Athena Diagnostics Neurology Test Requisition (October 2015)

Fields in red indicate required information

Please tear at perforation

Who Should Athena Contact with Questions About this Order?

Name _____ (First) _____ (Last)

Phone _____ Fax _____

Email _____

Patient Identification

Patient Name _____

Patient ID # (if available) _____ Sex: ☐ Male ☐ Female ☐ Unknown

DOB _____

Age _____

Mailing Address _____

_____

City _____

Province _____ Postal Code _____

Phone _____ ☐ Day ☐ Eve ☐ Cell

Patient Authorization to Use, Transmit and Retain Personal Health Information Outside of Canada. Your personal health information will be collected and used by Athena Diagnostics for diagnostic testing and analysis purposes. Your personal health information, including your blood specimen, will be transferred to and processed by Athena Diagnostics in its secure laboratory in Marlborough, Massachusetts. Athena Diagnostics will maintain your information on a confidential basis, and will not disclose your personal information except as required by applicable law, which may include lawful access by courts, governmental authorities or law enforcement in the US. **I consent to the foregoing:** ☐ **Yes** ☐ **No**

_____

Signature of Patient, Parent or Legally Authorized Representative Date

_____

Printed Name of Patient, Parent or Legally Authorized Representative Date

_____

Relationship to Patient if Signatory is Someone Other than Patient

Alternative Physician Attestation of Patient Authorization. I warrant that I have obtained written consent from the patient to use, transmit and retain patient's personal health information outside of Canada substantially as described in the above Patient Authorization.

_____

Medical Practitioner Signature Date

_____

Printed Name of Medical Practitioner

Billing Information

Please indicate responsible party (check only one):

☐ **Ministry of Health** *(Prior approval required before testing begins)*

☐ Prior Approval Included ☐ Prior Approval Pending

☐ **Hospital/Laboratory**

Athena Account # (if assigned) _____

Purchase Order # (if available) _____

Billing Contact _____

Email _____

Phone _____ Fax _____

Hospital/Lab Name _____

Address _____

City _____

Province _____ Postal Code _____

☐ **Self Pay** Payer Information:

Name _____ (First) _____ (Last)

Credit Card # _____

Credit Card Expiration Date _____ Security Code _____

Phone _____

Address _____

City _____

Province _____ Postal Code _____

Tests Ordered

Important: Write in the test code and test name.

Code _____ Name _____

Code _____ Name _____

Authorized Result Report Recipients Required Physician Information

Name _____ (First) _____ (Last)

Address _____

City _____

Province _____ Postal Code _____

Phone _____ Fax _____

Email _____

Indications for Testing (Check One)

☐ Diagnostic (symptomatic) ☐ Prenatal ☐ Family Testing

☐ Predictive (asymptomatic) ☐ Carrier

Physician Attestation of Informed Consent

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

I warrant that I have obtained both oral and written consent using the **Patient Informed Consent Form for Genetic Testing** provided by Athena Diagnostics. This written consent was signed by the person who is the subject of the test (or if that person lacks capacity to consent, signed by the person authorized to consent for that person).

_____

Medical Practitioner Signature Date

_____

Printed Name of Medical Practitioner NPI

Patient Informed Consent Form for Genetic Testing is available at AthenaDiagnostics.com/consent.

Specimen Collection Laboratory Information

Lab Name _____

Address _____

City _____

Province _____ Postal Code _____

Phone _____ Fax _____

Type of Specimen Date Collected _____

☐ Whole Blood ☐ Serum ☐ CSF ☐ Buccal Swabs

☐ Muscle ☐ CVS: Direct ☐ CVS: Cultured

☐ Amniotic Fluid: Direct ☐ Amniotic Fluid: Cultured

NOTE: Specimen tube(s) must be labeled with two of the following forms of identification: name, date of birth, patient ID no. These same two forms of ID must also be indicated on the test requisition.

Athena Diagnostics Neurology Testing Services (October 2015)

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Test Code		Pref. Spec.	Pref. Vol.	Tube Type
Cerebrovascular Disease (Stroke)				
☐ 421	Complete CADASIL Evaluation* (Notch3 Sequencing)	B	8 mL	L
☐ 442	HTRA1 DNA Sequencing Test* (CARASIL)	B	8 mL	L
☐ 424	COL4A1 DNA Sequencing Test* (CSVD)	B	8 mL	L
☐ **692**	**Complete Cerebral Cavernous Malformation (CCM) Evaluation*** (KRIT1 Seq./Del., CCM2 Seq./Del., PDCD10 Seq./Del.)	B	8 mL	L
☐ **683**	**KRIT1 (CCM1) Evaluation*** (KRIT1 Sequencing/Deletion)	B	8 mL	L
☐ **686**	**CCM2 Evaluation*** (CCM2 Sequencing/Deletion)	B	8 mL	L
☐ **689**	**PDCD10 (CCM3) Evaluation*** (PDCD10 Sequencing/Deletion)	B	8 mL	L
☐ 681	KRIT1 (CCM1) DNA Sequencing Test*	B	8 mL	L
☐ 682	KRIT1 (CCM1) Deletion Test*	B	8 mL	L
☐ 684	CCM2 DNA Sequencing Test*	B	8 mL	L
☐ 685	CCM2 Deletion Test*	B	8 mL	L
☐ 687	PDCD10 (CCM3) DNA Sequencing Test*	B	8 mL	L
☐ 688	PDCD10 (CCM3) Deletion Test*	B	8 mL	L
Dementia				
☐ **178**	**ADmark® Alzheimer's Evaluation*** (ApoE, Phospho-Tau, Total-Tau, Aß42) (Symptomatic for Dementia) (CSF must be in polypropylene tube and arrive on cold pack)	C B	2 mL 8 mL	P L
☐ 109	ADmark® ApoE Genotype Analysis & Interpretation* (Symptomatic for Dementia)	B	8 mL	L
☐ 177	ADmark® Phospho-Tau/Total-Tau/Aß42 CSF Analysis & Interpretation (CSF must be in polypropylene tube and arrive on cold pack or frozen)	C	2 mL	P
☐ **179**	**ADmark® Early-Onset Alzheimer's Evaluation*** (PS-1, APP Seq./Dup., PS-2)	B	8 mL	L
☐ 167	ADmark® PSEN1 DNA Sequencing Test*	B	8 mL	L
☐ 168	ADmark® APP DNA Sequencing Test and Duplication Test *	B	8 mL	L
☐ 169	ADmark® PSEN2 DNA Sequencing Test*	B	8 mL	L
☐ **281**	**Frontotemporal Dementia (FTD) Evaluation*** (MAPT, GRN, C9orf72)	B	8 mL	L
☐ 209	C9orf72 DNA Test*	B	8 mL	L
☐ 204	GRN DNA Sequencing Test*	B	8 mL	L
☐ 205	MAPT DNA Sequencing Test*	B	8 mL	L
☐ **1700**	**Autoimmune Rapidly Progressive Dementia Evaluation** (Hu, MaTa, CV2, Amphiphysin, GAD65, NMDA, VGKC, LGI1, CASPR2)	S	2 mL	R
☐ 1701	Recombx Hu Autoantibody Test (Autoimmune Rapidly Progressive Dementia)*	S	2 mL	R
☐ 1702	Recombx MaTa Autoantibody Test (Autoimmune Rapidly Progressive Dementia)*	S	2 mL	R
☐ 1703	Recombx CV2 Autoantibody Test (Autoimmune Rapidly Progressive Dementia)*	S	2 mL	R
☐ 1704	Amphiphysin Antibody Test (Autoimmune Rapidly Progressive Dementia)*	S	2 mL	R
☐ 1705	GAD65 Autoantibody Test (Autoimmune Rapidly Progressive Dementia)*	S	2 mL	R
☐ 1706	NMDA Receptor Autoantibody Test (Autoimmune Rapidly Progressive Dementia)*	S	2 mL	R
☐ 1707	VGKC Autoantibody Test (Autoimmune Rapidly Progressive Dementia)*	S	2 mL	R
☐ 1708	LGI1 Autoantibody Test (Autoimmune Rapidly Progressive Dementia)*	S	2 mL	R
☐ 1709	CASPR2 Autoantibody Test (Autoimmune Rapidly Progressive Dementia)*	S	2 mL	R
Developmental Disabilities				
☐ **788**	**Primary Microcephaly Evaluation*** (ASPM, MCPH1, WDR62)	B	8 mL	L
☐ 784	ASPM DNA Sequencing Test*	B	8 mL	L
☐ 786	MCPH1 DNA Sequencing Test*	B	8 mL	L
☐ 787	WDR62 DNA Sequencing Test*	B	8 mL	L
☐ 742	SHANK3 DNA Sequencing Test*	B	8 mL	L
☐ 724	SHANK2 DNA Sequencing Test*	B	8 mL	L
☐ 744	PTEN DNA Sequencing Test*	B	5 mL	L
☐ **795**	**Joubert Syndrome Evaluation*** (TMEM67, TMEM216, AHI1, CEP290, NPHP1, CC2D2A)	B	8 mL	L
☐ 792	TMEM67 DNA Sequencing Test*	B	8 mL	L
☐ 789	TMEM216 DNA Sequencing Test*	B	8 mL	L
☐ 790	AHI1 DNA Sequencing Test*	B	8 mL	L
☐ 791	CEP290 DNA Sequencing Test*	B	8 mL	L
☐ 793	NPHP1 DNA Deletion Test*	B	8 mL	L
☐ 794	CC2D2A DNA Sequencing Test*	B	8 mL	L
☐ 737	Smith-Lemli-Opitz Syndrome (DHCR7) DNA Test*	B	8 mL	L
☐ 729	Cohen Syndrome (COH1) DNA Seq. Test*	B	8 mL	L
☐ **153**	**Complete Rett Syndrome Evaluation*** (MECP2 Seq., MECP2 Duplication/Deletion)	B	8 mL	L
☐ 142	Rett Syndrome (MECP2) DNA Seq. Test*	B	8 mL	L
☐ 148	Rett Syndrome (MECP2) Dup./Del. Test*	B	8 mL	L
☐ **773**	**ARX Evaluation*** (ARX DNA Seq., ARX Dup./Del.)	B	8 mL	L
☐ 141	ARX DNA Sequencing Test*	B	8 mL	L
☐ 041	ARX Duplication/Deletion Test*	B	8 mL	L
☐ **785**	**CDKL5 Evaluation*** (CDKL5 Seq., CDKL5 Dup./Del.)	B	8 mL	L
☐ 149	CDKL5 DNA Sequencing Test*	B	8 mL	L
☐ 049	CDKL5 Duplication/Deletion Test*	B	8 mL	L
☐ 771	SYNGAP1 DNA Sequencing Test*	B	8 mL	L
☐ **7540**	**MEF2C Evaluation*** (MEF2C DNA Seq., MEF2C Del.)	B	4 mL	L
☐ 754	MEF2C DNA Sequencing Test*	B	4 mL	L
☐ 077	MEF2C Deletion Test*	B	4 mL	L
☐ **7410**	**FOXG1 Evaluation*** (FOXG1 DNA Seq., FOXG1 Del.)	B	4 mL	L
☐ 740	FOXG1 DNA Sequencing Test*	B	4 mL	L
☐ 074	FOXG1 Deletion Test*	B	4 mL	L
NOTE: Pediatric minimum for all Developmental Disabilities tests is 2 mL.				
Epilepsy				
☐ **5000**	**Epilepsy Advanced Sequencing Evaluation***	B	8 mL	L
☐ **5001**	**Epilepsy Advanced Sequencing Evaluation – Generalized, Absence, Focal and Myoclonus Epilepsies***	B	8 mL	L
☐ **5002**	**Epilepsy Advanced Sequencing Evaluation – Epileptic Encephalopathies***	B	8 mL	L
☐ **5003**	**Epilepsy Advanced Sequencing Evaluation – Neuronal Migration Disorders***	B	8 mL	L
☐ **5004**	**Epilepsy Advanced Sequencing Evaluation – Epilepsy in X-Linked Intellectual Disability***	B	8 mL	L
☐ **5005**	**Epilepsy Advanced Sequencing Evaluation – Neuronal Ceroid Lipofuscinosis***	B	8 mL	L
☐ **5006**	**Epilepsy Advanced Sequencing Evaluation – Epilepsy Associated with Migraine***	B	8 mL	L
☐ **5007**	**Epilepsy Advanced Sequencing Evaluation – Syndromic Disorders with Epilepsy***	B	8 mL	L
☐ **5008**	**Epilepsy Advanced Sequencing Evaluation – Infantile Spasms***	B	8 mL	L
Please see website for the list of genes in each panel.				
☐ **5100**	**Autoimmune Epilepsy Evaluation** GAD65 Neurological Syndrome, VGKC, CASPR2, LGI1, NMDA (NR1-subunit) Autoantibody Test	S	2 mL	R
☐ 5101	GAD65 Neurological Syndrome Autoantibody Test (Epilepsy) (Single)	S	2 mL	R
☐ 5102	VGKC Autoantibody Test (Epilepsy) (Single)	S	2 mL	R
☐ 5103	CASPR2 Autoantibody Test (Epilepsy) (Single)	S	2 mL	R
☐ 5104	LGI1 Autoantibody Test (Epilepsy) (Single)	S	2 mL	R
☐ 5105	NMDA Receptor (NR1-subunit) Autoantibody Test (Epilepsy) (Single)	S	2 mL	R
☐ **556**	**Complete Tuberous Sclerosis Evaluation*** (TSC1 Seq., TSC1 Del., TSC2 Seq., TSC2 Del.)	B	8 mL	L
☐ 521	TSC1 DNA Sequencing Test*	B	8 mL	L
☐ 508	TSC1 DNA Deletion Test*	B	8 mL	L
☐ 522	TSC2 DNA Sequencing Test*	B	8 mL	L
☐ 524	TSC2 DNA Deletion Test*	B	8 mL	L
☐ 523	TSC Familial DNA Seq. Mutation Evaluation* Proband Accession # _____ Relationship _____	B	8 mL	L
☐ **573**	**SCN1A Complete Evaluation*** (SCN1A Sequencing, SCN1A Deletion)	B	8 mL	L
☐ 537	SCN1A Deletion Test*	B	8 mL	L
☐ **674**	**CSTB (EPM1) Evaluation*** (CSTB (EPM1) DNA Test, CSTB (EPM1) Seq.)	B	10 mL	L
☐ 410	CSTB (EPM1) (Unverricht-Lundborg) DNA Test*	B	8 mL	L
☐ **797**	**ARX Evaluation*** (ARX Seq., ARX Dup./Del.)	B	8 mL	L
☐ **799**	**CDKL5 Evaluation*** (CDKL5 Seq., CDKL5 Dup./Del.)	B	8 mL	L
☐ 065	ARX Duplication/Deletion Test*	B	8 mL	L
☐ 067	CDKL5 Duplication/Deletion Test*	B	8 mL	L
☐ 549	Alexander Disease (GFAP) DNA Seq. Test*	B	8 mL	L
☐ 443	POLG DNA Seq. Test* (Alpers Syndrome)	B	8 mL	L
NOTE: Pediatric minimum for all Epilepsy tests is 2 mL.				
Family Testing				
☐ 185	Familial DNA Sequence Evaluation* This test detects previously identified sequence variants in at-risk family members. This test cannot be applied to the TTR gene. For Familial TSC mutations, please order Code 523. Proband Accession # _____ Relationship _____	B	8 mL	L
Hearing Loss				
☐ **329**	**Connexin Related Deafness Evaluation*** (Connexin 26, Connexin 30)	B	8 mL	L
☐ 321	Connexin 26 DNA Sequencing Test*	B	8 mL	L
☐ 319	Connexin 30 DNA Deletion Test*			
Leukodystrophy				
☐ 421	Complete CADASIL Evaluation* (Notch3 Sequencing)	B	8 mL	L
☐ **6106**	**Leukoencephalopathy with Vanishing White Matter Evaluation*** (EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5)	B	8 mL	L
☐ 6101	EIF2B1 DNA Sequencing Test*	B	8 mL	L
☐ 6102	EIF2B2 DNA Sequencing Test*	B	8 mL	L
☐ 6103	EIF2B3 DNA Sequencing Test*	B	8 mL	L
☐ 6104	EIF2B4 DNA Sequencing Test*	B	8 mL	L
☐ 6105	EIF2B5 DNA Sequencing Test*	B	8 mL	L
☐ 6107	ARSA DNA Sequencing Test*	B	8 mL	L
☐ 6108	ABCD1 DNA Sequencing Test*	B	8 mL	L
☐ **6110**	**PLP1 Evaluation*** (PLP1 Seq., PLP1 Dup.)	B	8 mL	L
☐ 6112	PLP1 DNA Sequencing Test*	B	8 mL	L
☐ 6111	PLP1 Duplication Test*	B	8 mL	L
☐ 6109	GJC2 DNA Sequencing Test*	B	8 mL	L
☐ 549	Alexander Disease (GFAP) DNA Seq. Test*	B	8 mL	L
Migraine				
☐ **190**	**Hemiplegic Migraine Evaluation*** (CACNA1A, ATP1A2, SCN1A)	B	8 mL	L
☐ 187	CACNA1A (FHM1) DNA Test*	B	8 mL	L
☐ 188	ATP1A2 (FHM2) DNA Test*			
☐ 189	SCN1A (FHM3) DNA Test* (Exons 3, 23, 26)			
Mitochondrial Disorders				
☐ **575**	**Common Mitochondrial Disorders Evaluation*** (POLG, MELAS, MERRF, NARP)	B	8 mL	L
☐ **576**	**Progressive External Ophthalmoplegia (PEO) Evaluation*** (POLG, TWINKLE, ANT1, OPA1, MELAS)	B	8 mL	L
☐ **577**	**Mitochondrial Neurogastrointestinal Encephalopathy (MNGIE) Evaluation*** (TYMP, RRM2B, MELAS)	B	8 mL	L
☐ **578**	**Mitochondrial Hepatoencephalopathic Evaluation*** (POLG, DGUOK, MPV17, TWINKLE)	B	8 mL	L
☐ **579**	**Mitochondrial Encephalomyopathic Evaluation*** (TK2, RRM2B, POLG)	B	8 mL	L
☐ **515**	**LHON mtDNA Evaluation*** (LHON 11778, 3460, 14484)	B	8 mL	L
☐ 474	POLG DNA Sequencing Test* (Related to all allelic disorders)	B	8 mL	L
☐ 479	TWINKLE (PEO1/C10orf2) DNA Seq. Test* (Related to mtDNA depletion)	B	8 mL	L
☐ 466	ANT1 (SLC25A4) DNA Sequencing Test* (Related to mtDNA depletion)	B	8 mL	L
☐ 469	OPA1 DNA Sequencing Test* (Related to mtDNA depletion)	B	8 mL	L
☐ 484	TYMP DNA Sequencing Test* (Related to mtDNA depletion)	B	8 mL	L
☐ 486	RRM2B DNA Sequencing Test* (Related to mtDNA depletion)	B	8 mL	L

Important: Please be sure to write in test code and test name in the Tests Ordered section on front.

Not all available tests are listed here. Please see our catalog or website for complete offering, as well as CPT codes for each test.

Important: Please be sure to write in test code and test name in the Tests Ordered section on front.

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Test Code		Pref. Spec.	Pref. Vol.	Tube Type
☐ 487	DGUOK DNA Sequencing Test* (Related to mtDNA depletion)	B	8 mL	L
☐ 488	MPV17 DNA Sequencing Test* (Related to mtDNA depletion)	B	8 mL	L
☐ 489	TK2 DNA Sequencing Test* (Related to mtDNA depletion)	B	8 mL	L
☐ 517	MELAS mtDNA Evaluation* (MELAS 3243, 3271, 3252, 3256, 3291, 13513)	B	8 mL	L
☐ 518	MERRF mtDNA Evaluation* (MERRF 8344, 8356, 8296, 8363)	B	8 mL	L
☐ 516	NARP mtDNA Evaluation* (NARP 8993)	B	8 mL	L
☐ 824	PDHA1 DNA Sequencing Test*	B	8 mL	L
Motor Neuron Diseases				
☐ **6520**	**Amyotrophic Lateral Sclerosis Advanced Sequencing Evaluation*** (ALS2, ANG, CHMPB2, C9ORF72, DCTN1, FIG4, FUS, OPTN, PFN1, SETX, SIGMAR1, SOD1, SQSTM1, TARDBP, UBQLN2, VAPB, VCP)	B	8mL	L
☐ **6522**	**Nonprevalent Amyotrophic Lateral Sclerosis Advanced Sequencing Evaluation*** (ALS2, ANG, CHMPB2, DCTN1, FIG4, FUS, OPTN, PFN1, SETX, SIGMAR1, SQSTM1, TARDBP, UBQLN2, VAPB, VCP)	B	8mL	L
☐ 670	C9orf72 DNA Test*	B	10 mL	L
☐ 620	SOD1 DNA Sequencing Test*	B	10 mL	L
☐ **6601**	**HSP, Common Sporadic Evaluation*** (SPAST, SPG7)	B	8 mL	L
☐ **6602**	**HSP, Supplemental Sporadic Evaluation*** (ATLN, REEP1, KIF5A, NIPA1, KIAA0196, BSCL2, HSPD1, RTN2, SLC33A1, SPG11, CYP7B1, ZFYVE26, ALS2, AP5Z1, FA2H, KIF1A, PNPLA6, SACS, SPG20, SPG21, L1CAM, PLP1)	B	8 mL	L
☐ **6610**	**HSP, Complete Dominant Evaluation*** (SPAST, ATLN, REEP1, KIF5A, NIPA1, KIAA0196, BSCL2, HSPD1, RTN2, SLC33A1)	B	8 mL	L
☐ **6611**	**HSP, Common Dominant Evaluation*** (SPAST, ATLN, REEP1, KIF5A)	B	8 mL	L
☐ **6612**	**HSP, Supplemental Dominant Evaluation*** (NIPA1, KIAA0196, BSCL2, HSPD1, RTN2, SLC33A1)	B	8 mL	L
☐ **6620**	**HSP, Complete Recessive Evaluation*** (SPG11, ZFYVE26, SPG7, CYP7B1, ALS2, AP5Z1, FA2H, KIF1A, PNPLA6, SACS, SPG20, SPG21)	B	8 mL	L
☐ **6621**	**HSP, Common Recessive Evaluation*** (SPG11, ZFYVE26, SPG7)	B	8 mL	L
☐ **6622**	**HSP, Supplemental Recessive Evaluation*** (CYP7B1, ALS2, AP5Z1, FA2H, KIF1A, PNPLA6, SACS, SPG20, SPG21)	B	8 mL	L
☐ **6630**	**HSP, Comprehensive Evaluation*** (SPAST, SPG7, ATLN, REEP1, KIF5A, NIPA1, KIAA0196, BSCL2, HSPD1, RTN2, SLC33A1, SPG11, CYP7B1, ZFYVE26, ALS2, AP5Z1, FA2H, KIF1A, PNPLA6, SACS, SPG20, SPG21, L1CAM, PLP1)	B	8 mL	L
☐ **6631**	**HSP, X-Linked Evaluation*** (L1CAM, PLP1)	B	8 mL	L
☐ **6509**	**SPG4 Evaluation***	B	8 mL	L
Individual HSP DNA Tests: ☐ 531 Atlastin (SPG3A)* ☐ 632 Paraplegin (SPG7)* ☐ 633 Spatacsin (SPG11)* ☐ 614 Spastizin/ZFYVE26 (SPG15)*		B	8 mL	L
☐ **214**	**SMA Plus (Reflexive)*** This is a reflexive test. Tests will be run in succession until either a positive result is detected or the profile is completed. Testing is performed in this order: 1. SMN1 Del.; 2. SMN1 Seq.	B	4 mL	L
☐ 111D	Spinal Muscular Atrophy Deletion - Diagnostic* (including SMN2 Copy Number)	B	4 mL	L
☐ 211	Spinal Muscular Atrophy - SMN1 DNA Seq. Test* (only order if deletion testing has already been performed)	B	4 mL	L
☐ 444	Spinal Muscular Atrophy - Carrier SMN1 Del. Test*	B	4 mL	L
☐ 117	Kennedy's Disease (SBMA) DNA Test*	B	8 mL	L
☐ **6521**	**Atypical Spinal Muscular Atrophy Advanced Sequencing Evaluation*** (BICD2, DYNC1H1, GARS, HSPB1, HSPB3, HSPB8, IGHMBP2, TRPV4, UBA1, VRK1)	B	8 mL	L
Movement Disorders				
☐ **6900**	**Ataxia, Complete Dominant Evaluation** (ATXN1, ATXN2, ATXN3, CACNA1A, ATXN7, TBP, ATXN8OS, ATXN10, PPP2R2B, ATN1, AFG3L2, KCNC3, PRKCG, SPTBN2, EEF2, FGF14, ITPR1, KCND3, PDYN, TGM6, TTBK2, VAMP1, KCNA1, CACNB4, SLC1A3, CACNA1A)	B	10 mL	L
☐ **6901**	**Ataxia, Common Repeat Expansion Evaluation** (ATXN1, ATXN2, ATXN3, CACNA1A, ATXN7, TBP, ATXN8OS, ATXN10)	B	8 mL	L
☐ **6903**	**Ataxia, Supplemental Dominant Evaluation** (AFG3L2, KCNC3, PRKCG, SPTBN2, EEF2, FGF14, ITPR1, KCND3, PDYN, TGM6, TTBK2, VAMP1, KCNA1, CACNB4, SLC1A3, CACNA1A)	B	8 mL	L
☐ **6910**	**Ataxia, Complete Recessive Evaluation** (FXN, APTX, ATM, SETX, TTPA, ADCK3, AFG3L2, ANO10, FLVCR1, GRM1, MRE11A, MTPAP, SACS, SYNE1, SYT14, TDP1, SIL1, POLG)	B	8 mL	L
☐ **6911**	**Ataxia, Supplemental Recessive Evaluation** (APTX, ATM, SETX, TTPA, ADCK3, AFG3L2, ANO10, FLVCR1, GRM1, MRE11A, MTPAP, SACS, SYNE1, SYT14, TDP1, SIL1, POLG)	B	8 mL	L
☐ **6912**	**Oculomotor Apraxia Ataxia Advanced Sequencing Evaluation** (APTX, SETX)	B	8 mL	L
☐ **6920**	**Episodic Ataxia Evaluation** (CACNB4, KCNA1, SLC1A3, CACNA1A)	B	8 mL	L
☐ **6930**	**Ataxia, Comprehensive Evaluation** (ATXN1, ATXN2, ATXN3, CACNA1A, ATXN7, TBP, ATXN8OS, ATXN10, PPP2R2B, ATN1, AFG3L2, KCNC3, PRKCG, SPTBN2, EEF2, FGF14, ITPR1, KCND3, PDYN, TGM6, TTBK2, VAMP1, KCNA1, CACNB4, SLC1A3, CACNA1A, FXN, APTX, ATM, SETX, TTPA, ADCK3, AFG3L2, ANO10, FLVCR1, GRM1, MRE11A, MTPAP, SACS, SYNE1, SYT14, TDP1, SIL1, POLG)	B	10 mL	L
☐ **349**	**Ataxia, Friedreich (FXN) Evaluation*** (FRDA/FXN Seq., FRDA/FXN Expansion)	B	8 mL	L
☐ **353**	**Ataxia-Telangiectasia (ATM) Evaluation*** (ATM Seq., ATM Dup./Del.)	B	8 mL	L
Individual Ataxia DNA Tests: ☐ 371 SCA1* ☐ 373 SCA6* ☐ 387 SCA10* ☐ 401 DRPLA* ☐ 283 TTPA (AVED)* ☐ 119 FRDA/FXN Expansion* ☐ 672 SCA2* ☐ 677 SCA7* ☐ 285 SCA12* ☐ 383 POLG1 (MIRAS)* ☐ 348 FRDA/FXN Seq.* ☐ 105 SCA3* ☐ 384 SCA8* ☐ 388 SCA17*		B	8 mL	L
☐ **402**	**Chorea Differential Evaluation*** (DRPLA, HD)	B	8 mL	L
☐ 116	Huntington's Disease DNA Test*	B	8 mL	L
☐ **639**	**Primary Dystonia Evaluation*** (DYT1, THAP1)	B	8 mL	L
☐ 626	Dystonia (DYT1) DNA Test*	B	8 mL	L
☐ 618	THAP1 (DYT6) DNA Sequencing Test*	B	8 mL	L
☐ **629**	**Complete Dopa-Responsive Dystonia (DYT5) Evaluation*** (GCH1 Seq., GCH1 Del., TH Seq.)	B	8 mL	L
☐ 637	GCH1 DNA Sequencing Test* (DYT5)	B	8 mL	L
☐ 638	GCH1 Deletion Test (DYT5)*	B	8 mL	L
☐ 634	TH DNA Sequencing Test (DYT5)*	B	8 mL	L
☐ 624	SGCE DNA Sequencing Test (DYT11)*	B	8 mL	L
☐ 627	SGCE Deletion Test (DYT11)*	B	8 mL	L
☐ 617	MR-1 (PNKD) DNA Sequencing Test*	B	8 mL	L
☐ **588**	**Complete Parkinsonism Evaluation*** (LRRK2, PARK2, PINK1, PARK7, SNCA)	B	8 mL	L
☐ 558	LRRK2 DNA Sequencing Test*	B	8 mL	L
☐ 559	PARK2 (Parkin) DNA Sequencing Test*	B	8 mL	L
☐ 040	PARK2 (Parkin) Duplication/Deletion Test*	B	8 mL	L
☐ 542	PINK1 DNA Sequencing Test*	B	8 mL	L
☐ 058	PINK1 Deletion Test*	B	8 mL	L
☐ 554	PARK7 (DJ1) DNA Sequencing Test*	B	8 mL	L
☐ 047	PARK7 (DJ1) Deletion Test*	B	8 mL	L
☐ 557	Alpha Synuclein (SNCA) DNA Seq. Test*	B	8 mL	L
☐ 059	Alpha Synuclein (SNCA) Dup./Del. Test*	B	8 mL	L
☐ 666	PRRT2 (Dyskinesia/IC) DNA Seq. Test*	B	8 mL	L
Multiple Sclerosis				
☐ 112	NAbFeron® (IFN-ß) Neutralizing Antibody Test	S	2 mL	R
☐ 197	TYSABRI® (Natalizumab) Antibody Test (must arrive on cold pack)	S	2 mL	R
☐ 193	Neuromyelitis Optica (NMO) Autoantibody Test	S	2 mL	R
Myasthenia Gravis				
☐ 482	MuSK Quantitative Titers Antibody Test	S	2 mL	R
☐ 483	AChR/MuSK Reflexive Antibody Test (Now with MuSK quantitative titers levels)	S	2 mL	R
☐ 1480	Titin Autoantibody Test	S	2 mL	R
Neuromuscular Disorders				
☐ **5501**	**Muscular Dystrophy Advanced Evaluation**	B	8 mL	L
☐ **5502**	**Congenital Muscular Dystrophy Advanced Sequencing Evaluation**	B	8 mL	L
☐ **5503**	**Congenital Myopathy Advanced Sequencing Evaluation**	B	8 mL	L
☐ **5504**	**Distal Myopathy Advanced Sequencing Evaluation**	B	8 mL	L
☐ **5505**	**Myofibrillar Myopathy Advanced Sequencing Evaluation**	B	8 mL	L
☐ **5506**	**Myotonic Syndromes Advanced Sequencing Evaluation**	B	8 mL	L
☐ **5507**	**Periodic Paralysis Advanced Sequencing Evaluation**	B	8 mL	L
☐ **5508**	**Malignant Hyperthermia Advanced Sequencing Evaluation**	B	8 mL	L
☐ **5518**	**Emery-Dreifuss Muscular Dystrophy Advanced Sequencing Evaluation**	B	8 mL	L
☐ **5519**	**Limb Girdle Muscular Dystrophy Advanced Evaluation**	B	8 mL	L
☐ **5511**	**Congenital Myasthenic Syndrome Advanced Sequencing Evaluation**	B	8 mL	L
☐ **5530**	**DMD Evaluation**	B	8 mL	L
☐ 5531	DMD Duplication/Deletion	B	8 mL	L
☐ 183	Partial DMD DNA Sequencing Only*	B	8 mL	L
☐ 100	Dystrophin Test	M	10 mg	C
☐ **207**	**Early-Onset Myotonia Evaluation*** (DM1, CLCN1, SCN4A)	B	8 mL	L
☐ 108	DM1 DNA Test*	B	8 mL	L
☐ 110	DM2 DNA Test* (DM2 testing is not recommended for patients with early onset myotonic dystrophy)			
☐ 128	CLCN1 DNA Test*			
☐ 146	SCN4A DNA Test*			
☐ **494**	**Neuromyotonia Evaluation** (CASPR2, VGKC Antibody Tests)	S	2 mL	R
☐ **585**	**CAPN3 Evaluation*** (includes CAPN3 Seq., CAPN3 Del.)	B	8 mL	L
Individual Limb Girdle Muscular Dystrophy Tests: ☐ 562 FKRP* ☐ 565 LMNA* ☐ 566 CAV3* ☐ 582 Sarcoglycan A Deletion Test* ☐ 583 Sarcoglycan G Deletion Test* ☐ 584 CAPN3 Deletion Test*		B	8 mL	L
☐ 561	Dysferlin Protein Blood Test* (must arrive on cold pack)	B	10 mL	L
☐ 571	Dysferlin Sequencing Test*	B	8 mL	L
☐ 405	FSHD Southern Blot Test*	B	15 mL	L
☐ 5905	FSHD Molecular Combing Test*	B	15 mL	L
☐ 300	OPMD DNA Test*	B	8 mL	L
☐ **490**	**Optic Atrophy Evaluation*** (OPA1)	B	8 mL	L
Neuro-Oncology				
☐ **648**	**Neurofibromatosis Type 1 (NF1) Evaluation*** (NF1 Sequencing, NF1 Deletion)	B	8 mL	L
☐ **645**	**Neurofibromatosis Type 2 (NF2) Evaluation*** (NF2 Seq., NF2 Dup./Del.)	B	8 mL	L
☐ 646	Neurofibromatosis Type 1 DNA Sequencing Test*	B	8 mL	L
☐ 647	Neurofibromatosis Type 1 Deletion Test*	B	8 mL	L
☐ 635	Neurofibromatosis Type 2 DNA Sequencing Test*	B	8 mL	L
☐ 644	Neurofibromatosis Type 2 Duplication/Deletion Test*	B	8 mL	L
Note: Additional specimens accepted. Please contact Lab Director.				
Paraneoplastic & Other Antibody Disorders of the CNS				
☐ **4500**	**Paraneoplastic Neurological Syndromes Initial Assessment (PNS-IA)** (Hu, Yo, CV2, MaTa, Ri, Amphiphysin)	S or C	2 mL or 2 mL	R P**
☐ **467**	**NeoComplete Paraneoplastic Evaluation with Recombx® (Reflexive)** Hu, Yo, Zic4, CV2, MaTa, Ri, CAR, VGCC, VGKC, Amphiphysin, gnAChR, NR1, GAD65 Neurological Syndrome, LGI1, CASPR2.	S	2 mL	R
☐ **438**	**NeoCerebellar Degeneration Paraneoplastic Evaluation with Recombx®** (Hu, Yo, Zic4, CV2, MaTa, Ri, Amphiphysin, GAD65 Neurological Syndrome)	S	2 mL	R
☐ **447**	**NeoEncephalitis Paraneoplastic Evaluation with Recombx®** (Hu, CV2, MaTa, VGKC, Amphiphysin, NR1, GAD65 Neurological Syndrome, LGI1, CASPR2)	S	2 mL	R
☐ **436**	**NeoSensory Neuropathy Paraneoplastic Evaluation with Recombx®** (Hu, CV2, Amphiphysin)	S	2 mL	R
☐ **494**	**Neuromyotonia Evaluation** (CASPR2, VGKC)	S	2 mL	R
Individual Recombx® Antibody Tests: ☐ 118 CAR ☐ 123 CV2 ☐ 120 Hu ☐ 122 MaTa ☐ 115 Ri ☐ 125 Yo ☐ 127 Zic4		S	2 mL	R

Athena Diagnostics Neurology Testing Services (October 2015)

Test Code		Pref. Spec.	Pref. Vol.	Tube Type
☐ 449	LGI1 Antibody Test	S	2 mL	R
☐ 499	CASPR2 Antibody Test	S	2 mL	R
☐ 419	NMDA Receptor (NR1-subunit) Antibody Test	S	2 mL	R
☐ 422	GAD65 Neurological Syndrome Antibody Test	S	2 mL	R
☐ 475	LEMS (VGCC) Antibody Test	S	2 mL	R
☐ 485	VGKC Antibody Test	S	2 mL	R
☐ 427	Amphiphysin Antibody Test	S	2 mL	R
☐ 428	Ganglionic AChR (gnAChR) Antibody Test	S	2 mL	R
Peripheral Neuropathy: Autoimmune				
☐ **287**	**SensoriMotor Neuropathy Evaluation** (Co-GM1 Quattro®, MAG 'Dual Antigen'®, Hu, GALOP™, Sulfatide)	S	2 mL	R
☐ **263**	**Sensory Neuropathy Evaluation** (MAG 'Dual Antigen'®, Hu, GALOP™, Sulfatide)	S	2 mL	R
☐ **288**	**Motor Neuropathy Evaluation** (Co-GM1 Quattro®, MAG 'Dual Antigen'®)	S	2 mL	R
☐ **289**	**Multifocal Motor Neuropathy Evaluation*** (Co-GM1 Quattro®, PMP22 Dup./Del.)	S B	2 mL 8 mL	R L
☐ 277	Co-GM1 Quattro® Antibody Test	S	2 mL	R
☐ 145	MAG 'Dual Antigen'® Autoantibody Test	S	2 mL	R
☐ 261	GALOP™ Antibody Test	S	2 mL	R
☐ 210	Sulfatide Autoantibody Test	S	2 mL	R
☐ 160	GQ1b Autoantibody Test	S	2 mL	R
☐ 278	GD1a Antibody Test	S	2 mL	R
☐ 272	Co-Asialo Antibody Test	S	2 mL	R
☐ 273	Co-GD1b Antibody Test	S	2 mL	R
☐ 271	Co-GM1 Antibody Test	S	2 mL	R
Peripheral Neuropathy: Hereditary				
☐ **4010**	**CMT Advanced Evaluation – Initial Genetic Assessment** (PMP22 Dup./Del., GJB1 (Cx32), MPZ, MFN2 Seq.)	B	8 mL	L
☐ **4011**	**CMT Advanced Evaluation – Nonprevalent Axonal** (GJB1 (Cx32) Del., NFL, GDAP1, GARS, RAB7, HSPB1, DNM2, YARS, LMNA, TRPV4, HSPB8 Seq.)	B	8 mL	L
☐ **4012**	**CMT Advanced Evaluation – Nonprevalent Demyelinating** (GJB1 (Cx32) Del., EGR2, LITAF, PMP22, PRX, GDAP1, DNM2, YARS SH3TC2, MTMR2, NDRG1, FGD4, FIG4, SBF2 Seq.)	B	8 mL	L
☐ **4013**	**CMT Advanced Evaluation – Nonprevalent** (GJB1 (Cx32) Del., PMP22, EGR2, LITAF, PRX, GDAP1, RAB7, GARS NFL, HSPB1, LMNA, FIG4, SH3TC2, DNM2, YARS, FGD4, NDRG1, TRPV4, HSPB8, MTMR2, SBF2 Seq.)	B	8 mL	L
☐ **4001**	**CMT Advanced Evaluation – Comprehensive (Reflexive)*** Testing is performed in this order: 1. PMP22 Dup./Del. If negative: 2. Cx32, PMP22, MFN2, MPZ, EGR2, LITAF, PRX, GDAP1, RAB7, GARS, NFL, HSPB1, LMNA, FIG4, SH3TC2, DNM2, YARS, FGD4, NDRG1, TRPV4, HSPB8, MTMR2, SBF2 DNA Seq.	B	8 mL	L
☐ **4002**	**CMT Advanced Evaluation – Dominant, Demyelinating (Reflexive)*** Testing is performed in this order: 1. PMP22 Dup./Del. If negative: 2. MPZ, PMP22 Seq., EGR2, LITAF, DNM2, YARS DNA Seq.	B	8 mL	L
☐ **4003**	**CMT Advanced Evaluation – Dominant, Axonal*** (MFN2, MPZ, RAB7, GARS, NFL, HSPB1, LMNA, DNM2, YARS, TRPV4, HSPB8)	B	8 mL	L
☐ **4004**	**CMT Advanced Evaluation – Recessive, Demyelinating*** (PRX, GDAP1, SBF2, SH3TC2, MTMR2, NDRG1, FGD4, FIG4)	B	8 mL	L
☐ **4005**	**CMT Advanced Evaluation – Dominant (Reflexive)*** Testing is performed in this order: 1. PMP22 Dup./Del. If negative: 2. MFN2, MPZ, PMP22 Seq., EGR2, LITAF, RAB7, GARS, NFL, HSPB1, DNM2, YARS, TRPV4, HSPB8 DNA Seq.	B	8 mL	L
☐ **4006**	**CMT Advanced Evaluation – Recessive*** (PRX, GDAP1, SBF2, LMNA, FIG4, SH3TC2, MTMR2, NDRG1, FGD4)	B	8 mL	L
☐ **4007**	**CMT Advanced Evaluation – Demyelinating (Reflexive)*** Testing is performed in this order: 1. PMP22 Dup./Del. If negative: 2. Cx32, MPZ, PMP22 Seq., EGR2, LITAF, PRX, GDAP1, DNM2, YARS, SH3TC2, MTMR2, NDRG1, FGD4, FIG4, SBF2 DNA Seq.	B	8 mL	L
☐ **4008**	**CMT Advanced Evaluation – Axonal*** (MFN2, Cx32, MPZ, RAB7, GARS, NFL, HSPB1, GDAP1, DNM2, YARS, LMNA, TRPV4, HSPB8)	B	8 mL	L
Individual CMT Tests:		B	8 mL	L
☐ 144	TRPV4*	☐ 463	HSPB8*	
☐ 354	MTMR2*	☐ 164	SBF2*	
☐ 394	NDRG1*	☐ 208	FGD4*	
☐ 253	DNM2*	☐ 468	YARS*	
☐ 221	GDAP1 (CMT2K, 4A)*	☐ 222	LITAF/SIMPLE (CMT1C)*	
☐ 223	MFN2 (CMT2A2)*	☐ 239	Periaxin (CMT4F)*	
☐ 247	PMP22 Seq.*	☐ 248	EGR2 (CMT1D)*	
☐ 249	NFL (CMT2E, 1F)*	☐ 131	PMP22 Dup./Del. (CMT1A)*	
☐ 134	MPZ (CMT1B, 2I, 2J)*	☐ 226	LMNA (CMT2B1, 4C1)*	
☐ 224	SH3TC2 (CMT4C)*	☐ 227	RAB7 (CMT2B)*	
☐ 225	FIG4 (CMT4J)*	☐ 228	GARS (CMT2D)*	
☐ 143	Cx32 Seq./Del. (CMTX)*	☐ 229	HSPB1 (CMT2F)*	
☐ **243**	**Complete HNPP Evaluation*** (PMP22 Sequencing, PMP22 Dup./Del.)	B	8 mL	L
☐ **245**	**Congenital Hypomyelination Evaluation*** (MPZ, EGR2)	B	8 mL	L
☐ **296**	**Entrapment Neuropathy Evaluation*** (PMP22 Seq., PMP22 Dup./Del., TTR)	B	8 mL	L
☐ 235	Amyloidosis Evaluation* (TTR)	B	8 mL	L
☐ 691	**Early-Onset HSAN Evaluation*** (NTRK1 and WNK1)	B	8 mL	L
☐ 698	**Late-Onset HSAN Evaluation*** (SPTLC1 and SPTLC2)	B	8 mL	L
☐ 551	SPTLC1 (HSAN I) DNA Sequencing Test*	B	8 mL	L
☐ 552	SPTLC2 (HSAN I) DNA Sequencing Test*	B	8 mL	L
☐ 553	WNK1 (HSAN II) DNA Sequencing Test*	B	8 mL	L
☐ 659	NTRK1 (HSAN IV) DNA Sequencing Test*	B	8 mL	L
☐ 660	ATL1 (HSAN I) DNA Sequencing Test*	B	8 mL	L
☐ 719	SEPT9 (HNA) DNA Sequencing Test*	B	8 mL	L

Neurome™ Neurological Exome	
"The Neurome™ Neurological Exome requires a separate requisition. Learn more at AthenaDiagnostics.com/Neurome	
☐ 1500	Neurome™ Neurological Exome (Proband)
☐ 1501	Neurome™ Neurological Exome (Trio)
☐ 1509	Family Testing Supporting Neurome™ Analysis

Specimen Requirements

Specimen Type:
B – Blood
C – CSF
M – Muscle Tissue
S – Serum

Tube Type:
L – Lavender
R – Red
C – Cryovial
P – Polypropylene CSF Transfer Tube

** CSF must be collected in a tube not containing additives.

NOTE: Specimen tube(s) must be labeled with two of the following forms of identification: name, date of birth, patient ID no. These same two forms of ID must also be indicated on the test requisition.

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Athena Diagnostics Endocrinology Testing Services (October 2015)

athena diagnostics

Test Code		Genes Included
Adrenal Disorders		
☐ **816**	**Primary Adrenal Insufficiency (Addison's disease)**	ABCD1, NR0B1, AIRE
☐ 812	Autoimmune Polyglandular Syndrome (AIRE) DNA Sequencing Test	
☐ 815	ABCD1 DNA Sequencing Test (X-linked Adrenoleukodystrophy)	
☐ 814	NR0B1/DAX1 DNA Sequencing Test (X-linked Adrenal Hypoplasia Congenita)	
☐ 881	Endocrine Hypertension (HSD11B2) DNA Sequencing Test (Apparent Mineralocorticoid Excess)	
☐ 855	PHEX DNA Sequencing Test (X-linked Hypophosphatemic Rickets)	
☐ 856	FGF23 DNA Sequencing Test (Autosomal Dominant Hypophosphatemic Rickets)	
☐ **879**	**Congenital Adrenal Hyperplasia Evaluation** CYP21A2 sequencing and deletion, CYP11B1 sequencing	
☐ 880	CYP21A2 (CAH) DNA Sequencing and Deletion Test	
	Required: 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_____	
☐ 875	CYP11B1 (CAH) DNA Sequencing Test	
☐ 874	Lipoid CAH (STAR) DNA Sequencing Test	
☐ 877	CYP17A1 DNA Sequencing Test	
☐ 878	HSD3B2 DNA Sequencing Test	
☐ 881	Endocrine Hypertension (HSD11B2) DNA Sequencing Test	
Bone Diseases		
☐ **860**	**Osteogenesis Imperfecta Evaluation**	COL1A1, COL1A2
☐ 861	COL1A1 (OI) DNA Sequencing Test	
☐ 862	COL1A2 (OI) DNA Sequencing Test	
☐ 811	Osteoporosis-Pseudoglioma (LRP5) DNA Sequencing Test	
☐ 821	Idiopathic Osteoporosis (LRP5) DNA Sequencing Test	
☐ **857**	**Hypophosphatemic Rickets Evaluation**	PHEX, FGF23
☐ 855	PHEX (Hypophosphatemic Rickets) DNA Sequencing Test	
☐ 856	FGF23 (Hypophosphatemic Rickets) DNA Sequencing Test	
Congenital Hyperinsulinism		
☐ **819**	**Congenital Hyperinsulinism Evaluation** GLUD1, GCK, KCNJ11, ABCC8	
	Indication for Study (check one or more below):	
	☐ Diazoxide Responsive	
	☐ Diazoxide Non-Responsive	
	☐ Hypoglycemic	
	☐ Large for Gestational Age (LGA)	
	☐ Other (describe) _____	
☐ 822	GLUD1 (CH) DNA Sequencing Test	
☐ 823	GCK (CH) DNA Sequencing Test	
☐ 826	KCNJ11 (CH) DNA Sequencing Test	
☐ 827	ABCC8 (CH) DNA Sequencing Test	
☐ **042**	**CH Parental Testing – To augment child/proband diagnosis**	
	For expedited diagnosis of proband, send parental testing samples as soon as possible and provide information below.	
	☐ Mother ☐ Father	
	Proband Name/Accession #_____	

Test Code		Genes Included
Diabetes		
☐ **8800**	**Monogenic Diabetes (MODY) 4-Gene Evaluation**	GCK, HNF1A, HNF1B, HNF4A
☐ **8801**	**Monogenic Diabetes (MODY) 3-Gene Evaluation**	GCK, HNF1A, HNF1B
☐ **8802**	**Monogenic Diabetes (MODY) 2-Gene Evaluation**	GCK, HNF1A
☐ **885**	**Monogenic Diabetes (MODY) 5-Gene Evaluation**	HNF1A, GCK, HNF4A, HNF1B, IPF1
☐ 802	HNF4A (MODY1) DNA Sequencing and Deletion Test	
☐ 803	GCK (MODY2) DNA Sequencing and Deletion Test	
☐ 804	HNF1A (MODY3) DNA Sequencing and Deletion Test	
☐ 834	IPF1 (MODY4) DNA Sequencing Test	
☐ 805	HNF1B (MODY5) DNA Sequencing and Deletion Test	
☐ 837	CEL (MODY8) DNA Sequencing Test	
☐ **882**	**Neonatal Diabetes Mellitus Evaluation**	IPF1, GCK, KCNJ11, INS, ABCC8
☐ 841	IPF1 (NDM) DNA Sequencing Test	
☐ 842	GCK (NDM) DNA Sequencing Test	
☐ 843	KCNJ11 (NDM) DNA Sequencing Test	
☐ 853	INS (NDM) DNA Sequencing Test	
☐ 876	ABCC8 (NDM) DNA Sequencing Test	
Nephrogenic Diabetes		
☐ **854**	**Nephrogenic Diabetes Insipidus Evaluation**	AVPR2, AQP2
☐ 851	Nephrogenic Diabetes Insipidus (AVPR2) DNA Sequencing Test	
☐ 852	Nephrogenic Diabetes Insipidus (AQP2) DNA Sequencing Test	
Familial Cancer Syndromes		
☐ 818	MEN1 (MEN1) DNA Sequencing Test	
☐ **889**	**Pheochromocytoma Evaluation**	RET, VHL, SDHB
☐ 813	MEN2 (RET) DNA Sequencing Test	
☐ 858	von Hippel-Lindau Syndrome (VHL) DNA Sequencing Test	
☐ 888	SDHB DNA Sequencing Test	
Familial Hypocalciuric Hypercalcemia		
☐ 829	Familial Hypocalciuric Hypercalcemia (CASR) DNA Sequencing Test	
Familial Testing – Targeted Analysis		
☐ **800**	**Familial DNA Sequence Evaluation** This test detects previously identified sequence variants in at-risk family members. This test is available for HNF4A, GCK, TCF1, IPF1, TCF2, COL1A1, COL1A2, MEN1, and RET mutations	
	Proband Accession #_____ Relationship _____	
Lipid Disorders		
☐ **895**	**Hypercholesterolemia Evaluation**	LDLR, APOB
☐ 894	LDLR (Hypercholesterolemia) DNA Sequencing Test	
☐ 893	APOB Mutation Analysis	
Obesity		
☐ **884**	**Early Onset Obesity Panel**	LEPR, MC4R
☐ 883	Early Onset Obesity (LEPR) DNA Sequencing Test	
☐ 640	Early Onset Obesity (MC4R) DNA Sequencing Test	
☐ **887**	**Bardet-Biedl Syndrome Evaluation**	BBS1, BBS2, BBS10
☐ 871	BBS1 (BBS) DNA Sequencing Test	
☐ 872	BBS2 (BBS) DNA Sequencing Test	
☐ 886	BBS10 (BBS) DNA Sequencing Test	

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Athena Diagnostics Endocrinology Testing Services (October 2015)

Test Code		Genes Included
Reproductive Disorders		
☐ 817	Male Precocious Puberty (LHCGR) DNA Sequencing Test	
☐ **679**	**Complete Kallmann/IHH Evaluation**	CHD7, KAL1, PROK2, PROKR2, FGF8, FGFR1, GnRHR, GnRH1, KISS1R, TACR3
☐ **667**	**Normosmic Kallmann/IHH Evaluation**	PROK2, PROKR2, FGFR1, GnRHR, GnRH1, TACR3, KISS1R
☐ **462**	**Anosmic Kallmann/IHH Evaluation**	KAL1, PROK2, PROKR2, FGF8, FGFR1, GnRHR, KISS1R
	☐ 173 KAL1 DNA Sequencing Test	
	☐ 175 PROK2 DNA Sequencing Test	
	☐ 180 PROKR2 DNA Sequencing Test	
	☐ 195 FGF8 DNA Sequencing Test	
	☐ 196 FGFR1 DNA Sequencing Test	
	☐ 279 GnRHR DNA Sequencing Test	
	☐ 343 GnRH1 DNA Sequencing Test	
	☐ 358 TACR3 DNA Sequencing Test	
	☐ 364 KISS1R DNA Sequencing Test	
	☐ 461 CHD7 DNA Sequencing Test	

Test Code		Genes Included
Short Stature		
☐ **865**	**Combined Pituitary Hormone Deficiency Evaluation**	PROP1, POU1F1
	☐ 863 PROP1 (CPHD) DNA Sequencing Test	
	☐ 864 POU1F1 (CPHD) DNA Sequencing Test	
☐ **848**	**Growth Hormone Deficiency (GHD) Evaluation**	GH1 and GHRHR Seq.; SHOX Seq. and Del.
	☐ 866 GH1 (GHD) DNA Sequencing Test	
	☐ 868 GHRHR (GHD) DNA Sequencing Test	
	☐ 847 SHOX (GHD) DNA Sequencing and Deletion Test	
☐ 867	GHR (SS) DNA Sequencing Test	
Noonan Syndrome		
☐ 846	Noonan Syndrome (PTPN11) DNA Sequencing Test	
☐ **658**	**KRAS/RAF1/SOS1 DNA Sequencing Evaluation**	SOS1, RAF1, KRAS
	☐ 662 SOS1 DNA Sequencing Test	
	☐ 663 RAF1 DNA Sequencing Test	
	☐ 664 KRAS DNA Sequencing Test	

Endocrinology Specimen Requirements & Shipping Information (applies to all Endocrinology tests)

Specimen Type: Whole blood, 8 mL in yellow or lavender top (pediatric minimum volume: 2 mL)

Stability: Hemolysis may compromise DNA recovery and integrity after 48 hrs. It is recommended to ship samples immediately after draw. Samples can be stored for short periods only. Send specimen overnight at room temperature.

Shipping: Send specimen overnight at room temperature. If you have any questions on sample requirements or shipping, contact our client service department at 800-394-4493, extension 2.

NOTE: Specimen tube(s) must be labeled with two of the following forms of identification: name, date of birth, patient ID no. These same two forms of ID must also be indicated on the test requisition.

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Athena Diagnostics Nephrology Testing Services (October 2015)

Test Code		Pref. Spec.	Pref. Vol.	Tube Type
Alport Syndrome				
☐	**759 Complete Alport Evaluation** (COL4A3,4,5 DNA Sequencing; COL4A5 Deletion Test)	B	20 mL	L
☐	755 COL4A5 DNA Sequencing and Deletion Test	B	20 mL	L
☐	756 COL4A5 Deletion Test	B	20 mL	L
☐	757 COL4A3 DNA Sequencing Test	B	20 mL	L
☐	758 COL4A4 DNA Sequencing Test	B	20 mL	L
Amyloidosis				
☐	235 Amyloidosis Evaluation (TTR)	B	20 mL	L
Bardet-Biedl Syndrome				
☐	**887 Bardet-Biedl Syndrome Evaluation** (BBS1, BBS2, BBS10)	B	10 mL	L
☐	871 BBS1 (BBS) DNA Sequencing Test	B	10 mL	L
☐	872 BBS2 (BBS) DNA Sequencing Test	B	10 mL	L
☐	886 BBS10 (BBS) DNA Sequencing Test	B	10 mL	L
Fanconi Syndrome				
☐	517 MELAS mtDNA Evaluation (MELAS 3243, 3271, 3252, 3256, 3291, 13513)	B	20 mL	L
Family Testing				
☐	**185 Familial DNA Sequence Evaluation** This test detects previously identified sequence variants in at-risk family members. Proband Accession # _____ Relationship _____	B	10 mL	L
Hereditary Renal Tubular Disorders				
☐	**767 Hereditary Renal Tubular Disorders Evaluation** (SLC12A1, KCNJ1, CLCNKB, BSND, SLC12A3)	B	10 mL	L
☐	762 SLC12A1 DNA Sequencing Test (Bartter type 1)	B	10 mL	L
☐	763 KCNJ1 DNA Sequencing Test (Bartter type 2)	B	10 mL	L
☐	764 CLCNKB DNA Sequencing Test (Bartter type 3)	B	10 mL	L
☐	765 BSND DNA Sequencing Test (Bartter type 4)	B	10 mL	L
☐	766 SLC12A3 DNA Sequencing Test (Gitelman)	B	10 mL	L
☐	825 Autosomal Dominant Hypocalcemia (CASR) Evaluation	B	10 mL	L
Monogenic Hypertension				
☐	**749 Monogenic Hypertension Evaluation** (SCNN1B, SCNN1G, CYP11B1, HSD11B2)	B	10 mL	L
☐	**747 Liddle's Syndrome Evaluation** (SCNN1B, SCNN1G)	B	10 mL	L
☐	**748 Pseudohypoaldosteronism Type 1 Evaluation** (SCNN1A, SCNN1B, SCNN1G)	B	10 mL	L
☐	772 SCNN1A DNA Sequencing Test	B	10 mL	L
☐	745 SCNN1B DNA Sequencing Test	B	10 mL	L
☐	746 SCNN1G DNA Sequencing Test	B	10 mL	L
☐	774 CYP11B1 DNA Sequencing. Test	B	10 mL	L
☐	775 HSD11B2 DNA Sequencing Test	B	10 mL	L
☐	779 CYP11B1/CYP11B2 Chimeric Gene Fusion Test	B	10 mL	L
Nephrogenic Diabetes Insipidus				
☐	**854 Nephrogenic Diabetes Insipidus Evaluation** (AVPR2, AQP2)	B	10 mL	L
☐	851 AVPR2 DNA Sequencing Test	B	10 mL	L
☐	852 AQP2 DNA Sequencing Test	B	10 mL	L
Nephronophthisis				
☐	750 NPH1 (Familial Juvenile Nephronophthisis (FJN)) Molecular Test	B	10 mL	L

Test Code		Pref. Spec.	Pref. Vol.	Tube Type
Nephrotic Syndrome				
☐	**722 Early Onset Nephrotic Syndrome Evaluation** (PLCE1, LAMB2, WT1, NPHS1, NPHS2)	B	10 mL	L
☐	**717 Inherited Focal and Segmental Glomerulosclerosis (FSGS) Evaluation** (INF2, ACTN4, TRPC6, NPHS2)	B	10 mL	L
☐	711 ACTN4 DNA Sequencing Test	B	10 mL	L
☐	712 TRPC6 DNA Sequencing Test	B	10 mL	L
☐	716 INF2 DNA Sequencing Test	B	10 mL	L
☐	718 PLCE1 DNA Sequencing Test	B	10 mL	L
☐	713 WT1 DNA Sequencing Test	B	10 mL	L
☐	714 LAMB2 DNA Sequencing Test	B	10 mL	L
☐	710 NPHS2 DNA Sequencing Test (Steroid Resistant Nephrotic Syndrome; Podocin)	B	10 mL	L
☐	730 NPHS1 DNA Sequencing Test (Congenital Nephrotic Syndrome; Nephrin)	B	10 mL	L
Polycystic Kidney Disease				
☐	**761 Complete PKD Evaluation** Step 1. PKD1/PKD2 Sequencing; Step 2. PKD1/PKD2 MLPA			
☐	725 PKDx® DNA Sequencing Test (PKD1 and PKD2 Sequencing)	B	10 mL	L
☐	728 PKDx® Familial Mutation Evaluation (PKD1 and PKD2 Single Exon Sequencing) Proband Accession # _____ Relationship _____	B	10 mL	L
☐	760 PKD Deletion Test (PKD1/PKD2 MLPA)	B	10 mL	L
Other Cystic Diseases				
☐	**556 Complete Tuberous Sclerosis Evaluation** (TSC1 Sequencing, TSC1 Deletion, TSC2 Sequencing, TSC2 Deletion)	B	20 mL	L
☐	521 TSC1 DNA Sequencing Test	B	20 mL	L
☐	508 TSC1 DNA Deletion Test	B	20 mL	L
☐	522 TSC2 DNA Sequencing Test	B	20 mL	L
☐	523 TSC Familial Mutation Evaluation (TSC1 and TSC2 Single Exon Sequencing) Proband Accession # _____ Relationship _____	B	10 mL	L
☐	524 TSC2 DNA Deletion Test	B	10 mL	L
☐	770 Hereditary Interstitial Kidney Disease (2 exon UMOD seq.)	B	10 mL	L
☐	836 TCF2 DNA Sequencing Test (Renal Cysts and Diabetes Syndrome (RCAD))	B	10 mL	L
Renal Cancer				
☐	**889 Pheochromocytoma Evaluation** (RET, VHL, SDHB)	B	10 mL	L
☐	813 MEN2 (RET) Evaluation	B	10 mL	L
☐	818 MEN1 (MEN1) Evaluation	B	10 mL	L
☐	888 SDHB DNA Sequencing Test	B	10 mL	L
☐	858 von Hippel-Lindau Syndrome (VHL) Evaluation	B	10 mL	L
Renal Cysts and Diabetes				
☐	776 HNF1ß DNA Sequencing and Deletion Evaluation (RCAD)	B	10 mL	L
Rickets				
☐	**857 Hypophosphatemic Rickets Evaluation** (PHEX, FGF23)	B	10 mL	L
☐	855 PHEX DNA Seq. Test (X-linked Hypophosphatemic Rickets)	B	10 mL	L
☐	856 FGF23 DNA Sequencing Test (Autosomal Dominant Hypophosphatemic Rickets)	B	10 mL	L

Specimen Requirements & Shipping Information

Specimen Type: B – Blood

Tube Type: L – Lavender C – Cryovial

Pediatric Minimum Volume: 2 mL (for blood tests)

Stability: Hemolysis may compromise DNA recovery and integrity after 48 hrs. It is recommended to ship samples immediately after draw. Samples can be stored for short periods only. Send specimen overnight at room temperature.

Shipping: Send specimen overnight at room temperature. If you have any questions on sample requirements or shipping, contact our client service department at 800-394-4493, extension 2.

NOTE: Specimen tube(s) must be labeled with two of the following forms of identification: name, date of birth, patient ID no. These same two forms of ID must also be indicated on the test requisition.

Athena Diagnostics, Inc., 200 Forest Street, 2nd Floor, Marlborough, MA 01752 • 800-394-4493 • Fax 774-843-3721 • AthenaDiagnostics.com

Athena Diagnostics Client Service Representatives are available from 8:30am to 6:30pm Eastern Time (U.S.).

Customers in the U.S. and Canada please call toll-free

800-394-4493

(Non-U.S. customers please call 508-756-2886 or fax 774-843-3721)

200 Forest Street, 2nd Floor
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