DISEASE:
Septo-optic dysplasia spectrum

NAME:	Septo-optic dysplasia spectrum
DESCRIPTION:	A rare clinically heterogeneous disorder characterized by the classical triad of optic nerve hypoplasia, pituitary hormone abnormalities and midline brain defects.
ORPHACODE:	3157
SYNONYMS:	De Morsier syndrome SOD Septo-optic dysplasia
XREF(S):	Orphanet ICD-10 MeSH OMIM MedDRA

ANALYTE(S):	PROKR2 SOX2 FGFR1 HESX1 OTX2 SOX3 ARNT2
CREATED:	13 May 2019 - 01:02
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Source URL: http://gentest.healthdata.be/disease/1245

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- Septo-optic dysplasia (HESX1 gene)

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- Centre de Génétique-Institut de Pathologie et de Génétique (IPG)
- Centrum Medische Genetica - UZ Brussel VUB

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- prokineticin receptor 2
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