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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	9/1/1966
Gender:	Female
Patient Identifiers:	01234567890ABCD, 012345
Visit Number (FIN):	01234567890ABCD
Collection Date:	01/01/2017 12:34

Hereditary Paraganglioma-Pheochromocytoma (SDHC) Sequencing and Deletion/Duplication

ARUP test code 2007117

HPGL-PCC (SDHC) Seq, DelDup Specimen	Whole Blood
HPGL-PCC (SDHC) Seq, DelDup Interp	Negative

Section 79-1 of New York State Civil Rights Law requires informed consent be obtained from all patients (or their legal guardians) prior to pursuing any diagnostic genetic testing or testing to assess carrier status. These forms must be kept on file by the ordering physician. Biochemical and DNA testing patient consent forms can be accessed from ARUP's web site: www.aruplab.com.

TEST PERFORMED - 2007117
TEST DESCRIPTION - Hereditary Paraganglioma-Pheochromocytoma (SDHC) Sequencing and Deletion/Duplication
INDICATION FOR TEST - Not Provided

RESULT
No pathogenic variants were detected in the SDHC gene.

INTERPRETATION
No pathogenic variants were detected in the SDHC gene 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 Sequence: GenBank # NM_003001.3 (SDHC)
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 Elaine Lyon, Ph.D.

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

BACKGROUND INFORMATION: Hereditary Paraganglioma-Pheochromocytoma (SDHC) Sequencing and Deletion/Duplication

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 SDHC, predispose to paraganglioma and pheochromocytoma.
INCIDENCE: About 1 in 300,000 per year.
INHERITANCE: Autosomal dominant.
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: 4 percent.
METHODOLOGY: Bidirectional sequencing of all coding regions and intron-exon boundaries of the SDHC gene; multiplex ligation-dependent probe amplification (MLPA) to detect large SDHC 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 SDHC 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 (SDHC) Seq, DelDup Specimen	17-227-106998	8/15/2017 11:49:00 AM	8/15/2017 11:49:50 AM	8/15/2017 2:49:37 PM
HPGL-PCC (SDHC) Seq, DelDup Interp	17-227-106998	8/15/2017 11:49:00 AM	8/15/2017 11:49:50 AM	8/15/2017 2:49:37 PM

END OF CHART

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