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

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.
### VERIFIED/REPORTED DATES

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\[ H=\text{High}, \ L=\text{Low}, \ *=\text{Abnormal}, \ C=\text{Critical} \]