



Clinical Feature, Cause and Outcome in Female Paediatric Child with Osseomuscular Type of Wilson's Disease

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Authors' contributions

This work was carried out in collaboration among all authors. All authors read and approved the final manuscript.

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Case Study

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ABSTRACT

Introduction: Genetically inherited diseases have grown in the last few decades. Wilson's disease is one of those, named after the U.S.-born British neurologist Dr. Samuel Alexander Kinnier Wilson.

Case Presentation: A 12 years old female child was admitted in A.V.B.R. hospital with the chief complaints of altered behaviour, speech disturbances, no physical coordination, uncontrolled movement since 2 to 3 months and fever since 2-3 days. Golden brown eye discoloration was present. After physical examination and investigation doctor diagnosed it as a case of Wilson's disease.

The Main Diagnosis, Therapeutic Intervention and Outcomes: After physical examination and investigation doctor diagnosed this as a case of Wilson's disease. Zinc and vitamin supplements were given for 7 days to enhance immunity. Beta-blocker was given For 7days twice a day, Tab. Trietinine 250 mg was given once a day for 7 days to remove the heavy metal i.e. copper. She took

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all treatment and outcome was good. Her signs and symptoms got reduced and she was able to do her routine activities.

Conclusion: The diagnosis of Wilson's disease relies largely on clinical examination and laboratory confirmation of abnormal metabolism of copper. This case responded well to all treatment and her recovery was good.

Keywords: Wilson's disease; management; copper; MRI; splenomegaly.

1. INTRODUCTION

Wilson's disease (WD) is an autosomal recessive disease that entails a hepatic lysosome defect in the transport of copper. estimated prevalence of approximately 1:30,000 This leads to the excess accumulation of copper in the liver, brain, kidney and skeletal system, most often affecting children or young adults and, if de-copper treatment is not adequately treated, frequently contributing to lethal pathways [1].

Wilson disease is an autosomal recessive disorder triggered by a gene mutation in the Wilson disease protein (ATP7B). A copy of the gene for a person to be affected [2].

It poses a broad spectrum of clinical symptoms in infancy, adolescence or adulthood [3]. The classic presentation involves the liver disease trio plus neurological and ophthalmological participation. In the paediatric age group, liver symptoms predominate. 10 to 25 percent of cases are neurological and usually detected in adults [4]

Any unexplained acute or chronic liver disorder should increase concern of Wilson's disease in children and adolescents [5].

2. PATIENT INFORMATION

Patient specific information: Patient admitted in Acharya Vinoba Bhave Rural Hospital with the complaints of altered behaviour, disturbances speech and movements , speech became slurred and dysarthric When attempting to walk, her gait was reported to be shuffling with a tendency to tip forward. fever since 8 days and abdominal pain which was intermittent, dull, around umbilicus since 2 days.

2.1 Primary Concern and Symptoms

Altered behaviour speech and movements, fever and abdominal pain which was intermitted, dull around umbilicus since 2 days. Golden brown

eye discoloration, these were the primary symptoms which was observed at the time of admission.

2.2 Medical, Family and Psychosocial History

Patient had medical history of jaundice at the age of 6 yrs. And history of convulsion. And splenomegaly. She took treatment for that. She belongs to nuclear family. There are five members in his family. All family members are healthy except the patient. Patient looks anxious, depressed and confused.

2.3 Relevant Past Intervention with Outcome

History of jaundice 2 years back for which she was hospitalized for 7 days. After ultrasonography splenomegaly was observed she took treatment for that. And her outcome was good. She had past history of fall and followed by loss of consciousness and convulsion. Computed tomography was done and she took all necessary treatment for convulsion. after that she did not had any episode of convulsion.

2.4 Physical Examination and Clinical Finding

General examination – She was unhealthy with thin body built, hygiene was not maintained, the weight was 25 kg. Her vital parameters are normal. Her milestone developments was normal. Kayser-Fleischer ring was present in the eye. Abdominal tenderness was present, child was anxious and depressed.

Timeline: 2 yrs ago she was admitted in the hospital for 7 days for the treatment of jaundice.

She had a history of convulsion 4 yrs back. She was also hospitalized for the treatment of convulsion. Currently she was admitted for the treatment of Wilson disease. Zinc and vitamin

supplementary was given for 7 days to enhance immune function. Beta-blocker was given For 7 days in twice a day, Tab. Trietinine 250 mg was given in ones a day for 7 days to remove the heavy metal i.e. copper.

3. DIAGNOSTIC ASSESSMENTS

On the basis of patient history, physical examination, abdominal palpation and In Ultra Sonography moderate splenomegaly was observed, magnetic resonance imaging of Brain—altered single intensities seen in ventrolateral part of bilateral thalami head of bilateral caudate and lentiform nuclei, mid brain Pons and medulla. Blood investigations was also done WBC Count (1500/cu mm) than normal, Platelet count was less 0.25 lacs cu mm RBS predominantly normocytic mildly hypochromic with mild anisopoiculocytosis. and other investigations was also done. In urine examination calcium oxalate level was 8- 10 crystals/HPF. A slit-lamp examination was done by the ophthalmologist.

3.1 Diagnostic Assessment

Diagnostic challenges: No any challenges reported during diagnostic evaluation.
Diagnosis: After physical examination and investigation doctor diagnosed it as a case of Wilson disease. Prognosis was good.

4. THERAPEUTIC INTERVENTION

Medical management was provided to the patient. Zinc and vitamin supplementary was given for 7 days to enhance immune function. Beta-blocker was given for 7 days in twice a day, Tab. Trietinine 250 mg was given in ones a day for 7 days to remove the heavy metal i.e. copper. Tablet Evian once a day, Tab. Quetiapine 12.5 mg was given. She was took all treatment and outcome was good. Her sign and symptoms was reduced, she was able to do her own activity. No any change in therapeutic intervention was reported.

5. FOLLOW-UP AND OUTCOMES

Clinical and patient assessment outcomes: Patient condition was improved.

Essential medical follow-up and other test results: to avoid disease progression and to try to avoid any signs and symptoms that have arisen

due to copper accumulation [6]. Doctor advised follow up after 1 month and advised ultrasonography, blood investigation and eye examination to know the further disease progression.

5.1 Intervention Adherence and Tolerability

Patient took all prescribed medications regularly. But sometime she was refused to take medication. She also followed the dietician advised. Dietician was advised copper free content food and rich in zinc supplementation. Her interventional adherence was satisfactory.

5.2 Adverse and Unanticipated Events

None were reported.

6. DISCUSSION

Patient was admitted in AVBR hospital with the chief complaints of altered behaviour speech and movements, fever since 8 days and abdominal pain which was intermitted, dull around umbilicus since 2 days. After physical examination and investigation doctor diagnosed as a case of Wilson disease. She undergone medical management vitamin supplementary, zing and beta blocker. She was reduced the symptoms and she was feel better. Life expectancy without care is estimated to be 40 years, but patients can have a normal lifespan with timely and effective treatment. The possibility of Wilson disease should be considered in any child more than one year old presenting with any form of liver disease ranging from asymptomatic with raised liver enzymes to decompensate cirrhosis. Children with Wilson disease are usually normal at birth and may remain healthy for a variable period of time; most cases present in the second and third decade of life [7].

Together with pharmacological management To avoid or minimise the harmful effects of copper accumulation in tissues, proper treatment should be initiated as early as possible [8].

For patients with serious liver failure who do not respond to treatment and for those with complications of portal hypertension, liver transplantation is indicated where presentation is form of collateral. In this case child response to the treatment was good. Along with the other investigations Another significant diagnostic tool is genetic examination, which may validate the diagnosis in incorrect cases [9]. Severe long-

term disabilities and life-threatening complications can be avoided by early diagnosis and treatment. When symptoms improve and tests indicate that copper has been reduced to a safe amount, maintenance therapy starts. Typically, maintenance treatment involves consuming zinc and taking either d-penicillamine or Trientine hydrochloride at low doses. A health care provider should monitor blood and urine to [10]. Especially when facilities for copper studies are not available, it can be difficult to develop a diagnosis of fulminant Wilson disease. A consanguineous marriage setting, death of a sib with similar ailment and clinical features of hepatocellular failure contribute to a high suspicion index. This suspicion was supported by the bilateral Kayser-Fleischer ring under the slit lamp examination [11]. When a diagnosis of Wilson's disease in an index patient is made, it is mandatory to examine his or her relatives [12-13]. Few of the related studies were reported by Kinyoki et al. [14] and De et al. [15]. Studies on different aspects of child health [16-17], nutrition and care [18-20] were reviewed [21-25].

7. CONCLUSION

As Wilson's disease is a rare disease the diagnosis is likely to be missed. There should be a high index of suspicion in all cases of liver cirrhosis with no clear cut etiologic 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. There is no way to prevent Wilson disease. But genetic counselling may help you find out if your current or future children are at risk for the disorder.

ETHICAL APPROVAL & CONSENT

As per international standard or university standard guideline parental consent and ethical approval has been collected and preserved by the authors.

COMPETING INTERESTS

Authors have declared that no competing interests exist.

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