ARUP LABORATORIES
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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	9/27/1970
Gender:	Male
Patient Identifiers:	01234567890ABCD, 012345
Visit Number (FIN):	01234567890ABCD
Collection Date:	00/00/0000 00:00

Gaucher Disease (GBA) Sequencing

ARUP test code 3001648

GBA FGS- Specimen	Whole Blood
GBA FGS Interpretation	Negative 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 patient's 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. 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: 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.

This test was developed and its performance characteristics determined by ARUP Laboratories. It has not been cleared or approved by the US Food and Drug Administration. This test was performed in a CLIA certified laboratory and is intended for clinical purposes.

Counseling and informed consent are recommended for genetic testing. Consent forms are available online.

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
GBA FGS- Specimen	24-030-400369	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
GBA FGS Interpretation	24-030-400369	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