THE NEUROPSYCHIATRIC MANIFESTATION OF WILSON’S DISEASE IN A TERTIARY CARE CENTRE IN INDIA: BEFORE AND AFTER TREATMENT

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ABSTRACT

Wilson’s disease is a neurological condition warranting immediate treatment. Being a rare genetic disorder it is characterized by excess copper stored in various body tissues, particularly the liver, brain and corneas of the eyes. The disease is progressive and, if left untreated, it may cause liver (hepatic) disease, central nervous system dysfunction, and death. Psychiatric manifestations may coexist like irritability, abnormal movements, outburst in teenagers or people in their twenties. Early diagnosis and treatment may prevent serious long-term disability and life threatening complications. Treatment is aimed at reducing the amount of copper that has accumulated in the body and maintaining normal copper levels thereafter.

KEYWORDS: Wilsons disease, neurological condition, copper, choreoathetoid movements, Penicillamine, benzodiazepines.

INTRODUCTION

Wilson’s disease is a rare inherited disorder affecting both the liver and the central nervous system first described by Wilson in 1912.1 The incidence of the disease is approximately 1 in 40,000 and is inherited in an autosomal recessive fashion. Over 200 mutations have been identified in the responsible gene (ATP7B), which is located on the long arm of chromosome 13.2 The gene encodes a membrane-bound copper-binding protein expressed primarily in the liver that is responsible for transporting copper into the secretory pathway for incorporation in apoceruloplasmin and excretion in bile.

In Wilson’s disease main sites for copper build up are in the liver and the brain.3 Hence liver disease and the neuropsychiatric manifestation are the main features which lead to the diagnosis. 50% of the patients have neuropsychiatric symptoms as the initial presentation ie mild cognitive deterioration, changes in personality, tremors, masked like facies, slurred speech, ataxia, seizures etc.4 As the disease involves multiple organ systems it is best treated by a multidisciplinary approach involving the pediatrician, neurologist and a Psychiatrist.5

Initial presentation

10 yrs old male child presented with abnormal involuntary movements of hands, occasional fall while walking, dribbling of saliva and slurring of speech for 9 months. The symptoms were insidious in onset and gradually progressive. Prenatal, Perinatal and Developmental History was essentially normal. Family History revealed that his younger sibling had died at 8 yrs of age with some liver disease.

On Examination Anthropometry Weight 21.8 kg (< -3 z score). Height 126cm (-22 to 32), OFC: 51 cm, BMI 13.7 (-1 to ~22). Vitals were Within Normal Limits. No palor/icterus/cyanosis/clubbing/lymphadenopathy /skin rash/oedema. No dysmorphic features. CNS: Conscious, Well oriented, Slurring of speech present, Choreoathetoid movement present in both UL Rt>Lt, Bulk. tone/reflexes normal. Cerebellar signs absent, Spine NAD. Eye examination revealed KF rings and MRI revealed hyperintense lesions in basal ganglia (suggestive of Wilsons). Urinary Copper was raised. A diagnosis of Wilsons disease was made and the patient was started on penicillamine and Zinc. The child showed good response and was discharged on the same medications.

Further course

Patient had worsening of the movements and changes in the personality in the form of emotional labiality and refusal to go to school. He was showed to another general hospital and was started on Tab clonazepam and Tab Trihexyphenidyl. Patient maintained improvement for some days but thereafter without any precipitator had worsening of the abnormal movements. Neurology and Psychiatry consult was sought Tab trihexyphenidyl was stopped and Clonazepam was continued. Patient showed symptomatic relief within few days. During present
admission, the patient reported with intermittent high-grade fever, with worsening of the choreoathetoid movements, with decreased responsiveness and irritability for 2 days duration. Patient was aggressively treated with antibiotics, Inj Midazolam, Antipyretics, Zinc and Tab Pencillamine. Psychiatric consult was sought for aggressiveness, irritability and worsening of choreoathetoid movements. Patient was treated with Inj Phenergan and Tab clonazepam and was advised that the worsening of symptoms is due to the deterioration in the general condition. With regular medications and good nursing care, over next few days the patient’s fever subsided on its own and the movements also spontaneously reduced.

CONCLUSION

Although WD is most frequently manifested with hepatic and neurological features related with chronic accumulation of copper, it has a wide clinical spectrum. The age of onset of neurological symptoms is frequently older compared to the age of onset of hepatic involvement. Although the diagnosis of WD is made in the first decade of life in children, the neurological symptoms of WD are mostly observed in the second decade. The course of the disease is fluctuating in nature and variable response to treatment; ultimately, however severe crippling results from spasticity and dystonia contractions. The prognosis is worse the younger the age of onset.

The treatment for Wilson’s disease primarily involves reduction of copper levels by chelation in refractory cases hepatic transplant can be an option.

During times of stress i.e., fever symptoms can worsen. Primary goal involves treating the cause of fever and secondary involves maintaining reduction in the copper levels. For Choreathetosis additional sedatives can be provided as sedation prevents exhaustion.

This case highlights that a neurological condition such as Wilson’s disease warrants a multidisciplinary approach involving the neurologist, psychiatrist, pediatrician, radiologist and ophthalmologist and the nursing staff. With this multipronged approach, such conditions can be well tackled and recovery can be good.

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REFERENCES