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

Multiple Endocrine Neoplasia Type 1 (MEN1) Sequencing and Deletion/Duplication

ARUP test code 2005360

MEN Type 1 (MEN1) Seq, Del/Dup Specimen	Whole Blood

MEN Type 1 (MEN1) Seq, Del/Dup Interp

Negative

TEST PERFORMED - 2005360
TEST DESCRIPTION - Multiple Endocrine Neoplasia Type 1 (MEN1) Sequencing and Deletion/Duplication
INDICATION FOR TEST - Confirm Diagnosis

RESULT
No pathogenic variants were detected in the MEN1 gene.

INTERPRETATION
No pathogenic variants were detected in the MEN1 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 multiple endocrine neoplasia type 1. 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. If there is suspicion for a hereditary cancer syndrome, consideration should be given to ordering the Hereditary Cancer Panel, Sequencing and Deletion/Duplication (ARUP test code 2012032). Genetic consultation is recommended.

COMMENTS
Reference Sequence: GenBank # NM_130799.2 (MEN1)
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

BACKGROUND INFORMATION: Multiple Endocrine Neoplasia Type 1 (MEN1) Sequencing and Deletion/Duplication
CHARACTERISTICS: Multiple Endocrine Neoplasia Type 1 (MEN1) syndrome can include multiple endocrine and non-endocrine tumors. Common MEN1-related endocrine tumors include parathyroid (90-95 percent), pancreatic islets (30-80 percent), and pituitary (15-90 percent). Non-endocrine tumors include facial angiofibroma, collagenoma, lipoma, meningioma, ependymoma, and leiomyoma. Primary hyperparathyroidism is the most common and often the first manifestation of MEN1. High mortality rates occur in persons with gastrinoma and carcinoid tumors.
INCIDENCE: Approximately 1 in 30,000.
INHERITANCE: Autosomal dominant.
PENETRANCE: Approximately 50 percent by age 20 and 95 percent by age 40.
CAUSE: Pathogenic MEN1 gene mutations.
CLINICAL SENSITIVITY: Approaches 94 percent.
METHODOLOGY: Bidirectional sequencing of the entire coding region and intron-exon boundaries of the MEN1 gene. Multiplex ligation-dependent probe amplification (MLPA) to detect large MEN1 coding region deletions/duplications.
ANALYTICAL SENSITIVITY AND SPECIFICITY: Approximately 98 percent.
LIMITATIONS: Diagnostic errors can occur due to rare sequence variations. Regulatory region mutations and deep intronic mutations will not be detected. The breakpoints of large deletions/duplications will not be detected. Mutations in genes other than MEN1 are not evaluated. 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
MEN Type 1 (MEN1) Seq, Del/Dup Specimen	20-015-403386	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
MEN Type 1 (MEN1) Seq, Del/Dup Interp	20-015-403386	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