ARUP LABORATORIES
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Tracy I. George, MD, Chief Medical Officer

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

Physician: Doctor, Example

Patient: Patient, Example

DOB	12/12/2016
Gender:	Male
Patient Identifiers:	01234567890ABCD, 012345
Visit Number (FIN):	01234567890ABCD
Collection Date:	01/01/2017 12:34

Legius Syndrome (SPRED1) Sequencing and Deletion/Duplication

ARUP test code 2008347

Legius Syndrome (SPRED1)Seq, DelDup Spcm	Whole Blood
Legius Syndrome (SPRED1)Seq, DelDup Int	Negative

TEST PERFORMED - 2002945
TEST DESCRIPTION - Legius Syndrome (SPRED1) Sequencing and Deletion/Duplication
INDICATION FOR TEST - Confirm Diagnosis

RESULT
No pathogenic variants were detected in the SPRED1 gene.

INTERPRETATION
No pathogenic variants were detected in the SPRED1 gene by sequencing the coding regions and intron/exon boundaries and by deletion/duplication analysis. This result significantly decreases but does not exclude a diagnosis of Legius 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. Genetic consultation is recommended.

COMMENTS
Reference Sequence: GenBank # NM_152594.2 (SPRED1)
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 [redacted]

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

BACKGROUND INFORMATION: Legius Syndrome (SPRED1) Sequencing and Deletion/Duplication

CHARACTERISTICS: Cafe au lait spots, axillary and inguinal freckling, learning disabilities and macrocephaly. Neurofibromas, lisch nodules and CNS tumors are not observed.
INCIDENCE: Unknown; may represent 0.5 percent of neurofibromatosis type 1 diagnoses or 8 percent of those with isolated cafe au lait spots.
INHERITANCE: Autosomal dominant.
CAUSE: Pathogenic SPRED1 gene mutations.
CLINICAL SENSITIVITY: Unknown.
METHODOLOGY: Bidirectional sequencing and multiplex ligation-dependent probe amplification (MLPA) of the entire coding region and intron-exon boundaries of the SPRED1 gene.
ANALYTICAL SENSITIVITY AND SPECIFICITY: 99 percent.
LIMITATIONS: Diagnostic errors can occur due to rare sequence variations. Regulatory region mutations and deep intronic mutations will not be detected. Deletion/duplication breakpoints will not be determined.

Counseling and informed consent are recommended for genetic testing. Consent forms are available online at www.aruplab.com.

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

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
Legius Syndrome (SPRED1)Seq, DelDup Spcm	17-037-401015	2/3/2017 1:04:00 PM	2/7/2017 4:51:32 AM	3/1/2017 7:37:00 AM
Legius Syndrome (SPRED1)Seq, DelDup Int	17-037-401015	2/3/2017 1:04:00 PM	2/7/2017 4:51:32 AM	3/1/2017 7:37:00 AM

END OF CHART

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