

## HOTLINE: Effective August 15, 2022

2008848 Holoprosencephaly Panel, Sequencing and Deletion/Duplication HPE PAN

Methodology: Massively Parallel Sequencing

Performed: Varies
Reported: 3 weeks

Specimen Required: Collect: Lavender or pink (EDTA) or yellow (ACD Solution A or B)

Specimen Preparation: Transport 3 mL whole blood. (Min: 2 mL)

Storage/Transport Temperature: Refrigerated.

<u>Unacceptable Conditions:</u> Serum or plasma; grossly hemolyzed or frozen specimens; saliva, buccal brush, or swab; FFPE tissue.

Stability (collection to initiation of testing): Ambient: 72 hours; Refrigerated: 1 week; Frozen: Unacceptable

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.

**CPT Code(s):** 81479

**HOTLINE NOTE:** Remove information found in the Remarks field.