

HOTLINE: Effective September 7, 2021

0051270 Galactosemia (GALT) 9 Mutations, Fetal

GALTDNA FE



Patient History for Fetal Molecular Testing



Additional Technical Information

Time Sensitive

Methodology: Polymerase Chain Reaction/Single Nucleotide Extension

Specimen Required: Collect: Cultured Amniocytes, Cultured CVS, or Amniotic fluid (direct)

AND Maternal Whole Blood Specimen: Lavender (EDTA), pink (K<sub>2</sub>EDTA), or yellow (ACD Solution A or B).

Specimen Preparation: Cultured Amniocytes or Cultured CVS: Transfer cultured amniocytes or cultured CVS to two T-25 flasks at 80% confluence. (Min: one T-25 flask at 80% confluence). Backup cultures must be retained at the client's institution until testing is complete. If the client is unable to culture amniocytes or CVS, this can be arranged by contacting ARUP Client Services at (800) 522-2787. Please contact an ARUP genetic counselor at (800) 242-2787 ext. 2141 prior to test submission.

Amniotic Fluid (direct): 10 milliliters

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

Storage/Transport Temperature: Cultured Amniocytes or Cultured CVS: CRITICAL ROOM TEMPERATURE. Must be

received within 48 hours of collection due to viability of cells.

Amniotic fluid (direct): Ship room temperature.

Maternal Whole Blood Specimen: Room temperature.

Remarks: Please contact an ARUP genetic counselor at (800) 242-2787 ext. 2141 prior to sample submission. Patient History Forms

are available on aruplab.com or by contacting ARUP Client Services at (800) 522-2787.

<u>Stability (collection to initiation of testing): Cultured Amniocytes or Cultured CVS: Room temperature: 48 hours; Refrigerated:</u>

Unacceptable; Frozen: Unacceptable

Amniotic Fluid (direct): Room temperature: 48 hours; Refrigerated: 72 hours; Frozen: Unacceptable

Maternal Whole Blood Specimen: Room temperature: 7 days; Refrigerated: 1 month; Frozen: Unacceptable