TEST CHANGE

Holoprosencephaly Panel, Sequencing and Deletion/Duplication, Fetal

2008863, HPE PAN FE

Specimen Requirements:	
Patient Preparation:	
Collect:	Fetal Specimen: Cultured ~~Two T-25 flasks at 90% confluent of cultured~~ amniocytes OR~~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 ~~filled with~~ Culture for Genetic Testing (3020627) to the initial order. ~~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: Specimen must be sent overnight to 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:	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~~
Methodology:	Massively Parallel Sequencing
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. Chromosome analysis, with or without reflex to genomic microarray, should be performed for the affected pregnancy before ordering this test. ~~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. ~~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:	

Refer to~~By~~ report