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
Zespół wad wrodzonych

Synonimy
Atrophia bulborum hereditaria
Choroba Norriego i Warburga
Dziedziczna atrofia opuszkowa
Ślepota Episkopi
Episkopi blindness
Norrie-Warburg disease

Kod ORPHA
649

Kod OMIM
310600

Kod ICD10
H35.5

Kod ICD11
LD21.Y

---

*Źródło

orphanet