DISEASE:
B4GALT1-CDG

NAME:	B4GALT1-CDG
DESCRIPTION:	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.
ORPHACODE:	79332
SYNONYMS:	Beta-1,4-galactosyltransferase deficiency CDG syndrome type IId CDG-IId CDG2D Carbohydrate deficient glycoprotein syndrome type IId Congenital disorder of glycosylation type 2d Congenital disorder of glycosylation type IId
XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	B4GALT1

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