

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
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Kod ORPHA

502423

Kod OMIM

617675

Kod ICD10

G71.3

Kod ICD11

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[*Źródło](#)

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