ARUP LABORATORIES
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Jonathan R. Genzen, MD, PhD, Chief Medical Officer

Client: Example Client ABC123
123 Test Drive
Salt Lake City, UT 84108
UNITED STATES

Physician: Doctor, Example

Patient: Patient, Example

DOB	9/26/1972
Gender:	Male
Patient Identifiers:	01234567890ABCD, 012345
Visit Number (FIN):	01234567890ABCD
Collection Date:	00/00/0000 00:00

Myotonic Dystrophy Type 1 (DMPK) CTG Expansion

ARUP test code 3001907

Myotonic Dystrophy (DM1) - Specimen	Whole Blood
Myotonic Dystrophy (DM1) - Allele 1	13 CTG repeats
Myotonic Dystrophy (DM1) - Allele 2	21 CTG repeats
Myotonic Dystrophy (DM1) Interpretation	See Note

RESULT
No CTG expansion present in the DMPK gene.

INTERPRETATION
This individual has two DMPK CTG repeats within the normal size range; thus, will neither be affected with nor transmit myotonic dystrophy type 1 (DM1).

RECOMMENDATIONS
Medical management should rely on clinical findings and family history. Genetic consultation is recommended.

COMMENTS
Reference Sequence: GenBank # NM_001081563.1

This result has been reviewed and approved by [redacted]

H=High, L=Low, *=Abnormal, C=Critical

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.

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.

H=High, L=Low, *=Abnormal, C=Critical

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
Myotonic Dystrophy (DM1) - Specimen	22-231-147435	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Myotonic Dystrophy (DM1) - Allele 1	22-231-147435	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Myotonic Dystrophy (DM1) - Allele 2	22-231-147435	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Myotonic Dystrophy (DM1) Interpretation	22-231-147435	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00

END OF CHART

H=High, L=Low, *=Abnormal, C=Critical