

0051265

Achondroplasia (*FGFR3*) 2 Mutations, Fetal

AD PCR FE

Performed: Varies
Reported: 2-7 days

Specimen Required: Collect: Fetal specimen: Amniotic fluid.

OR Cultured amniocytes: Two T-25 flasks at 80 percent confluency.

OR cultured CVS: Two T-25 flasks at 80 percent confluency.

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.

AND maternal whole blood specimen: Lavender (EDTA), pink (K₂EDTA), or yellow (ACD Solution A or B).

Specimen Preparation: **Amniotic fluid:** Transport 10 mL amniotic fluid in a sterile container. (Min: 5 mL).

Cultured amniocytes AND cultured CVS: Transport two T-25 flasks at 80 percent confluency filled with culture media. Backup cultures must be retained at the client's institution until testing is complete.

Maternal Whole Blood Specimen: Transport 3 mL whole blood. (Min: 1 mL)

Storage/Transport Temperature: **Amniotic fluid, cultured amniocytes and cultured CVS: CRITICAL ROOM TEMPERATURE.** Must be received within 48 hours of shipment due to lability of cells.

Maternal Whole Blood Specimen: Refrigerated.

Remarks: **Please contact an ARUP genetic counselor at 800-242-2787 x2141 prior to sample submission.** Patient History Form is available on the ARUP Web site or by contacting ARUP Client Services.

Unacceptable Conditions: Frozen specimens in glass collection tubes.

Stability (collection to initiation of testing): **Amniotic fluid, cultured amniocytes and cultured CVS:** Room Temperature: 48 hours; Refrigerated: Unacceptable; Frozen: Unacceptable

Maternal Whole Blood Specimen: Room Temperature: 72 hours; Refrigerated: 1 week; Frozen: 1 month

Interpretive Data:

Refer to report

This test was developed and its performance characteristics determined by ARUP Laboratories. It has not been cleared or approved by the U.S. Food and Drug Administration. This test was performed in a CLIA-certified laboratory and is intended for clinical purposes.

Counseling and informed consent are recommended for genetic testing. Consent forms are available online.