

TEST CHANGE

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

Specimen Requirements:

Patient Preparation:

Collect: Fetal Specimen: ~~Cultured Two T-25 flasks at 90% confluent of cultured~~ amniocytes ~~OR~~ cultured chorionic villi.

~~villus sampling (CVS).~~
AND Maternal ~~Whole Blood~~ Specimen: Refer to Maternal Cell Contamination, Maternal Specimen (0050608) for maternal specimen requirements. ~~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 T-25 flasks ~~of~~at 90 percent confluent ~~of~~ 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.

~~filled with culture media. Backup cultures must be retained at the client's institution until testing is complete. If ARUP receives cultured specimens a sample below the minimum confluence, Cell Culture for Genetic Testing (3020627 CG GRW&SND (0040182) will be added on by ARUP for an and additional fee. The client is responsible for maintaining backup cultures charges will apply. If clients are unable to culture specimens, CG GRW&SND should be added to initial order.~~

~~Maternal Whole Blood Specimen: Transport 3 mL whole blood (Min: 1 mL)~~

~~New York State Clients: Specimens must be sent overnight to the performing laboratory. For specimen requirements and direct submission instructions please contact ARUP Referral Testing at 800-242-2787 ext. 5161.~~

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.~~

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:

New York State Clients: 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
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

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

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*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.

~~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:

Refer to **By** report
