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Letter to the Editor

Osteopetrozis

Osteopetrosis

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Osteopetrosis is a descriptive term that refers to a group of rare, inherited bone disorders first described in 1904 by Albers Schönberg.[1] Three forms of this disease have been described: two lethal autosomal recessive forms and a benign autosomal dominant type (osteopetrosis tarda).[2] Herein, we present a case with the clinical and radiological signs of the autosomal dominant type of osteopetrosis.

A 28-year-old male patient was admitted to our clinic with complaints of neck and hip pain. He had approximately an eight-month history of problems with his neck, and the pain is his hip began before then. In addition, his past medical history was unremarkable. The patient's systemic examinations were normal, and a musculoskeletal system examination showed that his neck extension and lateral rotation was limited and painful and that his flexion was minimally limited. Furthermore, the patient's internal and external hip rotation was bilaterally limited. The range of motion in his back was normal, but the lumbar lordosis measurements were increased. Moreover, there was bilateral pes cavus deformity. The range of motion in the other joints was normal, and the neurological examination revealed nothing out of the ordinary. Laboratory analyses revealed the following irregularities: aspartate aminotransferase (AST) 47 U/l, hemoglobin (Hb) 11.8 g/dl, and erythrocyte sedimentation rate (ESR) 28 mm/h. All other results were normal. At the radiological evaluation, the pelvis showed increased density, and more bilateral joint space narrowing on the left side of the hip was observed. In addition, there was increased uniform density and sandwich vertebra sign (sclerotic dense bands parallel to the vertebrae end plates) at the lumbar vertebrae. There was also bilateral femural, tibial, and fibular cortical thickening along with dense, diffuse, symmetrical sclerosis, Erlenmeyer flask deformity, and cranium-based sclerosis. After evaluating all of the clinical and radiological findings, the patient was diagnosed with osteopetrosis tarda.

Due to the patient's anemia, he was referred to the hematology department. The patient was then discharged with recommendations of fracture protection and an exercise program.

Osteopetrosis is a rare hereditary condition characterized by an increase in bone density. The overall incidence of this disease is difficult to estimate, but in some studies, the incidence of autosomal recessive osteopetrosis (ARO) is 1: 250,000 births and for autosomal dominant osteopetrosis (ADO), it is 1: 20,000 births.[2,3]

In some cases, bone formation is normal, but there is a reduction in bone resorption. This occurs as a result of osteoclast differentiation and function failure.

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Additionally, the normal structural pattern of the bone is grossly altered, the cortices are thickened, the trabecular bone increases in size, and the bone marrow spaces are narrowed, which can cause secondary anemia. This can lead to excessive calcified tissue, and hypercalcification increases bone fragility and causes fractures.[4]

In the first months of life, ARO, a life-threatening condition, may occur. Macrocephaly and frontal bossing can also be found in the first year of life, which causes the typical face appearance in patients with this disease.[5] During that time, nerve foramens can be narrowed by the increased bone mass, causing blindness, deafness, and facial palsy. However, the most harmful complication is bone marrow suppression.[2]

In late childhood and adulthood, ADO has been noted. This disease is classically characterized by radiological features like sandwich vertebrae sign. There are also complications associated with ADO, including fractures, scoliosis, hip osteoarthritis (OA), and osteomyelitis.

Both clinical and radiological evaluations are used to diagnose osteopetrosis. The most common radiological signs are disseminated sclerosis at the skull, vertebrae, pelvis, or appendicular bones, "bone-in-bone" sign at the vertebra and phalanx, bone modeling defects at the metaphyses of long bones, for example a funnel-like appearance (Erlenmeyer flask deformity), and sandwich vertebra sign. In the absence of these typical radiological features, increased isoenzyme creatine kinase (CK)-BB and tartarate-resistant acid phosphatase levels can indicate this disease.[2]

Unfortunately, there is no effective medical management for osteopetrosis, but the treatment usually involves offering support for the management of symptomatic complications.

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