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Perspective

Differential Diagnosis and Brief Note on Von Willebrand Disease

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DESCRIPTION

Von Willebrand Disease (vWD) is a hemorrhagic condition characterized by a deficit or malfunction of the protein Von Willebrand Factor (*vWF*), which is genetically and clinically diverse. Primary hemostasis is compromised in vWD due to a dysfunctional *vWF* interaction between platelets and the vessel wall.

vWF is a large, multimeric glycoprotein that circulates in the plasma at around 10 mg/ml. *vWF* is released from platelets and endothelial cells' storage granules in response to a variety of triggers. It has two key functions in hemostasis. It first facilitates platelet adherence to vascular damage sites. Second, it attaches to the procoagulant protein factor VIII and stabilizes it (FVIII).

The three major categories of vWD are as follows:

- Type I: Partial quantitative *vWF* insufficiency
- Type II: Qualitative *vWF* deficiency
- Type III: Total quantitative *vWF* depletion

In the vast majority of instances, vWD is a genetic disorder. The *vWF* gene is found on chromosome 12 near the end of the short arm. The gene consists of 52 exons and spans 180 kb of the human genome, making it comparable in size to the FVIII gene. Megakaryocytes, endothelial cells, and potentially placental syncytiotrophoblasts are the only cells that express the *vWF* gene. On chromosome 22, there is a partial, nonfunctional duplication (pseudogene).

Type I vWD

A mild to severe quantitative shortage of *vWF* is caused by vWD type I. (i.e., about 20%-50% of normal levels). Type I is normally autosomal dominant in those with *vWF* levels less than 0.3 IU/ml; in people with levels greater than 0.3 IU/ml, mutations have variable penetrance.

Type II vWD

vWD type II is classified into kinds IIA, IIB, IIN, and IIM and is caused by qualitative *vWF* aberrations. The most prevalent qualitative aberration of *vWF*, vWD type IIA, is linked to the loss of big and medium-sized multimers selectively. The majority of instances are autosomal dominant.

The loss of big multimers in vWD type IIB occurs by a process different from that of type IIA. To date, observations have identified a key element of *vWF* that is implicated in *vWF* binding to the platelet receptor Glycoprotein Ib (GpIb). Each of the single amino acid alterations identified is thought to result in an increase of function, allowing *vWF* to bind to platelets spontaneously.

Typically, plasma *vWF* has little interaction with platelets until it comes into contact with an exposed subendothelial surface. Binding of *vWF* to collagen and/or other ligands within the wounded vessel wall is thought to cause a subsequent morphological shift, which enhances binding to the GpIb receptor.

Type III vWD

Patients with vWD type III, a significant statistical deficiency with little or no detectable plasma or platelet *vWF*, suffer from extreme bleeding problem. vWD type III appears to be caused by both parents inheriting a mutant *vWF* gene. In the clearest hypothesis, vWD type I would simply be the heterozygous version of vWD type III; however, inheritance patterns suggest that there is more complexity.

vWD type III is considerably more unusual than the model's projected incidence of 1 case per 40,000 people, with a frequency closer to 1 case per 1 million people. Although few mutations have been found in families with pure vWD type I, it has been theorized that certain vWD type I cases are caused by a mutant *vWF* subunit that interferes in a dominant, negative way with the normal allele, resulting in autosomal dominant inheritance.

Acquired vWD.

The formation of antibodies against *vWF* causes acquired vWD, an uncommon condition. Acquired vWD can be associated with

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a number of conditions, including lymphoproliferative, cardiovascular, and myeloproliferative illnesses, and it usually resolves after the disease is treated.

Diagnosis

Because bleeding is widespread and does not always imply an illness, mild types of von Willebrand disease can be difficult to detect. If your doctor suggests you have a bleeding issue, he or she may recommend you to a specialist in blood disorders (hematologist).

Your doctor will likely ask you specific questions about your medical history and check for bruises or other evidence of recent bleeding to determine if you have von willebrand disease.

- This test measures a specific protein to detect the level of von willebrand factor in your blood.
- A range of tests are available to determine how well the von willebrand factor functions in your coagulation process.
- This test determines whether your factor VIII levels and activity are abnormally low.
- This test examines the structure of von willebrand factor in your blood, as well as its protein complexes and how its molecules degrade. This data aids in determining the type of von willebrand disease you have.

Due to circumstances such as stress, exercise, infection, pregnancy, and drugs, the findings of these tests can change over time in the same person.