TEST CHANGE

Gamma Globin (HBG1 and HBG2) Sequencing

3001957, HBG FGS

Specimen Requirements:	
Patient Preparation:	
Collect:	Lavender (K2EDTA), Pink (K2EDTA), or Yellow (ACD Solution A or B).
Specimen Preparation:	Transport 3 mL whole blood. (Min: 1 mL)
Transport Temperature:	Refrigerated.
Unacceptable Conditions:	N/A
Remarks:	N/A
Stability:	Ambient: 1 week; Refrigerated: 1 month; Frozen: Unacceptable~~6 months~~
Methodology:	Polymerase Chain Reaction/Sequencing
Performed:	Varies
Reported:	~~Within~~ 2-3 weeks
Note:	
CPT Codes:	81479
New York DOH Approval Status:	This test is New York DOH approved.

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, and 5' proximal promoters 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.

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:

By report