TEST CHANGE

Gaucher Disease (GBA) Sequencing

3001648, GBA FGS

Specimen Requirements:	
Patient Preparation:	
Collect:	Lavender (K2EDTA or K3EDTA), pink (K2EDTA), ~~K3 EDTA)~~ or yellow~~Pink (K2 EDTA). Also acceptable: Yellow~~ (ACD s~~S~~olution A or B).
Specimen Preparation:	Transport 3 mL whole blood. (Min: 1 mL)
Transport Temperature:	Refrigerated.
Unacceptable Conditions:	
Remarks:	
Stability:	Ambient: 1 week; Refrigerated: 1 month; Frozen: 6 months
Methodology:	Polymerase Chain Reaction/Sequencing
Performed:	Varies~~Sun-Sat~~
Reported:	2-3 weeks
Note:	
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 variants, deep intronic variants, large deletions/duplications/insertions, gene conversion and complex gene events may not be detected.

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.

Reference Interval: