Platelet-Type von Willebrand Disease

National Cancer Institute

Source

National Cancer Institute. Platelet-Type von Willebrand Disease. NCI Thesaurus. Code C131681.

A rare autosomal dominant bleeding disorder characterized by abnormally enhanced binding of von Willebrand factor (VWF) by the platelet glycoprotein Ib receptor complex. Hemostatic function is impaired due to the removal of VWF multimers from the circulation. It is due to a mutation in the gene encoding for platelet glycoprotein Ib alpha, resulting in enhanced affinity for VWF. It is often misdiagnosed as type 2B von Willebrand disease due to similarities between these two conditions. Patients present with a mild thrombocytopenia with large platelets. Platelet aggregates are often visible in blood smears.