Opis choroby *

Definicja

A rare hereditary optic atrophy characterized by an early onset of bilateral optic nerve degeneration without other systemic features. Clinical manifestations include pallor of the optic disks, severe but slowly progressing visual impairment, and in some patients also paracentral scotoma, photophobia and dyschromatopsia.

Dane

Klasyfikacja
Choroba

Synonimy
Autosomal recessive non-syndromic optic atrophy
Autosomal recessive non-syndromic optic atrophy

Kod ORPHA
98676

Kod OMIM
616732

Kod ICD10
H47.2

Kod ICD11
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*Źródło

orphanet