Congenital coagulation disorder	Genetics	Presentation	Lab tests	Notes
Factor VIII deficiency (hemophilia A)	Inversion of intron 22 X-linked	Acute hemarthrosis (protease destruction of cartilage), chronic hemophilic arthropathy (subchondral cysts & loss of range of motion), intramusc hem, CNS bleeding, post-dental/surgical bleeding, naso-oral mucosal bleeds	Prolonged APTT, specific clot factor assay, normal PT, BT	Routine venepuctures, immuizations ok Factor replacement for invasive procedures X-lyonization
Factor IX deficiency (hemophilia B)	X-linked	Same as hemophilia A	Same as hem A	Same as hem A
Factor XI deficiency (hemophilia C)	Not sex-linked	Hemor after surgical or traumatic provocation, hemarthroses and intraarticular bleeds rare	Prolonged APTT, normal PT/TCT	Ashkenazi jews
Von Willebrand disease	Type 1	Mucocutaneous bleeding, superficial bruising, gingival bleeding, epistaxis, menorrhagia, GI bleeding, post-dental bleeding, post partum hemorrhage	Low ristocetin cofactor, low vWF ag & FVIII, normal vWf multimers, prolonged BT, APTT	*Partial deficiency*, auto dom Treat w/ DDAVP (vassopressin analogue), estrogen (increase endothelial synth) and cryo (vWF, FVIII, fibrinogen, fibronectin)
	Type 2A		Severely reduced ristocetin cofactor	*No large vWF multimers* Treat w/ cryo
	Type 2B		High ristocetin induced platelet aggreg (RIPA), low risto cofac, low ag	*Gain of function*, increased binding of vWF to GPIb Treat w/ cryo
	Type 2M		Low ristocetin cofactor	Lots of *uncleaved pro-vWF* Treat w/ cryo
	Type 2N		Normal vWF ag & ristocetin cofactor, low FVIII activity	*Defect in FVIII binding*, auto rec, like mild hem A Treat w/ cryo
	Type 3		Undetectable risto cofac & vWF, low FVIII	*Complete deficiency in vWF* function & ag, auto rec

Acquired bleeding disorder	Etiology	Pathology	Lab tests	Treatment
DIC	Acute (hemorrhagic): infec, obstretical, malignancy (APL), tissue injury (snake bite, heat stroke, aortic aneurysm, hemolytic transfusion reaction), homo prot C/S def, HITTS Subacute (prothrombotic): malignancy, obstretical, vascular	Loss of balance between clot promoting (thrombin) and lysing (plasmin) systems in vivo	D-dimer (gold standard), fibrin degradation products (FDP), fibrin monomer (weak), abnormal APTT, PT, platelet count, fibrinogen	Treat underlying condition Replacement therapy, heparin therapy for thrombosis
Liver disease	Alcoholism, immunologic, toxin, viral	All coag prot made in liver Structural manifestations promote bleeding, causing portal hypertension, varices, gastritis, hemmorhoids, platelet destruction	Prolonged APPT, PT, BT, TCT	
Vitamin K deficiency	Nutritionally depleted alcoholics, warfarin, abx (interfere w/ intestinal bact synth and absorption of vit K)	Vit K cofactor of enzymes that carboxylate a.a. in factors II, VII, IX, X, protein C/S, allowing them to bind to cell membranes		
Massive transfusion		Plasma and platelet dilution, increased anticoag sodium citrate dextrose and calcium depletion		FFP, calcium
Dysfibrinogemias	Acquired liver disease	Abnormal fibrinogens	Prolonged TCT, APTT, PT	