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Case Report

Wilson's disease

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Abstract:

Wilson's disease is a disorder of copper metabolism leading to accumulation of the metal in different organ. Hepatic manifestation tend to occurs in the first decade and neurological symptoms in the third decade. Neurological manifestation are said to worsen with chelation therapy.

Key Words: Wilson's disease, Liver Cirrhosis, Kayser-Fleischer ring, Head tremor.

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Introduction: Wilson's disease is a rare inherited disorder of copper metabolism with deposition of copper in the liver, brain, and other tissues with an incidence of one in 30,000 **[1]**. Initially there is deposition of the metal in the liver followed by its release into the circulation and there after chronic accumulation in the brain and other extra hepatic tissues. Liver cirrhosis occurs early. In the nervous system basal ganglia and midbrain are affected most frequently. According to a German study the patients who presented in first decade show predominantly hepatic manifestations while the patients with neurological symptoms presented during the third decade **[2]** .Untreated Wilson's disease has a progressive course and may be fatal. The disease tends to be under diagnosed and timely diagnosis is rewarding but remains a challenge.

Case Report: A boy of 9 years admitted with complaints of yellowish colouration of whole body for 2 months , which was gradually increasing associated with malaise ,nausea .There is no history of fever ,skin rash, contact with jaundice patient, infusion ,blood transfusion, intravenous drug abuse ,travel history or sexual exposure, weight loss ,convulsion ,head truma. His bowel and bladder habit was normal.

On general examination pt was icteric, BP - 100/70 mmhg.P-80 b/min, Spo2-98% in RT. On Per abdominal examination Abdomen soft & non tender, just palpable hepatomegaly, bowel sound present. On CNS examination GCS 15/15, higher cerebral function is intact, speech normal, cranial nerve examination including

Slit Lamp Examination: K-F ring absent, motor & sensory function intact .Other systemic examination – No abnormality was found.

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Laboratory Investigations Finding were:

Parameters	Values
Haemoglobin	11.5 g/dl
Red Blood Cell	3.97x 10^3 /Ul
White Blood Cell	6.6x10^3 /Ul
Neutrophils	34.7%
Lymphocytes	48.6%
Monocytes	5.5%
Eosinophils	10%
Basophils	1.2%
Platelet	107x10^3 /Ul
MCV	83 fl
MCHC	34.9 g/dl
PBF	Dimorphic Anaemia
Prothombin Time	22 Sec
Bleeding Time	3 min 30 sec
Clotting Time	6 min 30 sec
Haemoglobin Electrophoresis	Within Normal Limit
24 hours Urinary Copper	*98.37 ug/day*.(N.R : 3.00 – 50.00)
S. Bilirubin	7.2 mg/dl
S. Alt	259 U/L
HbsAg	Negative
Anti HAV IgM	Negative
Anti HEV IgM	Negative
Anti HCV	Negative
USG Of Whole Abdomen	Thick walled GB ,Mild Hepatomegaly Spleenic Volume – 295 cm3, Portal Vein Diameter – 8 mm.
Endoscopy Of GIT	1.Early Oesophageal 2 .Portal Hypertensive Gastropathy & Gastritis 3. Duodenal Polyp

On the basis of clinical & laboratory findings he was diagnosed as a patient of **Wilson disease** and treatment started.

Discussion: Wilson's disease is rare autosomal recessive disorder resulting in copper overload .Children with WD are usually normal at birth and may remain healthy for variable period of time most of case present in the second & third decade of life **[7].**The patients with Wilson's disease usually present with the liver disease and develop neurological manifestations later on. In our case it was typical presentation. The presenting symptom was yellowish coloration of skin & mucous membrane & there is no neurological anifestation. Thats why we could treat our patient with penecillamine .There are other case reports as well where the initial presentation pertained to nervous system without involvement of liver and the presenting age was over 40 **[3,4].** Paradoxically the neurological manifestations are said to become worse with penicillamine **[5,6]** . This is attributed to mobilization of copper from the liver with elevations in unbound copper which produces worsening of neurological symptoms. In different studies the initial neurological deterioration was observed in 30- 75% of patients following penicillamine therapy this was refuted by certain other reports [**7].** The tremor improved in our case after treatment with penicillamine and anticholinergics. On the other hand the hepatic manifestations were actually precipitated by the drug. This fact has never been reported before. The patient was not willing to take the chelation therapy but was convinced of its importance. After studying the literature and earlier case reports treatment was started with good results.

Conclusion: As Wilson's disease is a rare disease the diagnosis is likely to be missed. There should be a high index of suspicion in all cases of liver cirrhosis with no clear cut etiology or an isolated neurological symptom such as tremor. It is also important to warn patients not to stop therapy. The patient had many unusual features which are being reported and shared for future reference.

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