Case report on Wilson’s disease with hepatic encephalopathy and type 1 hepatorenal syndrome

Prajwal Bhagat¹, Shalini Moon², Roshan Umate³
¹Final Year Basic Bsc Student Smt. Radhikabai Meghe Memorial College Of Nursing Sawangi Meghe Wardha.
²Associate Professor, Smt. Radhikabai Meghe Memorial College Of Nursing Sawangi Meghe Wardha.
³Research Scientist, Jawaharlal Nehru Medical College, Datta Meghe Institute of Medical Sciences, Sawangi, Wardha, Maharashtra.
DOI: 10.47750/pnr.2022.13.S07

Abstract
An uncommon inherited condition of copper metabolism is known as Wilson disease. The copper-transporting enzyme ATP7B, which is mostly found in the liver and brain, has undergone several mutations that result in hereditary abnormality. Clinical symptoms are loss of vision for 2 days, and after eye examination, there was a yellowish discoloration in the eye. The patient did not want to eat anything due to the disease condition, and he has been having a loss of appetite for 2 days. Early infancy to young adulthood and even later ages can see the start of the condition. Clinical, biochemical, and molecular indicators are used in conjunction to make the clinical diagnosis. Early or even better pre-symptomatic disease stages are when chelating medicines and zinc salts are most useful as a treatment.

Keywords: Wilson’s disease, hemolytic anemia, hepatorenal syndrome, hepatic encephalopathy.

INTRODUCTION
Wilson’s disease (WD) is a copper transport condition brought on by ATP7B, a P-type ATPase that transports copper. The incidence of WD ranges from 1/50 to 10,000 live births globally. (1) Clinical manifestations of WD can take many different forms, although typically they present as hepatic or neuropsychiatric symptoms, with a wide variety of problems for each group. (2) However, clinical signs may not always be present in youngsters, which makes disease identification more challenging than it is in adults. Hepatic symptoms can range from asymptomatic mild biochemical changes to severe fulminant hepatic failure, cirrhosis, or acute, but primarily chronic, hepatitis. (3) Hepatic encephalopathy, also known as HE, is generally understood to be a brain malfunction brought on by liver insufficiency and/or portal-systemic blood shunting.(4)

Hepatorenal syndrome, a serious consequence of end-stage cirrhosis, is characterised by high kidney vasoconstriction, increased splanchnic blood flow, hyperdynamic condition, state of reduced central volume, activation of vasoconstrictor systems, and decreased GFR. In recent years, researchers have drawn attention to the role that systemic inflammation, a crucial aspect of cirrhosis, plays in the emergence of hepatorenal syndrome.(5)

CASE PRESENTATION
The 28-year-old young adult male visited the medicine department with a complaint of loss of vision for 2 days, and after eye examination, there was a yellowish discoloration in the eye. The patient did not want to eat anything due to the disease condition, and he has been having loss of appetite for 2 days, and has been having decreased talking for 2 days. The patient is a known case of Wilson's disease, and he was hospitalized 20 days ago for the treatment of Wilson's disease, taking a treatment of penicillamine, ceftriaxone, copper chelating agent, and healthy diet.
On examination, the patient was conscious and oriented. The abdomen was distended with shifting dullness and the spleen was palpable on per-abdomen examination. The cardiovascular, respiratory, and musculoskeletal systems were normal. No focal neurological deficits were observed.

On the ultrasonography of admission day, hepatomegaly, thickened gall bladder wall, dilated portal vein and cystitis were seen in the report, and blood values were also disturbed. These were Hb 5.6% decreases normal range in male is 13.2-16.6 grams/dl, urea 64 mg/dl normal range is 6-24mg/dl, creatinine 2.4 mg/dl 0.7-1.3 mg/dl, all values increased as compared to normal value. In the CT scan of the abdomen, hepatomegaly with mild ascites and features of portal hypertension and pseudocyst in the head of the pancreas appears normal. The rest of the parenchyma appears normal.

After all the associated diagnosis, the physician started the treatment of intravenous fluids, Inj. Ceftriaxone 1 gm, Inj. Pan 40mg, Inj. Emset, Inj. Thiamin 100mg, Inj. D25%, Tablets lasilactone, limcee. Udilive, syp. Dulphalac, and a healthy diet as per dietician.

**DISCUSSION**

Worldwide, there are cases of Wilson’s illness; the incidence is around 1 in 30,000 people, and the carrier rate is roughly 1 in 90. The copper-transporting P-type adenosine triphosphatase is encoded by the WD gene, also known as ATP7B, which is located on the chromosome.(6) The WD gene product protein is found in the trans-Golgi network and has been shown to have the ability to transport copper. More than 150 ATP7B mutations have so far been found, and their relative frequency has been characterised.(7-15)

Copper poisoning causes liver damage with a wide clinical range. Chronic hepatitis with steatosis and fibrosis, asymptomatic forms with high liver enzymes or enlarged liver, liver cirrhosis, and chronic liver failure are all possible manifestations. Some patients with asymptomatic illness up to puberty initially appear with fulminant liver failure. Acute renal failure, coagulopathy, and haemolytic anaemia (Coombs-negative) are frequently linked with it.(8) Patients with cirrhosis may lack zinc, an essential cofactor for the urea cycle enzymes, especially if they are malnourished. In experimental cirrhosis models, zinc supplementation increases the urea cycle's activity. (17-21)

Only 23 of the 70 individuals examined (33%) had Type-1 HRS that could be reversed throughout the course of the illness after a median of 4 days (range 2 to 13 days). Type-1 HRS was not reversible in the remaining 47 individuals (67%) of the group. Figure 1 depicts changes in serum creatinine levels in individuals with and without reversibility of type-1 HRS. The average peak value of blood creatinine concentration in individuals without reversibility was 4.61.9 mg/dL (within a range of 2.6 to 9.6 mg/dL). (22-28)

**CONCLUSION**

There is an accumulation of copper in liver, brain, and other important organs due to Wilson's disease, a rare genetic illness. Although younger and older persons can sometimes be affected, Wilson's disease is typically identified in patients between the ages of 5 and 35. Healthy nerves, strong bones, collagen, and the melanin pigment in the skin all grow in part due to copper. The majority of the time, extra copper is eliminated through a chemical your liver produces after being absorbed from food (bile). Nearly of patients with Wilson's disease may anticipate a full recovery from this typically deadly ailment if it is diagnosed and treated in a timely manner.

**REFERENCES**

4. Hepatic Encephalopathy [Internet]. NORD (National Organization for Rare Disorders). [cited 2022 Aug 30]. Available from:


