Von Willebrand disease:

الموضوع الرئيسي 1	most common inherited bleeding disorder increase in aPTT normal platelet count secondary decrease in factor VIII levels genetic mutations result in inherited deficiency — Gene is located on chromosome 12

الموضوع الرئيسي 2

Genatic

	Type-1	Type-3	Type-2
	autosomal dominant level of vWF in the blood range from 20%-50% of norma accounts for 70% of all cases reduced quantity of circulating vWF	autosomal recessive due to deletions or frameshift mutations with total deficiency for 5-10% of the cases reduced quantity of circulating vWF	autosomal dominant due to missense mutations resulting in nonfunctional vWF levels Accounts for 20% of all cases. associated with qualitative defects in vWF

Acquired vWD

This type of vWD in adults	
results after a diagnosis of an autoimmune disease, such as SLE, or from heart disease or some types of cancer	
Also, it can also occur after taking certain medications.	

Classic Hemophilia

	normal platelet count Normal bleeding time prolonged aPTT treated by: blood transfusion of concentrated plasma fraction containing factor VIII (with its associated dangers: a- Hepatitis or HIV/AIDS; b- Possibility of patients making auto-antibodies) treated by: cloning and expression of the gene for factorVIII(protein). (Through the DNA recombinant technology, the pure protein can be isolated and administered to patients with none of those dangers.)
Hemophilia A	most common blood clotting defect-permanent tendency for hemorrhage due to missing factor VIII of the intrinsic pathway or marked reduction of its activity X-linked recessive disorder The blood level of factor VIII in severe hemophilia A patient is less than 5% of normal. due to: an inversion mutation in intron 1 (5%) or 22 (45%) due to: Nonsense/stop mutations prevent factor production due to: Missense mutations may affect factor production , activity or half-life.
Hemophilia B	factor IX deficiency X-linked recessive disorder due: Most cases associated with point mutations due: Deletions in about 3% of cases due: Promoter mutations in about 2%
Hemophilia C	factor XI deficiency autosomal recessive disorder
Parahemophilia	deficiency of factor V autosomal recessive disorder

Thrombosis

Four primary influences that contribute to the pathogenesis:	a- Endothelial injury (dominant) b- Abnormal blood flow c- Hypercoagulability (less) d- Alteration of the coagulation pathways
الموضوع الرئيسي 3	young adults or teenagers develop venous thrombosis heterozygous for the deficiency with levels of functional protein C of 40 - 65%.
الموضوع الرئيسي 2	Defects of → protein C pathway mutation → changed amino acid serine into proline at position 270) → Protein C is involved in deactivation of blood] clotting factors (Va and VIIIa) Defects of → Factor V mutations → amino acid arginine 506 into glutamine; rendering factor V resistant to cleavage by protein C Defects of → prothrombin mutations → are common genetic risk factors for venous thrombosis.