Opis choroby *

Definicja

A form of autosomal dominant optic atrophy characterized by an early and bilateral optic atrophy leading to insidious visual loss of variable severity, followed by a late anterior and/or posterior cortical cataract. Additional features include sensorineural hearing loss and neurological signs such as tremor, extrapyramidal rigidity and absence of deep tendon reflexes. It is caused by mutations in the OPA3 gene (19q13.32).

Dane

Klasyfikacja
Choroba

Synonimy
Autosomal dominant optic atrophy type 3
Autosomalna dominująca atrofia nerwu wzrokowego typu 3
OPA3, autosomalna dominująca
OPA3, autosomal dominant

Kod ORPHA
67036

Kod OMIM
165300

Kod ICD10
H47.2

Kod ICD11
9C40.8

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*Źródło

orphanet