

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

*Źródło

orphanet