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ATYPICAL PRESENTATION OF WILSON DISEASE WITH EARLY ONSET NEUROLOGICAL SYMPTOMS.. A RARE CASE

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ABSTRACT Wilson disease is a rare autosomal recessive disorder that causes copper deposition in several organs primarily liver and brain resulting in neurological psychiatric and hepatolenticular disease. Early Clinical, neuroimaging and biochemical screening including liver biopsy remains standard for diagnosis. Typically, Wilson disease presents with hepatic symptoms like jaundice, and ultimately could lead to liver failure in early adulthood, followed by neuropsychiatric symptoms in later adulthood. As seen in this patient neurological symptom that is slurring of speech was directly due to wilson disease without any hepatic or psychiatric symptom. So any case which presented with isolated neuropsychiatric symptom could raise the suspicion of Wilson disease. Early diagnosis and treatment in this child resulted in good outcome and better prognosis.

KEYWORDS : Hepatolenticular disease, Serum ceruloplasmin, urinary copper, KF Ring, Neuropsychiatric symptoms

INTRODUCTION

Wilson disease is an autosomal recessive disorder of ATP7B [1]gene on chromosome 13q resulting in defective copper metabolism. [1] Copper excess deposition in several organs primarily liver and brain, eye resulting in hepatolenticular disease etc. [2]

In 1912 Samuel Ak Wilson described the condition as a familial hepatolenticular degeneration with neurological symptoms and potential fatal outcome. [3]If left untreated, Wilson`s disease can be fatal due to progressive liver damage, neurological deterioration, or other complications.

Wilson disease has varied spectrum of presentation depending upon area of copper deposition varying from being asymptotic to fetal hepatic failure or encephalopathy to suicidal tendency as psychiatric symptom. Hepatic symptom include jaundice pruritus lower limb edema to signs and symptom related to hepatic failure to, Ascitis, varices. Extra pyramidal symptoms include tremors, dysarthria, abnormalgait, behavioralproblem, depressions, anxiety, mood swings, psychosis poor vision.

Diagnosis of Wilson's disease is done through modified Leipzig score (TABLE1). This score include low ceruloplasmin level low 24 hrs urinary copper, KF ring, coombs negative hemolytic anemia, mutational analysis, liver biopsy s/o Wilson disease, typical MRI finding, neurobehavioural symptoms and history of wilson disease in family member. For diagnosis of score of more then 4 suggest wilson disease. 0

Therapeutic goal is to restore and maintain copper homeostasis. Among various cu chilators Penicillamine potentially and effectively improves hepatic and and reverse neuropsychiatric sign and symptoms in most patient and prevent in onset. Trientine and tetrahydromolybdate are alternate drug with similar mechanism of action but with more serious side effect like sideroblastic anemia and bone marrow suppression. Zinc also inhibit copper absorption in gastrointestinal tract maintaining negative cu balance. Hence can be used alone after pt is return to normal cu balance with chilation.

Table1: Leipzig Score[9]

PARAMETERS	POINTS
KF RING Present Absent	 2 0
Neuropsychiatric symptoms(or typical feature on brain MRI) Present Absent	 2 0
Coombs negative hemolytic anemia (plus high serum copper) Present absent	 1 0
24hrs urinary copper(uln40microg/d) normal 1-2×uln >2×uln	 0 1 2
Liver copper Normal(uln 50microg/g dry wt) 5×uln >5×uln	 -1 1 2
Genetic mutation Present on both chromosome Present on one chromosome absent	15% 4 1 0
Serum creulloplasmin Normal(0.2g/dl) 0.1-0.2 g/dl <0.1	 0 1 2

Case Presentation:

A 10year old female child born to G2P1L1 mother vial normal delivery 2nd by birth order Hindu by religion, who was previously normal presented with a 4 month history of slurred speech and disturbances in fluency.

The parents observed there was gradual decline in their child`s speech, characterized by increasing slurring and decreased fluency of words which was gradual and progressive in nature. When inquired from parents they confirmed that there is no history of any trauma. She had negative history of fever, ear infection, cold and cough, headache, projectile vomiting, jaundice, anorexia, vomiting, no history of drug intake or drug abuse and no other congenital anomaly. No history of any seizure was present in child. Child was hospitalized and routine investigations were send. Blood investigation were within normal limit, but patient had low ceruloplasmin level raising suspicion of wilson disease. So 24 hrs urinary copper was send and child was send for KF ring examination. Ophthalmology examination suggested KF ring like deposition on cornea and 24 hrs urinary copper showed >30. MRI was adviced which s/o hyperintensity in bilateral basal ganglia, bilateral thalamus extending to midbrain and pons, a finding consistent with Wilson`s disease, further supporting the diagnosis (IMAGE1, 2).

Parents were not ready for genetic analysis and liver biopsy due to financial burden. The diagnostic results as per above mention criteria were diagnostic of Wilson disease. The treatment goals are to reduce copper accumulation, manage symptoms and prevent disease progression. Low copper diet along with Chelation therapy, pyridoxine and zinc therapy was started. Targeted exercises to improve speech clarity, articulation and communication skills. Patient was called after 1 week and showed improvement in speech fluency.

Regular monitoring of liver function and copper levels were done. The patient showed significant improvement in symptoms, accompanied by a notable decrease in copper levels.

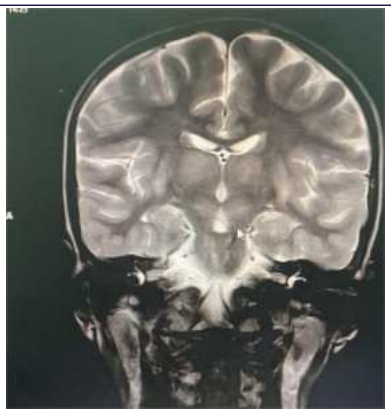

Image 1

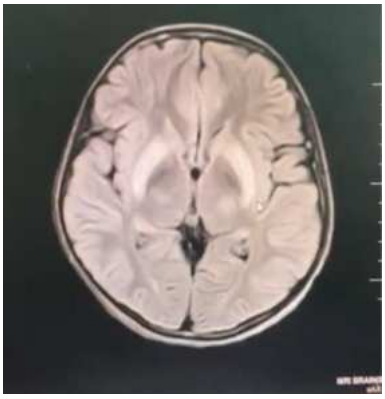

Image 2

DISCUSSION:

According to one recent study on Wilson disease on children of Egypt and India hepatic manifestation were more seen in children less then 10 yrs and lenticular symptom were seen in later adulthood. According to this study hepatic symptom were seen in <10 years: 83%; 10–18 years: 52%; >18 years: 24%, while neuropsychiatric presentation increased as age advanced (<10 years: 17%; 10–18 years: 48%; >18 years: 74%. this co relate to our patient age of onset of Wilson disease. Symptom onset to diagnosis was shorter in hepatic (6 months) than neuropsychiatric presentation (18 months) [4].

Another case record from AIIMS New Delhi of 25 Wilson disease follow up cases found both hepatic and neurological menifestation was earlier than found in other case record of western country giving an insight that Wilson disease can be diagnosed at early age. According to their study The age of onset of initial symptom ranged from 3 years to 11 years with a mean age of 7. 2 years. [5] The mean time to presentation after the onset of symptoms was 2 years (range 2 months to 6 years). In our case patient was diagnosed with in 4 month of onset of symptom.

A case study published in the Journal of Clinical Neuroscience highlights the importance of reports of a 17-year-old patient who presented with tremors, rigidity, and psychiatric symptoms, which were Research suggests that neuropsychiatric symptoms are common in Wilson's disease, affecting up to 70% of patients. A study published in the Journal of Neurology, Neurosurgery, and Psychiatric found that psychiatric symptoms were the primary presenting feature in 28% of patients with Wilson's disease[6]. To note that in our case neuropsychiatric symptom ie slurring of speech was primary symptom as mention in these study.

The pathophysiology of neuropsychiatric symptoms in Wilson's disease is thought to be related to copper accumulation in the basal ganglia and other brain regions[7]. A study published in the journal Neurology found that patients with Wilson's disease had significant abnormalities in brain MRI scans, including lesions in the basal ganglia and white matter[8]. As also seen in our case there was hyper dense deposition in internal capsule, pons and mid brain.

Early diagnosis and treatment of Wilson's disease are critical to preventing long-term neurological damage. A study published in the Journal of Hepatology found that zinc acetate therapy was effective in reducing copper accumulation and improving neurological symptoms in patients with Wilson's disease [9]. in our pt also zinc was started along with d penicillamine as it decreases copper absorption from gut.

CONCLUSION:

Typically, Wilson disease presents with hepatic symptoms in early adulthood, followed by neuropsychiatric symptoms in later adulthood. As seen in this patient neurological symptom that is slurring of speech was directly due to Wilson disease without any hepatic or psychiatric symptom.

So any case which presented with isolated neurological symptom could raise the suspicion of Wilson disease. Early diagnosis and treatment in this child resulted in good outcome and better prognosis.

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