Von Willebrand's disease (VWD) is an autosomally inherited bleeding disorder caused by a deficiency or abnormality of von Willebrand factor (VWF). Castaman and co-workers have recently reviewed the molecular basis of VWD in this journal. VWD has a prevalence of about 1% in the general population, but the figure for clinically relevant cases is lower (about 100/million inhabitants). Most cases appear to have a partial quantitative deficiency of VWF (type 1 VWD) with variable bleeding tendency, whereas qualitative variants (type 2 VWD), due to a dysfunctional VWF, are clinically more homogeneous. Type 3 VWD is rare and the patients have a moderate to severe bleeding diathesis because of the virtual absence of VWF, and a recessive pattern of inheritance.

In this issue, Hilbert and co-worker report studies on a new mutation in the VWF gene that causes type 2A VWD. These patients may be poorly responsive to desmopressin and should receive FVIII/VWF concentrates in cases of prolonged mucosal bleeding and major surgery. This study provides an example of the relevance of molecular studies for clinical practice.

Other studies on VWF or VWD recently published in this journal are listed below.1–8

References