

TEST CHANGE

Fragile X (*FMR1*) with Reflex to Methylation Analysis, Fetal

2009034, FX PCR FE

Specimen Requirements:

Patient Preparation:

Collect:

Fetal Specimen: Cultured amniocytes.
Amniocytes
~~AND Maternal Specimen: Refer to Maternal Cell Contamination, Maternal Specimen (0050608) for maternal specimen requirements. 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: Transfer cultured amniocytes. Cultured amniocytes are required for testing. If submitting uncultured (direct) amniotic fluid 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 initial order. Maternal Whole Blood Specimen: Transport 2 mL whole blood. (Min: 1 mL)~~

Transport Temperature:

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

Unacceptable Conditions:

Frozen specimens.

Remarks:

Methylation patterns may not be fully established in early gestation; thus, methylation testing performed on chorionic villus samples may not distinguish between premutation and full mutation alleles.

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: Room temperature: 2 days 48 hours;~~
~~Refrigerated: Unacceptable; Frozen: Unacceptable~~
~~Maternal Whole Blood Specimen: Room temperature: 7 days;~~
~~Refrigerated: 1 month; Frozen: Unacceptable~~

Methodology: Polymerase Chain Reaction (PCR) / Capillary Electrophoresis

Note: If a CGG repeat of 100 or greater is detected by PCR and capillary electrophoresis, methylation analysis will be added. Additional charges apply.

CPT Codes: 81243; 81265 Fetal Cell Contamination (FCC); if reflexed, add 81244

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

Interpretive Data:

Refer to report.

Phenotype	Number of CGG Repeats
Unaffected	<45
Intermediate	45-54
Premutation	55-200
Affected	>200

Reference Interval:

Refer to ~~By~~ report