







Erdheim-Chester disease in a psoriatic arthritis patient

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The Erdheim-Chester disease (ECD) is a multisystem myeloid clonal inflammatory disorder. It is characterized by abnormally high production and accumulation of foamy histiocytes and Touton giant cells in multiple organs and tissues.¹ All systems may be affected, resulting in an extremely variable clinic. The pathogenesis of ECD is characterized by cluster of differentiation (CD)68-positive and CD1a-negative histiocyte infiltration. Fifty-seven to 70% of patients have the BRAF V600E mutation, whereas only 20% of patients have the MAP2K1 mutation.²

A 40-year-old female presented to the emergency department with severe bone pain, numbness in the left leg, muscular weakness, and pain in the right gluteal region. Psoriatic arthritis (PsA) with skin and nail psoriasis, distal and proximal interphalangeal arthritis, dactylitis, and sacroiliitis was diagnosed 14 years ago. The patient was undergoing etanercept treatment for PsA. Multiple imaging procedures were conducted on the patient. Computed tomography revealed multiple abscesses in the bilateral lumbar and gluteal regions, pleural effusion and nodules in the thorax, increased sclerosis in the sacroiliac

joint, sacral wing, and both femoral head-neck junctions, and osteosclerosis areas in the right tibia metaphysis diaphysis. A muscle biopsy revealed the presence of infiltrating ECD. In the biopsy, there was an intense infiltration of histiocytes that stained positively for CD68 but negatively for S100 and CD1a (Figure 1). The patient did not possess a BRAF mutation. Etanercept was stopped, and six cycles of peginterferon alfa-2a were administered by the hematology clinic. The patient's C-reactive protein (CRP) levels returned to normal after receiving the medication, and clinical remission was achieved.

Erdheim-Chester disease is a disease that can affect all body systems. Typical bilateral osteosclerotic lesions of the long bones of the lower extremities, pleural effusion, vascular involvement, cardiovascular disease, and retroperitoneal fibrosis are the most frequent clinical manifestations. It is more prevalent in males than in females. Eighty percent or more of the patients have elevated CRP levels.³ In our patient, the presence of pleural effusion, involvement of the skeletal system, and elevated CRP levels were consistent with the literature. Interferon is the first-line treatment

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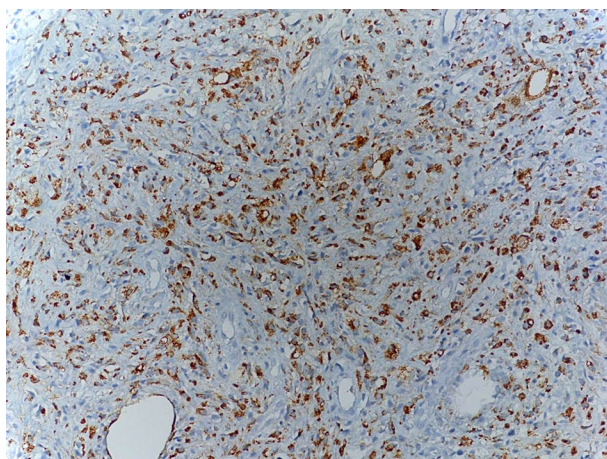


Figure 1. Histiocytes stained favorably for CD68 ($\times 200$).

for ECD as it has been found to improve overall survival. When interferon therapy has failed, anakinra and infliximab should be considered due to their excellent results.⁴

According to scientific literature, no association between rheumatological diseases and ECD has been reported. In addition, infiltrative ECD of the gluteal muscle observed in our patient is a rare entity. With this in mind, we wanted to share our experience. In patients with rheumatic disorders, it is difficult to differentiate between diseases affecting the musculoskeletal system. Clinicians frequently associate the symptoms with the underlying rheumatic disorder. However, diagnosing rare diseases requires a substantial amount of time. Patients may be exposed to unnecessary treatments, and their disease may progress as a result of a protracted process. Each time a new symptom manifests in one of our

patients, we must evaluate it separately from the underlying condition. Additional research will aid in the diagnosis of rare and difficult diseases.

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