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Tracy I. George, MD, Chief Medical Officer

Patient Report

Patient Age/Sex: 35 years Female

Specimen Collected: 08-Mar-22 10:41

Achondroplasia (FGFR3) 2 Mutations Fetal | **Received: 08-Mar-22 10:41** | **Report/Verified: 10-Mar-22 13:05**

Procedure	Result	Units	Reference Interval
Achondroplasia PCR Fetal Specimen	Whole Blood		
Achondroplasia PCR Maternal Contamination Study Fetal Spec	Negative [f1] [i1]		
	Fetal Cells [f2]		
Maternal Contam Study, Maternal Spec	Whole Blood [i2]		

Result Footnote

f1: Achondroplasia PCR

Indication for testing: Rule out achondroplasia.
Negative: The fibroblast growth factor receptor (FGFR) 3 gene mutations, G1138A (also known as c.1138G>A) and G1138C (also known as c.1138G>C), were not present. Therefore, this individual is unlikely to be affected with achondroplasia. Rarely, an affected person may have another mutation not detectable by this assay.

This result has been reviewed and approved by Yuan Ji, Ph.D.

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

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.
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

Test Information

i1: Achondroplasia PCR

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.