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How to estimate bleeding risk in mild bleeding disorders

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Abstract

The concept of mild bleeding disorders (MBD) has evolved in contrast to severe hemophilia A and B to indicate less severe disorders, characterized by the presence of more frequent and/or more prominent bleeding symptoms than in the normal population. These symptoms occur mostly after a recognizable challenge and do not lead to major discomfort or organ damage, even in the absence of specific medical intervention. However, it has become clear that, from the most severe to the mildest hemostatic disorders, there is a continuous spectrum of bleeding manifestations, which overlap with the occasional bleeding occurring in people without any identifiable hemostatic abnormality. By reviewing the principal hemorrhagic disorders we have tried to identify those entities that could fit a diagnosis of MBD and result, at the same time, in a net benefit for treatment or prophylaxis of patients rather than being simply accurate. This goal can usually be achieved by comparing the patient's phenotype with known nosological entities. However, limitations of this approach are evident, considering the paucity of clinical data and the biases of most published reports on the different disorders. In addition, in a partial deficiency of a clotting factor, a reliable relationship between the residual activity and bleeding severity is not invariably found. Molecular characterization of the defects is also generally useless. Accordingly, an accurate bleeding history in the propositus and his/her family remains of major importance. For this purpose, new standardized and possibly quantitative tools are being developed in several institutions. Innovative approaches, combining into a single probability phenotypic and genetic data, could possibly estimate better the bleeding risk in specific disorders.