Demande de test en neurologie d'Athena Diagnostics (Mai 2019)

Les champs en rouge indiquent les renseignements requis

athena diagnostics

Avec qui devrions-nous communiquer pour des questions relatives à cette commande ?

Nom _____ (Prénom / Nom de famille)

Téléphone _____ Télécopieur _____

Courriel _____

Identification du patient

Nom du patient _____

Identifiant (#) du patient, si disponible _____ Sexe : ☐ Homme ☐ Femme ☐ Inconnu

DDN _____

Âge _____

Adresse postale _____

Ville _____

Province _____ Code postal _____

Téléphone _____ ☐ Jour ☐ Soir ☐ Cellulaire

Autorisation du patient à utiliser, à transmettre et à conserver des renseignements personnels relatifs à sa santé à l'extérieur du Canada. Vos renseignements personnels relatifs à la santé seront recueillis et utilisés par Athena Diagnostics aux fins de tests de diagnostic et d'analyse. Vos renseignements personnels relatifs à la santé, y compris votre spécimen sanguin, seront transférés à Athena Diagnostics et traités par cette dernière dans son laboratoire sécurisé à Marlborough, au Massachusetts. Athena Diagnostics assurera la confidentialité de vos renseignements et ne divulguera pas vos renseignements personnels, sauf lorsqu'exigé autrement par les lois en vigueur, ce qui peut comprendre un accès légal par les tribunaux, les autorités gouvernementales ou les organismes d'application de la loi aux États-Unis. **Je consens aux dispositions précédentes :** ☐ **OUI** ☐ **NON**

_____
Signature du patient, du parent ou du représentant légalement autorisé — Date

_____
Nom en caractères d'imprimerie du patient, du parent ou du représentant légalement autorisé — Date

_____
Relation avec le patient si le signataire est une personne autre que le patient

Alternative : Attestation du médecin de l'autorisation du patient. Je déclare avoir obtenu le consentement écrit du patient à utiliser, à transmettre et à conserver les renseignements personnels relatifs à la santé du patient à l'extérieur du Canada, dans une large mesure tel que décrit dans l'autorisation du patient ci-dessus.

_____
Signature du médecin praticien — Date

_____
Nom en caractères d'imprimerie du médecin praticien

Renseignements sur la facturation

Veuillez indiquer la partie responsable (en cocher une seule) :

☐ **Ministère de la Santé** *(approbation requise avant le début des tests)*

☐ Approbation préalable incluse ☐ Approbation préalable en suspens

☐ **Approbation préalable en suspens**

Numéro de compte Athena (si attribué) _____

Numéro de commande (si disponible) _____

Coordonnées de facturation _____

Courriel _____

Téléphone _____ Télécopieur _____

Nom de l'hôpital/du laboratoire Adress _____

Ville _____

Province _____

Renseignements sur le payeur _____ Code postal _____

☐ autonome :

Nom _____ (Prénom / Nom de famille)

Numéro de carte de crédit _____

Date d'expiration de la carte de crédit _____ Code de sécurité _____

Téléphone _____

Adresse _____

Ville _____

Province _____ Code postal _____

Tests commandés

Important: inscrivez le code du test et le nom du test.

Code _____ Nom _____

Code _____ Nom _____

Destinataire (s) autorisé (s) du rapport de résultat Renseignements requis sur le médecin

Nom _____ (Prénom / Nom de famille)

Adresse _____

Ville _____

Province _____ Code postal _____

Téléphone _____ Télécopieur _____

Courriel _____

Indications pour tests (en cocher une)

☐ Diagnostique (symptomatique) ☐ Prénatal ☐ Test familial

☐ Prédictif (asymptomatique) ☐ Porteur

Attestation d'obtention de consentement éclairé par le médecin

Conformément à l'article 70G du chapitre 111 de la *Loi générale du Massachusetts*, et à l'article 79 1 de la *Loi de New York sur les droits civils*, la vérification du consentement éclairé du patient est requise pour les tests génétiques. De plus, les laboratoires de tests situés au Massachusetts exigent une reconnaissance signée du médecin praticien demandeur. La reconnaissance signée est requise pour effectuer les tests génétiques demandés. L'entreprise offre une Attestation-cadre de l'obtention du consentement éclairé par le médecin qui peut être signée pour toute commande future.

J'atteste que j'ai obtenu le consentement tant oral qu'écrit au moyen du **Formulaire de consentement éclairé du patient aux tests génétiques** fourni par Athena Diagnostics ou autre. Ce consentement écrit a été signé par la personne qui fera l'objet du test (ou, si cette personne n'est pas en mesure de consentir, signé par la personne autorisée à consentir en son nom).

_____
Signature du médecin praticien — Date

_____
Nom en caractères d'imprimerie du médecin praticien

Le Formulaire de consentement éclairé du patient aux tests génétiques est disponible à l'adresse AthenaDiagnostics.com/consent.

Laboratoire de prélèvement de spécimens

Nom du laboratoire _____

Adresse _____

Ville _____

Province _____ Code postal _____

Téléphone _____ Télécopieur _____

Type de spécimen **Date de prélèvement** _____

☐ Sang entier ☐ Sérum ☐ LCR

☐ Muscle ☐ PVC : direct ☐ PVC : de culture

☐ ADN*

*L'ADN doit être prélevé dans un laboratoire certifié CLIA ou dans un laboratoire répondant à des exigences équivalentes, tel que déterminé par le Cap et/ou le CMS.

CLIA# _____

NOTE : Les tubes de spécimen doivent être étiquetés et porter deux des identifications suivantes : nom, date de naissance, identifiant du patient. Ces mêmes deux formes d'identification doivent également être indiquées sur la demande de test.

Les tests réflexes seront effectués à titre onéreux

Services de test en neurologie d'Athena Diagnostics (Mai 2019)

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

Test Code		Pref. Spec.	Pref. Vol.	Tube Type
Cerebrovascular Disease (Stroke)				
☐ 1175	Notch3(CADASIL) Sequencing Test	B	8 mL	L
☐ 1149	HTRA1 (CARASIL) Sequencing Test	B	8 mL	L
☐ 1120	COL4A1 Sequencing Test (CSVD)	B	8 mL	L
☐ **1122**	**Complete CCM Sequencing and CNV Evaluation** (KRIT1 Seq./Del., CCM2 Seq./Del., PDCD10 Seq./Del.)	B	8 mL	L
☐ **1152**	**KRIT1 (CCM1) Seq. and CNV Evaluation**	B	8 mL	L
☐ **1106**	**CCM2 Seq. and CNV Evaluation**	B	8 mL	L
☐ **1179**	**PDCD10 (CCM3) Seq. and CNV Evaluation**	B	8 mL	L
Dementia				
☐ **178**	**ADmark® Alzheimer's Evaluation**	C	2 mL	P
	(ApoE, Phospho-Tau, Total-Tau, Aß42) (Symptomatic for Dementia) (CSF must be in polypropylene tube and arrive on cold pack or frozen)	B	8 mL	L
☐ 109	ADmark® ApoE Genotype Analysis & Interpretation (Symptomatic for Dementia)	B	8 mL	L
☐ 177	ADmark® Phospho-Tau/Total-Tau/Aß42 CSF Analysis & Interpretation (Symptomatic) (CSF must be in polypropylene tube and arrive on cold pack or frozen)	C	2 mL	P
☐ **179**	**ADmark® Early-Onset Alzheimer's Evaluation** (PSEN1, APP Seq./Dup., PSEN2)	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 (FTD) DNA Test	B	8 mL	L
☐ 204	GRN DNA Sequencing Test	B	8 mL	L
☐ 205	MAPT DNA Sequencing Test	B	8 mL	L
☐ **1711**	**Autoimmune Rapidly Progressive Dementia Evaluation with Recombx®** (Hu, MaTa, CV2, Amphiphysin, GAD65, NMDA, VGKC, LGI1, CASPR2)	S	2 mL	R
☐ 1714	Recombx® Hu Autoantibody Test (Autoimmune Rapidly Progressive Dementia)	S	2 mL	R
☐ 1716	Recombx® MaTa Autoantibody Test (Autoimmune Rapidly Progressive Dementia)	S	2 mL	R
☐ 1717	Recombx® CV2 Autoantibody Test (Autoimmune Rapidly Progressive Dementia)	S	2 mL	R
☐ 1718	Recombx® Amphiphysin Autoantibody 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				
☐ **1186**	**Primary Microcephaly Sequencing Evaluation** (ASPM, MCPH1, WDR62)	B	8 mL	L
☐ 1092	ASPM Sequencing Test	B	8 mL	L
☐ 1153	MCPH1 Sequencing Test	B	8 mL	L
☐ 1257	WDR62 Sequencing Test	B	8 mL	L
☐ 1193	SHANK3 Sequencing Test	B	8 mL	L
☐ 1192	SHANK2 Sequencing Test	B	8 mL	L
☐ 1190	PTEN 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
☐ 1256	VPS13B (COH1) Sequencing Test	B	8 mL	L
☐ **1155**	**MECP2 Sequencing and CNV Evaluation**	B	8 mL	L
☐ 148	Rett Syndrome (MECP2) Dup./Del. Test	B	8 mL	L
☐ **1038**	**ARX Seq. and CNV Evaluation (Intellectual Disability)**	B	8 mL	L
☐ **1114**	**CDKL5 Seq. and CNV Evaluation (Atypical Rett)**	B	8 mL	L
☐ 1194	SYNGAP1 Sequencing Test	B	8 mL	L
☐ **1166**	**MEF2C Sequencing and CNV Evaluation**	B	4 mL	L
☐ **1142**	**FOXG1 Sequencing and CNV Evaluation**	B	4 mL	L
NOTE: Pediatric minimum for all Developmental Disabilities tests is 2 mL.				
Epilepsy				
☐ **6000**	**Epilepsy Advanced Sequencing and CNV Evaluation**	B	8 mL	L
☐ **6008**	**Epilepsy Advanced Sequencing and CNV Evaluation - Generalized, Absence, Focal, Febrile and Myoclonic Epilepsies**	B	8 mL	L
☐ **6010**	**Epilepsy Advanced Sequencing and CNV Evaluation - Epileptic Encephalopathy**	B	8 mL	L
☐ **6018**	**Epilepsy Advanced Sequencing and CNV Evaluation - Developmental Brain Malformations**	B	8 mL	L
☐ **6019**	**Epilepsy Advance Sequencing and CNV Evaluation - Intellectual Disability**	B	8 mL	L
☐ **6022**	**Epilepsy Advanced Sequencing and CNV Evaluation - Neuronal Ceroid Lipofuscinosis**	B	8 mL	L
☐ **6023**	**Epilepsy Advanced Sequencing and CNV Evaluation - Epilepsy with Migraine**	B	8 mL	L
☐ **6033**	**Epilepsy Advanced Sequencing and CNV Evaluation - Syndromic Disorders**	B	8 mL	L
☐ **6038**	**Epilepsy Advanced Sequencing and CNV Evaluation - Infantile Spasms**	B	8 mL	L
Please see website for the list of genes in each panel.				
☐ **5120**	**Autoimmune Epilepsy Evaluation** GAD65, VGKC, CASPR2, LGI1, NMDA	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 Autoantibody Test (Epilepsy) (Single)	S	2 mL	R
☐ **1131**	**Complete Tuberous Sclerosis Seq. and CNV Evaluation** (TSC1 Seq., TSC1 Del., TSC2 Seq., TSC2 Del.)	B	8 mL	L
☐ 1245	TSC1 Sequencing Test	B	8 mL	L
☐ 1236	TSC1 CNV Test	B	8 mL	L
☐ 508	TSC1 Deletion Analysis	B	8 mL	L
☐ 1255	TSC2 Sequencing Test	B	8 mL	L
☐ 1254	TSC2 CNV Test	B	8 mL	L
☐ 524	TSC2 DNA Deletion Test	B	8 mL	L
☐ 523	TSC Familial DNA Seq. Mutation Evaluation	B	8 mL	L
	Proband Accession # _____			
	Relationship _____			
☐ **1129**	**SCN1A Seq. and CNV Evaluation**	B	8 mL	L
☐ 1191	SCN1A CNV Test	B	8 mL	L
☐ 537	SCN1A Deletion Test	B	8 mL	L
☐ **1133**	**CSTB (EPM1) Seq. and Repeat Expansion Evaluation**	B	8 mL	L
☐ 410	EPM1 DNA Test	B	8 mL	L
☐ **1036**	**ARX Seq. and CNV Evaluation (Epilepsy)**	B	8 mL	L
☐ **1115**	**CDKL5 Seq. and CNV Evaluation (Epilepsy)**	B	8 mL	L
☐ 4411	SLC2A1 DNA Sequencing Test	B	8 mL	L
☐ 1003	GFAP (Alexander Disease) 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
Genetic: Anti-Drug Antibody				
☐ 1181	AAV9 Antibody Test	S	2mL	R
Hearing Loss				
☐ **3029**	**Hearing Loss Advanced Seq. and CNV Evaluation** Please see website for the complete list of genes. This test is currently not available for New York patient testing.	B	8mL	L
☐ **329**	**Connexin Related Deafness Evaluation** (Connexin 26, Connexin 30)	B	8 mL	L
☐ 321	Connexin 26 (GJB2) DNA Sequencing Test	B	8 mL	L
☐ 319	Connexin 30 (GJB6) DNA Test	B	8 mL	L
Leukodystrophy				
☐ 1175	Notch3(CADASIL) Sequencing Test	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
☐ **1183**	**PLP1 Sequencing and CNV Evaluation**	B	8 mL	L
☐ 6109	GJC2 DNA Sequencing Test	B	8 mL	L
Migraine				
☐ **1148**	**Hemiplegic Migraine Seq. Evaluation** (CACNA1A, ATP1A2, SCN1A)	B	8 mL	L
☐ 1103	CACNA1A Sequencing Test	B	8 mL	L
☐ 1101	ATP1A2 Sequencing Test	B	8 mL	L
☐ 1136	SCN1A Sequencing Test (FHM) (Exons 3, 23, 26)	B	8 mL	L
Mitochondrial Disorders				
☐ **575**	**Common Mitochondrial Disorders Evaluation** (POLG, MELAS, MERRF, NARP)	B	8 mL	L
☐ **576**	**Progressive External Ophthalmoplegia 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

Note: Test requisitions become outdated. For the most accurate and up-to-date test offering, please visit AthenaDiagnostics.com.

athena diagnostics

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 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	8 mL	L
☐ 620	SOD1 DNA Sequencing Test	B	8 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** (SPAST)	B	8 mL	L
Individual HSP DNA Tests: ☐ 531 Atlastin (SPG3A) ☐ 632 Paraplegin (SPG7) ☐ 633 Spatacsin (SPG11) ☐ 614 ZFYVE26 (SPG15)		B	8 mL	L
☐ **214**	**SMA Plus** (Reflexive) Testing is performed in this order: 1. SMN1 Del./SMN2 Del.; 2. SMN1 Seq.	B	4 mL	L
☐ 111	SMA Diagnostic Test (including SMN2 Copy Number)	B	4 mL	L
☐ 211	SMN DNA Sequencing Test (only order if deletion testing has already been performed)	B	4 mL	L
☐ 444	SMA Carrier Screen SMN1 Del./SMN2 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)	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 (ATXN1) ☐ 672 SCA2 (ATXN2) ☐ 105 SCA3 (ATXN3) ☐ 373 SCA6 (CACNA1A) ☐ 677 SCA7 (ATXN7) ☐ 384 SCA8 (ATXN8OS) ☐ 387 SCA10 (ATXN10) ☐ 285 SCA12 (PPP2R2B) ☐ 388 SCA17 (TBP) ☐ 401 DRPLA ☐ 383 POLG1 (MIRAS) ☐ 283 TTPA (AVED) ☐ 348 FRDA/FXN Seq. ☐ 119 FRDA/FXN Expansion		B	8 mL	L
☐ **402**	**Chorea Differential Evaluation** (DRPLA, Huntington's Disease)	B	8 mL	L
☐ 116	Huntington Disease Repeat Expansion Test	B	8 mL	L
☐ **639**	**Isolated Dystonia Evaluation** (DYT1, THAP1)	B	8 mL	L
☐ 626	Dystonia (DYT1) DNA Test	B	8 mL	L
☐ 618	THAP1 DNA Sequencing Test (DYT6)	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 (DYT5A)	B	8 mL	L
☐ 638	GCH1 Deletion Test (DYT5A)	B	8 mL	L
☐ 634	TH DNA Sequencing Test (DYT5B)	B	8 mL	L
☐ 624	SGCE DNA Sequencing Test (DYT11)	B	8 mL	L
☐ 627	SGCE Deletion Test (DYT11)	B	8 mL	L
☐ 617	PNKD (MR-1) 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
☐ 1187	PRRT2 (Dyskinesia/IC) Seq. Test	B	8 mL	L
Multiple Sclerosis				
☐ **1284**	**NMO Spectrum Evaluation (AQP4, ELISA reflex to MOG, CBA)**	S	2 ml	R
☐ **1287**	**NMO Spectrum Evaluation (AQP4, CBA reflex to MOG, CBA)**	S	2 ml	R
☐ 1523	Myelin Oligodendrocyte Glycoprotein (MOG) Antibody, CBA with Reflex to Titer	S	2 ml	R
☐ 1282	Aquaporin-4 (AQP4) (NMO IgG) Antibody, CBA with Reflex to Titer	S	2 ml	R
☐ 193	Aquaporin-4 (AQP4) Antibody (NMO-IgG), ELISA	S	2 mL	R
☐ 112	NAbFeron® (INFB-1) Neutralizing Antibody Test	S	2 mL	R
☐ 197	TYSABRI® (Natalizumab) Antibody Test (See website for collection notes)	S	2 mL	R
Myasthenia Gravis				
☐ 482	MuSK Antibody Test	S	2 mL	R
☐ 1480	Titin Autoantibody Test	S	2 mL	R
☐ 1481	RyR Autoantibody Test	S	2 mL	R
☐ 1483	LRP4 Autoantibody Test	S	2 mL	R
☐ **1490**	**AChR-Seronegative Myasthenia Gravis Evaluation**	S	2 mL	R
☐ 1510	Acetylcholine Receptor Binding Antibody with Reflex to Musk Antibody	S	2mL	R
☐ 1511	Acetylcholine Receptor Binding Antibody with Reflex to MuSK/LRP4 Antibodies	S	2 mL	R
☐ 1513	Acetylcholine Receptor Binding Antibody	S	2 mL	R
☐ **1514**	**Myasthenia Gravis Panel 2**	S	2 mL	R
☐ 1516	Acetylcholine Receptor Blocking Antibody	S	1 mL	R
☐ 1517	Acetylcholine Receptor Modulating Antibody	S	1mL	R
☐ 1521	Myasthenia Gravis Panel 2 with Reflex to MuSK Antibody	S	3mL	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
☐ **5511**	**Congenital Myasthenic Syndrome 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
☐ **5530**	**DMD Evaluation**	B	8 mL	L
NOTE: Please see website for the list of genes in each panel.				
☐ 5531	DMD Duplication/Deletion	B	8 mL	L
☐ 183	DMD DNA Sequencing Test	B	8 mL	L
☐ 100	Dystrophin ProteinTest	M	10 mg	C
☐ **207**	**Early-Onset Myotonia Evaluation** (DM1, CLCN1, SCN4A)	B	8 mL	L
☐ 108	DMPK DNA Test (DM1)	B	8 mL	L
☐ 110	CNBP DNA Test (DM2) (DM2 testing is not recommended for patients with early onset myotonic dystrophy)			
☐ 128	CLCN1 DNA Sequencing Test			
☐ 146	SCN4A (Myotonia) DNA Sequencing Test			
☐ **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 SGCA Duplication/Deletion Test ☐ 583 SGCG Duplication/Deletion Test ☐ 584 CAPN3 Duplication/Deletion Test		B	8 mL	L
☐ 561	Dysferlin Protein Blood Test Sample must be received within 48 hours of collection Sample must arrive on cold pack Ship sample M-Th only	B	10 mL	L
☐ 571	Dysferlin Sequencing Test	B	8 mL	L
☐ 405	FSHD1 Southern Blot Test Sample must be received within 72 hours of collection Ship sample M-Th only	B	15 mL	L
☐ 300	OPMD Repeat Expansion 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.

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

Services de test en neurologie d'Athena Diagnostics (Mai 2019)

Test Code		Pref. Spec.	Pref. Vol.	Tube Type
Paraneoplastic & Other Antibody Disorders of the CNS				
☐ **4711**	**Paraneoplastic Neurological Syndromes Evaluation with Recombx®, Initial Assessment** (Hu, Yo, CV2, MaTa, Ri, Amphiphysin)	S or C	2 mL or 2 mL	R P
☐ **4620**	**NeoComplete Paraneoplastic Evaluation with Recombx®** Hu, Yo, Zic4, CV2, MaTa, Ri, CAR, VGCC, VGKC, Amphiphysin, gnAChR, NMDA, GAD65, LGI1, CASPR2.	S	2 mL	R
☐ **4640**	**Paraneoplastic Autoantibody Evaluation with Recombx®, CSF** Hu, Yo, Zic4, CV2, MaTa, Ri, CAR, Amphiphysin, NMDA, LGI1, CASPR2.	C	2ml	P
☐ **4724**	**NeoCerebellar Degeneration Paraneoplastic Profile with Recombx®** (Hu, Yo, Zic4, CV2, MaTa, Ri, Amphiphysin, GAD65 Neurological Syndrome)	S	2 mL	R
☐ **4722**	**NeoEncephalitis Paraneoplastic Evaluation with Recombx®** (Hu, CV2, MaTa, VGKC, Amphiphysin, GAD65, LGI1, NMDA, CASPR2)	S	2 mL	R
☐ **4725**	**NeoSensory Neuropathy Paraneoplastic Profile with Recombx®** (Hu, CV2, Amphiphysin)	S	2 mL	R
☐ **4727**	**Neuromyotonia Evaluation** (CASPR2, VGKC)	S	2 mL	R
Individual Recombx® Autoantibody Tests:		S	2 mL	R
☐ 4684 CAR ☐ 4681 CV2 ☐ 4682 Hu				
☐ 4683 MaTa ☐ 4685 Ri ☐ 4686 Yo ☐ 4689 Zic4				
☐ 449	LGI1 Antibody Test	S	2 mL	R
☐ 499	CASPR2 Antibody Test	S	2 mL	R
☐ 419	NMDA Receptor Autoantibody Test	S	2 mL	R
☐ 422	GAD65 Neurological Syndrome Antibody Test	S	2 mL	R
☐ 475	VGCC Type P/Q Autoantibody Test (LEMS)	S	2 mL	R
☐ 485	VGKC Antibody Test	S	2 mL	R
☐ 4674	Recombx® Amphiphysin Autoantibody Test	S	2 mL	R
☐ 428	Ganglionic AChR Antibody Test	S	2 mL	R
Peripheral Neuropathy: Autoimmune				
☐ **3100**	**SensoriMotor Neuropathy Profile with Recombx® - Complete** (Co-GM1 Quattro®, MAG 'Dual Antigen'®, Hu, GALOP™, Sulfatide)	S	2 mL	R
☐ **3148**	**Sensory Neuropathy Profile with Recombx®** (MAG 'Dual Antigen'®, Hu, GALOP™, Sulfatide)	S	2 mL	R
☐ **3163**	**Motor Neuropathy Profile - Complete** (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
☐ 3155	Co-GM1 Quattro® Autoantibody Test (Co-Asialo, GD1a, Co-GD1b and Co-GM1)	S	2 mL	R
☐ 3127	MAG 'Dual Antigen'® Autoantibody Test	S	2 mL	R
☐ 261	GALOP™ Autoantibody Test	S	2 mL	R
☐ 210	Sulfatide Autoantibody Test	S	2 mL	R
☐ 160	GQ1b Autoantibody Test	S	2 mL	R
☐ 278	GD1a Autoantibody Test	S	2 mL	R
☐ 272	Co-Asialo Autoantibody Test	S	2 mL	R
☐ 273	Co-GD1b Autoantibody Test	S	2 mL	R
☐ 271	Co-GM1 Autoantibody Test	S	2 mL	R
Peripheral Neuropathy: Hereditary				
☐ **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
☐ **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
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 PRX (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	TTR DNA Sequencing Test	B	8 mL	L
Peripheral Neuropathy: Hereditary Sensory Autonomic Neuropathy				
☐ 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

Type d'étude:
CSF M - Muscle Tissue
B - Blood
S - Serum

Tube Type
P - Polypropylène CSF Transfert Tube
R - Red
L - Lavender
C - Cryovial
** CSF doit être collecté dans un tube ne contenant pas d'additifs.

Services de test en nephrologie d'Athena Diagnostics (Mai 2019)

Test Code		Pref. Spec.	Pref. Vol.	Tube Type
Alport Syndrome				
☐ **759**	**Complete Alport Syndrome Evaluation** (COL4A3,4,5 DNA Sequencing; COL4A5 Deletion Test)	B	8 mL	L
☐ 755	COL4A5 Sequencing and Deletion Analysis	B	8 mL	L
☐ 756	COL4A5 Deletion Analysis	B	8 mL	L
☐ 757	COL4A3 DNA Sequencing Test	B	8 mL	L
☐ 758	COL4A4 DNA Sequencing Test	B	8 mL	L
Amyloidosis				
☐ 235	TTR DNA Sequencing Test	B	8 mL	L
Bardet-Biedl Syndrome				
☐ **887**	**Bardet-Biedl Syndrome Evaluation** (BBS1, BBS2, BBS10)	B	8 mL	L
☐ 871	BBS1 (BBS) DNA Sequencing Test	B	8 mL	L
☐ 872	BBS2 (BBS) DNA Sequencing Test	B	8 mL	L
☐ 886	BBS10 (BBS) DNA Sequencing Test	B	8 mL	L
Fanconi Syndrome				
☐ 517	MELAS mtDNA Evaluation (MELAS 3243, 3271, 3252, 3256, 3291, 13513)	B	8 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	8 mL	L
Hereditary Renal Tubular Disorders				
☐ **767**	**Hereditary Renal Tubular Disorders Evaluation** (SLC12A1, KCNJ1, CLCNKB, BSND, SLC12A3)	B	8 mL	L
☐ 762	SLC12A1 DNA Sequencing Test	B	8 mL	L
☐ 763	KCNJ1 DNA Sequencing Test	B	8 mL	L
☐ 764	CLCNKB DNA Sequencing Test	B	8 mL	L
☐ 765	BSND DNA Sequencing Test	B	8 mL	L
☐ 766	SLC12A3 DNA Sequencing Test	B	8 mL	L
☐ 825	CASR DNA Sequencing Test	B	8 mL	L
Monogenic Hypertension				
☐ **749**	**Monogenic Hypertension Evaluation** (SCNN1B, SCNN1G, CYP11B1, HSD11B2)	B	8 mL	L
☐ 747	Liddle's Syndrome Evaluation (SCNN1B, SCNN1G)	B	8 mL	L
☐ 748	Pseudohypoaldosteronism Type 1 Evaluation (SCNN1A, SCNN1B, SCNN1G)	B	8 mL	L
☐ 772	SCNN1A DNA Sequencing Test	B	8 mL	L
☐ 745	SCNN1B DNA Sequencing Test	B	8 mL	L
☐ 746	SCNN1G DNA Sequencing Test	B	8 mL	L
☐ 774	CYP11B1 DNA Sequencing. Test	B	8 mL	L
☐ 775	HSD11B2 DNA Sequencing Test	B	8 mL	L
☐ 779	CYP11B1/CYP11B2 Chimeric Gene Fusion Test	B	8 mL	L
Nephrogenic Diabetes Insipidus				
☐ **854**	**Nephrogenic Diabetes Insipidus Evaluation** (AVPR2, AQP2)	B	8 mL	L
☐ 851	AVPR2 DNA Sequencing Test	B	8 mL	L
☐ 852	AQP2 DNA Sequencing Test	B	8 mL	L
Nephronophthisis				
☐ 750	NPHP1 Deletion Test (Familial Juvenile Nephronophthisis)	B	8 mL	L

Test Code		Pref. Spec.	Pref. Vol.	Tube Type
Nephrotic Syndrome				
☐ **722**	**Early Onset Nephrotic Syndrome Evaluation** (PLCE1, LAMB2, WT1, NPHS1, NPHS2)	B	8 mL	L
☐ **717**	**Focal and Segmental Glomerulosclerosis (FSGS) Evaluation** (INF2, ACTN4, TRPC6, NPHS2)	B	8 mL	L
☐ 711	ACTN4 DNA Sequencing Test	B	8 mL	L
☐ 712	TRPC6 DNA Sequencing Test	B	8 mL	L
☐ 716	INF2 DNA Sequencing Test	B	8 mL	L
☐ 718	PLCE1 DNA Sequencing Test	B	8 mL	L
☐ 713	WT1 DNA Sequencing Test	B	8 mL	L
☐ 714	LAMB2 DNA Sequencing Test	B	8 mL	L
☐ 710	NPHS2 DNA Sequencing Test	B	8 mL	L
☐ 730	NPHS1 DNA Sequencing Test	B	8 mL	L
Polycystic Kidney Disease				
☐ 728	PKDx® Familial Mutation Evaluation (PKD1 and PKD2 Single Exon Sequencing) Proband Accession # _____ Relationship _____	B	8 mL	L
☐ 8100	Complete PKDx Evaluation	B	8 mL	L
☐ 8105	PKD1 Deletion Test	B	8 mL	L
☐ 8101	PKD1 DNA Sequencing and Deletion Evaluation	B	8 mL	L
☐ 8103	PKD1 DNA Sequencing Test	B	8 mL	L
☐ 8106	PKD2 Deletion Test	B	8 mL	L
☐ 8102	PKD2 DNA Sequencing and Deletion Evaluation	B	8 mL	L
☐ 8104	PKD2 DNA Sequencing Test	B	8 mL	L
Other Cystic Diseases				
☐ **556**	**Complete Tuberous Sclerosis Evaluation** (TSC1 Sequencing, TSC1 Deletion, TSC2 Sequencing, TSC2 Deletion)	B	8 mL	L
☐ 521	TSC1 DNA Sequencing Test	B	8 mL	L
☐ 508	TSC1 Deletion Test	B	8 mL	L
☐ 522	TSC2 DNA Sequencing Test	B	8 mL	L
☐ 523	TSC Familial Mutation Evaluation Proband Accession # _____ Relationship _____	B	8 mL	L
☐ 524	TSC2 DNA Deletion Test	B	8 mL	L
☐ 770	Hereditary Interstitial Kidney Disease (UMOD) DNA Sequencing Test	B	8 mL	L
☐ 836	TCF2 DNA Sequencing Test (Renal Cysts and Diabetes Syndrome (RCAD))	B	8 mL	L
Renal Cancer				
☐ **889**	**Pheochromocytoma Evaluation** (RET, VHL, SDHB)	B	8 mL	L
☐ 813	MEN2 (RET) DNA Sequencing Test	B	8 mL	L
☐ 818	MEN1 (MEN1) DNA Sequencing Test	B	8 mL	L
☐ 888	SDHB DNA Sequencing Test	B	8 mL	L
☐ 858	von Hippel-Lindau Syndrome (VHL) DNA Sequencing Test	B	8 mL	L
Renal Cysts and Diabetes				
☐ 776	HNF1ß DNA Sequencing and Deletion Evaluation (RCAD)	B	8 mL	L
Rickets				
☐ **857**	**Hypophosphatemic Rickets Evaluation** (PHEX, FGF23)	B	8 mL	L
☐ 855	PHEX (Hypophosphatemic Rickets) DNA Sequencing Test	B	8 mL	L
☐ 856	FGF23 (Hypophosphatemic Rickets) DNA Sequencing Test	B	8 mL	L

Exigences relatives aux échantillons: 8 ml (6 ml minimum) de sang total recueilli dans un tube EDTA (lavender-top).

REMARQUE: Le (s) tube (s) de l'échantillon doivent être étiquetés avec deux des formes suivantes d'identification: nom, date de naissance, quatre derniers chiffres de SS #, numéro d'identification du patient. Ces deux mêmes formes d'identité doivent également être indiquées sur la demande d'essai.

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

Services de tests d'endocrinologie d'Athena Diagnostics (Mai 2019)

athena diagnostics

Test Code	Test Name	Genes Included
Adrenal Disorders		
☐ **816**	**Primary Adrenal Insufficiency Evaluation**	ABCD1, NR0B1, AIRE
☐ 812	Autoimmune Polyglandular Syndrome (AIRE) Evaluation	
☐ 815	ABCD1 (Adrenoleukodystrophy) DNA Sequencing Test	
☐ 814	NR0B1 (Adrenal Hypoplasia Congenita) DNA Sequencing Test	
☐ **879**	**Congenital Adrenal Hyperplasia (CAH) Evaluation** CYP21A2 sequencing and deletion, CYP11B1 sequencing	
☐ 880	CYP21A2 (CAH) Evaluation	
	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) Evaluation	
Bone Diseases		
☐ **860**	**Osteogenesis Imperfecta Evaluation**	COL1A1, COL1A2
☐ 861	COL1A1 (OI) DNA Sequencing Test	
☐ 862	COL1A2 (OI) DNA Sequencing Test	
☐ 811	LRP5 (OPPG) DNA Sequencing Test	
☐ 821	LRP5 Idiopathic Osteoporosis (IOP) 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 (CHI) DNA Sequencing Test	
☐ 823	GCK (CHI) DNA Sequencing Test	
☐ 826	KCNJ11 (CHI) DNA Sequencing Test	
☐ 827	ABCC8 (CHI) DNA Sequencing Test	
☐ **42**	**CH Parental Testing – To augment child/proband diagnosis**	
	For expedited diagnosis of proband, send parental testing samples as soon as possible and provide information below.	
	☐ Mother ☐ Father	
	Proband Name/Accession #_____	

Test Code	Test Name	Genes Included
Diabetes		
☐ **885**	**Monogenic Diabetes (MODY) Five-Gene Evaluation**	HNF1A, GCK, HNF4A, HNF1B, IPF1
☐ **8800**	**Monogenic Diabetes (MODY) Four-Gene Evaluation**	HNF1A, GCK, HNF4A, HNF1B
☐ **8801**	**Monogenic Diabetes (MODY) Three-Gene Evaluation**	HNF1A, GCK, HNF1B
☐ **8802**	**Monogenic Diabetes (MODY) Two-Gene Evaluation**	HNF1A, GCK
☐ 802	HNF4A (MODY1) DNA Sequencing and Deletion Test	
☐ 803	GCK (MODY2) DNA Sequencing and Deletion Test	
☐ 804	TCF1 (MODY3) DNA Sequencing and Deletion Test	
☐ 834	IPF1 (MODY4) DNA Sequencing Test	
☐ 805	TCF2 (MODY5) DNA Sequencing and Deletion Test	
☐ 837	CEL (MODY8) Mutation Analysis	
☐ **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	AQP2 (Nephrogenic Diabetes Insipidus) DNA Sequencing Test	
Familial Cancer Syndromes		
☐ 818	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		
☐ **185**	**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 _____	
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	
Obesity		
☐ **884**	**Early Onset Obesity Evaluation**	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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Test Code	Test Name	Genes Included
Reproductive Disorders		
☐ 817	Male Precocious Puberty (LHCGR) DNA Sequencing Test	
☐ **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	
☐ **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

Test Code	Test Name	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 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 DNA Sequencing Test	

Exigences relatives aux échantillons: 8 ml (6 ml minimum) de sang total recueilli dans un tube EDTA (lavender-top).

REMARQUE: Le (s) tube (s) de l'échantillon doivent être étiquetés avec deux des formes suivantes d'identification: nom, date de naissance, quatre derniers chiffres de SS #, numéro d'identification du patient. Ces deux mêmes formes d'identité doivent également être indiquées sur la demande d'essai.

Athena Diagnostics, Inc., 200 Forest Street, 2nd Floor, Marlborough, MA 01752 • 1-800-394-4493 • Télécopieur 610-271-6085 • AthenaDiagnostics.com

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