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Paediatrics

THE STORY: NOT IN WORDS BUT THROUGH THE SLIT LAMP

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ABSTRACT Wilson disease (WD) is an inborn error of copper metabolism caused by a mutation to the copper-transporting gene ATP7B. The disease's mode of inheritance is autosomal recessive, and is characterized by excessive copper deposition mainly in the liver, cornea, kidney and brain. The prognosis depends on various factors like age, sex, organ involvement, time of diagnosis, early initiation of de-coppering therapy and extent of involvement in case of neurowilson disease. In WD excess copper accumulates in liver and gets redistributed to nervous system, cornea, kidneys and others. In first decade of life, hepatic involvement predominates but neurological manifestations occur in third or fourth decade.

Here we present a 12 years old male with primary neurological manifestation of Wilson disease. He presented with abnormal gait and inability to speak. On examination he also had Kayser- Fleischer (KF) ring in both eyes which clinically confirmed our diagnosis.

KEYWORDS : Wilson disease, Autosomal recessive, KF ring, D-Penicillamine, Zinc

INTRODUCTION

Wilson disease (hepatolenticular degeneration) is an autosomal recessive disorder, which is associated with degenerative changes in the brain, liver and cornea. 1 in 30,000 to 1 in 50,000 births are affected worldwide [1]. Genetic basis is traced to the ATP-7B gene locus on long arm of Chromosome 13 which is critical for biliary copper excretion and for copper incorporation into ceruloplasmin. Absence or malfunction of ATP7B results in decreased biliary copper excretion and diffuse accumulation of copper in the cytosol of hepatocytes [2].

Normally, copper loss occurs through bile and into faeces. In Wilson disease biliary excretion of copper is impaired and body copper progressively increases, especially in the liver, brain, kidneys and cornea. The serum ceruloplasmin is low and excessive copper exists in the plasma and urine [2, 3]. The excess copper leads to tissue injury and if not effectively treated, may lead to death [2, 3]. Besides the better known basal ganglia lesions, grey matter and white matter lesions may also occur [4,5].

Case Report

A 12 years old male, born of third degree consanguineous marriage presented with involuntary movements of upper limbs, inability to speak for the past four months. There was no history seizures, jaundice, bleeding diathesis, fever with rash, joint pain, drug intake or any bleeding disorders in the family.

His developmental milestones were normal he studies in 6th class with average school performance. On examination his vitals were stable. Liver was not palpable. KF ring was present on both eyes. Neurological examination showed dystonia, choreiform movements of limbs, brisk deep tendon reflexes with heel walking. Blood investigations revealed Hemoglobin 11.6gm/dl, WBC count-6000, platelet 2.3 lakhs/. Liver function test was normal. Serum ceruloplasmin levels- 0.7mg/dl and urinary copper was 256 mcg/24hours and oral D- Pencillamine challenge test showed copper excretion of 1876mcg/24hours. When the Ophthalmoscopic examination was done by slit lamp confirmed KF ring in the eyes.

With the clinical diagnosis of Wilson disease, MRI study of the brain was done to know the extent of involvement, which showed T2/FLAIR hyper intense signal in bilateral basal ganglia thalami, midbrain and bilateral superior cerebellar peduncles. Patient was diagnosed as Wilson disease and started on D-Penicillamine and zinc therapy. In spite of good compliance with de-coppering therapy with D- Penicillamine and zinc, he had progressive neurological deterioration in the form of progressive dystonia, dysarthria and difficulty in walking, so the drug was stopped and only zinc therapy was continued.

DISCUSSION

Clinical presentation of Wilson Disease is mostly hepatic, ranging from the asymptomatic to a fulminant variety with hepatitis, portal hypertension. Clinical presentation of Wilsons Disease is between 5 to 50 years [2]. However, early childhood Wilson disease usually presents with chronic liver disease or hemolytic anemia and neurological manifestations are rare before the age of ten years [1]. Clinically evident liver disease may precede neurologic manifestations by as much as 10 yr. After 20 years of age, neurologic symptoms predominate [5], but in our case neurologic symptoms are there without any hepatic involvement.

Diagnosis is based on clinical evaluation along with biochemical and neuroimaging confirmation. Biochemical studies may show a low serum ceruloplasmin level (<20 mg/ dl) and increased urinary copper excretion (more than >100 µg copper per 24 hours). Hepatic copper estimation, of more than 250 g/g of dry tissue (Normal 15-55 µg/g) is the most definitive method of diagnosis [2].

The characteristic 'face of giant panda' sign, feature of central pontine myelinolysis was noted [6]. So, the involvement of basal ganglia and midbrain is common finding in Wilson disease, however involvement of gray and white matter is a very rare finding which signifies copper toxicosis. The possible hypothesis for this signal changes are combination of demyelination, spongy degeneration, softening and cavitation [3, 4].

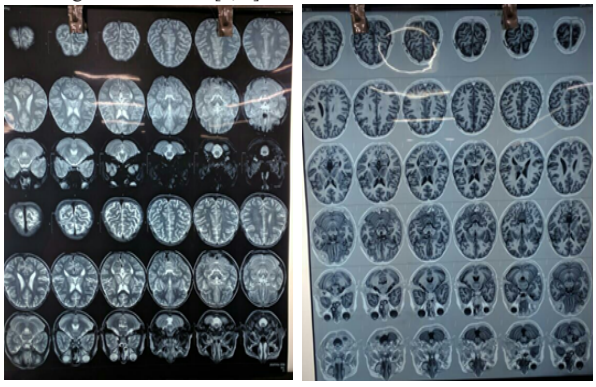

Figure 1and 2: MRI brain showing Symmetric white matter hyperintensity is noted in bilateral lentiform nuclei ,caudate nuclei, thalami , PAG matter , pons with relative hypointensity involving the central tegmental tracts giving it a miniature panda sign . Area of hyperintensity noted in left frontal subcortical white matter. Mild prominence of ventricles , cisterns and sulcal spaces s/o cerebral atrophy.

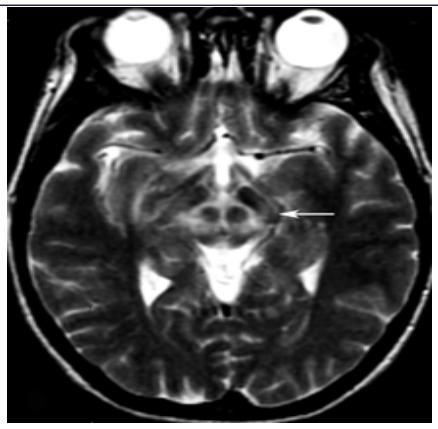

Figure 3: MRI Showing Face Of Gaint Panda

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