

## **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