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, thereby protecting it from being inactivated and cleared.

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

Type 1	Partial quantitative deficiency of qualitatively normal vWF
Type 2	Qualitative defects of vWF (abnormal function)
2A	Defective platelet-dependent vWF functions, associated with lack of larger multimers
2B	Heightened platelet-dependent vWF functions, associated with lack of larger multimers
2M	Defective platelet-dependent vWF functions, not associated with lack of larger multimers
2N	Defective vWF binding to factor VIII
Type 3	Severe or complete deficiency of vWF

Laboratory tests

Coagulation studies (a normal result does not exclude vWD)

Platelet Function Analysis (PFA)

Blood group and von Willebrand studies, which include:

- Factor VIII
- vWF Antigen
- vWF Ristocetin cofactor activity
- vWF Collagen Binding activity

(Medicare Australia requires each of these tests to be ordered individually)

Uncommonly, other tests may be used in order to sub-classify vWD:

- 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.