Muscular Dystrophy-Dystroglycanopathy (Congenital with Brain and Eye Anomalies) Type A, 3

National Cancer Institute

Source

National Cancer Institute. Muscular Dystrophy-Dystroglycanopathy (Congenital with Brain and Eye Anomalies) Type A, 3. NCI Thesaurus. Code C126740.

An autosomal recessive muscular dystrophy caused by mutations in the POMGNT1 gene. It is associated with characteristic brain and eye malformations, profound mental retardation, and death usually in the first years of life.