DISEASE:
Hypercoagulability syndrome due to glycosylphosphatidylinositol deficiency

NAME:	Hypercoagulability syndrome due to glycosylphosphatidylinositol deficiency
DESCRIPTION:	A rare congenital disorder of glycosylation characterized by cerebral and portal vein thrombosis, portal hypertension, macrocephaly, and persistent absence seizures. Additional reported features include mild to moderate global developmental delay and intellectual disability, as well as thrombocytopenia. Brain imaging may show variable stages of infarction and cerebral and cerebellar atrophy.
ORPHACODE:	83639
SYNONYMS:	Congenital disorder of glycosylation due to PIGM deficiency PIGM-CDG
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	PIGM PIGW
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