

Opis choroby *

Definicja

A rare mitochondrial disease characterized by adult onset of progressive external ophthalmoplegia, exercise intolerance, muscle weakness, manifestations of spinocerebellar ataxia (e.g. impaired gait, dysarthria) and mild motor peripheral neuropathy. Respiratory insufficiency has been reported in some cases.

Dane

Klasyfikacja

Choroba

Synonimy

Adult-onset CPEO with mitochondrial myopathy
CPEO o początku w wieku dorosłym z miopatią mitochondrialną

Kod ORPHA

329336

Kod OMIM

616479

Kod ICD10

G71.3

Kod ICD11

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*Źródło

orphanet