Spontaneous Haemophilia in a Genotypically Normal Female -A Family Study

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The diagnosis of spontaneous haemophilia A with severe antihæmophilic globulin deficiency (less than 1 %) was established in an 11-year-old girl with a normal female chromosomal pattern. Coagulation studies proved that the proposita is the offspring of a normal father with negative history for bleeding disorders and a normal mother who had an antihæmophilic globulin concentration of 200%. Family history revealed no bleeding disorder in 3 successive generations of the maternal side. This together with AHF activity exceeding slightly AHF-like antigen concentration provides good evidence against the carrier state of the mother. The possible mechanisms through which spontaneous haemophilia may occur in a genotypically normal female are discussed.