

**TEST CHANGE**

**Holoprosencephaly Panel, Sequencing and Deletion/Duplication**

2008848, HPE PAN

**Specimen Requirements:**

**Patient Preparation:**

**Collect:** Lavender or pink (EDTA) or yellow (ACD solution A or B).  
~~New York State Clients: Lavender (EDTA)~~

**Specimen Preparation:** Transport 3 mL whole blood. (Min: 2 mL)  
New York State Clients: 5 mL (Min: ~~3~~ 2 mL)

**Transport Temperature:** Refrigerated.

**Unacceptable Conditions:** Serum or plasma; grossly hemolyzed or frozen specimens; saliva, buccal brush, or swab; FFPE tissue.

**Remarks:**

**Stability:** Ambient: 72 hours; Refrigerated: 1 week; Frozen: Unacceptable  
New York State Clients: Ambient: ~~8 days~~24 hours; Refrigerated:  
~~8 days~~1 week; Frozen: ~~1 month~~Unacceptable

**Methodology:** Massively Parallel Sequencing

**Note:**

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.

When testing cord blood specimens, the presence of maternal cell contamination (MCC) is possible, which may impact result interpretation. If clinically warranted, testing for MCC is available, at a charge, through ARUP Laboratories.

**CPT Codes:** 81479

**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



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and its Department of Pathology*

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