Muscular Dystrophy-Dystroglycanopathy (Congenital with Brain and Eye Anomalies) Type A, 1

National Cancer Institute

Source

National Cancer Institute. Muscular Dystrophy-Dystroglycanopathy (Congenital with Brain and Eye Anomalies) Type A, 1. NCI Thesaurus. Code C128118.

An autosomal recessive muscular dystrophy caused by mutations in the POMT1 gene, encoding protein O-mannosyl-transferase 1. It is associated with characteristic brain and eye malformations, profound mental retardation, and early death.