

Zespół Ehlersa i Danlosa miopatyczny

Kod Orpha: 536516 Kod OMIM:

Opis choroby *

Definicja

A rare systemic disease characterized by congenital muscle hypotonia and/or muscle atrophy that improves with age, proximal joint contractures (knee, hip, elbow), and hypermobility of distal joints. Additional features include soft, doughy skin, atrophic scarring, delayed motor development, and myopathic findings in muscle biopsy. Abnormal craniofacial features have been reported in some patients. Molecular testing is obligatory to confirm the diagnosis.

Dane

Klasyfikacja

Choroba

Synonimy

EDS/myopathy overlap syndrome

Myopathic EDS

EDS/myopathy overlap syndrome

Myopathic EDS

Kod ORPHA

536516

Kod OMIM

-

Kod ICD10

Q79.6

Kod ICD11

-

[*Źródło](#)

orphanet

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.