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Client: ARUP Example Report Only
500 Chipeta Way
Salt Lake City, UT 84108
UNITED STATES

Physician: TEST, DR

Patient: Test, GBA2

DOB
Gender: Female
Patient Identifiers: 18262
Visit Number (FIN): 18483
Collection Date: 8/22/2019 08:55

Gaucher Disease (GBA) Sequencing

ARUP test code 3001648

GBA FGS- Specimen	Whole Blood

GBA FGS Interpretation

Negative

TEST PERFORMED - 3001648
TEST DESCRIPTION - Gaucher Disease (GBA) Sequencing
INDICATION FOR TEST - Confirm Diagnosis

RESULT
No pathogenic variants were detected in the GBA gene.

INTERPRETATION
No pathogenic variants were detected in the GBA gene using bidirectional sequencing of all coding regions and intron/exon boundaries. This result significantly reduces the likelihood that this individual is affected with, or a carrier of, Gaucher disease. Please refer to the background information included in this report for the clinical sensitivity and limitations of this test.

RECOMMENDATIONS
This result should be combined with the patients clinical findings and glucocerebrosidase activity level for optimal interpretation.

COMMENTS
Reference Sequence: GenBank # NM_001005741.2 (GBA)
Nucleotide numbering begins at the "A" of the ATG initiation codon.

This result has been reviewed and approved by Rong Mao, M.D.

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

BACKGROUND INFORMATION: Gaucher Disease (GBA) Sequencing

CHARACTERISTICS: Gaucher disease (GD) is a lysosomal storage disorder with phenotypes ranging from perinatal lethality to lack of symptoms. There are three GD subtypes. Type 1 GD manifests with bone disease, hepatosplenomegaly, anemia, thrombocytopenia, and lung disease but no central nervous system (CNS) involvement. Type 2 GD exhibits CNS symptoms before age 2 and rapidly progresses resulting in death by age 4. Type 3 GD presents as early as age 2 with CNS symptoms that slowly progress resulting in death during the third or fourth decade.
INCIDENCE: 1 in 900 Ashkenazi Jewish individuals; approximately 1 in 57,000 to 1 in 75,000 in general population.
INHERITENCE: Autosomal recessive.
CAUSE: Two pathogenic GBA variants on opposite chromosomes.
CLINICAL SENSITIVITY: 99 percent.
METHODOLOGY: Long range PCR followed by bidirectional sequencing of all coding regions and intron-exon boundaries of the GBA gene.
ANALYTICAL SENSITIVITY AND SPECIFICITY: approximately 99 percent.
LIMITATIONS: Diagnostic errors can occur due to rare sequence variations. Regulatory region variants, deep intronic variants, large deletions/duplications/insertions, gene conversion and complex gene events may 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
GBA FGS- Specimen	19-234-102575	8/22/2019 8:55:00 AM	8/22/2019 10:47:53 AM	8/22/2019 12:22:00 PM
GBA FGS Interpretation	19-234-102575	8/22/2019 8:55:00 AM	8/22/2019 10:47:53 AM	8/22/2019 12:22:00 PM

END OF CHART

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