Thrombophilia

Disorders of haemostasis that predispose to thrombosis (prothrombotic states).

Causes

Hereditary

- Factor V Leiden mutation Activated protein C (APC) resistance: commonest inherited
- Protein C deficiency
- Protein S deficiency
- Antithrombin III deficiency
- Hyperhomocysteinaemia
- Prothrombin G200210A polymorphism
- Elevated levels of Factors I, II, VIII, IX and XI
- Elevated levels of von-Willibrand Factor (vWF)
- Dysfibrinogenaemias

Acquired

- Antiphospholipid antibodies, e.g. antiphospholipid syndrome, SLE or Hughes' Syndrome
- Cancer (increases risk x 7)
- Hyperhomocysteinaemia due to mild folic acid, Vitamin B12 or B6 deficiency
- Some persistent inflammatory conditions e.g. IDB
- Myeloproliferative disorders.
- Thrombocytosis
- Polycythaemia
- Paroxysmal nocturnal haemoglobinuria
- Behçet's disease
- Nephrotic syndrome
- Heart failure
- Recent myocardial infarction or stroke

Presentation

- Venous thromboembolism occurring at a young age.
- Thromboses affecting more than one site (typically the larger vessels)
- Recurrent miscarriage
- Pre-eclampsia
- Family history of thromboembolism

Investigations

Bloods: FBC+film, ESR/CRP, clotting screen, ANA, Factor V Leiden, antithrombin III, homocysteine

Genetic tests: Factor V Leiden, anti-thrombin III deficiency, prothrombin G20210A, protein C, and protein S mutations

Investigations for malignancy, cardiac/liver disease, nephrotic syndrome as appropriate

Management

Supportive:- Mobilisation, Adequate hydration , TEDS, OCP/HRT advice

Drugs: **aspirin**, LMWH periop or in pregnancy. **Warfarin** init. for 3mo if thrombotic episode.

Prognosis

Depends on cause & thrombosis size/site. Mortality/morbidity higher with PE, CVA or MI.