Opis choroby *

Definicja

A rare mitochondrial myopathy characterized by motor developmental delay (in infancy), growth impairment and mostly proximal muscle weakness caused by a muscular dystrophy. Muscle biopsy presents myopathic abnormalities and decreased mtDNA content. Electromyography (EMG) shows a myopathic process and serum creatine kinase is increased. The disease is also characterized by early onset non-progressive cerebellar atrophy (particularly cerebellar vermis and hemispheres), corticospinal tract dysfunction, and global or partial cerebral atrophy on brain MRI. Additionally, some patients presented with cognitive deficiencies, skeletal abnormalities, tremors, and retinopathy.

Dane

Klasyfikacja
Choroba

Synonimy
Mitochondrial myopathy-cerebellar atrophy-pigmentary retinopathy syndrome
Mitochondrial myopathy-cerebellar atrophy-pigmentary retinopathy syndrome

Kod ORPHA
502423

Kod OMIM
617675

Kod ICD10
G71.3

Kod ICD11
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*Źródło

orphanet