

Opis choroby *

Definicja

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.

Dane

Klasyfikacja

Wada morfologiczna

Kod ORPHA

137902

Kod OMIM

165550

Kod ICD10

Q07.8

Kod ICD11

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*Źródło

orphanet