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Case Report

Psychiatry/Mental Health Section

Obsessive Compulsive Disorder as a Rare Manifestation of Wilson's Disease

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ABSTRACT

Wilson's disease is a rare autosomal recessive disorder of copper metabolism, that causes an impairment of cellular copper transport. An individual's inability to excrete excess copper in bile leads to accumulation of copper in organs like the brain, liver and cornea. It can manifest with symptoms of hepatic impairment and liver failure. It can also present with neurological symptoms like rigidity, gait disturbances, dystonia, dysarthria and chorea. Frequently certain psychiatric manifestations have been reported like mood changes, personality changes, cognitive impairment, phobias, psychosis, anxiety, impulsive and compulsive behaviours . It's rare for a Wilson's disease patient to present with only Obsessive Compulsive Disorder (OCD). In the present case report, a 15-year-old male presented in the Outpatient Department with complaints of repetitive intrusive thoughts that he was unable to voluntarily control and compulsions of washing and preoccupation with checking and symmetrical organising with a deteriorating scholastic performance. The only positive findings were Kayser-Fleischer rings, low serum ceruloplasmin and mild cerebellar atrophy. The patient started showing symptomatic improvement with pharmacological treatment comprising of zinc, D-penicillamine, and fluoxetine and diet modification.

Keywords: Cerebellar atrophy, Compulsive acts, Kayser-fleischer rings, Obsessions, Tremors

CASE REPORT

A 15-year-old male school student presented in the Outpatient Department (OPD) with his mother (informant) with a chief complaint of inability to concentrate due to repetitive thoughts, that he was unable to voluntarily control, since last one and a half years. Patients's academic performance was poor due to his excessive preoccupation with hand washing and cleanliness. Thoughts of getting contaminated and skin coming in contact with dirt compelled him to do so, in a repetitive manner. The patient also had to repeatedly check the locks and bolts of the doors in his room and house even though he knew they were securely locked. His worries kept on returning even though he tried to distract himself with other things. Patient felt compelled to arrange his books and other stationeries in a symmetrical fashion and always wants to do things in perfect, symmetrical and organised manner. The patient's attitude had become argumentative and irritable, many times over trivial issues. The patient has normal birth and developmental history.

Patient's illness had worsened in the last three months to an extent that was hampering his day-to-day activities leading to social impairment in various facets of his life for example interaction with his friends, family and performance in the school. No history of psychiatric illness or substance abuse, significant family history or any genetic disorder was present.

On examination, the patient was oriented to time, place, person and was cooperative. He was alert and conscious. Rapport could be established with ease and eye contact was maintained throughout the interview. Vitals including Blood Pressure (BP), Pulse Rate (PR), Respiratory Rate (RR), temperature and Oxygen Saturation (SpO_2) were normal. On general physical examination, liver and spleen were not palpable. There was no abnormality detected in the Cardiovascular System (CVS) and respiratory system. The Central Nervous System (CNS) examination revealed coarse postural tremors. There was no presence of rigidity, dystonia, gait disturbances or bradykinesia. His deep and superficial reflexes were preserved.

Mental Status Examination (MSE) revealed a well-groomed male, dressed appropriately for his socio-economic status and age with normal psychomotor activity. The patient was cooperative and communicative without any presence of tics, jerky movements, mannerisms, excoriations or bald spots. The patient's speech was spontaneous, relevant and coherent. There was no circumstantiality of speech detected. His mood was irritable and congruent to thought since he was unable to control the repetitive thoughts that intruded his mind. He had obsessions of contamination and pathological doubt and compulsions of washing, checking and arranging things in a symmetrical fashion. Patient recognises the thoughts to be his own. There was no loosening of associations or ideas of persecution and he had no hallucinations, illusions or derealisations. He had average intellectual capacity with average general fund of knowledge. His judgment and insight were preserved.

The patient was sent to Ophthalmology Department and Kayser-Fleischer (KF) rings were observed in both the corneas on general examination and slit-lamp examination. Baseline investigations were done and there was no abnormality detected in patients Liver Function Test (LFT), Kidney Function Test (KFT), Thyroid Function Test (TFT), Fasting Blood Sugar (FBS), Post-prandial Blood Sugar (PPBS), Complete Blood Count (CBC), Bleeding Time (BT), Clotting Time (CT), Prothrombin Time {PT (International normalised ratio)}. Ultrasonography Whole Abdomen (USG W/A) was normal. A low serum ceruloplasmin level of 16 mg/dL was a positive laboratory finding. Twenty-four hour urine copper content was increased to 110 µg/24 hours. The Computed Tomography Scan (CT scan) of brain revealed cerebellar atrophy. The patient refused to do Magnetic Resonance Imaging (MRI) of brain due to financial constraints. Liver biopsy for copper quantification (quantitative assay) could not be done.

The patient was diagnosed to have Wilson's disease with OCD. He was started on zinc (50 mg three times daily) and D-penicillamine (250 mg 2 capsules three times daily) and fluoxetine (20 mg once daily) with appropriate diet changes to lower the copper load (avoidance of shell fish, oysters, crab meat, organ meats like liver and kidney, dried fruits, cocoa, dried beans, lotus stem, radish, beet root, high copper fruits like water chestnut, grapes and litchi, beverages like soya milk, horlicks, coconut water etc., and avoidance of storing food and cooking in copper and brass vessels) and advised regular follow-up in the OPD. The family members were

advised to undergo genetic counselling including his two siblings. However, the patient was lost to follow-up after a few visits.

DISCUSSION

Wilson's disease is often described as hepatoneurological disease, which encompasses liver along with brain. So, the focus is on signs of Extra Pyramidal Symptoms (EPS), like tremors and dystonia, as well as on liver disease [1,2]. Wilson's disease is a rare inherited autosomal recessive genetic neurodegenerative abnormality that causes an impairment of cellular copper transport. The copper accumulation occurs in various organs, primarily the liver and the brain, and the clinical symptoms are the result of direct or indirect of copper accumulation, leading to organ dysfunction [3,4].

A study shows that in almost 100% cases, over the course of disease, a patient of Wilson's disease, presents with psychiatric symptoms like affective, behavioural, personality, psychotic, anxiety, and cognitive. There is data suggesting that upto 30% of patients with the disease initially manifest with psychiatric symptoms [3,5,6]. Many a times, manifestations of initial psychiatric symptoms can occur in childhood like a decline in school performances, intellectual and cognitive impairment, anxiety, irritability, impulsiveness or inappropriate behaviour [7-9].

Mostly, the clinical symptoms are non specific making diagnosis difficult including often misdiagnosis, for example in cases of isolated OCD and anorexia-nervosa [2,10]. While the clinical hepatic symptoms of Wilson's disease is potentially treatable with anti-copper agents, there are many problems related to treatment of clinical symptoms outside the hepatic ones [11].

In Wilson's disease, patients manifesting with psychiatric symptoms may have a negative impact on the recovery and survival of the affected patients [2,3]. There are certain drawbacks faced in the current scenario for those, who present with psychiatric manifestations of the disease. The patients may remain undiagnosed and misdiagnosed. Other factors that hamper the therapy are refusal by the patient or the caregiver to acknowledge that the problem even exists, non compliance and difficulty in treatment adherence [2,9,10].

CONCLUSION(S)

There is a dearth of studies regarding the psychiatric manifestations of Wilson's disease, especially a presentation of stand-alone OCD. The psychiatric manifestations of Wilson's disease being varied and non specific adds to the difficulty in early diagnosis and appropriate management. The present case report was attempted to add to the existing pool of literature of psychiatric aspects like OCD in Wilson's disease, which may help in future early diagnosis and encourage further exploration in this challenging aspect.

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