TEST CHANGE

Skeletal Dysplasia Panel, Sequencing and Deletion/Duplication, Fetal

2012010, SKEL FE

Specimen Requirements:	
Patient Preparation:	
Collect:	Fetal Sspecimen: Two (2) T-25 flasks at 9080% confluent of cultured amniocytes or cultured chorionic villus sampling (CVS). AND Maternal Whole Blood Specimenwhole blood specimen: Lavender (EDTA), pink (K2EDTA), or yellow (ACD Ssolution A or B).
Specimen Preparation:	Cultured Aamniocytes or Ccultured CVS: Fill flasks with culture media. Transport two (2) T-25 flasks at 90 percent80% 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 ARUP receives a sample below the minimum confluence, CG GRW&SND (0040182) will be added on by ARUP, and additional charges will apply. If clients areclient is unable to culture specimens, CG GRW&SND should be addedamniocytes, this can be arranged by contacting ARUP Client Services at (800) 522-2787 ext. 2141 prior to initial order.test submission. Maternal Whole Blood Specimenwhole blood specimen: Transport 3 mL whole blood. (Min: 12 mL)).
Transport Temperature:	Cultured Aamniocytes or Ccultured CVS: CRITICAL ROOM TEMPERATURE. Must be received within 48 hours of shipment due to viability of cells. Maternal Specimen: Room temperature.
Unacceptable Conditions:	
Remarks:	
Stability:	Cultured Aamniocytes or Ccultured CVS: Room temperature: 48 hours; Refrigerated: Unacceptable; Frozen: Unacceptable Maternal Whole Blood Specimenblood specimen: Room temperature: 7 days; Refrigerated: 1 month; Frozen: Unacceptable
Methodology:	Massively Parallel Sequencing
Performed:	Varies
Reported:	2-34 weeks, if culture is required an additional 1 to 2 weeks is required for processing time.

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 two T-25 flasks at 90 percent confluent. Cell culture time is independent of testing turnaround time. Maternal specimen is recommended for proper test interpretation. Order Maternal Cell Contamination.
CPT Codes:	81405; 81408; 81479; 81265
New York DOH Approval Status:	Specimens from New York clients will be sent out to a New York DOH approved laboratory, if possible.

Interpretive Data:

Refer to report.

This test was developed and its performance characteristics determined by ARUP Laboratories. It has not been cleared or approved by the US Food and Drug Administration. This test was performed in a CLIA certified laboratory and is intended for clinical purposes.

Counseling and informed consent are recommended for genetic testing. Consent forms are available online.

Reference Interval:

By report