Case Report

An uncommon presentation of Wilson's disease as Polyarthritis

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Abstract

Wilson's disease is an inherited disorder of copper homeostasis seen in 1 in 30,000 individuals. The clinical presentation of this disease is variable and includes chronic hepatitis, hepatic steatosis and cirrhosis in young adults. The neurological presentation include speech disorders, neuro-psychiatric manifestations and various movement disorders. The diagnosis is made by demonstration of reduced serum ceruloplasmin levels, increased urinary excretion of copper, the presence of Kayser-Fleischer rings in the cornea of eyes and elevated hepatic copper level. Here we report a case, who presented to us with polyarthritis an uncommon presentation.& subsequently confirmed to have wilson's disease.

KEY words: Polyarthritis, K-F ring, Hepatosplenomegaly, Ceruloplasmin

Introduction:

Wilson's disease is an autosomal recessive disorder caused by mutations in the ATP7B gene, a membrane-bound copper-transporting ATPase. The clinical manifestations are caused by copper toxicity and primarily involve the liver and brain. ATP7B protein deficiency impairs biliary copper excretion, resulting in positive copper balance, hepatic copper accumulation and copper toxicity from oxidant damage. Clinically it presents as hepatitis, cirrhosis, or as hepatic decompensation, neurologic & psychiatric manifestations. The classical finding of Wilson's disease in the eyes is golden-brown Keyser-Fleischer (K-F) ring, which is due to deposition of copper in descemet membrane of cornea.

The rare manifestations of the disease reported are repeated spontaneous abortions, Cholelithiasis, nephrolithiasis haematuria & Renal tubular acidosis(16%). Osteoarthritis of knees, hips and wrists is seen in patients over 20 yrs. age in 20-50% patients. The spine and appendicular joints are affected. Juvenile polyarthritis, recurrent dislocations and fractures are described. Chondro-calcinosis and metabolic bone disease with osteomalacia and osteopenia are seen. Sunflower cataract can be seen in eyes. Generalised hyperpigmentation of skin is described. We here report a case of wilson's disease who clinically presented predominently as Polyarthritis

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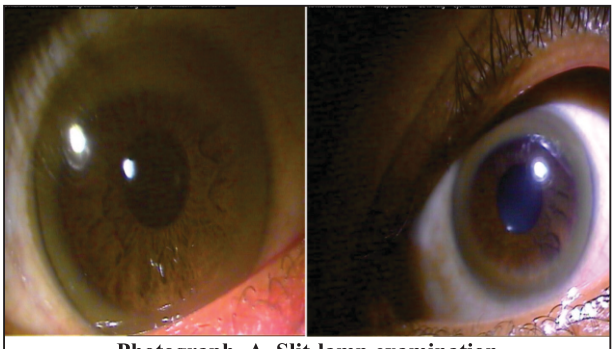
Case Report:

18 years adolescent boy was admitted with history of-Pain in both hip and knee joints and difficulty in walking & getting up from squatting position of 1 year duration. He also noticed tremors of both hands, deterioration of handwriting & inability to perform fine work with hands. The mother noticed emotional and behavioural problems in him since the same duration. There was no history of motor weakness of any limb or sensory impairement or loss of consciousness or seizures. Past history did not reveal any specific events. Family history was non contributory.

On general Examination his vitals were stable. There were no signs of hepatocellular failure. On locomotor system examination both Hip & knee joints were swollen with painful & restricted movements. Considering his hand tremors & emotional behavioural problems possibility of Wilson's disease was kept. Subsequently diagnosis confirmed on the basis of presence of circumferential



Photograph- A. Slit-lamp examination showing circumferential K-F ring

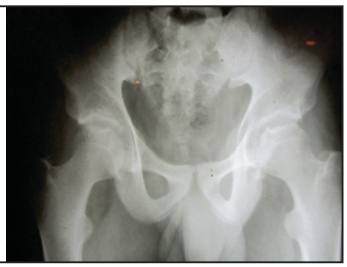
golden-brown K-F ring on slit lamp examination of eyes. (**Photograph –A).**

His abdominal examination revealed Hepato-splenomegaly. Cardiovascular&respiratory system was normal. Except dysarthria& tremors, no other neurological deficit was present.

On investigation his routine hemogram was normal.& had normal Renal and liver functions His RA factor was negative. Sickling test was Negative and Hb electrophoresis revealed normal pattern. Serum Calcium was 10mg/dl and phosphorus was 3.6mg/dl. Serum Ceruloplasmin levels was <0.004g/L (N. 0.2to0.6g/l)& Serum Copper was 32µg/dl (N. 56-110µg/dl). Free Copper value was 31.988 (Total S. copper- 3x S. ceruloplasmin) normal value=5-15µg/dl & 24 hours urinary copper – 477.63µg/day (N. 30-80µg/d, >100µg/dl virtually diagnostic). X ray Chest and ECG were normal.

X-ray both hips**:** showed flattening of superior articular surfaces of both femoral heads, sclerosis of subchondral

Fig 1.
X-Ray Pelvis with both Hips Shows flattening of both heads of femur & sclerosis of subchondral bone



bone giving focal defects in articular margins and joint spaces widening. **(Fig .1)**

X-ray knee joints: shows irregular articular margins of

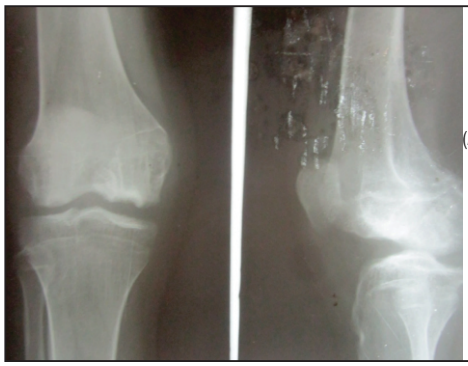


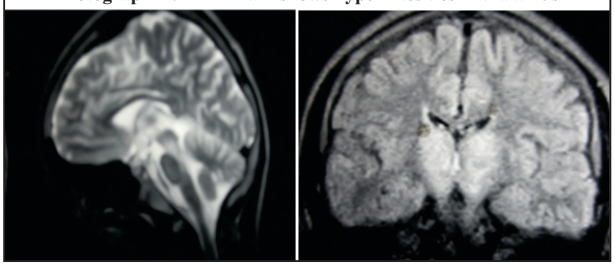
Fig 2.
X-Ray Knee (AP & Lateral views) shows irregular articular margins, focal defects & sclerosis of both bones

lower end of femora involving medial condyle, focal defects and sclerosis **(Fig .2)**

MRI Brain showed hyperintensities in thalamus on T-2 weighted images, no changes seen in basal ganglia**.(Photograph-B)**USG abdomen showed hepato-splenomegaly with normal echotexture of liver & no ascites. Upper GI endoscopy was normal.

Thus the diagnosis of Wilson's disease was confirmed patient was put on tablets of Zinc acetate (Galzin) in doses of 50 mg three times a day& tablet of trihexyphenidyl 2mg

Photograph-B. MRI Brain shows hyperintesities in thalamus



three times a day for tremors. The use of newer drug trientine has a high risk of worsening the neurologic condition hence in our patient it was not advised. He is under our follow-up for last 3 months and showing some improvement in tremors. At present he is receiving only symptomatic treatment and physiotherapy for polyarthritis.

Discussion:

The common features of Wilson's disease are hepatic and neuropsychiatric. The rare manifestations are important because they may not give the clue and the diagnosis may be totally missed. High index of suspicion in patients of hepatosplenomegaly and neuro-psychiatric problems with eye examination for K-F ring can be very helpful. All clinical manifestations of Wilson's disease can be prevented if disease is diagnosed early before functional impairment occurs. It is reversible even after severe impairment occurs but it is not treatable if irreversible damage is inflicted on liver and brain. Polyarthritis is mentioned as one of the uncommon presentations of Wilson's disease, seen in about 20-50% cases [1.] which was present in the present case as predominant clinical manifestation A G Bearn, MD. In 1957 reported abnormal X-ray findings as severe osteoarthritis and bone fragmentation in 13 out of 19 patients of Wilson's disease [2.] Similarly osteoarticular changes and synovial biopsy findings were described by P.Kaklamanis & M.Spengos (1973)[3.,4] Major abnormalities were seen in only 5 patients over age of 21 yrs. Multiple calcified loose bodies at wrists, premature degenerative arthritis of knees with chondromalacia of patella and chondrocalcinosis of knees was seen in the study. Rheumatological features described as osteopenia on X-rays and arthritis are seen in 20-50% patients[5]. They are thought be due to accumulation of copper similar to arthropathy in haemochromatosis. The synovial copper deposition as explanation of arthropathy of Wilson's disease is reported by Krammer V, Weinberger A and Yarom R.(1993)[6]. There is thickening of synovial membrane and the lymphoid and plasma cell infiltration. Energy Dispensive X-ray (EDX) microanalysis showed copper pigmentation in high concentration in synovial membrane[7] Similar type of arthropathic presentation was reported by Misra AK, Biswas A, Ganguly G (2004)[8.]