Opis choroby *

Definicja

A rare congenital myopathy characterized ultrastructurally by the presence of tubular aggregates in the subsarcolemmal region of the muscle fiber. It most commonly presents with slowly progressive proximal muscle weakness predominantly of the lower limbs, periodic paralysis, post-exertion muscle cramps, and muscular pain. Ocular anomalies like ophthalmoplegia or pupillary abnormalities may be associated. The intensity of the symptoms is variable, cases with normal muscle strength but myalgia or fatigue, as well as clinically asymptomatic cases have been described.

Dane

Klasyfikacja
Choroba

Kod ORPHA
2593

Kod OMIM
615883

Kod ICD10
G71.2

Kod ICD11
8C72.Y

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*Źródło

orphanet