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
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*Źródło

orphanet