

## 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  
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Myopathic EDS

#### Kod ORPHA

536516

#### Kod OMIM

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#### Kod ICD10

Q79.6

#### Kod ICD11

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#### [\\*Źródło](#)

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