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CASE REPORT

Isolated neuropsychiatric manifestation in Wilson's disease

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ABSTRACT

One of the rare genetic disorders of copper metabolism in Wilson's disease (WD) presents various clinical manifestations at the time of diagnosis. Usually, it involves the liver during the early course of the disease and later with neuropsychiatric involvement.

Here, we present a case of a 21-year-old lactating woman who presented only neuropsychiatric manifestations with neither hepatic manifestations nor a family history of any neuropsychiatric illness. However, we have excluded the involvement of the liver following the standard protocol of the treatment.

For diagnosis of the WD, a high degree of suspicion is required.

Keywords: *Copper metabolism; lactation, autosomal recessive.*

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INTRODUCTION

Wilson's illness is a rare autosomal recessive condition that Samuel Alexander Kinnier Wilson first identified in 1912.[1] Wilson's disease is estimated to afflict one in 30,000 newborns.[2] The disorder is caused by a mutation in the gene that codes for ATPase copper-transporting beta (ATP7B), a membrane-bound copper-transporting ATPase.[3] The human body's liver produces and secretes apoceruloplasmin, which binds to copper to generate ceruloplasmin as part of normal physiology. It eliminates the excess copper through bile (Cu).[4] The gene mutation reduces copper excretion into the bile by preventing copper from incorporating with apoceruloplasmin, leading to decreased copper excretion into the bile. Hepatic dysfunction is brought on by Cu accumulation in the liver. As copper accumulates in other organs like the brain, kidneys, and cornea, it becomes poisonous and causes various clinical symptoms. Wilson's disease typically manifests as neuropsychiatric symptoms in adults and as hepatic symptoms in youngsters.[4,5] More than 60% of patients had an abnormal liver function at the time of diagnosis, 40% to 60% had neurological symptoms, and up to 10% to 25%

had psychiatric symptoms.[5] When copper (Cu) builds up in the cornea, Kayser-Fleischer (K-F) rings are seen. Twenty to thirty per cent of people without symptoms and ninety to one hundred per cent of patients with apparent neurological symptoms have K-F Ring. Although neurological and psychological symptoms can manifest at any stage of the illness, they usually manifest in the last stages once the liver fails.[6,7]

Here we present an atypical case of Wilson's Disease presenting with neuropsychiatric signs as the primary sign before exhibiting any significant symptoms of liver dysfunction.

CASE REPORT

A 21year old lactating lady presented with slurring of speech for four months, which was insidious in onset and gradually progressive. It was associated with the drooling of saliva. She had difficulty swallowing, which was more to liquid than solid. She also complains of weakness in her whole body with no distal or proximal predominance. She noticed that she had difficulty in writing and holding objects, and difficulty in walking, with a history of frequent falls. Her face retained a wry smile, and she was emotionally labile. Her developmental milestones were typical, and she had a normal vaginal delivery without complications. There is no history of any Jaundice, hepatitis, or palpable lymphadenopathy. On examination, her tongue retracted posteriorly (**Figure 1**).

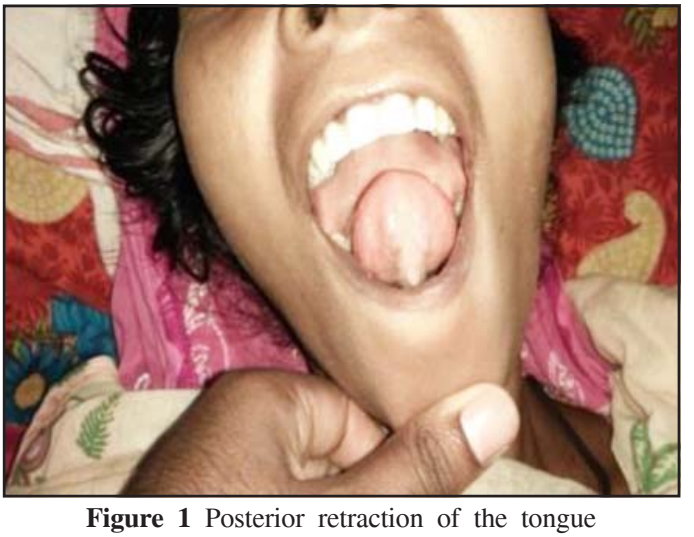

Figure 1 Posterior retraction of the tongue

A brown ring was seen in the bilateral cornea, confirmed to be Kayser-Fleischer Ring on slit-lamp examination (**Figure 2**).

On higher mental examination patient had speech impairment which was dysarthric with normal intellect and no hallucination, delusion, or illusion. Cranial nerve functions were intact, and a batwing type of tremor was present. On

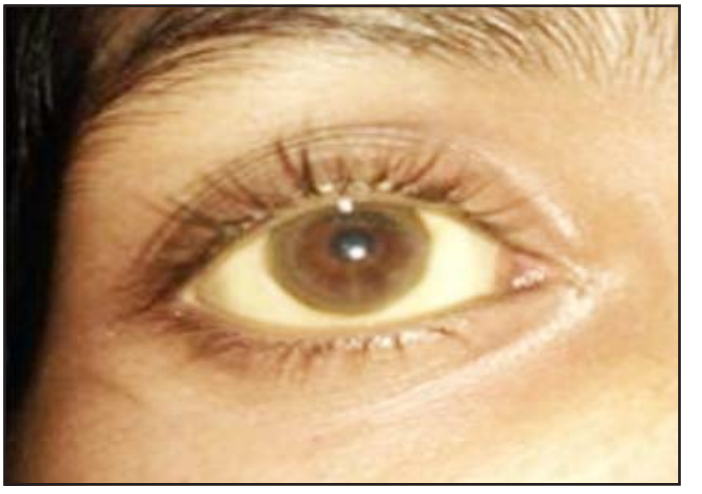

Figure 2 Kayser Fleischer Ring

brain imaging (**Figure 3**), T2/FLAIR hyperintensities were seen in the bilateral caudate nucleus, lentiform nucleus, ventro lateral thalamic and midbrain tectum. They showed subtle diffusion restriction on DWI suggestive of Wilson's Disease. Serum ceruloplasmin was 8mg/dl, and 24-hour urine copper was 116 ì g. She was diagnosed with WD based on Leipzig's criteria (score 4). However, her Liver Function Tests were normal in terms of Synthetic function and metabolic function. Her Ultrasonography of the Abdomen shows normal hepatic echotexture with normal shape and size. Fibroscan was done, and the value was 2 kPa, within the normal range. Liver Biopsy was planned but could not be done because the patient have not consented to the procedure. She was started on D-Penicillamine and zinc, and her symptoms improved on medications.

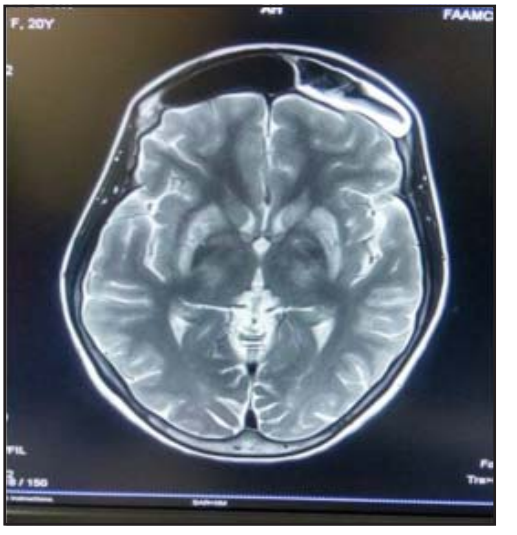

Figure 3 MRI image of Brain

DISCUSSION

This patient experienced neuropsychiatric symptoms rather than any liver issues. She was diagnosed with Wilson's

Disease in light of her serum ceruloplasmin levels, the Kayser-Fleischer ring's presence, and MRI results. Hepatic manifestation of Wilson's Disease often manifests earlier than neurological manifestation, and neuropsychiatric symptoms may first manifest in the early stages of Wilson's disease before neurological ones. In roughly 20% of instances, neurological and hepatic dysfunction occurs before psychiatric symptoms.[6] Her symptoms were relieved by Penicillamine and Zinc treatment, and no unfavourable side effects were identified.

CONCLUSION

Not all patients of WD with neuropsychiatric symptoms present with coexisting hepatic dysfunction at the time of diagnosis. It is important to emphasize this critical aspect of diagnosis, as neuropsychiatric symptoms without typical clinical liver abnormality can be fatal if not treated appropriately. Since timely diagnosis and management of WD hold prognostic value in reducing disease impact, clinicians should be vigilant in identifying similar atypical scenarios.

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