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

Physician: Doctor, Example

Patient: Patient, Example

DOB	8/19/1988
Gender:	Female
Patient Identifiers:	01234567890ABCD, 012345
Visit Number (FIN):	01234567890ABCD
Collection Date:	00/00/0000 00:00

Duchenne/Becker Muscular Dystrophy (DMD) Deletion/Duplication

ARUP test code 2011235

Duchenne/Becker MD (DMD) DelDup Specimen	Whole Blood

Duchenne/Becker MD (DMD) DelDup Interp

Negative

RESULT
No pathogenic variants were detected in the DMD gene.

INTERPRETATION
No pathogenic variants were detected in the DMD gene by deletion/duplication analysis. This result reduces the likelihood that this individual is a carrier of Duchenne/Becker muscular dystrophy. Please refer to the background information included in this report for the clinical sensitivity and limitations of this test.

RECOMMENDATIONS
Medical screening and management should rely on clinical findings and family history. Because large deletions and duplications account for only a subset of causative variants in the DMD gene, consideration should be given to ordering Duchenne/Becker Muscular Dystrophy (DMD) Sequencing (ARUP test code 2011153). Genetic consultation is recommended.

COMMENTS
Reference Sequence: GenBank # NM_004006.2

This result has been reviewed and approved by [redacted]

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

Background information for Duchenne/Becker Muscular Dystrophy (DMD) Deletion/Duplication:
Characteristics: Symptoms of Duchenne muscular dystrophy (DMD) usually begin before age 6 and include fatigue, learning difficulties, muscle weakness (beginning in legs and pelvis), progressive difficulty walking with wheelchair needed at approximately 12 years and breathing difficulties and heart disease by age 20 years. Symptoms of Becker muscular dystrophy (BMD) are similar to DMD but start later and progress at a slower rate. Dilated cardiomyopathy has been observed in nearly all affected males and many female carriers of DMD and BMD.
Incidence: DMD: 1 in 3,500 male births, BMD: 1 in 19,000 male births.
Inheritance: X-linked; de novo mutations occur in one-third of cases.
Penetrance: Males: 100 percent. Females: Varies with X-chromosome inactivation.
Cause: Pathogenic DMD mutations.
Clinical Sensitivity: DMD: 55-75 percent, BMD: 75-90 percent.
Methodology: Multiplex ligation-dependent probe amplification (MLPA) to detect large exonic deletions/duplications.
Analytical Sensitivity and Specificity: Greater than 99 percent.
Limitations: DMD base pair substitutions, small deletions/duplications, deep intronic, and regulatory region mutations will not be detected. Breakpoints for large deletions/duplications will not be determined. Diagnostic errors can occur due to rare sequence variation.

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.

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
Duchenne/Becker MD (DMD) DelDup Specimen	24-113-400031	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Duchenne/Becker MD (DMD) DelDup Interp	24-113-400031	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