Factor V Leiden

Factor V Leiden (rs6025) is a variant (mutated form) of human factor V (one of several substances that helps blood clot), which causes an increase in blood clotting (hypercoagulability). With this mutation, protein C, an anticoagulant protein (which normally inhibits the pro-clotting activity of factor V), is not able to bind normally to Factor V, leading to a hypercoagulable state, i.e., an increased tendency for the patient to form abnormal and potentially harmful blood clots.

Factor V Leiden is the most common hereditary hypercoagulability (prone to clotting) disorder amongst ethnic Europeans.

It is named after the Dutch city Leiden, where it was first identified in 1994 by Prof R. Bertina et al [1].

Factor V Leiden (FVL) may be more common in patients with cerebral venous thrombosis (CVT). However, the association between FVL and CVT varied depending on the geographic origin of the studies [2].

Polymorphisms in the Factor V Leiden gene are associated with the atypical timing of IVH, suggesting an as yet unknown environmental trigger. The methylenetetrahydrofolate reductase (MTHFR) variants render neonates more vulnerable to cerebral injury in the presence of perinatal hypoxia. The present study demonstrates that the MTHFR 677C>T polymorphism and low 5-min Apgar score additively increase the risk of IVH. Finally, review of published preclinical data suggests the stressors of delivery result in hemorrhage in the presence of mutations in collagen 4A1, a major structural protein of the developing cerebral vasculature. Maternal genetics and fetal environment may also play a role [3].

1)
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2)
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3)
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