HOTLINE: Effective May 16, 2022

0051265 Achondroplasia (*FGFR3*) 2 Mutations, Fetal AD PCR FE

Specimen Required: Collect: **Fetal specimen**: **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 amniocytes or CVS, this can be arranged by contacting ARUP Client Services at (800) 522-2787.
AND maternal whole blood specimen: Lavender (EDTA), pink (K_2EDTA), or yellow (ACD Solution A or B).
Specimen Preparation: **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: **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): **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:

Background information for Achondroplasia (*FGFR3*) 2 Mutations, Fetal:
Characteristics: Short stature with disproportionately short arms and legs, a large head, usually normal life span and intelligence; increased risk for death in infancy from compression of spinal cord and/or upper airway obstruction.
Incidence: 1:25,000.
Inheritance: Autosomal dominant; 80 percent arise from de novo mutations.
Penetrance: 100 percent.
Cause: Pathogenic *FGFR3* gene mutation.
Clinical Sensitivity: Two mutations, c.1138G>A and c.1138G>C, in the *FGFR3* gene account for greater than 99 percent of cases.
Methodology: Polymerase chain reaction (PCR) and fluorescence monitoring.
Analytical Sensitivity and Specificity: Greater than 99 percent.
Limitations: Mutations other than c.1138G>A and c.1138G>C will not be detected. Diagnostic errors can occur due to rare sequence variations.

For quality assurance purposes, ARUP Laboratories will confirm the above result at no charge following delivery. Order Confirmation of Fetal Testing and include a copy of the original fetal report (or the mother's name and date of birth) with the test submission. Please contact an ARUP genetic counselor at (800) 242-2787 extension 2141 prior to specimen submission.

This test was developed and its performance characteristics determined by ARUP Laboratories. It has not been cleared or approved by the US 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.