DISEASE:
Semilobar holoprosencephaly

NAME:	Semilobar holoprosencephaly
DESCRIPTION:	A form of holoprosencephaly characterized by fusion of the left and right frontal and parietal lobes with only a posterior interhemispheric fissure. Craniofacial features variably include ocular hypotelorism, midline cleft lip (complete or partial) and a flat nose.
ORPHACODE:	220386
XREF(S):	Orphanet OMIM OMIM OMIM ICD-10 OMIM

ANALYTE(S):	STAG2 FGFR1 SMC1A STIL PTCH1 SHH SIX3 TGIF1 ZIC2 GLI2 CRIPTO FOXH1 FGF8 DISP1 CDON NODAL DLL1 GAS1
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RELATED CONTENT

Related Genetic Tests

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

Related Laboratories

- Centre de Génétique Humaine - Erasme ULB
- Centre de Génétique Médicale UCL
- Centrum Medische Genetica - UZ Antwerpen

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
- structural maintenance of chromosomes 1A
- STAG2 cohesin complex component
- 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
- Rare epilepsy with developmental delay (> 240 genes) - UZA
