ARUP LABORATORIES | aruplab.com
500 Chipeta Way, Salt Lake City, Utah 84108-1221
phone: 801-583-2787, toll free: 800-522-2787
Tracy I. George, MD, Chief Medical Officer

Patient Report

Client: UU University Division Validation
50 N. Medical Drive
Salt Lake City, UT 84132-
USA

Provider: 9999-ARUP -ARUP,

Patient: **SS NGS, CERTVAL1**
DOB: 09-Jun-98
Gender: Male
Patient Identifiers: 558815
Visit Number (FIN): 581975
Client Supplied ID:

Specimen Collected: 24-Feb-21 16:52

Stickler Syndrome by NGS **|Received: 24-Feb-21 16:52** **Report/Verified: 24-Feb-21 16:55**

	Result	Units	Reference Interval
Stickler Syndrome Specimen	Whole Blood		
Stickler Syndrome Interp	Negative [i1]		

Test Information

i1: Stickler Syndrome Interp

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%
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

Unless otherwise indicated, testing performed at:
ARUP Laboratories
500 Chipeta Way, Salt Lake City, UT 84108
Laboratory Director: Tracy I. George, MD

ARUP Accession: n/a
Report Request ID: 13707367
Printed: 24-Feb-21 16:57

ARUP LABORATORIES | aruplab.com
500 Chipeta Way, Salt Lake City, Utah 84108-1221
phone: 801-583-2787, toll free: 800-522-2787
Tracy I. George, MD, CMO

Patient:	**SS NGS, CERTVAL1**
DOB:	09-Jun-98
Patient Identifiers:	558815

Test Information

i1: Stickler Syndrome Interp

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.

Unless otherwise indicated, testing performed at:
ARUP Laboratories
500 Chipeta Way, Salt Lake City, UT 84108
Laboratory Director: Tracy I. George, MD