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A RARE CASE OF ISOLATED NEUROLOLOGICAL WILSON'S DISEASE

General Medicine

Dr. Tushti Panchani* 3rd year MD Medicine, Department of Medicine P.D.U. Medical College Rajkot. *Corresponding Author
Dr. Kamesh Malik 3rd year MD Medicine Department of Medicine P.D.U. Medical College Rajkot.
Dr. Hiren Makvana Assistant Prof. Medicine Department of Medicine P.D.U. Medical College Rajkot.

ABSTRACT

A 17 year old male patient presented with abnormal, purposeless and involuntary jerky all four limb movements associated with difficulty in speaking since 10 days. Patient had history of unable to lift glass and difficulty in doing some routine activity since 1 year. Cognition of this patient was also impaired. There was no significant family history and past history.On examination, patient was conscious but cognitive function was impaired with disarticulated speech. Tone and power of all limbs was found to be normal but Babinski sign was positive. Both upper limb remained in abducted postures with jerky movements (Wing beating tremor). There was circumferential golden brown ring seen in cornea (Kayser-Fleischer ring). On investigation, S.Bilirubin Direct/Total- 0.1/0.3 , ALT-15 , ALP-68 , AST-100 , 24 hours urinary copper- 779 , Serum ceruloplasmin- 1% and on MRI Brain , bilateral Caudate and lentiform nucleus was relatively reduced in volume as compared to age with box out appearance of bilateral frontal horns. Diffuse slow waves on EEG. So diagnosis of Wilsons disease was made and patient put on Zinc , Tab Penicillamine, and Trihexyphenidyl.

KEYWORDS

INTRODUCTION:

Wilson's disease is a rare inherited disorder occuring in approximately one in 30,000 to 40,000 people worldwide(1) with CNS involvement seen in 50% cases (2). It is inherited as an autosomal recessive trait. Genetic disease is determined by two genes, one received from the father and one from the mother. Researchers have determined that Wilson disease is caused by disruption or changes (mutations) of the ATP7B gene (3), which plays an important role in the movement of excess copper from the liver to the bile to eventually be excreted from the body through the intestines.

Some individuals with Wilson disease may have only abnormalities of liver function test and may show no other symptoms until many years later. A minority of affected individuals may experience severe liver failure. This happens most frequently in people with Wilson's disease during adolescence and more commonly in women. These individuals may rapidly develop signs and symptoms of liver disease, often associated with anemia due to breakdown of red blood cells (hemolysis) and mental confusion. In these young patients, the characteristic rusty-brown deposits in the cornea of eye (Kayser-Fleischer rings) may not yet be present.

In some patients, liver disease does not reveal itself, and the patient develops neurologic (brain-related) symptoms. Common neurological symptoms of Wilson disease that may appear and progress with time include tremor, involuntary movements, difficulty swallowing (dysphagia), difficulty speaking and poor articulation (dysarthria), lack of coordination, spasticity, dystonic postures, and muscle rigidity. Almost all affected individuals with the neurological symptoms of Wilson's disease have Kayser-Fleischer rings in their eyes.

CASE REPORT:

A 17 year old male patient presented with abnormal, purposeless and involuntary jerky all four limb movements associated with difficulty in speaking since 10 days. Patient had history of unable to lift glass and difficulty in doing some routine activity since 1 year. Cognition of this patient was also impaired. There was no history of loss of consciousness, frothing from mouth, dysphagia, facial deviation, yellowish discoloration of sclera , hepatosplenomegaly and skin changes. There was no significant family history or history of similar illness.

On examination, patient was conscious but cognitive function was impaired with disarticulated speech. Tone and power of all limbs was found to be normal but Babinski sign was positive. Both upper limb remained in abducted postures with jerky movements (Wing beating tremor). There was circumferential golden brown ring seen in cornea (Kayser-Fleischer ring).

Investigation :

Hb-11.8	TLC-7000	PC-2.13lakh
S. creat-1.1	Bilirubin Direct/	Total- 0.1/0.3
ALT-15	ALP-68	AST-100

24 hours urinary copper- 779
Serum ceruloplasmin- 1%.
USG Abdomen - NAD

MRI Brain - Hyperintensity in bilateral cerebellar peduncle, thalami, gangliocapsular region, midbrain, Pons and bilateral Caudate and lentiform nucleus which was relatively reduced in volume as compared to age with box out appearance of bilateral frontal horns.

EEG- Diffuse slow waves

Treatment : Diagnosis of Wilson's disease was made and started Zinc 50mg TDS, Tab Penicillamine 250mg OD, and Trihexyphenidyl 2mg QDS.

Fig 1. Kayser-Fleischer ring

Fig 2. Bat-wing tremor

DISCUSSION:

Despite a long history, Wilson's disease, an autosomal recessive disease caused by mutations in the ATP7B gene, remains a commonly misdiagnosed rare disease among Indian population. Mutations in ATP7B result in abnormal copper metabolism and subsequent toxic accumulation of copper. Clinical manifestations of neurologic Wilson's disease include variable combinations of dysarthria, dystonia, tremor, and choreoathetosis. Among neurodegenerative diseases, it is unusual in that misdiagnosis and delay in treatment are clinically relevant because treatments can prevent and cure Wilson's disease, if they are given appropriately. If left untreated, Wilson's disease progresses to hepatic failure or severe neurologic disability and death, while those adequately treated have normal life spans. This review focuses on the neurologic features of Wilson's disease, its diagnosis, and treatment options.

CONCLUSION:

It is important to diagnose Wilson disease as early as possible. Permanent neurologic dysfunction and serious liver disease may be

avoided with early diagnosis and treatment.

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