

Interpretive Data:**Background Information:** Beta Globin (*HBB*) Sequencing**Characteristics:** Structural hemoglobinopathies or thalassemias (insufficient or absent beta-chain production).**Incidence:** Varies with ethnicity.**Inheritance:** Usually autosomal recessive, infrequently autosomal dominant.**Cause:** Pathogenic variants in the *HBB* gene.**Clinical Sensitivity:** Up to 97 percent, depending upon ethnicity.**Methodology:** Bidirectional sequencing of the *HBB* coding regions, intron-exon boundaries, 5' proximal promoter and untranslated region, 3' polyadenylation signal, and intronic variants c.93-21 (IVS-I-110), c.316-197 (IVS-II-654), c.316-146 (IVS-II-705), c.316-106 (IVS-II-745), and c.316-86_316-85 (IVS-II-765 L1).**Analytical sensitivity:** 99 percent.**Limitations:** Diagnostic errors can occur due to rare sequence variations. Large deletions and variants in distal regulatory elements are not detected.See Compliance Statement C: www.aruplab.com/CS**HOTLINE NOTE:** Remove information found in the Reference Interval field.