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

Physician: Doctor, Example

Patient: Patient, Example

DOB	12/31/1752
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	**Deletion** *

TEST PERFORMED 2011231
TEST DESCRIPTION - Duchenne/Becker Muscular Dystrophy (DMD) Deletion/Duplication, Fetal
INDICATION FOR TESTING - Prenatal Diagnosis

RESULT
One pathogenic variant was detected in the DMD gene.

DNA VARIANT
Classification: Pathogenic
Gene: DMD
Nucleic Acid Change: Deletion of exons 49-51 Hemizygous In-frame deletion

INTERPRETATION
According to information provided to ARUP, the mother of this fetus carries a pathogenic deletion of DMD exons 49-51. The pathogenic familial deletion of exons 49-51 was detected in this prenatal sample by deletion/duplication analysis. This deletion is not expected to alter the reading frame. This deletion was reported to be associated with Becker muscular dystrophy in the family; therefore, this fetus is predicted to be affected.

Evidence for variant classification: The deletion of DMD exons 49-51 is reported in the literature in individuals with Duchenne muscular dystrophy or Becker muscular dystrophy (Esposito 2017, Kaspar 2009, Ling 202, Mital 1998, Rani 2013). This deletion is

not predicted to alter the reading frame of the Dystrophin protein. Consistent with the DMD reading frame hypothesis (Monaco 1988), this variant is predicted to be associated with the milder Becker muscular dystrophy (MIM: 300376). Based on available information, this deletion is considered to be pathogenic.

RECOMMENDATIONS
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

Note: A familial positive control was not tested.

REFERENCES
Esposito et al. Precise mapping of 17 deletion breakpoints within the central hotspot deletion region (introns 50 and 51) of the DMD gene. J Hum Genet. 2017 Dec;62(12):1057-1063. PMID: 28878337

Kaspar et al. Analysis of dystrophin deletion mutations predicts age of cardiomyopathy onset in becker muscular dystrophy. Circ Cardiovasc Genet. 2009; 2(6): 544-551.

Ling 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; 2(1): 90-95.

Mital et al. Molecular characterisation of Duchenne muscular dystrophy and phenotypic correlation. J Neurol Sci. 1998; 157(2): 179-186.

Rani et al. Mutation spectrum of dystrophin gene in malaysian patients with Duchenne/Becker muscular dystrophy. J Neurogenet. 2013; 27(1-2): 11-15.

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	22-116-104468	4/26/2022 11 01:00 AM	4/26/2022 11:02:06 AM	4/29/2022 12:54:00 PM
Maternal Contam Study, Maternal Spec	22-116-104468	4/26/2022 11 01:00 AM	4/26/2022 11:02:06 AM	4/29/2022 12:54:00 PM
DMD DelDup Fetal Specimen	22-116-104468	4/26/2022 11 01:00 AM	4/26/2022 11:02:06 AM	4/29/2022 12:54:00 PM
Duchenne/Becker DelDup Fetal Interp	22-116-104468	4/26/2022 11 01:00 AM	4/26/2022 11:02:06 AM	4/29/2022 12:54:00 PM

END OF CHART

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