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

Physician: Doctor, Example

[redacted]

Li-Fraumeni (TP53) Sequencing and Deletion/Duplication

ARUP test code 2009313

Li-Fraumeni (TP53) Seq, DelDup Spcm	Whole Blood
Li-Fraumeni (TP53) Seq, DelDup Interp	Negative

TEST PERFORMED - 2009313
TEST DESCRIPTION - Li-Fraumeni (TP53) Sequencing and Deletion/Duplication
INDICATION FOR TEST - Confirm Diagnosis

RESULT
No pathogenic variants were detected in the TP53 gene.

INTERPRETATION
No pathogenic variants were detected in the TP53 gene by sequencing all coding regions and intron-exon boundaries or by deletion/duplication analysis. This result significantly decreases the probability of, but does not exclude, a diagnosis of Li-Fraumeni 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_ 000546.5 (TP53)
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 [redacted]

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

BACKGROUND INFORMATION: Li-Fraumeni (TP53) Sequencing and Deletion/Duplication
CHARACTERISTICS: Predisposition for developing early-onset and multiple primary cancers, particularly soft tissue and bone sarcomas, adrenocortical carcinoma, brain tumors, premenopausal breast cancer, and other malignancies.
PREFALENCE: 1 in 5,000 - 1 in 20,000.
INHERITANCE: Autosomal dominant.
PENETRANCE: Approximately 50 percent by age 30 years and 90 percent by age 60 years.
CAUSE: Pathogenic germline mutations in the TP53 gene.
CLINICAL SENSITIVITY: 80 percent for individuals meeting classic Li-Fraumeni syndrome (LFS) criteria.
METHODOLGY: Bidirectional sequencing of all coding regions and intron-exon boundaries of the TP53 gene; Multiplex Ligation-dependent Probe Amplification (MLPA) to detect large TP53 deletions/duplications.
ANALYTICAL SENSITIVITY AND SPECIFICITY: Greater than 95 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.

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
Li-Fraumeni (TP53) Seq, DelDup Spcm	20-204-400922	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Li-Fraumeni (TP53) Seq, DelDup Interp	20-204-400922	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