

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

Physician: Doctor, Example

**Patient: Patient, Example**

**DOB:** 6/25/2014  
**Gender:** Male  
**Patient Identifiers:** 01234567890ABCD, 012345  
**Visit Number (FIN):** 01234567890ABCD  
**Collection Date:** [REDACTED]

**Achondroplasia (FGFR3) 2 Mutations**

ARUP test code 0051266

Achondroplasia PCR Specimen

DNA

Achondroplasia PCR

**HTG1138A \***

Section 79-1 of New York State Civil Rights Law requires informed consent be obtained from patients (or their legal guardians) prior to pursuing genetic testing. These forms must be kept on file by the ordering physician. Consent forms for genetic testing are available at www.aruplab.com. Incidental findings are not reported unless clinically significant but are available upon request.

Indication for testing: Rule out achondroplasia.

Heterozygous G1138A: This sample is positive for one copy of the G1138A mutation in the fibroblast growth factor receptor (FGFR) 3 gene consistent with a diagnosis of achondroplasia.

This result has been reviewed and approved by Elaine Lyon, Ph.D.

**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:** PCR and fluorescent hybridization probes.

**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.

Test developed and characteristics determined by ARUP Laboratories. See Compliance Statement C: aruplab.com/CS

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

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
Achondroplasia PCR Specimen	18-164-146660	6/13/2018 12:00:00 PM	6/21/2018 12:05:22 PM	6/26/2018 8:06:00 AM
Achondroplasia PCR	18-164-146660	6/13/2018 12:00:00 PM	6/21/2018 12:05:22 PM	6/26/2018 8:06:00 AM

END OF CHART

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