DISEASE:
Microform holoprosencephaly

NAME:	Microform holoprosencephaly
DESCRIPTION:	A benign form of holoprosencephaly characterized by midline defects without the typical HPE defect in brain cleavage and which can variably manifest with microcephaly, hypotelorism, midline cleft lip and/or flat nose, choanal stenosis, pyriform sinus stenosis, coloboma as well as a single median maxillary incisor.
ORPHACODE:	280200
SYNONYMS:	HPE, minor form HPE-L Holoprosencephaly, minor form Holoprosencephaly-like Microform HPE
XREF(S):	Orphanet OMIM OMIM ICD-10 OMIM OMIM

ANALYTE(S):	FGFR1 SUFU PTCH1 SIX3 TGIF1 ZIC2 GLI2 CRIPTO FOXH1 FGF8 DISP1 CDON NODAL DLL1 GAS1 SHH
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RELATED CONTENT

Related Genetic Tests

- Brain malformations (gene panel)
- cleft lip with/whitout cleft palate (virtual gene panel)

Related Laboratories

- Centre de Génétique Humaine - Erasme ULB
- Centre de Génétique Médicale UCL

Related Analytes

- cell adhesion associated, oncogene regulated
- cripto, EGF-CFC family member
- dispatched RND transporter family member 1
- delta like canonical Notch ligand 1
- fibroblast growth factor 8
- fibroblast growth factor receptor 1
- forkhead box H1
- growth arrest specific 1
- GLI family zinc finger 2
- nodal growth differentiation factor
- patched 1
- sonic hedgehog signaling molecule
- SIX homeobox 3

- SUFU negative regulator of hedgehog signaling
- TGFB induced factor homeobox 1
- Zic family member 2

Related Gene Panels

- Brain malformations (34 genes) - ULB
- Cleft lip and palate / dysmorphic facial features / craniofacial anomalies (255 genes)) - UCL
- Hypogonadotropic Hypogonadism/Kallmann (61 genes) - ULG
