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A CASE REPORT ON WILSON'S DISEASE WITH PREDOMINANT NEUROPSYCHIATRIC MANIFESTATIONS

Gastroenterology

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ABSTRACT

Wilson's disease also known as Hepatolenticular degeneration is an inherited disorder of copper metabolism. Treatment guidelines suggest use of anti copper agents for management. The primary manifestation of Wilson's disease will be psychiatricdeterioration such as declining in scholastic performance. However, treatment with psychiatric drugs is of greater concern due to susceptibility for greater side effects. Here we present a case of Wilson's disease in a female patient.

KEYWORDS

Wilson's disease, Kayser Fleischer ring, Neuropsychiatric symptoms

INTRODUCTION

Wilson disease is a rare genetic disorder that affects approximately 1 in 30,000 individuals.[1] It can be defined as an inherited disorder in which excessive amounts of copper gets accumulated in the body, especially in the liver, brain, and eyes. The various signs and symptoms of Wilson disease usually begins to appear between the ages of 6 and 45, but mostly begin during the teenage years.[1,2] The features of Wilson disease primarily include a combination of liver disease and neurological as well as psychiatric problems. The initial feature of Wilson disease in affected children and young adults is mostly the liver disease. Individuals diagnosed with this condition at an older age usually do not have symptoms of liver problems, but some may have very mild liver disease. Nervous system or psychiatric problems are the most common in individuals diagnosed with Wilson disease in adulthood and most common in young adults with the condition. In many patients with Wilson disease, copper gets deposited in the the cornea of the eyes which forms a green-to-brownish coloured ring called the Kayser-Fleischer ring, that surrounds the colored portion of the eyes.[3]

Wilson disease is inherited in an autosomal recessive pattern that means both copies of the gene in each cell will be having mutations. The parents of the affected individual will each carry one copy of the mutated gene, but they do not show any signs and symptoms of the condition.

Wilson disease can be diagnosed based upon a thorough clinical evaluation, a complete patient history review as well as specialized tests. Such tests includes the slit-lamp examination of the eyes that reveals the presence of Kayser-Fleischer rings, blood serum tests that shows low levels of ceruloplasmin, a copper protein and tests that reveal abnormally high levels of copper excreted through urine.[4] A liver biopsy for copper analysis may be necessary to confirm a diagnosis of Wilson disease in some patients. DNA analysis can also be used for diagnosing affected patients.

Treatment for Wilson disease is mostly life-long and usually aimed at lowering copper levels to nontoxic levels in the body and also at preventing the progression of the disease and trying to reverse any signs and symptoms that occurred due to copper accumulation in the body. Treatment can be divided into three parts: initially, the treatment of symptomatic patient, second includes the maintenance therapy after copper has been reduced in affected tissues, and finally, maintenance therapy may be used from the beginning in asymptomatic patients.[5]

Case Description

A 22 year old female patient came to the hospital for hepatologic evaluation. She was diagnosed with Wilson's disease from a local hospital and came to us for further management. Her personal and social history were found to be normal but she had family history of the disease to a distant relative and also her parents had consanguineous marriage which elevated risk of having the disease.

The patient was apparently normal till one year back. Her scholastic performance was excellent till the age of 19, from where she started showing symptoms of the disease. The first symptom to be noted was agitation followed by various other neuropsychiatric signs for which she was taken to a psychiatrist. She had difficulty in speech for one year, difficulty in chewing and swallowing for 8 months, tremor of both upper limb and lower limb and recurrent falls for 7 months, stiffness of limbs and neck for 6 months. She was under psychiatry treatment for a couple of months which further worsened the situation. She had marfanoid habitus with difficulty in initiating speech and stammering which worsened gradually and slowness in her activities.. She also started developing bouts of irritant cough while she tried to talk. Patient had decreased intake of food due to difficulty in navigating tongue to mix food and swallow. The patient started using gestures to communicate. There was history of tremors over bilateral upper limbs, both action and rest tremor. There is history stiffness and abnormal posturing of upper limbs and foot during walking. She appears to be stiff and she has difficulty to change dress by herself. There was no history of memory loss, emotional incontinence or limb weakness.

Gradually the patient started to be hypophonic. At the time of admission, the patient was conscious and oriented with a GCS score of 15/15 and the right and left cranial nerves were normal. On slit lamp examination, KF ring was observed 5 months ago. On general examination the patient appeared to have wing beating tremor over both hands, facial expression compatible with risus sardonicus- which is a peculiar distorted grin that occur due to contraction of facial muscles, chorea over trunk and limbs and athetosis. She was then undergone various differential diagnosis tests for Wilson's disease like Serum Copper and 24 hour urine copper test which was found to be elevated and she was thus diagnosed with Wilson's disease. Her vitals were found to be normal throughout her course in hospital. Her lab investigations were normal except declined serum albumin (3.1 mg/dl), urea (16 mg/dl), serum ceruloplasmin (<3mg/dl) and serum copper (16.7 mcg/dl) and elevated 24 hour urine copper (267 mcg/L). HBsAg, HCV, HIV test were negative. USG Abdomen and Pelvis shown coarse echotexture of liver and presence of bilateral multiple renal calculi, largest measuring 3.3 mm noted in mid pole. The uterus was small and anteverted with dimension of 4.3×1.3×2 cm. MRI of brain has shown T2 hyperintensities involving bilateral basal ganglia, thalami, mid brain and pons. (Figure.1)

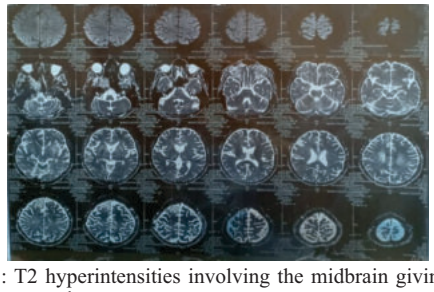

Figure.1: T2 hyperintensities involving the midbrain giving rise to face of giant panda appearance.

During the course of hospital stay, the patient was treated with chelating agent T.Pencillamine 250mg BD which was augmented to TDS since her serum ceruloplasmin levels were <3mg/dl, T Zinc Sulphate 50 mg as maintenance therapy in neurological and hepatic presentation, T Pacitane (Trihexyphenidyl) 2mg ½ - ½ -0 which was changed to 1-1-0 since she had tremors, Cap. Becosule 0-1-0 and T.Pyridoxine 10mg BD which was started on the 4th day by the neurologist as she showed activity intolerance related to the disease condition. Apart from the medical treatment she was given regular physiotherapy of Joint ROM + stretching gait training which indulged a huge change in her gait and movements. Also she was given copper free Wilson's diet which also augmented her recovery. She was better and hence discharged with the same drugs along with regular physiotherapy.

DISCUSSION:

Wilson's disease includes hepatic, neurological and psychiatric manifestations. In most of the cases, psychiatric symptoms precedes hepatic symptoms, however in this patient the hepatic symptoms preceded psychiatric manifestations.[6,7] MRI scan shows cerebral atrophy and symmetrical hyperintensity or mixed intensity in T2 weighted images changes in white matter of the cerebral hemispheres, basal ganglia, thalamus, cerebellum a brainstem.[8] Impairment of functional status in patients attribute to reduced quality of life. Recovery of patients with neuropsychiatric symptoms are gradual Lesions in putamen are responsible for the personality changes and loss of emotional control. [9]

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