluation	Please see website for the complete list	Individual Dopa-Responsive Dystonia single gene tests:	
☐ 6610 HSP, Complete Dominant Evaluation	of genes.	Only order single gene tests when not ordering the panel. Gamma 637 GCH1 DNA Sequencing Test	DYT5A
☐ 6611 HSP, Common Dominant Evaluation	SPAST, ATLN, REEP1, KIF5A	☐ 638 GCH1 Deletion Analysis	DYT5A
☐ 6612 HSP, Supplemental Dominant Evaluation	BSCL2, HSPD1, KIAA0196, NIPA1, RTN2, SLC33A1	☐ 634 TH DNA Sequencing Test	DYT5B
☐ 6620 HSP, Complete Recessive Evaluation	Please see website for the complete list	☐ 624 SGCE DNA Sequencing Test	DYT11
0020 Tion, complete recessive Evaluation	of genes.	☐ 627 SGCE Deletion Analysis	DYT11
☐ 6621 HSP, Common Recessive Evaluation	SPG11, ZFYVE26, SPG7	☐ 617 PNKD (MR-1) DNA Sequencing Test ☐ 588 Complete Parkinsonism Evaluation	LRRK2, PARK2, PINK1, PARK7, SNCA
☐ 6622 HSP, Supplemental Recessive Evaluation	Please see website for the complete list	Individual Parkinsonism single gene tests:	
	of genes.	Only order single gene tests when not ordering the panel.	050 411 0 1: (0104) 5 (517)
☐ 6631 HSP, X-Linked Evaluation	L1CAM, PLP1		059 Alpha Synuclein (SNCA) Dup /Del. Test 559 PARK2 (Parkin) DNA Sequencing Test
☐ 6509 SPG4 Evaluation	SPAST	☐ 040 PARK2 (Parkin) Duplication/Deletion Test ☐	554 PARK7 (DJ1) DNA Sequencing Test
Movement Disorders: Molecular Genetics		☐ 047 PARK7 (DJ1) Deletion Test ☐ 058 PINK1 Deletion Test	542 PINK1 DNA Sequencing Test
Individual HSP DNA Tests:		☐ 1187 PRRT2 (Dyskinesia/IC) Seq. Test	
Only order single gene tests when not ordering the panel.	SPG3A	Multiple Sclerosis/Demylenating Diseases: Immuno	plogy
☐ 531 Atlastin	SPG7 SPG11	☐ 1287 NMO Spectrum Evaluation	AQP4, CBA reflex to MOG, CBA
G32 Paraplegin	SPG15	☐ 1282 Aquaporin-4 (AQP4) (NMO IgG) Antibody,	Cerebrospinal Fluid (CSF) is an
G33 Spatacsin		CBA with Reflex to Titer	acceptable sample type.
☐ 614 ZFYVE26 ☐ 117 Kennedy's Disease (SBMA) DNA Test		☐ 1523 Myelin Oligodendrocyte Glycoprotein (MOG) Antibody, CBA with Reflex to Titer	Cerebrospinal Fluid (CSF) is an acceptable sample type.
☐ 6930 Ataxia, Comprehensive Evaluation	Please see website for the complete list	☐ 1284 NMO Spectrum Evaluation	AQP4, ELISA reflex to MOG, CBA
,	of genes. Test 6930 contains all genes included in the sub-panels.	☐ 193 Aquaporin-4 (AQP4) Antibody (NMO-IgG), ELISA	
	NOTE: Only select sub-panels if 6930 is	☐ 112 NAbFeron® (INFB-1) Neutralizing Antibody Test	
	not ordered. Cannot be performed on saliva.	☐ 197 TYSABRI® (Natalizumab) Antibody Test	See website for collection notes
COOO Ata 'a Canadata Bandarat E al al'an		Myasthenia Gravis: Immunology	
☐ 6900 Ataxia, Complete Dominant Evaluation ☐ 6901 Ataxia, Common Repeat Expansion	Please see website for the complete list	☐ 1521 Myasthenia Gravis Panel 2 with Reflex to MuSK Antibody	
Evaluation Geographic Geographics Evaluation Geographics Geographics Evaluation Geographics Geographics	of genes. Cannot be performed on saliva.	☐ 1514 Myasthenia Gravis Panel 2	Includes AChR Binding / Blocking / Modulating Antibody
Evaluation	Carnot be performed on saliva.	☐ 1490 MuSK and LRP4	
☐ 6910 Ataxia, Complete Recessive Evaluation		☐ 1510 Acetylcholine Receptor Binding Antibody with Reflex to Musk Antibody	
☐ 6911 Ataxia, Supplemental Recessive Evaluation	Please see website for the complete list of genes.	☐ 1511 Acetylcholine Receptor Binding Antibody with Reflex to MuSK/LRP4 Antibodies	
☐ 6912 Oculomotor Apraxia Ataxia Advanced Sequencing Evaluation	APTX, SETX	Individual Myasthenia Gravis single antibody tests:	
☐ 6920 Episodic Ataxia Evaluation	CACNB4, KCNA1, SLC1A3, CACNA1A	Only order single antibody tests when not ordering the 1513 Acetylcholine Receptor Binding Antibody	corresponding panel option(s). 1483 LRP4 Autoantibody Test
☐ 349 Ataxia, Friedreich (FXN) Evaluation	FRDA/FXN Seq., FRDA/FXN Expansion	☐ 1516 Acetylcholine Receptor Blocking Antibody	☐ 1481 RyR Autoantibody Test
☐ 353 Ataxia-Telangiectasia (ATM) Evaluation	ATM Seq., ATM Dup./Del.	☐ 1517 Acetylcholine Receptor Modulating Antibody ☐ 482 MuSK Antibody Test	☐ 1480 Titin Autoantibody Test

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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)	Test Code	Test Name	Additional Information (Genes, Antibodies, Comments)
	evelopmental Disorders: Molecular Genetic	, ,	Neurom	uscular Disorders: Molecular Genetics (Con	, , ,
1186	Primary Microcephaly Sequencing Evaluation			OPMD Repeat Expansion Test	Cannot be performed on saliva.
Individua	al Primary Microcephaly single gene tests:			OPA1 DNA Sequencing Test (optic atrophy)	Related to optic atrophy.
	al Primary Microcephaly single gene tests: er single gene tests when not ordering the panel. 92 ASPM Sequencing Test	153 MCPH1 Sequencing Test		Oncology: Molecular Genetics	Treated to optic attorny.
<u> </u> 12	57 WDR62 Sequencing Test			Neurofibromatosis Type 1 (NF1)	
<u> </u>	SHANK3 Sequencing Test		L 040	Evaluation	NF1 Sequencing, NF1 Deletion
	SHANK2 Sequencing Test		Individus	al NF1 single gene tests:	
	PTEN Sequencing Test Joubert Syndrome Evaluation			er single gene tests. er single gene tests when not ordering the panel.	
Individua	al Jouhert Syndrome single gene tests:			7 Neurofibromatosis Type 1 Deletion Test	
Only orde	er single gene tests when not ordering the panel.			6 Neurofibromatosis Type 1 DNA Sequencin	g Test
79	0 AHI1 DNA Sequencing Test ☐ 79 1 CEP290 DNA Sequencing Test ☐ 79		□ 645	Neurofibromatosis Type 2 (NF2)	
78	9 TMEM216 DNA Sequencing Test 79	92 TMEM67 DNA Sequencing Test		Evaluation	NF2 Seq., NF2 Dup./Del.
	MECP2 Sequencing and CNV Evaluation		Individua	al NF2 single gene tests:	
☐ 11°	14 CDKL5 Seq. and CNV Evaluation		Only orde	er single gene tests when not ordering the panel.	
	(Atypical Rett) 8 Rett Syndrome (MECP2) Dup./Del. Test			Neurofibromatosis Type 2 DNA Sequencin	
	Smith-Lemli-Opitz Syndrome (DHCR7)			4 Neurofibromatosis Type 2 Duplication/Dele	
131	DNA Sequencing Test		Paraneo	oplastic & Other Antibody Disorders of the C	
□ 1256	VPS13B (COH1) Sequencing Test		□ 4711	Paraneoplastic Neurological Syndromes	Cerebrospinal Fluid (CSF) is an
□ 1038	ARX Seq. and CNV Evaluation (Intellectual			Evaluation with Recombx®, Initial Assessment	acceptable sample type. Amphiphysin, CV2, Hu, MaTa, Ri, Yo
	Disability)		□ 4620	NeoComplete Paraneoplastic Evaluation	Amprilphysin, CV2, Hu, Mara, Ni, 10
	SYNGAP1 Sequencing Test MEF2C Sequencing and CNV Evaluation		4020	with Recombx®	
	FOXG1 Sequencing and CNV Evaluation		□ 4640	Paraneoplastic Autoantibody Evaluation with	* NOTE: Cerebrospinal Fluid (CSF) is an
	uscular Disorders: Molecular Genetics			Recombx®, CSF *	acceptable sample type
	Muscular Dystrophy Advanced Evaluation		□ 4724	NeoCerebellar Degeneration Paraneoplastic	Diagon and website for the complete list
□ 5502	Congenital Muscular Dystrophy Advanced			Profile with Recombx®	Please see website for the complete list of antibodies.
	Sequencing Evaluation		□ 4722	NeoEncephalitis Paraneoplastic Evaluation	of antibodics.
5503	Congenital Myopathy Advanced Sequencing Evaluation	Please see website for the complete list		with Recombx®	
□ 5504	Distal Myopathy Advanced Sequencing	of genes.	□ 4725	NeoSensory Neuropathy Paraneoplastic	Cerebrospinal Fluid (CSF) is an
	Evaluation			Profile with Recombx®	acceptable sample type.
□ 5505	Myofibrillar Myopathy Advanced Sequencing Evaluation			<u> </u>	Amphiphysin, CV2, Hu
□ 5506	Myotonic Syndromes Advanced Evaluation	Please see website for the complete list		Neuromyotonia Evaluation	CASPR2, VGKC
	mystoriis syriaisiniss ravanssa Evaldation	of genes.		dual antibody Tests: order single antibody tests when not ordering t	the corresponding panel option(s)
		Cannot be performed on saliva.	,	• ,	☐ 4681 Recombx® CV2 Autoantibody Test *
□ 5507	Periodic Paralysis Advanced Sequencing Evaluation			GAD65 Neurological Syndrome Antibody Test	
□ 5508	Malignant Hyperthermia Advanced				4683 Recombx [®] MaTa Autoantibody Test *
	Sequencing Evaluation				4684 Recombx® CAR (Anti-Recoverin)
□ 5511	Congenital Myasthenic Syndrome Advanced Sequencing Evaluation	Please see website for the complete list		VGCC Type P/Q Autoantibody Test (LEMS)	Autoantibody Test *
☐ 5518	Emery-Dreifuss Muscular Dystrophy	of genes.			4685 Recombx® Ri Autoantibody Test *
3310	Advanced Sequencing Evaluation				☐ 4686 Recombx® Yo Autoantibody Test *☐ 4689 Recombx® Zic4 Autoantibody Test *
□ 5519	Limb Girdle Muscular Dystrophy Advanced			Cerebrospinal Fluid (CSF) is an acceptable sa	•
	Evaluation		NOTE.	Corospinar ridia (COI) is an acceptable so	ample type for these tests.
Individua	al Limb Girdle Muscular Dystrophy Tests: er single gene tests when not ordering the par	201	Periphe	ral Neuropathy (Hereditary): Molecular Gen	etics
		584 CAPN3 Duplication/Deletion Test	4001	CMT Advanced Evaluation	Testing is performed in this order:
		562 FKRP DNA Sequencing Test		Comprehensive (Reflexive)	1. PMP22 Dup./Del. If negative: 2. Cx32,
		582 SGCA Duplication/Deletion Test			PMP22, MFN2, MPZ, EGR2, LITAF, PRX, GDAP1, RAB7, GARS, NFL,
	3 SGCG Duplication/Deletion Test	·			HSPB1, LMNA, FIG4, SH3TC2, DNM2,
<u>5530</u>	DMD Evaluation				YARS, FGD4, NDRG1, TRPV4, HSPB8,
Only orde	al DMD Evaluation single gene tests: er single gene tests when not ordering the panel.				MTMR2, SBF2 DNA Seq.
l	3 DMD DNA Seguencing Test		□ 40	002 CMT Advanced Evaluation – Dominant,	Testing is performed in this order: 1.
□ 55	31 DMD Duplication/Deletion Test			Demyelinating (Reflexive)	PMP22 Dup./Del. If negative: 2. MPZ,
□ 207	Early-Onset Myotonia Evaluation	DM1, CLCN1, SCN4A			PMP22 Seq., EGR2, LITAF, DNM2,
1 11 11	15.10.14.1.1.1	Cannot be performed on saliva.		NOO OMTA I are also also also also also also also also	YARS DNA Seq.
	al Early-Onset Myotonia single gene tests: er single gene tests when not ordering the panel.			103 CMT Advanced Evaluation – Dominant, Axonal	Diogno and wobaits for the complete list
	8 CLCN1 DNA Sequencing Test				Please see website for the complete list of genes.
	6 SCN4A (Myotonia) DNA Sequencing Test		40	004 CMT Advanced Evaluation – Recessive, Demyelinating	or gorios.
	DMPK DNA Test (DM1)	Cannot be performed on saliva.	<u> </u>	005 CMT Advanced Evaluation – Dominant	Testing is performed in this order: 1.
	CNBP DNA Test (DM2) (DM2 testing is not	35.55 25.55	1 -10	(Reflexive)	PMP22 Dup./Del. If negative: 2. MFN2,
	recommended for patients with early onset	Cannot be performed on saliva.			MPZ, PMP22 Seq., EGR2, LITAF, RAB7,
	myotonic dystrophy)				GARS, NFL, HSPB1, DNM2, YARS,
	CAPN3 Evaluation	Includes CAPN3 Seq., CAPN3 Del.			TRPV4, HSPB8 DNA Seq.
□ 571	Dysferlin DNA Sequencing Test		40	006 CMT Advanced Evaluation – Recessive	Please see website for the complete list of genes.

Reflexive testing is performed at an additional charge.





MOLECULAR GENETICS SPECIMEN REQUIREMENTS: Specimen Type = Blood, Volume = 8 mL, Tube Type = Lavender Top.

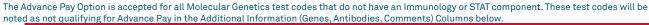
NOTE1: Saliva is acceptable for most genetic tests. Call Athena at 1-800-394-4493 to order Saliva kits and see the Additional Information Column for exceptions. NOTE2: The pediatric minimum is 2 mL for Neurodevelopmental Disorders & Epilepsy.

NOTE3: Copy Number Variant (CNV) is equivalent to Deletion and Duplication.

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

	lease Refer to the Additional Information (G	eperation type and the second	elow for s		
Test Code	Test	Additional Information	Test	Test Name	Additional Information
	Name	(Genes, Antibodies, Comments)	Code		(Genes, Antibodies, Comments)
	eral Neuropathy (Hereditary): Molecular Gen			ral Neuropathy (Autoimmune): Immunology	
	007 CMT Advanced Evaluation – Demyelinating (Reflexive)	Testing is performed in this order: 1. PMP22 Dup./Del. If negative: 2. Cx32,	3100	SensoriMotor Neuropathy Profile with Recombx® - Complete	GM1 Quattro [®] , MAG 'Dual Antigen' [®] , Hu, GALOPTM, Sulfatide
		MPZ, PMP22 Seq., EĞR2, LITAF, PRX, GDAP1, DNM2, YARS, SH3TC2,	□ 3148	Sensory Neuropathy Profile with Recombx®	(MAG 'Dual Antigen'®, Hu, GALOPTM, Sulfatide)
		MTMR2, NDRG1, FGD4, FIG4, SBF2 DNA Seq	□ 3163	Motor Neuropathy Profile - Complete	GM1 Quattro®, MAG 'Dual Antigen'®
	008 CMT Advanced Evaluation – Axonal	51010004	□ 289	Multifocal Motor Neuropathy Evaluation	Requires both Serum and whole blood.
	010 CMT Advanced Evaluation – Initial Genetic Assessment		□ 3155	Co-GM1 Quattro® Autoantibody Test	GM1 Quattro®, PMP22 Dup./Del. (Asialo, GD1a, GD1b and GM1)
□ 4	011 CMT Advanced Evaluation – Nonprevalent Axonal	Please see website for the complete list	Individu	al Peripheral Neuropathy antibody tests:	
<u></u> 4	012 CMT Advanced Evaluation –	of genes.	Only ord	der the single antibody tests when not ordering	the corresponding panel option(s).
	Nonprevalent Demyelinating		3′	127 MAG 'Dual Antigen'® Autoantibody Test [☐ 272 Asialo Autoantibody Test
∐4	013 CMT Advanced Evaluation – Nonprevalent				□ 273 GD1b Autoantibody Test
Individu	ual CMT single gene tests:			•	•
	der single gene tests when not ordering the panel of	or sub-panels.		•	271 GM1 Autoantibody Test
1	43 CX32 Seq./Del. (CMTX) ☐ 253 DNM2	☐ 248 EGR2 (CMT1D)		•	☐ 4682 Recombx® Hu Autoantibody Test *
□ 2	08 FGD4 □ 225 FIG4 (CN	MT4J) ☐ 228 GARS (CMT2D)	□ 27	78 GD1a Autoantibody Test	
	21 GDAP1 (CMT2K, 4A)	*	* NOTE:	: Cerebrospinal Fluid (CSF) is an acceptable sa	ample type for these tests.
	34 MPZ (CMT1B, 2I, 2J) 354 MTMR2	:MT2B1, 4C1)	Spinal I	Muscular Atrophy (SMA): Molecular Genetic	s
		Oup./Del. (CMT1A) 247 PMP22 Seq.	·		
□ 2	39 PRX (CMT4F)		□ 5056	SMA Carrier Screen (New York)	Does not qualify for the Advance Pay Option. Test Codes are for New York State Clients
	35 TTR DNA Sequencing Test ☐ 468 YARS	NTRK1 and WNK1	□ 5026	SMA Diagnostic (New York)	ordering SMA testing. 4 mL (2 mL minimum) whole blood collected in an
					EDTA (lavender-top) tube.
	Complete HNPP Evaluation Congenital Hypomyelination Evaluation	PMP22 Sequencing, PMP22 Dup./Del. MPZ, EGR2	□ 5070	SMA Plus (New York)	Pediatric (0-3 years): 2 mL (1 mL minimum).
☐ 296	<u> </u>	PMP22 Seq., PMP22 Dup./Del., TTR	□ 214	SMA Plus (Reflexive)	
	eral Neuropathy (Hereditary Sensory Autono	omic Neuropathy): Molecular Genetics	□ 111	Spinal Muscular Atrophy-Diagnostic	
	al Early-Onset HSAN single gene tests: der single gene tests when not ordering the panel.				
6	59 NTRK1 (HSAN IV) DNA Sequencing Test		□ 444	Spinal Muscular Atrophy-Carrier	December of facility Advances De
□ 5	53 WNK1 (HSAN II) DNA Sequencing Test		□ 211	Spinal Muscular Atrophy - SMN1 DNA	Does not qualify for the Advance Pay Option.
	Late-Onset HSAN Evaluation	SPTLC1 and SPTLC2		Sequencing Test	'
	ual Late-Onset HSAN single gene tests:		□ 6521	Atypical SMA Advanced Sequencing	Test 214 includes 111 with reflex to 211.
	der single gene tests when not ordering the panel.			Evaluation	
	51 SPTLC1 (HSAN I) DNA Sequencing Test				
	52 SPTLC2 (HSAN I) DNA Sequencing Test				
	ATL1 (HSAN I) DNA Sequencing Test				
<u> </u>	SEPT9 (HNA) DNA Sequencing Test				
		RENAL GENE			
Test Code	Test Name	Additional Information (Genes, Antibodies, Comments)	Test Code	Test Name	Additional Information (Genes, Antibodies, Comments)
Alport	Syndrome: Molecular Genetics		Heredita	ary Renal Tubular Disorders: Molecular Gen	
□ 759	Complete Alport Syndrome Evaluation	COL4A3,4,5 DNA Sequencing; COL4A5 Deletion Test	☐ 767	Hereditary Renal Tubular Disorders Evaluation	SLC12A1, KCNJ1, CLCNKB, BSND, SLC12A3
	ial Alport Syndrome single gene tests:			al Hereditary Renal Tubular Disorder single ger	ne tests:
	der single gene tests when not ordering the panel. 57 COL4A3 DNA Sequencing Test	COL4A4 DNA Sequencing Test		er single gene tests when not ordering the panel.	
	57 COL4A3 DNA Sequencing Test 758 COL4A5 Deletion Analysis 755	COL4A4 DNA Sequencing Test COL4A5 Sequencing and Deletion Analysis			764 CLCNKB DNA Sequencing Test
	idosis: Molecular Genetics	ool in to ooquonomig and personally manyor			762 SLC12A1 DNA Sequencing Test
	TTR DNA Sequencing Test			66 SLC12A3 DNA Sequencing Test	
				CASR DNA Sequencing Test	
	-Biedl Syndrome: Molecular Genetics Bardet-Biedl Syndrome Evaluation	BBS1, BBS2, BBS10	Monoge	enic Hypertension: Molecular Genetics	CONNIAD CONNIAC CVD44D4
Individu	ial Bardet-Biedl Syndrome single gene tests:	DD31, DD32, DD310	□ 749	Monogenic Hypertension Evaluation	SCNN1B, SCNN1G, CYP11B1, HSD11B2
□8		872 BBS2 (BBS) DNA Sequencing Test	□ 747	Liddle's Syndrome Evaluation	SCNN1B, SCNN1G
	86 BBS10 (BBS) DNA Sequencing Test Testing		□ 748	Pseudohypoaldosteronism Type 1 Evaluation	SCNN1A, SCNN1B, SCNN1G
_	Familial DNA Sequence Evaluation	This test detects previously identified		al Monogenic Hypertension single gene tests:	
00	Proband Accession #	sequence variants in at-risk family	Only ord	er single gene tests when not ordering the panel.	
		members. For Familial PKD1 and PKD2		 79 CYP11B1/CYP11B2 Chimeric Gene Fusion 74 CYP11B1 DNA Sequencing Test 	n Test □ 775 HSD11B2 DNA Sequencing Test
	Relationship	variants, please order Code 728.			☐ 745 SCNN1B DNA Sequencing Test

Reflexive testing is performed at an additional charge.





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 (C			
Test Test Code Name	Additional Information (Genes, Antibodies, Comments)	Test Test Code Name	Additional Information (Genes, Antibodies, Comments)
Nephrogenic Diabetes Insipidus: Molecular Geneti		Other Cystic Diseases: Molecular Genetics	(
☐ 854 Nephrogenic Diabetes Insipidus		☐ 1131 Complete Tuberous Sclerosis	
Evaluation	AVPR2, AQP2	Sequencing and CNV Evaluation	TSC1 & TSC2
Individual Nephrogenic Diabetes Insipidus single gene	tests:		
Only order single gene tests when not ordering the panel.		Individual Tuberous Sclerosis single gene tests:	
	51 AVPR2 DNA Sequencing Test	Only order single gene tests when not ordering the panel.	
Nephrotic Syndrome: Molecular Genetics		☐ 1236 TSC1 CNV Test ☐ 1	254 TSC2 CNV Test
☐ 722 Early Onset Nephrotic Syndrome	PLCE1, LAMB2, WT1, NPHS1, NPHS2	☐ 508 TSC1 Deletion Analysis (for NYS Only) ☐ 5	
Evaluation	T LOET, LAWIDZ, WTT, NI TIOT, NI TIOZ		
Individual Early Onset Nephrotic Syndrome tests:			255 TSC2 Sequencing Test
Only order single gene tests when not ordering the panel.		☐ 523 TSC Familial Mutation Evaluation	
i =	712 TRPC6 DNA Sequencing Test	Proband Accession # Relationship	_
	713 WT1 DNA Sequencing Test	'	-
· ·	710 NPHS2 DNA Sequencing Test	☐ 770 Hereditary Interstitial Kidney Disease	
☐ 730 NPHS1 DNA Sequencing Test		(UMOD) ĎNA Sequencing Ťest	
☐ 717 Focal and Segmental Glomerulosclerosis	INF2, ACTN4, TRPC6, NPHS2	Renal Cancer: Molecular Genetics	DET VIII ODLID
(FSGS) Evaluation		□ 889 Pheochromocytoma Evaluation	RET, VHL, SDHB
Individual FSGS single gene tests:		Individual Pheochromocytoma single gene tests:	
Only order single gene tests when not ordering the panel.		Only order single gene tests when not ordering the panel.	
	710 NPHS2 DNA Sequencing Test	☐ 813 MEN2 (RET) DNA Sequencing Test	☐ 888 SDHB DNA Sequencing Test
Polycystic Kidney Disease: Molecular Genetics		☐ 858 von Hippel-Lindau Syndrome (VHL) DNA	Sequencing lest
☐ 728 PKDx® Familial Mutation Evaluation	Does not qualify for the Advance Pay	☐ 818 MEN1 DNA Sequencing Test	
Proband Accession #	Option.	Renal Cysts and Diabetes: Molecular Genetics	
Relationship	PKD1 and PKD2 Variants	☐ 776 HNF1ß DNA Sequencing and Deletion	
☐ 8100 Complete PKDx Evaluation	Does not qualify for the Advance Pay	Evaluation (RCAD)	
·	Option.	Rickets: Molecular Genetics	
Individual PKDx single gene tests:		☐ 857 Hypophosphatemic Rickets Evaluation	PHEX, FGF23
Only order single gene tests when not ordering the panel.		Individual Hypophosphatemic Rickets single gene test	ts:
☐ 8105 PKD1 Deletion Test		Only order single gene tests when not ordering the panel.	
☐ 8101 PKD1 DNA Sequencing and Deletion Evaluation	Does not qualify for the Advance Pay	, , ,	A Coguencina Test
☐ 8103 PKD1 DNA Sequencing Test	Option.	☐ 856 FGF23 (Hypophosphatemic Rickets) DNA	•
☐ 8106 PKD2 Deletion Test		☐ 855 PHEX (Hypophosphatemic Rickets) DNA	Sequencing Test
☐ 8102 PKD2 DNA Sequencing and Deletion Evaluation			
☐ 8102 PKD2 DNA Sequencing and Deletion Evaluation	ENDOCRINE GE	ENETIC TESTING	
□ 8102 PKD2 DNA Sequencing and Deletion Evaluation □ 8104 PKD2 DNA Sequencing Test Test Test	Additional Information	Test Test	Additional Information
□ 8102 PKD2 DNA Sequencing and Deletion Evaluation □ 8104 PKD2 DNA Sequencing Test Test Test Code Name		Test Test Code Name	Additional Information (Genes, Antibodies, Comments)
□ 8102 PKD2 DNA Sequencing and Deletion Evaluation □ 8104 PKD2 DNA Sequencing Test Test Test Code Name Adrenal Disorders: Molecular Genetics	Additional Information (Genes, Antibodies, Comments)	Test Test Code Name Bone Diseases: Molecular Genetics (Continued)	(Genes, Antibodies, Comments)
□ 8102 PKD2 DNA Sequencing and Deletion Evaluation □ 8104 PKD2 DNA Sequencing Test Test Test Code Name Adrenal Disorders: Molecular Genetics □ 816 Primary Adrenal Insufficiency Evaluation	Additional Information (Genes, Antibodies, Comments) ABCD1, NR0B1, AIRE	Test Test Code Name Bone Diseases: Molecular Genetics (Continued) Individual Hypophosphatemic Rickets single gene test	(Genes, Antibodies, Comments)
□ 8102 PKD2 DNA Sequencing and Deletion Evaluation □ 8104 PKD2 DNA Sequencing Test Test Test Code Name Adrenal Disorders: Molecular Genetics □ 816 Primary Adrenal Insufficiency Evaluation Individual Primary Adrenal Insufficiency single gene te	Additional Information (Genes, Antibodies, Comments) ABCD1, NR0B1, AIRE	Test Test Code Name Bone Diseases: Molecular Genetics (Continued) Individual Hypophosphatemic Rickets single gene test Only order single gene tests when not ordering the panel. — 856 FGF23 (Hypophosphatemic Rickets) DNA	ts: A Sequencing Test
□ 8102 PKD2 DNA Sequencing and Deletion Evaluation □ 8104 PKD2 DNA Sequencing Test Test Test Code Name Adrenal Disorders: Molecular Genetics □ 816 Primary Adrenal Insufficiency Evaluation Individual Primary Adrenal Insufficiency single gene te Only order single gene tests when not ordering the panel.	Additional Information (Genes, Antibodies, Comments) ABCD1, NR0B1, AIRE sts:	Test Test Code Name Bone Diseases: Molecular Genetics (Continued) Individual Hypophosphatemic Rickets single gene test Only order single gene tests when not ordering the panel. 856 FGF23 (Hypophosphatemic Rickets) DNA 855 PHEX (Hypophosphatemic Rickets) DNA	ts: A Sequencing Test
□ 8102 PKD2 DNA Sequencing and Deletion Evaluation □ 8104 PKD2 DNA Sequencing Test Test Test Code Name Adrenal Disorders: Molecular Genetics □ 816 Primary Adrenal Insufficiency Evaluation Individual Primary Adrenal Insufficiency single gene te Only order single gene tests when not ordering the panel. □ 815 ABCD1 (Adrenoleukodystrophy) DNA Sec	Additional Information (Genes, Antibodies, Comments) ABCD1, NR0B1, AIRE sts: uencing Test	Test Test Code Name Bone Diseases: Molecular Genetics (Continued) Individual Hypophosphatemic Rickets single gene test Only order single gene tests when not ordering the panel. 856 FGF23 (Hypophosphatemic Rickets) DNA 855 PHEX (Hypophosphatemic Rickets) DNA 811 LRP5 (OPPG) DNA Sequencing Test	ts: A Sequencing Test
□ 8102 PKD2 DNA Sequencing and Deletion Evaluation □ 8104 PKD2 DNA Sequencing Test Test Code Name Adrenal Disorders: Molecular Genetics □ 816 Primary Adrenal Insufficiency Evaluation Individual Primary Adrenal Insufficiency single gene te Only order single gene tests when not ordering the panel. □ 815 ABCD1 (Adrenoleukodystrophy) DNA Sec □ 812 Autoimmune Polyglandular Syndrome (Al	Additional Information (Genes, Antibodies, Comments) ABCD1, NR0B1, AIRE sts: uencing Test RE) Evaluation	Test Test Code Name Bone Diseases: Molecular Genetics (Continued) Individual Hypophosphatemic Rickets single gene test Only order single gene tests when not ordering the panel. 856 FGF23 (Hypophosphatemic Rickets) DNA 855 PHEX (Hypophosphatemic Rickets) DNA 811 LRP5 (OPPG) DNA Sequencing Test 821 LRP5 Idiopathic Osteoporosis (IOP) DNA	ts: A Sequencing Test
□ 8102 PKD2 DNA Sequencing and Deletion Evaluation □ 8104 PKD2 DNA Sequencing Test Test Code Name Adrenal Disorders: Molecular Genetics □ 816 Primary Adrenal Insufficiency Evaluation Individual Primary Adrenal Insufficiency single gene te Only order single gene tests when not ordering the panel. □ 815 ABCD1 (Adrenoleukodystrophy) DNA Sec □ 812 Autoimmune Polyglandular Syndrome (AI □ 814 NROB1 (Adrenal Hypoplasia Congenita) D	Additional Information (Genes, Antibodies, Comments) ABCD1, NR0B1, AIRE sts: uencing Test RE) Evaluation NA Sequencing Test	Test Code Name Bone Diseases: Molecular Genetics (Continued) Individual Hypophosphatemic Rickets single gene test Only order single gene tests when not ordering the panel. □ 856 FGF23 (Hypophosphatemic Rickets) DNA □ 855 PHEX (Hypophosphatemic Rickets) DNA □ 811 LRP5 (OPPG) DNA Sequencing Test □ 821 LRP5 Idiopathic Osteoporosis (IOP) DNA Sequencing Test	ts: A Sequencing Test
□ 8102 PKD2 DNA Sequencing and Deletion Evaluation □ 8104 PKD2 DNA Sequencing Test Test Code Name Adrenal Disorders: Molecular Genetics □ 816 Primary Adrenal Insufficiency Evaluation Individual Primary Adrenal Insufficiency single gene te Only order single gene tests when not ordering the panel. □ 815 ABCD1 (Adrenoleukodystrophy) DNA Sec □ 812 Autoimmune Polyglandular Syndrome (Al	Additional Information (Genes, Antibodies, Comments) ABCD1, NR0B1, AIRE sts: uencing Test RE) Evaluation NA Sequencing Test Includes CYP21A2 sequencing and	Test Code Name Bone Diseases: Molecular Genetics (Continued) Individual Hypophosphatemic Rickets single gene test Only order single gene tests when not ordering the panel. □ 856 FGF23 (Hypophosphatemic Rickets) DNA □ 855 PHEX (Hypophosphatemic Rickets) DNA □ 811 LRP5 (OPPG) DNA Sequencing Test □ 821 LRP5 Idiopathic Osteoporosis (IOP) DNA Sequencing Test Congenital Hyperinsulinism: Molecular Genetics	ts: A Sequencing Test Sequencing Test
□ 8102 PKD2 DNA Sequencing and Deletion Evaluation □ 8104 PKD2 DNA Sequencing Test Test Code Name Adrenal Disorders: Molecular Genetics □ 816 Primary Adrenal Insufficiency Evaluation Individual Primary Adrenal Insufficiency single gene te Only order single gene tests when not ordering the panel. □ 815 ABCD1 (Adrenoleukodystrophy) DNA Sec □ 812 Autoimmune Polyglandular Syndrome (Al □ 814 NR0B1 (Adrenal Hypoplasia Congenita) □ ■ 879 Congenital Adrenal Hyperplasia (CAH) ■ Evaluation	Additional Information (Genes, Antibodies, Comments) ABCD1, NR0B1, AIRE sts: uencing Test RE) Evaluation NA Sequencing Test Includes CYP21A2 sequencing and deletion, CYP11B1 sequencing	Test Code Name Bone Diseases: Molecular Genetics (Continued) Individual Hypophosphatemic Rickets single gene test Only order single gene tests when not ordering the panel. □ 856 FGF23 (Hypophosphatemic Rickets) DNA □ 855 PHEX (Hypophosphatemic Rickets) DNA □ 811 LRP5 (OPPG) DNA Sequencing Test □ 821 LRP5 Idiopathic Osteoporosis (IOP) DNA Sequencing Test	ts: A Sequencing Test Sequencing Test Does not qualify for the Advance Pay
□ 8102 PKD2 DNA Sequencing and Deletion Evaluation □ 8104 PKD2 DNA Sequencing Test Test Code Name Adrenal Disorders: Molecular Genetics □ 816 Primary Adrenal Insufficiency Evaluation Individual Primary Adrenal Insufficiency single gene te Only order single gene tests when not ordering the panel. □ 815 ABCD1 (Adrenoleukodystrophy) DNA Sec □ 812 Autoimmune Polyglandular Syndrome (Al □ 814 NR0B1 (Adrenal Hypoplasia Congenita) □ 879 Congenital Adrenal Hyperplasia (CAH) Evaluation Individual CAH single gene tests: Only order single gene tests when not ordering the panel.	Additional Information (Genes, Antibodies, Comments) ABCD1, NR0B1, AIRE sts: uencing Test RE) Evaluation NA Sequencing Test Includes CYP21A2 sequencing and deletion, CYP11B1 sequencing Required for tests 879, 880, 875: Indication for Study (check one or more below):	Test Code Name Bone Diseases: Molecular Genetics (Continued) Individual Hypophosphatemic Rickets single gene test Only order single gene tests when not ordering the panel. □ 856 FGF23 (Hypophosphatemic Rickets) DNA □ 855 PHEX (Hypophosphatemic Rickets) DNA □ 811 LRP5 (OPPG) DNA Sequencing Test □ 821 LRP5 Idiopathic Osteoporosis (IOP) DNA Sequencing Test Congenital Hyperinsulinism: Molecular Genetics	ts: A Sequencing Test Sequencing Test
□ 8102 PKD2 DNA Sequencing and Deletion Evaluation □ 8104 PKD2 DNA Sequencing Test Test Code Name Adrenal Disorders: Molecular Genetics □ 816 Primary Adrenal Insufficiency Evaluation Individual Primary Adrenal Insufficiency single gene te Only order single gene tests when not ordering the panel. □ 815 ABCD1 (Adrenoleukodystrophy) DNA Sec □ 812 Autoimmune Polyglandular Syndrome (Al □ 814 NR0B1 (Adrenal Hypoplasia Congenita) D □ 879 Congenital Adrenal Hypoplasia (CAH) ■ Evaluation Individual CAH single gene tests: Only order single gene tests when not ordering the panel. □ 875 CYP11B1 (CAH) DNA Sequencing Test	Additional Information (Genes, Antibodies, Comments) ABCD1, NR0B1, AIRE sts: uencing Test RE) Evaluation NA Sequencing Test Includes CYP21A2 sequencing and deletion, CYP11B1 sequencing Required for tests 879, 880, 875: Indication for Study (check one or more below):	Test Code Name Bone Diseases: Molecular Genetics (Continued) Individual Hypophosphatemic Rickets single gene test Only order single gene tests when not ordering the panel. □ 856 FGF23 (Hypophosphatemic Rickets) DNA □ 855 PHEX (Hypophosphatemic Rickets) DNA □ 811 LRP5 (OPPG) DNA Sequencing Test □ 821 LRP5 Idiopathic Osteoporosis (IOP) DNA Sequencing Test Congenital Hyperinsulinism: Molecular Genetics	ts: A Sequencing Test Sequencing Test Does not qualify for the Advance Pay Option. GLUD1, GCK, KCNJ11, ABCC8 Indication for Study (check one or more
□ 8102 PKD2 DNA Sequencing and Deletion Evaluation □ 8104 PKD2 DNA Sequencing Test Test Code Name Adrenal Disorders: Molecular Genetics □ 816 Primary Adrenal Insufficiency Evaluation Individual Primary Adrenal Insufficiency single gene te Only order single gene tests when not ordering the panel. □ 815 ABCD1 (Adrenoleukodystrophy) DNA Sec □ 812 Autoimmune Polyglandular Syndrome (Al □ 814 NR0B1 (Adrenal Hypoplasia Congenita) □ 879 Congenital Adrenal Hyperplasia (CAH) Evaluation Individual CAH single gene tests: Only order single gene tests: Only order single gene tests: Only DNA Sequencing Test □ 875 CYP211B1 (CAH) DNA Sequencing Test □ 880 CYP21A2 (CAH) Evaluation	Additional Information (Genes, Antibodies, Comments) ABCD1, NR0B1, AIRE sts: uencing Test RE) Evaluation NA Sequencing Test Includes CYP21A2 sequencing and deletion, CYP11B1 sequencing Required for tests 879, 880, 875: Indication for Study (check one or more below):	Test Code Name Bone Diseases: Molecular Genetics (Continued) Individual Hypophosphatemic Rickets single gene test Only order single gene tests when not ordering the panel. □ 856 FGF23 (Hypophosphatemic Rickets) DNA □ 855 PHEX (Hypophosphatemic Rickets) DNA □ 811 LRP5 (OPPG) DNA Sequencing Test □ 821 LRP5 Idiopathic Osteoporosis (IOP) DNA Sequencing Test Congenital Hyperinsulinism: Molecular Genetics	Genes, Antibodies, Comments
□ 8102 PKD2 DNA Sequencing and Deletion Evaluation □ 8104 PKD2 DNA Sequencing Test Test Code Name Adrenal Disorders: Molecular Genetics □ 816 Primary Adrenal Insufficiency Evaluation Individual Primary Adrenal Insufficiency single gene te Only order single gene tests when not ordering the panel. □ 815 ABCD1 (Adrenoleukodystrophy) DNA Sec □ 812 Autoimmune Polyglandular Syndrome (Al □ 814 NR0B1 (Adrenal Hypoplasia Congenita) D □ 879 Congenital Adrenal Hypoplasia (CAH) ■ Evaluation Individual CAH single gene tests: Only order single gene tests when not ordering the panel. □ 875 CYP11B1 (CAH) DNA Sequencing Test	Additional Information (Genes, Antibodies, Comments) ABCD1, NR0B1, AIRE sts: uencing Test RE) Evaluation NA Sequencing Test Includes CYP21A2 sequencing and deletion, CYP11B1 sequencing Required for tests 879, 880, 875: Indication for Study (check one or more below): Family history of CAH Virilization (ambiguous genitalia) Salt Wasting	Test Code Name Bone Diseases: Molecular Genetics (Continued) Individual Hypophosphatemic Rickets single gene test Only order single gene tests when not ordering the panel. □ 856 FGF23 (Hypophosphatemic Rickets) DNA □ 855 PHEX (Hypophosphatemic Rickets) DNA □ 811 LRP5 (OPPG) DNA Sequencing Test □ 821 LRP5 Idiopathic Osteoporosis (IOP) DNA Sequencing Test Congenital Hyperinsulinism: Molecular Genetics	Genes, Antibodies, Comments
□ 8102 PKD2 DNA Sequencing and Deletion Evaluation □ 8104 PKD2 DNA Sequencing Test Test Code Name Adrenal Disorders: Molecular Genetics □ 816 Primary Adrenal Insufficiency Evaluation Individual Primary Adrenal Insufficiency single gene te Only order single gene tests when not ordering the panel. □ 815 ABCD1 (Adrenoleukodystrophy) DNA Sec □ 812 Autoimmune Polyglandular Syndrome (Al □ 814 NR0B1 (Adrenal Hypoplasia Congenita) □ 879 Congenital Adrenal Hyperplasia (CAH) Evaluation Individual CAH single gene tests: Only order single gene tests: Only order single gene tests: Only DNA Sequencing Test □ 875 CYP211B1 (CAH) DNA Sequencing Test □ 880 CYP21A2 (CAH) Evaluation	Additional Information (Genes, Antibodies, Comments) ABCD1, NR0B1, AIRE sts: uencing Test RE) Evaluation NA Sequencing Test Includes CYP21A2 sequencing and deletion, CYP11B1 sequencing Required for tests 879, 880, 875: Indication for Study (check one or more below):	Test Code Name Bone Diseases: Molecular Genetics (Continued) Individual Hypophosphatemic Rickets single gene test Only order single gene tests when not ordering the panel. □ 856 FGF23 (Hypophosphatemic Rickets) DNA □ 855 PHEX (Hypophosphatemic Rickets) DNA □ 811 LRP5 (OPPG) DNA Sequencing Test □ 821 LRP5 Idiopathic Osteoporosis (IOP) DNA Sequencing Test Congenital Hyperinsulinism: Molecular Genetics	Genes, Antibodies, Comments
□ 8102 PKD2 DNA Sequencing and Deletion Evaluation □ 8104 PKD2 DNA Sequencing Test Test Code Name Adrenal Disorders: Molecular Genetics □ 816 Primary Adrenal Insufficiency Evaluation Individual Primary Adrenal Insufficiency single gene te Only order single gene tests when not ordering the panel. □ 815 ABCD1 (Adrenoleukodystrophy) DNA Sec □ 812 Autoimmune Polyglandular Syndrome (Al □ 814 NR0B1 (Adrenal Hypoplasia Congenita) □ 879 Congenital Adrenal Hyperplasia (CAH) Evaluation Individual CAH single gene tests: Only order single gene tests: Only order single gene tests: Only DNA Sequencing Test □ 875 CYP211B1 (CAH) DNA Sequencing Test □ 880 CYP21A2 (CAH) Evaluation	Additional Information (Genes, Antibodies, Comments) ABCD1, NR0B1, AIRE sts: uencing Test RE) Evaluation NA Sequencing Test Includes CYP21A2 sequencing and deletion, CYP11B1 sequencing 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	Test Code Name Bone Diseases: Molecular Genetics (Continued) Individual Hypophosphatemic Rickets single gene test Only order single gene tests when not ordering the panel. □ 856 FGF23 (Hypophosphatemic Rickets) DNA □ 855 PHEX (Hypophosphatemic Rickets) DNA □ 811 LRP5 (OPPG) DNA Sequencing Test □ 821 LRP5 Idiopathic Osteoporosis (IOP) DNA Sequencing Test Congenital Hyperinsulinism: Molecular Genetics	Genes, Antibodies, Comments
□ 8102 PKD2 DNA Sequencing and Deletion Evaluation □ 8104 PKD2 DNA Sequencing Test Test Code Name Adrenal Disorders: Molecular Genetics □ 816 Primary Adrenal Insufficiency Evaluation Individual Primary Adrenal Insufficiency single gene te Only order single gene tests when not ordering the panel. □ 815 ABCD1 (Adrenoleukodystrophy) DNA Sec □ 812 Autoimmune Polyglandular Syndrome (Al □ 814 NR0B1 (Adrenal Hypoplasia Congenita) □ 879 Congenital Adrenal Hyperplasia (CAH) Evaluation Individual CAH single gene tests: Only order single gene tests: Only order single gene tests: Only DNA Sequencing Test □ 875 CYP211B1 (CAH) DNA Sequencing Test □ 880 CYP21A2 (CAH) Evaluation	Additional Information (Genes, Antibodies, Comments) ABCD1, NR0B1, AIRE sts: uencing Test RE) Evaluation NA Sequencing Test Includes CYP21A2 sequencing and deletion, CYP11B1 sequencing 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)	Test Code Name Bone Diseases: Molecular Genetics (Continued) Individual Hypophosphatemic Rickets single gene test Only order single gene tests when not ordering the panel. □ 856 FGF23 (Hypophosphatemic Rickets) DNA □ 855 PHEX (Hypophosphatemic Rickets) DNA □ 811 LRP5 (OPPG) DNA Sequencing Test □ 821 LRP5 Idiopathic Osteoporosis (IOP) DNA Sequencing Test Congenital Hyperinsulinism: Molecular Genetics □ 819 Congenital Hyperinsulinism Evaluation	Genes, Antibodies, Comments
□ 8102 PKD2 DNA Sequencing and Deletion Evaluation □ 8104 PKD2 DNA Sequencing Test Test Code Name Adrenal Disorders: Molecular Genetics □ 816 Primary Adrenal Insufficiency Evaluation Individual Primary Adrenal Insufficiency single gene te Only order single gene tests when not ordering the panel. □ 815 ABCD1 (Adrenoleukodystrophy) DNA Sec □ 812 Autoimmune Polyglandular Syndrome (Al □ 814 NR0B1 (Adrenal Hypoplasia Congenita) □ 879 Congenital Adrenal Hyperplasia (CAH) Evaluation Individual CAH single gene tests: Only order single gene tests: Only order single gene tests: Only DNA Sequencing Test □ 875 CYP211B1 (CAH) DNA Sequencing Test □ 880 CYP21A2 (CAH) Evaluation	Additional Information (Genes, Antibodies, Comments) ABCD1, NR0B1, AIRE sts: uencing Test RE) Evaluation NA Sequencing Test Includes CYP21A2 sequencing and deletion, CYP11B1 sequencing 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	Test Code Name Bone Diseases: Molecular Genetics (Continued) Individual Hypophosphatemic Rickets single gene test Only order single gene tests when not ordering the panel. □ 856 FGF23 (Hypophosphatemic Rickets) DNA □ 855 PHEX (Hypophosphatemic Rickets) DNA □ 811 LRP5 (OPPG) DNA Sequencing Test □ 821 LRP5 Idiopathic Osteoporosis (IOP) DNA Sequencing Test Congenital Hyperinsulinism: Molecular Genetics □ 819 Congenital Hyperinsulinism Evaluation	Genes, Antibodies, Comments
□ 8102 PKD2 DNA Sequencing and Deletion Evaluation □ 8104 PKD2 DNA Sequencing Test Test Code Name Adrenal Disorders: Molecular Genetics □ 816 Primary Adrenal Insufficiency Evaluation Individual Primary Adrenal Insufficiency single gene te Only order single gene tests when not ordering the panel. □ 815 ABCD1 (Adrenoleukodystrophy) DNA Sec □ 812 Autoimmune Polyglandular Syndrome (Al □ 814 NR0B1 (Adrenal Hypoplasia Congenita) □ 879 Congenital Adrenal Hypoplasia (CAH) Evaluation 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 □ 877 CYP17A1 DNA Sequencing Test □ 881 Endocrine Hyportension (HSD11B2)	Additional Information (Genes, Antibodies, Comments) ABCD1, NR0B1, AIRE sts: uencing Test RE) Evaluation NA Sequencing Test Includes CYP21A2 sequencing and deletion, CYP11B1 sequencing 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	Test Code Name Bone Diseases: Molecular Genetics (Continued) Individual Hypophosphatemic Rickets single gene test Only order single gene tests when not ordering the panel. □ 856 FGF23 (Hypophosphatemic Rickets) DNA □ 811 LRP5 (OPPG) DNA Sequencing Test □ 821 LRP5 Idiopathic Osteoporosis (IOP) DNA Sequencing Test ■ 819 Congenital Hyperinsulinism: Molecular Genetics □ 819 Congenital Hyperinsulinism Evaluation	Comments Comments
□ 8102 PKD2 DNA Sequencing and Deletion Evaluation □ 8104 PKD2 DNA Sequencing Test Test Code Name Adrenal Disorders: Molecular Genetics □ 816 Primary Adrenal Insufficiency Evaluation Individual Primary Adrenal Insufficiency single gene te Only order single gene tests when not ordering the panel. □ 815 ABCD1 (Adrenoleukodystrophy) DNA Sec □ 812 Autoimmune Polyglandular Syndrome (Al □ 814 NR0B1 (Adrenal Hypoplasia Congenita) D ■ 879 Congenital Adrenal Hyperplasia (CAH) ■ Evaluation 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 □ 877 CYP17A1 DNA Sequencing Test □ 881 Endocrine Hypertension (HSD11B2) Evaluation	Additional Information (Genes, Antibodies, Comments) ABCD1, NR0B1, AIRE sts: uencing Test RE) Evaluation NA Sequencing Test Includes CYP21A2 sequencing and deletion, CYP11B1 sequencing 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	Test Code Name Bone Diseases: Molecular Genetics (Continued) Individual Hypophosphatemic Rickets single gene test Only order single gene tests when not ordering the panel. □ 856 FGF23 (Hypophosphatemic Rickets) DNA □ 815 PHEX (Hypophosphatemic Rickets) DNA □ 811 LRP5 (OPPG) DNA Sequencing Test □ 821 LRP5 Idiopathic Osteoporosis (IOP) DNA Sequencing Test Congenital Hyperinsulinism: Molecular Genetics □ 819 Congenital Hyperinsulinism Evaluation Individual Congenital Hyperinsulinism Evaluation	Genes, Antibodies, Comments
□ 8102 PKD2 DNA Sequencing and Deletion Evaluation □ 8104 PKD2 DNA Sequencing Test Test Code Name Adrenal Disorders: Molecular Genetics □ 816 Primary Adrenal Insufficiency Evaluation Individual Primary Adrenal Insufficiency single gene te Only order single gene tests when not ordering the panel. □ 815 ABCD1 (Adrenoleukodystrophy) DNA Sec □ 812 Autoimmune Polyglandular Syndrome (Al □ 814 NR0B1 (Adrenal Hypoplasia Congenita) □ 879 Congenital Adrenal Hyperplasia (CAH) Evaluation 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 □ 881 Endocrine Hypertension (HSD11B2) Evaluation □ 878 HSD3B2 DNA Sequencing Test	Additional Information (Genes, Antibodies, Comments) ABCD1, NR0B1, AIRE sts: uencing Test RE) Evaluation NA Sequencing Test Includes CYP21A2 sequencing and deletion, CYP11B1 sequencing 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	Test Code Name Bone Diseases: Molecular Genetics (Continued) Individual Hypophosphatemic Rickets single gene test Only order single gene tests when not ordering the panel. □ 856 FGF23 (Hypophosphatemic Rickets) DNA □ 811 LRP5 (OPPG) DNA Sequencing Test □ 821 LRP5 Idiopathic Osteoporosis (IOP) DNA Sequencing Test Congenital Hyperinsulinism: Molecular Genetics □ 819 Congenital Hyperinsulinism Evaluation Individual Congenital Hyperinsulinism Evaluation Individual Congenital Hyperinsulinism Sequencing Test Only order single gene tests when not ordering the panel. Single gene tests for the CH Panel, do not qualify for 1 □ 822 GLUD1 (CHI) DNA Sequencing Test □ 826 KCNJ11 (CHI) DNA Sequencing Test	Comments Comments
□ 8102 PKD2 DNA Sequencing and Deletion Evaluation □ 8104 PKD2 DNA Sequencing Test Test Code Name Adrenal Disorders: Molecular Genetics □ 816 Primary Adrenal Insufficiency Evaluation Individual Primary Adrenal Insufficiency single gene te Only order single gene tests when not ordering the panel. □ 815 ABCD1 (Adrenoleukodystrophy) DNA Sec □ 812 Autoimmune Polyglandular Syndrome (Al □ 814 NR0B1 (Adrenal Hypoplasia Congenita) □ 879 Congenital Adrenal Hyperplasia (CAH) Evaluation 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 □ 877 CYP17A1 DNA Sequencing Test □ 878 HSD3B2 DNA Sequencing Test □ 878 HSD3B2 DNA Sequencing Test □ 874 Lipoid CAH (STAR) DNA Sequencing Test	Additional Information (Genes, Antibodies, Comments) ABCD1, NR0B1, AIRE sts: uencing Test RE) Evaluation NA Sequencing Test Includes CYP21A2 sequencing and deletion, CYP11B1 sequencing 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	Test Code Name Bone Diseases: Molecular Genetics (Continued) Individual Hypophosphatemic Rickets single gene test Only order single gene tests when not ordering the panel. □ 856 FGF23 (Hypophosphatemic Rickets) DNA □ 855 PHEX (Hypophosphatemic Rickets) DNA □ 811 LRP5 (OPPG) DNA Sequencing Test □ 821 LRP5 Idiopathic Osteoporosis (IOP) DNA Sequencing Test Congenital Hyperinsulinism: Molecular Genetics □ 819 Congenital Hyperinsulinism Evaluation Individual Congenital Hyperinsulinism Evaluation Individual Congenital Hyperinsulinism Single gene test Only order single gene tests when not ordering the panel. Single gene tests for te CH Panel, do not qualify for the Sequencing Test □ 822 GLUD1 (CHI) DNA Sequencing Test □ 826 KCNJ11 (CHI) DNA Sequencing Test □ 42 CH Parental Testing – To augment child/	Comments Comments
□ 8102 PKD2 DNA Sequencing and Deletion Evaluation □ 8104 PKD2 DNA Sequencing Test Test Code Name Adrenal Disorders: Molecular Genetics □ 816 Primary Adrenal Insufficiency Evaluation Individual Primary Adrenal Insufficiency single gene tes Only order single gene tests when not ordering the panel. □ 815 ABCD1 (Adrenoleukodystrophy) DNA Sec □ 812 Autoimmune Polyglandular Syndrome (Al □ 814 NR0B1 (Adrenal Hypoplasia Congenita) D 879 Congenital Adrenal Hyperplasia (CAH) Evaluation 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 □ 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	Additional Information (Genes, Antibodies, Comments) ABCD1, NR0B1, AIRE sts: uencing Test RE) Evaluation NA Sequencing Test Includes CYP21A2 sequencing and deletion, CYP11B1 sequencing 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	Test Code Name Bone Diseases: Molecular Genetics (Continued) Individual Hypophosphatemic Rickets single gene test Only order single gene tests when not ordering the panel. □ 856 FGF23 (Hypophosphatemic Rickets) DNA □ 811 LRP5 (OPPG) DNA Sequencing Test □ 821 LRP5 Idiopathic Osteoporosis (IOP) DNA Sequencing Test Congenital Hyperinsulinism: Molecular Genetics □ 819 Congenital Hyperinsulinism Evaluation Individual Congenital Hyperinsulinism Evaluation Individual Congenital Hyperinsulinism Sequencing Test Only order single gene tests when not ordering the panel. Single gene tests for the CH Panel, do not qualify for 1 □ 822 GLUD1 (CHI) DNA Sequencing Test □ 826 KCNJ11 (CHI) DNA Sequencing Test	Genes, Antibodies, Comments
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□ 8102 PKD2 DNA Sequencing and Deletion Evaluation □ 8104 PKD2 DNA Sequencing Test Test Code Name Adrenal Disorders: Molecular Genetics □ 816 Primary Adrenal Insufficiency Evaluation Individual Primary Adrenal Insufficiency single gene te Only order single gene tests when not ordering the panel. □ 815 ABCD1 (Adrenoleukodystrophy) DNA Sec □ 812 Autoimmune Polyglandular Syndrome (Al □ 814 NR0B1 (Adrenal Hypoplasia Congenita) □ 879 Congenital Adrenal Hyperplasia (CAH) Evaluation 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 □ 881 Endocrine Hypertension (HSD11B2) Evaluation □ 878 HSD3B2 DNA Sequencing Test □ 874 Lipoid CAH (STAR) DNA Sequencing Test □ 875 Osteogenesis Imperfecta Evaluation Individual Osteogenesis Imperfecta single gene tests:	Additional Information (Genes, Antibodies, Comments) ABCD1, NR0B1, AIRE sts: uencing Test RE) Evaluation NA Sequencing Test Includes CYP21A2 sequencing and deletion, CYP11B1 sequencing 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	Test Code Name Bone Diseases: Molecular Genetics (Continued) Individual Hypophosphatemic Rickets single gene test Only order single gene tests when not ordering the panel. □ 856 FGF23 (Hypophosphatemic Rickets) DNA □ 855 PHEX (Hypophosphatemic Rickets) DNA □ 811 LRP5 (OPPG) DNA Sequencing Test □ 821 LRP5 Idiopathic Osteoporosis (IOP) DNA Sequencing Test Congenital Hyperinsulinism: Molecular Genetics □ 819 Congenital Hyperinsulinism Evaluation Individual Congenital Hyperinsulinism Evaluation Individual Congenital Hyperinsulinism Single gene test Only order single gene tests when not ordering the panel. Single gene tests for te CH Panel, do not qualify for the Sequencing Test □ 822 GLUD1 (CHI) DNA Sequencing Test □ 826 KCNJ11 (CHI) DNA Sequencing Test □ 42 CH Parental Testing – To augment child/	Comments Comments
□ 8102 PKD2 DNA Sequencing and Deletion Evaluation □ 8104 PKD2 DNA Sequencing Test Test Code Name Adrenal Disorders: Molecular Genetics □ 816 Primary Adrenal Insufficiency Evaluation Individual Primary Adrenal Insufficiency single gene te Only order single gene tests when not ordering the panel. □ 815 ABCD1 (Adrenoleukodystrophy) DNA Sec □ 812 Autoimmune Polyglandular Syndrome (Al □ 814 NR0B1 (Adrenal Hypoplasia Congenita) DI 879 Congenital Adrenal Hyperplasia (CAH) Evaluation 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 □ 877 CYP17A1 DNA Sequencing Test □ 881 Endocrine Hypertension (HSD11B2) Evaluation □ 878 HSD3B2 DNA Sequencing Test □ 874 Lipoid CAH (STAR) DNA Sequencing Test □ 875 Osteogenesis Imperfecta Evaluation Individual Osteogenesis Imperfecta single gene tests: Only order single gene tests when not ordering the panel.	Additional Information (Genes, Antibodies, Comments) ABCD1, NR0B1, AIRE sts: uencing Test RE) Evaluation NA Sequencing Test Includes CYP21A2 sequencing and deletion, CYP11B1 sequencing 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 COL1A1, COL1A2	Test Code Name Bone Diseases: Molecular Genetics (Continued) Individual Hypophosphatemic Rickets single gene test Only order single gene tests when not ordering the panel. □ 856 FGF23 (Hypophosphatemic Rickets) DNA □ 855 PHEX (Hypophosphatemic Rickets) DNA □ 811 LRP5 (OPPG) DNA Sequencing Test □ 821 LRP5 Idiopathic Osteoporosis (IOP) DNA Sequencing Test Congenital Hyperinsulinism: Molecular Genetics □ 819 Congenital Hyperinsulinism Evaluation Individual Congenital Hyperinsulinism Evaluation Individual Congenital Hyperinsulinism Single gene test Only order single gene tests when not ordering the panel. Single gene tests for te CH Panel, do not qualify for the Sequencing Test □ 822 GLUD1 (CHI) DNA Sequencing Test □ 826 KCNJ11 (CHI) DNA Sequencing Test □ 42 CH Parental Testing – To augment child/	Genes, Antibodies, Comments
□ 8102 PKD2 DNA Sequencing and Deletion Evaluation □ 8104 PKD2 DNA Sequencing Test Test Code Name Adrenal Disorders: Molecular Genetics □ 816 Primary Adrenal Insufficiency Evaluation Individual Primary Adrenal Insufficiency single gene te Only order single gene tests when not ordering the panel. □ 815 ABCD1 (Adrenoleukodystrophy) DNA Sec □ 812 Autoimmune Polyglandular Syndrome (Al □ 814 NR0B1 (Adrenal Hypoplasia Congenita) D 879 Congenital Adrenal Hyperplasia (CAH) Evaluation 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 ■ 877 CYP17A1 DNA Sequencing Test □ 881 Endocrine Hypertension (HSD11B2) Evaluation □ 878 HSD3B2 DNA Sequencing Test □ 874 Lipoid CAH (STAR) DNA Sequencing Test ■ 874 Lipoid CAH (STAR) DNA Sequencing Test ■ 876 Osteogenesis Imperfecta Evaluation Individual Osteogenesis Imperfecta single gene tests: Only order single gene tests when not ordering the panel.	Additional Information (Genes, Antibodies, Comments) ABCD1, NR0B1, AIRE sts: uencing Test RE) Evaluation NA Sequencing Test Includes CYP21A2 sequencing and deletion, CYP11B1 sequencing 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	Test Code Name Bone Diseases: Molecular Genetics (Continued) Individual Hypophosphatemic Rickets single gene test Only order single gene tests when not ordering the panel. □ 856 FGF23 (Hypophosphatemic Rickets) DNA □ 855 PHEX (Hypophosphatemic Rickets) DNA □ 811 LRP5 (OPPG) DNA Sequencing Test □ 821 LRP5 Idiopathic Osteoporosis (IOP) DNA Sequencing Test Congenital Hyperinsulinism: Molecular Genetics □ 819 Congenital Hyperinsulinism Evaluation Individual Congenital Hyperinsulinism Evaluation Individual Congenital Hyperinsulinism Single gene test Only order single gene tests when not ordering the panel. Single gene tests for te CH Panel, do not qualify for the Sequencing Test □ 822 GLUD1 (CHI) DNA Sequencing Test □ 826 KCNJ11 (CHI) DNA Sequencing Test □ 42 CH Parental Testing – To augment child/	Comments Comments

Reflexive testing is performed at an additional charge.

QD91640-ADX 8/25





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)	Test Code	Test Name	Additional Information (Genes, Antibodies, Comments)
		(Genes, Antibodies, Comments)		n Syndrome: Molecular Genetics	(Ceries, Antibodies, Comments)
Diabetes: Molecular Genetics			Noonan Syndrome (PTPN11) DNA	T	
	Monogenic Diabetes (MODY) Five-Gene Evaluation	HNF1A (TCF1), GCK, HNF4A, HNF1B (TCF2), IPF1		Sequencing Test	
	Monogenic Diabetes (MODY) Four-Gene Evaluation	HNF1A (TCF1), GCK, HNF4A, HNF1B (TCF2)		KRAS/RAF1/SOS1 DNA Sequencing Evaluation	SOS1, RAF1, KRAS
□ 8801	Monogenic Diabetes (MODY) Three-Gene Evaluation	HNF1A (TCF1), GCK, HNF1B (TCF2)		al KRAS/RAF1/SOS1 single gene tests: der single gene tests when not ordering the panel.	
□ 8802	Monogenic Diabetes (MODY) Two-Gene Evaluation	HNF1A (TCF1), GCK	□ 6	62 SOS1 DNA Sequencing Test	G63 RAF1 DNA Sequencing Test
□ 80	3 GCK (MODY2) DNA Sequencing and			y: Molecular Genetics	
	Deletion Test		□ 884	Early Onset Obesity Evaluation	LEPR, MC4R
□ 80	12 HNF4A (MODY1) DNA Sequencing and Deletion Test			al Early Onset Obesity single gene tests: der single gene tests when not ordering the panel.	
□ 83	4 IPF1 (MODY4) DNA Sequencing Test		□ 6	40 Early Onset Obesity (MC4R) DNA Sequer	ncing Test
	4 TCF1 (MODY3) DNA Sequencing and	HNF1A (TCF1)	□ 8	83 Early Onset Obesity (LEPR) DNA Sequen	cing Test
	Deletion Test	, ,		Bardet-Biedl Syndrome Evaluation	BBS1, BBS2, BBS10
□ 80	TCF2 (MODY5) DNA Sequencing and Deletion Test	HNF1B (TCF2)		al Bardet-Biedl Syndrome single gene tests: der single gene tests when not ordering the panel.	
□ 837	CEL (MODY8) Mutation Analysis				372 BBS2 (BBS) DNA Sequencing Test
□ 882	Neonatal Diabetes Mellitus Evaluation	IPF1, GCK, KCNJ11, INS, ABCC8		86 BBS10 (BBS) DNA Sequencing Test	, , , ,
Individual Neonatal Diabetes Mellitus single gene tests:		Reprod	luctive Disorders: Molecular Genetics		
	er single gene tests when not ordering the panel.		□ 679	Complete Kallmann/IHH Evaluation	
		842 GCK (NDM) DNA Sequencing Test	Individu	al Kallmann/IHH single gene tests:	
		341 IPF1 (NDM) DNA Sequencing Test	Only order single gene tests when not ordering the panel.		
	3 KCNJ11 (NDM) DNA Sequencing Test		□ 4		95 FGF8 DNA Sequencing Test
	genic Diabetes: Molecular Genetics		□ 1		43 GnRH1 DNA Sequencing Test
□ 854	Nephrogenic Diabetes Insipidus	AVPR2, AQP2			73 KAL1 DNA Sequencing Test
1 11 11	Evaluation	·			75 PROK2 DNA Sequencing Test
	al Nephrogenic Diabetes Mellitus single gene t	ests:			58 TACR3 DNA Sequencing Test
	er single gene tests when not ordering the panel. 2 AQP2 (Nephrogenic Diabetes Insipidus) D	INA Coguencing Toot		Anosmic Kallmann/IHH Evaluation	Please see website for the complete list of
	il Nephrogenic Diabetes Insipidus (AVPR2)		☐ 667		genes.
	Cancer Syndromes: Molecular Genetics	DNA Sequencing lest	□ 817	, , , , , , , , , , , , , , , , , , , ,	
□ 818	MEN1 DNA Sequencing Test			Sequencing Test	
	Pheochromocytoma Evaluation	DET VIII OBUB		Stature: Molecular Genetics	
	al Pheochromocytoma single gene tests:	RET, VHL, SDHB	□ 865	Combined Pituitary Hormone Deficiency Evaluation	PROP1, POU1F1
	er single gene tests when not ordering the panel.		Individu	al Pituitary Hormone Deficiency single gene te	sts:
	3 MEN2 (RET) DNA Sequencing Test	1888 SDHB DNA Sequencing Test		der single gene tests when not ordering the panel.	
	8 von Hippel-Lindau Syndrome (VHL) DNA		□ 8	64 POU1F1 (CPHD) DNA Sequencing Test	
	Hypocalciuric Hypercalcemia: Molecular (□ 8	63 PROP1 (CPHD) DNA Sequencing Test	
	Familial Hypocalciuric Hypercalcemia (CASR) DNA Sequencing Test		□ 848	Growth Hormone Deficiency Evaluation	GH1 and GHRHR Seq.; SHOX Seq. and Del.
Family 7			Individu	al Growth Hormone Deficiency single gene tes	ts:
	Familial DNA Sequence Evaluation	This test detects previously identified		der single gene tests when not ordering the panel.	
	Proband Accession #	sequence variants in at-risk family members.		66 GH1 (GHD) DNA Sequencing Test 47 SHOX (GHD) DNA Sequencing and Delet	
	Relationship	Internacia.		GHR DNA Sequencing Test	
					*