Pathogenesis and Diagnostics Factors of von Willebrand Disease

Abstrak :

von Willebrand disease (vWD) is an autosomal inherited bleeding disorder caused by a deficiency or abnormality of von Willebrand factor (vWF). vWF is a large multimeric glycoprotein that mediates platelet adhesion at the site of vessel injury. It also protects factor VIII from proteolytic degradation in the circulation. vWD has a prevalence of about 1% in the general population but less than 10% have bleeding symptoms. Bleeding symptoms are usually mucocutaneous and post surgical with varying severity. This disorder can result from either a quantitative (types 1 and 3) or qualitative (type 2) defect in vWF. Type 2 vWD has been further classified into four distinct subtypes; 2A, 2B, 2M and 2N. The diagnosis of vWD requires attention to personal and family history of excessive bleeding and confirmation by laboratory evaluation. A mild chronic thrombocytopenia is often seen in type 2B vWD. Patients with mild vWD often have both a normal bleeding time and normal APTT. Specific tests for vWD diagnosis involve vWF antigen level, vWF activity (ristocetin cofactor), and factor VIII activity. Once a diagnosis is established, additional tests that aid in classifying the type of vWD include ristocetin-induced platelet aggregation and vWF multimer analysis.

Keyword :

Diagnostics factor, von Willebrand disease.