Congenital Muscular Dystrophy-Dystroglycanopathy with Mental Retardation Type B2

National Cancer Institute

Source

National Cancer Institute. Congenital Muscular Dystrophy-Dystroglycanopathy with Mental Retardation Type B2. NCI Thesaurus. Code C126690.

An autosomal recessive inherited congenital muscular dystrophy caused by mutations in the POMT2 gene. It is characterized by mental retardation and mild structural brain abnormalities resulting from defective glycosylation of alpha-dystroglycan.