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

Physician: Doctor, Example

Patient: Patient, Example

DOB	4/9/2000
Sex:	Female
Patient Identifiers:	01234567890ABCD, 012345
Visit Number (FIN):	01234567890ABCD
Collection Date:	01/01/2017 12:34

Duchenne/Becker Muscular Dystrophy (DMD) Deletion/Duplication, Fetal

ARUP test code 2011231

Maternal Contamination Study Fetal Spec	Fetal Cells Single fetal genotype present; no maternal cells present. Fetal and maternal samples were tested using STR markers to rule out maternal cell contamination.
Maternal Contam Study, Maternal Spec	Whole Blood For quality assurance purposes, ARUP Laboratories will confirm the above result at no charge following delivery. Order Confirmation of Fetal Testing and include a copy of the original fetal report (or the mother's name and date of birth) with the test submission. Please contact an ARUP genetic counselor at (800) 242-2787 extension 2141 prior to specimen submission.
DMD DelDup Fetal Specimen	Cultured Amnio
Duchenne/Becker DelDup Fetal Interp	Negative TEST PERFORMED - 2011231 TEST DESCRIPTION - Duchenne/Becker Muscular Dystrophy (DMD) Deletion/Duplication, Fetal INDICATION FOR TESTING - Prenatal Diagnosis for DMD/BMD. RESULT Negative for the requested pathogenic variant in the DMD gene. INTERPRETATION According to information provided to ARUP, the mother of this fetus is a carrier of a pathogenic deletion of exons 3-55 in the DMD gene. The familial pathogenic deletion of exons 3-55 in the DMD gene was not detected in this prenatal sample by deletion/duplication analysis; therefore, this fetus is predicted to be unaffected. Evidence for variant classification: The deletion of DMD exons 3-55 deletes 52/79 exons but is not predicted to alter the DMD reading frame. A similar overlapping in-frame deletion (exons 3-51), has been reported in the ClinVar database (Variation ID:526179), and other large in-frame deletions are associated with DMD/BMD (Den Dunnen 1989, Ling 2020, Takeshima 2010). Based on available information, the deletion of DMD exons 3-55 is classified as pathogenic. However, this variant was not detected in this prenatal sample. RECOMMENDATIONS

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

Genetic consultation is recommended. For quality assurance purposes, ARUP Laboratories will confirm the above result at no charge following delivery. Order Confirmation of Fetal Testing and include a copy of the original fetal report (or the mother's name and date of birth) with the test submission. Please contact an ARUP genetic counselor (800-242-2787 ext 2141) prior to specimen submission.

COMMENTS
Reference Sequence: GenBank # NM_004006.2

REFERENCES
American Academy of Pediatrics Section on Cardiology and Cardiac Surgery. Cardiovascular health supervision for individuals affected by Duchenne or Becker muscular dystrophy. Pediatrics. 2005; 116(6):1569-73.

Den Dunnen JT et al. Topography of the Duchenne muscular dystrophy (DMD) gene: FIGE and cDNA analysis of 194 cases reveals 115 deletions and 13 duplications. Am J Hum Genet. 1989 Dec;45(6):835-47. PMID: 2573997.

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.

Takeshima Y et al. Mutation spectrum of the dystrophin gene in 442 Duchenne/Becker muscular dystrophy cases from one Japanese referral center. J Hum Genet. 2010 Jun;55(6):379-88. PMID:20485447.

This result has been reviewed and approved by [redacted]

Background information for Duchenne/Becker Muscular Dystrophy (DMD) Deletion/Duplication, Fetal:
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.

For quality assurance purposes, ARUP Laboratories will confirm the above result at no charge following delivery. Order Confirmation of Fetal Testing and include a copy of the original fetal report (or the mother's name and date of birth) with the test submission. Please contact an ARUP genetic counselor at (800) 242-2787 extension 2141 prior to specimen submission.

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
Maternal Contamination Study Fetal Spec	21-270-157586	9/27/2021 12:23:00 PM	10/15/2021 1:48:33 PM	10/20/2021 3:08:00 PM
Maternal Contam Study, Maternal Spec	21-270-157586	9/27/2021 12:23:00 PM	10/15/2021 1:48:33 PM	10/20/2021 3:08:00 PM
DMD DelDup Fetal Specimen	21-270-157586	9/27/2021 12:23:00 PM	10/15/2021 1:48:33 PM	10/26/2021 7:24:00 PM
Duchenne/Becker DelDup Fetal Interp	21-270-157586	9/27/2021 12:23:00 PM	10/15/2021 1:48:33 PM	10/26/2021 7:24:00 PM

END OF CHART