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	Unknown
Gender:	Unknown
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 — **Positive** *

Section 79-1 of New York State Civil Rights Law requires informed consent be obtained from patients (or their legal guardians) prior to pursuing genetic testing. These forms must be kept on file by the ordering physician. Consent forms for genetic testing are available at www.aruplab.com. Incidental findings are not reported unless clinically significant but are available upon request.

Result: ONE PATHOGENIC MUTATION DETECTED IN THE STK11 GENE

Pathogenic Mutation:
Nucleic Acid Change: c.179dupA; Heterozygous
Predicted effect: Frameshift

INTERPRETATION: One pathogenic STK11 gene mutation, c.179dupA, was detected by sequencing; no STK11 mutations were detected by deletion/duplication analysis. This mutation has been previously published in a patient diagnosed with Peutz-Jeghers syndrome (see reference). This result is consistent with a diagnosis of Peutz-Jeghers syndrome; clinical manifestations are variable and age-dependent. This individuals offspring have a 50 percent chance of inheriting the causative mutation.

RECOMMENDATIONS: Genetic consultation is indicated, including a discussion of medical screening and management. At-risk family members should be offered a clinical evaluation for Peutz-Jeghers syndrome. If it is unclear whether or not they are affected, targeted testing for the identified mutation should be considered (Familial Mutation, Targeted Sequencing, ARUP test code 2001961).

REFERENCE:
Aretz et al. (2005) High proportion of large genomic STK11 deletions in Peutz-Jeghers syndrome. Hum Mutat. 26(6):513-9.

REFERENCE SEQUENCE: GenBank # NM_000455.4
Nucleotide numbering begins at the A of the ATG initiation codon

This result has been reviewed and approved by Yuan Ji, 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	20-091-101487	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
PJS (STK11) Seq, DelDup Interp	20-091-101487	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