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

Synonimy

Choroba

Congenital muscular dystrophy with ITGA7

deficiency

Wrodzona dystrofia mięśniowa z niedoborem

ITGA7

Kod ORPHA

34520

Kod OMIM

Kod ICD10

20 613204

G71.2

Kod ICD11 8C70.6

*Źródło

orphanet