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HEMOLYSIS IN A PATIENT WITH WILSON'S DISEASE: A RARE PRESENTATION

DR. FAIZA BHUTTO
SICHN Hospital Karachi

Corresponding author:
Dr. Faiza Bhutto, faizabhutto62@gmail.com

ABSTRACT

Wilson's disease is an uncommon autosomal recessive inherited disease that is characterized by abnormal copper metabolism with toxic accumulation of copper in a number of organs. Hepatic, neurological and psychiatric symptoms are frequently observed, whereas hemolytic anemia is an often forgotten sign. We report here the case of a young man with Wilson's disease who presented with acute hemolysis. This case underscores the importance of inclusion of Wilson's disease in the differential diagnosis of idiopathic hemolytic anemia in young patient and emphasizes the importance of prompt diagnosis and management for a better long-term outcome.

Keywords: Wilson disease, Copper deposition disease and Hemolysis

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INTRODUCTION:

Wilson's disease (WD), or hepatolenticular degeneration, occurs due to mutations in ATP7B gene which disrupts the execcretion of copper via the biliary system. Thereafter, the excess copper is deposited in the liver, brain, cornea, kidneys, and red blood cells leading to numerous clinical presentations. The hepatic findings usually predominate in the young; neurological features tend to appear later. A rare, but potentially life-threatening, presentation is Coombs-negative haemolytic anaemia which can precede hepatic or neurological manifestations. Sudden free copper exposure to intra-vascular system, commonly from hepatic injury, can cause acute hemolysis. Early identification of this presentation may allow diagnosis and treatment with chelating agents or zinc therapy.

Case Presentation:

Patient Profile

A 12-year-old male was admitted to the emergency department during 2023 due to fatigue, dark urine, and yellow discoloration of eyes and skin for 3 days. There was no fever, abdominal pain, recent drug ingestion, or transfusion. There were no pre-existent records of illness, or comparable episodes. There was no history of liver disease or hemolytic disorder in his family.

Clinical Examination:

On examination, the patient was conscious and alert, but pale and icteric. Vital signs were normal: pulse 92/min, BP 110/70 mm Hg, respiratory rate 18/min, temperature 98.6°F. The patient did not have initial evidence of lymphadenopathy or organomegaly. On abdominal examination, mild hepatomegaly (liver span 15 cm) was observed. Neurological examination was unremarkable.

Investigations:

Initial laboratory results were as follows:
Hemoglobin: 8.2 g/dL
Total bilirubin: 5.4 mg/dL (Indirect: 4.2 mg/dL)
Reticulocyte count: 7.5%
Lactate dehydrogenase (LDH): 870 U/L
Haptoglobin: Undetectable
Peripheral smear: Polychromasia, schistocytes, no spherocytes
Direct Coombs test: Negative
Liver enzymes: AST 82 U/L, ALT 95 U/L, ALP 210 U/L
INR: 1.6
Serum ceruloplasmin: 6 mg/dL (normal 20-40)
24-hour urinary copper: 168 mcg (elevated)
Slit-lamp exam: Kayser-Fleischer rings present bilaterally.

Abdominal ultrasonography revealed mild hepatomegaly with increased echogenicity but no splenomegaly or ascites. MRI brain was normal.

Diagnosis:

A diagnosis of Wilson's disease with acute hemolytic anemia was considered in light of clinical signs, laboratory findings, and ocular findings. This diagnosis was further reinforced by low ceruloplasmin level, increased urinary excretion of copper, the finding of Kayser-Fleischer rings and abnormal liver enzymes. Hemolysis was proven by increased LDH, reticulocytosis, and indirect hyperbilirubinemia, and the negative Coombs test was indicative of a not immunological cause.

Management:

The patient was initiated on D-penicillamine, a copper chelating agent, at a dose of 250 mg twice daily, and pyridoxine (Vitamin B6). He was also recommended low copper diet (shelled fish, nuts, chocolate, and mushroom). Supportive care for hemolysis consisted of intravenous hydration and folic acid. The patient was hemodynamically stable and did not need transfusion.
His hemoglobin levels remained stable, and serum bilirubin began to trend downward over 10 days. Liver enzymes showed mild improvement. The patient was educated regarding the lifelong requirement of treatment and monitoring, and zinc acetate was proposed as maintenance therapy after initial chelation. The patient and his family were referred for genetic counseling and screening.

DISCUSSION:

Wilson's disease presents either hepatic (40-60%), neurological (30-40%), or psychiatric (10-15%) symptoms [1]. Hemolytic AHA is encountered in 10-15% of patients and is occasionally the initial clinical manifestation [2]. It is frequently misdiagnosed as autoimmune hemolytic anemia or viral hepatitis, which causes delayed treatment.
Hemolysis with Wilson's disease is of the intravascular type and Coombs-negative variety, secondary to a toxic effect of the free copper entering into the blood stream as a result of hepatic damage. The additional copper produces reactive oxygen species, which are detrimental to red blood cell membranes and lead to cell hemolysis [3].
In this situation, no evidence of splenomegaly, spherocytes, or Coombs positivity excluded hereditary spherocytosis or an autoimmune pathogenesis. The marked elevation of bilirubin with high reticulocyte count raised suspicion of hemolysis, and copper studies helped establish the diagnosis as Wilson's disease.
Kayser-Fleischer rings, observed in 90-100% of patients with a neurological presentation and up to

50% with hepatic disease, are very suggestive [4]. Slit-lamp evaluation should always be part of the diagnosis, particularly in underprivileged areas.

Management is based on life-long copper chelation therapy with penicillamine, trientine, and zinc salts interfering with gastrointestinal absorption of copper [5]. Liver transplantation is potentially curative, and indicated in cases of profound hepatic failure or advanced cirrhosis [6].

Patients who develop hemolysis respond well to medical treatment if diagnosed early as in our case. However, late diagnosis may result in irreversible hepatic and/or neurological injury.

This case highlights the need for early diagnosis of rare but clinically significant presentations such as hemolysis. It is important to detect the disease among children with unexplained hemolytic anemia and abnormal liver test, so as to prevent the long-term complications [7].

CONCLUSION:

Wilson's disease, although an uncommon cause, should be included in the differential diagnosis for unexplained hemolysis, particularly in young patients with liver disease. The first and unique symptom could be an absence of Coombs-positive hemolytic anemia. Early diagnosis with ceruloplasmin levels, urinary copper excretion and slit-lamp examination is essential. The early commencement of chelation therapy is able to revert hematologic disturbances and to postpone the progression of the disease.

Informed written consent from patient and family was acquired regarding this case report to be published. All attempts have been made to protect patient's identity.

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