Considerations on Combined Factor VII and Factor VIII Defect

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We have read with great interest the paper by Machin and Muller [5] on a new family with combined factor VII and factor VIII deficiency. It always gives a great satisfaction to find a confirmation of personal observations made years ago [2].

The family presented by the authors seems a case of type II combined factor VII and factor VIII deficiency. The term type II should be limited to those patients showing a similar depression in factor VII and factor VIII and no heterozygosis for factor VII deficiency in the relatives. On the contrary, type I combined deficiency should be limited to those cases representing a causal association of heterozygosis for factor VII deficiency and hemophilia A [4]. The patient studied by Gaston et al. [1] belongs perhaps to this

Table I. Tentative classification of combined deficiency of factor VII and factor VIII in two groups

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group. We say 'perhaps' because, unfortunately, no family members were studied. The main features of the two forms are summarized in table I.

A similar classification has been proposed for combined factor V and factor VIII hereditary transmission [3]. At this stage, at least four genes or systems of genes seem involved, namely: (1) An x-linked gene controlling factor VIII activation, deficient or abnormal in hemophilia A. (2) An auto-somal gene controlling protein synthesis, deficient or abnormal in von Willebrand's disease. (3) An autosomal recessive gene controlling factor V and factor VIII activation, deficient or abnormal in combined factor V and factor VIII defect. (4) An auto-somal-dominant gene controlling factor VII and factor VIII activation, deficient or abnormal in combined factor VII and factor VIII activation. The genetic plot has really thickened.

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