A Rare Case of Wilson’s Disease in a 17 Years Old Girl

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Authors’ contributions

This work was carried out in collaboration between both authors. Both authors read and approved the final manuscript.

ABSTRACT

Introduction: Wilson’s disease (WD) is an autosomal recessive disorder involving cellular copper transport. A defect in biliary excretion leads to accumulation of copper in the liver, causing progressive liver injury and cirrhosis. Approximately 1 in 40,000 people have Wilson’s Disease. It affects both men and women equally. Symptoms appear between ages 5 and 35.

Case Presentation: This is a case of 17 years old girl came with complained of dysphagia, irritability, breathlessness, weakness in both upper and lower limb at left side for 6 months. She had difficulty in going up and coming down a staircase and in getting up from the squatting position. She could walk with support for the first 6 months of his illness but later, had required crutches. After her admission diagnostic evaluation was done, and in the findings was chest x-ray was done which was normal. ECG showing normal sinus rate and rhythm. Ophthalmology call was done and slit lamp examination was noted which was showing KF ring was present bilaterally. MRI brain suggested symmetrical altered signal intensity lesion in bilateral thalami and lentiform nucleus, midbrain and pons appearing hyper intense on T2/FLAIR sequences in bilateral gangliocapsular region (mainly in putamen) and thalami as well as midbrain and dorsal aspect of pons as described above. A possibility of Wilson’s Disease can be considered, blood test show that Hb was decrease that is -9.7gm%, S.G.O.T was 34.0U/L, S.G.P.T was 32.0U/L, was normal at the time of discharge early ambulation, nutrition, psychological support was given.

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Therapeutic interventions and outcome: In the present case received syndopa 110/4, BD, orally. Tablet Zinc, OD, orally. Tablet Pan D, OD, orally., inj. Dexamethasone 2 mg I.V, T.D.S., Syrup Sumax, 10ml, BD. Now the patient condition is stable.

Conclusion: This presented case of Wilson's Disease is a rare disease condition. It is an autosomal recessive disease. In this case study, author mainly focus on expert medical management and excellent nursing care which leads to fast recovery of patient. After conversation with patient her response was positive and after nursing management and treatment she was discharged with satisfaction of recovery.

Keywords: Wilson’s disease; slow stepping gait; irritability; cataract.

1. INTRODUCTION

Copper metabolism problem Wilson's disease was identified for the first time in 1912. Wilson's disease is characterized by hepatic and neurological symptoms, such as dystonia and parkinsonism [1]. Wilson disease (WD) is a neurodegenerative ailment that manifests as tremor, bradykinesia, stiffness, dystonia, chorea, dysarthria, and dysphagia, as well as a variety of other neurologic symptoms that can lead to misdiagnosis [2]. Dietary modifications and medication are usually used to treat Wilson's disease. Dietary modifications include avoiding copper cookware and eating a low-copper diet. Chelating drugs like trientine and d-penicillamine, as well as zinc supplements, are used [3]. Copper is essential for the formation of healthy nerves, bones, collagen, and the pigment melanin in the skin. Copper is normally taken from meals and eliminated via a liver-produced substance (bile). Copper, on the other hand, isn't adequately removed in persons with Wilson's disease, and it builds up, potentially to dangerous levels. Wilson's illness is treatable if caught early enough, and many people with it have normal lives [4].

2. PATIENT CASE HISTORY

2.1 Patient's Identification Data

A 17 years old girl admitted in A.V.B. R. Hospital with complained of dysphagia, irritability, breathlessness, weakness in both upper and lower limb at left side for 6 months.

2.2 Medical History

She had trouble ascending and descending a staircase, as well as rising from a crouching position. She was able to walk with assistance for the first six months of his sickness, but later needed crutches. After her admission diagnostic evaluation was done, and in the findings was chest x-ray was done which was normal. ECG showing normal sinus rate and rhythm. Ophthalmology call was done and slit lamp examination was noted which was showing KF ring was present bilaterally. MRI brain suggested symmetrical altered signal intensity lesion in bilateral thalamus and lentiform nuclei, midbrain and pons appearing hyper intense on T2/FLAIR sequences in bilateral gangliocapsular region (mainly in putamen) and thalamus as well as midbrain and dorsal aspect of pons as described above. She had no any medical history like DM, Hypertension, TB, and Asthma. She lives in nuclear family in their family four members are living together including patient and she was belonging from middle class family and in their house all facilities are available like electricity, water supply from municipality etc. Her monthly income was 10000/- per month. She maintains good interpersonal relationship with others. She is taking only vegetarian diet. She doesn't have any allergic reaction from any food and no any history of any bad habits like chewing tobacco, smoking etc.

2.3 Physical Examination

Patient general examination was state of health was unhealthy, conscious, Body built thin, Posture erect, and hygiene was good. General parameter height was 154 cm, weight 40 kg. Vital sign is Temperature 98˚c, Pulse 84 b/m, Respiration – 20 b/m, BP – 120/80 mmHg. In physical examination patient condition was poor, she was having difficulty in walking, inability to talk properly.

2.4 Investigation

2.4.1 Blood investigations

In complete blood count- hemoglobin level was 9.7gm% which was decreased, RBC count 5.51 milli/cu mm (3.80-5.80milli/cu mm), Platelet count
was 2,93,000/cu mm (1,50,000 3,90,000/cu.mm) and M.C.H, M.C.V, Total WBC count was normal. Prothrombin time (PT) 01.07(13.40), APTT control - 30, APTT - patient - 30.10, Urea 25, Creatinine 0.6. Sodium 140, Potassium - 3.8 which was normal. Alkaline phosphatase 113. Biochemistry report - S.G.O.T was 34U/L (upto46U/L), S.G.P.T was 32 (Upto 49U/L). RBS - Glucose - Plasma Random: 76 mg/dl

2.4.2 M.R. I brain

MRI brain study reveal areas of increased signal intensity on T2/ FLAIR sequences in bilateral gangliocapsular region (mainly in putamen) and thalami as well as midbrain and dorsal aspect of pons as described above. A possibility of Wilson disease can be considered. Need further evaluation. Which was shown in Fig.

2.5 Echocardiography

2.5.1 Therapeutic intervention

General measures: to check the vital signs and airway. Continue observation and record a heart’s electrical function.

2.5.2 Pharmacological management

The drugs are used Tab syndopa, BD, orally. Action – Levodopa gets converted into Dopamine. This dopamine gives relief from symptoms of Parkinson’s. Carbidopa helps in maintaining an optimal level of levodopa by inhibiting its breakdown by enzymes before it reaches the brain. Tablet Zinc, OD, orally. Action - In Wilson’s illness, zinc, as well as other anticopper medicines, is totally successful in reducing copper levels and toxicity. Zinc has an extremely low toxicity compared to other anticopper medicines. Tablet Pan D, OD, orally. Action - Pan D capsule has combined action of its two ingredients pantoprazole and domperidone. Pantoprazole inhibits a pump called the proton pump in the stomach wall, responsible for the secretion of stomach acids for digestion and leads to reduced acid production. Domperidone stops vomiting. It increases the movement in the stomach and bowel so that acid does not flow back into the food pipe. Inj. Dexamethasone, 2mg, I.V. T.D.S Action-Dexamethasone is used to treat conditions such as arthritis, blood/hormone disorders, allergic reactions, skin diseases, eye problems, breathing problems, bowel disorders, cancer, and immune system disorders. It is also used as a test for an adrenal gland disorder (Cushing’s syndrome). Syrup Sumax, 10 ml. Orally. Action-This medication is used to treat ulcers in the intestines. Sucralfate forms a coating over ulcers, protecting the area from further injury. This helps ulcers heal more quickly. At the time of discharge early ambulation, nutrition, psychological support was given & the abdominal suture was removed. Now the patient condition is stable.

2.6 Nursing Management

2.6.1 Nursing Diagnosis: Impaired breathing pattern related to compression of diaphragm

Expected Outcome: Patient’s breathing pattern is effectively maintained as evidenced by normal respiratory rate, depth and absence of use of accessory muscles for respiration.

Intervention: Assessed the patient’s respiratory rate, rhythm, depth, SaO₂ [2], use of accessory muscles for breathing. The child’s respiratory rate was 40/min, saturation was 92/min, auscultation revealed decreased air entry in the left side of the lungs, and chest X-ray revealed left pleural effusion. Assessed the patient for precipitating factors and identified the patient to have abdominal distention which was the major cause for the breathing difficulty. Positioned him in semi-Fowler’s with supportive devices such as pillows in order to facilitate better lung expansion. Administered Oxygen 2L/min via nasal prongs in order to prevent hypoxia and hypoxemia. Taught the patient deep breathing exercises.

Fig. 1. Kayser-Fleischer Ring
Evaluation: patient’s breathing pattern was effectively maintained as evidenced by improvement in the saturation to 98% and decrease in the respiratory rate. Patient remained comfortable after interventions.

2. Nursing Diagnosis: Fluid volume excess related to cellular swelling secondary to liver disease and hypotonic over hydration.

Expected Outcome: Patient maintains adequate fluid volume as evidenced by resolution of edema and ascites.

Interventions: Assessed the patient’s daily weight, abdominal girth, intake and output, breath sounds, blood pressure, respiratory rate and edema and identified the patient to have pedal edema, negative fluid balance, crackles, fluid thrill, tachycardia and diminished air entry. Assessed the diet pattern and restricted fluids to prevent overload and edema. Elevated oedematous extremities to promote venous return to heart and thereby to prevent edema. Taught importance of fluid restriction to prevent further fluid overload.

Evaluation: Patient’s status had not improved; edema and ascites did not reduce during the period of hospital stay.

3. Nursing Diagnosis: Fatigue related to anemia

Expected Outcome: Patient remains energetic and verbalizes having sufficient energy to complete desired activities. Anemia is corrected.

2.7 Interventions

Assessed patient’s energy levels, Haemoglobin, ability to perform ADL, calorie intake, and fluid balance. Identified haemoglobin level of 9.7 gm/dL, negative fluid balance and decreased ability to perform activities of daily living. Administered iron supplements and taught on iron rich diet and vitamin C rich diet to improve the haemoglobin levels. Taught the patient energy conservation techniques and assisted in performance of activities of daily living. Assisted the family to plan for a high calorie nutritive diet with fluid, sodium and potassium restriction.

Evaluation: Patient demonstrated sufficient energy to perform desired ADL.

3. DISCUSSION

In our patient, Wilson's disease manifested itself in the form of a sunflower cataract and Parkinson's disease. The Sonnenblumenkatarakt, also known as the sunflower cataract, is caused by the deposition of copper in the lens. Following the intra-ocular localization of a foreign material containing copper, similar cataracts have been reported. Patients with Wilson's disease (WD) frequently have Parkinson's disease (PD). Furthermore, most PD patients have low ceruloplasmin levels, which is a symptom of Wilson's disease. WD is a recessive autosomal disorder.

The patients with Wilson’s disease usually present with the liver disease and develop neurological manifestations later on. But in our case, it was the reverse. The presenting symptom was tremor. There are other case reports as well where the initial presentation pertained to nervous system without involvement of liver and the presenting age was over 40 [5,6]. Paradoxically the neurological manifestations are said to become worse with penicillamine. This is
attributed to mobilization of copper from the liver with elevations in unbound copper which produces worsening of neurological symptoms. In different studies the initial neurological deterioration was observed in 30-75% of patients following penicillamine therapy [7,8]. This was refuted by certain other reports [9]. The tremor improved in our case after treatment with penicillamine and anticholinergics. On the other hand, the hepatic manifestations were actually precipitated by the drug. This fact has never been reported before. The patient was not willing to take the chelation therapy but was convinced of its importance. After studying the literature and earlier case reports treatment was started with good results. In a review of 51 consecutive patients with Wilson's disease the mean age for initial symptoms was 15.5 years [10]. At the age of seventeen, our patient developed Wilson's illness. Cirrhosis usually causes an uneven surface on the liver. Wilson's illness can be difficult to diagnose, especially when copper investigations aren’t possible. A high index of suspicion is raised in the presence of a consanguineous marriage, the death of a sibling with a similar illness, and clinical symptoms of hepatocellular failure. Under slit lamp examination, bilateral KF rings verified this suspicion. If you don't search for KF rings under a slit lamp, you might miss them. For the time being, zinc therapy appears to be a viable choice [11,12], as well as maintenance therapy in Wilson's disease [13]. Screening of siblings and parents for early diagnosis and genetic counselling is essential. Non-invasive screening methods such as KF rings, high liver enzymes, elevated urine copper, and raised plasma no ceruloplasmin copper are insufficient to detect all asymptomatic Wilson's disease cases.

4. CONCLUSION

Wilson’s illness is a rare disease, thus it's likely that the diagnosis will go unnoticed. There should be a high index of suspicion in all cases of liver cirrhosis with no clear cut etiology or an isolated neurological symptom such as tremor. It is also important to warn patients not to stop therapy. The patient had many unusual features which are being reported and shared for future reference.

CONSENT

While preparing case report and for publication patient’s informed consent has been taken from patient and parents also.

ETHICAL APPROVAL

As per international standard or university standard written ethical approval has been collected and preserved by the author(s).

COMPETING INTERESTS

Authors have declared that no competing interests exist.

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