

HOTLINE: Effective September 7, 2021

0051508

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

TD PAN FE

Specimen Required: Collect: Cultured Amniocytes, Cultured CVS, or Amniotic fluid (direct).

Maternal **Whole Blood** Specimen: Lavender (EDTA), pink (K₂EDTA), or yellow (ACD Solution A or B).

Specimen Preparation: **Cultured Amniocytes or Cultured CVS:** Transfer cultured amniocytes or cultured CVS to two T-25 flasks at 80% confluence. (Min: one T-25 flask at 80% confluence). Backup cultures must be retained at the client's institution until testing is complete. **If the client is unable to culture amniocytes or CVS, this can be arranged by contacting ARUP Client Services at (800) 522-2787. Please contact an ARUP genetic counselor at (800) 242-2787 ext. 2141 prior to test submission.**

Amniotic Fluid (direct): 10 milliliters

Maternal Whole Blood Specimen: Transport 2 mL whole blood (Min: 1mL).

Storage/Transport Temperature: **Cultured Amniocytes or Cultured CVS:** **CRITICAL ROOM TEMPERATURE.** Must be received within 48 hours of collection due to viability of cells.

Amniotic fluid (direct): Ship room temperature.

Maternal Whole Blood Specimen: Room temperature.

Remarks: Please contact an ARUP genetic counselor at (800) 242-2787 ext. 2141 prior to sample submission.. Patient History Form is available on the ARUP Web site or by contacting ARUP Client Services at (800) 522-2787.

Stability (collection to initiation of testing): **Cultured Amniocytes or Cultured CVS:** Room temperature: 48 hours; Refrigerated: Unacceptable; Frozen: Unacceptable

Amniotic Fluid (direct): Room temperature: 48 hours; Refrigerated: 72 hours; Frozen: Unacceptable

Maternal Whole Blood Specimen: Room temperature: 7 days; Refrigerated: 1 month; Frozen: Unacceptable