HOTLINE: Effective July 5, 2019

New Test Available Now **3001907 Myotonic Dystrophy Type 1 (*DMPK*) CTG Expansion DM1 PCR**

Methodology: Polymerase Chain Reaction/Capillary Electrophoresis
Performed: Varies
Reported: 7-10 days

Specimen Required: Collect: Lavender (K_2EDTA), Pink (K_2EDTA), or Yellow (ACD Solution A or B).
Specimen Preparation: Transport 5 mL whole blood. (Min: 3 mL)
Storage/Transport Temperature: Refrigerated.
Stability (collection to initiation of testing): Ambient: 1 week; Refrigerated: 1 month; Frozen: 6 months

Reference Interval: By report

Interpretive Data:
Background Information for Myotonic Dystrophy Type 1 (*DMPK*):
Characteristics: Myotonic dystrophy type 1 (DM1) is a multisystem disorder characterized by myotonic myopathy with involvement of the eye, heart, endocrine system and central nervous system. Clinical findings span a continuum from mild to severe, with overlap in the three recognized clinical subtypes of DM1: mild, classic and congenital. Mild DM1 is adult-onset and features include mild myotonia and premature cataracts or baldness. Onset of classic DM1 is typically between 10-30 years of age and findings include distal muscle weakness, myotonia, cataracts, GI disturbances, and cardiac conduction abnormalities. Congenital DM1 may present prenatally with polyhydramnios and reduced fetal movement, and postnatal features commonly include infantile hypotonia, respiratory insufficiency, facial diplegia, and intellectual disability.
Prevalence: 1:20,000.
Inheritance: Autosomal dominant.
Penetrance: Age-related, approaches 100 percent by age 50.
Cause: Expanded number of CTG repeats in the *DMPK* gene.
Normal: 5-34 CTG repeats, stably transmitted, not associated with DM1 manifestations.
Premutation: 35-49 CTG repeats, may be unstably transmitted, not associated with DM1 manifestations.
Full-penetrance disease allele: 50 or more CTG repeats, unstably transmitted, associated with DM1 manifestations.
Clinical Sensitivity: >99 percent for DM1.
Methodology: Triplet repeat-primed polymerase chain reaction (PCR) followed by size analysis using capillary electrophoresis to assess the CTG repeat in the *DMPK* 3' untranslated region. Specific allele sizing estimates cannot be determined for CTG repeats of >150. Repeat sizing precision is approximately +/- 2 repeats for alleles with 5-24 repeats and +/- 4 repeats for alleles with 77 to 150 repeats.
Analytical Sensitivity and Specificity: 99 percent.
Limitations: Diagnostic errors can occur due to rare sequence variations. This assay will not detect myotonic dystrophy type 2.

Phenotype	Number of CTG Repeats
Normal allele	Less than or equal to 34
Premutation	35 - 49
Mild	50 – approx. 150
Classic	approx.100 - approx 1000
Congenital	>1000

See Compliance Statement C: www.aruplab.com/CS

CPT Code(s): 81234

New York DOH approval pending. Call for status update.

HOTLINE NOTE: Refer to the Test Mix Addendum for interface build information.