Opis choroby *

Definicja

An extremely rare multiple mitochondrial DNA deletion syndrome with markedly decreased deoxyguanosine kinase (DGUOK) activity in skeletal muscle characterized by a highly variable phenotype. Clinical manifestations include progressive external ophthalmoplegia, mitochondrial myopathy, recurrent rhabdomyolysis, lower motor neuron disease, mild cognitive impairment, sensory axonal neuropathy, optic atrophy, ataxia, hypogonadism and/or parkinsonism.

Dane

Klasyfikacja
Choroba

Synonimy
Adult-onset multiple mtDNA deletion syndrome due to DGUOK deficiency
Zespół mnogich delecji mtDNA z powodu deficytu DGUOK o początku w wieku dorosłym

Kod ORPHA
329314

Kod OMIM
617070

Kod ICD10
G71.3

Kod ICD11
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*Źródło

orphanet