

Client: Example Client ABC123
123 Test Drive
Salt Lake City, UT 84108
UNITED STATES

Physician: Doctor, Example

Patient: Patient, Example

DOB: Unknown
Gender: Female
Patient Identifiers: 01234567890ABCD, 012345
Visit Number (FIN): 01234567890ABCD
Collection Date: 00/00/0000 00:00

Achondroplasia (FGFR3) 2 Mutations

ARUP test code 0051266

Achondroplasia PCR Specimen whole Blood

Achondroplasia PCR

HMG1138A *

Indication for testing: Rule out achondroplasia.
Homozygous G1138A (also known as c.1138G>A): This sample is positive for two copies of the G1138A (also known as c.1138G>A) mutation 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 [REDACTED]

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: >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.

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

H=High, L=Low, *=Abnormal, C=Critical

VERIFIED/REPORTED DATES				
Procedure	Accession	Collected	Received	Verified/Reported
Achondroplasia PCR Specimen	23-228-102481	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Achondroplasia PCR	23-228-102481	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00

END OF CHART

H=High, L=Low, *=Abnormal, C=Critical

Unless otherwise indicated, testing performed at:

ARUP LABORATORIES | 800-522-2787 | aruplab.com
500 Chipeta Way, Salt Lake City, UT 84108-1221
Jonathan R. Genzen, MD, PhD, Laboratory Director

Patient: Patient, Example
ARUP Accession: 23-228-102481
Patient Identifiers: 01234567890ABCD, 012345
Visit Number (FIN): 01234567890ABCD
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