Opis choroby *

Definicja

Progressive external ophthalmoplegia-myopathy-emaciation syndrome is a rare mitochondrial oxidative phosphorylation disorder due to nuclear DNA anomalies characterized by progressive external ophthalmoplegia without diplopia, cerebellar atrophy, proximal skeletal muscle weakness with generalized muscle wasting, profound emaciation, respiratory failure, spinal deformity and facial muscle weakness (manifesting with ptosis, dysphonia, dysphagia and nasal speech). Intellectual disability, gastrointestinal symptoms (e.g. nausea, abdominal fullness, and loss of appetite), dilated cardiomyopathy and renal colic have also been reported.

Dane

Klasyfikacja
Choroba

Synonimy
Mitochondrial DNA maintenance syndrome due to MGME1 deficiency
PEO - miopatia - wyniszczenie
Zespół utrzymania mitochondrialnego DNA z powodu niedoboru MGME1
Zespół utrzymania mtDNA z powodu niedoboru MGME1
PEO-myopathy-emaciation syndrome
mtDNA maintenance syndrome due to MGME1 deficiency

Kod ORPHA
352447

Kod OMIM
615084

Kod ICD10
G71.3

Kod ICD11
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*Źródło

orphanet