

TEST CHANGE

Familial Targeted Sequencing, Fetal

3005869, FAM NGS FE

Specimen Requirements:

Patient Preparation:

Collect:

Fetal Specimen: Cultured amniocytes OR cultured chorionic villi.

Maternal Specimen: Refer to Maternal Cell Contamination, Maternal Specimen (0050608) for maternal specimen requirements. Submit fetal specimen and maternal whole blood specimen. To avoid delays due to inappropriate sample submission, contact ARUP's genetic counselors at 800-242-2787 ext. 2141 for specimen requirements prior to sending samples.

New York State Clients: ARUP cannot facilitate testing for New York patients. Please work directly with a New York approved laboratory.

Specimen Preparation:

Cultured Amniocytes or Cultured CVS: Fill flasks with culture media. Transport two T-25 flasks of 90 percent confluent 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.

If ARUP receives cultured specimens below the minimum confluence, Cell Culture for Genetic Testing (3020627) will be added by ARUP for an additional fee. The client is responsible for maintaining backup cultures.

Transport Temperature:

Cultured Amniocytes or Cultured CVS: CRITICAL ROOM TEMPERATURE. Must be received within 48 hours of shipment due to viability of cells.

Unacceptable Conditions:

Remarks:

Documentation of the familial gene variants from a relative's laboratory test report is required to perform testing. Testing will begin upon receipt of all necessary components, including a ~~clinical~~ ~~an original~~ laboratory report detailing the familial variant(s) to be tested. 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 ~~A maternal specimen is~~ recommended for ~~genetic testing.~~ ~~proper fetal test interpretation.~~ Order

~~Maternal Cell Contamination on the maternal specimen (ARUP test code 0050608).~~

Stability: Cultured Amniocytes or Cultured CVS: Room temperature: 48 hours; Refrigerated: Unacceptable; Frozen: Unacceptable

Methodology: Massively Parallel Sequencing

Note: Only the requested sequence variant(s) of interest and other incidentally detected pathogenic or likely pathogenic sequence variants related to the condition in the gene of interest will be reported. ~~Reported times are based on receiving the cultured fetal sample (at 90 percent confluency) and required documentation. Backup cultures must be retained at the client's institution until testing is complete. If the client is unable to culture the fetal sample, this can be arranged by contacting ARUP Client Services at 800-522-2787 prior to test submission. Cell culture time is independent of testing turnaround time. Maternal specimen is recommended for proper test interpretation. Order Maternal Cell Contamination, Maternal Specimen on the maternal specimen.~~

CPT Codes: 81403; 81265 Fetal Cell Contamination (FCC)

New York DOH Approval Status: Specimens from New York clients will be sent out to a New York DOH approved laboratory, if possible.

Interpretive Data:

Refer to report.

Reference Interval:

Refer toBy report