

Opis choroby *

Definicja

A rare genetic isolated inherited retinal disorder characterized by primary cone degeneration with significant secondary rod involvement, with a variable fundus appearance. Typical presentation includes decreased visual acuity, central scotoma, photophobia, color vision alteration, followed by night blindness and loss of peripheral visual field.

Dane

Klasyfikacja

Choroba

Kod ORPHA

1872

Kod OMIM

616502

Kod ICD10

H35.5

Kod ICD11

9B70

*Źródło

orphanet