# Bleeding Disorders (Diatheses)



## Congenital bleeding disorders

***Von Willebrand's disease*** - most common 1:1000. Mainly AD. M=F. Usually mild without spont bleeding. ↓Prod or abnormality of vWF (req for normal plt fn and stabilisation of factor VIII).
***Haemophilia A (factor VIII deficiency) & B (factor IX deficiency or Christmas disease)*** - X-linked recessive. Incid: A > B (1:10,000 vs 1:60,000). Severe (<2% factor – spont muscle & jt bleeding → to crippling joint disease), mod (2-5%) & mild (bleeding only post trauma or Sx).
***Other genetic disorders*** – rarer, AR e.g. prothrombin (factor II) deficiency is 1:2,000,000.
***Glanzmann's thrombasthenia and Bernard-Soulier's disease*** - Rare AR diseases affecting platelet membrane glycoproteins and causing abnormal platelet binding and aggregation

## Acquired disorders

- Liver disease and cirrhosis → ↓synthesis of clotting proteins and thrombocytopenia.
- Vit K def from dietary def, GI malabsorption or absence of gut bacteria in newborns.
- Shock, sepsis or malignancy → ↑bleeding tendency, often by DIC
- Renal disease causes platelet dysfunction and reduced aggregation.
- AutoAb to coag factors (e.g. in lymphoma and SLE) or to plts (as in ITP).
- Amyloidosis where factor X deficiency occurs as well as infiltration of blood vessels.
- Old age, prolonged steroid use and vitamin C deficiency all ↓integrity of blood vessel wall.

## Presentation

*Haemophilias* – onset in early childhood, dental bleeds, haematomas & haemarthrosis.
*vWD* – often asymptomatic, post-surg, mucocutaneous bleeding common. Spont. bleeding rare.

## Investigations

*Bloods:* FBC and film, APPT (↑with factor VIII and IX def), INR (↑ with factors I, II, V, VII and X def), fibrinogen, FDPs, bleeding time (↑ with vWD)
*Gene & specific factors assays*: VIII & IX for haemophilias; VIII & vWF & ristocetin in vWD

| Acquired Disease | Platelet Count | INR | APTT | Thrombin time |
|---|---|---|---|---|
| Liver disease | Low | Prolonged | Prolonged | Normal (rarely prolonged) |
| DIC | Low | Prolonged | Prolonged | Grossly prolonged |
| Massive transfusion | Low | Prolonged | Prolonged | Normal |
| Oral anticoagulants | Normal | Grossly prolonged | Prolonged | Normal |
| Heparin | Normal (rarely low) | Mildly prolonged | Prolonged | Prolonged |
| Circulating anticoagulant | Normal | Normal or prolonged | Prolonged | Normal |

## Management

Haemophilia

- Care via a regional haemophilia centre.
- Early factor VIII (1U/kg →↑level by 2%) or IX (1U/kg →↑level by 1%) transfusions if bleeding, aim for 30-40% in mild, >50% in mod & 100% in sev bleeding.
- Symptomatic relief - analgesia, rest, physiotherapy etc - for muscle/jt bleeds.
- Prophylaxis: **Tranexamic acid** may prevent bleeding after minor ops in those with mild haemophilia A/B or **desmopressin** (DDAVP) for mild or moderate haemophilia A only.
- Gene therapy – not yet.

Von Willebrand's disease

- Mild cases are treated with **desmopressin**, **tranexamic acid** and OCP for menorrhagia.
- Severe cases may need vWF concentrate.
- Avoid NSAIDs and aspirin

Acute ITP - rarely needs treating but steroids may be useful.
If inherited disorder: genetic counselling and prenatal diagnosis.