LETTER TO EDITOR

Wilson Disease : another unusual presentation

Dear Editor,

The case report of a 12 year old child with unusual manifestations of Wilson Disease (WD) is interesting [1]. As widely known, the hepatic disease occurs predominantly in childhood and adolescence, while, the neurologic manifestations predominate in the second decade of life [2]. However, Singh *et al* [1] reported that the disease does not always follow this pattern. We here in report a similar case of Wilson disease in a child presenting with neurologic manifestations, but without any clinical features of liver involvement.

A 12 year old boy, born of a non-consanguineous marriage, presented with abnormal posture, rigidity, tremor and loss of speech for 3 months. There was history of gradual loss of walking, dysarthria and drooling of saliva for about one year, but with no history of seizures, jaundice, hematemesis, melena, or any drug intake prior to the onset of symptoms. On general examination, vitals were stable and there was no evidence of pallor, icterus, clubbing or lymphadenopathy. Neurologic examination revealed the child to be apathetic with loss of speech. Pupils were equal in size, reactive to light and accommodation. Visual fields were full to confrontation. A brown pigmentation in the cornea near the limbus was noticed in both eyes. Muscle tone was increased in all the four limbs (lead pipe rigidity) and plantar response was extensor bilaterally. On abdominal examination, liver and spleen were not palpable. Respiratory and cardiovascular system examination revealed no abnormality. Slit lamp examination of the eyes confirmed the presence of Keyser-Fleischer (KF) ring in both eyes (Fig 1).

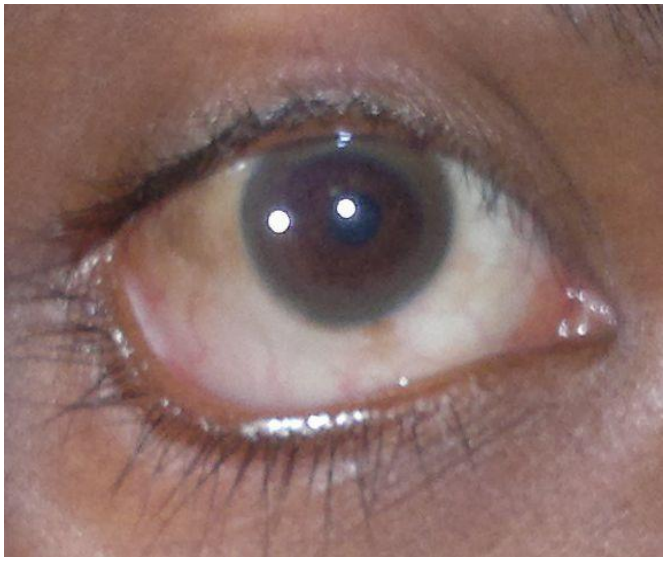

Figure 1. Kayser- Fleischer ring seen as a brownish pigmentation around the periphery of the cornea

Routine blood investigations' including hemogram and cell counts were normal. Liver function tests showed a serum bilirubin level of 0.6 mg/dL, Aspartate transaminase (AST) 50 IU/L, Alanine transaminase (ALT) 54 IU/L and prothrombin time of 15 seconds. Serum ceruloplasmin level was 0.1g/L and 24 hour urinary copper excretion was 110 µg/day. Ultrasonogram of the abdomen showed a normal liver size, with coarse echotexture and minimally dilated portal vein and a mildly enlarged spleen with mildly dilated splenic vein. Magnetic resonance imaging (MRI) of the brain revealed high signal hyperintensities in the basal ganglia and putamen region of brain, bilaterally on T2 weighted images.

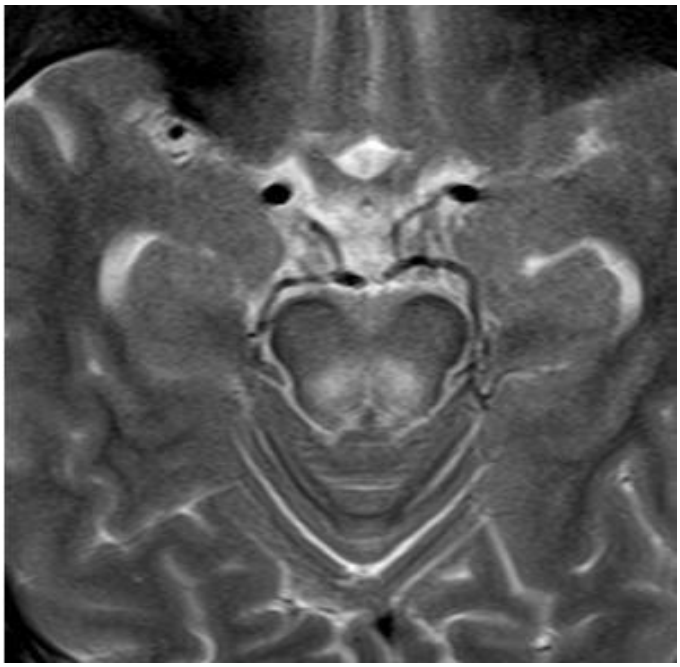

Figure 2. Axial T2WI at midbrain level : high T2 signal in the tegmentum showing face of giant panda (normal signal intensity in red nuclei against background of the hyperintense signal in tegmentum)

On the basis of these findings, the child was diagnosed as a case of WD and treatment was initiated accordingly with D-penicillamine and other supportive treatment. After about one month of this therapy, the child's condition began to improve, as the dystonia decreased and the child was able to speak and walk with support.

Diagnosis of WD in the present case was made on the basis of established criteria in a child with neuropsychiatric disorder with or without liver disease, i.e., presence of KF rings, low serum ceruloplasmin level (<20mg/dL) and a 24-hour urine copper exceeding 40 µg/day [3]. Besides the above mentioned criteria, the MRI of the brain showed characteristic giant panda appearance which further strengthens the diagnosis [4]. The recommended treatment of a symptomatic patient includes a chelating agent like D-penicillamine and agents that reduce intestinal absorption of copper [5]. As recommended, we gave our patient D-penicillamine, zinc and vitamin B_6. The initial response to therapy in the present case has been satisfactory, but a close follow-up is being done for the overall prognosis.

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Unusual presentation of this disease has a lesson for all of us; as such a case may lead to delay in diagnosis and consequently may alter the prognosis substantially.

References

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Competing Interests

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Sincerely,

Anita Kumari, Satish C Agrawal, Manu P Singh, Anant VS Rathee

Department of Pediatrics, SRMS Institute of Medical Sciences, Bareilly, Uttar Pradesh, India.
Email: anitaneo@gmail.com

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