TEST CHANGE

Galactosemia (GALT) 9 Mutations, Fetal

0051270, GALTDNA FE

Specimen Requirements:	
Patient Preparation:	
Collect:	Fetal: Cultured Amniocytes, Cultured CVS~~, or Direct Amniotic Fluid (direct)~~ AND Maternal Whole Blood Specimen: Lavender (EDTA), pink (K2EDTA), 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 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 a sample below the minimum confluence, 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. ~~Direct Amniotic Fluid: 10 mL~~ Maternal Whole Blood Specimen: 2 mL whole blood (Min: 1 mL).
Transport Temperature:	Cultured Amniocytes or Cultured CVS: CRITICAL ROOM TEMPERATURE. Must be received within 48 hours of collection due to viability of cells. ~~Direct Amniotic Fluid: Ship room temperature.~~ Maternal Whole Blood Specimen: Room temperature
Unacceptable Conditions:	
Remarks:	
Stability:	Cultured Amniocytes or Cultured CVS: Room temperature: 48 hours; Refrigerated: Unacceptable; Frozen: Unacceptable ~~Direct Amniotic Fluid: Room temperature: 48 hours; Refrigerated: 72 hours; Frozen: Unacceptable~~ Maternal Whole Blood Specimen: Room temperature: 7 days; Refrigerated: 1 month; Frozen: Unacceptable
Methodology:	Polymerase Chain Reaction (PCR)~~)/~~Single Nucleotide Extensions
Performed:	Sun-Sat
Reported:	5-7 days
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. ~~Background Information for Galactosemia (GALT) 9 Mutations:~~
~~Characteristics: Affected infants present at 3-14 days old with poor feeding, vomiting, diarrhea, jaundice, lethargy progressing to coma, and abdominal distension with hepatomegaly usually followed by progressive liver failure. Patients with galactosemia are also at increased risk for E. coli or other Gram negative neonatal sepsis. Diagnosis is made by measuring GALT enzyme activity in red blood cells.~~
~~Incidence: Approximately 1 in 30,000 to 60,000 for classic galactosemia in Caucasians; varies in other populations.~~
~~Inheritance: Autosomal recessive.~~
~~Penetrance: 100 percent for severe GALT mutations.~~
~~Cause: Mutations in the GALT gene.~~
~~Mutations Tested: Seven GALT gene mutations (Q188R, S135L, K285N, T138M, L195P, Y209C, and IVS2-2 A>G) and two variants (N314D and L218L).~~
~~Clinical Sensitivity: Approaches 80 percent in Caucasians but reduced in other ethnic groups.~~
~~Methodology: Polymerase chain reaction followed by single nucleotide extension (SNE) and capillary electrophoresis.~~
~~Analytical Sensitivity: 99 percent for mutations listed.~~
~~Limitations: GALT gene mutations, other than the 9 targeted, will not be detected. Diagnostic errors can occur due to rare sequence variations.~~

Reference Interval:

By report