HOTLINE: Effective November 18, 2019

New Test **3001957** **Gamma Globin (*HBG1* and *HBG2*) Sequencing** **HBG FGS**

Additional Technical Information

Out of Pocket Estimator

Patient History for *HBG* Testing

Methodology: Polymerase Chain Reaction/Sequencing
Performed: Varies
Reported: Within 2 weeks

Specimen Required: Collect: Lavender (K_2EDTA), Pink (K_2EDTA), or 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: By report

Interpretive Data: Background information for Gamma Globin (*HBG1* and *HBG2*) Sequencing:
Characteristics: Variants in the gamma globin genes, *HBG1* and *HBG2*, may occasionally result in either a quantitative defect (gamma thalassemia or nondeletional hereditary persistence of fetal hemoglobin) or a qualitative abnormality (gamma variant). Gamma variants resulting in unstable, high- and low-oxygen affinity or M hemoglobin variants may result in hemolytic anemia/hyperbilirubinemia, erythrocytosis/cyanosis, or methemoglobinemia in neonates, respectively. Clinical symptoms related to gamma globin variants commonly resolve after the first six months of life given the switch from fetal hemoglobin expression to adult hemoglobin expression.
Incidence: Unknown.
Inheritance: Autosomal dominant.
Cause: Pathogenic germline variants in *HBG1* or *HBG2*.
Clinical Sensitivity: Unknown. Gamma globin variants are a rare cause of neonatal hemolytic anemia, cyanosis, erythrocytosis, or methemoglobinemia.
Methodology: Long range PCR followed by nested PCR and bidirectional sequencing of all coding regions, intron/exon boundaries, proximal promoters, and 5' and 3' untranslated regions of the *HBG1* and *HBG2* genes.
Analytical Sensitivity and Specificity: 99 percent.
Limitations: Diagnostic errors can occur due to rare sequence variations or repeat element insertions. Large deletions/duplications, distal regulatory region variants, deep intronic variants, and hybrid gene events will not be detected.

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.