Client: ARUP Example Report Only
500 Chipeta Way
Salt Lake City, UT 84108
UNITED STATES

Physician: arup, arup

Patient: EXAMPLE, POSITIVE

DOB	
Gender:	Female
Patient Identifiers:	32864
Visit Number (FIN):	33174
Collection Date:	9/27/2021 08:56

Distal Arthrogryposis Panel, Sequencing

ARUP test code 3003917

Distal Arthrogryposis Specimen	Whole Blood

Distal Arthrogryposis Interp

Positive

INDICATION FOR TESTING
Distal arthrogryposis.

RESULT
One pathogenic variant was detected in the MYH3 gene.

PATHOGENIC VARIANT
Gene: MYH3 (NM_002470.3)
Nucleic Acid Change: c.2015G>A; Heterozygous
Amino Acid Alteration: p.Arg672His
Inheritance: Autosomal Dominant

INTERPRETATION
One copy of a pathogenic variant, c.2015G>A; p.Arg672His, was detected in the MYH3 gene by massively parallel sequencing and confirmed by Sanger sequencing. Pathogenic MYH3 variants are inherited in an autosomal dominant manner and are associated with distal arthrogryposis (MIM: 193700). This result is consistent with a diagnosis of distal arthrogryposis, Freeman-Sheldon syndrome type. Offspring of this individual have a 50 percent chance of inheriting the pathogenic variant.

No additional pathogenic variants were identified in the targeted genes by massively parallel sequencing. Please refer to the background information included in this report for a list of the genes analyzed and limitations of this test.

Evidence for variant classification:
The MYH3 c.2015G>A; p.Arg672His variant is one of the most common pathogenic variants reported in patients with distal arthrogryposis, Freeman-Sheldon syndrome type. Clinical manifestations are highly variable.

RECOMMENDATIONS
Genetic consultation is indicated, including a discussion of medical screening and management. At-risk family members should be offered testing for the identified pathogenic MYH3 variant (Familial Mutation, Targeted Sequencing, ARUP test code 2001961).

COMMENTS
Likely benign and benign variants are not included in this report.

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

REFERENCES
Toydemir R et al. (2006) Mutations in embryonic myosin heavy chain (MYH3) cause Freeman-Sheldon syndrome and Sheldon-Hall syndrome. Nat Genet. 38(5):561-5.

This result has been reviewed and approved by [redacted]

BACKGROUND INFORMATION: Distal Arthrogryposis Panel, Sequencing

CHARACTERISTICS: Distal arthrogryposes (DA) are a subset of arthrogryposis disorders that involve contractures of the distal parts of the limbs. The contractures are congenital but typically do not have primary neurologic and/or muscle disease; the shared findings among DA include a consistent pattern of hand and foot involvement, limited involvement of the proximal joints, and variable expressivity. There are multiple types of DA caused by different genes (genetic heterogeneity).
PREVALENCE: 1 in

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
Distal Arthrogryposis Specimen	21-270-103033	9/27/2021 8:56 00 AM	9/27/2021 8:56:58 AM	9/27/2021 8:59:00 AM
Distal Arthrogryposis Interp	21-270-103033	9/27/2021 8:56 00 AM	9/27/2021 8:56:58 AM	9/27/2021 8:59:00 AM

END OF CHART

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