

## 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