DISEASE:
Midline interhemispheric variant of holoprosencephaly

NAME:	Midline interhemispheric variant of holoprosencephaly
DESCRIPTION:	Midline interhemispheric variant of holoprosencephaly (MIH) or syntelencephaly is a form of holoprosencephaly (HPE; see this term) characterized by non-separation of the posterior frontal and parietal lobes, normally-formed callosal genu and splenium, absence of the callosal body, normally-separated hypothalamus and lentiform nucleus, and frequent heterotopic gray matter.
ORPHACODE:	93926
SYNONYMS:	MIH MIH type HPE MIHF MIHV Middle interhemispheric fusion variant Middle interhemispheric variant of holoprosencephaly Syntelencephaly
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
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RELATED CONTENT

Related Genetic Tests

- Brain malformations (gene panel)

Related Laboratories

- Centre de Génétique Humaine - Erasme ULB

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

- Hypogonadotropic Hypogonadism/Kallmann (61 genes) - ULG
