Opis choroby *

Definicja

A rare genetic skeletal muscle disease characterized by childhood onset of exercise-induced progressive impairment of muscle relaxation, stiffness, cramps, and myalgia, predominantly in the arms, legs, and face (eyelids), and, biochemically, by a reduced sarcoplasmic reticulum Ca(2+)-ATPase activity. Symptoms improve after a few minutes of rest and may be exacerbated by cold. The term Brody syndrome refers to a clinically distinguishable subset of patients without ATP2A1 mutations, with adolescence or adult onset and selective muscular involvement, in which myalgia is more common.

Dane

Klasyfikacja
Choroba

Kod ORPHA
53347

Kod OMIM
601003

Kod ICD10
G71.8

Kod ICD11
8C7Y

---

*Źródło

orphanet