

TEST CHANGE

Galactosemia (GALT) 9 Mutations, Fetal

0051270, GALTDNA FE

Specimen Requirements:

Patient Preparation:

Collect:

Fetal Specimen: Cultured amniocytes OR cultured chorionic villus sampling (CVS).
Maternal Specimen: Refer to Maternal Cell Contamination, Maternal Specimen (0050608) for maternal specimen requirements. Fetal: Cultured Amniocytes, Cultured CVS AND Maternal Whole Blood Specimen: Lavender (EDTA), pink (K2EDTA), or yellow (ACD solution A or B).

Specimen Preparation:

Transport: Two T-25 flasks of 80% confluent Cultured Amniocytes or Cultured CVS: Transfer cultured amniocytes OR Two T-25 flasks of 80% confluent cultured chorionic villus sampling (CVS).
Cultured amniocytes or or cultured CVS is required for testing. If submitting uncultured (direct) amniotic fluid or (direct) CVS and testing is desired on a cultured specimen, add Cell Culture for Genetic Testing (3020627). If transporting flasks, the client is responsible for maintaining backup to two T-25 flasks at 80 percent confluence. (Min: one T-25 flask at 80 percent confluence) Backup cultures must be retained at the client's institution, until testing is complete. If ARUP receives cultured fetal cells a sample below the minimum confluence, Cell Culture for Genetic Testing (3020627 Cytogenetics Grow and Send (ARUP test code 0040182) will be added on by ARUP, and additional charges will apply. If clients are unable to culture specimens, Cytogenetics Grow and Send should be added to the initial order.

Maternal Whole Blood Specimen: 2 mL whole blood (Min: 1 mL).

Transport Temperature:

Preferred transport temp: Cultured Amniocytes or Cultured CVS: CRITICAL ROOM TEMPERATURE. Must be received within 48 hours of collection due to viability of cells.
Maternal Whole Blood Specimen: Room temperature.
Preferred shipment: Within two days of collection or confluence.

Unacceptable Conditions:

Frozen specimens.

Remarks:

Counseling and informed consent are recommended for genetic testing. Consent forms are linked above.
New York Clients: informed consent is required with submission.

Stability: ~~Cultured Amniocytes or Cultured CVS: Room temperature: 2 days; Refrigerated: Unacceptable; Frozen: Unacceptable~~
~~Maternal Whole Blood Specimen: Room temperature: 1 week; Refrigerated: 1 month; Frozen: Unacceptable~~

Methodology: Polymerase Chain Reaction (PCR) / Single Nucleotide Extensions

Note: This test is offered to individuals with a known familial mutation(s).

CPT Codes: 81401; 81265 Fetal Cell Contamination (FCC)

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

Interpretive Data:

Refer to report.

Reference Interval:

Refer to report