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PATIENT REPORT

Patient Age/Sex: Unknown

Specimen Collected: 05-Sep-23 17:04

Myotonic Dystrophy Type 1 (DMPK) |Received: 05-Sep-23 17:04 Report/Verified: 05-Sep-23 17:16

Procedure	Result	Units	Reference Interval
Myotonic Dystrophy (DM1) - Specimen	Whole Blood		
Myotonic Dystrophy (DM1) -Allele 1	10	CTG repeats	
Myotonic Dystrophy (DM1) -Allele 2	65	CTG repeats	
Myotonic Dystrophy (DM1) Interpretation	See Note [f1 i1]		

Result Footnote

f1: Myotonic Dystrophy (DM1) Interpretation

RESULT
One pathogenic expansion and one allele in the normal size range were detected in the DMPK gene.

INTERPRETATION
One pathogenic, full-penetrance disease allele was detected in the DMPK gene. This result is consistent with a diagnosis of myotonic dystrophy type 1 (DM1). Clinical manifestations are highly variable and age-dependent.

Offspring of this individual have a 50 percent chance of inheriting the expanded allele. Full-penetrance disease alleles may expand during transmission, including expansion to repeat sizes associated with congenital DM1. Expansions associated with congenital DM1 most commonly occur when the expanded allele is maternally transmitted. Relatives of this individual, including offspring, are at risk for developing DM1 including the congenital form of DM1.

RECOMMENDATIONS
Genetic consultation, including a discussion of medical screening and management, is indicated. At-risk adult family members and symptomatic family members of any age should be offered Myotonic Dystrophy Type 1 (DMPK) CTG Expansion (ARUP test code 3001907).

COMMENTS
Reference Sequence: GenBank # NM_001081563.1

This result has been reviewed and approved by [redacted]

Test Information

i1: Myotonic Dystrophy (DM1) Interpretation

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

Test Information

i1: Myotonic Dystrophy (DM1) Interpretation

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.

This test was developed and its performance characteristics determined by ARUP Laboratories. It has not been cleared or approved by the US Food and Drug Administration. This test was performed in a CLIA certified laboratory and is intended for clinical purposes.

Counseling and informed consent are recommended for genetic testing. Consent forms are available online.