Opis choroby *

Definicja

Congenital muscular dystrophy with hyperlaxity is a rare, genetic neuromuscular disease characterized by congenital hypotonia, generalized, slowly progressive muscular weakness, and proximal joint contractures with distal joint hypermobility and hyperlaxity. Scoliosis or rigidity of the spine and delayed motor milestones are also frequently reported. Other manifestations include a long myopathic face and, in rare cases, respiratory failure, mild to moderate intellectual deficiency and short stature. Ambulation may be impaired with time.

Dane

Klasyfikacja
Choroba

Synonimy
CMDH
CMDH

Kod ORPHA
371007

Kod OMIM
-

Kod ICD10
G71.2

Kod ICD11
-

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*Źródło

orphanet