

TEST CHANGE

Gaucher Disease (GBA) Sequencing

3001648, GBA FGS

Specimen Requirements:

Patient Preparation:

Collect: Lavender (K2 or K3EDTA), pink (K2EDTA), or yellow (ACD solution A or B).
New York State Clients: Lavender (EDTA)

Specimen Preparation: Transport 3 mL whole blood. (Min: 1 mL)
New York State Clients: 5 mL (Min: 2 mL)

Transport Temperature: Refrigerated.
New York State Clients: Ambient

Unacceptable Conditions:

Remarks:

Stability: Ambient: 1 week; Refrigerated: 1 month; Frozen: Unacceptable
New York State Clients: Ambient: 4 days 24 hours; Refrigerated: Unacceptable 1 week; Frozen: Unacceptable

Methodology: Polymerase Chain Reaction (PCR) / Sequencing

Note:

When testing cord blood specimens, the presence of maternal cell contamination (MCC) is possible, which may impact result interpretation. If clinically warranted, testing for MCC is available, at a charge, through ARUP Laboratories.

CPT Codes: 81479

New York DOH Approval Status: Specimens from New York clients will be sent out to a New York DOH approved laboratory, if possible.

Interpretive Data:

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

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variants, deep intronic variants, large deletions/duplications/insertions, gene conversion, and complex gene events may not be detected.

Reference Interval:

Refer to report
