ARUP LABORATORIES | www.aruplab.com
500 Chipeta Way, Salt Lake City, Utah 84108-1221
phone: (801) 583-2787, toll free: (800) 522-2787
Julio C. Delgado, MD, MS, Director of Laboratories

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

Physician: Doctor, Example

Patient: Patient, Example

DOB	4/18/1951
Gender:	Female
Patient Identifiers:	01234567890ABCD, 012345
Visit Number (FIN):	01234567890ABCD
Collection Date:	01/01/2017 12:34

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	Negative
Gaucher Disease (GBA), Interpretation	See Note

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

Positive: One mild pathogenic variant, p.N409S (c.1226A>G), was detected in the GBA gene; therefore, this individual is at least a carrier of Gaucher disease type 1. This individuals 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. Genetic counseling is recommended.

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

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.

Test developed and characteristics determined by ARUP Laboratories. See Compliance Statement C: aruplab.com/CS

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
Gaucher Disease (GBA), Specimen	18-011-401577	1/11/2018 9:58:00 AM	1/12/2018 6:54:07 PM	1/16/2018 3:38:00 PM
Gaucher Disease (GBA), Allele 1	18-011-401577	1/11/2018 9:58:00 AM	1/12/2018 6:54:07 PM	1/16/2018 3:38:00 PM
Gaucher Disease (GBA), Allele 2	18-011-401577	1/11/2018 9:58:00 AM	1/12/2018 6:54:07 PM	1/16/2018 3:38:00 PM
Gaucher Disease (GBA), Interpretation	18-011-401577	1/11/2018 9:58:00 AM	1/12/2018 6:54:07 PM	1/16/2018 3:38:00 PM

END OF CHART

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