TEST CHANGE

Skeletal Dysplasia Panel, Sequencing and Deletion/Duplication, Fetal

2012010, SKEL FE

Specimen Requirements:	
Patient Preparation:	
Collect:	Fetal Specimen: Cultured amniocytes OR cultured chorionic villi. Maternal Specimen: Refer to Maternal Cell Contamination, Maternal Specimen (0050608) for maternal specimen requirements.
Specimen Preparation:	Cultured Amniocytes or Cultured CVS: Fill flasks with culture media. Transport two T-25 flasks of 90 percent confluent cultured amniocytes or two T-25 flasks of 90% cultured chorionic villi sampling (CVS). This assay is not performed on direct amniotic fluid or direct chorionic villi specimens. Clients submitting direct amniotic fluid and direct chorionic villi must add Cell Culture for Genetic Testing (3020627) to the initial order. If ARUP receives cultured specimens below the minimum confluence, Cell Culture for Genetic Testing (3020627) will be added by ARUP for an additional fee. The client is responsible for maintaining backup cultures.
Transport Temperature:	Cultured Amniocytes or Cultured CVS: CRITICAL ROOM TEMPERATURE. Must be received within 48 hours of shipment due to viability of cells.
Unacceptable Conditions:	
Remarks:	Patient history forms and informed consent documents are available by selecting the links above or by contacting ARUP Client Services. Counseling and informed consent are recommended for genetic testing. New York Clients: Informed consent is required with specimen submission.
Stability:	Cultured Amniocytes or Cultured CVS: Room temperature: 48 hours; Refrigerated: Unacceptable; Frozen: Unacceptable. New York State Clients: Specimens must be received at performing laboratory within 24 hours of shipping. Older specimens will be evaluated by performing laboratory for acceptability. For specimen requirements and direct submission instructions please contact ARUP Referral Testing at 800-242-2787 ext. 5161.
Methodology:	Massively Parallel Sequencing

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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.
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.

Reference Interval:

Refer to report