

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

Physician: arup, arup

**Patient: EXAMPLE, NEGATIVE**

**DOB**

**Gender:** Female

**Patient Identifiers:** 30801

**Visit Number (FIN):** 31107

**Collection Date:** 6/8/2021 16:03

## Heterotaxy & Situs Inversus Panel, Sequencing

ARUP test code 3002682

Heterotaxy and Situs Inversus Specimen whole Blood

Heterotaxy and Situs Inversus Interp

Negative

INDICATION FOR TESTING  
Situs inversus totalis.

**RESULT**

No pathogenic variants were detected in any of the genes tested.

**INTERPRETATION**

No pathogenic variants were identified by massively parallel sequencing of the coding regions and exon-intron boundaries of the genes tested. This result decreases the likelihood of, but does not exclude, a heritable laterality defect. Please refer to the background information included in this report for a list of the genes analyzed and limitations of this test.

**RECOMMENDATIONS**

Medical screening and management of this individual should rely on clinical findings and family history. Genetic consultation is recommended.

**COMMENTS**

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

This result has been reviewed and approved by [REDACTED]

**BACKGROUND INFORMATION:** Heterotaxy and Situs Inversus Panel, Sequencing

**CHARACTERISTICS:** Laterality defects such as heterotaxy and situs inversus are developmental defects characterized by the abnormal placement of the abdominal (visceral) organs.

**EPIDEMIOLOGY:** Heterotaxy syndrome affects approximately 1 in 10,000 individuals. This condition is causative of about 3 percent of congenital heart defects cases.

**CAUSE:** Pathogenic germline variants in genes associated with left-right symmetry in early embryo development.

**INHERITANCE:** Varies

**PENETRANCE:** Varies; some associated genes exhibit reduced penetrance.

**GENES TESTED:** ANKS6\*, ARL2BP, ARMC4\*, CCDC103\*, CCDC114\*, CCDC151, CCDC39, CCDC40\*, CFAP298\*, CFAP53, CRELD1, DNAAF1, DNAAF2, DNAAF3, DNAAF4, DNAAF5\*, DNAH1, DNAH11, DNAH5, DNAIL1, DNAI2\*, DNAL1, FOXH1, GATA4, GATA6\*, INVS, LRRC6, MMP21, NKX2-5, NME8, NODAL, PIH1D3, PKD1L1\*, SPAG1\*, ZIC3, ZMYND10

\*One or more exons are not covered by sequencing for the

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

Unless otherwise indicated, testing performed at:

**ARUP LABORATORIES | 800-522-2787 | aruplab.com**  
500 Chipeta Way, Salt Lake City, UT 84108-1221  
Tracy I. George, MD, Laboratory Director

Patient: EXAMPLE, NEGATIVE  
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indicated gene; see limitations section below.  
METHODODOLOGY: Capture of all coding exons and exon-intron junctions of the targeted genes, followed by massively parallel sequencing. Sanger sequencing was performed as necessary to fill in regions of low coverage and confirm reported variants.  
ANALYTICAL SENSITIVITY/SPECIFICITY: The analytical sensitivity of this test is approximately 99 percent for single nucleotide variants (SNVs) and greater than 93 percent for insertions/duplications/deletions from 1-10 base pairs in size. Variants greater than 10 base pairs may be detected, but the analytical sensitivity may be reduced.  
LIMITATIONS: A negative result does not exclude a heritable laterality defect. This test only detects variants within the coding regions and intron-exon boundaries of the targeted genes. Regulatory region variants and deep intronic variants will not be identified. Deletions/duplications/insertions of any size may not be detected by massively parallel sequencing. Diagnostic errors can occur due to rare sequence variations. In some cases, variants may not be identified due to technical limitations in the presence of pseudogenes, repetitive, or homologous regions. This assay may not detect low-level mosaic or somatic variants associated with disease. Interpretation of this test result may be impacted if this patient has had an allogeneic stem cell transplantation. Noncoding transcripts were not analyzed.

The following regions are not sequenced due to technical limitations of the assay:

ANKS6(NM\_173551) exon(s) 1  
ARMC4(NM\_001290020) exon(s) 9  
ARMC4(NM\_001290021) exon(s) 13  
ARMC4(NM\_001312689) exon(s) 4  
ARMC4(NM\_018076) exon(s) 9  
CCDC103(NM\_001258397) exon(s) 4  
CCDC114(NM\_001364171) exon(s) 3  
CCDC114(NM\_001364171) partial exon(s) 4(Chr19:48822049-48822069)  
CCDC40(NM\_001243342) exon(s) 18  
CFAP298(NM\_001350335) partial exon(s) 5(Chr21:33975399-33975450)  
CFAP298(NM\_001350337) partial exon(s) 6(Chr21:33974534-33974561)  
DNAAF5(NM\_017802) exon(s) 1  
DNAI2(NM\_001353167) exon(s) 13  
GATA6(NM\_005257) partial exon(s) 2(Chr18:19751812-19751963)  
PKD1L1(NM\_138295) partial exon(s) 8(Chr7:47955029-47955060)  
SPAG1(NM\_001374321) partial exon(s) 11(Chr8:101225456-101225529)  
SPAG1(NM\_003114) partial exon(s) 11(Chr8:101225456-101225529)  
SPAG1(NM\_172218) partial exon(s) 11(Chr8:101225456-101225529)

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.

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VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
Heterotaxy and Situs Inversus Specimen	21-159-122657	6/8/2021 4 03:00 PM	6/8/2021 4:04:02 PM	6/8/2021 4:09:00 PM
Heterotaxy and Situs Inversus Interp	21-159-122657	6/8/2021 4 03:00 PM	6/8/2021 4:04:02 PM	6/8/2021 4:09:00 PM

END OF CHART

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