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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	Unknown
Gender:	Unknown
Patient Identifiers:	01234567890ABCD, 012345
Visit Number (FIN):	01234567890ABCD
Collection Date:	00/00/0000 00:00

Inherited Insulin Resistance Syndromes (INSR) Sequencing

ARUP test code 2006274

INSR Sequencing Specimen	Blood
INSR Sequencing Interpretation	Negative

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TEST PERFORMED - 2006274
TEST DESCRIPTION - Inherited Insulin Resistance Syndrome (INSR)
Sequencing
INDICATION FOR TEST - Not Provided

RESULT
No pathogenic variants were detected in the INSR gene.

INTERPRETATION
No pathogenic variants were detected in the coding region or
intron/exon boundaries of the INSR gene by sequencing. This
reduces the chance that this individual is affected with or a
carrier of an inherited insulin resistance syndrome.

RECOMMENDATIONS
Diagnosis of inherited insulin resistance syndromes should rely
on clinical symptoms and biochemical/functional assay results.
Medical management should rely on clinical and biochemical
findings.

COMMENTS
Reference Sequence: GenBank # NM_000208.2 (INSR)
Nucleotide numbering begins at the "A" of the ATG initiation
codon.
Likely benign and benign variants are not included in this
report.

This result has been reviewed and approved by Rong Mao, M.D.
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H=High, L=Low, *=Abnormal, C=Critical

BACKGROUND INFORMATION: Insulin Resistance Conditions (INSR) Sequencing:

CHARACTERISTICS: Extreme insulin resistance is characterized by abnormal glucose homeostasis and hyperinsulinemia, leading eventually to ketoacidosis. Donohue syndrome (leprechaunism), Rabson-Mendenhall syndrome, and Type A insulin resistance are all caused by INSR gene mutations, although severity and survival varies greatly among conditions. Symptoms may include intrauterine growth restriction, failure to thrive after birth, characteristic dysmorphic features, lack of subcutaneous fat, acanthosis nigricans, enlargement of genitalia in males and females, cystic ovaries and amenorrhea in females, premature and dysplastic dentition, and pineal hyperplasia.
INCIDENCE: Unknown; rare.
INHERITANCE: Autosomal recessive (Donohue and Rabson-Mendenhall syndromes). Type A insulin resistance can be autosomal recessive or dominant.
CAUSE: Pathogenic INSR gene mutations.
CLINICAL SENSITIVITY: Predicted to be greater than 90 percent in individuals with a clinical diagnosis.
METHODOLOGY: Bidirectional sequencing of the entire coding region and intron/exon boundaries of the INSR gene.
ANALYTICAL SENSITIVITY AND SPECIFICITY: 99 percent.
LIMITATIONS: Diagnostic errors can occur due to rare sequence variations. Regulatory region mutations, deep intronic mutations, and large deletions/duplications will not be detected. Mutations in genes other than INSR are not evaluated.

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
INSR Sequencing Specimen	20-056-112512	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
INSR Sequencing Interpretation	20-056-112512	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