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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** | 12/9/2021 |
| **Sex:** | Male |
| **Patient Identifiers:** | 01234567890ABCD, 012345 |
| **Visit Number (FIN):** | 01234567890ABCD |
| **Collection Date:** | 01/01/2017 12:34 |

## 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

TEST PERFORMED - 2011235
TEST DESCRIPTION - Duchenne/Becker Muscular Dystrophy (DMD) Deletion/Duplication
INDICATION FOR TESTING - Predictive Testing

RESULT
Negative for the requested pathogenic variant in the DMD gene.

INTERPRETATION
According to information provided to ARUP, this individual has family history of Becker Muscular Dystrophy (BMD) and several family members with a deletion of exons 14-15 in the DMD gene. The pathogenic familial deletion of exons 14-15 in the DMD gene was not detected by deletion/duplication analysis. This variant was reported to be associated with BMD in the family; therefore, this individual is predicted to be unaffected.

Evidence for variant classification: The deletion of DMD exons 14-15 is reported in the literature in an individual with Duchenne muscular dystrophy and an individual with Becker muscular dystrophy (Ling 2020, Taylor 2007). This deletion is not predicted to alter the DMD reading frame, predicting the milder Becker muscular dystrophy (Monaco 1988). Based on available information, this deletion is classified as pathogenic. However, this variant was not detected in this individual.

RECOMMENDATIONS
Genetic consultation is recommended.

A familial positive control was not available. Because the variant was identified in a family member by another clinical laboratory and/or by a different testing methodology, testing of a positive familial control is a recommended quality measure to confirm that this assay is able to identify the requested familial variant. ARUP will test a control sample from a family member known to harbor the familial variant at no additional cost within 30 days of this report to confirm this negative result. Contact an ARUP genetic counselor (800-242-2787 x2141) to submit a positive familial control.

COMMENTS
Reference Sequence: GenBank # NM_004006.2

Note: A positive familial control was not tested.

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

REFERENCES
Ling C et al. Exonic rearrangements in DMD in Chinese Han individuals affected with Duchenne and Becker muscular dystrophies. Hum Mutat. 2020 Mar 41(3):668-677. PMID: 31705731.

Monaco AP et al. An explanation for the phenotypic differences between patients bearing partial deletions of the DMD locus. Genomics. 1988 Jan;2(1):90-5. PMID: 3384440.

Taylor PJ et al. Measurement of the clinical utility of a combined mutation detection protocol in carriers of Duchenne and Becker muscular dystrophy. J Med Genet. 2007 Jun 44(6):368-72. PMID: 17259292.

This result has been reviewed and approved by [redacted]

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 | 22-073-102427 | 3/14/2022 10:43:00 AM | 3/15/2022 2:14:14 PM | 3/27/2022 8:13:00 AM |
| Duchenne/Becker MD (DMD) DelDup Interp | 22-073-102427 | 3/14/2022 10:43:00 AM | 3/15/2022 2:14:14 PM | 3/27/2022 8:13:00 AM |

END OF CHART

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