

TEST CHANGE

Alpha Globin (*HBA1* and *HBA2*) Sequencing and Deletion/Duplication, Fetal

3019566, HBA FGA FE

Specimen Requirements:

Patient Preparation:

Collect:

Fetal Specimen: ~~Cultured Two T-25 flasks at 90% confluent of cultured amniocytes~~ OR cultured chorionic villi.

~~villus sampling (CVS).~~

~~AND Maternal Whole Blood Specimen: Refer to Maternal Cell Contamination, Maternal Specimen (0050608) for maternal specimen requirements. Lavender (EDTA), pink (K2EDTA), or yellow (ACD solution A or B).~~

Specimen Preparation:

Cultured Amniocytes or Cultured CVS: Fill flasks with culture media. Transport two T-25 flasks ~~of~~ at 90 percent confluent ~~of~~ cultured amniocytes or two T-25 flasks of 90% cultured chorionic villi sampling (CVS).

This assay is not performed on direct amniotic fluid or direct chorionic villi specimens. Clients submitting direct amniotic fluid and direct chorionic villi must add Cell Culture for Genetic Testing (3020627) to the initial order.

~~filled with culture media. Backup cultures must be retained at the client's institution until testing is complete. If ARUP receives cultured specimens a sample below the minimum confluence, Cell Culture for Genetic Testing (3020627) Cytogenetics Grow and Send (0040182) will be added on by ARUP for an additional fee. The client is responsible for maintaining backup cultures. charges will apply. If clients are unable to culture specimens, Cytogenetics Grow and Send should be added to initial order.~~

~~Maternal Whole Blood Specimen: Transport 3 mL whole blood. (Min: 1 mL).~~

Transport Temperature:

Cultured Amniocytes or Cultured CVS: CRITICAL ROOM TEMPERATURE. ~~Separate specimens must be submitted when multiple tests are ordered.~~ Must be received within 48 hours of shipment due to viability of cells.

~~Maternal specimen: Refrigerated~~

Unacceptable Conditions:

~~Frozen specimens~~

Remarks:

Patient history forms and informed consent documents are available by selecting the links above or by contacting ARUP Client Services. Counseling and informed consent are recommended for genetic testing. New York Clients: Informed consent is required with specimen submission.

Stability: Cultured Amniocytes or Cultured CVS: Room temperature: 48 hours; Refrigerated: Unacceptable; Frozen: Unacceptable
~~Maternal Whole Blood Specimen: Room temperature: 7 days; Refrigerated: 1 month; Frozen: Unacceptable~~

Methodology: Qualitative Multiplex Ligation-Dependent Probe Amplification (MLPA) / Qualitative Sequencing

Note: ~~Reported times are based on receiving the two T-25 flasks at 90 percent confluent. Cell culture time is independent of testing turnaround time. Maternal specimen is recommended for proper test interpretation. Order Maternal Cell Contamination.~~

CPT Codes: 81259; 81269; 81265

New York DOH Approval Status: This test is New York DOH approved.

Interpretive Data:

Refer to report

Reference Interval:

~~Refer to~~By report