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Combined factor V and VIII deficiency (Review)

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Abstract

This review summarizes current data on the pathomechanisms and new genetic findings of combined factor V and VIII deficiency (CF5F8D). Congenital haemorrhagic disorders characterized by deficiency of two clotting factors comprise an interesting group. Among dual coagulation disorders, CF5F8D is the most common type. For the first time combined factor V and VIII deficiency (F5F8D) was reported by Oeri et al in 1954. That is distinct from the coinheritance of both FV deficiency (parahaemophilia) and FVIII deficiency (haemophilia A) that has been reported in four families. Individuals who present with this phenotype have between 5 and 30% of normal plasma levels of FV and FVIII antigen and activity, whereas the level of other plasma proteins are not altered. Total numbers of affected individuals are less than 150 cases all over the world. At first it was assumed that deficiency of protein C inhibitor was a responsible cause, but further investigations revealed that it was due to mutations called ERGIC-53 and LMAN-1.

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Author keywords

Factor V deficiency; Factor VIII deficiency; Hemorrhagic disorder

Indexed Keywords

EMTREE drug terms: binding protein; blood clotting factor 5; blood clotting factor 8 concentrate; desmopressin; endoplasmic reticulum golgi intermediate compartment protein 53; lectine mannose binding protein 1; unclassified drug

EMTREE medical terms: bleeding; blood clotting factor 5 deficiency; demography; gene mutation; genetics; hemophilia A; human; inheritance; pathogenesis; phenotype; plasma transfusion; review

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