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

Physician: Doctor, Example

Patient: Patient, Example

DOB	Unknown
Gender:	Unknown
Patient Identifiers:	01234567890ABCD, 012345
Visit Number (FIN):	01234567890ABCD
Collection Date:	00/00/0000 00:00

Heterotaxy & Situs Inversus Panel, Sequencing

ARUP test code 3002682

Heterotaxy and Situs Inversus Specimen	Whole Blood

Heterotaxy and Situs Inversus Interp

Negative

NDICATION 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, DNAI1, 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 indicated gene; see limitations section below.

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

METHODOLOGY: 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.

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

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
Heterotaxy and Situs Inversus Specimen	22-182-102807	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Heterotaxy and Situs Inversus Interp	22-182-102807	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00

END OF CHART

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