

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

Physician: Doctor, Example

**Patient: Patient, Example**

**DOB:** 4/14/2019  
**Gender:** Male  
**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 Negative

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 [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	23-227-114631	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Achondroplasia PCR	23-227-114631	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: 23-227-114631  
Patient Identifiers: 01234567890ABCD, 012345  
Visit Number (FIN): 01234567890ABCD  
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