Opis choroby *

Definicja

A rare congenital muscular dystrophy characterized by prominent axial hypotonia, predominantly proximal muscle weakness in upper limbs and distal in lower limbs, joint contractures (initially distal, later proximal), spinal rigidity, and progressive respiratory insufficiency, in the presence of moderately elevated serum creatine kinase. Cardiac arrhythmias and sudden death have also been reported.

Dane

Klasyfikacja
Choroba

Synonimy
L-CMD
L-CMD
Wrodzona dystrofia mięśniowa związana z LMNA
LMNA-related congenital muscular dystrophy

Kod ORPHA
157973

Kod OMIM
613205

Kod ICD10
G71.2

Kod ICD11
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*Źródło

orphanet