

TEST CHANGE

Achondroplasia (FGFR3) 2 Mutations, Fetal

0051265, AD PCR FE

Specimen Requirements:

Patient Preparation:

Collect:

Fetal Specimen: Amniotic fluid OR cultured amniocytes OR cultured chorionic villus sampling (CVS).

Maternal Specimen: Refer to Maternal Cell Contamination, Maternal Specimen (0050608) for maternal specimen requirements. Amniotic fluid OR cultured amniocytes OR cultured CVS: Two T-25 flasks at 80 percent confluency. AND maternal whole blood: Lavender (K2 or K3EDTA), pink (K2EDTA), or yellow (ACD solution A or B).

If the client is unable to culture, order test Cytogenetics Grow and Send (ARUP test code 0040182) in addition to this test and ARUP will culture upon receipt (culturing fees will apply). If you have any questions, contact ARUP's Genetics Processing at 800-522-2787 ext. 3301.

Specimen Preparation:

Amniotic fluid: Transport: 10 mL amniotic fluid in a sterile container (min: 5 mL); OR

Two T-25 flasks of 80% confluent cultured amniocytes OR Two T-25 flasks of 80% confluent cultured chorionic villus sampling (CVS).

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: Fill flasks with culture media. Backup cultures must be retained at the client's institution. If ARUP receives cultured fetal cells below minimum confluence, Cell Culture for Genetic Testing (3020627) will be added by ARUP. -until testing is complete. AND maternal whole blood: transport 2 mL whole blood (min: 1 mL).

Transport Temperature:

Preferred transport temp: Amniotic fluid, cultured amniocytes or cultured CVS: CRITICAL ROOM TEMPERATURE. Must be received within 2 days of shipment due to liability of cells. Maternal whole blood: Room temperature.

Preferred shipment: Within two days of collection or confluence.

Unacceptable Conditions:

Frozen specimens in glass collection tubes.

Remarks:

Patient History Form is available on the ARUP Web site or by contacting ARUP Client Services.

Counseling and informed consent are recommended for

genetic testing. Consent forms are linked above.
New York Clients: **i**nformed consent is required with
submission.

Stability: ~~Fetal Specimen~~-Room temperature: 2 days; Refrigerated: 2
days; Frozen: Unacceptable
~~Maternal whole blood: Room Temperature: 3 days;
Refrigerated: 1 week; Frozen: Unacceptable~~

Methodology: Polymerase Chain Reaction (PCR) / Fluorescence Monitoring /
Fragment Analysis

Note:

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