

Catatonias: A Rare and Unusual Presentation of Wilson's Disease

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Abstract: Wilson's disease is a rare autosomal recessive condition involving abnormal copper accumulation in the body due to a defect in the ATP7B gene, which encodes a membrane bound copper binding protein. It presents with hepatic, neurological, and behavioural manifestations.

The authors report a case of a 14 years old female who presented catatonic symptoms and was later diagnosed with Wilson's disease. Though uncommon, a diagnosis of Wilson's disease should be considered in the evaluation of adolescents and young adults presenting with psychiatric manifestations and/or neurological features.

Key words: Catatonia, Wilson's disease, adolescents.

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I. Introduction

Wilson's disease is a rare autosomal recessive condition involving abnormal copper accumulation in the body due to a defect in the ATP7B gene, which encodes a membrane bound copper binding protein.[1] It frequently manifests with psychiatric and behavioural symptoms ranging from 30% to 100%.[2] A recent review of literature showed that 30 to 40 % of patients of Wilson's disease have psychiatric manifestations at the time of diagnosis, and 20% patients would have seen a psychiatrist.[3] Failure to diagnose is known to be the principal cause of death in Wilson's disease.[4]

Here, we report a patient who presented with predominant catatonic symptoms with neurological signs leading to diagnosis of Wilson's Disease. The discussion is from a psychiatrist's perspective.

II. Case Report

A 14-year-old girl born out of non-consanguineous parentage, with no family history of psychiatric illness, presented with insidious onset of abnormal body posturing, inability to walk, inability to talk, drooling of saliva and lack of appetite which progressed gradually over 3 months.

Physical examination showed mild anaemia, drooling of saliva and posturing. Systemic examination was normal except for rigidity in all limbs and exaggerated deep tendon reflexes of bilateral lower limbs.

Mental status examination revealed profound psychomotor retardation, mutism, echopraxia, negativism and prolonged abnormal posturing. Bush Francis Catatonia Rating scale was applied on the first day of admission and a score of 18 was obtained.

Ophthalmological evaluation revealed bilateral Kayser-Fleischer ring. Neuroimaging revealed T1 hypointense, T2 and flair high signal intensity focus noted involving bilateral basal ganglia and internal capsule suggestive of metabolic disorders or toxic encephalopathy. Ultrasound abdomen showed coarse and heterogeneous liver echotexture suggestive of liver parenchymal disease. Serum ceruloplasmin was low, <0.08 g/L (0.15-0.50).

Considering the diagnostic formulation, the patient was provisionally diagnosed according to DSM-5 as catatonia due to other medical reasons and started on Inj Lorazepam.

Keeping in view of the above findings, and after consultation with a Neurologist, the patient was diagnosed as a case of Wilson's disease.

III. Discussion

Wilson's disease can present with hepatic, neurological, and behavioural symptoms. The exact lifetime prevalence of psychiatric disorders is not known but is estimated to range between 30-100%. Psychiatric manifestations may precede neurological signs in the early stages of Wilson's Disease.[2]

In a study of 136 patients with Wilson's disease, an initial diagnosis of organic disease other than Wilson's was made in 25.7%, psychiatric illness in 23.5%, seizure disorder in 19.1% and Wilson's disease in 31.6% patients.[5]

In another study of 307 patients of Wilson's Disease, a diagnostic error was made by referring doctors in 192 patients. Lack of awareness among health professionals about the varied presentations of Wilson's can have prognostic implications. [6]

Catatonia is a syndrome associated with a variety of psychiatric, and neurological disorders. Serious medical illness might underlie new onset catatonia and psychiatrists must be vigilant of such presentations.[7] Importance of early diagnosis of Wilson's Disease cannot be stressed enough as outcome can be fatal in the absence of specific treatment. [8]

IV. Conclusion

Catatonia is a rare presenting symptom of Wilson's Disease and it should be considered as a possibility in young patients presenting with catatonia, especially if co-morbid with neurological symptoms. Early diagnosis and prompt management is associated with better overall outcome.

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