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

Physician: Doctor, Example

**Patient: Patient, Example**

| | |
|---|---|
| **DOB** | 5/21/1966 |
| **Gender:** | Female |
| **Patient Identifiers:** | 01234567890ABCD, 012345 |
| **Visit Number (FIN):** | 01234567890ABCD |
| **Collection Date:** | 00/00/0000 00:00 |

## Primary Ciliary Dyskinesia Panel, Sequencing

ARUP test code 3001621

| | |
|---|---|
| Primary Ciliary Dyskinesia Specimen | Whole Blood |

Primary Ciliary Dyskinesia Interp

Negative

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

INTERPRETATION
No pathogenic variants were detected in any of the genes tested. This result decreases the likelihood of, but does not exclude, a diagnosis of primary ciliary dyskinesia. Please refer to the background information included in this report for a list of the genes analyzed, methodology, and limitations of this test.

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

COMMENTS
Likely benign and benign variants are not reported.
Variants in the following region(s) may not be detected by NGS with sufficient confidence in this sample due to technical limitations: None

This result has been reviewed and approved by [redacted]

BACKGROUND INFORMATION: Primary Ciliary Dyskinesia Panel, Sequencing

CHARACTERISTICS: Primary ciliary dyskinesia (PCD, also known as Kartagener syndrome) is a rare inherited condition that results from an underlying defect in the structure or function of motile cilia, impacting multiple body systems. Patients with PCD typically first present with neonatal respiratory distress, chronic oto-sinopulmonary disease, and year-round wet coughing. Approximately half of patients with PCD will have a laterality defect such as situs inversus totalis or heterotaxy. PCD is also associated with infertility and ectopic pregnancy due to ciliary dysfunction.

PREVALENCE: Approximately 1 in 16,000

CAUSE: Pathogenic germline variants in genes associated with structure and function of the motile cilia

INHERITANCE: Autosomal recessive; rare X-linked recessive forms have been reported

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

PENETRANCE: 100 percent

GENES TESTED: ARMC4*, CCDC103*, CCDC114*, CCDC151, CCDC39, CCDC40*, CCDC65, CCNO, CFAP298*, DNAAF1, DNAAF2, DNAAF3, DNAAF4, DNAAF5*, DNAH1, DNAH11, DNAH5, DNAI1, DNAI2*, DNAL1, DRC1, GAS8, LRRC6, MCIDAS, NME8, PIH1D3, RSPH1, RSPH3, RSPH4A, RSPH9, SPAG1*, ZMYND10
*One or more exons are not covered by sequencing for the indicated gene; see limitations section below.

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. Human genome build 19 (Hg 19) was used for data analysis.

ANALYTICAL SENSITIVITY/SPECIFICITY: The analytical sensitivity of sequencing 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 diagnosis of primary ciliary dyskinesia. 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. 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:
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 exons(s) 4(Chr19:48822049-48822069)
CCDC40(NM_001243342) exon(s) 18
CFAP298(NM_001350335) partial exons(s) 5(Chr21:33975399-33975450)
CFAP298(NM_001350337) partial exons(s) 6(Chr21:33974534-33974561)
DNAAF5(NM_017802) exon(s) 1
DNAI2(NM_001353167) exon(s) 13
SPAG1(NM_001374321) partial exons(s) 11(Chr8:101225456-101225529)
SPAG1(NM_003114) partial exons(s) 11(Chr8:101225456-101225529)
SPAG1(NM_172218) partial exons(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.

VERIFIED/REPORTED DATES

| Procedure | Accession | Collected | Received | Verified/Reported |
|---|---|---|---|---|
| Primary Ciliary Dyskinesia Specimen | 24-324-400928 | 00/00/0000 00:00 | 00/00/0000 00:00 | 00/00/0000 00:00 |
| Primary Ciliary Dyskinesia Interp | 24-324-400928 | 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