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Images in Rheumatology

# The view of joints in the Wilson's disease

Bengisu Aslan[1], Hatice Erdoğan[2], Veli Yazısız[1]

A 57-year-old female presented with pain in the metacarpophalangeal (MCP) joints of the wrist and knee. She was diagnosed with Wilson's disease 25 years ago. She had undergone liver transplantation, with a liver from a living donor two years ago because of cirrhosis associated with Wilson's disease. In addition, she had undergone hip replacement surgery for femoral neck fracture one and a half years ago. The patient had had muscle-joint pains for a long time. Rheumatoid factor and anticyclic citrullinated peptide tests were negative. Erythrocyte sedimentation rate and C-reactive protein levels were normal. Plain radiographs of the joints showed erosions and osteoarthritic changes. Although the view of distal and proximal interphalangeal joints was normal, irregularity of articular surfaces and joint space narrowing was observed in the MCP joints of both hands, especially in the right 3 and 4 joints. Chondrocalcinosis, bone fragmentations, changes of articular surfaces, and osteophyte protrusions were seen in the wrist joints (Figure 1). Classical osteoarthritic changes such as joint space narrowing in the medial compartments, subchondral sclerosis, and marginal osteophytes were visible in the radiograph of the knees (Figure 2a). Spine radiographs had shown spinal osteoarthrosis with Schmorl's nodes, narrowing of the intervertebral disc spaces, and osteophytes at the edges of the vertebral body (Figure 2b). Degenerative changes and irregularity of trochanters were seen on the right hip joint and the left hip joint was replaced with a prosthesis because of traumatic fracture (Figure 2c). Although some radiographic features were resembling osteoarthritis, the patient was diagnosed with arthropathy related to Wilson's disease.

Wilson's disease is a recessively inherited autosomal disease caused by mutations in the ATP7B gene, which leads to impaired copper excretion into the bile and causes a combination of hepatic, neurologic, and psychiatric symptoms. It has a disabling and fatal course if the diagnosis is overlooked and treatment is not initiated. The estimate prevalence ratio for this disease is 1:30,000 - 1:50,000 in the USA, Europe, and Asia (1). Joint involvements and radiological abnormalities have been described in clinical case studies. Early osteoarthritic changes were reported at the joints, especially at the knee, hip, and wrist joints (2). In addition, Wilson's disease is associated with premature osteoarthritis at the joints that are unaffected in patients with classic osteoarthritis, such as the MCP joints (3). Other abnormalities include joint space narrowing in the medial compartments, subchondral sclerosis, osteophytic protrusions at bone ends, and bunches of tongue-like osteophytes at the joint margin of the knees; irregularity of femoral trochanters; and osteochondritis, reduction of intervertebral joint spaces, osteoar-

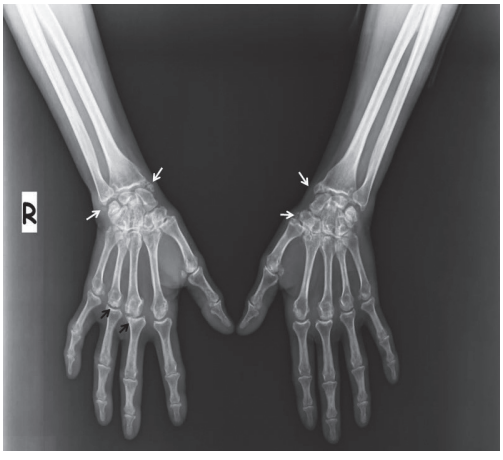


**Figure 1.** Plain radiographs of wrists and hand joints.


**Figure 2. a-c.** Plain radiographs of knee (a), lumbar spine (b), and pelvis (c).

**ORCID iDs of the authors:**
B.A. 0000-0001-6892-1090;
H.E. 0000-0002-1231-3640;
V.Y. 0000-0002-3176-4850.

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[1] Division of Rheumatology, Department of Internal Medicine, Akdeniz University School of Medicine, Antalya, Turkey
[2] Department of Internal Medicine, Akdeniz University School of Medicine, Antalya, Turkey

*Address for Correspondence:*
Veli Yazısız; Division of Rheumatology, Department of Internal Medicine, Akdeniz University School of Medicine, Antalya, Turkey

E-mail: drvyazisiz@yahoo.com.tr

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throsis, and a tendency of squaring of vertebral bodies at the spine. Although the exact pathogenic mechanisms responsible for joint changes are unknown, high levels of copper were found in synovial biopsies, and copper and sulfur deposits were found in the cartilage biopsies (4, 5).

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