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Case Report

Avascular necrosis of femoral head in Wilson's disease

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Abstract

Introduction: Wilson disease is an autosomal recessive disorder which is due to mutation in gene encoding for ATPase. Due to excessive copper accumulation in various organs, it can present with spectrum of hepatic, neurological, osteoarticular changes most common being arthiritis. Diagnosis is confirmed on clinical features and genetic testing.
Case presentation: We present a case of 25year old male, diagnosed with Wilson's disease on genetic testing with complaints of both hip pain and restricted movements. Further radiological evaluation was suggestive of avascular necrosis of both hips (grade 3 on left and grade 2 on right – Ficat-Arlet staging). He underwent core decompression of right hip which relieved his symptoms of pain on right side and was counselled for bilateral total hip arthroplasty at a later date.
Conclusion: With varied clinical manifestations of Wilson's disease being encountered, avascular necrosis of femoral head should be kept as a differential diagnosis in cases of hip pain.
Keywords: Wilson's disease; Avascular Necrosis; Arthiritis.

1.Introduction

Wilson's disease (WND) is a rare autosomal recessive disorder that is caused by abnormal copper metabolism; its prevalence is approximately 30 cases per million people [1-4]. The gene responsible for WND was first identified in 1993 and encodes a copper-transporting P-type ATPase (*ATP7B*) [10]. It is located on chromosome 13 (13q14.3-q21.1). The excessive copper accumulation in various organs primarily the liver, brain, kidney, and cornea, results in a spectrum of hepatic and neurologic abnormalities [5-7]. Clinical presentation is highly heterogeneous [8,9]; patients can present with hepatic symptoms, neurologic symptoms, or both. The age of onset ranges from 2 to 70 years. Most of patients have degenerative changes of major joints and spine which leads to disability [11]. Diagnosis of WND is based on clinical symptoms (hepatic symptoms, neurologic symptoms, and cornea Kayser-Fleischer ring) and biochemical tests (elevated 24-h urinary copper, low plasma ceruloplasmin, and elevated liver copper concentrations) [1,2]. However, biochemical markers can be misleading [1,2], rendering WND diagnosis difficult in the absence of typical symptoms. For that reason, genetic testing has become the method of choice to establish a precise diagnosis [2,3].

2. Case Presentation

A 25 years old male, presented with history of insidious onset slowly progressive nonradiating pain in both hip with difficulty in walking since 6 months. Over the past two years patient gave history of intermittent tremulousness of all four limbs both at rest and in posture. Patient also complained of behavioral disturbance, cognitive decline, convulsion and weakness of both lower limb.

Physical examination revealed mild pallor, lack of jaundice and skin pigmentation. Neurological survey disclosed that he was a right handed individual with normal higher mental function (MMSE score being 28). Slit lamp examination demonstrated the presence of clear Kayser Fleischer ring. Fundus oculi and cranial nerve examination were normal.

Motor system examination showed mild rigidity and bradykinesia involving all four limbs. He had grade 4 power in lower limb, brisk deep tendon reflexes, and bilateral flexor plantar response. There were coarse tremors in distal part of upper and lower limbs, present both at rest and in posture, with no terminal accentuation during action. There was no evidence of dystonia.

Examination of musculoskeletal system showed wasting of both lower limbs. No flexion deformity at hip joint. Tenderness was present over both hips. There was painful restriction of movements of both hip. Right lower limb showed differential rotation in flexion and extension suggestive of sectoral involvement of right femoral head.

Investigations revealed Hb-11.0 gm/dl, with normal total and differential WBC counts, ESR 22 mm in the 1st hour, reticulocyte count 2%, and platelet count 1,50,000/cu mm. Routine urinalysis was normal. Blood biochemistry including blood sugar, urea and creatinine were normal. The serum calcium was 9.1 mg/dl and serum phosphate was 4.1 mg/dl. Liver function test revealed serum albumin 3.2 gm/dl, globulin 4.9 gm/dl, total serum bilirubin 0.5 mg/dl, SGOT 46 IU/ml, SGPT 44 IU/ml, and serum alkaline phosphatase 137 IU/l. Collagen vascular profile including C-reactive protein, Rheumatoid factor, and anti-nuclear factor was negative. The Coomb's test was negative. 24 hours urinary copper excretion was 110 micrograms (normal<40gm/24 hrs.).

USG of abdomen revealed heterogeneous echotexture of liver parenchyma and splenomegaly. CT scan of brain showed prominent sulci suggesting generalized cortical atrophy and hypodensity in the right basal ganglia region.

X-ray pelvis with both hips shows cortical collapse of left femoral head with sclerosis and subchondral cysts in right femoral head. MRI shows avascular necrosis grade III (Ficat and Arlet stage) of Left femoral head with grade II changes in right femoral head

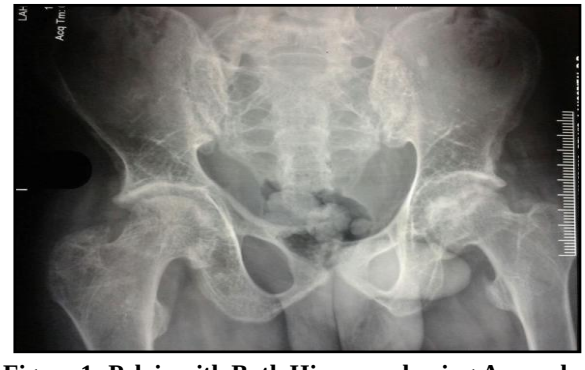

Figure 1: Pelvis with Both Hip xray shoving Avascular Necrosis of Femoral Head

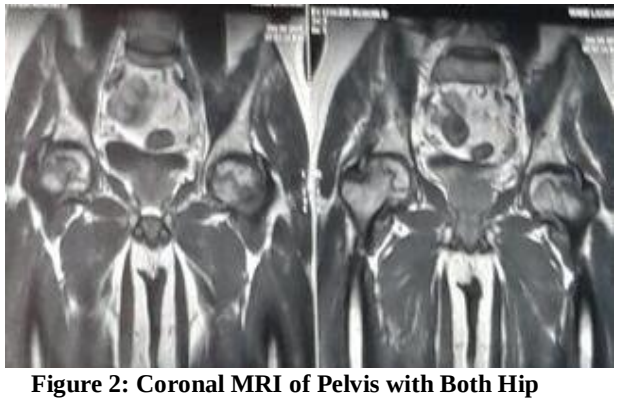

Figure 2: Coronal MRI of Pelvis with Both Hip

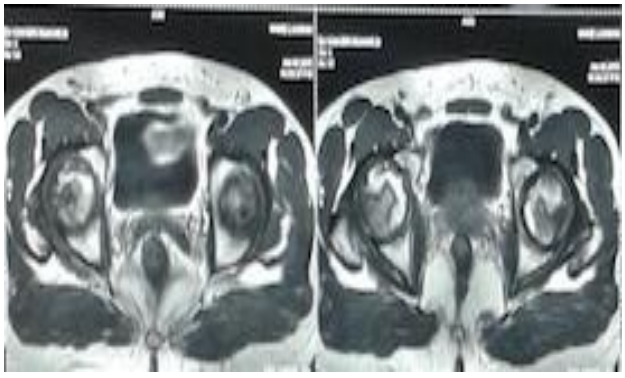

Figure 3: Axial MRI of Pelvis with Both Hip

3. Discussion

Wilson disease affecting multiple systems can present with wide variety of symptoms. Excess copper deposition in various organs is leading cause for various manifestations. The mechanism of most osteoarticular changes in Wilson's disease is unclear: copper overloading, renal tubular dysfunction, chronic spasticity and tremor, and liver failure have all been held responsible [12,13]. The radiographic evidence of osteoporosis is present in up to 88% of persons with Wilson's disease resulting in spontaneous fractures [11]. Joint involvement, especially at the knees, is also common and joint pain may be the presenting symptom of Wilson's disease [14]. Radiological evidence of vertebral column abnormalities is evident in 20 to 33% of individuals with Wilson's disease [15]. The most common radiological abnormality is a generalized increase of radiolucency, interpreted as skeletal demineralization followed by premature osteoarthrosis. Changes in the spine were common and included osteochondritis, reduction of intervertebral joint spaces, osteoarthrosis, and a tendency towards squaring of vertebral bodies. Other bony changes included fluffy irregularity of femoral trochanters, osteochondritis dissecans of the knees, osteophytic protrusions at bone ends, and bunch of tongue-like osteophytes at joint margins. Joint hypermobility was also observed and episodes of acute polyarthritis with serological changes were also seen [11].

Our patient presented with avascular necrosis of both hips which is unusual presentation of Wilson's disease. He underwent right hip core decompression which relieved his symptoms. Patient was counseled regarding future requirement of bilateral Total hip Arthroplasty.

4. Conclusion

Wilson disease being a multi system disorder has varied manifestations especially arthrosis of large joints but can also present as avascular necrosis of femoral head which might be the cause of pain in that region.

Clinical Message

With varied clinical manifestations of Wilson's disease being encountered, avascular necrosis of femoral head should be kept as a differential diagnosis in cases of hip pain. As arthropathy being the most likely presentation of hip pain in this disease, MRI would be an investigation that would clinch the diagnosis.

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