Case Report

Wilson's Disease: A Case Report

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Abstract:

Wilson's disease (WD) is a disorder of copper metabolism, which results in accumulation of copper in different organs. We report a case of 12 year old female was admitted to our hospital with complaints of fever, cough, cold and pain in abdomen since 12 days associated with yellowish discolouration of eyes and skin. Ophthalmological examination showed Kayser-Fleischer rings on both the cornea. Though Wilson's disease is a rare entity but it needs quick recognition and prompt management, to prevent the hepatic encephalopathy which can be life threatening.

Key words: Wilson's disease, Kayser-Fleischer ring, Hepatic encephalopathy, Conjugated hyperbilirubinemia, Ceruloplasmin, Haemolysis

Introduction:

Wilson's disease (WD) is rare inherited disorder of copper metabolism. The condition was first defined by Dr. Samuel Alexander Kinnier Wilson in 1912.[1] In Wilson's disease there is defective biliary excretion of copper leads to its accumulation, particularly in liver and brain.[2,3] Wilson's disease is due to mutations of the ATP7B gene on chromosome 13, which encodes a copper-transporting P-type ATPase (ATP7B) residing in the trans-Golgi network of hepatocytes.[4,5] ATP7B is responsible for transporting copper from intracellular chaperone proteins into the secretory pathway, both for excretion into bile and for incorporation into apo-ceruloplasmin for the synthesis of functional ceruloplasmin.[4, 5] Clinical presentation can vary widely, but the key features of Wilson's disease are liver disease and cirrhosis, neuropsychiatric disturbances, Kayser–Fleischer rings in Desçemet's membrane of the cornea, and acute episodes of hemolysis often in association with acute liver failure. Wilson's disease is not just a disease of children and young adults, but may present at any age.[6]

Case report:

A 12 year old female was admitted to our hospital with Chief complaints of fever, cough, cold and pain in abdomen since 12 days associated with yellowish discolouration of eyes and skin since 4 days.

Patient was apparently alright 12 days ago when she developed fever which was of sudden onset, moderate grade mainly at night and not relieved on medications, fever was associated with chills and rigors. Cough was dry in nature relieved on medication and associated with cold with nasal blockage and relieved on medication. She developed yellowish discolouration of eyes and yellow colour urination. No History of loose stools was present, patient was suffering from vomiting since 12 days which was whitish in the start then yellowish in colour. Vomiting occurred immediately after feed since the past 2 days.

There was no significant past history and family history. On general examination the anthropometry was as follows, weight was 12.8kg, height was 100cm, chest circumference was 50cm, mid arm circumference was 58cm.

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ISSN No. *: (p) 2348-523X, (o) 2454-1982*

DOI: 10.46858/vimshsj.10106

Date of Published : *31st March 2023*

On abdominal examination abdomen was soft and was tender in the right hypochondrium, liver was slightly palpable (grade 1), while CVS, RS and CNS systemic examination were normal. Patient was admitted in PICU and started on, intravenous injection meropenem, and vancomycin in hepatic doses were given for 2 days along with 3 bags of Fresh frozen plasma, and also intravenous injection of vitamin K for 3 days, ondensetron and pantoprazole was given. Tablet doxycycline was given for 2 days, syrup lactulose was given I/V/O hyperammonemia. Patient had one episode of convulsion which was characterized by up rolling of eyes followed by unconsciousness which lasted for 2 mins after which injection leviteracetam was started.

USG of abdomen and pelvis shows acalculus cholecystitis along with Mild ascites.

Ophthalmological examination showed Kayser-Fleischer rings on both the cornea.

On blood investigation, complete blood count was normal. The serum ceruloplasmin levels were decreased, observed value: 14.6 mg/dl (normal limits-30-65). Activated partial thromboplastin time was increased, observed value:70.6 sec (normal limits:24-36 secs). Prothrombin time was also increased, observed value:56.3 secs (normal limits: 10.3-13.9 secs). Total bilirubin was 19.42mg/dl (normal-0.3-1.2mg/dl)

The patient was falling under the King's college criteria for acute liver failure,[7] hence referred to higher center for further management.

Discussion:

Although the form of the disease initially described was predominantly neurological [1], the disease manifestations can be pleomorphic, and although the correlation mutation-predominant manifestation has been elusive,[8,9] clinical forms of the disease tend to cluster and wide geographical differences exist.WD may Be predominantly hepatic, neurological or psychiatric, and manifestations of disease may range from an asymptomatic state to life-threatening fulminant hepatic Failure.[10] Acute transient hepatitis is the mode of presentation in 25% of those in whom hepatic symptoms herald disease onset. Wilson's disease can also make its appearance as acute fulminant Hepatitis. The mortality rate with this mode of presentation is alarmingly high; individuals typically are younger than 30 years and two-thirds are female.[11] A severe Coombs-negative hemolytic anemia, presumably due to intravascular hemolysis triggered by the sudden release of massive amounts of copper into the bloodstream from the failing liver, is often present.[12]

The average age of neurological dysfunction is 18.9 years, although neurological symptoms may appear as early as age 6 years.[13] Tremor, which may be resting, postural, or kinetic, is the most frequent initial neurological feature of Wilson's disease. Proximal upper extremity tremor may Take on a coarse, "wing-beating" appearance, but Wilson's disease tremor may also be distal and quite small in Amplitude. Dysarthria is also common in persons with Wilson's disease and may possess either an extrapyramidal or a cerebellar character. A peculiar "whispering dysphonia" has been described in Wilson's disease, as has a laugh in which most of the sound is generated during inspiration. Gait abnormalities are a frequent component of Neurological Wilson's disease; both extrapyramidal and Cerebellar patterns may develop.

Psychiatric symptoms appear at some point in time in most individuals with Wilson's disease, and most frequently in persons who also display neurological dysfunction.

Kayser-Fleischer rings are formed by deposition of copper within Descemet's membrane. Excess Copper is actually deposited throughout the cornea in Wilson's disease, but it is only in Descemet's membrane that sulfur-copper complexes are formed, producing the Visible copper deposits.[14] Kayser-Fleischer rings are Almost always bilateral, but unilateral formation has been Reported. The color of the rings can range from gold to brown to green; consequently, they can be difficult to see in individuals with brown irises.

Other manifestations of disease may include renal abnormalities such as hypercalciuria, nephrocalcinosis, nephrolithiasis, and aminoaciduria, cardiomyopathy with arrhythmias, autonomous nervous alterations, gigantism, hyoparathyroidism, osteoarthritis, pathological fractures and pancreatitis

Conclusion:

Considering the rarity of Wilson's disease its manifestations likely to be missed. In all cases of liver cirrhosis with an undetermined cause or a solitary neurological symptom like tremor, there should be a high degree of suspicion. Identifying disease in its early stage not only help to initiate appropriate management but also help to reduce the mortality especially in resource poor set up.

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