ARUP LABORATORIES
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Jonathan R. Genzen, MD, PhD, Chief Medical Officer

Client: Example Client ABC123
123 Test Drive
Salt Lake City, UT 84108
UNITED STATES

Physician: Doctor, Example

Patient: Patient, Example

DOB	12/5/1941
Gender:	Male
Patient Identifiers:	01234567890ABCD, 012345
Visit Number (FIN):	01234567890ABCD
Collection Date:	00/00/0000 00:00

Gaucher Disease (GBA), 8 Variants

ARUP test code 0051438

Gaucher Disease (GBA), Specimen	Whole Blood	
Gaucher Disease (GBA), Allele 1	c.1226A>G	*
Gaucher Disease (GBA), Allele 2	c.1226A>G	*
Gaucher Disease (GBA), Interpretation	See Note	

Indication for testing: Carrier screening or diagnostic testing for Gaucher disease.

Positive: Two copies of the mild pathogenic variant, p.N409S (c.1226A>G), were detected in the GBA gene. Individuals homozygous for this variant are predicted to be affected with Gaucher disease type 1; however, some affected individuals may be asymptomatic. Genetic consultation is indicated. This individual's reproductive partner should be offered screening for the disorder. At-risk family members should be offered testing to determine carrier status for the identified variant. This result has been reviewed and approved by [redacted]

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

BACKGROUND INFORMATION: Gaucher Disease (GBA), 8 Variants

CHARACTERISTICS: Gaucher disease affects lysosomal storage and has extreme symptom variability, ranging from perinatal lethality to asymptomatic individuals. Three Gaucher subtypes have been identified based on symptom characteristics. Type 1 is characterized by bone disease, hepatosplenomegaly, anemia, thrombocytopenia, and lung disease. Individuals with Type 1 disease do not have primary central nervous system (CNS) involvement. Type 2 is characterized by CNS symptoms displaying before age 2, which progresses rapidly resulting in death by age 4. In Type 3 disease, individuals may present as early as age 2 with display CNS symptoms. However, Type 3 disease progresses slowly, usually resulting in death during the third or fourth decade of life.
INCIDENCE: 1 in 900 in Ashkenazi Jewish individuals.
INHERITANCE: Autosomal recessive.
CAUSE: GBA pathogenic variants.
VARIANTS TESTED: c.115+1G>A, p.L29Afs (c.84dupG), p.N409S (c.1226A>G), c.1263_1317del55, p.V433L (c.1297G>T), p.D448H (c.1342G>C), p.L483P (c.1448T>C), and p.R535H (c.1604G>A).
CLINICAL SENSITIVITY: 90 percent in Ashkenazi Jewish individuals; 55 percent in other ethnicities.
METHODOLOGY: Polymerase chain reaction (PCR) and fluorescence monitoring.
ANALYTICAL SENSITIVITY AND SPECIFICITY: Greater than 99 percent.
LIMITATIONS: Variants other than those tested will not be detected. Diagnostic errors can occur due to rare sequence variations.

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
Gaucher Disease (GBA), Specimen	23-249-130758	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Gaucher Disease (GBA), Allele 1	23-249-130758	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Gaucher Disease (GBA), Allele 2	23-249-130758	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Gaucher Disease (GBA), Interpretation	23-249-130758	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