

A Case Study On Wilson's Disease (Westphal- Strümpell Pseudosclerosis)

Dr .Vishal Sadatia*, Dr.Ritesh Vekariya*, Dr.Manish Bavaliya*, Dr.Mehul Patel**

Post Graduate*, Assistant Professor**, Medicine Department, C.U.Shah Medical College, Surendranagar

Abstracts: Wilson disease of Progressive Lenticular Degeneration is a familial nervous disease associated with Cirrhosis of the Liver. It has been observed that there is an increased copper content of the liver and brain. Increased excretion of copper in patients with Wilson disease & increased even more after administration of the chelating agent British anti-Lewisite (BAL). Ceruloplasmin, serum protein that binds copper, is reduced. The deposition of copper in tissues is the cause of virtually all the manifestations of the disease in Liver Blood Kidney & Brain We have observed three different cases of different clinical presentations.[Sadatia Vet al NJIRM 2012; 3(2) : 177-179]

Key words: Cirrhosis, Kayser-Fleischer rings & cerebral damage

Author for correspondence: Dr.Vishal Sadatia, Department of Medicine, C.U.SHAH Medical college, Surendranagar 363001 E- mail: drvvsadatia@gmail.com

Introduction: Wilson disease is transmitted as an autosomal recessive trait. The gene, called ATP7B (homologous with the ATP7A gene, Menkes disease) codes for a membrane bound, copper-binding ATPase. Inadequate functioning of the ATPase enzyme reduces excretion of copper in the bile^{1,2}.

In Wilson's disease there are two fundamental disturbances of copper metabolism:

(1) A reduced rate of incorporation of copper into ceruloplasmin³.

(2) A reduction in biliary excretion of copper.

Case Report 1: A 30 years old male patient presented with complaints short time episodes of difficulty in standing & walking without support and simultaneous loss of postural tone since last 6 months. These complaints were transient and it will be resolved within 30 minutes & precipitated after sometime. He is also having difficulty in speech during these episodes. In between these episodes he becomes alright without any residual defect. Patient had a fever with vomiting before 1 year and he had taken treatment for it then after he developed these episodes.

Examination: Patient was conscious & oriented, poorly nourished & vitally stable. No icterus or pallor. No clubbing or cyanosis. There was no any abnormality in cardiovascular, respiratory or gastrointestinal system. In central nervous system there is presence of involuntary

movements like chorea & gait abnormality in the form of ataxic gait.

Investigations: Routine blood investigations & other blood indices were normal.

- S.bilirubin 1.23(total), 0.33(direct), 0.90(indirect)
- SGPT 51 IU/lit
- Alkaline phosphatase 112.5 IU/lit
- Prothrombin time with INR 16.5/1.20
- Serum HIV NEGATIVE
- Serum protein 5.7(total), 3.3(albumin), A:G 1:1.3
- ESR - 6
- CSF examination was NORMAL.
- Serum ceruloplasmin 6.1 mg/dl
- Serum copper 54.33 mg/dl
- 24 hour urinary copper 150mg/day
- FUNDOSCOPY- Normal
- SLIT LAMP EXAMINATION- KAYSER FLEISHER RING is present.
- LIVER BIOPSY- INCREASED COPPER CONTENT in liver cells.
- MRI BRAIN – NORMAL

Treatment:- After confirming diagnosis patient was being treated with copper chelating agent D-PENICILLAMINE 500mg TID and PYRIDOXINE 40mg OD with ZINC supplementation. Follow up is being taken regularly for next 1 year. Patient is gradually improved and still continuing treatment.

Case Report 2:

A 34 year old male presented with the complaints of distension of abdomen, swelling over feet, difficulty in breathing since 15 days. Patient was having left sided hemiplegia secondary to right thalamic infarct. No significant past history.

Examination: On general examination there was raised JVP, oedema feet, and anaemia present. In respiratory system examination there was tachypnea & bilateral pleural effusion. On abdominal examination there were signs of ascitis. On central nervous system examination there were signs of UMN lesion in left upper & lower extremity.

Investigations: Increased serum copper level (60 mg/dl) with decreased ceruloplasmin (8.3mg/dl) level. Patient was having normal SGPT & bilirubin. MRI BRAIN :- s/o right thalamic infarct

Treatment: Patient had been started D-PENICILLAMINE. But before we discharged him, he died because of acute liver failure.

Case Report 3 :

A 19 year old male patient was admitted with the complaints of abdominal distension, loose motion, vomiting & low grade fever since one month. All of the complaints were together associated with mild jaundice. No significant past history.

Examination: Patient was having icterus & pallor. In systemic examination patient was having Ascitis, hepatomegaly, splenomegaly.

Investigations: Routine investigations were normal except anaemia (microcytic hypochromic). LFTs were deranged in the form of elevated bilirubin, SGPT (150 IU/L), decreased Serum protein. Serum copper was elevated (76 mg/dl), Serum ceruloplasmin was decreased (10 mg/dl). FUNDOSCOPY: - KF ring was seen on slit lamp examination. CT SCAN :- Mild hydrocephalus

Treatment: Patient had been treated with ZINC, D-PENICILLAMINE and SUPPLEMENTARY drugs. Patient

was well upto 6 months then he died due to hepatic encephalopathy.

Discussion: A neurologic disorder had been described previously under the title of "tetanoid chorea" & "pseudosclerosis"⁴.

None of them recognized the association with cirrhosis. The clinical studies clearly established that the pseudosclerosis was the same disease as the one that had been described by Wilson⁵. Interestingly, none of these authors, including Wilson, noticed the golden-brown (Kayser-Fleischer) corneal ring, the one pathognomonic sign of the disease. The onset of neurologic symptoms is usually in the second decade. Half of patients are symptomatic by age 15 years, but exceptional cases, including two under our care, had their first clinical manifestations as late as their mid-thirties. In all instances the initial event is a deposition of copper in the liver, leading to an acute or chronic hepatopathy and eventually to multilobular cirrhosis and splenomegaly.

In childhood, attacks of jaundice, unexplained hepatosplenomegaly or hypersplenism with thrombocytopenia and bleeding. Rarely there is clear evidence of cirrhosis alone.

The first neurologic manifestation is most often extra pyramidal^{5,7}. Typical presentations are tremor of a limb or of the head and generalized slowness of movement (i.e. parkinsonian syndrome) or slowness of movement of the tongue, lips, pharynx, larynx, and jaws, resulting in dysarthria, dysphagia, and hoarseness. There may be slowness of finger movement and occasionally choreic movements or dystonic postures of the limbs. As the disease progresses, dysphagia and drooling, rigidity and slowness of movements of the limbs, flexed limb postures, fixity of facial muscles, giving an appearance of grinning or a "vacuous smile". When the limbs are outstretched to a coarse, it produces "wing-beating" movement.

KF ring is virtually always present once the neurologic signs become manifest. A slit-lamp examination may be necessary for their early detection.

We got 3 cases of Wilson's disease amongst them one was presented with Wilson's chorea which is rare primary presentation, second one presented with stroke and third one with long standing jaundice with complications.

References:

1. Scheinberg IH, Sternlieb I. Wilson's disease. In: Smith LH Jr, ed. Major problems in internal medicine. Vol 23. Philadelphia: WB Saunders Company, 1984.
2. Brewer GJ. Neurologically presenting Wilson's disease: epidemiology, pathophysiology and treatment. *CNS Drugs*. 2005; 19(3):185-92.
3. Sundaresan S, Eapen CE, Shaji RV, Chandy M, Kurian G, Chandy G. Screening for mutations in ATP7B gene using conformation-sensitive gel electrophoresis in a family with Wilson's disease. *Med Sci Monit*. Mar 2007; 13(3):CS38-40.
4. Sir William Gowers, S. A. Kinnier Wilson, Progressive lenticular degeneration: a familial nervous disease associated with cirrhosis of the liver, *Brain* 2009: 132; 1997–2001 | 1997.
5. Allan H. Ropper, Raymond Delacy Adams, Mourice Victor. The inherited metabolic diseases of the nervous system in Adams and Victor's principles of neurology, 9th edition, McGraw-Hill, 2009, 37 940-941.
6. Owen CA Jr, Ludwig J. Inherited copper toxicosis in Bedlington terriers: Wilson's disease (hepatolenticular degeneration). *Am J Pathol*. Mar 1982; 106(3):432-4.
7. Sinha S, Taly AB, Ravishankar S, Prashanth LK, Vasudev MK. Central pontine signal changes in Wilson's disease: distinct MRI morphology and sequential changes with de-coppering therapy. *J Neuroimaging*. Oct 2007; 17(4):286-91.
8. Ala A, Walker AP, Ashkan K, Dooley JS, Schilsky ML. Wilson's disease. *Lancet*. Feb 3 2007; 369(9559):397-408.
9. Eisenbach C, Sieg O, Stremmel W, Encke J, Merle U. Diagnostic criteria for acute liver failure due to Wilson disease. *World J Gastroenterol*. Mar 21 2007; 13(11):1711-4.
10. Merle U, Schaefer M, Ferenci P, Stremmel W. Clinical presentation, diagnosis and long-term outcome of Wilson's disease: a cohort study. *Gut*. Jan 2007; 56(1):115-20.