

New Test **3001648** **Gaucher Disease (GBA) Sequencing** **GBA FGS**



Additional Technical Information



Patient History for Gaucher Disease Sequencing



Out of Pocket Estimator

Methodology: Polymerase Chain Reaction/Sequencing
Performed: Sun-Sat
Reported: 2-3 weeks

Specimen Required: Collect: Lavender (K₂ or K₃ EDTA) or Pink (K₂ EDTA). Also acceptable: Yellow (ACD Solution A or B).
Specimen Preparation: Transport 3 mL whole blood. (Min: 1 mL)
Storage/Transport Temperature: Refrigerated.
Stability (collection to initiation of testing): Ambient: 1 week; Refrigerated: 1 month; Frozen: 6 months

Reference Interval:

Background information for 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.

Inheritance: 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.

Interpretive Data: See Compliance Statement C: www.aruplab.com/CS

CPT Code(s): 81479

New York DOH approval pending. Call for status update.

HOTLINE NOTE: Refer to the Test Mix Addendum for interface build information.