Wilson's Disease - A Case Report

Harharpreet Kaur[1], Kiranjit Kaur[2], Navneet Sharma[3], Kiran Kumar[4]

ABSTRACT

Introduction: Wilson's disease (WD) is a disorder of copper metabolism leading to the accumulation of this metal in different organs. Hepatic manifestations tend to occur in the first decade and neurological symptoms in the third decade. Neurological manifestations are said to worsen with chelation therapy.

Case report: In our patient however the initial manifestation was head tremor at the age of 43 years which improved with treatment. The patient for some reason stopped the therapy for 8 years after which he decided to resume it only to precipitate the liver cirrhosis clinically –something that has not been reported earlier. The diagnosis was missed initially. However treatment produced good results.

Conclusion: The case also serves as a reminder not to dismiss this disease as a rare theoretical possibility but to suspect it in a case of liver cirrhosis of unknown etiology or when the patient presents with an obscure isolated neurological sign such as tremor. Delayed recognition of the disease or stopping therapy can lead to a progression of the disease. The patient had many unusual features which are being reported for future reference by researchers and practioners.

Keywords: Wilson's disease, Head tremor, Liver cirrhosis

INTRODUCTION

Wilson's disease is a rare inherited disorder of copper metabolism with deposition of copper in the liver, brain, and other tissues with an incidence of one in 30,000.[1] Initially there is deposition of the metal in the liver followed by its release into the circulation and thereafter chronic accumulation in the brain and other extrahepatic tissues. Liver cirrhosis occurs early. In the nervous system basal ganglia and midbrain are affected most frequently. According to a German study the patients who presented in first decade show predominantly hepatic manifestations while the patients with neurological symptoms presented during the third decade.[2] Untreated Wilson's disease has a progressive course and may be fatal. The disease tends to be underdiagnosed and timely diagnosis remains a challenge.

CASE REPORT

Our patient was a 55 year old male who presented in the out patient department with a history of swelling of the abdomen and was provisionally assigned a diagnosis of liver cirrhosis on basis of clinical findings and the ultrasound report. Liver showed coarse echotexture. Portal vein was dilated. Collaterals were seen and gross ascites was present. However the etiology of cirrhosis was not obvious. The patient denied history of alcohol intake and was negative for hepatitis B and C. A head tremor was noticed at that time but no correlation was made. The patient promised to return for further investigation and was put on diuretics. He improved and returned after a week and confessed that he had been diagnosed earlier as a case of Wilson's disease 12 years back at the age of 43 years when he had reported to a hospital for some knee ailment. At that time the attending doctors noticed his head and hand tremor and sent him for MRI following which the eye specialist reported a Kayser-Fleischer ring. Further biochemical investigations confirmed the diagnosis. At that time the patient did not have any clinically apparent liver disease.

The patient took penicillamine therapy for 4 years with improvement in his tremor but it reappeared on discontinuation of the drug as he moved to another place where it was not available and was off the drug for 8 years. A few months ago he had opportunity to visit a bigger city where he could access the drug and he restarted the treatment. Within 10 days he began to lose his appetite and weight and developed swelling of the abdomen due to ascites. He attributed it to the chelating drug. He also had head tremor and slurring of speech. Rest of the neurological examination was unremarkable. Slit lamp examination revealed Kayser-Fleischer rings in both eyes.(Fig 1)MRI showed T2 and FLAIR hyperintensity seen in tegmentum of midbrain with sparing of red nucleus around cerebral aqueduct and ventrolateral part of bilateral thalami. (Fig 2) Basal cisterns and ventricular system showed mild dilatation.T2 and FLAIR

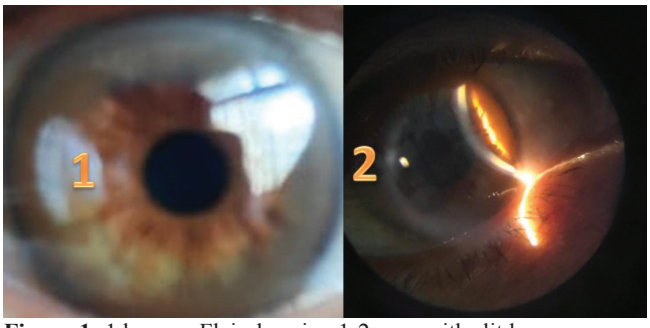

Figure-1: 1 kayser- Fleischer ring 1-2 seen with slit lamp

[1]Professor, Department of Medicine, [2]Professor, Department of Biochemistry, [3]Assistant Professor, Department of Radiology, [4]Professor, Department of Medicine, Maharshi Markandeshwar, Medical College, Kumarhatti, Distt Solan, Himachal Pradesh, India

Corresponding author: Dr Harharpreet Kaur, Professor Medicine, MM Medical College, Kumarhatti, Distt. Solan, Himachal Pradesh, India

How to cite this article: Harharpreet Kaur, Kiranjit Kaur, Navneet Sharma, Kiran Kumar. Wilson's disease - a case report. International Journal of Contemporary Medical Research 2019;6(7):G42-G44.

DOI: http://dx.doi.org/10.21276/ijcmr.2019.6.7.33

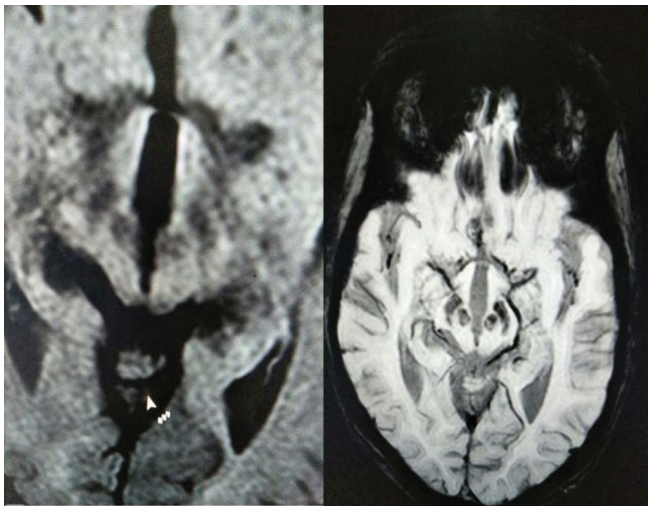

Figure-2: Axial FLAIR and gradient images of midbrain showing hyperintensity throughout the mesencephalon with sparing of red nucleus, substantia nigra and superior colliculus which are hypointense giving giant panda sign.

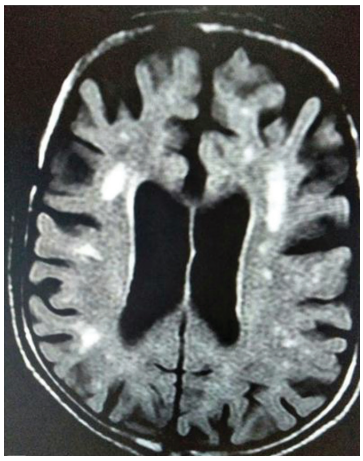

Figure-3: FLAIR axial image showing prominent sulci and lateral ventricles suggestive of brain atrophy and non specific focal hyperintensities in subcortical white matter.

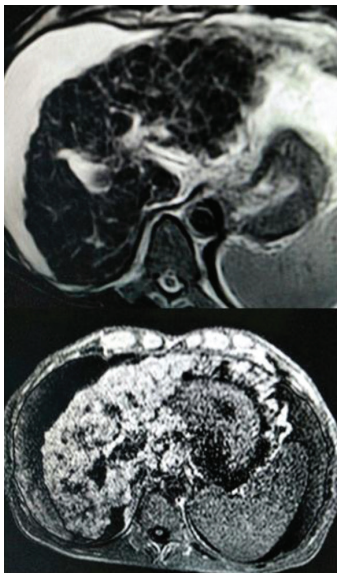

Figure-4: Axial T2WI and T1WI MR images of abdomen showing macronodular cirrhosis of liver as evident by T2 hypointense nodules, irregular hepatic outline with ascites.

hyperintensities were seen in subcortical and periventricular white matter of bilateral cerebral hemispheres. (Fig 3). MR images of abdomen showed macronodular cirrhosis of liver. (Fig 4) Biochemical studies including serum ceruloplasmin, serum copper and 24 hours urinary copper were supportive of the diagnosis of Wilson's disease. Penicillamine therapy 1 gm daily was restarted. Other medications included Tab Zn 50mg daily, Tab pyridoxin 25mg daily, Tab pacitane 2mg twice daily, Tab torsemide and Tab propranolol. Within 2-3 days patient improved. He regained his appetite and sense of well being.The ascites and tremor showed marked improvement.

DISCUSSION

The patients with Wilson's disease usually present with the liver disease and develop neurological manifestations later on. But in our case it was the reverse. The presenting symptom was tremor.There are other case reports as well where the initial presentation pertained to nervous system without involvement of liver and the presenting age was over 40.[3,4]

Paradoxically the neurological manifestations are said to become worse with penicillamine. This is attributed to mobilization of copper from the liver with elevations in unbound copper which produces worsening of neurological symptoms. In different studies the initial neurological deterioration was observed in 30- 75% of patients following penicillamine therapy.[5,6] This was refuted by certain other reports.[7] The tremor improved in our case after treatment with penicillamine and anticholinergics. On the other hand the hepatic manifestations were actually precipitated by the drug. This fact has never been reported before. The patient was not willing to take the chelation therapy but was convinced of its importance. After studying the literature and earlier case reports treatment was started with good results.

CONCLUSION

As Wilson's disease is a rare disease the diagnosis is likely to be missed. There should be a high index of suspicion in all cases of liver cirrhosis with no clear cut etiology or an isolated neurological symptom such as tremor. It is also important to warn patients not to stop therapy. The patient had many unusual features which are being reported and shared for future reference.

REFERENCES

1. Patil M, Sheth KA, Krishnamurthy AC et-al. A review and current perspective on Wilson disease. J Clin Exp Hepatol. 2013;3: 321-36.
2. Merle U, Schaefer M, Ferenci P, Stremmel W. Clinical presentation, diagnosis and long-term outcome of Wilson's disease: a cohort study. Gut. 2007;56:115-20.
3. Taksande et al. MR Imaging of The Wilson's Disease-A Case Report. www.jkscience.org 2009; 11:150-151J.
4. Ferenci P Czlonkowska A Merie U Ferenc S Gromadzka G Yurdaydin C et al.. Late onset Wilson's disease. Gastroenterology 2007; 132:1294–8.
5. Jayantee Kalita, Kumar Vijay et al. Worsening of Wilson Disease following Penicillamine Therapy Eur

Neurol 2014;71:126–131.

6. Medici V, Trevisan CP, D'Inca R, Barollo M, Zancan L, Fagiuoli S, et al: Diagnosis and management of Wilson's disease: results of a single center experience. J Clin Gastroenterol 2006;40:936–941.
7. Lange J: Long-term treatment of Wilson's disease with D -penicillamine. Report on 20 cases. Dtsch Med Wochenschr 1967; 92: 1657–1662.

Source of Support: Nil; **Conflict of Interest:** None

Submitted: 24-06-2019; **Accepted:** 28-06-2019; **Published:** 25-07-2019