SCHOOL OF MEDICINE
INDIANA UNIVERSITY

Department of Medical and Molecular Genetics
Division of Diagnostic Genomics

Laboratory Test Directory

Myotonic Dystrophy Type 1, *DMPK* PCR

CPT Code(s): 81403

Service Code (IU Health): 53025516

Ordering Recommendation: *DMPK* mutation analysis is recommended for an individual with a clinical diagnosis of myotonic dystrophy type 1(DM-1); Carrier identification in individuals with a positive family history of DM-1.

Synonyms: DM-1 mutation analysis, *DMPK* CTG genotyping, CTG repeats

Methodology: Triplet Repeat Primed PCR (TP-PCR) and capillary electrophoresis. Southern analysis is performed as a send-out test, as needed, to further characterize expanded/abnormal alleles.

Performed: Mon-Fri

Reported: 6-9 days

Specimen Requirements

Patient Preparation: None required for whole blood

Collect: Lavender (EDTA) tubes; buccal swab; DNA

Specimen Volume: Blood: 2-6 mL whole blood; Buccal swab (Lab provides the collection tube)

Storage/Transport: Refrigerated/Room temperature

Unacceptable Conditions: Grossly hemolyzed or clotted

Remarks:

Stability: 2 weeks refrigerated; 1 month frozen

Reference Interval: by report

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Interpretive Data

Characteristics: Myotonic dystrophy (DM1) is a multisystem neuromuscular disorder with 3 distinct presentations. The mild form has cataracts and mild myotonia but life span is normal. Classic DM1 is characterized by muscle weakness and wasting, myotonia, cataracts, and often cardiac conduction abnormalities. Life span can be reduced. Congenital DM1 phenotypically presents with hypotonia at birth, often with respiratory insufficiency and early death. Other phenotypic characteristics may include intellectual disability.

Inheritance: Autosomal dominant

Cause: This disease is the result of an expansion of a CTG tri-nucleotide repeat found in the (*DMPK*) gene. The CTG repeat size varies from: 5-34 repeats (normal); 35-49 repeats (potentially unstable allele / "premutation"); 50 or more repeats (abnormal, mild to severe symptoms). Thus, normal patients should have two alleles each with sizes not more than 34 CTG repeats.

Limitations: Low level (<5%) mixture/mosaicism may not be detected. Although rare, false positive or false negative results may occur. All results should be interpreted in context of clinical findings, relevant history, and other laboratory data.

References:

1. Singh S, Zhang A, DlouhyS and Bai S. Detection of large expansions in myotonic dystrophy type 1 using triplet primed PCR. Front. Genet., doi: 10.3389/fgene.2014.00094. 2014
2. Bird T. Myotonic Dystrophy Type 1. GeneReviews. Last Update: 2013.
3. International Myotonic Dystrophy Consortium. New nomenclature and DNA testing guidelines for myotonic dystrophy type 1 (DM1). Accessed 9-25-12. 2000