Opis choroby *

Definicja

A rare congenital muscular dystrophy characterized by neonatal hypotonia, life-threatening respiratory failure, and feeding difficulties, furthermore by delayed motor development, severe muscle weakness predominantly affecting axial muscles (leading to poor head control, rigid cervical spine, and severe scoliosis), generalized joint laxity with no or mild contractures, as well as dry skin with follicular hyperkeratosis. Serum creatine kinase is normal or slightly elevated. Muscle biopsy shows fiber size variability, rounded fibers with mild increase of endomysial connective tissue and adipose replacement, abundant minicore lesions, increase of centrally located nuclei, angular fibers, and cap lesions.

Dane

Klasyfikacja
Choroba

Synonimy
Congenital muscular dystrophy, Davignon-Chauveau type
Wrodzona dystrofia mięśniowa, typ Davignon i Chauveau

Kod ORPHA
486815

Kod OMIM
617066

Kod ICD10
G71.0

Kod ICD11
-

---

*Źródło

orphanet