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A Case Report on Wilson's DiseaseAnd Its Management.

Kranti Khobragade[1], Dipali Ghungrud[2], Indu Alwadkar[3], Roshan Umate[4], Swapna morey[5]
1] G.N.M. 2rd year, Florence Nightingale Training College of Nursing. Datta Meghe Institute of Medical Science (D.U.) Sawangi (M) Wardha, Email: - krantikhobragade9@gmail.com, Mobile No: - 7038918591.
2] Nursing Tutor, Florence Nightingale Training College of Nursing Sawangi (Meghe) Wardha, ghungruddeepali@gmail.com-8698232366
3] Principal, Florence Nightingale Training College of Nursing Sawangi (Meghe) Wardha, indu.alwadkar@gmail.com-9960278995
4] Research Consultant, Department of Research and Development, Jawaharlal Nehru Medical College, Datta Meghe Institute of Medical Sciences, Wardha.
5] Clinical Instructor, Department of Medical-Surgical Nursing, Smt. Radhikabai Meghe Memorial College of Nursing, Datta Meghe Institute of Medical Sciences, Sawangi, Wardha, Maharashtra.

ABSTRACT:
Introduction: Wilson's diseaseis a genetic disorder in one in 30,000 people. German pathologist Friedrich Theodor von Fredric initially identified this disease in 1854 and gave it the name Samuel Wilson Disease in honour of a British neurologist. Wilson's illness is an extremely uncommon inherited condition. The liver, brain, and other important organs are overly Copper accumulated in this condition. Between the ages of five and 35, disease occurs. Children between the ages of 5 and 35 are typically diagnosed with Wilson's disease, and symptoms usually manifest. Young and older people can both be affected by the disease. Hepatolenticular Degeneration Syndrome and Copper Storage Disease are other names for this condition.
Case presentation: In this case, a 14-year-old male child was admitted to the PICU at a rural hospital in Wardha. Irritability is his main complaint. He later complained of weakness in his left upper and lower limbs, slurred speech, jaundice (a six-day yellowing of the skin and whites of the eyes), golden-brown eye discolouration (Kayser-Fleischer rings) fluid accumulation in the legs or abdomen. Difficulties with speech, swallowing, or motor coordinationbegin in the ninth year of life. Wilson's illness was found after a physical examination and laboratory studies; the patient received treatment, i.e. Tab. Penicillamine, Tab.Trihexyphenidyl, Tab.Haloperidol 5mg, and Tab Clonazepam 0.5mg.
Therapeutic Intervention:The doctor's conclusion following the patient's history, physical examination, and an investigation was Wilson's disease. The patient was treated with Medication Tab.Penicillamine, Tab-Trihexiphenydyl, Tab-Haloperidol 5mg, and Tab. Clonazepam 0.5mg.
Nursing Management: Administered intravenous fluid and monitored vital signs every two hours. Administered medication as per doctor's orders.
Conclusion: Timely treatment and management of disease complications can be prevented.

Keywords: Wilson'sdisease, genetic disorder, excessive accumulation of Copper, health problems.

INTRODUCTION:

Wilson's disease is a genetic disorder with cirrhosis, Kayser-Fleischer rings in the cornea, and degenerative abnormalities in the brain were first identified in 1912. Due to altered copper incorporation into hepatic proteins such as ceruloplasmin, Copper is thickly accumulated in the cytoplasm of hepatocytes. Copper is transferred to other organs as liver cells become stressed, which is harmful[1].
The Tran's membrane copper transporters, cytosolic copper carriers, copper storage proteins, and copper-requiring enzymes control copper homeostasis in the liver. Hepatocytes produce a variety of proteins, including the Copper-binding protein ferroxidase ceruloplasmin (CP), factor VIII (blood coagulation), cytochrome c oxidase (mitochondrial respiration), superoxide dismutase (free radical defence), and other less common proteins[2].
It has been observed that 1 in 50,000–100,000 people are affected by this condition. The gene in question can be found in chromosomal region 13 q 14. It can be presented in a variety of ways. At specific periods, the focus shifts to different sections of the body. The liver is primarily engaged in children, resulting in the hepatic form, and as the years pass, neuropsychiatric abnormalities become increasingly prominent, resulting in the neurological type.[1,2]

A hereditary disease causes an overabundance of Copper in the organs. Copper isn't adequately excreted in Wilson's illness, so it builds up and can be dangerous. Between the ages of 12 and 23, symptoms usually appear. Swelling, weariness, stomach pain, and uncontrollable or ill-coordinated movements are some symptoms[3].
Medications that cause the organs to release Copper into the circulation are frequently used in treatment. It can be removed from the body through the kidneys once it has entered the bloodstream. Copper is an important trace element that is required for crucial enzyme activity. Excess Cu, on the other hand, disrupts the redox balance in cells and tissues, resulting in significant toxicity[4].The Copper-transporting ATPase ATP7B is required for the removal of extra Copper from organisms. This disorder is caused by a mutation in the copper-transporting ATPase ATP7B, which is required for ceruloplasmin 4-5 copper loading and biliary copper secretion[4].

PATIENT INFORMATION

Patient-Specific Information:

Here, we report a case of a 14-year-old male child who was admitted to the PICU of the rural hospital Wardha. Patient's main complaints were itchiness and weakness in his left upper and lower limbs, slurred speech, and yellowing of the skin and whites of his eyes (jaundice) that had been present for six days. The eyes' discolouration is of a golden-brown hue (Kayser-Fleischer rings). It was an accumulation of fluid in the legs or abdomen. Speech, swallowing, or physical coordination problems and difficulty occur in the ninth year of age. The patient had a history of Wilson's disease; diagnosed with this disease after a physical examination and laboratory investigations. Tran's membrane Cu transporters (CTR1 and ATP7B), cytosolic Cu carriers (chaperones), Cu storage proteins, and Cu-requiring enzymes help maintain Cu homeostasis in the liver. This patient was hospitalized in the PICU for this reason. Factor VIII (blood coagulation), cytochrome c oxidase (mitochondrial respiration), superoxide dismutase 1 (free radical defence), the Cu-binding protein ferroxidase ceruloplasmin (CP), and other less abundant proteins are all made by hepatocytes. To treat all of these issues, doctors gave the patient tablets of haloperidol (5 mg) and clonazepam (0.5 mg).

Primary concern symptoms of the patient:

The patient came to the medicine OPD with complaints of irritation, weakness in the left upper and lower limbs, slurring of speech, and other symptoms. Since six days, the patient had skin problems, and whites of the eyes have been found. The golden-brown eyes were present —fluid accumulation in the legs or abdomen. Beginning in the ninth year of life, the patient had speech difficulty, swallowing, or physical coordination. The patienthad to swallow, or physical coordination problems had to begin in the ninth year of life. His general health condition was not good. Therefore he was transferred to the PICU to manage his body temperature, cold and cough.Wilson's disease had been present in the patient since he was nine. The patient belongs to a joint family. There were no comorbidities or hereditary illnesses present in his family, such as hypertension or diabetes. The patient maintained good communication with doctors and doctors, and nurses also developed a positive relationship with the patient and his family members.

Relevant past Medicalinterventions and outcomes:

Wilson's disease was a significant factor in the current case. He continuously took the medication.

Clinical finding:

<u>General Appearance</u>
Body built - Thin
General condition: Unsatisfactory
State of consciousness: Patient was fully aware about time, place and person.
Pallor: Present
<u>Vital Signs</u>
Blood pressure: 109/57 mm of hg
Temperature: 99ºF
Pulse: 156 beats per minute
Respiration: 18breaths per minute
SpO_2:97%
CVS: S1S2+
P/A: Softnon-tender

DIAGNOSTIC EVALUATION:

The patient was conscious and aware of the place and time at the time of the physical examination. The patient's blood pressure readings were 104/70 mmHg, HR-72 beats/minute, and 97 percent Spo2 at room temperature. Treatment for the current complaints included strictly two-hourly TPR/BP charting and Spo2 monitoring.The patient underwent a regular investigation, with the following findings: 10.6% haemoglobin, 5.15 red blood cells, and 11,600 white blood cells, 20.6% HCT, 25.9% MCHC, 68 MCV, and 18.5 MCH.2.41 total platelets and 9.1 calcium.In the kidney function test, the values for urea were 25 mg/dl, creatinine was 0.7 mg/dl, sodium level was 139 MEq/l, and potassium was 3.8 MEq/l.

TRERAPEUTIC MANAGEMENT:

Present case took the medical treatment with Tab.Clonazepamlm 0.5mg, Tab. Penicillin, Tab.Trihenlip, Tab. Haloperidol 5mg,

NURSING MANAGEMENT:

Provided intravenous fluid to maintain electrolytes balance. Monitoring vital signs every 2 hours. Maintain intake and output chart.

DISCUSSION:

A 14-year-old child was admitted to the PICU at Wardha, rural hospital. With complaints of fluid accumulation in the legs or belly, weakness of the left upper and lower limbs, slurred speech, yellowish colour of the skin and eye whites during the past six days, and golden-brown staining of the eyes. Beginning in their ninth year of life, the patient started having issues with speech, swallowing, or physical coordination. After a physical examination and laboratory investigation, the patient regularly took medication, and his disease outcome was moderate. For medical assistance, he was admitted to a rural hospital.

Wilson disease is an autosomal recessive genetic disorder that is passed down from one generation to the next. Genetic disorders are determined by two genes, one from the father and the other from the mother. Recessive genetic illnesses can occur when a person inherits two faulty copies of the same gene from each parent. One healthy gene and one undesirable gene recipients will be carriers but not show symptoms. There is a 25% risk that two carriers may convey the mutant gene to their unborn child during each pregnancy, causing the infant to be affected. Each pregnancy has a 50% chance of giving birth to a child who shares the same gene as either parent. The possibility that a child will inherit both parents' healthy genes[5-15].

Men and women are both at risk. Consanguineous couples have a higher chance of sharing a genetic disease than unrelated couples do, which increases the possibility that their offspring will be born with a recessive genetic disorder. Wilson's illness is brought on by mutations in the ATP7B gene, which is important in the transport of excess Copper from the liver to the bile, where it is ultimately removed from the body through the intestines. More than 300 unique mutations have been found in the ATP7B gene[16].Males and females are equally affected by Wilson disease, which is a rare condition. The illness affects people of various racial and ethnic backgrounds. Though figures vary, it is believed that one in 30,000 to 40,000 persons worldwide are affected by Wilson's illness. According to estimates, one in every 90 people carries the illness gene. The United States has between 2,000 and 3,000 cases; however, there may be more affected individuals who have been misdiagnosed with other neurological, hepatic, or psychological conditions.Wilson's illness typically begins with liver disease and progresses to neurological signs later. In our case, though, it was the opposite[17-25].

The tremor was the first symptom. Other case reports show that the initial presentation was related to the neurological system without involving the liver, and that the patient was over 40 years old.[9,10] Penicillamine, strangely enough, is supposed to make neurological signs worse. This is due to the mobilization of Copper from the liver, which causes an increase in unbound Copper, causing neurological symptoms to worsen. Following penicillamine medication, 30-75 percent of patients experienced initial neurological impairment, according to several studies.[23-28]

Several other reports provided evidence to the contrary. According to one estimate, 9,000 people in the United States are estimated to be affected with the tremor's condition, which was improved in our case by penicillamine and anticholinergic medication.The medicine, on the other hand, triggered the hepatic symptoms. This is the first time this information has been made public. The patient was adamant about not taking the chelation therapy, although he understood its need. Doctors began treatment with favourable results after analyzing the literature and previous case reports[29-36].

CONCLUSION:

Wilson's disease is an autosomal recessive condition that can be challenging to diagnose. However, early detection and treatment can prevent more complications and enhance the patient's quality of life.

REFERENCES:

1. Raju K, Bangalore GN, Thuruvekere SN, Pathavanalli VN. Wilson's disease: A Clinical autopsy case report with literature review. Journal of Natural Science, Biology, and Medicine. 2015 Jan; 6(1):248.
2. Cao C, Colangelo T, Dhanekula RK, Brandt D, Laothamatas I, Thapar M, Herrine SK, Civan JM. A rare case of Wilson disease in a 72-year-old patient. ACG Case Reports Journal. 2019 Mar; 6(3):1.
3. Malbul K, Katwal S, Khetan S, Aryal N. A case report on the serendipitous diagnosis of Wilson's disease in a child with brucellosis and pseudomonal infection. Clinical Case Reports. 2021 Jun;9(6):e04178.
4. Cumings JN (1948). "The copper and iron content of brain and liver in the normal and in Hepato-lenticular degeneration". Brain. 71 (Dec): 410–15. doi:10.1093/brain/71.4.410. PMID 18124738.
5. McIntyre N, Clink HM, Levi AJ, Cumings JN, Sherlock S (February 1967). "Hemolytic anaemia in Wilson's disease". N. Engl. J. Med. 276 (8): 439–44. doi:10.1056/NEJM196702232760804. PMID 6018274.
6. Cumings JN (March 1951). "The effects of B.A.L. in hepatolenticular degeneration". Brain. 74 (1): 10–22. doi:10.1093/brain/74.1.10. PMID 14830662.
7. Denny-Brown D, Porter H (December 1951). "The effect of BAL (2,3-dimercaptopropanol) on hepatolenticular degeneration (Wilson's disease)". N. Engl. J. Med. 245 (24): 917–25. doi:10.1056/NEJM195112132452401. PMID 14882450
8. Walshe JM (January 1956). "Wilson's disease; new oral therapy". Lancet. 270 (6906): 25–26. doi:10.1016/S0140-6736(56)91859-1. PMID 13279157
9. Walshe JM (March 1982). "Treatment of Wilson's disease with trientine (triethylene tetramine) dihydrochloride". Lancet. 1 (8273): 643–47. doi:10.1016/S0140-6736(82)92201-2. PMID 6121964. S2CID 205999334
10. Brewer GJ (January 2000). "Recognition, diagnosis, and management of Wilson's disease". Proc. Soc. Exp. Biol. Med. 223 (1): 39–46. doi:10.1046/j.1525-1373.2000.22305.x. PMID 10632959. Archived from the original on 2008-04-09. Retrieved 2008-05-20.
11. Harper PL, Walshe JM (December 1986). "Reversible pancytopenia secondary to treatment with tetrathiomolybdate". Br. J. Haematol. 64 (4): 851–53. doi:10.1111/j.1365-2141.1986.tb02250.x. PMID 3801328. S2CID 1154.
12. Brewer GJ. Wilson Disease. In: NORD Guide to Rare Disorders. Philadelphia, PA: Lippincott Williams & Wilkins; 2003:506
13. Mishra, P., Vagha, S., Hiwale, K., Shukla, S., 2020a. Jessner's Lymphocytic Infiltrate - A Rare Case Report. JOURNAL OF EVOLUTION OF MEDICAL AND DENTAL SCIENCES-JEMDS 9, 3898–3899. https://doi.org/10.14260/jemds/2020/854
14. Mishra, P., Vagha, S., Shukla, S., Acharya, S., Goyal, A., 2020b. Assessment of Cytokeratin Expression in Carcinoma Breast. JOURNAL OF EVOLUTION OF MEDICAL AND DENTAL SCIENCES-JEMDS 9, 2545–2549. https://doi.org/10.14260/jemds/2020/553
15. Mishra, S., Darda, P., Naqvi, W.M., Sahu, A., 2020. Regaining activities of daily living in patient with middle cerebral artery stroke- A case report. MEDICAL SCIENCE 24, 1731–1737.
16. Mistry, R.A., Pisulkar, S.K., Borle, A.B., Godbole, S.R., 2020. Evaluating Efficacy of P-6 vs. CV-24 Acupressure Points in Controlling Severity of Gag Reflex. JOURNAL OF CLINICAL AND DIAGNOSTIC RESEARCH 14, ZC1–ZC3. https://doi.org/10.7860/JCDR/2020/42672.13416
17. Modak, A., Rao, S., Singam, A., Choudhari, M., 2020. Role of an Anaesthesiologist in the Management of an Embolised ASD Closure Device - Journey from Cathlab to OT. JOURNAL OF EVOLUTION OF MEDICAL AND DENTAL SCIENCES-JEMDS 9, 3175–3177. https://doi.org/10.14260/jemds/2020/696
18. Mohan, M., Punam, S., 2020. Efficacy of Nitya Virechana with TrivritChurna in the Management of Stage 1 Essential Hypertension - A Pilot Study. INTERNATIONAL JOURNAL OF AYURVEDIC MEDICINE 11, 249–254.
19. Moon, S.H., Gawande, S., 2020. Assess the Effectiveness of Breast Crawl on the Intensity of Episiotomy Pain snd Behavioral Response Among Parturient Mothers During Episiotomy Suturing. BIOSCIENCE BIOTECHNOLOGY RESEARCH COMMUNICATIONS 13, 263–267. https://doi.org/10.21786/bbrc/13.15/45

20. Morey, A., Madke, B., Singh, A., 2020. Effect of Isotretinoin on Thyroid Function Test in Acne Patients. JOURNAL OF CLINICAL AND DIAGNOSTIC RESEARCH 14. https://doi.org/10.7860/JCDR/2020/43032.14012
21. Motwani, N., Ikhar, A., Nikhade, P., Chandak, M., Khatod, S., Zamare, V., 2020. Management of mid mesial canal in mandibular first molar - A case report. MEDICAL SCIENCE 24, 3753–3758.
22. Motwani, N.M., Ikhar, A., Nikhade, P., Chandak, M., Khatod, S., Rathi, S., Rathi, C., 2020. Dentinal Pre-Treatment in Restorative Dentistry. JOURNAL OF EVOLUTION OF MEDICAL AND DENTAL SCIENCES-JEMDS 9, 804–809. https://doi.org/10.14260/jemds/2020/174
23. Mounika, P., Shrivastava, D.S., Diwani, D., 2020. Umbilical Metastasis Secondary to Ovarian Carcinoma - A Rare Case of Sister Mary Joseph Nodule. JOURNAL OF EVOLUTION OF MEDICAL AND DENTAL SCIENCES-JEMDS 9, 3343–3344. https://doi.org/10.14260/jemds/2020/734
24. Mukherji, R., Sune, P., 2020. Contrast Sensitivity and Visual Acuity Before and After Nd:YAG Capsulotomy in Patients with Posterior Capsular Opacification: A Prospective Study. JOURNAL OF CLINICAL AND DIAGNOSTIC RESEARCH 14. https://doi.org/10.7860/JCDR/2020/43120.13590
25. Muley, P.A., Thakkar, K.R., Gehlot, P.K., Vankudre, A.J., Muley, P.P., Barde, P.B., 2020. Association between Body Mass Index and Cognitive Functions in Medical Students. JOURNAL OF CLINICAL AND DIAGNOSTIC RESEARCH 14, CC8–CC14. https://doi.org/10.7860/JCDR/2020/45081.14256
26. Mundada, A., Vaidya, V., Lamture, Y., 2020. Anorexia in acute appendicitis: A non-specific factor with significant accuracy in diagnosis. MEDICAL SCIENCE 24, 2812–2816.
27. Mundada, A.V., Lamture, Y., Yeola, M., 2020. Scoring Systems in Acute Appendicitis - A Review. JOURNAL OF EVOLUTION OF MEDICAL AND DENTAL SCIENCES-JEMDS 9, 3881–3886. https://doi.org/10.14260/jemds/2020/850
28. Mundhe, S.S., Ade, V., 2020. Study on the Effect of Varun TwakLepa in Management of Vyanga. INTERNATIONAL JOURNAL OF AYURVEDIC MEDICINE 11, 241–248.
29. Murarka, S.P., Shrivastav, S., Kamble, R., Bidwai, P.S., Gosavi, S., Merchant, Z., 2020. Aesthetic management of missing anterior teeth in Binder Syndrome - A Case report. MEDICAL SCIENCE 24, 3370–3375.
30. Nagpure, P., Modak, A., Singam, A., Shrey, S., Latwal, B., Jejani, A., 2020a. Effectiveness of Intra-Articular Ropivacaine for Post-Operative Analgesia in Arthroscopic Knee Surgeries. JOURNAL OF EVOLUTION OF MEDICAL AND DENTAL SCIENCES-JEMDS 9, 324–328. https://doi.org/10.14260/jemds/2020/74
31. Nagpure, P., Modak, A., Singam, A., Shrey, S., Latwal, B., Jejani, A., 2020b. Relationship between Perceived Social Support and Severity of Symptoms in Persons with Somatoform Disorder. JOURNAL OF EVOLUTION OF MEDICAL AND DENTAL SCIENCES-JEMDS 9, 324–328.
32. Nagpure, S.P., Wagh, V.K., 2020. Scleral Dellen after Routine Uneventful Pterygium Surgery. JOURNAL OF EVOLUTION OF MEDICAL AND DENTAL SCIENCES-JEMDS 9, 1935–1937. https://doi.org/10.14260/jemds/2020/420
33. Naik, S., Kumar, S., Hulkoti, V., Mishra, A., Hathi, D., Pratapa, S., 2020. Acute organophosphate poisoning presenting with Cerebral Infarction: Association or Chance? MEDICAL SCIENCE 24, 393–396.
34. Nambiar, K.M., Kamble, R., Suroliya, K.P., Ahuja, M.M., Daigavane, P.S., Vora, S.M., 2020. Alveolar Bone Grafting - A Summary of History and Current Literature on the Technique. JOURNAL OF EVOLUTION OF MEDICAL AND DENTAL SCIENCES-JEMDS 9, 2652–2657. https://doi.org/10.14260/jemds/2020/576
35. Naqvi, W.M., Sahu, A., 2020. Paradigmatic Shift in the Education System in a Time of COVID 19. JOURNAL OF EVOLUTION OF MEDICAL AND DENTAL SCIENCES-JEMDS 9, 1974–1976. https://doi.org/10.14260/jemds/2020/430
36. Narayane, M.M., Pohekar, S.B., 2020. Effectiveness of Planned Teaching Program on Knowledge Regarding Management of Leukaemia among Leukaemia Patients. JOURNAL OF CLINICAL AND DIAGNOSTIC RESEARCH 14. https://doi.org/10.7860/JCDR/2020/44771.14056