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 generalized skeletal muscle weakness, muscle atrophy, sensory axonal neuropathy, ataxia, cardiomyopathy, and psychiatric symptoms. It is usually more severe than autosomal dominant form.

Dane

Klasyfikacja
Choroba

Synonimy
arPEO
arPEO

Kod ORPHA
254886

Kod OMIM
617069

Kod ICD10
H49.4

Kod ICD11
9C82.0

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*Źródło

orphanet