DISEASE:
Septopreoptic holoprosencephaly

NAME:	Septopreoptic holoprosencephaly
DESCRIPTION:	A rare subtype of holoprosencephaly characterized by midline fusion limited to the septal and/or preoptic regions of the telencephalon without a significant frontal neocortical fusion. Midline craniofacial malformations are generally mild and include solitary median maxillary incisor and pyriform sinus stenosis. Other reported manifestations include language delay, learning difficulties, and behavioral disorders. Imaging reveals abnormal fornix, absent or hypoplasic anterior corpus callosum, and unpaired anterior cerebral artery.
ORPHACODE:	280195
SYNONYMS:	Septopreoptic HPE
XREF(S):	Orphanet OMIM OMIM OMIM ICD-10

ANALYTE(S):	STIL PTCH1 SHH SIX3 TGIF1 ZIC2 GLI2 CRIPTO FOXH1 FGF8 DISP1 CDON NODAL DLL1 GAS1
CREATED:	13 May 2019 - 01:02
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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
- 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
- STIL centriolar assembly protein

- TGFB induced factor homeobox 1
- Zic family member 2

Related Gene Panels

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