DISEASE:
Lobar holoprosencephaly

NAME:	Lobar holoprosencephaly
DESCRIPTION:	A form of holoprosencephaly characterized by separation of the right and left cerebral hemispheres and lateral ventricules with some continuity only across the frontal neocortex, especially rostrally and ventrally. Craniofacial features are variable may include ocular hypotelorism, midline cleft lip (complete or partial) and/or flat nose amongst other features.
ORPHACODE:	93924
XREF(S):	Orphanet OMIM ICD-10 OMIM OMIM

ANALYTE(S):	STIL FGFR1 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)
- 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

- 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
