Congenital muscular dystrophy with intellectual disability

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Congenital muscular dystrophy with intellectual disability. ORPHA:370968

Congenital muscular dystrophy with intellectual disability is a rare, genetic, congenital muscular dystrophy due to dystroglycanopathy disorder characterized by a wide phenotypic spectrum which includes hypotonia and muscular weakness present at birth or early infancy and delayed or arrested motor development, associated with mild to severe intellectual disability and variable brain abnormalities on neuroimaging studies. Feeding difficulties, joint and spinal deformities, respiratory insufficiency, and ocular anomalies (e.g. strabismus, retinal dystrophy, oculomotor apraxia) may be associated. Decreased or absent alpha-dystroglycan on immunohistochemical muscle staining and elevated serum creatine kinase are observed.