NEURO WILSON DISEASE- A CASE REPORT

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ABSTRACT

BACKGROUND
Wilson disease is an autosomal recessive hereditary disease characterised by deficiency of ceruloplasmin with more pronounced involvement of the liver, eyes and brain. We report here a case of Wilson disease presenting with only neurological manifestations without hepatic involvement.

Clinical History
A 12-year-old female, fourth child born to third-degree consanguineous parents had been brought to Paediatric Neurology with complaints of slurred speech, tremors in both extremities and worsening academic performance. Clinical findings – palpable liver, bilateral KF rings. Patient was investigated and the results were: CBC showed anaemia and thrombocytopenia, Liver and Renal function tests were normal with abnormal serum ceruloplasmin levels.

CONCLUSION
Neurological feature may be the only presenting manifestation of Wilson disease even in the absence of clinical evidence of hepatic involvement. Along with clinical profile, laboratory investigations are also essential for diagnosis.

KEYWORDS
Ceruloplasmin, Copper, Kayser-Fleischer Rings, Anaemia, Thrombocytopenia, Hepatomegaly, Dysarthria.


BACKGROUND
Wilson disease, an autosomal recessive disorder is caused due to mutation in the ATP7B gene resulting in impaired biliary excretion of copper. (Prevalence rate of one per 30,000). Hence copper gets accumulated first in liver and then in the brain and other tissues resulting in hepatic, neurological, psychiatric, ophthalmological, and other derangements. Neurological dysfunction constitutes as initial clinical manifestation in 30% of individuals with Wilson disease.3

Case Report
A 12-year-old female, fourth child born to third-degree consanguineous parents was brought to Paediatric Neurology Outpatient Department with complaints of slurred speech, tremors of both extremities, abdominal pain, h/o drooping of right shoulder and worsening academic performance.

Family History
Other siblings are normal, but a niece with h/o similar complaints is on medical treatment.

On Examination
Pallor, hepatomegaly, drooping of right shoulder, tremors, dysarthria and gait disturbance were present. Bilateral KF rings. USG showed mild hepatosplenomegaly.

MRI Brain Report
Non-suppressible hyperintensities in bilateral globus pallidus, caudate nucleus, thalamus, midbrain and pons.

Slit-Lamp Examination
Showed bilateral KF rings.

Investigations
1. CBC - Anaemia and Thrombocytopenia.
2. Liver function test- Normal.
3. Renal function test- Normal.
5. ABG- Normal
6. Serum Copper- 81 mcg low normal range. (Colorimetry).
7. Serum Ceruloplasmin- 17 mg/dL reduced. (Nephelometry).
8. 24 hrs. Urinary Copper- 24 mcg/24 hours normal. (Colorimetric-Esterified Glycerol derivative).

Treatment

Showed Bilateral KF Rings
DISCUSSION
Wilson disease is an inborn error of copper metabolism leading to accumulation of copper in the liver in the early part of the illness, and thereafter in the brain, eye and other tissues. Clinical manifestations take 4 to 5 years for allowing copper to accumulate to toxic levels. Various hepatic forms like acute hepatitis, chronic hepatitis, cirrhosis of liver, acute fulminant hepatic failure can occur in early childhood. During the hepatic stage, Kayser-Fleischer ring may be absent. Neurological onset in Wilson disease has been recorded in children as young as 6 years and in adults as old as 52 years. Neurological symptomatology is generally limited to the motor system, presenting as extrapyramidal or cerebellar dysfunction. Neurological symptoms are usually secondary to cerebral copper accumulation, which is sufficient to destroy the nerve cells. The patient predominantly presents with dystonia, tremor, dysphasia, dysarthria, gait and limb ataxia, and neuropsychiatric manifestations. Kayser-Fleischer rings are present in almost all the patients in the neurological stage of the disease. In our case, neurological manifestations were the presenting feature without any association of hepatic involvement detected clinically and by liver function studies. Importantly, many patients with neuropsychiatric manifestations give past or concurrent history or have biochemical evidence of liver disease. The remarkable recovery in respect of extrapyramidal and pyramidal dysfunction with penicillamine therapy supported the diagnosis. The unusual feature was the presence of neurological manifestations without any clinical involvement of liver and abnormal liver function tests.

CONCLUSION
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REFERENCES