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Wilson's Disease Presenting as Lower Cranial Nerve Involvement- Gold is Not Glittering

Mansi Patel[1], Anusha Gupta[1], Kolluru Karthik Raja[1], Sunil Kumar[2], Sourya Acharya[2]

[1]Post Graduate Resident, [2]Professor,
Department of Medicine, Jawaharlal Nehru Medical College, Datta Meghe Institute of Medical Sciences (Deemed to be University), Wardha, Maharashtra, India.

Corresponding Author: Anusha Gupta

ABSTRACT

Wilson disease is a rare genetic disorder characterized by excess copper stored in various body tissues, particularly the liver, brain and cornea of the eyes. The disease is progressive and if left untreated, it may cause liver disease, central nervous system dysfunction and death. Early diagnosis and treatment may prevent serious long-term disability and life-threatening complications. We report a patient presenting with lower motor nerve involvement and was diagnosed to have Wilson's disease.

Keywords: Wilson's disease, Kayser-Fleischer ring, lower cranial nerves.

INTRODUCTION

Wilson's disease (WD) is an autosomal-recessive disorder of copper metabolism caused by mutations in the *ATP7B* gene having wide range of clinical manifestations in all age groups (Ala et al.,2007). Positive family history, suspicion of Kayser-Fleischer ring (seen in more than 90% of neurologic cases), and any evidence of liver disease should immediately raise the possibility of WD, regardless of the neurologic presentation(O Brien et al.,2014). Neurological symptoms in Wilsons disease is caused by nervous tissue damage that is primarily a consequence of extrahepatic copper deposition. Excess copper deposition is toxic to the brain tissue, as it may cause cell injury and inflammation via various mechanisms including mitochondrial toxicity, oxidative stress, cell membrane damage, cross-linking of DNA and enzyme inhibition. Neurological symptoms are largely extra pyramidal (Coffey et al.,2013) and Lower cranial nerve involvement is rarely reported in literature. This case report highlights about Wilson disease in young girl who presented with weakness of tongue movements, difficulty in swallowing and dysarthria which can be a source of great discomfort to the patient.

CASE REPORT

A17 years old young female patient presented with complaints of involuntary movements in left upper and lower limb, dysphagia and slurring of speech. There was no history of tremors, headache, seizure, loss of consciousness. No history of deviation of angle of mouth, drooling of saliva. No h/o oral or nasal regurgitation. There was no any sensory involvement. There was no history of drug intake, fever, jaundice, joint pains or rashes. On general examination patient had pulse of 98/min and blood pressure was 100/80mmhg. Patient was malnourished and having difficulty in swallowing more to solids than liquids. Cranial nerve examination reveals absent gag reflex.

Laboratory investigations revealed following values: Haemoglobin of 7.5 gm%, MCV of 70 and white blood cell count of 14000 per cumm. Serum Albumin 3.9 gm per dL, Total serum protein 7.1 gm per dL,

Aspartate transferase and Alanine transferase were 27 and 21 IU/L respectively, urea 25 mg per dL, creatinine 0.6 mg per dL, serum Na^+ and K^+ were 140 and 3.8 meq per L respectively. Slit lamp examination reveals Kayser-Fleischer (KF) Ring as shown in the figure-2. 24-hour urinary copper reveal 6.1mcg/24hrs. MRI brain along with cervical spine screening was done which reveals symmetrical altered signal intensity lesion in bilateral thalami and lentiform nucleus, midbrain(cerebral peduncles) and pons appearing hyper intense on T2WI as shown in figure-1.

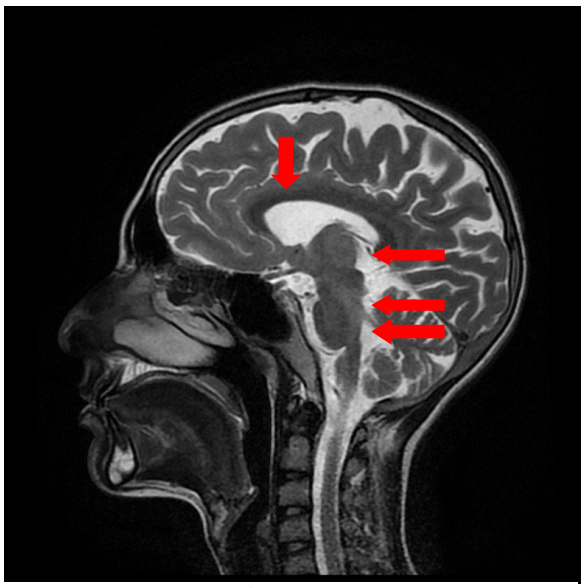

Figure 1: sagittal section of MRI brain on T2WI reveals hyper intensity in thalamus, midbrain and pons

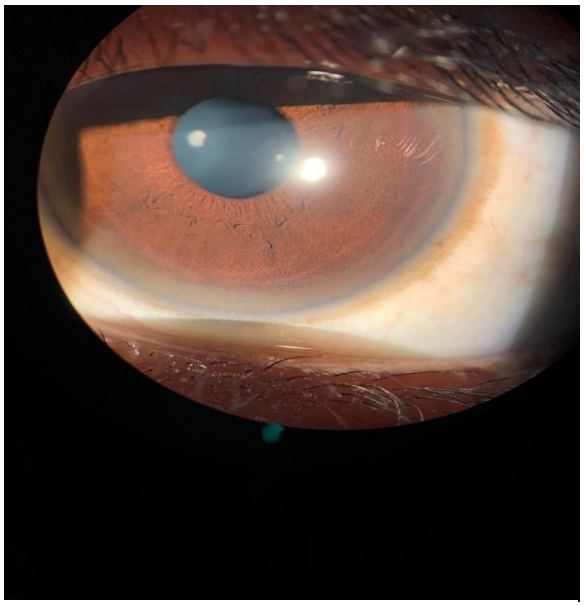

Figure 2: slit lamp examination reveals brown ring (Kayser-Fleischer ring) in the peripheral cornea.

DISCUSSION

Wilson's disease is a rare autosomal-recessive disorder with a prevalence rate of 30 cases per million. The average age of symptom onset in persons who present with neurological dysfunction is 18.9 years although it may appear as early as 6 years (Fenu et al.,2012). The most common initial neurological manifestation of Wilson's disease is tremor, which may be resting, postural, or kinetic and predominantly involves the proximal upper extremity. Neurological involvement is a rare manifestation of Wilson's disease and there are only three to four cases have been reported in the past (Czlonkowska et al.,2017).

Very important ophthalmologic feature of Wilson's disease is the Kayser-Fleischer (KF) ring. Copper is deposited in Descemet's membrane within the cornea and may be visible as a greenish-brown opacity at the periphery of each iris.

Our patient had dysphagia and slurring of speech. Isolated bulbar involvement can lead to drooling of saliva, dysarthria and difficulty in swallowing. It may not be an uncommon manifestation, but is often missed leading to a delay in diagnosis of Wilson's disease.

Dysarthria are caused by the damage of basal ganglia, cerebellar nuclei and their tracts and possibly also cortico-bulbar tracts leading to pseudobulbar features (Pfeiffer et al.,2007).

As far as dysphagia is concerned, any phase of the swallowing act can be affected including oral, preparation/ chewing, oral transit, and swallowing itself. Dysphagia may emerge due to impairment of muscle tone (e.g., in oro-facial dystonia), incoordination, slowness and weakness of deglutition muscles. Drooling, along with dysarthria and "wing beating" tremor, belongs to the most prominent and characteristic symptoms of WD and defined as involuntary flow of saliva from the mouth, drooling affects approximately 70% of neuro-WD patients (Acharya et al.,2014). Very often it is the consequence of

dysphagia and/or the inability to retain saliva within the mouth due to orofacial dystonia. This typically occurs in patients with "open mouth smile".

CONCLUSION

To conclude, this case highlights one of the rare neurological manifestations of Wilson's disease in the form of lower cranial nerve involvement like dysphagia, dysarthria and drooling of saliva. It is important to be aware of these neurological symptoms in Wilson's disease, as it can commonly be missed and an early diagnosis can be extremely rewarding in this potentially treatable condition.

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