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PANCYTOPENIA IN A YOUNG FEMALE: THINK DIFFERENTLY

General Medicine

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ABSTRACT

Wilson's disease is an autosomal recessive disease characterised by a copper transport defect due to ATP7B gene mutations. It usually affects children or young individuals and causes excess copper accumulation in the liver, brain, kidneys, and skeletal system. Hepatic and/or neurological involvement usually accounts for the majority of the clinical symptoms. Pancytopenia is an uncommon early manifestation of Wilson's disease. Here we are reporting a case of a 21 year old female with pancytopenia. Chronic liver disease changes in young age raised the possibility of wilson's disease, which was confirmed through the presence of kayser fleischer ring, raised 24 hour urine copper and low ceruloplasmin level.

KEYWORDS

Wilson's disease, KF ring, Pancytopenia, Ceruloplasmin

INTRODUCTION

Wilson disease (also known as hepatolenticular degeneration) is a genetic disorder of copper metabolism which is inherited as an autosomal recessive pattern that leads to impaired function of the intracellular copper transporter ATP7B[1]. The global prevalence of Wilson's disease is 12.7 per 100000[2]. Copper builds up in the liver and other tissues as a result of decreased biliary excretion (eg, brain, cornea). Majority of patients have liver involvement, which could range from asymptomatic elevations in liver biochemistries (eg, serum aminotransferases, bilirubin) to cirrhosis or acute liver failure. Some patients exhibit symptoms related to neurologic involvement[3]. The other less common manifestations of Wilson's disease include musculoskeletal and haematological forms. Here we are reporting a case of a 21 year old female presented with pancytopenia as the initial manifestation of Wilson's disease.

Case Report

A 21 year old female, LLB student, studying in Punjab, came to Medicine OPD with complaints of fever, headache, bodyache, fatigue - 5 days duration. There was no history of abdominal pain, vomiting, jaundice, bleeding manifestations, oliguria or altered sensorium. On examination she was conscious and oriented. Her vitals were stable, general examination was within normal limits except for pallor. Abdominal examination showed mild tenderness in the right hypochondriac region. Other systemic examinations were within normal limits.

The blood parameters showed a WBC count 2,900/mm3, haemoglobin 10.7 g/dL, normal MCV and platelet count 80,000/mm3. The differential leukocyte count showed 39% lymphocytes and 58% neutrophils. ESR was 30 mm/hr. Urine microscopy and urinalysis was normal. Reticulocyte count was 2%. LDH, DCT,ICT were negative. Serum Iron was low with elevated serum ferritin. Blood biochemistry revealed total serum protein 6.8 g/dL, albumin 3.9 g/dL, total bilirubin 0.54mg/dL and direct bilirubin 0.27 mg/dL, aspartate aminotransferase (AST) 126 IU/L, alanine aminotransferase (ALT) 107 IU/L , blood urea 30.5 mg/dL and serum creatinine 0.5 mg/dL. The serum electrolytes were within normal limits. The partial thromboplastin time was 37.9 seconds and the prothrombin time was 15.8 seconds. INR value was 1.18. Viral markers were all negative. Malarial parasite smear, malaria card tests were negative. Blood smear showed normocytic normochromic anaemia, leukopenia and moderate thrombocytopenia. Blood culture, urine culture, sputum culture, Sputum Truenat MTB were negative. Other possible infectious etiologies were excluded by testing antibodies to Dengue, Leptospirosis and WIDAL. Gamma GT was 187 IU/L. (Normal <40). RA factor, TSH, Serum IGG, ANA IF were negative. Abdominal ultrasonography showed Chronic Liver disease changes in the form of nodular liver margin, coarse echotexture, left and caudate lobe hypertrophy. Shear wave Elastography of the right lobe of liver yielded median value of 20.9 kPa- suggestive of clinically significant portal hypertension.

CLD in this age group raised the suspicion. Serum Ceruloplasmin level was found to be 11.2 mg/dl. (Normal value is 16-45). A bilateral Kayser- Fleischer (KF) ring was revealed in ophthalmological examination (figure 1 & figure 2). 24 hour urine copper was 419 mcg/L(Normal level is 2-80). Hence the diagnosis of Wilson's disease was established. Genetic study was done which confirmed ATP 7B gene mutation. Started oral zinc therapy. MRI Brain was normal. OGD scopy was done to assess variceal status. Showed Lower esophageal varices with no RCS, Patchy areas of erythematous mucosa in fundus, Mild portal gastropathy and antral erosions. And she was started on Cardivas 3.125 mg OD.

Pneumococcal/ Influenza vaccine, Hepatitis B vaccine, Hepatitis A vaccine and Varicella Zoster vaccinations were given. In view of CLD bystanders also have been primed regarding the option of liver transplant in future. She was advised to avoid chocolates, nuts, shellfish, red meat and mushrooms. Explained regarding family screening of first and second degree relatives.

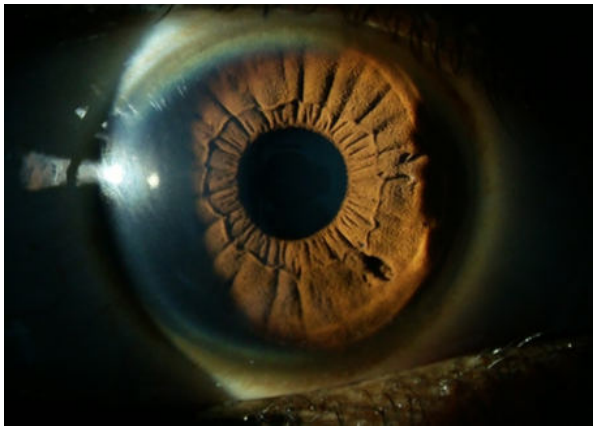

Figure 1:- KF ring in slit lamp examination

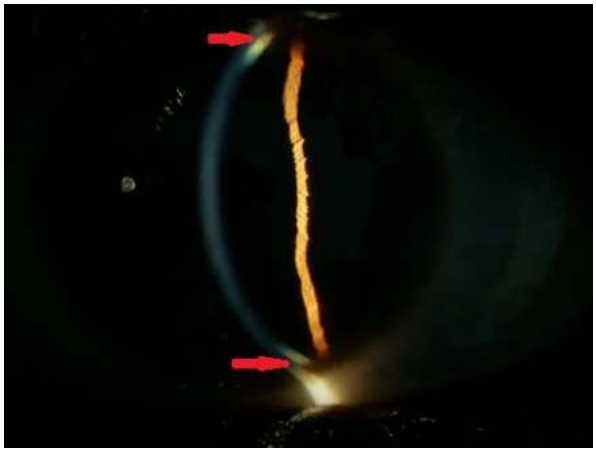

Figure- 2: Red arrows showing the KF ring

DISCUSSION

Wilson's disease is a rare inherited familial disorder characterised by progressive neurological features with evidence of cirrhosis and Kayser- Fleischer ring in the cornea. It results from a mutation in the ATP7B gene located on chromosome 13[3]. Because it takes time for copper to build up to toxic levels in the liver till such an age, the condition does not show clinical symptoms before the age of 4-5 years[4]. Clinical features in Wilson's disease includes different spectrum of liver diseases (Acute hepatitis, Chronic hepatitis, Decompensated or compensated cirrhosis, Acute fulminant hepatic failure, Persistent transaminases elevation), Neurological manifestations, Psychiatric manifestations, Eye changes, and Haematological manifestations[5]. Most important complication is Wilsonian fulminant hepatic failure.

Based on the clinical and biochemical parameters, diagnosis was made. Hematological manifestations may be the initial sign of disease, although they are uncommon. Pancytopenia has rarely been reported as an initial symptom like in our case[6]. Pancytopenia in our case can be attributed to chronic liver disease. Hemolytic anemia in wilson's disease is due to increased copper accumulation in the RBC'S causing damage to the cell membrane. The presence of a Kayser-Fleischer (KF) ring in the cornea is a useful indicator of severe copper overload. KF rings are present in 95% of patients with neurological symptoms, in 50–60% of patients without neurological symptoms and in only 10% of asymptomatic siblings. If KF rings are present and/or serum ceruloplasmin levels are low, additional tests for diagnosis are not necessary.

In our case, a 21-year-old female presented with pancytopenia. Chronic liver parenchymal changes at young age raised suspicion of Wilson's disease. An ophthalmological examination detected Kayser-Fleischer (KF) rings. Subsequent laboratory tests showed slightly elevated serum levels of alanine aminotransferase and aspartate aminotransferase, along with low ceruloplasmin levels. Elevated 24-hour urinary copper excretion confirmed the diagnosis.

Acknowledgments
None
Conflict Of Interest
None
Informed Consent
Written informed consent was taken

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