MAN1B1-CDG

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. MAN1B1-CDG. ORPHA:397941

MAN1B1-CDG is a form of congenital disorders of N-linked glycosylation characterized by intellectual disability, delayed motor development, hypotonia and truncal obesity. Additional features include slight facial dysmorphism (hypertelorism, downslanting palpebral fissures, large, low-set ears, hypoplastic nasolabial fold, thin upper lip), hypermobility of the joints and skin laxity. The disease is caused by mutations in the gene MAN1B1 (9q34.3).