

# Miopatia Brody'ego

Kod Orpha: 53347 Kod OMIM: 601003

## 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

## Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.