Review Article

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Wilson's disease

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Abstract

Wilson's Disease is an autosomal-recessive disorder caused by mutation in the ATP7B gene which impairs copper excretion from the bile. Impaired copper transport and decrease copper secretion into bile which leads copper accumulation, first in the liver but ultimately in the brain and other tissues, produces clinical manifestations that may include hepatic, neurological, psychiatric and ophthalmological. Treatment is only palliative and intended to restore and maintain copper balance.

Key words: Ceruloplasmin, copper, Wilson's disease

INTRODUCTION

Wilson's disease is also known as hepatocellular degeneration. It is an inherited disease that causes too much of copper that accumulate in the organ. Diatery source of copper content includes shellfish, liver, nuts, bran and organ meat.[1] Normally the copper is absorbed from the stomach and proximal small intestine and is rapidly taken into the liver, where it is stored and incorporated into ceruloplasmin, which is secreted into blood for the function of the body. Any alteration of copper absorption and metabolism cause overload of copper in the organs which impair the function of the organs.

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DEFINITION

Wilson's disease (*hepatocellular degeneration*) is a rare but important autosomal recessive disorder of copper metabolism that is caused by variety of mutations in the ATP7B gene on chromosome 13.[2]

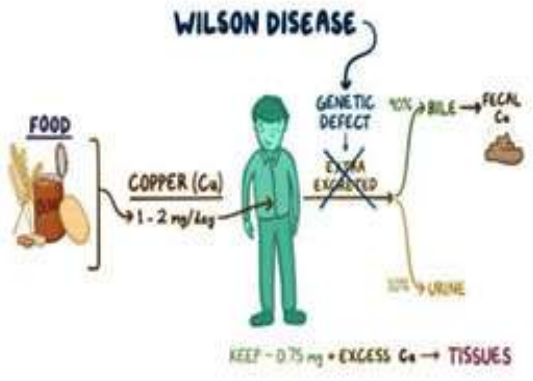
FUNCTION OF COPPER

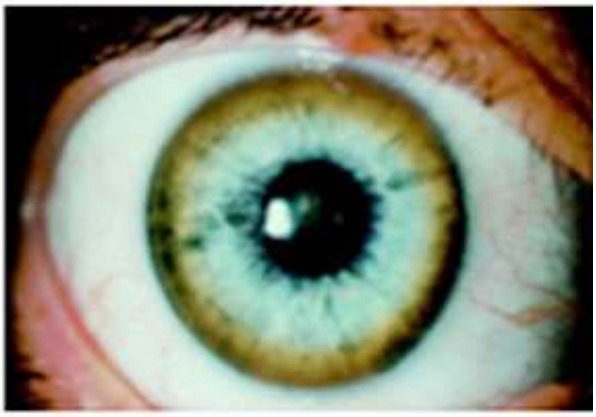
Copper is an enzyme which support for iron absorption and helps to form RBCs. It also help maintain healthy blood vessels, nerves, immune system and bone.[1]

EPIDEMIOLOGY

According to the World Health Organization, WD affects between 1/10,000 and 1/30,000 people worldwide. WD makes up 7.6–19.7% of juvenile liver disorders in tertiary hepatobiliary centres in India. A referral neurology centre registered 15 to 20 new cases of WD each year.[3]

Wilson's disease is diagnosed between the ages of 5 and 45 years, but it can affect younger and older people, as well.[2]





Kayser- Fleischer ring

NORMAL COPPER METABOLISM

Normal dietary Cu intake is 1.5–5 mg in 24 hours, 25–40% is absorbed from the duodenum which is stored by enterocytes and bound to metallothioneins in a non toxic form. 50–60% Cu is unabsorbed and excreted in faeces. From the intestine, 75% flows through the portal system with albumin or transcuprein and is taken up by the liver. The remaining 25% is bound to albumin in the circulation. In the liver, 20% of Cu is re-excreted back into the gastrointestinal tract through bile and 80% is transported to the periphery, bound to ceruloplasmin. The biliary excretion is approximately 2.5 mg/dl. Cu excretion is more through faeces than the urinary excretion.[3]

PATHOPHYSIOLOGY

Genetic defect (ATP7B gene-which transport copper).

Impaired copper transport and decrease copper secretion into bile.

These all interferes with incorporation of copper into the copper protein ceruloplasmin.

Copper overload mainly in hepatic system.

Hepatic fibrosis ultimately cause cirrhosis.

Then the copper diffuse into the blood, then into other tissues. (Basal ganglia of the brain, kidney, skeleton, reproductive organs).

Which leads to manifestations such as hemolytic anaemia, kayser-fleischer rings.[2,4]

STAGING

Stage 1: Initial period of accumulation of copper within hepatic binding sites.

Stage 2: The acute redistribution of copper within liver and subsequent release into circulation.

Stage 3: Chronic accumulation of copper in the brain and other extrahepatic tissue, with progressive disease eventually leading to fatal symptoms.

Stage 4: Restoration of copper balance by use of long term chelation therapy.[3]

SIGNS AND SYMPTOMS

Wilson's disease manifests clinically as hepatic and psychological issues. Most common symptoms are jaundice, ascitis, hepatomegaly, edema, and variceal haemorrhage and common hepatic presentation is chronic active hepatitis,which leads to cirrhosis with fulminant liver failure.[5]

Symptoms usually arises between the ages of 5-45 years. Hepatic disease occurs predominantly in childhood and early adolescence, although it can be present in adult in their fifties.

Liver Dseases

Acute hepatitis may progress into fulminant liver failure.

Chronic hepatitis leads to cirrhosis.

Massive haemolysis and renal tubulopathy due to free copper into blood stream.

Neurological disease

Tremor, choreoathetosis, dystonia, Parkinsonism and dementia, unusual clumsiness.

Kayser-Fleischer ring-greenish-brown or dark rings that appear to encircle the iris of the eye.[2]

DIAGNOSIS

- Low ceruloplasmin (less than 20 mg/dl, Referencerange 20-40 mg/dl) high free copper concentration.
- Urine/ 24 hrs urine collection.
- High urine copper excretion greater than 0.6 micromol/24 hrs (38 micromol/24hr).
- More than 25 micromole/24hrs of D-penicillamine.[2]
- Hepatic copper concentration of a liver biopsy specimen is >250mcg/g of dry weight (normal: 15- 55mcg/g).
- Kayser-Fleischer rings in cornea and Urine: 24 hours urine collection.

MANAGEMENT

The goal of treatment is to reduce the amount of copper in the tissues by chelation therapy:

- Copper binding agents: Penicillamine 1.5g/ day.
- Alternative choices: Trientinedi hydrochloride (1.2-2.4g/day) and Zinc 50mg 8 hrly.
- Liver transplantation.[2]
- A low-copper diet is recommended to reduce dietary copper intake to 1 mg/day. Foods to be avoided include: Chocolate, Dried fruit, Liver, Nuts, Shellfish.
- Demineralized drinking water can be suggested for patients as drinking water is high in copper.[5]

PROGNOSIS

Prognosis is better, if treatment is started before the irreversible damage to the organs.[2]

CONCLUSION

Wilson's disease (WD), which is defined by poor copper metabolism, can have a range of clinical effects, including asymptomatic conditions to fulminant hepatic failure, chronic liver disease with or without cirrhosis, and neurological and psychiatric manifestations. Chelators like D-penicillamine and trientine are used as part of treatment plans, while zinc salts act as methallothionein inducers, which encourage a negative copper balance and a decrease in free plasmatic copper.

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