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.				
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S44955		ADmark® Phospho-Tau/Total-Tau/Ab42 CSF Analysis & Interpretation (Symptomatic)	4/20/2015	2
S49327		**ADmark® PS-1 DNA Sequencing Test**	4/20/2015	2
S49335		**Complete HNPP Evaluation**	4/20/2015	3
S48185		**Dystonia (DYT1) DNA Test**	4/20/2015	3
S49489		Familial Amyotrophic Lateral Sclerosis (SOD1) DNA Test	4/20/2015	4
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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 presenilin 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.