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Ochronotic Spondyloarthropathy

Okronotik Spondiloartropati

Aslı GENÇAY CAN, Canan ÇELİK

Bursa Şevket Yılmaz Training and Research Hospital, Department of Physical Medicine and Rehabilitation, Bursa, Turkey

Ochronosis is a rare autosomal recessive metabolic disorder that is caused by a lack of the homogentisic acid (HGA) oxidase enzyme (1). The deficiency results in the accumulation of HGA within connective tissues, such as hyaline cartilages, intervertebral discs, tendons, skin, and sclera (2). The other common clinical manifestations are degeneration of intervertebral discs and large joints, dark urine on addition of alkali, renal stones, and cardiac involvement (1).

There is no effective specific treatment. The main objective is to prevent ochronotic arthropathy that may require joint replacements (3,4). The patients must be followed up for possible involvement of the renal and cardiovascular systems (4). At present, the only treatment available is symptomatic. Rest, analgesics, exercise, and physiotherapy can help relieve symptoms of arthropathy (1).

A 50-year-old man was referred to our clinic with pain in the left knee for 2 years and intermittent low back pain for 10 years. The pain was exacerbated by movement and relieved by resting. He denied morning stiffness. He noticed auricular pigmentation of almost 20 years in duration and urinary darkening. His brother also suffered from auricular pigmentation and chronic low back pain. On physical examination, we found blue-gray pigmentation on the ears. The spine flexion was slightly restricted and painful. The knees had a normal range of motion with crepitus. The neurologic examination was normal. Clinical examination of the cardiovascular and respiratory systems revealed no abnormality. Complete blood cell count, C-reactive protein (CRP), serum glucose, calcium, phosphorus, urea, creatinine, ferritin, liver and renal function tests, parathyroid hormone, thyroid-stimulating hormone, and free T4 levels were all within normal limits. Erythrocyte sedimentation rate (ESR) was slightly increased (36 mm/h). Urine analysis was found to be normal. Radiograph of the lumbar spine showed multilevel intervertebral disc calcifications, narrowed intervertebral spaces, vertebral body squaring, and marginal osteophytes (Figure 1). Radiograph of the left knee showed medial tibiofemoral joint space narrowing and osteophyte of the medial tibial plateau. An echocardiogram revealed no heart abnormalities. An abdominal ultrasound revealed no kidney stones or visceral abnormalities. Based on these findings, we diagnosed the patient as having ochronosis. The diagnosis was confirmed by high 24-hour urinary levels of HGA (670.3 mmol/mmol of creatinine; N<2 mmol/mmol of creatinine). We educated the patient about proper body alignment and treated him with analgesics, exercises, and electrotherapy.

Ochronotic arthropathy is the most common musculoskeletal involvement of ochronosis. It is age-associated progressive degenerative arthropathy. It develops typically after 40 years old and results from HGA accumulation in the cartilage. Subsequently, the cartilage turns dark and loses its elasticity. The intervertebral discs are the most frequently involved, where it results in calcification and intervertebral disc space narrowing. Severe spinal involvement can resemble ankylosing spondylitis due to formation of bony bridges between vertebral bodies, rigid spine, and morning stiffness (4,5). The physician should also consider diffuse idiopathic skeletal hyperostosis (DISH) as a differential diagnosis. In our case, a radiograph of the lumbar spine typically demonstrated multilevel disc calcifications. There was no sacroiliac involvement in the anteroposterior pelvis radiography. The patient had an almost normal range of motion in the spine. As a result of these findings, we did not consider ankylosing spondylitis or DISH. Ochronotic spinal arthropathy is clinically and radiologically similar to idiopathic osteoarthritis but differs in

Address for Correspondence / Yazışma Adresi: Aslı Gencay Can, Bursa Şevket Yılmaz Training and Research Hospital, Department of Physical Medicine and Rehabilitation, Bursa, Turkey Phone: +90 533 232 95 72 E-mail: asligencay@yahoo.com

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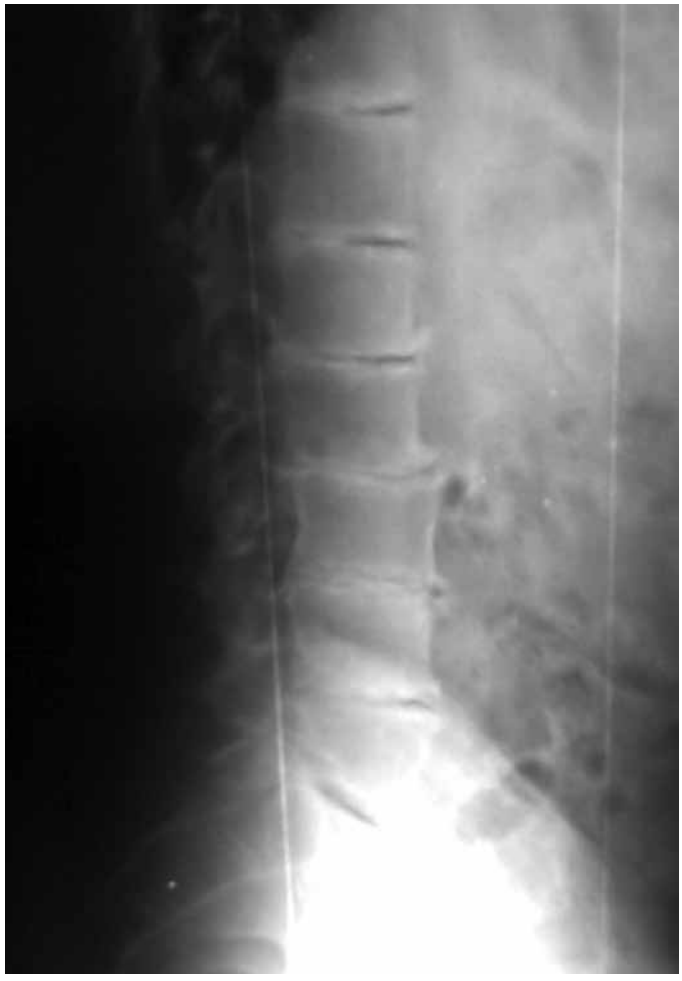

Figure 1. Characteristic appearance of ochronosis with narrowing of the disc spaces, disc calcifications, and sclerosis of the vertebral margins

predominant thoracolumbar involvement at a relatively young age. Extensive calcification of the intervertebral discs can also be seen in hemochromatosis, hyperparathyroidism, and calcium pyrophosphate deposition disease (CPPD). Differential diagnosis of these diseases is based on history, clinical examination, laboratory findings, and radiologic appearance.

Ochronotic arthropathy can affect large peripheral joints. The knee joint is one of the most commonly involved joints. Meniscal calcifications, loose bodies, marginal osteophytes, subchondral sclerosis, subchondral cysts, and asymmetric joint space narrowing may develop. The hips, shoulders, and other joints may also be involved (3,4). It can resemble osteoarthritis and inflammatory arthritis, such as rheumatoid arthritis and CPPD (5). Ochronotic arthropathy is more severe, with less osteophytosis at a relatively young age than is seen in osteoarthritis (2). In contrast to rheumatoid arthritis, small joints of the hand and foot are generally spared, and the synovial fluid is non-inflammatory (3). It differs from CPPD in that it causes more marked calcification in intervertebral discs rather than in peripheral joints. In our patient, we detected degenerative changes in his left knee radiography. There was no effusion or heat. ESR and CRP levels were normal. According to these findings, we excluded inflammatory arthritis. However, we could not exclude idiopathic osteoarthritis with these findings. Ochronosis should be considered in the differential diagnosis of severe spinal calcification and degeneration a relatively young age.

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