B4GALT1-CDG

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. B4GALT1-CDG. ORPHA:79332

B4GALT1-CDG is a congenital disorder of glycosylation characterised by macrocephaly due to Dandy-Walker malformation, hydrocephaly, hypotonia, myopathy and coagulation anomalies. To date, only one case has been reported. The syndrome is associated with mutations in the GALT1 gene (localised to region q13 of chromosome 9) leading to a deficiency in the Golgi apparatus enzyme beta-1,4-galactosyl transferase.