

# Von Willebrand disease

**Von Willebrand disease** (vWD) is an inherited autosomal disorder with a wide phenotypic spectrum, and variability in its clinical presentation.

The disease is caused by the quantitative deficiency or dysfunction of von Willebrand factor (vWF).

Von Willebrand factor has two main functions in haemostasis:

- It is an adhesion protein in plasma, essential for platelet-plug formation and diverting circulating platelets to the sites of vascular injury, particularly through larger multimers.
- It forms a non-covalent complex with Factor VIII in plasma, protecting FVIII from inactivation and clearance.

Patients with von Willebrand disease may notice frequent or prolonged epistaxis, easy bruising, menorrhagia, and excessive or prolonged bleeding after surgery. They may also describe a family history of bleeding symptoms.

## Von Willebrand disease

<b>Type 1</b>	Partial quantitative deficiency of vWF
<b>Type 2</b>	Qualitative defects of vWF (abnormal function)
<b>2A</b>	Defective vWF functions, associated with reduced/absent larger multimers
<b>2B</b>	Increased vWF affinity for platelets associated with reduced/absent larger multimers
<b>2M</b>	Reduced vWF interaction with platelets and collagen not associated with reduction in larger multimers
<b>2N</b>	Defective vWF binding to Factor VIII
<b>Type 3</b>	Severe or complete deficiency of vWF

## Laboratory tests

Coagulation studies (a normal result does not exclude vWD)

Platelet count

Von Willebrand studies, which can include:

- Factor VIII
- vWF antigen
- vWF activity
- vWF collagen binding

Other tests may be used in order to sub-classify vWD, including:

- Ristocetin-induced platelet aggregation
- vWF multimer analysis
- Factor VIII binding assay

Repeat testing is often advised as vWF is an acute phase reactant and levels fluctuate. Elevated levels can be seen with inflammation, infection, exercise, stress, pregnancy, and OCP use.

Collection at a designated SNP von Willebrand collection centre is recommended. A list of locations is available on the SNP website: [snp.com.au/locations](http://snp.com.au/locations) (use “von Willebrand Screen” specialised test filter).