Opis choroby *

Definicja

Congenital muscular dystrophy type 1B is a rare, genetic neuromuscular disorder characterized by proximal and symmetrical muscle weakness (particularly of neck, sternomastoid, facial and diaphragm muscles), spinal rigidity, joint contractures (Achilles tendon, elbows, hands), generalized muscle hypertrophy and early respiratory failure (usually in the first decade of life). Patients typically present delayed motor milestones and grossly elevated serum creatine kinase levels, and with disease progression, forced expiratory abdominal squeeze and nocturnal hypoventilation.

Dane

Klasyfikacja
Choroba

Synonimy
CMD1B
CMD1B
MDC1B
MDC1B

Kod ORPHA
98893

Kod OMIM
604801

Kod ICD10
G71.2

Kod ICD11
8C70.6

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*Źródło

orphanet