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

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, Fetal

ARUP test code 0051265

Achondroplasia PCR Fetal Specimen	Cultured Amnio
Achondroplasia PCR, Fetal Interp	HTG1138A *

Indication for testing: Rule out achondroplasia.
Heterozygous G1138A (also known as c.1138G>A): This sample is positive for one copy of the G1138A (also known as c.1138G>A) mutation in the fibroblast growth factor receptor (FGFR) 3 gene consistent with a diagnosis of achondroplasia.

This result has been reviewed and approved by [redacted]

BACKGROUND INFORMATION: Achondroplasia (FGFR3) 2 Mutations, Fetal
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)/Fluorescent Monitoring/Fragment Analysis.
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 or maternal cell contamination of the fetal specimen.

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.

Maternal Contamination Study Fetal Spec	Fetal Cells

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

	Single fetal genotype present; no maternal cells present. Fetal and maternal samples were tested using STR markers to rule out maternal cell contamination.
Maternal Contam Study, Maternal Spec	Whole Blood

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
Achondroplasia PCR Fetal Specimen	24-019-112398	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Achondroplasia PCR, Fetal Interp	24-019-112398	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Maternal Contamination Study Fetal Spec	24-019-112398	00/00/0000 00:00	00/00/0000 00:00	00/00/0000 00:00
Maternal Contam Study, Maternal Spec	24-019-112398	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