# DISEASE:
# Myopathic Ehlers-Danlos syndrome

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| **NAME:** | Myopathic Ehlers-Danlos syndrome |
| **DESCRIPTION:** | 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. |
| **ORPHACODE:** | 536516 |
| **SYNONYMS:** | EDS/myopathy overlap syndrome<br>Myopathic EDS |
| **XREF(S):** | Orphanet |
| **ANALYTE(S):** | COL12A1 |
| **CREATED:** | 17 Jul 2019 - 01:41 |
| **CHANGED:** | 22 Jun 2023 - 16:14 |

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# RELATED CONTENT

## Related Genetic Tests

- Ehlers-Danlos syndroom, EDS (gene panel)

## Related Laboratories

- Centrum Medische Genetica - UZ Gent

## Related Analytes

- collagen type XII alpha 1 chain

## Related Gene Panels

- Ehlers-Danlos syndrome -UGent

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