Opis choroby *

Definicja

A rare genetic, neuro-ophthalmological disease characterized by progressive weakness of the external eye muscles, resulting in bilateral ptosis and diffuse symmetric ophthalmoparesis. Additional signs may include skeletal muscle weakness, cataracts, hearing loss, sensory axonal neuropathy, ataxia, parkinsonism, cardiomyopathy, hypogonadism and depression. It is usually less severe than autosomal recessive form.

Dane

Klasyfikacja
Choroba

Synonimy
adPEO
adPEO

Kod ORPHA
254892

Kod OMIM
157640

Kod ICD10
H49.4

Kod ICD11
9C82.0

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*Źródło

orphanet