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WILSON'S DISEASE – A CASE REPORT

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ABSTRACT

Wilson's disease (WD) is a disorder of copper metabolism leading to accumulation of Copper(Cu) in various organs in the body. Liver manifestations tend to occur in the first decade of life and neurological manifestations in the third decade of life. Neurological manifestations tend to worsen with chelation therapy.

Keywords: Wilson's disease, Liver Cirrhosis, Kayser – Fleischer rings, Head tremors

INTRODUCTION

Wilson's disease is an autosomal recessive disorder characterised by degenerative changes in the brain, cirrhosis of liver and Kayser – Fleischer (KF) rings in the cornea and was first described in 1912 **[1]**. It results from altered binding of copper with hepatic proteins such as ceruloplasmin resulting in accumulation of copper in the hepatocytes and later as liver cells become overloaded, copper is deposited in other tissues to which it is toxic. Its incidence is reported as 1 in 50,000–100,000 **[2]**. The gene associated with the condition is located on the chromosome region 13q14 **[3]**. In children the liver is mainly involved hence it is called hepatic form and as years go on,

neuropsychiatric changes become increasingly important, so it is called neurological form **[4]**.

Case Report

History

Our patient was a 43 year old male who came to the outpatient department with a history of swelling of abdomen and was provisionally assigned a diagnosis of liver cirrhosis on the basis of clinical findings and ultrasonogram report. Liver showed coarse echotexture. Portal vein was dilated. Collaterals were seen and gross ascites was seen. However, the etiology of cirrhosis was not obvious. The patient denied history of alcohol intake and was tested negative for hepatitis B and C.

Examination

A head and hand tremor was noticed and an MRI was taken, following which an eye examination reported a Kayser-Fleischer ring. Further biochemical investigations confirmed the diagnosis. At that time the patient did not have any clinically apparent liver disease.He also had slurring of speech. Rest of the neurological examination was normal. Slit lamp examination revealed Kayser- Fleischer rings in both eyes.MRI revelaedT2 and FLAIR hyperintensities seen in the tegmentum of midbrain with sparing of the red nucleus around cerebral aqueduct and the ventrolateral part of bilateral thalamus. Basal cisterns and ventricular system showed mild dilatation.T2 and FLAIR hyperintensities were also seen in subcortical and periventricular white matter of bilateral cerebral hemispheres. MRI of abdomen showed macronodular cirrhosis of liver. Biochemical studies such as serum ceruloplasmin, serum copper and 24 hours urinary copper were also suggestive of the diagnosis of Wilson's disease.

Course

The patient was started on Penicillamine therapy 1 gm daily. Other medications included Tab. Zinc 50mg daily, Tab. Pyridoxine 25mg daily, Tab. Pacitane 2mg twice daily, Tab. Torsemide and Tab. Propranolol. Within 2-3 days patient’s symptoms improved. He regained his appetite and sense of wellbeing. The ascites and tremor showed marked improvement after treatment.

DISCUSSION

Patients with Wilson’s disease usually present with liver disease and develop neurological manifestations as disease progresses. But in our case it was the opposite. The presenting symptom was tremor.There are several other case reports where the initial presentation was of the

nervous system without involvement of the liver and presenting age was over 40 **[5, 6]**. Paradoxically the neurological manifestations tend to become worse with penicillamine. This is attributed to the mobilization of copper from liver with elevation in levels of unbound copper which leads to worsening of neurological symptoms. In different studies, the initial neurological deterioration was observed in 30 to 75% of patients following onset of penicillamine therapy **[7, 8]**. The tremor improved in our patient after starting treatment with penicillamine and anticholinergics. On the other hand, hepatic manifestations were actually precipitated by the start of the drug. The patient was not willing to take chelation therapy but was made aware of its importance. After studying the literature and earlier case reports of Wilson's disease, treatment was started showing good results.

CONCLUSION

As Wilson's disease is a rare disease the diagnosis is more likely to be missed, unless observed with a keen eye. There should be a high index of suspicion in all the cases of liver cirrhosis with no clear cut cause or an isolated neurological symptom such as a tremor. It is also important to advice patients not to stop therapy.

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