

TEST CHANGE

Holoprosencephaly Panel, Sequencing and Deletion/Duplication, Fetal

2008863, HPE PAN FE

Specimen Requirements:

Patient Preparation:

Collect:

Fetal Specimen: Two (2) T-25 flasks at ~~90~~80% confluent of cultured amniocytes or cultured chorionic villus sampling (CVS). AND ~~Ma~~AND Maternal **Whole Blood Specimen**~~whole blood specimen~~: Lavender (EDTA), pink (K2EDTA), or yellow (ACD ~~S~~solution A or B).

Specimen Preparation:

Cultured ~~A~~amniocytes or ~~C~~eultured CVS: Fill flasks with culture media. Transport two (2) T-25 flasks at ~~90 percent~~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 ARUP receives a sample below~~ If the minimum confluence, CG GRW&SND (0040182) will be added on by ARUP, and additional charges will apply. If clients are ~~client is~~ unable to culture ~~specimens, CG GRW&SND should be added~~ amniocytes, this can be arranged by contacting ARUP Client Services at (800) 522-2787 ext. 2141 prior to initial order. ~~test submission.~~ Maternal **Whole Blood Specimen**~~whole blood specimen~~: Transport 3 mL whole blood. (Min: ~~1~~3 mL)

Transport Temperature:

Cultured ~~A~~amniocytes or ~~C~~eultured CVS: CRITICAL ROOM TEMPERATURE. Must be received within 48 hours of shipment due to viability of cells. Maternal Specimen: Room ~~T~~emperature

Unacceptable Conditions:

Remarks:

Stability:

Cultured ~~A~~amniocytes or ~~C~~eultured CVS: Room temperature: 48 hours; Refrigerated: Unacceptable; Frozen: Unacceptable
Maternal **Whole Blood Specimen**~~whole blood specimen~~: Room temperature: 7 days, Refrigerated: 1 month, Frozen: Unacceptable

Methodology:

Massively Parallel Sequencing

Performed:

Varies

Reported:

2-~~3~~4 weeks; if culture is required, an additional 1 to 2 weeks is required for processing time.

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 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: 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.

Patient History forms are available online at www.aruplab.com.

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