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Muscular Dystrophy-Dystroglycanopathy (Limb-Girdle) Type C, 1

National Cancer Institute

Source

National Cancer Institute. *Muscular Dystrophy-Dystroglycanopathy (Limb-Girdle) Type C, 1*. NCI Thesaurus. Code C133730.

An autosomal recessive limb-girdle muscular dystrophy caused by mutations in the POMT1 gene, encoding protein O-mannosyl-transferase 1. It is characterized by mental retardation without structural brain abnormalities and limb-girdle muscular dystrophy.