3.1.4

Inherited (Primary) Disorders Involving Platelets

Von Willebrand Disease

Von Willebrand disease is the most common hereditary blood-clotting disorder. People with von Willebrand disease have low levels of vWF. Symptoms of this condition include easy bruising, frequent nose bleeds, bleeding gums, heavy menstrual periods, and more than average blood loss during childbirth. There are a variety of mutations that may arise in the vWF gene which lead to different types of this disease. Type I von Willebrand disease is the most common type (85%) and involves production of vWF that works correctly, but there just isn't enough of it found in the blood. Type II is less common (15%) and involves the production of vWF that is not functional.

One important treatment for type I von Willebrand disease is **desmopressin acetate**. This is also known as D-amino D-arginine vasopressin (**DDAVP**). DDAVP binds to the antidiuretic hormone (or vasopressin) receptors called V2 receptors on endothelial cells and causes them to release 3-5 times more vWF than normal. This treatment is not helpful with type II von Willebrand disease because the vWF in those cases is not functional, so releasing more of it through the use of DDAVP is not effective.

Genetic Deficiencies with Platelets

There are some genetic deficiencies of certain platelet receptors that bring about bleeding disorders. GPIIb/IIIa (fibrinogen receptor) deficiency is known as **Glanzmann thrombasthenia**. A deficiency of GP1b (vWF receptor) is known as **Bernard-Soulier syndrome**. Both lead to less blood coagulation than normal and increased bleeding risk.