Opis choroby *

Definicja

A rare genetic neuromuscular disease characterized by late onset of mild, progressive, proximal muscle weakness, severe myalgias during and after exercise, and susceptibility to rhabdomyolysis. Intellectual disability is mild or absent. There are no abnormalities of the skin. Muscle biopsy shows focal depletion of mitochondria especially at the center of muscle fibers, surrounded by enlarged mitochondria at the periphery.

Dane

Klasyfikacja

Choroba

Kod ORPHA 521305

Kod OMIM 600706

Kod ICD10 G72.8

Kod ICD11

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*Źródło

orphanet