Wilson's Disease

(X) Hepatolenticular Degeneration (Y)

- Autosomal recessive disorder characterized by increased Copper in Blood due to ATP7B Gene mutation

Copper (Cu) 1-2 mg/day → Excrete / Keep 0-75 mg excess
- 90% Bile → Fecal Cu
- 10% Urine.

Pathophysiology

→ Copper → Enterocyte → Portal vein → liver → Hepatocyte

→ In Hepatocyte → ATP7B Gene make the Cu to be bind to Apoceruloplasmin after binding called as ceruloplasmin and excreted through Bile.

↓

Gene mutation of ATP7B

↓

Copper react with Hydrogen peroxide

↓

Free radicals → Deposit in Tissue.

Clinical Features Hepatolenticular degeneration

Liver → Acute Hepatitis
↓
Fulminant liver Failure with massive hemolysis and renal tubulopathy.

Chronic Hepatitis
↓
Cirrhosis of Liver.

Brain → Basal ganglia
↓
movement disorder
Tremor, Parkinsonism

Cerebral Cortex
↓
Dementia.

Eyes → Kayser Fleischer rings.
- Copper deposit in Descemet membrane of Cornea.

→ Most important only clinical sign in 60% of adults.

→ Greenish brown discoloration in the Copper periphery of corneal margin.

→ Sometimes noted only by slit examination.

Investigations

→ ↓ Serum Ceruloplasmin

→ ↑ Serum Copper

→ ↑ Urinary Copper Excretion.

Complication:

1) Hepatosplenomegaly

2) Renal disease due to Cu in proximal tubule

3) Hemolytic anemia due to excess Cu

Treatment:

- Liver transplant in acute fulminant liver failure + cirhorris.
- Penicillamine & Zinc.