

Opis choroby *

Definicja

A rare mitochondrial disease characterized by bilateral auditory neuropathy and optic atrophy. Patients present hearing and visual impairment in the first or second decade of life, while psychomotor development is normal. Bilateral retinitis pigmentosa has been reported in association.

Dane

Klasyfikacja

Choroba

Kod ORPHA

542585

Kod OMIM

617717

Kod ICD10

E88.8

Kod ICD11

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*Źródło

orphanet