Opis choroby *

Definicja

Early-onset X-linked optic atrophy is a rare form of hereditary optic atrophy, seen in only 4 families to date, with an onset in early childhood, characterized by progressive loss of visual acuity, significant optic nerve pallor and occasionally additional neurological manifestations, with females being unaffected.

Dane

Klasyfikacja
Choroba

Synonimy
Non-Leber type optic atrophy with early-onset
OPA2
Zanik nerwu wzrokowego typu 2
OPA2
Optic atrophy type 2

Kod ORPHA
98890

Kod OMIM
311050

Kod ICD10
H47.2

Kod ICD11
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*Źródło

orphanet