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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:	01/01/2017 12:34

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 by Karl Voelkerding, M.D.

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	6/1/2017 8:18:00 PM	6/6/2017 1:01:09 PM	6/14/2017 5:15:17 PM
HPGL-PCC (SDHB,C,D) Seq, DelDup Interp	17-152-124845	6/1/2017 8:18:00 PM	6/6/2017 1:01:09 PM	6/14/2017 5:15:17 PM

END OF CHART

H – high L – low * – abnormal C – critical