ARUP LABORATORIES | aruplab.com
500 Chipeta Way, Salt Lake City, Utah 84108-1221
phone: 801-583-2787, toll free: 800-522-2787
Jonathan R. Genzen, MD, PhD, Chief Medical Officer

PATIENT REPORT

Patient Age/Sex: 31 years Female

Specimen Collected: 11-Sep-23 15:21

Achondroplasia (FGFR3) 2 Mutations Fetal | **Received: 11-Sep-23 15:23** **Report/Verified: 12-Sep-23 13:48**

Procedure	Result	Units	Reference Interval
Achondroplasia PCR Fetal Specimen	Cultured Amnio		
Achondroplasia PCR,Fetal Interp	**HTG1138A** * f1 i1		
Maternal Contamination Study Fetal Spec	Fetal Cells f2		
Maternal Contam Study,Maternal Spec	Whole Blood		

Result Footnote

f1: Achondroplasia PCR, Fetal Interp

Indication for testing: Rule out achondroplasia.
Heterozygous G1138A (also known as c.1138G>A): This sample is positive for one copy of the G1138A (also known as c.1138G>A) mutation in the fibroblast growth factor receptor (FGFR) 3 gene consistent with a diagnosis of achondroplasia.

This result has been reviewed and approved by [redacted]

f2: Maternal Contamination Study Fetal Spec

Single fetal genotype present; no maternal cells present. Fetal and maternal samples were tested using STR markers to rule out maternal cell contamination.

Test Information

i1: Achondroplasia PCR, Fetal Interp

BACKGROUND INFORMATION: 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: >99 percent
METHODOLOGY: Polymerase chain reaction (PCR) and fluorescent monitoring
ANALYTICAL SENSITIVITY AND SPECIFICITY: >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 variants or maternal cell contamination of the fetal specimen.

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.

Test Information

i1: Achondroplasia PCR, Fetal Interp

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

*=Abnormal, #=Corrected, C=Critical, f=Result Footnote, H-High, i-Test Information, L-Low, t-Interpretive Text, @=Performing lab