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

Client: ARUP Example Report Only
500 Chipeta Way
Salt Lake City, UT 84108-
USA

Provider: .108 -TEST,

Patient: **AD PCR FE, POSITIVE**
DOB:
Sex: Female
Patient Identifiers: 40650
Visit Number (FIN): 40975
Client Supplied ID:

Specimen Collected: 19-Sep-22 16:22

Achondroplasia (FGFR3) 2 Mutations Fetal | **Received: 19-Sep-22 16:38** **Report/Verified: 19-Sep-22 16:46**

Procedure	Result	Units	Reference Interval
Achondroplasia PCR Fetal Specimen	Amniotic fluid		
Achondroplasia PCR	**AD HT/HT** * f1 i1		
Maternal Contamination Study Fetal Spec	**Maternal Cells** * f2		
Maternal Contam Study,Maternal Spec	Whole Blood i2		

Result Footnote

f1: Achondroplasia PCR

Indication for testing: Rule out achondroplasia.
Compound Heterozygote: This sample is positive for one copy of both the G1138A (also known as c.1138G>A) and G1138C (also known as c.1138G>C) mutations in the fibroblast growth factor receptor (FGFR) 3 gene. This is associated with a lethal skeletal dysplasia with clinical findings more severe than achondroplasia.

This result has been reviewed and approved by Rong Mao, M.D.

f2: Maternal Contamination Study Fetal Spec

Single maternal genotype; no fetal cells present. Fetal and maternal samples were tested using STR markers to rule out maternal cell contamination. Results were identical to the maternal genotype indicating that no fetal cells were present in the sample. Please request an additional sample.

Test Information

i1: Achondroplasia PCR

BACKGROUND INFORMATION: Achondroplasia (FGFR3) 2 Mutations

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.

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

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Patient: **AD PCR FE, POSITIVE**
DOB:
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Test Information

i1: Achondroplasia PCR

METHODOLOGY: Polymerase chain reaction (PCR) and fluorescent 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.

i2: Maternal Contam Study, Maternal Spec

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.