

Opis choroby *

Definicja

Myopathy with hexagonally cross-linked tubular arrays is a rare, congenital, non-dystrophic, mild, slowly progressive, proximal myopathy characterized by exercise intolerance and post-exercise myalgia without rhabdomyolysis, associated with highly organized hexagonally cross-linked tubular arrays in skeletal muscle biopsy. Additional features may include muscle atrophy (or diffuse hypotrophy), myalgia with or without muscular weakness, paresis of truncal and limb-girdle musculature, minimal ptosis, lumbar hyperlordosis, decreased deep tendon reflexes, contractures and pes equinovarus.

Dane

Klasyfikacja

Choroba

Kod ORPHA

171889

Kod OMIM

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Kod ICD10

G71.2

Kod ICD11

8C72.0Y

*Źródło

orphanet