## Laboratory Update





## 3/19/2015 - Special Communication, Quest Diagnostics Nichols Institute, Valencia

ADmark® ApoE Genotype Analysis & Interpretation (Symptomatic) 4/20/2015 2  ADmark® Phospho-Tau/Total-Tau/Ab42 CSF Analysis & Interpretation (Symptomatic) 4/20/2015 2  ADmark® PS-1 DNA Sequencing Test 4/20/2015 2  Complete HNPP Evaluation 4/20/2015 3	TEST CHANGES  Please Note: Not all test codes assigned to each assay are listed in the table of contents.  Please refer to the complete listing on the page numbers indicated.				
S44955   ADmarks Phosphor Tau/Total-Tau/Ab42 CSF Analysis & Interpretation (Symptomatic)   2	Test Code	Former Test Code	Test Name	Effective Date	Page #
Symptomatic    2   2   349327   ADmark® PS-1 DNA Sequencing Test   4/20/2015   2   3   3   3   3   3   3   3   3   3	S4631 <u>5</u>			4/20/2015	2
Complete NNPP Evaluation   4/20/2015   3	S4495 <u>5</u>		, , , , , , , , , , , , , , , , , , , ,	4/20/2015	2
Dystonia (DYT1) DNA Test	S49327		ADmark® PS-1 DNA Sequencing Test	4/20/2015	2
## Familial Amyotrophic Lateral Sclerosis (SOD1) DNA Test	S49335		Complete HNPP Evaluation	4/20/2015	3
Familial Hemiplegic Migraine Type I (CACNA1A) DNA Test   4/20/2015   4	S48185		Dystonia (DYT1) DNA Test	4/20/2015	3
SAMESSO         GALOP™ Autoantibody Test         4/20/2015         4           S44395         LHON mtDNA Evaluation         4/20/2015         5           S51269         MELAS mtDNA Evaluation         4/20/2015         5           S50699         MFN2 DNA Sequencing Test         4/20/2015         5           S51162         Monogenic Diabetes (MODY) Evaluation         4/20/2015         6           S48727         MPZ DNA Sequencing Test         4/20/2015         7           S49558         NeoCorebellar Degeneration Paraneoplastic Evaluation with Recombx™         4/20/2015         7           S49558         NeoComplete Paraneoplastic Evaluation with Recombx™         4/20/2015         7           S11636         NeoComplete Paraneoplastic Evaluation with Recombx™         4/20/2015         7           S11632         NeoEncephalitis Paraneoplastic Profile with Recombx         4/20/2015         8           S12438         Neuromyelitis Optica (NMO) AQP4 Autoantibody Test         4/20/2015         8           S490078         Notch3 (CADASIL) DNA Sequencing Test         4/20/2015         8           S493997         OPMD DNA Test         4/20/2015         9           S44815         Recombx™ CAR (Anti-Recoverin) Autoantibody Test         4/20/2015         9           S44815<	S49489		Familial Amyotrophic Lateral Sclerosis (SOD1) DNA Test	4/20/2015	4
LHON mtDNA Evaluation 4/20/2015 5  351269 MELAS mtDNA Evaluation 4/20/2015 5  351269 MFN2 DNA Sequencing Test 4/20/2015 5  350699 MFN2 DNA Sequencing Test 4/20/2015 5  351162 Monogenic Diabetes (MODY) Evaluation 4/20/2015 6  3549727 MPZ DNA Sequencing Test 4/20/2015 6  3549727 MMSK Antibody Test 4/20/2015 7  3549558 NeoCerebellar Degeneration Paraneoplastic Evaluation with 4/20/2015 7  3649558 NeoCerebellar Degeneration Paraneoplastic Evaluation with 4/20/2015 7  37549558 NeoCerebellar Degeneration Paraneoplastic Evaluation with 4/20/2015 7  376262 NeoEncephalitis Paraneoplastic Profile with Recombx 4/20/2015 8  376263 Neuromyelitis Optica (NMO) AQP4 Autoantibody Test 4/20/2015 8  376278 Notch3 (CADASIL) DNA Sequencing Test 4/20/2015 8  37630078 Notch3 (CADASIL) DNA Sequencing Test 4/20/2015 9  37630078 Notch3 (CADASIL) DNA Sequencing Test 4/20/2015 10  37630078 Notch3 (CADASIL) DNA Sequencing Test 4/20/2015 11  37630078 Notch3 (CADASIL) DNA Sequencing Test 4/20	S52411		Familial Hemiplegic Migraine Type I (CACNA1A) DNA Test	4/20/2015	4
MELAS mtDNA Evaluation 4/20/2015 5  \$552689	S46850		GALOP™ Autoantibody Test	4/20/2015	4
MFN2 DNA Sequencing Test  Monogenic Diabetes (MODY) Evaluation  4/20/2015  6  S81162  Monogenic Diabetes (MODY) Evaluation  4/20/2015  6  S818727  MPZ DNA Sequencing Test  4/20/2015  6  S848727  MuSK Antibody Test  4/20/2015  7  NeoCerebellar Degeneration Paraneoplastic Evaluation with  4/20/2015  7  NeoComplete Paraneoplastic Evaluation with Recombx™  4/20/2015  7  NeoEncephalitis Paraneoplastic Evaluation with Recombx™  4/20/2015  8  NeoFincephalitis Paraneoplastic Profile with Recombx  4/20/2015  8  Neuromyelitis Optica (NMO) AQP4 Autoantibody Test  4/20/2015  8  Notch3 (CADASIL) DNA Sequencing Test  4/20/2015  8  S49097  OPMD DNA Test  4/20/2015  9  S44815  Recombx™ CAR (Anti-Recoverin) Autoantibody Test  4/20/2015  9  S49448  Recombx™ CAR (Anti-Recoverin) Autoantibody Test  4/20/2015  10  S49890  Recombx™ MaTa Autoantibody Test  4/20/2015  10  S50992  SCA1 DNA Test  4/20/2015  11  S50992  SCA2 DNA Test  4/20/2015  12  SCA3 (Machado-Joseph Disease) DNA Test  4/20/2015  12	S44395		LHON mtDNA Evaluation	4/20/2015	5
Monogenic Diabetes (MODY) Evaluation	S51269		MELAS mtDNA Evaluation	4/20/2015	5
MPZ DNA Sequencing Test  MPZ DNA Sequencing Test  MuSK Antibody Test  MuSK Antibody Test  NeoCerebellar Degeneration Paraneoplastic Evaluation with Recombx™  Recombx™  NeoComplete Paraneoplastic Evaluation with Recombx™  4/20/2015  7  NeoEncephalitis Paraneoplastic Evaluation with Recombx	S50699		MFN2 DNA Sequencing Test	4/20/2015	5
MuSK Antibody Test   4/20/2015   7	<u>S51162</u>		Monogenic Diabetes (MODY) Evaluation	4/20/2015	6
NeoCerebellar Degeneration Paraneoplastic Evaluation with   4/20/2015   7	<u> </u>		MPZ DNA Sequencing Test	4/20/2015	6
Recombx™         /           01636         NeoComplete Paraneoplastic Evaluation with Recombx™         4/20/2015         7           01622         NeoEncephalitis Paraneoplastic Profile with Recombx         4/20/2015         8           052438         Neuromyelitis Optica (NMO) AQP4 Autoantibody Test         4/20/2015         8           050078         Notch3 (CADASIL) DNA Sequencing Test         4/20/2015         8           049097         OPMD DNA Test         4/20/2015         9           0543020         PMP22 Duplication/Deletion Test         4/20/2015         9           0544815         Recombx™ CAR (Anti-Recoverin) Autoantibody Test         4/20/2015         9           054948         Recombx™ CV2 Antibody Test         4/20/2015         10           054949         Recombx™ MaTa Autoantibody Test         4/20/2015         10           0551087         Recombx™ Zic4 Antibody Test         4/20/2015         10           050092         SCA1 DNA Test         4/20/2015         11           050091         SCA2 DNA Test         4/20/2015         11           050091         SCA3 (Machado-Joseph Disease) DNA Test         4/20/2015         12           050090         SCA6 DNA Test         4/20/2015         12	S50437		MuSK Antibody Test	4/20/2015	7
NeoEncephalitis Paraneoplastic Profile with Recombx         4/20/2015         8           352438         Neuromyelitis Optica (NMO) AQP4 Autoantibody Test         4/20/2015         8           A50078         Notch3 (CADASIL) DNA Sequencing Test         4/20/2015         8           A50079         OPMD DNA Test         4/20/2015         9           S43020         PMP22 Duplication/Deletion Test         4/20/2015         9           S44815         Recombx™ CAR (Anti-Recoverin) Autoantibody Test         4/20/2015         9           S49448         Recombx™ CV2 Antibody Test         4/20/2015         10           S48890         Recombx™ MaTa Autoantibody Test         4/20/2015         10           S51087         Recombx™ Zic4 Antibody Test         4/20/2015         10           S50092         SCA1 DNA Test         4/20/2015         11           S50091         SCA2 DNA Test         4/20/2015         11           S50094         SCA3 (Machado-Joseph Disease) DNA Test         4/20/2015         12           S60090         SCA6 DNA Test         4/20/2015         12	<u>S49558</u>			4/20/2015	7
Neuromyelitis Optica (NMO) AQP4 Autoantibody Test	91636		NeoComplete Paraneoplastic Evaluation with Recombx <sup>™</sup>	4/20/2015	7
A50078         Notch3 (CADASIL) DNA Sequencing Test         4/20/2015         8           349097         OPMD DNA Test         4/20/2015         9           343020         PMP22 Duplication/Deletion Test         4/20/2015         9           344815         Recombx™ CAR (Anti-Recoverin) Autoantibody Test         4/20/2015         9           349448         Recombx™ CV2 Antibody Test         4/20/2015         10           348890         Recombx™ MaTa Autoantibody Test         4/20/2015         10           351087         Recombx™ Zic4 Antibody Test         4/20/2015         10           350092         SCA1 DNA Test         4/20/2015         11           350091         SCA2 DNA Test         4/20/2015         11           350094         SCA3 (Machado-Joseph Disease) DNA Test         4/20/2015         12           350090         SCA6 DNA Test         4/20/2015         12	91622		NeoEncephalitis Paraneoplastic Profile with Recombx	4/20/2015	8
649097       OPMD DNA Test       4/20/2015       9         843020       PMP22 Duplication/Deletion Test       4/20/2015       9         844815       Recombx™ CAR (Anti-Recoverin) Autoantibody Test       4/20/2015       9         849448       Recombx™ CV2 Antibody Test       4/20/2015       10         848890       Recombx™ MaTa Autoantibody Test       4/20/2015       10         851087       Recombx™ Zic4 Antibody Test       4/20/2015       10         850092       SCA1 DNA Test       4/20/2015       11         850095       SCA2 DNA Test       4/20/2015       11         850091       SCA2 DNA Test       4/20/2015       12         850090       SCA6 DNA Test       4/20/2015       12	S52438		Neuromyelitis Optica (NMO) AQP4 Autoantibody Test	4/20/2015	8
S43020       PMP22 Duplication/Deletion Test       4/20/2015       9         S44815       Recombx™ CAR (Anti-Recoverin) Autoantibody Test       4/20/2015       9         S49448       Recombx™ CV2 Antibody Test       4/20/2015       10         S48890       Recombx™ MaTa Autoantibody Test       4/20/2015       10         S51087       Recombx™ Zic4 Antibody Test       4/20/2015       10         S50092       SCA1 DNA Test       4/20/2015       11         S50095       SCA17 DNA Test       4/20/2015       11         S50091       SCA2 DNA Test       4/20/2015       11         S50094       SCA3 (Machado-Joseph Disease) DNA Test       4/20/2015       12         S50090       SCA6 DNA Test       4/20/2015       12	\5007 <u>8</u>		Notch3 (CADASIL) DNA Sequencing Test	4/20/2015	8
Recombx™ CAR (Anti-Recoverin) Autoantibody Test       4/20/2015       9         849448       Recombx™ CV2 Antibody Test       4/20/2015       10         848890       Recombx™ MaTa Autoantibody Test       4/20/2015       10         851087       Recombx™ Zic4 Antibody Test       4/20/2015       10         850092       SCA1 DNA Test       4/20/2015       11         850375       SCA2 DNA Test       4/20/2015       11         850091       SCA2 DNA Test       4/20/2015       11         850094       SCA3 (Machado-Joseph Disease) DNA Test       4/20/2015       12         850090       SCA6 DNA Test       4/20/2015       12	<u> </u>		OPMD DNA Test	4/20/2015	9
S49448       Recombx™ CV2 Antibody Test       4/20/2015       10         S48890       Recombx™ MaTa Autoantibody Test       4/20/2015       10         S51087       Recombx™ Zic4 Antibody Test       4/20/2015       10         S50092       SCA1 DNA Test       4/20/2015       11         S50375       SCA17 DNA Test       4/20/2015       11         S50091       SCA2 DNA Test       4/20/2015       11         S50094       SCA3 (Machado-Joseph Disease) DNA Test       4/20/2015       12         S50090       SCA6 DNA Test       4/20/2015       12	<u>543020</u>		PMP22 Duplication/Deletion Test	4/20/2015	9
S48890       Recombx™ MaTa Autoantibody Test       4/20/2015       10         S51087       Recombx™ Zic4 Antibody Test       4/20/2015       10         S50092       SCA1 DNA Test       4/20/2015       11         S50375       SCA17 DNA Test       4/20/2015       11         S50091       SCA2 DNA Test       4/20/2015       11         S50094       SCA3 (Machado-Joseph Disease) DNA Test       4/20/2015       12         S50090       SCA6 DNA Test       4/20/2015       12	S4481 <u>5</u>		Recombx™ CAR (Anti-Recoverin) Autoantibody Test	4/20/2015	9
851087       Recombx™ Zic4 Antibody Test       4/20/2015       10         850092       SCA1 DNA Test       4/20/2015       11         850375       SCA17 DNA Test       4/20/2015       11         850091       SCA2 DNA Test       4/20/2015       11         850094       SCA3 (Machado-Joseph Disease) DNA Test       4/20/2015       12         850090       SCA6 DNA Test       4/20/2015       12	<u> </u>		Recombx™ CV2 Antibody Test	4/20/2015	10
SES0092       SCA1 DNA Test       4/20/2015       11         SES0375       SCA17 DNA Test       4/20/2015       11         SES0091       SCA2 DNA Test       4/20/2015       11         SES0094       SCA3 (Machado-Joseph Disease) DNA Test       4/20/2015       12         SES0090       SCA6 DNA Test       4/20/2015       12	S48890		Recombx™ MaTa Autoantibody Test	4/20/2015	10
SE0375       SCA17 DNA Test       4/20/2015       11         SE0091       SCA2 DNA Test       4/20/2015       11         SE0094       SCA3 (Machado-Joseph Disease) DNA Test       4/20/2015       12         SE0090       SCA6 DNA Test       4/20/2015       12	<u>851087</u>		Recombx™ Zic4 Antibody Test	4/20/2015	10
350091       SCA2 DNA Test       4/20/2015       11         350094       SCA3 (Machado-Joseph Disease) DNA Test       4/20/2015       12         350090       SCA6 DNA Test       4/20/2015       12	550092		SCA1 DNA Test	4/20/2015	11
SE0094         SCA3 (Machado-Joseph Disease) DNA Test         4/20/2015         12           SE0090         SCA6 DNA Test         4/20/2015         12	<u>850375</u>		SCA17 DNA Test	4/20/2015	11
SCA6 DNA Test 4/20/2015 12	<u>550091</u>		SCA2 DNA Test	4/20/2015	11
	<u>850094</u>		SCA3 (Machado-Joseph Disease) DNA Test	4/20/2015	12
Sensory Neuropathy Evaluation 4/20/2015 12	<u>550090</u>		SCA6 DNA Test	4/20/2015	12
	349551		Sensory Neuropathy Evaluation	4/20/2015	12

<u>91566</u>	SHOX (GHD) DNA Sequencing and Deletion Test	4/20/2015	13
<u>\$42575</u>	TTR DNA Sequencing Test	4/20/2015	13
<u>S51255</u>	VGKC Antibody Test	4/20/2015	14

## **Test Changes**

The following test changes will be effective on the dates indicated below. **Please note information that is changing appears in bold text in this update.** Former test names and test codes have been italicized.

ADmark® ApoE Genotype Analysis & Interpretation (Symptomatic)		
Clinical Significance	Detection of ApoE2, E3, E4 alleles	
Effective Date	4/20/2015	
Former Test Name	APO E Genotype (For Late-Onset Alzeimer's Disease)	
Test Code	S46315	
Specimen Requirements	8 mL (6 mL minimum) whole blood collected in an EDTA (lavender-top) tube	
	Buccal swab is not acceptable	
Instructions	Higher blood volumes ensure adequate DNA quantity, which varies with WBC, specimen condition, and need for confirmatory testing.	
Specimen Stability	Room temperature and Refrigerated: 10 days Frozen: Call Lab	
Methodology	Restriction Fragment Length Polymorphism (RFLP)	
Performing Site	Athena Diagnostics, Inc.	

ADmark® Phospho-Tau/Total-Tau/Ab42 CSF Analysis & Interpretation (Symptomatic)	
Clinical Significance	Correlates levels of Phosphorylated-Tau protein, Total-Tau protein and AB42 peptide in CSF
Effective Date	4/20/2015
Test Code	S44955
Specimen Requirements	2 mL (0.5 mL minimum) CSF collected in a Polypropylene transfer tube
Transport Temperature	Refrigerated
Specimen Stability	Room temperature: 72 hours Refrigerated: 21 days Frozen: 4 months
Methodology	Enzyme-Linked Immunosorbent Assay (ELISA)
Performing Site	Athena Diagnostics, Inc.

ADmark® PS-1 DNA Sequencing Test		
Clinical Significance	Detects sequence variations in the presentlin 1 (PS-1) gene	
Effective Date	4/20/2015	
Former Test Name	Admark Symptomatic PS-1	
Test Code	S49327	
CPT Codes	81405	

Specimen Requirements	8 mL (6 mL minimum) whole blood collected in an EDTA (lavender-top) tube
Instructions	Higher blood volumes ensure adequate DNA quantity, which varies with WBC, specimen condition, and need for confirmatory testing.
Transport Temperature	Room temperature
Specimen Stability	Room temperature and Refrigerated: 10 days Frozen: Call Lab
Methodology	Sanger Sequencing
Performing Site	Athena Diagnostics, Inc.

Complete HNPP Evaluation		
Clinical Significance	Detects duplications/deletions and sequence variants in the PMP22 gene	
Effective Date	4/20/2015	
Former Test Name	HNPPC Evaluation	
Test Code	S49335	
CPT Codes	81324, 81325	
Specimen Requirements	8 mL (6 mL minimum) whole blood collected in an EDTA (lavender-top) tube Pediatric: 2 mL (1 mL minimum)  ACD-A (yellow-top) tube is not acceptable	
Instructions	Higher blood volumes ensure adequate DNA quantity, which varies with WBC, specimen condition, and need for confirmatory testing. Patients, 0-3 years have higher WBC, yielding more DNA per mL of blood.	
Transport Temperature	Room temperature	
Specimen Stability	Room temperature and Refrigerated: 10 days Frozen: Unacceptable	
Methodology	Next Generation Sequencing and Multiplex Ligation-dependent Probe Amplification (MLPA)	
Performing Site	Athena Diagnostics, Inc.	

Dystonia (DYT1) DNA Test		
Clinical Significance	Detects GAG deletion in DYT1 gene	
Effective Date	4/20/2015	
Former Test Name	Dystonia	
Test Code	S48185	
CPT Codes	81400	
Specimen Requirements	8 mL (6 mL minimum) whole blood collected in an EDTA (lavender-top) tube Pediatric: 2 mL (1 mL minimum)	
Instructions	Higher blood volumes ensure adequate DNA quantity, which varies with WBC, specimen condition, and need for confirmatory testing. Patients, 0-3 years have higher WBC, yielding more DNA per mL of blood.	
Transport Temperature	Room temperature	
Specimen Stability	Room temperature and Refrigerated: 10 days Frozen: Unacceptable	
Methodology	Polymerase Chain Reaction (PCR) and Fragment Size Analysis	
Performing Site	Athena Diagnostics, Inc.	

Familial Amyotrophic Lateral Sclerosis (SOD1) DNA Test		
Clinical Significance	Detects mutations in the superoxide dismutase (SOD1) gene. 20% of adult-onset familial ALS cases are associated with SOD1 mutations.	
Effective Date	4/20/2015	
Test Code	S49489	
CPT Codes	81404	
Specimen Requirements	8 mL (6 mL minimum) whole blood collected in an EDTA (lavender-top) tube	
Instructions	Higher blood volumes ensure adequate DNA quantity, which varies with WBC, specimen condition, and need for confirmatory testing.	
Transport Temperature	Room temperature	
Specimen Stability	Room temperature and Refrigerated: 10 days Frozen: Unacceptable	
Set-up/Analytic Time	Report available: 28-56 days	
Methodology	Sanger Sequencing	
Performing Site	Athena Diagnostics, Inc.	

Familial Hemiplegic Migraine Type I (CACNA1A) DNA Test		
Clinical Significance	Detects sequence variants in the CACNA1A gene	
Effective Date	4/20/2015	
Test Code	S52411	
Specimen Requirements	8 mL (6 mL minimum) whole blood collected in an EDTA (lavender-top) tube Pediatric: 2 mL (1 mL minimum)	
Instructions	Higher blood volumes ensure adequate DNA quantity, which varies with WBC, specimen condition, and need for confirmatory testing. Patients, 0-3 years have higher WBC, yielding more DNA per mL of blood.	
Transport Temperature	Room temperature	
Specimen Stability	Room temperature and Refrigerated: 10 days Frozen: Unacceptable	
Methodology	Next Generation Sequencing	
Performing Site	Athena Diagnostics, Inc.	

GALOP™ Autoantibody Test		
Clinical Significance	Detection of serum IgM antibodies binding to GALOP antigen	
Effective Date	4/20/2015	
Former Test Name	GALOP Autoantibodies	
Test Code	S46850	
CPT Codes	83520 (x2)	
Specimen Requirements	2 mL (0.5 mL minimum) serum	
Transport Temperature	Refrigerated	
Specimen Stability	Room temperature: 72 hours Refrigerated: 21 days Frozen: 4 months	
Set-up/Analytic Time	Report available: 7-14 days	

Methodology	Enzyme-Linked Immunosorbent Assay (ELISA)
Performing Site	Athena Diagnostics, Inc.

LHON mtDNA Evaluation	
Clinical Significance	Detection of point mutations in mitochondrial genes: a G to A at base pair 11778 in the ND4 gene; a T to C at base pair 14484 in the ND6 gene; and a G to A at base pair 3460 in the ND1 gene
Effective Date	4/20/2015
Former Test Name	Lebers Hered Optic Neuropathy DNA Mutation
Test Code	S44395
Specimen Requirements	8 mL (6 mL minimum) whole blood collected in an EDTA (lavender-top) tube Pediatric: 2 mL (1 mL minimum)
Instructions	Higher blood volumes ensure adequate DNA quantity, which varies with WBC, specimen condition, and need for confirmatory testing. Patients, 0-3 years have higher WBC, yielding more DNA per mL of blood.
Transport Temperature	Room temperature
Specimen Stability	Room temperature and Refrigerated: 10 days Frozen: Unacceptable
Methodology	Polymerase Chain Reaction (PCR) and Restriction Fragment Length Polymorphism (RFLP)
Performing Site	Athena Diagnostics, Inc.

MELAS mtDNA Evaluation	
Clinical Significance	Detects six mtDNA point mutations linked to MELAS syndrome
Effective Date	4/20/2015
Test Code	S51269
CPT Codes	81401
Specimen Requirements	Preferred: 8 mL (6 mL minimum) whole blood collected in an EDTA (lavender-top) tube Pediatric: 2 mL (1 mL minimum)  Acceptable: 100 mg Muscle tissue collected in an Cryovial
Instructions	Higher blood volumes ensure adequate DNA quantity, which varies with WBC, specimen condition, and need for confirmatory testing. Patients, 0-3 years have higher WBC, yielding more DNA per mL of blood.
Transport Temperature	Room temperature
Specimen Stability	Room temperature and Refrigerated: 10 days Frozen: Unacceptable
Methodology	Polymerase Chain Reaction (PCR) and Restriction Fragment Length Polymorphism (RFLP)
Performing Site	Athena Diagnostics, Inc.

MFN2 DNA Sequencing Test	
Clinical Significance	Detects sequence variants in the MFN2 gene
Effective Date	4/20/2015
Test Code	S50699
Specimen Requirements	8 mL (6 mL minimum) whole blood collected in an EDTA (lavender-top) tube Pediatric: 2 mL (1 mL minimum)

	ACD-A (yellow-top) tube is not acceptable
Instructions	Higher blood volumes ensure adequate DNA quantity, which varies with WBC, specimen condition, and need for confirmatory testing. Patients, 0-3 years have higher WBC, yielding more DNA per mL of blood.
Transport Temperature	Room temperature
Specimen Stability	Room temperature and Refrigerated: 10 days Frozen: Unacceptable
Methodology	Next Generation Sequencing
Performing Site	Athena Diagnostics, Inc.

Monogenic Diabetes (MODY) Evaluation	
Clinical Significance	Detects deletions in the HNF4A, GCK, HNF1A, HNF1B genes and mutations in the HNF4A, GCK, HNF1A, HNF1B and IPF1
Effective Date	4/20/2015
Test Code	S51162
CPT Codes	81404 (x2), 81405 (x2), 81406 (x2), 81479
Specimen Requirements	8 mL (6 mL minimum) whole blood collected in an EDTA (lavender-top) tube Pediatric: 2 mL (1 mL minimum)
	ACD (yellow-top) tubes are not acceptable
Instructions	Higher blood volumes ensure adequate DNA quantity, which varies with WBC, specimen condition, and need for confirmatory testing. Patients, 0-3 years have higher WBC, yielding more DNA per mL of blood.
Transport Temperature	Room temperature
Specimen Stability	Room temperature and Refrigerated: 10 days Frozen: Unacceptable
Set-up/Analytic Time	Report available: 28-42 days
Methodology	Sanger Sequencing and Multiplex Ligation-dependent Probe Amplification (MLPA)
Performing Site	Athena Diagnostics, Inc.

MPZ DNA Sequencing Test	
Clinical Significance	Detects sequence variations in the Myelin Protein Zero gene
Effective Date	4/20/2015
Test Code	S48727
CPT Codes	81405
Specimen Requirements	8 mL (6 mL minimum) whole blood collected in an EDTA (lavender-top) tube Pediatric: 2 mL (1 mL minimum)
	ACD-A (yellow-top) tubes are not acceptable
Instructions	Higher blood volumes ensure adequate DNA quantity, which varies with WBC, specimen condition, and need for confirmatory testing. Patients, 0-3 years have higher WBC, yielding more DNA per mL of blood.
Transport Temperature	Room temperature
Specimen Stability	Room temperature and Refrigerated: 10 days Frozen: Unacceptable
Methodology	Next Generation Sequencing
Performing Site	Athena Diagnostics, Inc.

MuSK Antibody Test	
Clinical Significance	Detection of antibodies to muscle-specific receptor tyrosine kinase (MuSK) (titer test)
Effective Date	4/20/2015
Test Code	S50437
Specimen Requirements	2 mL (0.5 mL minimum) serum
Specimen Stability	Room temperature: 72 hours Refrigerated: 21 days Frozen: 4 months
Set-up/Analytic Time	Report available: 7-14 days
Methodology	Radioimmuno Assay (RIA)
Performing Site	Athena Diagnostics, Inc.

NeoCerebellar Degeneration Paraneoplastic Evaluation with Recombx™	
Clinical Significance	Detection of anti-CV2, anti-Hu, anti-Ma, anti-Ta, anti-Ri, anti-Yo and anti-Zic4 antibodies using recombinant human antigens; anti-GAD65 antibodies using ELISA
Effective Date	4/20/2015
Test Code	S49558
CPT Codes	83516 (x2), 84181, 84182 (x5)
Specimen Requirements	2 mL (0.5 mL minimum) serum
Transport Temperature	Refrigerated
Specimen Stability	Room temperature: 72 hours Refrigerated: 21 days Frozen: 4 months
Methodology	Enzyme-Linked Immunosorbent Assay (ELISA) and Western Blot
Performing Site	Athena Diagnostics, Inc.

NeoComplete Paraneoplastic Evaluation with Recombx™	
Clinical Significance	Detection of anti-Hu, anti-CV2, anti-Yo, anti-Ri, anti-CAR, anti-Ma, anti-Ta and anti-Zic4 antibodies using recombinant human antigens; antibodies to voltage gated calcium channels (VGCC), P/Q type and voltage gated potassium channels (VGKC) and Ganglionic AChR using RIA, Amphiphysin using Immunoblot, NMDA receptor (NR1-subunit), LGI1, and CASPR2 using IFA, GAD65 using ELISA
Effective Date	4/20/2015
Test Code	91636
CPT Codes	83516 (x2), 83519 (x3), 84181, 84182 (x6), 86255 (x3)
Specimen Requirements	2 mL (0.5 mL minimum) serum
Transport Temperature	Refrigerated
Specimen Stability	Room temperature: 72 hours Refrigerated: 21 days Frozen: 4 months
Methodology	Western Blot, Immunofluorescence Assay (IFA), Radioimmuno Assay (RIA) and Enzyme-Linked Immunosorbent Assay (ELISA)
Performing Site	Athena Diagnostics, Inc.

NeoEncephalitis Paraneoplastic Profile with Recombx	
Clinical Significance	Detection of anti-Hu, anti-CV2, anti-Ma, and anti-Ta antibodies using recombinant human antigens; antibodies to voltage gated potassium channels (VGKC) using RIA and Amphiphysin using Immunoblot, anti-GAD65 antibodies using ELISA, anti-NMDA receptor (NR1-subunit antibodies),anti- LGI1, and anti-CASPR2 antibodies using IFA
Effective Date	4/20/2015
Former Test Name	Neo Encephalitis Paraneoplastic Evaluation w/Recombx
Test Code	91622
CPT Codes	83516 (x2), 83519, 84181, 84182 (x2), 86255 (x3)
Specimen Requirements	2 mL (0.5 mL minimum) serum
Transport Temperature	Refrigerated
Specimen Stability	Room temperature: 72 hours Refrigerated: 21 days Frozen: 4 months
Methodology	Western Blot, Immunofluorescence Assay (IFA), Radioimmuno Assay (RIA), Enzyme-Linked Immunosorbent Assay (ELISA) and Immunoblot
Performing Site	Athena Diagnostics, Inc.

Neuromyelitis Optica (NMO) AQP4 Autoantibody Test	
Effective Date	4/20/2015
Former Test Name	Neuromyelitis Optica (NMO)
Test Code	S52438
Specimen Requirements	2 mL (0.5 mL minimum) serum
Transport Temperature	Refrigerated
Specimen Stability	Room temperature: 72 hours Refrigerated: 21 days Frozen: 4 months
Performing Site	Athena Diagnostics, Inc.

Notch3 (CADASIL) DNA Sequencing Test	
Clinical Significance	Detects sequence variants in the Notch3 gene
Effective Date	4/20/2015
Former Test Name	Alt-Notch 3 DNA Sequencing Test
Test Code	A50078
Specimen Requirements	8 mL (6 mL minimum) whole blood collected in an EDTA (lavender-top) tube Pediatric: 2 mL (1 mL minimum)
Instructions	Higher blood volumes ensure adequate DNA quantity, which varies with WBC, specimen condition, and need for confirmatory testing. Patients, 0-3 years have higher WBC, yielding more DNA per mL of blood.
Transport Temperature	Room temperature
Specimen Stability	Room temperature and Refrigerated: 10 days Frozen: Unacceptable
Methodology	Next Generation Sequencing
Performing Site	Athena Diagnostics, Inc.

OPMD DNA Test	
Clinical Significance	Detects GCG trinucleotide expansions in the PABP2 gene
Effective Date	4/20/2015
Former Test Name	Oculopharyngeal Muscular Dystrophy DNA
Test Code	S49097
CPT Codes	81401
Specimen Requirements	8 mL (6 mL minimum) whole blood collected in an EDTA (lavender-top) tube Pediatric: 2 mL (1 mL minimum)
Instructions	Higher blood volumes ensure adequate DNA quantity, which varies with WBC, specimen condition, and need for confirmatory testing. Patients, 0-3 years have higher WBC, yielding more DNA per mL of blood.
Transport Temperature	Room temperature
Specimen Stability	Room temperature and Refrigerated: 10 days Frozen: Unacceptable
Reference Range	Normal: 6 GCG trinucleotide repeats
Methodology	Repeat Expansion Detection by PCR
Performing Site	Athena Diagnostics, Inc.

PMP22 Duplication/Deletion Test	
Clinical Significance	Detects rearrangements in the PMP22 gene. This test performs both duplication and deletion analysis to detect the cause of CMT1A and HNPP.
Effective Date	4/20/2015
Former Test Name	PMP22 DNA Duplication/Deletion Test (CMT1A)
Test Code	S43020
CPT Codes	81324
Specimen Requirements	8 mL (6 mL minimum) whole blood collected in an EDTA (lavender-top) tube
Instructions	Higher blood volumes ensure adequate DNA quantity, which varies with WBC, specimen condition, and need for confirmatory testing. Patients, 0-3 years have higher WBC, yielding more DNA per mL of blood.
Transport Temperature	Room temperature
Specimen Stability	Room temperature and Refrigerated: 10 days Frozen: Unacceptable
Methodology	Multiplex Ligation-dependent Probe Amplification (MLPA)
Performing Site	Athena Diagnostics, Inc.

Recombx™ CAR (Anti-Recoverin) Autoantibody Test	
Clinical Significance	Detection of anti-recoverin antibodies using recombinant human antigens
Effective Date	4/20/2015
Former Test Name	CAR Autoantibody Test
Test Code	S44815
Specimen Requirements	2 mL (0.5 mL minimum) serum
Specimen Stability	Room temperature: 72 hours

	Refrigerated: 21 days Frozen: 4 months
Set-up/Analytic Time	Report available: 7-14 days
Methodology	Western Blot
Performing Site	Athena Diagnostics, Inc.

Recombx™ CV2 Antibody Test	
Clinical Significance	Detection of anti-CV2 autoantibodies using recombinant human antigens
Effective Date	4/20/2015
Former Test Name	CV2 Autoantibody Test
Test Code	S49448
Specimen Requirements	Preferred: 2 mL (0.5 mL minimum) serum  Acceptable: 2 mL (0.5 mL minimum) CSF collected in a tube not containing additives
Specimen Stability	Room temperature: 72 hours Refrigerated: 21 days Frozen: 4 months
Set-up/Analytic Time	Report available: 7-14 days
Methodology	Western Blot
Performing Site	Athena Diagnostics, Inc.

Recombx™ MaTa Autoantibody Test	
Clinical Significance	Detection of anti-Ma and anti-Ta antibodies using recombinant human antigens
Effective Date	4/20/2015
Former Test Name	MA/TA Antibody
Test Code	S48890
Specimen Requirements	2 mL (0.5 mL minimum) serum
Specimen Stability	Room temperature: 72 hours Refrigerated: 21 days Frozen: 4 months
Set-up/Analytic Time	Report available: 7-14 days
Methodology	Western Blot
Performing Site	Athena Diagnostics, Inc.

Recombx™ Zic4 Antibody Test	
Clinical Significance	Detection of anti-Zic4 antibodies using recombinant human antigens
Effective Date	4/20/2015
Former Test Name	ZIC4 AB
Test Code	S51087
Specimen Requirements	2 mL (0.5 mL minimum) serum
Specimen Stability	Room temperature: 72 hours

	Refrigerated: 21 days Frozen: 4 months
Set-up/Analytic Time	Report available: 7-14 days
Reference Range	Negative
Methodology	Western Blot
Performing Site	Athena Diagnostics, Inc.

SCA1 DNA Test	
Clinical Significance	Detects CAG triplet repeat expansion in the SCA1 gene
Effective Date	4/20/2015
Former Test Name	Spinocerebellar Ataxia Type 1 DNA Test
Test Code	S50092
Specimen Requirements	8 mL (6 mL minimum) whole blood collected in an EDTA (lavender-top) tube Pediatric: 2 mL (1 mL minimum)
Instructions	Higher blood volumes ensure adequate DNA quantity, which varies with WBC, specimen condition, and need for confirmatory testing. Patients, 0-3 years have higher WBC, yielding more DNA per mL of blood.
Specimen Stability	Room temperature and Refrigerated: 10 days Frozen: Unacceptable
Methodology	Repeat Expansion Detection by PCR
Performing Site	Athena Diagnostics, Inc.

SCA17 DNA Test	
Clinical Significance	Detects CAG/CAA triplet repeat expansion in the SCA17 gene
Effective Date	4/20/2015
Former Test Name	Spinocerebellar Ataxia Type 17
Test Code	S50375
Specimen Requirements	8 mL (6 mL minimum) whole blood collected in an EDTA (lavender-top) tube Pediatric: 2 mL (1 mL minimum)
Instructions	Higher blood volumes ensure adequate DNA quantity, which varies with WBC, specimen condition, and need for confirmatory testing. Patients, 0-3 years have higher WBC, yielding more DNA per mL of blood.
Transport Temperature	Room temperature
Specimen Stability	Room temperature and Refrigerated: 10 days Frozen: Unacceptable
Methodology	Repeat Expansion Detection by PCR
Performing Site	Athena Diagnostics, Inc.

SCA2 DNA Test	
Clinical Significance	Detects CAG triplet repeat expansion in the SCA2 gene
Effective Date	4/20/2015
Former Test Name	Spinocerebellar Ataxia Type 2
Test Code	S50091
Specimen Requirements	8 mL (6 mL minimum) whole blood collected in an EDTA (lavender-top) tube

	Pediatric: 2 mL (1 mL minimum)
Instructions	Higher blood volumes ensure adequate DNA quantity, which varies with WBC, specimen condition, and need for confirmatory testing. Patients, 0-3 years have higher WBC, yielding more DNA per mL of blood.
Transport Temperature	Room temperature
Specimen Stability	Room temperature and Refrigerated: 10 days Frozen: Unacceptable
Methodology	Repeat Expansion Detection by PCR
Performing Site	Athena Diagnostics, Inc.

SCA3 (Machado-Joseph Disease) DNA Test	
Clinical Significance	Detects CAG triplet repeat expansion in the SCA3 gene
Effective Date	4/20/2015
Former Test Name	Spinocerebellar Ataxia Type 3
Test Code	S50094
Specimen Requirements	8 mL (6 mL minimum) whole blood collected in an EDTA (lavender-top) tube Pediatric: 2 mL (1 mL minimum)
Instructions	Higher blood volumes ensure adequate DNA quantity, which varies with WBC, specimen condition, and need for confirmatory testing. Patients, 0-3 years have higher WBC, yielding more DNA per mL of blood.
Transport Temperature	Room temperature
Specimen Stability	Room temperature and Refrigerated: 10 days Frozen: Unacceptable
Methodology	Repeat Expansion Detection by PCR
Performing Site	Athena Diagnostics, Inc.

SCA6 DNA Test	
Clinical Significance	Detects CAG triplet repeat expansion in the SCA6 gene
Effective Date	4/20/2015
Former Test Name	Spinocerebellar Ataxia Type 6
Test Code	S50090
Specimen Requirements	8 mL (6 mL minimum) whole blood collected in an EDTA (lavender-top) tube Pediatric: 2 mL (1 mL minimum)
Instructions	Higher blood volumes ensure adequate DNA quantity, which varies with WBC, specimen condition, and need for confirmatory testing. Patients, 0-3 years have higher WBC, yielding more DNA per mL of blood.
Transport Temperature	Room temperature
Specimen Stability	Room temperature and Refrigerated: 10 days Frozen: Unacceptable
Methodology	Repeat Expansion Detection by PCR
Performing Site	Athena Diagnostics, Inc.

Sensory Neuropathy Evaluation	
Clinical Significance	Detection of anti-MAG, anti-SGPG and anti-Sulfatide antibodies. Detection of anti-Hu antibodies using recombinant human antigens. Detection of serum IgM antibodies binding to GALOP antigen.
Effective Date	4/20/2015

Former Test Name	Sensory Neuropathy Profile - "XP"
Test Code	S49551
CPT Codes	83516, 83520 (x6)
Specimen Requirements	2 mL (0.5 mL minimum) serum
Transport Temperature	Refrigerated
Specimen Stability	Room temperature: 72 hours Refrigerated: 21 days Frozen: 4 months
Set-up/Analytic Time	Report available: 7-14 days
Methodology	Enzyme-Linked Immunosorbent Assay (ELISA) and Western Blot
Performing Site	Athena Diagnostics, Inc.

SHOX (GHD) DNA Sequencing and Deletion Test	
Clinical Significance	Detects deletions and sequence variations in the SHOX gene
Effective Date	4/20/2015
Test Code	91566
CPT Codes	81405, 81479
Specimen Requirements	8 mL (6 mL minimum) whole blood collected in an EDTA (lavender-top) tube Pediatric: 2 mL (1 mL minimum)
Instructions	Higher blood volumes ensure adequate DNA quantity, which varies with WBC, specimen condition, and need for confirmatory testing. Patients, 0-3 years have higher WBC, yielding more DNA per mL of blood.
Transport Temperature	Room temperature
Specimen Stability	Room temperature and Refrigerated: 10 days Frozen: Unacceptable
Methodology	Multiplex Ligation-dependent Probe Amplification (MLPA) and Sanger Sequencing
Performing Site	Athena Diagnostics, Inc.

TTR DNA Sequencing Test	
Clinical Significance	Detects sequence variants in the Transthyretin (TTR) gene
Effective Date	4/20/2015
Former Test Name	Amyloidosis/TTR MET-30
Test Code	S42575
Specimen Requirements	8 mL (6 mL minimum) whole blood collected in an EDTA (lavender-top) tube Pediatric: 2 mL (1 mL minimum)
Instructions	Higher blood volumes ensure adequate DNA quantity, which varies with WBC, specimen condition, and need for confirmatory testing. Patients, 0-3 years have higher WBC, yielding more DNA per mL of blood.
Transport Temperature	Room temperature
Specimen Stability	Room temperature and Refrigerated: 10 days Frozen: Unacceptable
Methodology	Sanger Sequencing
Performing Site	Athena Diagnostics, Inc.

VGKC Antibody Test	
Clinical Significance	Detection of antibodies to voltage gated potassium channels (VGKC)
Effective Date	4/20/2015
Test Code	S51255
Specimen Requirements	2 mL (0.5 mL minimum) serum
Transport Temperature	Refrigerated
Specimen Stability	Room temperature: 72 hours Refrigerated: 21 days Frozen: 4 months
Set-up/Analytic Time	Report available: 7-14 days
Units Of Measure	pMol/L
Methodology	Radioimmuno Assay (RIA)
Performing Site	Athena Diagnostics, Inc.