

Wilson's Disease With Anasarca: A Rare Presentation

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Abstract : Wilson's disease (WD) is an autosomal recessive inherited disorder of hepatic copper metabolism, caused by mutation in the copper-transporting gene, ATP7B leading to reduced incorporation of copper into ceruloplasmin and reduced hepatic biliary copper excretion. WD has varied clinical manifestations, most common being liver disease in younger age group and neuropsychiatric disturbances in older children. The diagnosis depends primarily on the clinical features, presence of the Kayser-Fleischer (KF) ring and biochemical parameters. We report a case of 8 year old female child who presented with anasarca and icterus for more than 3 months, which is suggestive of chronic liver disease. Ophthalmological examination showed bilateral KF ring and sunflower cataract. Investigations revealed hypoalbuminemia, hypertransaminemia with low serum ceruloplasmin and high urinary copper level, hence a diagnosis of Wilson's disease was made. Out of the various presentations of WD, anasarca is the rarest, which our patient had, this makes our case unique.

Keywords : Wilson's disease, Anasarca, Kayser-Fleischer ring, Sunflower cataract.

I. Introduction

Case Report

An eight years old female child born of non consanguineous marriage presented with anasarca and icterus for 3 months. The edema initially started as periorbital puffiness and gradually progressed to all over the body. On examination, her vitals were stable. Child had pallor, icterus and generalized edema which was pitting type. Liver was 2cm palpable below right subcostal margin with a span of 8cm, smooth surface, soft to firm in consistency and no evidence of free fluid in abdomen.

Ophthalmological examination revealed KF ring (figure 1) and sunflower cataract (Figure 2). On further investigation, haemoglobin-9.7, MCV-102.3. Peripheral smear showed macrocytic anemia, reticulocyte count was normal and Direct Coomb's Test was negative. Liver function showed high serum bilirubin (2.5 mg/dl with direct bilirubin 1.5 mg/dl), low albumin (2.1 gm/dl) and elevated enzymes. Coagulation profile was deranged. Urine examination was normal and USG abdomen confirmed hepatomegaly with mild ascites. Serum ceruloplasmin was low and urine copper level was high. ANA, HIV, HBsAG, HCV and HAV were negative. Liver biopsy was not done as the parents were not willing.

The diagnosis of WD was made on the basis of clinical and laboratory findings. She was started on D Penicillamine, zinc acetate, vitamin B6 and was put on copper free diet. With the above treatment, child improved in general well being and decrease in edema. Family screening was negative for WD.

II. Discussion

Wilson's disease or hepatolenticular degeneration is a rare disorder of copper transport in hepatic cells resulting in abnormal deposition of copper in liver, eyes, basal ganglia and other tissues¹. Reported incidence of Wilson's disease is 1:30,000. The abnormal gene for Wilson disease is localized to the long arm of chromosome 13 and encodes for ATP7B, which is mainly expressed in hepatocytes and is necessary for copper incorporation into ceruloplasmin and biliary copper excretion².

WD initially presents with hepatic manifestations in younger children and predominantly neurological and neuropsychiatric illness in later decade of life³. The most common presenting neurologic feature is asymmetric tremors whereas dysarthria, speech changes, parkinsonian features and incoordination can also occur. Psychiatric manifestations include impulsive behavior, phobias and depression. In hepatic manifestations, WD can present either as chronic active hepatitis, cirrhosis or fulminant hepatic failure⁴. Our patient was in pre adolescent age group presented as chronic active hepatitis. Edema was attributed to low protein, especially albumin due to decreased synthetic function of liver. She also had anemia with macrocytosis, which is a known fact in chronic liver disease⁵. To the best of our search, anasarca is associated with WD but as an initial presentation is rare. She had KF ring and sunflower cataract. KF ring and sunflower cataract is due to deposition of copper in the descemet's membrane and lens respectively. Sunflower cataract is also a rare manifestation of WD⁶. Early detection of KF ring can lead to early diagnosis and management.

Treatment of WD includes restriction of dietary copper intake <1mg/day and pharmacological treatment with chelating agent, D penicillamine in a dose of 20mg/kg/day or trientine at a dose of 20mg/kg/day⁷. They also require zinc supplement lifelong as it impairs decreased absorption of copper from gastrointestinal tract. Vitamin B6 should also be supplemented as penicillamine is an antimetabolite for this vitamin. Liver transplantation is indicated in patients with decompensated cirrhosis and fulminant liver disease⁸. In the mean while, Molecular Adsorbents recirculating system (MARS), an extracorporeal liver support system can be used to act as a bridge to liver transplant⁹.

III. Conclusion

A high index of suspicion for WD is required when a patient presents with chronic liver disease. Early diagnosis and initiation of treatment can prevent catastrophic outcome.

Declarations

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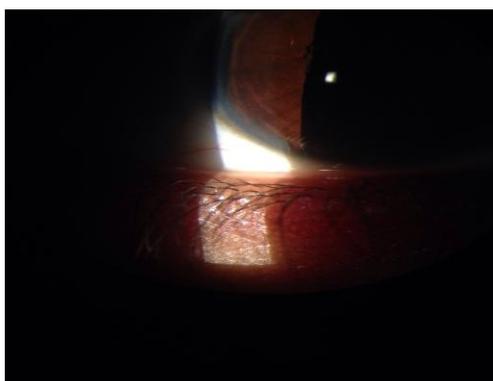


Figure 1 - Showing Kayser-Fleischer ring



Figure 2 – Showing Sunflower cataract