

500 Chipeta Way, Salt Lake City, Utah 84108-1221

phone: 801-583-2787, toll free: 800-522-2787

Tracy I. George, MD, Chief Medical Officer

Patient Age/Sex: 32 years Male

Specimen Collected: 08-Mar-22 10:39

Achondroplasia (FGFR3) 2 Mutations	Received: 08-Mar-22 10:39	Report/Verified: 10-Mar-22 12:58
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Procedure	Result	Units	Reference Interval
Achondroplasia PCR Specimen	Whole Blood		
Achondroplasia PCR	Negative ^{f1 i1}		

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.

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 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.

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

Unless otherwise indicated, testing performed at:**ARUP Laboratories**

500 Chipeta Way, Salt Lake City, UT 84108

Laboratory Director: Tracy I. George, MD

ARUP Accession: 22-067-900084**Report Request ID:** 15080545**Printed:** 10-Mar-22 14:52

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Test Information

i1: Achondroplasia PCR

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

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