**Wilson's Disease**

**Clinical features:**
- Presentation varies from acute hepatitis (mainly children) which may proceed to fulminant hepatic failure or chronic hepatitis &/or cirrhosis.
- Children usually present with hepatic problems.
- Young adults present with neurological problems (e.g. Tremor, dysarthria, dyskinesia, and eventually dementia).

**Signs & symptoms:**
- Those associated with liver disease (hepatomegaly, pruritis, jaundice, affected clotting)
- Associated with neurological signs due to basal ganglia involvement.
- Keyser-Fleischer rings (copper deposit in the cornea)

**Definition:**
- Wilson's disease is an inborn error of copper metabolism whereby there is copper deposition in the liver, basal ganglia, and the cornea.

**Pathophysiology:**
- Dietary copper normally absorbed from stomach & proximal small intestine.
- Transported to the liver loosely bound to albumin.
- Bound to caeruloplasmin in the liver and secreted into the blood.
- Copper is normally excreted into bile

- Wilson's disease is Autosomal recessive.
- Defect is on chromosome 13.
- Results in a failure of both incorporation and biliary excretion of copper.

**Investigations:**
- Serum copper & caeruloplasmin (reduced/may be normal)
- Urinary copper (raised)
- LFTs
- Liver biopsy
- Haemolysis & anaemia may be present
- Genetic analysis

**Management:**
- Chelation:
  - Penicillamine 1-1.5g OD is effective in chelating copper.
  - Monitor urinary copper and adjust dose accordingly (usually at 2-3 years)
  - Screening of family members should be offered.

**Prognosis:**
- Prognosis is good with early Dx.
- If neurological damage has occurred at Dx then this is irreversible.
- Fulminant hepatic failure & cirrhosis are indications for liver transplant.

**Side effects penicillamine:**
- Occurs in 10% of patients
  - Blood disorders
  - Rashes
  - Gi upset

All young patients with liver disease should be screened for Wilson's as it is potentially treatable.