

HOTLINE: Effective August 15, 2022

2012010 Skeletal Dysplasia Panel, Sequencing and Deletion/Duplication, Fetal

SKEL FE

Specimen Required: Collect: Fetal Specimen: Four (4) T-25 flasks at 80% confluent of cultured amniocytes or cultured chorionic villus sampling (CVS).

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

Specimen Preparation: Cultured Amniocytes or Cultured CVS: Fill flasks with culture media. Transport four (4) T-25 flasks at 80 percent confluent of cultured amniocytes or cultured CVS filled with culture media. Backup cultures must be retained at the client's institution until testing is complete. If the client is unable to culture amniocytes, this can be arranged by contacting ARUP Client Services at (800) 522-2787 ext. 2141 prior to test submission.

Maternal Whole Blood Specimen: Transport 3 mL whole blood (Min: 2 mL)

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

Maternal Specimen: Room temperature.

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

Unacceptable; Frozen: Unacceptable

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

Note: Genes Tested: AGPS; ALPL; ARSL; CANT1; CCN6; CILK1; COL1A1; COL1A2*; COL2A1; COL10A1; COL11A1; COL11A2; COMP; CRTAP; DDR2; DLL3; DYM*; DYNC2H1; EBP; EVC; EVC2; FGFR1*; FGFR2; FGFR3; FKBP10; FLNA; FLNB; GDF5; GNPAT; HSPG2; IFT80; INPPL1; LBR; LIFR; NEK1*; NPR2; P3H1; PCNT; PEX7; POR*; PPIB; PTH1R; RUNX2; SERPINH1; SLC26A2; SLC35D1; SMARCAL1; SOX9; TRIP11; TRPV4; TTC21B; WDR19; WDR35

Reported times are based on receiving the four T-25 flasks at 80 percent confluent. Cell culture time is independent of testing turnaround time. Maternal specimen is recommended for proper test interpretation. Order Maternal Cell Contamination.

CPT Code(s): 81405; 81408; 81479; 81265 Fetal Cell Contamination (FCC)

^{*} One or more exons are not covered by sequencing and/or deletion/duplication analysis for the indicated gene; see Additional Technical Information.