Wilson's Disease in a Young Boy with Hepatic Failure and Vitamin - D Deficiency

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INTRODUCTION: Wilson's disease is a rare metabolic disorder involving copper metabolism, may present with hepatic, neurological and psychiatric manifestations. The degeneration was first described in 1912 by Kinnear Wilson as progressive lenticular degeneration. Wilson's disease affects 1 in 30,000–100,000 individuals and usually presents in the second to third decades of life. WD has worldwide frequency of 1 in 40,000 and a carrier frequency of 1 in 9. WD has worldwide frequency of 1 in 40,000 and a carrier frequency of 1 in 9. WD has worldwide frequency of 1 in 40,000 and a carrier frequency of 1 in 9.

CASE REPORT: A 14 years boy from uzbekistan came to our Asian tertiary care hospital,Faridabad,India with left sided posturing and weakness since 2 years which started in left lower limb followed by left arm,difficulty in walking,difficulty in chewing and swallowing,tremors in both hands,speech disturbed,initially it started with an episode of jaundice. VITALS: BP 100/60mmhg, pulse 76/min, afebrile, respiratory rate 22/min, CVS: S1S2+LUNGS: b/l clear; P/ABDO: soft, non tender, spleen palpable, bowel sounds+: CNS: conscious, coherent, oriented, following commands, EPs examination: cog wheel rigidity, lead pipe rigidity, bradykinesia, foot dystonia present, cranial nerves normal, corneal examination bilateral KA ring seen, power bilateral symmetrical, EOM: Slow. ON INVESTIGATIONS:-HB 10.7 TLC 2500 Platelet 38000 PT 13.4 INR 1.02 APTT 33.2 Creatinine 0.4 BUN 13 sodium 132 Potassium 3.7 Chloride 108 S.Bilirubin 2.19 S.Protein 6 S.Albumin 4 SGOT 1309 SGPT 1548 S.Vitamin D 9.8 and S.vitamin - B 12- 431, G6PD normal coombs test both direct and indirect negative, reticulocyte count 1.5 peripheral smear showed microrycic normochromic leucopenia, reduced platelets on smear. MRI brain scan showed near symmetrical areas of altered signal intensity involving bilateral lentiform nuclei, bilateral thalami, midbrain and pons, with few tiny foci of restricted diffusion in the pons along with mild atrophic changes in the brain likely possibility of Wilson's disease. USG whole abdomen showed liver with diffuse coarse and heterogenous echotexture suggestive of chronic liver disease. Portal vein appear prominent, 13-14 mm, gall bladder is partially distended with edematous wall, retroperitoneum is obscured by bowel gases, spleen markedly enlarged in size-marked splenomegaly, splenic vein is prominent. Collaterals seen near splenic hilum and porta hepatitis region. Slit lamp examination showed bilateral KF ring. IMD panel showed methionine raised. Serum copper and ceruloplasmin levels were low and 24 hours urinary copper was high. Patient was consulted by physician, gastroenterologist, physiotherapist and ophthalmologist and advise followed. Patient was treated with tab syndopa, tab zinconia, calcio salt, tab laretol, tab pencillamine, tab benadon, tab folvite.

DISCUSSION: Wilson's disease is an inherited disorder leading to accumulation of copper in tissues, mainly in the liver and brain. Genetic defect is in the gene coding ATPase type P (ATP7B). The inheritance is autosomal recessive. Up to now, more than 500 mutations causing Wilson's disease were described. The manifestation of Wilson's disease is usually hepatic or neurologic. Hepatic form is manifested by acute or chronic hepatitis, steatosis or cirrhosis. Neurologic involvement is manifested usually after 20 years of age by motor disturbances (tremor, disturbed speech, problems with writing), which could progress into severe extrapyramidal syndrome with tremor, rigidity, dysarthria, dysphagia and muscle contracture. The neurological features of WD are primarily due to the deposition of copper in the lenticular nuclei, although areas like the brainstem and cerebellum can be affected.
Patients typically present in the second to the fourth decades of life with liver disease, a neurological disease of the movement disorder type, or a wide array of behavioural disturbances.

Once the disease comes under consideration, a series of diagnostic steps can be undertaken which almost always establish or rule out the diagnosis of Wilson's disease. These include urine copper, blood ceruloplasmin, slit lamp examination for Kayser-Fleischer rings, and liver biopsy with quantitative copper assay. Confirmation of diagnosis is done by hepatic copper concentration in liver biopsy or by genetic examination.

Serum ceruloplasmin and/or serum copper levels are less sensitive and specific in identifying patients with ALF-WD than other available tests. Serum copper was the most useful biochemical test in diagnosing Wilson's disease before death. More readily available laboratory tests including alkaline phosphatase, bilirubin and serum aminotransferases by contrast provides the most rapid and accurate method for diagnosis of Acute liver failure due to WD. The alkaline phosphatase-total bilirubin ratio of less than 2.0 provided 100% sensitivity and specificity in identifying fulminant hepatic failure caused by Wilson's disease from other types of fulminant hepatic failure.

One can use scoring system proposed by the 8th International Meeting of Wilson Disease and Menkes Disease to make a definite diagnosis in our patient. the brain.

Because the manifestations of Wilson's disease are so protean, and the disease masquerades so well as something else, recognition of the possibility of Wilson's disease is a major problem, leading to serious underdiagnosis of the disease. There are some diseases of copper transport such as Menkes disease, Occipital horn syndrome, Indian childhood cirrhosis, Neurodegenerative diseases which should be differentiated from WD

Fulminant hepatic failure with Wilson's disease differed from idiopathic fulminant hepatic failure by the following biochemical findings: (a) higher copper levels in serum, urine and liver; (b) less pronounced elevations of transaminase levels; (c) higher concentrations of total bilirubin; and (d) lower hemoglobin values.

Excellent therapies exist for both the prophylaxis and treatment of Wilson's disease. The longer recognition and diagnosis are delayed, the greater the risk of permanent damage to liver and/or brain. The availability of effective therapy and the risks in delay or therapy make the earliest possible diagnosis critical.

Diet: In general, a diet low in copper-containing foods is recommended with the avoidance of mushrooms, nuts, chocolate, dried fruit, liver, and shellfish.

Treatment is based on chelating agents decreasing the copper content by excretion into urine (D-penicillamine, trientine) or on agents preventing absorption of copper from food (zinc, ammonium-tetrathiomolybdate). Patients with asymptomatic Wilson's disease have to be treated as well.. Liver transplantation is indicated in patients with fulminant liver failure or decompensated cirrhosis. Screening in families of affected patients (all siblings) is obvious.

Prognosis

Left untreated, Wilson's disease tends to become progressively worse and is eventually fatal. With early detection and treatment, most of those affected can live relatively normal lives. Liver and neurologic damage that occurs prior to treatment may improve, but it is often permanent. Fulminant Wilson's disease (FWD) is rare and fatal condition in children unless liver transplantation is performed.

CONCLUSION: High degree of suspicion is needed in children presenting with liver dysfunction and/or extrapyramidal neurological features. If diagnosed early and properly managed, WD is one of the more easily treated inborn errors of metabolism.

KEYWORDS: Copper metabolism, Kayser-Fleischer ring, Wilson’s disease, Liver failure

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