

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

Physician: Doctor, Example

Patient: Patient, Example

DOB: 11/13/2018
Gender: Male
Patient Identifiers: 01234567890ABCD, 012345
Visit Number (FIN): 01234567890ABCD
Collection Date: 01/01/2017 12:34

Thanatophoric Dysplasia, Types 1 and 2 (FGFR3) 13 Mutations

ARUP test code 0051506

Thanatophoric Dysplasia I/II Specimen whole Blood

Thanatophoric Dysplasia I/II

Negative

Negative: None of the targeted fibroblast growth factor receptor (FGFR) 3 gene mutations were identified. These mutations are responsible for approximately 99% of thanatophoric dysplasia types I and II.

This result has been reviewed and approved by [REDACTED]

BACKGROUND INFORMATION: Thanatophoric Dysplasia Types I and II (FGFR3) 13 Mutations

CHARACTERISTICS: Micromelia macrocephaly short ribs and a narrow thorax; TD I has bowed femurs and TD II has straight femurs and a cloverleaf skull; death usually occurs from respiratory insufficiency within hours or days of birth.
INCIDENCE: 1 in 20,000 to 50,000 births.
INHERITANCE: Autosomal dominant; arising from a de novo mutation.
CAUSE: Fibroblast growth factor receptor 3 (FGFR3) gene mutations.
MUTATIONS TESTED: c.742C>T (R248C), c.746C>G (S249C), c.1108G>T (G370C), c.1111A>T (S371C), c.1118A>G (Y373C), c.2419T>G (X807G), c.2419T>A (X807R), c.2420G>T (X807L), c.2420G>C (X807S), c.2421A>T (X807C), c.2421A>C (X807C) and c.2421A>G (X807W) in TD I and c.1948A>G (K650E) in TD II.
CLINICAL SENSITIVITY AND SPECIFICITY: 99 percent.
METHODOLOGY: Polymerase chain reaction (PCR) and single nucleotide extension followed by capillary electrophoresis.
ANALYTICAL SENSITIVITY AND SPECIFICITY: 99 percent.
LIMITATIONS: Mutations other than those targeted in FGFR3 will not be detected analytical sensitivity may be compromised by rare primer site mutations. 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

Unless otherwise indicated, testing performed at:

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
Thanatophoric Dysplasia I/II Specimen	18-318-127737	11/14/2018 1:55:00 PM	11/15/2018 7:24:00 PM	11/19/2018 3 25:00 PM
Thanatophoric Dysplasia I/II	18-318-127737	11/14/2018 1:55:00 PM	11/15/2018 7:24:00 PM	11/19/2018 3 25:00 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: Patient, Example
ARUP Accession: 18-318-127737
Patient Identifiers: 01234567890ABCD, 012345
Visit Number (FIN): 01234567890ABCD
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