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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	9/10/2013
Gender:	Male
Patient Identifiers:	01234567890ABCD, 012345
Visit Number (FIN):	01234567890ABCD
Collection Date:	01/01/2017 12:34

PTEN-Related Disorders (PTEN) Sequencing

ARUP test code 2002722

PTEN FGS Specimen	Whole Blood

PTEN-Related Disorders Interpretation

Negative

TEST PERFORMED - 2002722
TEST DESCRIPTION - PTEN-Related Disorders (PTEN) Sequencing
INDICATION FOR TEST - Not Provided

RESULT
No pathogenic variants were detected in the PTEN gene.

INTERPRETATION
No pathogenic variants were detected in the PTEN gene by sequencing the coding region, intron/exon boundaries and promoter. However, a diagnosis of PTEN hamartoma tumor syndrome (PHTS) has not been excluded, since up to 10 percent of individuals with Bannayan-Riley-Ruvalcaba syndrome (BRRS) and rare individuals with Cowden syndrome (CS) carry large gene deletions not detectable by sequence analysis.

RECOMMENDATIONS
Medical screening and management should rely on clinical findings and family history. PTEN deletion/duplication analysis may be considered (PTEN Deletion/Duplication; ARUP test code 2002726). Genetic consultation is recommended.

COMMENTS
Reference Sequence: GenBank # NM_000314.4 (PTEN)
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 Steven Steinberg, Ph.D.

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

BACKGROUND INFORMATION: PTEN-Related Disorders (PTEN) Sequencing

CHARACTERISTICS OF PTEN HAMARTOMA TUMOR SYNDROME (PHTS):
Clinical findings are highly variable and include Cowden syndrome (CS), Bannayan-Riley-Ruvalcaba syndrome(BRRS), Proteus syndrome (PS) and Proteus-like syndrome (PSL).
CS: Multiple hamartoma syndrome with increased risk for malignant and benign tumors of the breast, thyroid and endometrium. Other associated findings include macrocephaly and mucocutaneous lesions (facial trichilemmomas, palmoplantar keratoses and papillomatous papules).
BRRS: Characterized by macrocephaly, intestinal hamartomatous polyposis, lipomas, hemangiomas and pigmented macules of the glans penis.
PS: A progressive disorder demonstrating mosaic distribution of associated lesions. Findings include hamartomatous tissue overgrowth, hyperostoses, connective tissue and epidermal nevi, dysregulated adipose tissue, vascular malformations and other congenital malformations.
PSL: Describes individuals with significant features of PS who do not meet clinical diagnostic criteria for PS.
INCIDENCE: At least 1 in 200,000 for CS; PS is rare with approximately 120 reported cases; unknown for other PTEN-associated conditions.
INHERITANCE: Autosomal dominant. All mutations causing PS and 50-90 percent causing CS are de novo.
CAUSE: Pathogenic PTEN gene mutations.
PENETRANCE: 99 percent by 30 years of age for CS.
CLINICAL SENSITIVITY: 80 percent for CS, 60 percent for BRRS, 50 percent for PSL and 20 percent for PS.
METHODOLOGY: Bidirectional sequencing of the PTEN promoter, coding region and intron-exon boundaries.
ANALYTICAL SENSITIVITY AND SPECIFICITY: 99 percent.
LIMITATIONS: Diagnostic errors can occur due to rare sequence variations. Some regulatory region mutations, deep intronic mutations, and large deletion/duplications will not be detected.

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

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
PTEN FGS Specimen	18-106-400869	4/13/2018 2:00:00 PM	4/17/2018 7:19:41 AM	4/25/2018 2:08:00 PM
PTEN-Related Disorders Interpretation	18-106-400869	4/13/2018 2:00:00 PM	4/17/2018 7:19:41 AM	4/25/2018 2:08:00 PM

END OF CHART

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