

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

Physician: Doctor, Example

**Patient: Patient, Example**

**DOB:** 12/23/1953  
**Gender:** Male  
**Patient Identifiers:** 01234567890ABCD, 012345  
**Visit Number (FIN):** 01234567890ABCD  
**Collection Date:** 00/00/0000 00:00

**Hereditary Paraganglioma-Pheochromocytoma (SDHB, SDHC, and SDHD) Sequencing and Deletion/Duplication Panel**

ARUP test code 2007167

HPGL-PCC (SDHB,C,D) Seq, DelDup Specimen whole blood

HPGL-PCC (SDHB,C,D) Seq, DelDup Interp

Negative

TEST PERFORMED - 2007167  
TEST DESCRIPTION - Hereditary Paraganglioma-Pheochromocytoma (SDHB, SDHC, and SDHD) Sequencing and Deletion/Duplication Panel  
INDICATION FOR TEST - Confirm Diagnosis

**RESULT**

No pathogenic variants were detected in the SDHB, SDHC and SDHD genes.

**INTERPRETATION**

No pathogenic variants were detected in the SDHB, SDHC, and SDHD genes by sequencing all coding regions and intron-exon boundaries or by deletion/duplication analysis. This result decreases the probability of, but does not exclude, a diagnosis of Hereditary Paraganglioma-Pheochromocytoma syndrome. Please refer to the background information included in this report for the clinical sensitivity and limitations of this test.

**RECOMMENDATIONS**

Medical screening and management should rely on clinical findings and family history. If there is suspicion for a hereditary cancer syndrome, consideration should be given to ordering the Cancer Panel, Hereditary, Sequencing and Deletion/Duplication, 47 Genes (ARUP test code 2012032). Genetic consultation is recommended.

**COMMENTS**

Reference Sequences: GenBank # NM\_003000.2 (SDHB), NM\_003001.3 (SDHC), NM\_003002.2 (SDHD)  
Nucleotide numbering begins at the "A" of the ATG initiation codon.  
Benign variants are not included in this report but are available upon request.

This result has been reviewed and approved [REDACTED]

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

**BACKGROUND INFORMATION:** Hereditary Paraganglioma-Pheochromocytoma (SDHB, SDHC, and SDHD) Sequencing and Deletion/Duplication Panel

**CHARACTERISTICS:** Hereditary paraganglioma-pheochromocytoma (PGL/PCC) syndromes are characterized by paragangliomas (neuroendocrine tumors of the autonomic nervous system) and pheochromocytomas (paragangliomas of the adrenal medulla). Pathogenic germline mutations in a number of genes, including SDHB, SDHC, and SDHD, predispose to paraganglioma and pheochromocytoma with risk of malignant transformation.

**INCIDENCE:** About 1 in 300,000 per year.

**INHERITANCE:** Autosomal dominant; parent of origin effect for SDHD.

**CAUSE:** Pathogenic succinate dehydrogenase, subunits B, C, and D (SDHB, SDHC, and SDHD) gene mutations. Mutations in other genes, including TMEM127, EGLN1, MAX, SDHA, and SDHAF2, may also be causative.

**CLINICAL SENSITIVITY:** 26-30 percent.

**METHODOLOGY:** Bidirectional sequencing of all coding regions and intron-exon boundaries of the SDHB, SDHC, and SDHD genes; Multiplex Ligation-dependent Probe Amplification (MLPA) to detect large SDHB, SDHC, and SDHD deletions /duplications.

**ANALYTICAL SENSITIVITY AND SPECIFICITY:** Sequencing: 99 percent; MLPA: 90 and 99 percent, respectively.

**LIMITATIONS:** Diagnostic errors can occur due to rare sequence variations. Regulatory region mutations and deep intronic mutations will not be detected. The breakpoints of large deletions/duplications will not be determined. Mutations in genes other than SDHB, SDHC, and SDHD are not evaluated.

Test developed and characteristics determined by ARUP Laboratories. See Compliance Statement C: aruplab.com/CS

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
HPGL-PCC (SDHB,C,D) Seq, DelDup Specimen	17-152-124845	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
HPGL-PCC (SDHB,C,D) Seq, DelDup Interp	17-152-124845	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