HOTLINE: Effective August 15, 2022

2008863 Holoprosencephaly Panel, Sequencing and Deletion/Duplication, Fetal HPE PAN FE

Methodology: Massively Parallel Sequencing

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 (K_2EDTA), 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: 3 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: Determine the etiology of holoprosencephaly in an affected pregnancy or determine if parents of an affected pregnancy are carriers. Chromosome analysis should be performed in an affected pregnancy before ordering this test.

Genes tested: *CDON*; *FGFR1**; *GLI2*; *PTCH1*; *SHH*; *SIX3*; *TGIF1*; *ZIC2**

* 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 (4) 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): 81479; 81265 Fetal Cell Contamination (FCC)

HOTLINE NOTE: Remove information found in the Remarks field.