

WILSON'S DISEASE

4th yr MBCHB 2021

WILSON'S DISEASE

- ❖ This is a genetic disorder in which excess copper builds up in the body
 - ❖ There is an inherited defect in the ATP7B gene. This gene provides instructions for making a protein called copper transporting ATPase 2 which plays a role in transporting copper from the liver to other parts of the body.
 - ❖ It is an autosomal recessive disorder, hence both parents must pass one gene to the offspring to have disease
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- Occurs in females in equal proportion to males: But females more likely to develop acute liver failure > males

Symptoms & Signs

Symptoms:

Liver related symptoms include:- (Liver involvement is typically seen in children & young adults with the disease)

- ❖ Yellowness of skin & itchiness
- ❖ Jaundice
- ❖ Cirrhosis
- ❖ Liver failure s/s

Physical signs:

- ❖ Check Eyes for Kayser Fleischer rings(green to brownish ring round the cornea-scleral junction)
- ❖ CNS signs of basal ganglion involvement

Wilson's disease: Diagnosis:

- ❖ Blood: Liver biochemistry

 - Serum ceruloplasmin levels ↓

 - Level of copper in blood ↓

- ❖ Urine: 24 hr urine for copper usually ↑

- ❖ Liver biopsy for copper assessment

- ❖ Hemolysis & anemia may be present

- ❖ Imaging: MRI & CT scan of brain

- ❖ Eye examination: Slit lamp to check for Kayser Fleischer rings in the cornea

- ❖ Genetic testing: Check for genetic mutation that causes Wilson's disease

Wilson's Disease: Complications

- ❖ Liver cirrhosis/Liver failure
- ❖ Persistent neurological problems:
 - Tremors
 - Involuntary movements
- ❖ Psychological problems
- ❖ Hemolysis

Wilson's Disease: Treatment

- ❖ No cure for Wilson's disease

Life long Rx is necessary

- ❖ Dietary advise to ↓ copper rich foods: e.g.: dried fruits, liver, shell fish, mushroom, nuts, chocolate

- ❖ Chelating agents: D penicillamine for life

- ❖ All siblings & children of patients be screened(ATP7B mutation) Treat even asymptomatic cases if have evidence of cu accumulation

- ❖ Liver transplant: In fulminant hepatic failure & decompensated cirrhosis