

Insurance and Advance Pay Test Requisition

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

Client Services is available Monday through Friday from 8:30 AM to 9:00 PM EST at 1.800.394.4493, option 2

Patient Information	on					
Patient Name						
	ole)					
	Sex designated at birth: Male Female					
	Other Phone #2					
	ther than English					
Ordering Account						
Ordering physician na	ame:					
Phone	FaxEmail					
NPI#	Athena Account # (if assigned)					
Reporting preference	: 🗆 Fax 🗆 Email					
Send additional repo	rt copies to:					
Clinician/Facility						
City, State, Zip						
Phone	Fax					
Email						
Test Information						
the test list. Call Client Services at	est code, name and acceptable specimen options. Specimen requirements are referenced at the top of t 1.800.394.4493, option 2 for additional details. ed for billing insurance):					
Test Code	Test Name					

Clinical Informatio	n	,	,			, , ,
Clinical diagnosis:						
Age at Initial Presentat Ancestral Background African Hispanic Caribbean	(check all that apply): Asian: East Native American	☐ Asian: Southeast☐ Ashkenazi Jewish☐ Middle Eastern		☐ Central/South Ame☐ Asian: Indian☐ Pacific Islander	rican	
Other:						
Indications for genetic						
☐ Diagnostic (sympton☐ Carrier		ve (asymptomatic) testing/single site	☐ Pren	atal (Contact Athena p	rior to sendi	ng)
Relationship to Proban	nd:					
If performed at Athena	, provide relative's acce	ession #				
If performed at anothe	r lab, a copy of the relat	tive's report is required.				
Please attach detailed	medical records and fa	amily history information	on.			
Specimen Informat	tion					
Specimen Type: Date	sample obtained:	/////				
☐ Whole Blood	Serum	☐ Cerebrospinal Fluid	(CSF)	☐ CVS: Cultured		
☐ Amniotic Fluid: Cultu	ured	☐ Saliva (Not availabl	e for all te	ests)		
☐ DNA* source:		Concentration	າ			ug/ml
*DNA must be extracted	d at a CLIA-certified or a	laboratory meeting equiv	alent requ	uirements (as determine	ed by CAP an	d/or CMS).
Other** source (prov	vide specimen type):					
**Contact Athena prio	<u>r to sending</u> specimen t	types not listed above.				
		s sample stored?		D □ Refrigerated □] Frozen	
Date of most recent tra	ansfusion/transplant:_	//				
Statement of Medical In accordance with Mass verification of patient in Massachusetts require required to complete the Consent (PAIC) at any Q Prior to ordering genetic their authorized represe forms as part of the pat (including Medicare and the test ordered. I under of the patient consister necessary for the diagnused in the medical mathe test ordered. I confrequested herein consister Please sign, date and in information is not provipayor request.	Necessity and Informed seachusetts General Law of formed consent is required a signed acknowledgment of genetic testing ordered uest lab. In the second of t	d Consent: A Chapter 111, Section 7 Aired for genetic testing. Bent from the ordering me Bed if you have not previous A listed above, I have obtate A applicable state law and A available to Athena Dia Bel necessity requirements Bel of the ase, illness, impairment Bet those tests which are Bed in the Ordering Physic Bed in the Ordering Physic Bel (MD, DO, NP) to docume Bed to provide medical recommendations.	Additional edical pracusty signed in a sign door regular gnostics of the consistic endical for the test or the tes	Illy, testing laboratories ctitioner. The signed acid a blanket Physician A ned, written consent for ations, and I will maintage appropriate the testing of the diagram of	located in knowledgment testation of the form from the ain all writtenst. Many pay ulatory requipatory	ent is f Informed patient (or n consent yers irements for reatment medically sults will be uirements for r the test(s) ote that if the
Medical Practitioner S	ignature:			Date	/	/
Medical Practitioner C	redentials:					

Payment Option	Selection and Details:		
Please check the	preferred payment option and comple	ete the corresponding section.	
*To be completed	d by patient/guardian, and signature is	required.	
For Billing inquir	ies provide contact information.		
Name:			
Mobile Phone (inc	ludes texts)	Email	
☐ Option 1: Ins	surance Pay (Please provide a photocop	y of the front and back of ALL insure	ance cards, including secondary)
Name of Ins	ured:		
Relationshi	p to Patient: 🗌 Self 🗌 Parent 🗌 Spouse 🛭	Other	
Insurance C	ompany:	Member ID#	Group ID#
Does the pa	tient have secondary insurance? \square Yes [□No	
	ompany:		
	or authorization # (please attach referra		
NOTE: AAP is n Please see the billing/athena To expedite and the ann	thena Alliance Program (AAP) Pate of available for Advance Pay/self-paying Athena Website for further information of alliance-program) consideration for AAP eligibility, please pual income of your household (Annual hold their dependents with income).	clients. bout the AAP offering (https://www.provide the number of household n	nembers (including yourself)
about test co	qualify for AAP and you do not want to have ost and Advance Pay. Please see the Advanc a Diagnostics does not hold Immunology te	ce Pay Section.	place order on hold to have discussion
I hereby ack of any and a nor employed me unless I an AAP app required pri Beneficiary received, in insurance of insurance of test not cover I am a New and a series of any any and any and a series of any and any and a series of any and a series of any and a series of any	nowledgement chowledge that the above information is all financial records necessary to verify the doby the physician who ordered the test provide alternative information. For more ication separately, go to www.athenadia or to genetic testing. I understand that if Notice (ABN) is required prior to the test cluding, without limitation, medical information and its authorized representatives arrier to directly pay Athena for the service of	ne above information. I hereby acking. The contact information above e detailed information on the AAP gnostics.com. For Medicaid and Miny physician ordered genetic test proceeding. I authorize Athena Distriction, which includes laboratory as necessary for reimbursement. I ces rendered. I understand that I wor and submit an AAP application.	nowledge that I am neither related to a will be used to communicate with program or to complete and send ledicare beneficiaries, payment is ting and I have Medicare, an Advanced iagnostics to release information test results, to my health plan/ I further authorize my health plan/ may be responsible for portions of this
*Signature of Pat	ent/Responsible Party:		Date
□ Option 2: Atl	nena Advance Pay Program Only a	vailable for genetic testing	
Please see the insurance-billi	Athena website for further information a ng/athena-advance-pay-program)	bout the Advance Pay offering (http	J
self-pay pat	n for this testing to be submitted for reimb ient for this testing. If I have insurance co unce for this testing or provide me with inf	verage, I acknowledge and agree Ath	nena Diagnostics will not submit a claim
to pay the ar	to Advance Pay, I understand that I will be nount due within 30 days of Athena receiviert/Responsible Party:	ng my sample, I will be charged the f	
	a Diagnostics at 1.800.394.4493, option y amount within 30 Days of specimen red		ing this option, I agree to pay the full
if the test re the 20% dis reflexive, pl	s that have multiple phases (reflexive co esult meets the criteria to reflex/move to count on the reflexive component of the ease call 1.800.394.4493, option 4.	the next phase. By electing the Adordered test. For questions on wh	dvance Pay Option you will still receive nether or not the ordered test code is
and/or mob (normal me the patient	il address or mobile telephone number i ile telephone number, the patient conser ssage and data rates may apply). The me may call 1.800.394.4493, option 4.	nts to receive calls, emails and/or t	text messages to collect payment
Mobile Phone:	Email: _		Reviewed December 2023

Reviewed December 2023

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 (Genes, Antibodies, Comments) Column below for specimen requirement exceptions

NEUROLOGY GENETIC & IMMUNOLOGY TESTING					
Test Code	Test Name	Additional Information (Genes, Antibodies, Comments)	Test Code	Test Name	Additional Information (Genes, Antibodies, Comments)
Cerebro	vascular Disease (Stroke): Molecular Gener	tics	Epilepsy	y: Molecular Genetics (Continued)	
	Notch3 (CADASIL) Sequencing Test HTRA1 (CARASIL) Sequencing Test			Complete Tuberous Sclerosis Sequencing and CNV Evaluation	Full Sequencing of TSC1 & TSC2
☐ 1120 ☐ 1122	COL4A1 Sequencing Test (CSVD) Complete CCM Sequencing and CNV Evaluation		Only orde	al Tuberous Sclerosis single gene tests: er single gene tests when not ordering the panel. 236 TSC1 CNV Test	254 TSC2 CNV Test 24 TSC2 DNA Deletion Test (for NYS Only)
Only orde	al CCM single gene tests: er single gene tests when not ordering the panel.	1400 COMO Core and CNN/Firely Star	□ 12		255 TSC2 Sequencing Test
<u> </u>	52 KRIT1 (CCM1) Seq. and CNV Evaluation 79 PDCD10 (CCM3) Seq. and CNV Evaluation a: Molecular Genetics		523	Proband Accession #	
	ADmark® Alzheimer's Evaluation	Does not qualify for the Advance Pay Option.		Relationship	
		Molecular Genetics Component(s): ApoE Immunology Component(s): AB42,		SCN1A Seq. and CNV Evaluation al SCN1A tests:	
		Phospho-Tau & Total-Tau. Specimen Requirements:		91 SCN1A CNV Test 53 CSTB (EPM1) Sequencing and Repeat	37 SCN1A Deletion Test
		Cerebrospinal Fluid (CSF) 2 mL in Polypropylene Tube and must arrive on		Expansion Evaluation	Cannot be done on saliva. Repeat Expansion Testing
		cold pack or frozen. Whole blood 8 mL (6 mL minimum) in		0 EPM1 DNA Test	Cannot be done on saliva.
		Lavender top (EDTA) tube. Cannot be performed with Saliva		ARX Seq. and CNV Evaluation (Epilepsy) CDKL5 Seq. and CNV Evaluation (Epilepsy)	
		sample type.	4411	SLC2A1 DNA Sequencing Test	
□ 10	9 ADmark® ApoE Genotype Analysis & Interpretation (Symptomatic for Dementia)		□ 1003	GFAP (Alexander Disease) Seq. Test	
□ 179	ADmark® Early-Onset Alzheimer's			POLG DNA Seq. Test (Alpers Syndrome)	
•	Evaluation	PSEN1, APP Seq./Dup., PSEN2		y: Immunology	
Only orde	II ADmark® Early-Onset Alzheimer's single gener single gene tests when not ordering the panel. ADmark® APP DNA Sequencing Test and If ADmark® PSEN1 DNA Sequencing Test ADmark® PSEN2 DNA Sequencing Test ADmark® PSEN2 DNA Sequencing Test		□ 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)		
	Frontotemporal Dementia (FTD) Evaluation	MAPT, GRN, C9orf72	□ 51	O1 GAD65 Neurological Syndrome Autoantibo O4 LGI1 Autoantibody Test (Epilepsy) (Single)	
	I FTD single gene tests:			NMDA Receptor Autoantibody Test (EpilepVGKC Autoantibody Test (Epilepsy) (Single	
	er single gene tests when not ordering the panel. 9 C9orf72 (FTD) DNA Test 20	04 GRN DNA Sequencing Test	Family 1		
	5 MAPT DNA Sequencing Test	or and 210 reequentsing reed	□ 185	Familial DNA Sequence Evaluation	This test detects previously identified sequence variants in at-risk family
	a: Immunology ADmark® Phospho-Tau/Total-Tau/Aß42 CSF	Analysis & Interpretation (Symptomatic) Specimen Type = Cerebrospinal Fluid (CSF)			members. For Familial TSC variants, please order Code 523.
		Volume = 2 mL Tube Type = Polypropylene Tube			Proband Accession #
☐ 1711	Autoimmune Rapidly Progressive	Must arrive on cold pack or frozen.			Relationship
· · · · ·	Dementia Evaluation with Recombx®			ology: Anti-Drug Antibody	
Individua	Il Autoimmune Dementia single antibody tests:			AAV9 Antibody Test	Does not qualify for the Advance Pay Option.
Only orde	er single autoantibody tests when not ordering the	panel.		ystrophy: Molecular Genetics	FIF2D4 FIF2D2 FIF2D4
□ 17	14 Recombx [®] Hu Autoantibody Test*	Recombx® Amphiphysin Autoantibody Test*		Leukoencephalopathy with Vanishing White Matter Evaluation	EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5
		S NMDA Receptor Autoantibody Test* 3 LGI1 Autoantibody Test*		al Leukoencephalopathy with Vanishing White I er single gene tests when not ordering the panel.	watter single gene tests.
	09 CASPR2 Autoantibody Test*	S LGIT Autoantibody Test			6102 EIF2B2 DNA Seguencing Test
	Cerebrospinal Fluid (CSF) is an acceptable sa	ample type for these tests.	1	, ,] 6104 EIF2B4 DNA Sequencing Test
Epileps	y: Molecular Genetics		1	105 EIF2B5 DNA Sequencing Test	3
☐ 6000	Epilepsy Advanced Sequencing and CNV Evaluation			PLP1 Sequencing and CNV Evaluation	
6r	18 Developmental Brain Malformations			ABCD1 DNA Sequencing Test	
	23 Epilepsy with Migraine	Test 6000 contains all genes included in the sub-panels.		ARSA DNA Sequencing Test GJC2 DNA Sequencing Test	
□ 60	10 Epileptic Encephalopathy	NOTE O L. L. L. MODES		Notch3(CADASIL) Sequencing Test	
□ 60	08 Generalized, Absence, Focal, Febrile and Myoclonic Epilepsies	NOTE: Only select sub-panels if 6000 is not ordered.	Migraine	e: Molecular Genetics	
□ 60	38 Infantile Spasms	Please see website for the list of genes	<u>1148</u>	Hemiplegic Migraine Sequencing Evaluation	CACNA1A, ATP1A2, SCN1A
□ 60	19 Intellectual Disability	in each panel	Individua	al Hemiplegic Migraine single gene tests:	
	22 Neuronal Ceroid Lipofuscinosis		□ 11		103 CACNA1A Sequencing Test
<u> </u>	33 Syndromic Disorders			36 SCN1A Sequencing Test (FHM)	

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 (Genes, Antibodies, Comments) Column below for specimen requirement exceptions.

Test Test Code Name	Additional Information (Genes, Antibodies, Comments)	Test Code	Test Name	Additional Information (Genes, Antibodies, Comments)
Motor Neuron Diseases: Molecular Genetics	(**************************************		ent Disorders: Molecular Genetics (Continu	(
☐ 6520 Amyotrophic Lateral Sclerosis Advanced			al Isolated Dystonia single gene tests:	
Evaluation ☐ 6522 Nonprevalent Amyotrophic Lateral Sclerosis	Please see website for the complete list of genes.	Only ord	er single gene tests when not ordering the panel. 26 Dystonia (DYT1) DNA Test 18 THAP1 DNA Sequencing Test	
Advanced Sequencing Evaluation 670 C9orf72 DNA Test			Complete Dopa-Responsive Dystonia	GCH1 Seg., GCH1 Del., TH Seg.
☐ 620 SOD1 DNA Sequencing Test		Individue	(DYT5) Evaluation	, , ,
☐ 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 ord	al Dopa-Responsive Dystonia single gene tests: er single gene tests when not ordering the panel. 37 GCH1 DNA Sequencing Test 38 GCH1 Deletion Analysis 34 TH DNA Sequencing Test	DYT5A DYT5A DYT5B
G601 USB Common Charadia Evaluation	not ordered. SPAST, SPG7		SGCE DNA Sequencing Test	DYT11
6601 HSP, Common Sporadic Evaluation	· · · · · · · · · · · · · · · · · · ·	☐ 627 ☐ 617	SGCE Deletion Analysis PNKD (MR-1) DNA Sequencing Test	DYT11
☐ 6602 HSP, Supplemental Sporadic Evaluation	Please see website for the complete list of genes.	□ 588		LRRK2, PARK2, PINK1, PARK7, SNCA
☐ 6610 HSP, Complete Dominant Evaluation ☐ 6611 HSP, Common Dominant Evaluation	SPAST, ATLN, REEP1, KIF5A		al Parkinsonism single gene tests:	
☐ 6612 HSP, Supplemental Dominant Evaluation	BSCL2, HSPD1, KIAA0196, NIPA1, RTN2, SLC33A1	☐ 55 ☐ 55	58 LRRK2 DNA Sequencing Test	059 Alpha Synuclein (SNCA) Dup./Del. Test 559 PARK2 (Parkin) DNA Sequencing Test
☐ 6620 HSP, Complete Recessive Evaluation	Please see website for the complete list of genes.	□ 04	10 PARK2 (Parkin) Duplication/Deletion Test	554 PARK7 (DJ1) DNA Sequencing Test 542 PINK1 DNA Sequencing Test
☐ 6621 HSP, Common Recessive Evaluation	SPG11, ZFYVE26, SPG7		PRRT2 (Dyskinesia/IC) Seq. Test	
☐ 6622 HSP, Supplemental Recessive Evaluation			e Sclerosis/Demylenating Diseases: Immuno	ology
☐ 6631 HSP. X-Linked Evaluation	of genes. L1CAM, PLP1		NMO Spectrum Evaluation	AQP4, CBA reflex to MOG, CBA
☐ 6509 SPG4 Evaluation	SPAST		Aquaporin-4 (AQP4) (NMO IgG) Antibody,	Cerebrospinal Fluid (CSF) is an
Movement Disorders: Molecular Genetics	SPASI		CBA with Reflex to Titer	acceptable sample type.
Individual HSP DNA Tests:		□ 1523	Myelin Oligodendrocyte Glycoprotein (MOG)	Cerebrospinal Fluid (CSF) is an
Only order single gene tests when not ordering the panel.		T 1001	Antibody, ČBA with Reflex to Titer NMO Spectrum Evaluation	acceptable sample type. AQP4. ELISA reflex to MOG. CBA
531 Atlastin	SPG3A	□ 1204	Aquaporin-4 (AQP4) Antibody (NMO-lgG),	AQP4, ELISATEIIEX (0 MOG, CBA
☐ 632 Paraplegin	SPG7	193	ELISA	
☐ 633 Spatacsin	SPG11	□ 112	NAbFeron® (INFB-1) Neutralizing Antibody Test	
☐ 614 ZFYVE26	SPG15	□ 197	TYSABRI® (Natalizumab) Antibody Test	See website for collection notes
☐ 117 Kennedy's Disease (SBMA) DNA Test		Myasth	enia Gravis: Immunology	
☐ 6930 Ataxia, Comprehensive Evaluation	Please see website for the complete list of genes. Test 6930 contains all genes		Myasthenia Gravis Panel 2 with Reflex to MuSK Antibody	
	included in the sub-panels. NOTE: Only select sub-panels if 6930 is		Myasthenia Gravis Panel 2	Includes AChR Binding / Blocking / Modulating Antibody
	not ordered.		MuSK and LRP4	
	Cannot be performed on saliva.	1310	Acetylcholine Receptor Binding Antibody with Reflex to Musk Antibody	
☐ 6900 Ataxia, Complete Dominant Evaluation		□ 1511	Acetylcholine Receptor Binding Antibody	
☐ 6901 Ataxia, Common Repeat Expansion Evaluation	Please see website for the complete list of genes.		with Reflex to MuSK/LRP4 Antibodies al Myasthenia Gravis single antibody tests:	
☐ 6903 Ataxia, Supplemental Dominant Evaluation	Cannot be performed on saliva.	Only ord	der single antibody tests when not ordering the Acetylcholine Receptor Binding Antibody	☐ 1483 LRP4 Autoantibody Test
☐ 6910 Ataxia, Complete Recessive Evaluation			Acetylcholine Receptor Blocking Antibody Acetylcholine Receptor Modulating Antibody	☐ 1481 RyR Autoantibody Test☐ 1480 Titin Autoantibody Test
☐ 6911 Ataxia, Supplemental Recessive Evaluation	Please see website for the complete list of genes.	☐ 482	MuSK Antibody Test evelopmental Disorders: Molecular Genetic	•
☐ 6912 Oculomotor Apraxia Ataxia Advanced	APTX, SETX		Primary Microcephaly Sequencing Evaluation	
Sequencing Evaluation		Individua	al Primary Microcephaly single gene tests:	Ther in, mer irr, WBROL
G920 Episodic Ataxia Evaluation	CACNB4, KCNA1, SLC1A3, CACNA1A	Only ord	er single gene tests when not ordering the panel.	
349 Ataxia, Friedreich (FXN) Evaluation	FRDA/FXN Seq., FRDA/FXN Expansion		J92_ASPM Sequencing Test □ 1° 257_WDR62 Sequencing Test	153 MCPH1 Sequencing Test
353 Ataxia-Telangiectasia (ATM) Evaluation	ATM Seq., ATM Dup./Del.		SHANK3 Sequencing Test	
Individual Ataxia single gene DNA Tests: Only order single gene tests when not ordering the panel			SHANK2 Sequencing Test	
or sub-panels.			PTEN Sequencing Test	
☐ 401 DRPLA ☐ 119 FRDA/FXN Expansion)			Joubert Syndrome Evaluation	
☐ 348 FRDA/FXN Seg. ☐ 383 POLG1 (MIRAS)	SCA8 and SCA10 test cannot be		al Joubert Syndrome single gene tests:	
☐ 371 SCA1 (ATXN1) ☐ 672 SCA2 (ATXN2)	performed on saliva.	Only ord	er single gene tests when not ordering the panel. 30 AHI1 DNA Sequencing Test	94 CC2D2A DNA Sequencing Test
☐ 105 SCA3 (ATXN3) ☐ 373 SCA6 (CACNA1A)		79	91 CEP290 DNA Sequencing Test 79	
☐ 677 SCA7 (ATXN7) ☐ 384 SCA8 (ATXN8OS)				92 TMEM67 DNA Sequencing Test
☐ 387 SCA10 (ATXN10) ☐ 285 SCA12 (PPP2R2B)			MECP2 Sequencing and CNV Evaluation	
☐ 388 SCA17 (TBP) ☐ 283 TTPA (AVED)			14 CDKL5 Seq. and CNV Evaluation	
☐ 402 Chorea Differential Evaluation (DRPLA, Huntington's Disease)	Cannot be performed on saliva.		(Atypical Rett) Rett Syndrome (MECP2) Dup./Del. Test	
☐ 116 Huntington Disease Repeat Expansion Test	Cannot be performed on saliva.	□ 737	Smith-Lemli-Opitz Syndrome (DHCR7)	
Solated Dystonia Evaluation	DYT1, THAP1	□ 40F0	DNA Sequencing Test	
1 000 ISUIALEU DYSLUINA EVAIUALIUN	DITI, INAFI	1256	VPS13B (COH1) Sequencing Test	l

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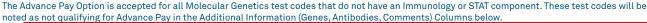
NOTE2: The pediatric minimum is 2 mL for Neurodevelopmental Disorders & Epilepsy.

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)
Neurode	evelopmental Disorders: Molecular Genetic		Paraneo	pplastic & Other Antibody Disorders of the	CNS: Immunology
	ARX Seq. and CNV Evaluation (Intellectual Disability)	(00000000000000000000000000000000000000		Paraneoplastic Neurological Syndromes	Cerebrospinal Fluid (CSF) is an
	SYNGAP1 Sequencing Test			Evaluation with Recombx®, Initial Assessment	acceptable sample type. Amphiphysin, CV2, Hu, MaTa, Ri, Yo
	MEF2C Sequencing and CNV Evaluation FOXG1 Sequencing and CNV Evaluation		□ 4620	NeoComplete Paraneoplastic Evaluation with Recombx®	
	uscular Disorders: Molecular Genetics		□ 4640	Paraneoplastic Autoantibody Evaluation with	* NOTE: Cerebrospinal Fluid (CSF) is
	Muscular Dystrophy Advanced Evaluation		1 4040	Recombx [®] , CSF *	an acceptable sample type
	Congenital Muscular Dystrophy Advanced Sequencing Evaluation		□ 4724	NeoCerebellar Degeneration Paraneoplastic Profile with Recombx®	I loade dee website for the complete list
□ 5503	Congenital Myopathy Advanced Sequencing Evaluation	Please see website for the complete list	□ 4722	NeoEncephalitis Paraneoplastic Evaluation with Recombx®	of antibodies.
□ 5504	Distal Myopathy Advanced Sequencing Evaluation	of genes.	□ 4725	NeoSensory Neuropathy Paraneoplastic Profile with Recombx®	Cerebrospinal Fluid (CSF) is an acceptable sample type.
	Myofibrillar Myopathy Advanced Sequencing Evaluation		□ 4727	Neuromyotonia Evaluation	Amphiphysin, CV2, Hu CASPR2, VGKC
□ 5506	Myotonic Syndromes Advanced Evaluation	Please see website for the complete list of genes.	Indiv	dual antibody Tests:	
		Cannot be performed on saliva.	□ 419	order single antibody tests when not ordering NMDA Receptor Autoantibody Test*	☐ 4681 Recombx® CV2 Autoantibody Test *
□ 5507	Periodic Paralysis Advanced Sequencing Evaluation		☐ 422 ☐ 428	GAD65 Neurological Syndrome Antibody Test	 ☐ 4682 Recombx® Hu Autoantibody Test * ☐ 4683 Recombx® MaTa Autoantibody Test *
□ 5508	Malignant Hyperthermia Advanced Sequencing Evaluation		1 449	LGI1 Antibody Test* VGCC Type P/Q Autoantibody Test (LEMS)	4684 Recombx® CAR (Anti-Recoverin) Autoantibody Test *
□ 5511	Congenital Myasthenic Syndrome Advanced Sequencing Evaluation	Please see website for the complete list	∟ 485	VGKC Antibody Test	☐ 4685 Recombx® Ri Autoantibody Test * ☐ 4686 Recombx® Yo Autoantibody Test *
<u></u> 5518	Emery-Dreifuss Muscular Dystrophy	of genes.	4674	Recombx® Amphiphysin Autoantibody Test *	4689 Recombx [®] Zic4 Autoantibody Test *
☐ EE10	Advanced Sequencing Evaluation Limb Girdle Muscular Dystrophy Advanced			Cerebrospinal Fluid (CSF) is an acceptable s	
0019	Evaluation			ral Neuropathy (Hereditary): Molecular Gen	
Only ord	al Limb Girdle Muscular Dystrophy Tests: er single gene tests when not ordering the par 3 Calpain 3 DNA Sequencing Test 5 6 CAV3 DNA Sequencing Test 5 5 LMNA DNA Sequencing Test 5	nel. 584 CAPN3 Duplication/Deletion Test 562 FKRP DNA Sequencing Test 582 SGCA Duplication/Deletion Test	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,
	3 SGCG Duplication/Deletion Test DMD Evaluation				MTMR2, SBF2 DNA Seq.
	al DMD Evaluation single gene tests:		□ □ 40	002 CMT Advanced Evaluation – Dominant,	Testing is performed in this order: 1. PMP22 Dup./Del. If negative: 2. MPZ,
Only orde	re single gene tests when not ordering the panel. 3 DMD DNA Sequencing Test 31 DMD Duplication/Deletion Test			Demyelinating (Reflexive)	PMP22 Seq., EGR2, LITAF, DNM2, YARS DNA Seq.
207	Early-Onset Myotonia Evaluation	DM1, CLCN1, SCN4A		003 CMT Advanced Evaluation – Dominant, Axonal	Please see website for the complete list
Individua	al Early-Onset Myotonia single gene tests:	Cannot be performed on saliva.	□ 40	004 CMT Advanced Evaluation – Recessive,	of genes.
Only orde	er single gene tests when not ordering the panel. 8 CLCN1 DNA Sequencing Test 6 SCN4A (Myotonia) DNA Sequencing Test		<u></u> 40	Demyelinating 005 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,
	DMPK DNA Test (DM1) CNBP DNA Test (DM2) (DM2 testing is not	Cannot be performed on saliva.			GARS, NFL, HSPB1, DNM2, YARS,
110	recommended for patients with early onset myotonic dystrophy)	Cannot be performed on saliva.		006 CMT Advanced Evaluation – Recessive	TRPV4, HSPB8 DNA Seq. Please see website for the complete list of genes.
□ 585	CAPN3 Evaluation	Includes CAPN3 Seq., CAPN3 Del.	□ 40	007 CMT Advanced Evaluation – Demyelinating (Reflexive)	Testing is performed in this order: 1. PMP22 Dup./Del. If negative: 2. Cx32,
	Dysferlin DNA Sequencing Test			,	MPZ. PMP22 Sea., EGR2, LITAF.
□ 405	FSHD1 Southern Blot Test	Specimen Type: Whole Blood Specimen Requirements: 10 mL			PRX, GDAP1, DNM2, YARS, SH3TC2, MTMR2, NDRG1, FGD4, FIG4, SBF2 DNA Seg
		(7 mL minimum) whole blood collected in two (lavender-top) EDTA tubes		008 CMT Advanced Evaluation – Axonal	DIAN OCY
		Sample must be received within 72		010 CMT Advanced Evaluation – Initial	-
		hours of collection and refrigerated. Ship sample M-Th only		Genetic Assessment	
		Cannot be performed on saliva or extracted DNA.	□ 40	011 CMT Advanced Evaluation – Nonprevalent Axonal	Please see website for the complete list
□ 300	OPMD Repeat Expansion Test	Cannot be performed on saliva.	□ 40	012 CMT Advanced Evaluation –	of genes.
	OPA1 DNA Sequencing Test (optic atrophy)	Related to optic atrophy.		Nonprevalent Demyelinating	_
Neuro-C	Incology: Molecular Genetics		□ 40	113 CMT Advanced Evaluation –	
□ 648	Neurofibromatosis Type 1 (NF1) Evaluation	NF1 Sequencing, NF1 Deletion	Individue	Nonprevalent al CMT single gene tests:	
Individua	al NF1 single gene tests:	•	Only ord	er single gene tests when not ordering the panel of	or sub-panels.
Only orde	er single gene tests when not ordering the panel.		<u> </u> 14	I3 CX32 Seq./Del. (CMTX) ☐ 253 DNM2	☐ 248 EGR2 (CMT1D)
☐ 64	 Neurofibromatosis Type 1 Deletion Test Neurofibromatosis Type 1 DNA Sequencir 	ng Test		08 FGD4 ☐ 225 FIG4 (CI 21 GDAP1 (CMT2K, 4A) ☐ 229 HSPB1 (
	Neurofibromatosis Type 2 (NF2) Evaluation	NF2 Seq., NF2 Dup./Del.	☐ 22	22 LITAF/SIMPLE (CMT1C) ☐ 226 LMNA (C 34 MPZ (CMT1B, 2I, 2J) ☐ 354 MTMR2	
Individua	al NF2 single gene tests:	1	□ 24	I9 NFL (CMT2E, 1F) ☐ 131 PMP22 □	Dup./Del. (CMT1A) 247 PMP22 Seq.
Only orde	er single gene tests when not ordering the panel.	a Toot	☐ 23	39 PRX (CMT4F) □ 227 RAB7A (CMT2B) ⊂ ☐ 164 SBF2
☐ 64	 Neurofibromatosis Type 2 DNA Sequencir Neurofibromatosis Type 2 Duplication/Dele 	ig rest etion Test		24 SH3TC2 (CMT4C)	

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 (Genes, Antibodies, Comments) Column below for specimen requirement exceptions Test Test **Additional Information** Test Test **Additional Information** Code Name (Genes, Antibodies, Comments) Code Name (Genes, Antibodies, Comments) Peripheral Neuropathy (Hereditary): Molecular Genetics (Continued) Spinal Muscular Atrophy (SMA): Molecular Genetics Does not qualify for the Advance Pay Option. ☐ 691 Early-Onset HSAN Evaluation NTRK1 and WNK1 5056 SMA Carrier Screen (New York) Test Codes are for New York State Clients 243 Complete HNPP Evaluation PMP22 Sequencing, PMP22 Dup./Del. ordering SMA testing. **245** Congenital Hypomyelination Evaluation MPZ, EGR2 ☐ 5026 SMA Diagnostic (New York) 4 mL (2 mL minimum) whole blood collected in ☐ 296 Entrapment Neuropathy Evaluation PMP22 Seq., PMP22 Dup./Del., TTR an EDTA (lavender-top) tube. ☐ 5070 SMA Plus (New York) Pediatric (0-3 years): 2 mL (1 mL minimum). Peripheral Neuropathy (Hereditary Sensory Autonomic Neuropathy): Molecular Genetics Individual Early-Onset HSAN single gene tests: 7 214 SMA Plus (Reflexive) Only order single gene tests when not ordering the panel.

— 659 NTRK1 (HSAN IV) DNA Sequencing Test 111 Spinal Muscular Atrophy-Diagnostic Spinal Muscular Atrophy-Carrier ☐ 553 WNK1 (HSAN II) DNA Sequencing Test 211 Spinal Muscular Atrophy - SMN1 DNA SPTLC1 and SPTLC2 ☐ 698 Late-Onset HSAN Evaluation Does not qualify for the Advance Pay Sequencing Test Individual Late-Onset HSAN single gene tests: Option. 6521 Atypical SMA Advanced Sequencing Only order single gene tests when not ordering the panel.
 □ 551 SPTLC1 (HSAN I) DNA Sequencing Test
 □ 552 SPTLC2 (HSAN I) DNA Sequencing Test
 Evaluation Test 214 includes 111 with reflex to 211. ☐ 660 ATL1 (HSAN I) DNA Sequencing Test ☐ 719 SEPT9 (HNA) DNA Sequencing Test Peripheral Neuropathy (Autoimmune): Immunology GM1 Quattro®, MAG 'Dual Antigen'®, Hu, ☐ 3100 SensoriMotor Neuropathy Profile with Recombx® - Complete GALOPTM, Sulfatide (MAG 'Dual Antigen'®, Hu, GALOPTM, ☐ 3148 Sensory Neuropathy Profile with Recombx® Sulfatide) ☐ 3163 Motor Neuropathy Profile - Complete GM1 Quattro®, MAG 'Dual Antigen'® Requires both Serum and whole blood. ☐ 289 Multifocal Motor Neuropathy Evaluation

	GM1 Quattro [®] , PMP22 Dup./Del.		
☐ 3155 Co-GM1 Quattro® Autoantibody Test	(Asialo, GD1a, GD1b and GM1)		
☐ 210 Sulfatide Autoantibody Test	272 Asialo Aŭtoantibody Test 273 GD1b Autoantibody Test 271 GM1 Autoantibody Test 4682 Recombx® Hu Autoantibody Test *		
	RENAL GENI	ETIC TESTING	
Test Test	Additional Information	Test Test	Additional Information
Code Name	(Genes, Antibodies, Comments)	Code Name	(Genes, Antibodies, Comments)
Alport Syndrome: Molecular Genetics		Monogenic Hypertension: Molecular Genetics	
☐ 759 Complete Alport Syndrome Evaluation	COL4A3,4,5 DNA Sequencing; COL4A5 Deletion Test		SCNN1B, SCNN1G, CYP11B1, HSD11B2
Individual Alport Syndrome single gene tests:		☐ 747 Liddle's Syndrome Evaluation	SCNN1B, SCNN1G
Only order single gene tests when not ordering the panel. 757 COL4A3 DNA Sequencing Test 758 758 COL4A5 Deletion Analysis 755		Evaluation	SCNN1A, SCNN1B, SCNN1G
Amyloidosis: Molecular Genetics		Individual Monogenic Hypertension single gene tests:	
☐ 235 TTR DNA Sequencing Test		Only order single gene tests when not ordering the panel. 779 CYP11B1/CYP11B2 Chimeric Gene Fusion	Total
Bardet-Biedl Syndrome: Molecular Genetics			☐ 775 HSD11B2 DNA Sequencing Test
☐ 887 Bardet-Biedl Syndrome Evaluation	BBS1, BBS2, BBS10		☐ 745 SCNN1B DNA Sequencing Test
Individual Bardet-Biedl Syndrome single gene tests:		☐ 746 SCNN1G DNA Sequencing Test	_ : : : : : : : : : : : : : : : : : : :
Only order single gene tests when not ordering the panel.		Nephrogenic Diabetes Insipidus: Molecular Genetic	·s
☐ 871 BBS1 (BBS) DNA Sequencing Test ☐ 886 BBS10 (BBS) DNA Sequencing Test	372 BBS2 (BBS) DNA Sequencing Test	☐ 854 Nephrogenic Diabetes Insipidus	AVPR2, AQP2
Family Testing:		Evaluation Individual Nephrogenic Diabetes Insipidus single gene	tooto
☐ 185 Familial DNA Sequence Evaluation	This test detects previously identified	Only order single gene tests when not ordering the panel.	lesis.
Proband Accession #	sequence variants in at-risk family		51 AVPR2 DNA Sequencing Test
Relationship	members. For Familial PKD1 and PKD2	Nephronophthisis: Molecular Genetics	
· Colditorionip	variants, please order Code 728.	☐ 750 NPHP1 Deletion Test (Familial Juvenile	
		Nephronophthisis)	
Hereditary Renal Tubular Disorders: Molecular Ger	etics	Nephrotic Syndrome: Molecular Genetics	
☐ 767 Hereditary Renal Tubular Disorders Evaluation	SLC12A1, KCNJ1, CLCNKB, BSND, SLC12A3	☐ 722 Early Onset Nephrotic Syndrome Evaluation	PLCE1, LAMB2, WT1, NPHS1, NPHS2
	ne tests: 764	☐ 718 PLCE1 DNA Sequencing Test ☐	712 TRPC6 DNA Sequencing Test 713 WT1 DNA Sequencing Test 710 NPHS2 DNA Sequencing Test
For additional information on test-specific genes and re	quirements (preferred volume, specimen, an	d tube type), please visit AthenaDiagnostics.com or call 1.80	00.394.4493. Reviewed December 2023

Reflexive testing is performed at an additional charge.





oted 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.

Ple	<u>ease Refer to the Additional Information (C</u>	<u>ienes, Antibodies, Comments) Column b</u>	e <u>low for s</u>	specimen requirement exceptions.	
Test	Test	Additional Information	Test	Test	Additional Information
Code	Name	(Genes, Antibodies, Comments)	Code	Name	(Genes, Antibodies, Comments)
Nephrotic Syndrome: Molecular Genetics (Continued)			Other C	Cystic Diseases: Molecular Genetics (Contin	ued)
	Focal and Segmental Glomerulosclerosis (FSGS) Evaluation al FSGS single gene tests:	INF2, ACTN4, TRPC6, NPHS2	□ 523	TSC Familial Mutation Evaluation Proband Accession # Relationship	
	er single gene tests when not ordering the panel.			<u> </u>	
	16 INF2 DNA Sequencing Test	710 NPHS2 DNA Sequencing Test	□ 770	Hereditary Interstitial Kidney Disease (UMOD) DNA Sequencing Test	
	stic Kidney Disease: Molecular Genetics		Renal C	Cancer: Molecular Genetics	
□ 728	PKDx® Familial Mutation Evaluation	Does not qualify for the Advance Pay Option.	□ 889	Pheochromocytoma Evaluation	RET, VHL, SDHB
	Proband Accession #Relationship	PKD1 and PKD2 Variants		al Pheochromocytoma single gene tests: ler single gene tests when not ordering the panel.	
8100	Complete PKDx Evaluation	Does not qualify for the Advance Pay Option.		13 MEN2 (RET) DNA Sequencing Test	☐ 888 SDHB DNA Sequencing Test
	al PKDx single gene tests:		☐ 858 von Hippel-Lindau Syndrome (VHL) DNA Sequencing Test		
	er single gene tests when not ordering the panel. 105 PKD1 Deletion Test		□ 8	18 MEN1 DNA Sequencing Test	
	103 PKD1 Deletion Test 101 PKD1 DNA Sequencing and Deletion Evaluation		Renal C	Cysts and Diabetes: Molecular Genetics	
□ 8 <i>′</i> □ 8 <i>′</i>	103 PKD1 DNA Sequencing Test 106 PKD2 Deletion Test	Does not qualify for the Advance Pay Option.	□ 776	HNF1ß DNA Sequencing and Deletion Evaluation (RCAD)	
	102 PKD2 DNA Sequencing and Deletion Evaluation		Rickets	: Molecular Genetics	
	104 PKD2 DNA Sequencing Test		□ 857	Hypophosphatemic Rickets Evaluation	PHEX, FGF23
Other Cystic Diseases: Molecular Genetics □ 1131 Complete Tuberous Sclerosis				al Hypophosphatemic Rickets single gene tests	3:
1131	Sequencing and CNV Evaluation	TSC1 & TSC2	1 1	ler single gene tests when not ordering the panel.	0
Individual Tuberous Sclerosis single gene tests:				FGF23 (Hypophosphatemic Rickets) DNAPHEX (Hypophosphatemic Rickets) DNA	
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					

	524 TSC2 DNA Deletion Test (for NYS Only) 1255 TSC2 Sequencing Test		
	ENDOCRINE GE	NETIC TESTING	
Test Test	Additional Information	Test Test	Additional Information
Code Name	(Genes, Antibodies, Comments)	Code Name	(Genes, Antibodies, Comments)
Adrenal Disorders: Molecular Genetics		Congenital Hyperinsulinism: Molecular Genetics	
☐ 816 Primary Adrenal Insufficiency Evaluation	ABCD1, NR0B1, AIRE	☐ 819 Congenital Hyperinsulinism Evaluation	Does not qualify for the Advance Pay
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			Option. GLUD1, GCK, KCNJ11, ABCC8 Indication for Study (check one or more below): Diazoxide Responsive Diazoxide Non-Responsive
□ 879 Congenital Adrenal Hyperplasia (CAH) Evaluation	Includes CYP21A2 sequencing and deletion, CYP11B1 sequencing		☐ Hypoglycemic ☐ Large for Gestational Age (LGA) ☐ Other (describe)
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)	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. B22 GLUD1 (CHI) DNA Sequencing Test B23 GCK (CHI) DNA Sequencing Test B26 KCNJ11 (CHI) DNA Sequencing Test B27 ABCC8 (CHI) DNA Sequencing Test CHI Parental Testing – To augment child/Does not qualify for the Advance	
□ 877 CYP17A1 DNA Sequencing Test □ 881 Endocrine Hypertension (HSD11B2) Evaluation	elevated concentration in serum	proband diagnosis	Option. For expedited diagnosis of proband, send parental testing samples as soon as possible and provide information below. Mother Father Proband Name/Accession #
☐ 878 HSD3B2 DNA Sequencing Test		Diabetes: Molecular Genetics	Trobalia Hallion Goodeleli II
☐ 874 Lipoid CAH (STAR) DNA Sequencing Test		885 Monogenic Diabetes (MODY) Five-Gene	HNF1A (TCF1), GCK, HNF4A.
Bone Diseases: Molecular Genetics	1001414 001410	Evaluation	HNF1B (TCF2), IPF1
☐ 860 Osteogenesis Imperfecta Evaluation Individual Osteogenesis Imperfecta single gene tests	COL1A1, COL1A2	☐ 8800 Monogenic Diabetes (MODY) Four-Gene Evaluation	HNF1A (TCF1), GCK, HNF4A, HNF1B (TCF2)
Only order single gene tests when not ordering the panel 861 COL1A1 (OI) DNA Sequencing Test	□ 862 COL1A2 (OI) DNA Sequencing Test	☐ 8801 Monogenic Diabetes (MODY) Three-Gene Evaluation	HNF1A (TCF1), GCK, HNF1B (TCF2)
☐ 857 Hypophosphatemic Rickets Evaluation Individual Hypophosphatemic Rickets single gene tes	PHEX, FGF23	☐ 8802 Monogenic Diabetes (MODY) Two-Gene Evaluation	HNF1A (TCF1), GCK
Only order single gene tests when not ordering the panel 856 FGF23 (Hypophosphatemic Rickets) DNA 855 PHEX (Hypophosphatemic Rickets) DNA	A Sequencing Test	□ 803 GCK (MODY2) DNA Sequencing and Deletion Test □ 802 HNF4A (MODY1) DNA Sequencing and Deletion Test	
☐ 811 LRP5 (OPPG) DNA Sequencing Test		834 IPF1 (MODY4) DNA Sequencing Test	
□ 821 LRP5 Idiopathic Osteoporosis (IOP) DNA Sequencing Test		□ 804 TCF1 (MODY3) DNA Sequencing and Deletion Test	HNF1A (TCF1)

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

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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 Test	Additional Information	Test Test	Additional Information
Code Name	(Genes, Antibodies, Comments)	Code Name	(Genes, Antibodies, Comments)
Diabetes: Molecular Genetics (Continued)		Obesity: Molecular Genetics	
☐ 805 TCF2 (MODY5) DNA Sequencing and	HNF1B (TCF2)	☐ 884 Early Onset Obesity Evaluation	LEPR, MC4R
Deletion Test		Individual Early Onset Obesity single gene tests:	
☐ 837 CEL (MODY8) Mutation Analysis		Only order single gene tests when not ordering the part	
■ 882 Neonatal Diabetes Mellitus Evaluation	IPF1, GCK, KCNJ11, INS, ABCC8	☐ 640 Early Onset Obesity (MC4R) DNA Sec	
Individual Neonatal Diabetes Mellitus single gene tests	3:	☐ 883 Early Onset Obesity (LEPR) DNA Seq	uencing Test
Only order single gene tests when not ordering the panel.		☐ 887 Bardet-Biedl Syndrome Evaluation	BBS1, BBS2, BBS10
	3 842 GCK (NDM) DNA Sequencing Test3 841 IPF1 (NDM) DNA Sequencing Test	Individual Bardet-Biedl Syndrome single gene tests	5:
843 KCNJ11 (NDM) DNA Sequencing Test	J 641 IFFT (NDIVI) DIVA Sequending Test	Only order single gene tests when not ordering the part	
Nephrogenic Diabetes: Molecular Genetics		☐ 871 BBS1 (BBS) DNA Sequencing Test	☐ 872 BBS2 (BBS) DNA Sequencing Test
1 0		☐ 886 BBS10 (BBS) DNA Sequencing Test	
☐ 854 Nephrogenic Diabetes Insipidus Evaluation	AVPR2, AQP2	Reproductive Disorders: Molecular Genetics	
Individual Nephrogenic Diabetes Mellitus single gene t	teete:	☐ 679 Complete Kallmann/IHH Evaluation	
Only order single gene tests when not ordering the panel.	.6515.	Individual Kallmann/IHH single gene tests:	
852 AQP2 (Nephrogenic Diabetes Insipidus))NA Sequencing Test	Only order single gene tests when not ordering the pa	
☐ 851 Nephrogenic Diabetes Insipidus (AVPR2)	DNA Sequencing Test		☐ 195 FGF8 DNA Sequencing Test
Familial Cancer Syndromes: Molecular Genetics	3		☐ 343 GnRH1 DNA Sequencing Test ☐ 173 KAL1 DNA Sequencing Test
818 MEN1 DNA Sequencing Test			☐ 175 RALT DNA Sequencing Test
■ 889 Pheochromocytoma Evaluation	RET, VHL, SDHB		☐ 358 TACR3 DNA Sequencing Test
•	REI, VIIE, SDIIB	462 Anosmic Kallmann/IHH Evaluation	
Individual Pheochromocytoma single gene tests:		667 Normosmic Kallmann/IHH Evaluation	Please see website for the complete list of
Only order single gene tests when not ordering the panel. 813 MEN2 (RET) DNA Sequencing Test	☐ 888 SDHB DNA Seguencing Test		genes.
□ 858 von Hippel-Lindau Syndrome (VHL) DNA		☐ 817 Male Precocious Puberty (LHCGR) DNA	
	· · ·	Sequencing Test	
Familial Hypocalciuric Hypercalcemia: Molecular	Senetics	Short Stature: Molecular Genetics 865 Combined Pituitary Hormone Deficience	
☐ 829 Familial Hypocalciuric Hypercalcemia (CASR) DNA Seguencing Test		Evaluation	PROP1, POU1F1
Family Testing		Individual Pituitary Hormone Deficiency single gen	tests:
☐ 185 Familial DNA Sequence Evaluation	This test detects previously identified	Only order single gene tests when not ordering the part	
· ·	sequence variants in at-risk family	☐ 864 POU1F1 (CPHD) DNA Sequencing Te	
Proband Accession #	members.	☐ 863 PROP1 (CPHD) DNA Sequencing Tes	
Relationship		☐ 848 Growth Hormone Deficiency Evaluatio	n GH1 and GHRHR Seq.; SHOX Seq.
Noonan Syndrome: Molecular Genetics			and Del.
□ 846 Noonan Syndrome (PTPN11) DNA		Individual Growth Hormone Deficiency single gene	17 7 7
Sequencing Test		Only order single gene tests when not ordering the particular of t	
☐ 658 KRAS/RAF1/SOS1 DNA Sequencing		☐ 866 GH1 (GHD) DNA Sequencing Test	
Evaluation	SOS1, RAF1, KRAS	☐ 847 SHOX (GHD) DNA Sequencing and D	eletion Test
Individual KRAS/RAF1/SOS1 single gene tests:		☐ 867 GHR DNA Sequencing Test	
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			