Congenital muscular dystrophy with integrin alpha-7 deficiency

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Congenital muscular dystrophy with integrin alpha-7 deficiency. ORPHA:34520

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.