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An Elusive Case of Wilson Disease: No KF Rings, Multiple Atypical Features

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Abstract

Wilson disease is a rare genetic autosomal recessive disorder of Copper metabolism associated with ATP 7B gene on Chromosome 13, that often manifests in the form of hepatic dysfunction in earlier stages and neuropsychiatric manifestations in later stages with evidence Kayser Fleischer ring. Early diagnosis of Wilson disease hold a prognostic value and delay in diagnosis of WD or atypical presentations adds complexities or challenges in diagnosis. We report a middle aged female who presented with neuropsychiatric symptoms for six years with mild hepatic dysfunction undergone evaluation and diagnosed as Wilson disease based on scoring system. Interestingly our case does not have Kayser Fleischer ring, which is Pathognomic of Wilson disease. More than one century after the first description of Wilson disease by Sir K. Wilson, understanding and management of Wilson disease have improved but challenges in diagnosis of atypical presentation of Wilson disease makes delay in diagnosis

and thereby affecting the prognosis. In case of atypical presentation we can use Scoring system to diagnose Wilson disease.

Keywords: Wilson disease, Kayser Fleischer Ring, Copper metabolism

Introduction

Wilson disease is a rare Autosomal Recessive disorder cause by mutation of ATP 7B gene of Chromosome 13 resulting in impaired copper excretion into bile and failure to incorporate Copper to Ceruloplasmin[1]. Wilson description of "Progressive Lenticular Degeneration: A Familial Nervous Disease associated with Cirrhosis of the Liver" appeared in 1912 [2]. A Similar neurologic disorder had been described previously by Gowers (1906) under the title of "Tetanoid Chorea" and by Westphal (1883) and Strumpell (1898) as "Pseudo sclerosis". Copper deposited in tissues is responsible for the hepatic and neurological changes, the greenish brown pigmented rings in the periphery of the cornea (Kayser – Fleischer rings) [3] and the lesion in kidneys and other organs. Tissue damage leads to cirrhosis of liver and bilateral degeneration of basal ganglia. The Wilson disease is found worldwide, with an average prevalence of 1 in 30,000- 50,000. The carrier frequency is 1 in 90 [4]. Most patients present between 3 -55 years of age.

Clinical presentation can vary widely, but the key features of Wilson disease are liver disease and cirrhosis, Neuropsychiatric disturbances, Kayser–Fleischer rings in Descement's membrane of the Cornea, and acute episodes of hemolysis often in association with acute liver failure. Wilson disease is not just a disease of children and young adults, but may present at any age [EASL]

Signs of liver disease are nonspecific, but any liver disease of unknown origin should be considered as Wilson disease until proved otherwise. Diagnostic vigilance is important because Kayser–Fleischer rings may be absent in up to 50% of patients with Wilson disease affecting the liver. About one-third of patients initially present with psychiatric abnormalities.

A delay in diagnosing Wilson disease in patients with neuropsychiatric presentations is frequent. Here we have a case of middle aged female with predominant neuro psychiatric manifestations and discussed below.

Case Report

28 year old female patient diagnosed as psychiatric disorder for 6 years with history of intake of antipsychotics for a few months after diagnosis 6 years back presented with complaints of yellowish discolouration of urine for 1 week and decreased food intake. Patient had history of self talk, unexplained cry, hallucinations with episodes of mania and depression for the past 6 years. Patient also had history of jaundice 6 years back and subsided with native medications. Initially patient had good academics during the high school and higher secondary education and patient academic activities declined and stopped the college studies from second year. Patient is able to do all the activities of her own initially and for the past 2 years she is dependent on her mother and unable to perform the daily activities on her own. Her mother denies no other family members have similar complaints. On further evaluation patient is concious, oriented, depressed mood and irritable occasionally. Her grooming, hygiene and self-care is poor. She is pale and icteric. Neurological examination reveals bradykinesia, dystonia and resting tremor was present. No other significant neurological signs present. No neurocutaneous markers or signs of liver cell failure present. No evidence of KF ring or catatarct on ophthal/slit lamp examination.

Investigations

Hemoglobin	8.1g/dl	PT/aPTT/ INR	14.3/23.3/1.07
Total count	12600cells/cmm	T3/T4/TSH	50/9.0/2.3
Platelet /Hct	2.47 lakhs/30.3	HIV	Non Reactive
MCV/MCH/MCHC	97/33/29	HBsAg/Anti HCV	Negative/Negative
ESR/CRP	60/38	HAV IgM/HEV Igm	Negative/Negative
RBS/HbA1C	118mg/dl/ 4.7%	DCT/ICT	Negative/Negative
Urea/creatinine	19/0.9 mg/dl	CSF protein	87mg/dl
Sodium/ potassium	134/4.2	CSF glucose	69mg/DL
Calcium/LDH	8.6/508	CSF globulin	Negative
Bilirubin-Total/direct/ indirect	3.1/1.82/1.25	CSF Encephalitis panel	Negative
SGOT/SGPT/ALP	22/33/70	CSF Cytology	Acellular
T.Protein/Alb/Glob	4.88/2.38/2.5	CSF C/S	Negative
Sr. Vit B12	220 pg/mL	Sr. Vit D3	62 ng/mL
Peripheral smear study	Mild anisopoikilocytosis, microcytic hypochromic RBCs admixed with macrocytes & macroovalocytes WBCs & platelet adequate and normal in shape, Reticulocyte count 1.8%		
USG Abdomen & Pelvis	Grade III fatthy liver No other significant abnormality		
ECHO	Normal study , LVEF 65%		
MRI Brain	Mild diffuse cerebral atrophy & bilateral basal ganglia calcification		

Liver biopsy: Steatohepatitis with focal perivenular fibrosis. NAS CRN: 4 Fibrosis: 1a

Discussion

Wilson disease (WD) is an autosomal recessive genetic disorder of copper metabolism which leads to toxic accumulation of copper in the liver, nervous system and other organs [5]. Disease causing mutations on both alleles of ATP 7B resulting WD, in which defective biliary excretion of copper and the absence of holo ceruloplasmin complex (five molecules of copper bound to ceruloplasmin) lead to copper accumulation in body organs [6]. Heterozygotes have no disease symptoms. Increased levels of non-protein-bound toxic copper within the hepatocytes lead to hepatitis and cell death, with subsequent release of copper into the circulation. Since the maturation process of ceruloplasmin requires functional intact ATPase 7B, decreased mature ceruloplasmin is secreted into the circulation due to decreased or absent ATPase 7B activity in hepatocytes.

Wilson disease is not just a disease of children and young adults, but may present at any age [7]. Wilson disease may present symptomatically at any age, although the majority presents between ages 5 and 35. The youngest patient reported with cirrhosis due to Wilson's disease was 3-years-old [8]. About 3% of patients present beyond the fourth decade, either with hepatic or neurologic disease [7]. The oldest patients diagnosed were in their eighth decade [9].

The major manifestations of Wilson disease are hepatic, neurological, neuro psychiatric, pure psychiatric and asymptomactic. Kayser- Fleischer Ring is golden brown colour peripheral deposition of copper in the descement membrane and diagnosed on slit lamp examination. The rings are present in 95% of neurological involvement and 40-50% of hepatic involvement [10]. Both male and female are equally involved. The incidence of hepatic involvement is common in female patient and neurological /neuropsychiatry involvement most common in males.

Hepatic manifestation of Wilson disease range from asymptomatic to progressive jaundice, ascites, hepatic encephalopathy, cirrhosis, coagulopathy & Fulminant hepatic failure. Hepatic destruction is due to rampant apoptosis, presumably due to accumulation of copper [11]. Acute Coombs Negative intravascular hemolysis is due to destruction of erythrocytes by a sudden flux of copper from leaky hepatocytes [12]. Liver biopsy may show simple steatosis to steatohepatitis, autoimmune hepatitis [13] or cirrhosis.

The neurological manifestation of Wilson Disease is classified as three syndromes, based on signs & symptoms as Tremor & ataxia, bradykinesia, and dystonia [14]. Movement disorders are often associated with dysarthria, gait and postural disturbances, drooling and dysphagia. Tremor may affect single limb or the whole body, and it resembles a combination of

Cerebellar dysfunction and Parkinsonism. Postural tremor is commonest and there is wild 'wing beating tremor' or flapping of arms which may injure chest or abdomen [15]. KF rings are present in almost all patient with Neurological presentation of Wilson disease [16].

The psychiatric symptoms of Wilson disease such as mood disturbances, abnormal behaviour (increase irritability or disinhibition), anxiety, depression are present [17]. About one third patient have declining school performance, personality changes, impulsiveness, labile mood, sexual exhibitionism and inappropriate behaviour may be observed [18]. Less common manifestations include acromegaly, azure lunulae (bluish discolouration of lunula), renal abnormalities (aminoaciduria, tubular acidosis, hypercalciuria and nephrocalcinosis)[19], cardiomyopathy[20], myopathy[21], chondrocalcinosis, osteoarthritis [22], bone demineralisation [23], risk of fracture [24], hypoparathyroidism[25], pancreatitis[26], infertility or repeated miscarriages [27].

In our case the patient had neurological, neuropsychiatry manifestations and biochemical abnormalities. The serum ceruloplasmin level is low normal of 26 mg/dl (20-80mg/dl) , increased 24 hours urine copper 146.98 µg/L (2.0- 80 µg/L), decreased serum copper level 68 mcg/dl (80-155 mcg/dl) with absent KF ring. This makes the diagnostic challenge of Wilson disease. So we proceeded with the Scoring system used for diagnosis of Wilson disease such as Leipzig score [28] and Ferrenci scoring system [29].

Modified LEIPZIG Score

PARAMETER	SCORE	PATIENT
KAYSER FLEISCHER RING (PRESENT /ABSENT)	2/0	0
Serum CERULOPLASMIN mg/dl (>20/11-20/6-11/0-5)	0/1/2/3	0 (26)
24 hour urine Copper mcg (in absence of cholestasis) >100/40-100/<40	2/1/0	2(146.98)
Coombs negative Hemolytic anemia (present/absent)	1/0	1
Mutation Analysis (on both chromosome/ one chromosome/ absent or not done)	4/1/0	0
Liver Biopsy suggestive of wilson's disease with orcein or rhodamine staining (present/absent)	1/0	0
Neurobehavioural symptoms(present/ absent)	2/0	2
Typical MRI finding (Present/ absent)	1/0	0
Family history of Wilson disease (present /absent)	1/0	0
score		5

Evaluation: Score >_4 diagnosis established

3 probable

<2 unlikely

Serum ceruloplasmin is the major carrier of copper in blood. Ceruloplasmin is an acute phase reactant possessing a ferroxidase activity [30]. Serum ceruloplasmin concentrations are elevated in acute inflammation, hyper estrogenemia states. The levels are typically low in neurologic Wilson disease, but may be low normal in about half of patient with active Wilson disease[31]. A prospective study on serum ceruloplasmin, as a screening test for Wilson disease in patients referred with liver disease, showed that subnormal ceruloplasmin had a positive predictive value of only 6%. In children with Wilson disease, 15–36% had ceruloplasmin in the normal range [32]. In one series, 12 out of 55 Wilson disease patients had normal ceruloplasmin and no Kayser–Fleischer rings[33]. In our patient have low normal serum ceruloplasmin levels with no KF rings.

Although a disease of copper overload, the total serum copper (which includes copper incorporated in ceruloplasmin) in Wilson disease is usually decreased in proportion to the decreased ceruloplasmin in the circulation. In patients with severe liver injury, serum copper may be within the normal range, independent of whether serum ceruloplasmin levels are elevated or low[31]. The serum non ceruloplasmin-bound copper concentration has been proposed as a diagnostic test for

Wilson's disease55. Our patient had low serum copper levels.

The amount of copper excreted in urine in a 24 hour period may be helpful for diagnosing Wilson disease and monitoring treatment [31]. In untreated patient the 24 hour urinary copper excretion reflects the amount of non ceruloplasmin bound copper in the circulation. Urinary copper excretion with D-penicillamine administration was thought to be a useful diagnostic test. This test has only been standardized in a pediatric population in which 500 mg of D-penicillamine was administered orally at the beginning and again 12 h later during the 24-hour urine collection, irrespective of body weight [34]. Compared with a spectrum of other liver diseases, including autoimmune hepatitis, primary sclerosing cholangitis, and acute liver failure, a clear differentiation was found when more than 25 μ mol/24 h was excreted. The penicillamine challenge test has been used in adults, but many of the reported results of this test utilized different dosages and timing for administration of the D-penicillamine [35]. Thus, this test is not recommended for diagnosis of Wilson disease in adults. In our patient had 24 hour urine copper of 146.98μg/L which is diagnostic of Wilson disease.

The liver biopsy is only required if the clinical signs and non-invasive test do not allow a final diagnosis or if there is suspecion of other or additional liver pathologies[36]. The earliest histologic abnormalities include mild steatosis (micronodular and macronodular), glycogenated nucleic in hepatocytes and focal hepatocellular necrosis [36]. Frequently this will be misdiagnosed as Non Alcoholic Fatty Liver disease or Non Alcoholic Steato Hepatitis. The biopsy may also show features of auto immune hepatitis or chronic active hepatitis. Almost half of the patient has cirrhosis at the time of diagnosis [37]. Apoptosis of hepatocytes is a prominent feauture during acute injury [38]. In our patient have NASH pattern on liver biopsy. Detection of copper in hepatocytes by routine histochemical evaluation is highly variable. Especially in early stages of the disease, copper is mainly present in the cytoplasm bound to metallothionein and is not histochemically detectable [39]. The amount of copper varies from nodule to nodule in the cirrhotic liver and may vary from cell to cell in pre-cirrhotic stages. The absence of histo chemically identifiable copper does not exclude Wilson disease. Lysosomal copper complexes can be stained by various methods, including the rhodamine or orcein stain.

Hepatic copper accumulation is the hallmark of Wilson disease. However specific stains like rhodamine or orcein reveal copper stores in less than 10% of patients because they can detect only lysosomal copper depositions. Hepatic copper content more than 4 μmol/g dry weight is considered as best biochemical evidence for Wilson disease.

The neuroimaging MRI brain in Wilson disease may detect structural abnormalities in the Basal Ganglia [40]. The most frequent findings are increased density on T2 MRI in basal Ganglia. A characteristic finding in Wilson disease is the "face of Giant Panda sign" [41] but found only in minority of patients. Besides this sign hyperintensities in tectal plate and central pons and simultaneous involvement of Basal Ganglia, thalamus and brainstem are virtually pathognomic of Wilson disease[42]. Our patient has calcification of bilateral basal ganglia with cerebral atrophy.

Direct molecular-genetic diagnosis is difficult because of more than 500 possible mutations; except for a few more frequent mutations, each of which is rare[43]. Furthermore, most patients are compound heterozygotes (i.e. carry

two different mutations). Comprehensive molecular-genetic screening takes several months, which makes this an impractical method. Nevertheless, it is reasonable to perform molecular analysis of the ATP 7B gene in any patient who has a provisional diagnosis of Wilson disease, both for confirmation purposes and to facilitate the subsequent screening of family members.

Treatment

A number of drugs are available for the treatment of Wilson disease, including D penicillamine, trientine, zinc, tetrathiomolybdate, and dimercaprol. Copper Chelation can be done with D.Pencillamine and Trientene, which is used for Initial treatment as well as maintainance. D.Pencillamine also enhances Urinary excretion of Copper and dose is 1 to 1.5gm/day in 3 to 4 divided doses per oral available as capsule. Worsening of neurologic symptoms has been reported in 10–50% of patients treated with D-penicillamine during the initial phase of treatment [44]. Trientene was introduced in 1969 alternative to D- Penicillamine, acts as a copper Chelator and also promotes urinary copper excretion. Trientene are given in dose of 900 to 2700mg/day in 2 or 3 divided doses, with 900- 1500mg/day for maintainance therapy. Zinc interferes with the uptake of copper from the gastrointestinal tract [45]. Zinc is used for maintainance. Different zinc salts (sulphate, acetate, gluconate) are used. The recommended dose is 150mg elemental zinc/day (for children <50kg in body weight 75mg) administered in three divided doses, 30min before meals. Although zinc is currently reserved for maintenance treatment, it has also been used as first-line therapy, most commonly for asymptomatic or presymptomatic patient. The outcome of exclusive zinc therapy was generally good in cases of neurologic disease. A less satisfactory outcome in hepatic disease may relate to less efficient de-coppering. Liver transplantation46 is indicated in acute liver failure, primary treatment failure and non adherence to hepatic decompensation.

Conclusion

Wilson Disease can present with atypical features, making clinical diagnosis challenging. The use of standardized scoring systems provides valuable support in identifying such cases. Even in patients with predominant neuropsychiatric symptoms, Wilson Disease must be actively ruled out. Early recognition is crucial, as delays in diagnosis can significantly postpone clinical improvement. Timely evaluation and treatment therefore remain essential to optimize patient recovery.

No single investigation will be diagnostic of Wilson disease. In a suspected case with symptoms of hepatic, neurologic or neuropsychiatric manifestations predominantly of young age should undergo complete evaluation of Wilson disease and diagnose based on the Scoring system (Leipzig [28] and ferrenci Scoring [29]), so by earlier the diagnosis better the prognosis. In our patient there has been delay of around 6 years to diagnose the Wilson disease.

In some rare cases, the first manifestations of the disease can be psychiatric which, according to the literature, accounts for only 10% of the cases. The disease can be revealed by isolated behavioral problems, an irrational syndrome, a schizophrenic syndrome, or a manic-depressive syndrome.

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