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Bulbar Palsy as a Rare Presentation of Wilson's Disease: A Case Report

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1. Introduction

Wilson's disease is an AR disorder caused by mutations in ATP7B gene located on chromosome 13q14 which leads to decreased production of ceruloplasmin and increased free copper. This excess copper is deposited in brain, liver, eyes, etc. Here we report a case of bulbar palsy who was subsequently diagnosed as Wilson's disease.

2. Case Report

20yr old male presented with tremulousness of body for 3 years, difficulty in swallowing and drooling of saliva for 1yr, nasal regurgitation of fluids for 8months, difficulty in walking for 6months and urinary incontinence for 2months. On examination, patient was hemodynamically stable with normal higher mental functions having more dysphagia to liquids than solids, dysarthria, absent jaw jerk, absent gag reflex, rigidity in all 4 limbs, bilateral plantar extensor and exaggerated DTR's with presence of wing beating tremors.

Investigations: AST 46IU/L, ALT 16IU/L, AST/ALT ratio 3, ALP 46IU/L, T. Bilirubin (0.5mg/dl), ALP/T. Bilirubin ratio 92, Sr. ceruloplasmin of 4.36 (normal >20)

Slit lamp examination: KF rings in Descemet's membrane.

MRI brain: T2 and FLAIR hyperintensities in bilateral basal ganglia, thalamus, pons with restrictive diffusion.

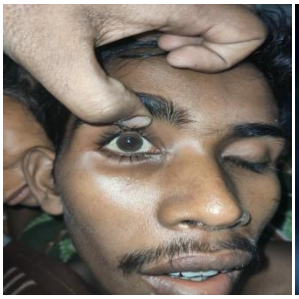
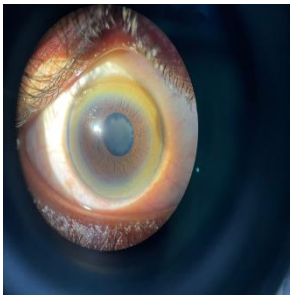
Usg abdomen revealed features of hepatic parenchymal disease.

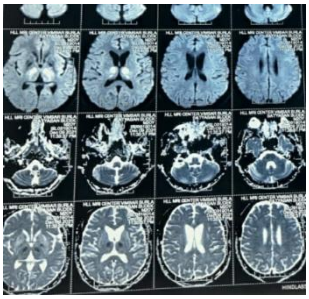
Leipzig score was 7.

Hence patient was diagnosed as a case of Wilson's disease and started on zinc and d - penicillamine.

3. Discussion

- Wilson's disease is a rare AR disorder which commonly presents between 3 to 50 years of age. (1)
- GI manifestations include acute & chronic hepatitis & cirrhosis of liver with or without decompensation. (3)
- Neuropsychiatric manifestations includes tremors, dystonia, parkinsonian type of rigidity, depression, phobia etc. Tremors are the usual initial neurological manifestation in Wilson's disease.30 - 40% of these patients develop cirrhosis. (3)
- KF ring is a very important ophthalmologic feature which is present in 99% of neurological Wilson's disease and 30 - 50% of the hepatic Wilson's. (4)
- Copper deposition in basal ganglia, cerebellar nuclei & their tracts and corticobulbar tracts can results in pseudobulbar palsy. (1)
- But our patient had bulbar palsy in the form of drooling of saliva, dysphagia to liquids, nasal regurgitation of fluids and dysarthria which is rare and leads to delay in diagnosis of Wilson's disease. (1)
- Leipzig's score is used to diagnose Wilson's disease.
- Neurological Wilson's disease is treated with chelating agents like d - penicillamine, trientene and by addition of zinc tablet to reduce copper absorption from the intestine.
- Our case was treated with oral d - penicillamine 250mg qid zinc 50mg tds but no significant improvement were observed.



4. Conclusion

Although tremors and abnormal movements are the initial manifestations of Wilson's disease, bulbar symptoms especially in middle aged individuals must also be evaluated in the line of Wilson's disease.