Opis choroby *

Definicja

A rare, syndromic, hereditary optic neuropathy disorder characterized by early-onset, severe, progressive visual impairment, optic disc pallor and central scotoma, variably associated with dyschromatopsia, auditory neuropathy (e.g. mild progressive sensorineural hearing loss), sensorimotor axonal neuropathy and, occasionally, moderate hypertrophic cardiomyopathy.

Dane

Klasyfikacja
Choroba

Kod ORPHA
227976

Kod OMIM
612989

Kod ICD10
H47.2

Kod ICD11
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*Źródło

orphanet