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