

Opis choroby *

Definicja

A rare developmental defect during embryogenesis characterized by abnormal retinal development with congenital blindness. Common associated manifestations include sensorineural hearing loss and developmental delay, intellectual disability and/or behavioral disorders.

Dane

Klasyfikacja	Synonimy
Zespół wad wrodzonych	Atrophia bulborum hereditaria
	Choroba Norriego i Warburga
	Dziedziczna atrofia opuszkowa
	Ślepotą Episkopi
	Episkopi blindness
	Norrie-Warburg disease

Kod ORPHA
649

Kod OMIM
310600

Kod ICD10
H35.5

Kod ICD11
LD21.Y

*Źródło

orphanet