

Client: ARUP Physician Services  
321 TESTING ANSR EXTRACT  
Salt Lake City, NY 84108  
UNITED STATES

Physician: DR, TEST

**Patient: AD PCR FE, 0051265 NEGATIVE**

**DOB**

**Sex:** Female

**Patient Identifiers:** 610374

**Visit Number (FIN):** 634694

**Collection Date:** 5/19/2022 12:19

**Achondroplasia (FGFR3) 2 Mutations, Fetal**

ARUP test code 0051265

Achondroplasia PCR Fetal Specimen      Amniotic fluid

Achondroplasia PCR      Negative

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](http://www.aruplab.com). Incidental findings are not reported unless clinically significant but are available upon request.

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: 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.

**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  
Tracy I. George, MD, Laboratory Director

Patient: AD PCR FE, 0051265 NEGATIVE  
ARUP Accession: 22-139-108315  
Patient Identifiers: 610374  
Visit Number (FIN): 634694  
Page 1 of 2 | Printed: 6/1/2022 7:54:38 AM

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.

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

Maternal Contamination Study Fetal Spec

See Note

Maternal Contam Study, Maternal Spec

Whole Blood

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.

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
Achondroplasia PCR Fetal Specimen	22-139-108315	5/19/2022 12:19:00 PM	5/19/2022 3:08:38 PM	5/25/2022 1:01:00 PM
Achondroplasia PCR	22-139-108315	5/19/2022 12:19:00 PM	5/19/2022 3:08:38 PM	5/25/2022 1:01:00 PM
Maternal Contamination Study Fetal Spec	22-139-108315	5/19/2022 12:19:00 PM	5/19/2022 3:08:38 PM	5/25/2022 1:01:00 PM
Maternal Contam Study, Maternal Spec	22-139-108315	5/19/2022 12:19:00 PM	5/19/2022 3:08:38 PM	5/25/2022 1:01:00 PM

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  
Tracy I. George, MD, Laboratory Director

Patient: AD PCR FE, 0051265 NEGATIVE  
ARUP Accession: 22-139-108315  
Patient Identifiers: 610374  
Visit Number (FIN): 634694  
Page 2 of 2 | Printed: 6/1/2022 7:54:38 AM