

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