Hypercoagulability syndrome due to glycosylphosphatidylinositol deficiency

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Hypercoagulability syndrome due to glycosylphosphatidylinositol deficiency. ORPHA:83639

The combination of a propensity for venous thrombosis and seizures has been reported in two unrelated kindreds. Transmission is autosomal recessive. It results from a point mutation of PIGM, which reduces transcription of PIGM and blocks mannosylation of glycosylphosphatidylinositol (GPI), leading to partial but severe deficiency of GPI.