

HOTLINE: Effective November 14, 2022

2012010

Skeletal Dysplasia Panel, Sequencing and Deletion/Duplication, Fetal

SKEL FE

Specimen Required: Collect: **Fetal specimen:** Two (2) 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 two (2) T-25 flasks at 80% 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*

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

Reported times are based on receiving the four T-25 flasks at 80% 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