

## Opis choroby \*

### Definicja

Spinal muscular atrophy-progressive myoclonic epilepsy syndrome is characterized by hereditary myoclonus and progressive distal muscular atrophy. Less than 10 cases have been reported. Treatment with clonazepam results in complete and lasting improvement of the myoclonus.

### Dane

#### Klasyfikacja

Choroba

#### Synonimy

Hereditary myoclonus-progressive distal muscular atrophy syndrome  
Zespół Jankovica i Rivera  
Jankovic-Rivera syndrome  
SMA-PME

#### Kod ORPHA

2590

#### Kod OMIM

159950

#### Kod ICD10

G25.3

#### Kod ICD11

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#### \*Źródło

orphanet