Incidental Finding of Wilson Disease in Acute Liver Disease—A Case Report

INTRODUCTION

Wilson’s disease is an autosomal recessive disorder due to mutation in ATP 7B gene on chromosome 13 that results in an impaired copper excretion by the liver. As a result, copper gets deposited in the liver, brain etc., in toxic concentration and causes tissue damage. Wilson’s disease is a disease of adolescence, 50% presenting with hepatic disease as the first disease manifestation.

Hepato-lenticular degeneration (Wilson disease) occurs more frequently in male, is usually detected during adolescence, half of the patients have onset before 16 years of age. Forty percent patients first show hepatic dys-function, 40% neurological symptoms and 20% with psychiatric disorder or behavioural disorder. Here we present a case report of an adolescent boy who presented to us with fever, jaundice and altered sensorium.

CASE REPORT

A 16 year old boy presented with a 7 days history of fever which was intermittent associated with chills, pain in abdomen, vomiting, swelling over the bilateral lower limbs and face, itching all over the body, history of yellowish discoloration of sclera and skin and high colored urine and 1 day history of altered sensorium and irritability. There was no history of loose motion, haematemesis, malena, skin rashes, bleeding from orifices or seizure. There was no history of breathlessness, chest pain or bowel and bladder complaints. He had no history of intake of outside food or water, no history of travel, no history of any drug with known hepato-toxicity in the last three months and was non-alcoholic. No past history suggestive of jaundice. His parents were non-consanguineously married and neither he nor his siblings had a history of any hepatic or neurological manifestation suggesting Wilson’s disease. As the patient was having altered sensorium and irritable, he was being admitted in ICU for observation and investigations.

Examination

On physical examination the patient had altered sensorium and irritable. His heart rate was 88/min, BP was 110/80 mm of Hg, respiratory rate was 16/minute, temperature was 98.3 F, had mild icterus and moderate pallor. There was edema over the bilateral lower limbs and face. No signs suggestive of hepatic encephalopathy and chronic liver disease. Examinations of CVS, RS, and CNS were within normal limits.

On Per Abdomen Examination Soft, Distended, Hepato-spleenomegaly with no signs of Ascites were noticed.
Patient's serum ferritin level was normal, ANA – negative, MCV – 108.3 fl, High liver copper levels (>250 μg/g dry wt). Kayser–Fleischer rings in eyes. Low serum ceruloplasmin levels (<20 mg%). Delay in diagnosis of WD is observed across the health care levels. Blood urea – 22 mg%, S. creatinine - 0.7 mg%, 24 hour urinary copper levels was 387 μg/day. So it was are given Radioisotope copper studies using 64 Cu, 67 Cu.

HIV- Negative and serology for viral markers:negative.

Patient’s serum ferritin level was normal, ANA was negative, Serum for copper levels was 64.98 μg/dL, Serum ceruloplasmin level was 10.40 mg% (20–60 mg).

S. Ammonia-180 mg%.

24 hour urinary copper levels was 387 μg/day (3.00–50.00 μg/day), results were rechecked and repeat 24 hour urinary copper levels were 715.66 μg/ day (3.00–50.00 μg/day).

Radiological investigations

Chest X-ray-WNL.

USG abdomen- Spleenomegaly with Liver parenchymal disease (s/o cirrhosis) with signs of portal hypertension.

CECT (abdomen + pelvis) with contrast: liver parenchymal disease with changes of portal hypertension.

MRI Brain: Bilateral symmetrical hyperintensity noted in globus pallidus and tegmentum of mid brain especially in the region of substantia nigra on T1WI. These appear T2W hypointense with subtle blooming on GRE. Bilateral symmetrical hyperintensity in globus pallidus and mid brain. This is consistent with clinical history of Wilson’s disease.

Slit lamp examination showed Kayser-Fleisher ring in both eyes.

Keeping the patient’s clinical profile and investigations in mind, differential diagnosis of Malaria, dengue, haemolytic disorder, viral and autoimmune hepatitis for Wilson’s disease, as it is also increased in chronic cholestasis and Indian childhood cirrhosis. To summarise, Wilson’s disease should be suspected in any young patient who presents with hepatitis or fulminant hepatic failure.

Increased hepatic copper content is not specific for Wilson’s disease, as it is also increased in chronic cholestasis and Indian childhood cirrhosis. To summarise, Wilson’s disease should be suspected in any young patient who presents with hepatitis or fulminant hepatic failure.

REFERENCES


