
DISEASE:
Isolated optic nerve hypoplasia/aplasia

NAME:	Isolated optic nerve hypoplasia/aplasia
DESCRIPTION:	A rare genetic optic nerve disorder characterized by visual impairment or blindness resulting from varying degrees of underdevelopment of the optic nerve or even complete absence of the optic nerve, ganglion cells, and central retinal vessels. It may be unilateral, typically with otherwise normal brain development, or bilateral with accompanying severe and widespread congenital malformations of the central nervous system.
ORPHACODE:	137902
XREF(S):	<u>Orphanet</u> <u>OMIM</u> <u>ICD-10</u>
ANALYTE(S):	<u>PAX6</u>
CREATED:	13 May 2019 - 01:02
CHANGED:	01 May 2022 - 06:55

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