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

Physician: Doctor, Example

Patient: Patient, Example

DOB	3/7/2021
Gender:	Female
Patient Identifiers:	01234567890ABCD, 012345
Visit Number (FIN):	01234567890ABCD
Collection Date:	00/00/0000 00:00

Stickler Syndrome Panel, Sequencing

ARUP test code 3001613

Stickler Syndrome Specimen	Whole Blood
Stickler Syndrome Interp	Negative INDICATION FOR TESTING Confirm Diagnosis 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 diagnosis of Stickler syndrome or a related disorder. Please refer to the background information included in this report for a list of the genes analyzed and the 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 included in this report. This result has been reviewed and approved by Yuan Ji, Ph.D.

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

BACKGROUND INFORMATION: Stickler Syndrome Panel, Sequencing

CHARACTERISTICS: Stickler syndrome and related disorders are a group of connective tissue disorders characterized by ocular abnormalities, hearing loss, and skeletal or joint problems.
INCIDENCE: Approximately 1/7500 to 1/9000 newborns
CAUSE: Pathogenic germline variants in certain genes associated with collagen formation.
INHERITANCE: Most cases are autosomal dominant; there are rare autosomal recessive causes.
PENETRANCE: 100 percent
CLINICAL SENSITIVITY: Variable, dependent on phenotype
GENES TESTED: COL11A1, COL11A2, COL2A1, COL9A1, COL9A2, COL9A3, VCAN
METHODOLOGY: Capture of all coding exons and exon-intron junctions of the targeted genes, followed by massively parallel sequencing. Sanger sequencing is performed as necessary to fill in regions of low coverage and confirm reported variants. Human genome build 19 (Hg 19) is used for data analysis.
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 diagnosis of Stickler syndrome or a related disorder. 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 and breakpoints of large deletions/duplications will not be determined. Deletions/duplications/insertions of any size may not be detected by massive 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. Non-coding transcripts are not analyzed.

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
Stickler Syndrome Specimen	21-068-111040	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Stickler Syndrome Interp	21-068-111040	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