ARUP LABORATORIES | www.aruplab.com
500 Chipeta Way, Salt Lake City, Utah 84108-1221
phone: (801) 583-2787, toll free: (800) 522-2787
Julio C. Delgado, MD, MS, Director of Laboratories

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

Physician: Doctor, Example

Patient: Patient, Example

DOB	[redacted]
Gender:	Male
Patient Identifiers:	01234567890ABCD, 012345
Visit Number (FIN):	01234567890ABCD
Collection Date:	00/00/0000 00:00

Peutz-Jeghers Syndrome (STK11) Sequencing and Deletion/Duplication

ARUP test code 2008398

PJS (STK11) Seq, DelDup Spcm	Whole Blood
PJS (STK11) Seq, DelDup Interp	Negative

TEST PERFORMED - 2008398
TEST DESCRIPTION - Peutz-Jeghers Syndrome (STK11) Sequencing and Deletion/Duplication
INDICATION FOR TEST - Confirm Diagnosis

RESULT
No pathogenic variants were detected in the STK11 gene.

INTERPRETATION
No pathogenic variants were detected in the STK11 gene by sequencing and deletion/duplication analysis. This result significantly decreases the likelihood, but does not exclude a diagnosis of Peutz-Jeghers 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. If there is suspicion for a hereditary gastrointestinal cancer syndrome, consideration should be given to ordering the Hereditary Gastrointestinal Cancer Panel, Sequencing and Deletion/Duplication (ARUP test code 2013449).

COMMENTS
Reference Sequence: GenBank # NM_000455.4 (STK11)
Nucleotide numbering begins at the "A" of the ATG initiation codon.
Likely benign and benign variants are not reported.

This result has been reviewed and approved by Steven Steinberg, Ph.D.

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

INTERPRETIVE INFORMATION: Peutz-Jeghers Synd (STK11) Seq, DelDup

Background Information for Peutz-Jeghers Syndrome (STK11) Sequencing and Deletion/Duplication
Characteristics: Hamartomatous polyps of the gastrointestinal tract and hyperpigmented macules around the buccal mucosa, eyes, nostrils, perianal area, and fingers. Epithelial malignancies including colorectal, gastric, pancreatic, breast, ovarian, sex cord tumors with annular tubules and adenoma malignum of cervix. Cumulative risk for any cancer is 17 percent by age 40, 31 percent by age 50, 60 percent by age 60 and 85 percent by age 70.
Inheritance: Autosomal dominant.
Cause: Pathogenic STK11 gene mutations.
Clinical Sensitivity: About 99 percent in individuals with a family history and about 91 percent in those without a family history.
Methodology: Bidirectional sequencing and multiplex ligation-dependent probe amplification (MLPA) of the entire coding region and intron-exon boundaries of the STK11 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. This assay is not designed to detect somatic variants associated with malignancy. Interpretation of this test result may be impacted if the patient has had an allogeneic stem cell transplantation.

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

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
PJS (STK11) Seq, DelDup Spcm	19-343-400173	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
PJS (STK11) Seq, DelDup Interp	19-343-400173	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