

Genetics of Bleeding Disorders

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ABSTRACT Genetic bleeding disorders form an important presentation among various genetic disorders occurring in children. A prevalence of 6 per 100,000 population has been reported in India. Bleeding disorders constitute a heterogeneous group of disorders with varying clinical presentations. The common bleeding disorders are Hemophilia A and B, von Willebrand disease, and inherited thrombocytopenias. In addition, there are a number of other defects of coagulation pathway and platelet function, about which only limited knowledge is available. This review article attempts to organize the information available regarding the clinical presentations, molecular defects and methods available for molecular diagnosis of these disorders.