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Evidence-based diagnosis of type 1 von Willebrand disease: a Bayes theorem approach

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Abstract

The diagnosis of type 1 von Willebrand disease (VWD) is based on the presence of bleeding symptoms, reduced von Willebrand factor (VWF) levels, and autosomal inheritance of the phenotype. To better appreciate the contribution of clinical and laboratory data to the final diagnosis of VWD, we computed the likelihoods of having VWD as a function of the bleeding score (LR(score)), of VWF level (LR(VWF)), and of number of first-degree family members with reduced VWF levels (LR(family)). The 3 likelihoods were therefore combined using the Bayes theorem, giving the final probability (odds) of having VWD. LR(family) and LR(VWF) were the 2 factors mostly influencing the final probability of having VWD. Data from the present study provide an evidence-based description of the minimal criteria for the diagnosis of type 1 VWD. As an example, presence of VWF levels lower than 40 IU/dL in at least 2 family members (including the proband) and a bleeding score of at least 1 were found to be required for a final odd of VWD higher than 2.0 (false-positive rate less than one-half). Validation of this approach and of its clinical utility is, however, required by analysis in other cohorts of well-characterized type 1 VWD patients.

Link all'articolo: <https://pubmed.ncbi.nlm.nih.gov/18187661/>