

## 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

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### \*Źródło

orphanet