

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

Physician: arup, arup

**Patient: Genomics, SHD NGS 2**

**DOB**

**Sex:** Male

**Patient Identifiers:** 36456

**Visit Number (FIN):** 36775

**Collection Date:** 2/23/2022 12:03

**Hereditary Paranglioma-Pheochromocytoma (SDHA, SDHB, SDHC, and SDHD) Sequencing and Deletion/Duplication**

ARUP test code 3004480

SDH Specimen

whole blood

SDH Interp

Negative

**RESULT**

No pathogenic variants were detected in SDHA, SDHB, SDHC, or SDHD genes.

**INTERPRETATION**

No pathogenic variants were identified by massively parallel sequencing of the coding regions and exon-intron boundaries of the SDHA, SDHB, SDHC, or SDHD genes. No large exonic deletions or duplications were identified in the genes tested. This result decreases the likelihood of, but does not exclude, a diagnosis of hereditary paraganglioma-pheochromocytoma. 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 should rely on clinical findings and family history. If suspicion remains for a hereditary cancer syndrome, consideration should be given to ordering the Hereditary Cancer Panel, Sequencing and Deletion/Duplication (ARUP test code 2012032). Genetic consultation is recommended.

**COMMENTS**

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

**BACKGROUND INFORMATION:** Hereditary

Paranglioma-Pheochromocytoma  
(SDHA, SDHB, SDHC, and SDHD) Panel,  
Sequencing and Deletion/Duplication

**CHARACTERISTICS:** Hereditary paraganglioma-pheochromocytoma (PGL/PCC) syndromes are familial cancer syndromes characterized by neuroendocrine tumors: paragangliomas (neuroendocrine tumors of the autonomic nervous system) and pheochromocytomas (paragangliomas of the adrenal medulla). Pathogenic germline variants in SDHA, SDHB, SDHC, and SDHD, among several other genes, predispose individuals to paraganglioma and pheochromocytoma with an increased risk for malignancy.

**CAUSE:** Pathogenic germline variants in succinate dehydrogenase, subunits A, B, C, and D (SDHA, SDHB, SDHC, and SDHD), and other genes

**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: Genomics, SHD NGS 2  
ARUP Accession: 22-054-107086  
Patient Identifiers: 36456  
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INHERITANCE: Autosomal dominant; parent-of-origin effect for SDHD

PENETRANCE: Variable and age dependent

CLINICAL SENSITIVITY: 22-45 percent

GENES TESTED: SDHA\* (NM\_004168), SDHB (NM\_003000), SDHC (NM\_003001), SDHD (NM\_003002)

\* - One or more exons are not covered by sequencing, and deletion/duplication detection is not available for this gene; see limitations section below.

METHODOLOGY: Probe hybridization-based 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 to confirm reported variants that do not meet acceptable quality metrics. Human genome build 19 (Hg 19) was used for data analysis. Multiplex ligation-dependent probe amplification (MLPA) of the targeted genes.

ANALYTICAL SENSITIVITY/SPECIFICITY: The analytical sensitivity is approximately 99 percent for single nucleotide variants (SNVs) and greater than 93 percent for insertions/duplications/deletions (indels) from 1-10 base pairs in size. Indels greater than 10 base pairs may be detected, but the analytical sensitivity may be reduced. Specificity is greater than 99.9 percent for all variant classes. The analytical sensitivity for MLPA is greater than 99 percent.

LIMITATIONS: A negative result does not exclude a diagnosis of hereditary paraganglioma-pheochromocytoma. This test only detects variants within the coding regions and intron-exon boundaries of the SDHA, SDHB, SDHC, and SDHD genes. Deletions/duplications/insertions of any size may not be detected by massively parallel sequencing. Regulatory region variants and deep intronic variants will not be identified. Precise breakpoints for large deletions or duplications are not determined in this assay and single exon deletions/duplications may not be detected based on the breakpoints of the rearrangement. The actual breakpoints for the deletion or duplication may extend beyond or be within the exon(s) reported. Diagnostic errors can occur due to rare sequence variations. In some cases, variants may not be identified due to technical limitations caused by the presence of pseudogenes, repetitive, or homologous regions. This test is not intended to detect low-level mosaic or somatic variants, gene conversion events, complex inversions, translocations, mitochondrial DNA (mtDNA) mutations, or repeat expansions. 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:  
SDHA(NM\_004168) exon 14

This test was developed and its performance characteristics determined by ARUP Laboratories. It has not been cleared or approved by the U.S. 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
SDH Specimen	22-054-107086	2/23/2022 12:03:00 PM	2/23/2022 12:03:49 PM	2/23/2022 12:21:00 PM
SDH Interp	22-054-107086	2/23/2022 12:03:00 PM	2/23/2022 12:03:49 PM	2/23/2022 12:21:00 PM

END OF CHART

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