Opis choroby *

Definicja

Congenital muscular dystrophy with integrin alpha-7 deficiency is a rare, genetic, congenital muscular dystrophy due to extracellular matrix protein anomaly characterized by early motor development delay and muscle weakness with mild elevation of serum creatine kinase, that may be followed by progressive disease course with predominantly proximal muscle weakness and atrophy, motor development regress, scoliosis and respiratory insufficiency.

Dane

Klasyfikacja
Choroba

Synonimy
Congenital muscular dystrophy with ITGA7 deficiency
Wrodzona dystrofia mięśniowa z niedoborem ITGA7

Kod ORPHA
34520

Kod OMIM
613204

Kod ICD10
G71.2

Kod ICD11
8C70.6

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*Źródło

orphanet