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

ARUP test code 0051508

Thanatophoric Dysplasia I/II, Fetal Spec
Cultured Amnio

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 Yuan Ji, Ph.D.

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 (X807T), c.2420G>T (X807L), c.2420G>C (X807S), c.2421A>T (X807C), c.2421A>C (X807T) 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

Maternal Contamination Study Fetal Spec
Unknown Origin

H=High, L=Low, *=Abnormal, C=Critical
Single genotype. A maternal specimen was not submitted for correlation. The fetal sample was tested using STR markers to rule out maternal cell contamination. Only a single genotype was detected. Testing a maternal sample can confirm that this genotype is from the fetus.

**INTERPRETIVE INFORMATION: Maternal Cell Contamination, Fetal Specimen**

Please refer to fetal report for interpretation.

**Maternal Contam Study, Maternal Spec**

Not Received

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

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