

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

Physician: Doctor, Example

Patient: Patient, Example

DOB: 8/22/1986
Gender: Male
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 Negative

Gaucher Disease (GBA), Allele 2 Negative

Gaucher Disease (GBA), Interpretation

See Note

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

Negative: This sample is negative for the eight pathogenic variants tested in the GBA gene. If this is an asymptomatic individual of Ashkenazi Jewish descent, his/her risk of being a carrier of Gaucher disease is reduced from 1 in 15 to approximately 1 in 140.

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	18-142-116525	5/22/2018 1:39:00 PM	5/23/2018 4:22:27 PM	5/25/2018 6:07:00 PM
Gaucher Disease (GBA), Allele 1	18-142-116525	5/22/2018 1:39:00 PM	5/23/2018 4:22:27 PM	5/25/2018 6:07:00 PM
Gaucher Disease (GBA), Allele 2	18-142-116525	5/22/2018 1:39:00 PM	5/23/2018 4:22:27 PM	5/25/2018 6:07:00 PM
Gaucher Disease (GBA), Interpretation	18-142-116525	5/22/2018 1:39:00 PM	5/23/2018 4:22:27 PM	5/25/2018 6:07:00 PM

END OF CHART

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