## 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 <i>ATP2A1</i> 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