



VON WILLEBRAND DISEASE PROFILE

*BloodCenter of Wisconsin
Hemostasis Reference Laboratory specializes
in comprehensive testing for von Willebrand disease.*

BACKGROUND:

von Willebrand disease (VWD) is a common bleeding disorder characterized by either quantitative or qualitative defects of von Willebrand factor (VWF). Screening tests for VWD include Factor VIII activity, VWF antigen, and VWF GPIbM activity. Discrepancies between VWF antigen and activity levels suggest a variant of VWD. Correct diagnosis of variant VWD is essential to providing effective treatment.

Profile Tests:

von Willebrand Profile

Includes VWF Antigen, VWF GPIbM Activity, VWF Multimers, and Factor VIII Activity. This set of tests provides sufficient information to support a clinical diagnosis of VWD.

Additional testing for classification of variant VWD

VWF Collagen Binding (VWF:CB)

In vivo, collagen binding by VWF facilitates platelet adhesion at sites of blood vessel injury. The collagen binding assay tests this capacity, and discrepancy between VWF:CB and VWF antigen levels is a sensitive screen for von Willebrand disease subtypes in which large molecular weight multimers are missing (type 2A, 2B and platelet-type VWD).

VWD Type 2N Binding (VWF-FVIII Binding)

In Type 2N VWD, the binding of factor VIII to VWF is reduced. In patients with reduced levels of factor VIII, the Type 2N Binding Assay differentiates type 2N VWD from mild hemophilia A or hemophilia A carrier state.

VWD Type 2B Binding (VWF-Platelet Binding)

Type 2B VWD is caused by an increase in VWF binding to platelets, and a resulting loss of larger VWF multimers. The Type 2B Binding Assay differentiates type 2B VWD from platelet-type VWD.

VWF Propeptide Antigen

Patients with increased clearance of VWF will have normal VWF propeptide levels and a VWF antigen level that is decreased compared to the VWF propeptide antigen. This has been observed in some congenital von Willebrand disease variants (1C, 2A, 2B) and in a subset of individuals with acquired von Willebrand disease.

Unlike Factor VIII inhibitors in hemophilia A, VWF antibodies usually cause increased clearance of VWF from circulation rather than inhibiting VWF function. Therefore, most VWF antibodies are not detected in the standard mixing studies used for coagulation factor inhibitors. Measurement of VWF propeptide is helpful in diagnosing some cases of acquired von Willebrand disease.

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Sequence Analysis

Some cases of variant VWD may require DNA sequencing to confirm the diagnosis. Type 2N Sequence Analysis will identify pathogenic variants in the *VWF* exons that encode factor VIII binding domains. *VWF* Exon 28 Sequence Analysis will identify all pathogenic variants associated with type 2B, the vast majority associated with 2M, and 70-80% of pathogenic variants associated with type 2A VWD. Additional sequencing for type 2A VWD is available. VWD Platelet-Type Sequence Analysis identifies pathogenic variants in *GP1BA* platelet glycoprotein Ib alpha associated with platelet type VWD, and VWD Type 1C Sequence Analysis identifies pathogenic variants in *VWF* associated with the 1C phenotype.

Other Tests for VWD

VWF Inhibitor
VWF Propeptide Antigen
VWF Ristocetin Cofactor Activity

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