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# Erdheim Chester Disease (ECD): A Case Report

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Erdheim Chester disease (ECD) is very rare non-Langerhans cell histiocytosis (LCH) which occurs in the skeletal system and multiple organs. As it is progressive, sometimes it causes fatal results. However, it is often misdiagnosed as LCH or multiple bone metastasis and, thus, is very difficult to diagnose. In Korea, only 10 cases were first reported in 1999. In particular, there have been a few orthopedic approaches or reports in English-speaking literatures, and no report has been issued in Korea. The authors performed bone biopsy in patients with knee and lower extremity pain who were referred for the integrated treatment. We attempts to report this diagnosis experience with literature review.

**Key word**: Langerhans cell histiocytosis

This rare disease is characterized by a symmetrical sclerosis at the diametaphyseal portions of the lower extremities with extraskeletal involvement. Many internal organs and tissue sites, including the kidney and retroperitoneum, lung, pericardium, skin, orbit, and brain may occur. The symptoms and clinical manifestations depend on the organ involved. The prognosis is poor with progressive disease with resultant organ dysfunction. We report a case of Erdheim Chester disease. The patient was informed that the data concerning this case would be submitted for publication.

## Case Report

A 69-year-old female patient was referred for both knees and lower extremities pains which began 6 years ago and were recently serious. She has taken medication for diabetes and hypertension for 20 years and was in the hospital for polyuria and pyuria in the internal medicine department of this hospital. Soft tissue invasion around both kidneys (Fig. 1) and soft-tissue lesions around right coronary artery were found on abdominal CT (computed tomography) and their assessment was in progress.

Biopsy was preformed for soft-tissue lesions around both kidneys under the ultrasonography guide and the pathologic features revealed collagenous fibroadipose tissue with lymphoplasma and histiocytes. The result immunohistochemical staining were a positive for CD68, and negative for S-100, CD1a (Fig. 2). PET CT (Positron Emission Tomography) showed high metabolic lesions of SUV 4.7 in heart, kidney, bilateral distal femoral, and proximal tibia (Fig. 3) and X-ray also showed osteosclerotic and osteolytic lesions (Fig. 4). Thus, this department and the departments of hematology-oncology and cardiology implemented the integrated treatment under the suspicion of hematologic malignancy or multiple bone metastasis. While the

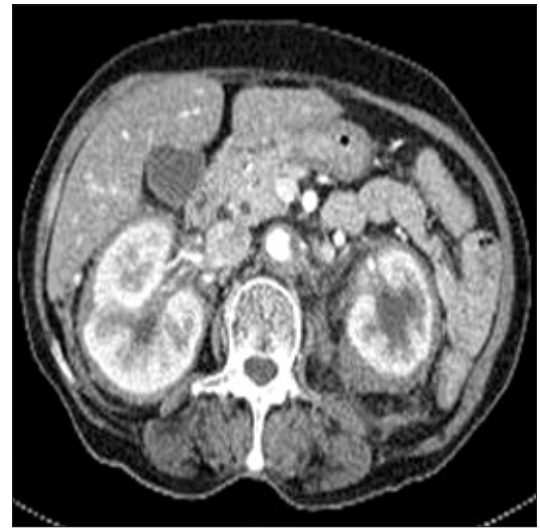

Figure 1. On computed tomography, there are soft tissue invasions around both kidney.

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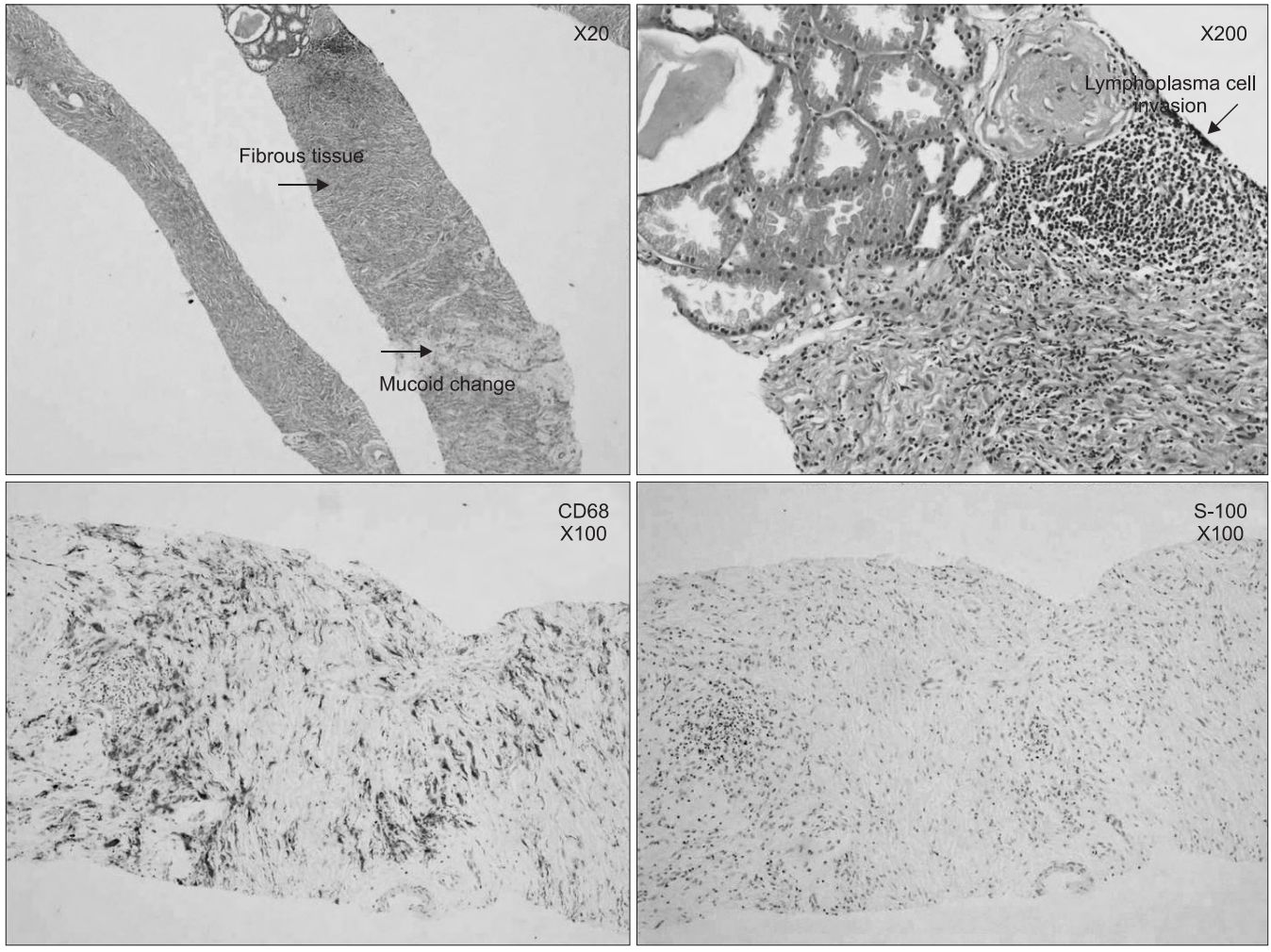


**Figure 2.** On histologic finding, pathologic features revealed collagenous fibroadipose tissue with lymphoplasma and histiocytes. The result immunohistochemical staining were a positive for CD68, and negative for S-100, CD1a.

department of hematology-oncology considered bone marrow examination and the department of cardiology considered biopsy, this department performed biopsy under the suspicion of multiple bone metastasis and lymphoma. The bone biosy revealed an intraosseous fibrosis (Fig. 5). We diagnosed the patient with ECD based on the above clinical and biopsy findings. She is under internal treatment.

## Discussion

Erdheim Chester disease (ECD) is very rare non-Langerhans cell histiocytosis which invade the skeletal system and multiple organs. It was first introduced by Chester[1] and is histologically characterized by the accumulation of lipid laden foamy macrophage.[2,3] Although the cause of disease has not been established yet, the high turnover of LDL seems to play an important role. In Korea, 10 cases[4] were first reported. There have been a few orthopedic approaches or reports in English-speaking literatures, and no report has been issued in Korea.

Since the clinical characteristics of ECD are much unknown and it is difficult to diagnose, clinical suspicion is important. Most cases are developed after forty years of age and are characterized by osteosclerotic lesions which are well marked by bilateral symmetrical boundary of metaphysis of long bone. It may occur in ribs, sacrum, head, neck, and spine. It is rare to occur in the spine in case of extremities[3] and its occurrence in head and neck is associated with diabetes insipidus. Patients clinically have arthralgia, keloid formation, and a systemic symptom which invades several organs such as heart, liver, lung, and kidney.[5,6] It is known to be accompanied by extraskeletal symptoms[2] in about its 50%. A clinical prognosis is determined by invaded organs and if it invades the blood vessel system, it shows a bad prognosis.[7,8]

ECD is progressive disease. Generally, patients are often seen in

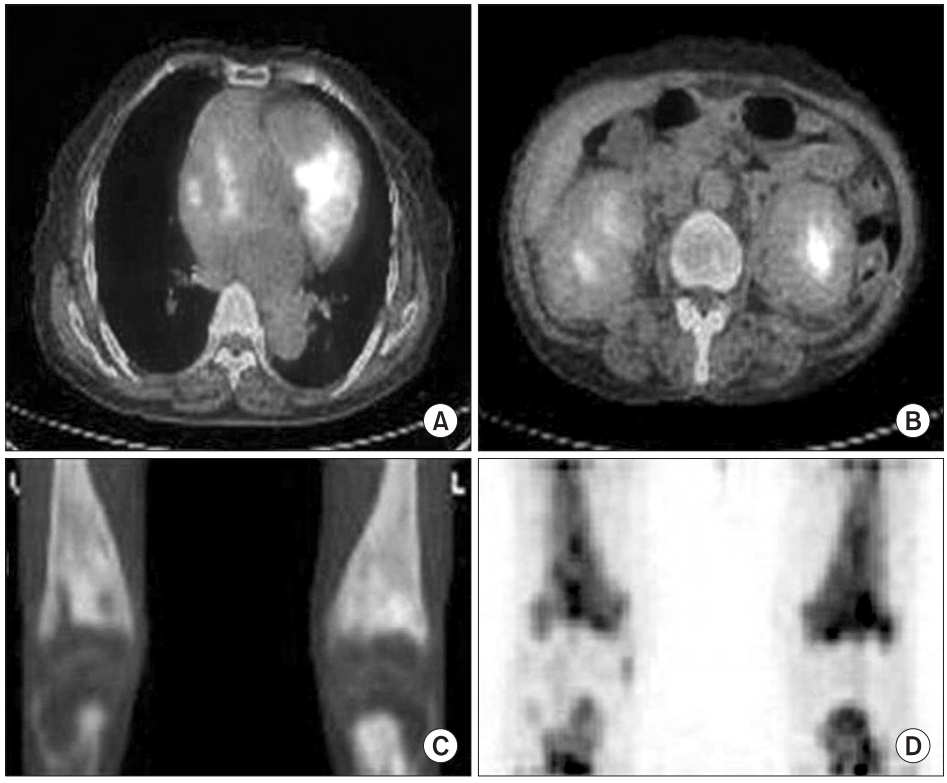


Figure 3. On PET CT, it showed high metabolic lesions of SUV 4.7 in heart (A), kidney (B), bilateral distal femoral, and proximal tibia (C, D).

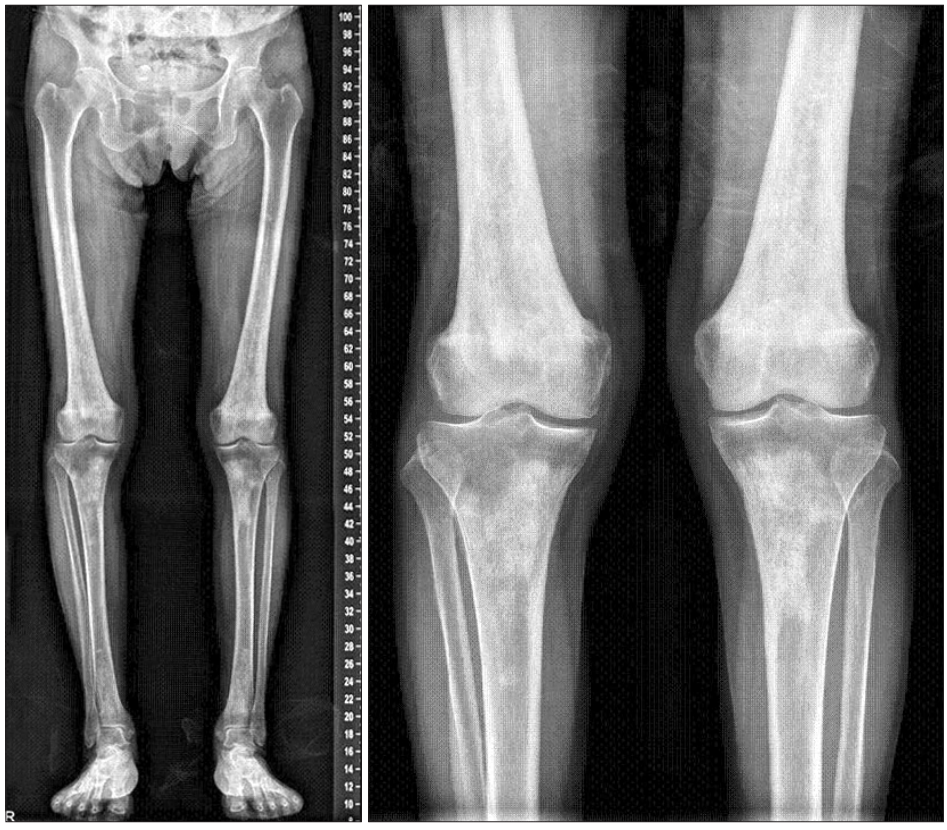

Figure 4. X-ray also showed osteosclerotic and osteolytic lesions on both distal femur and proximal tibia.

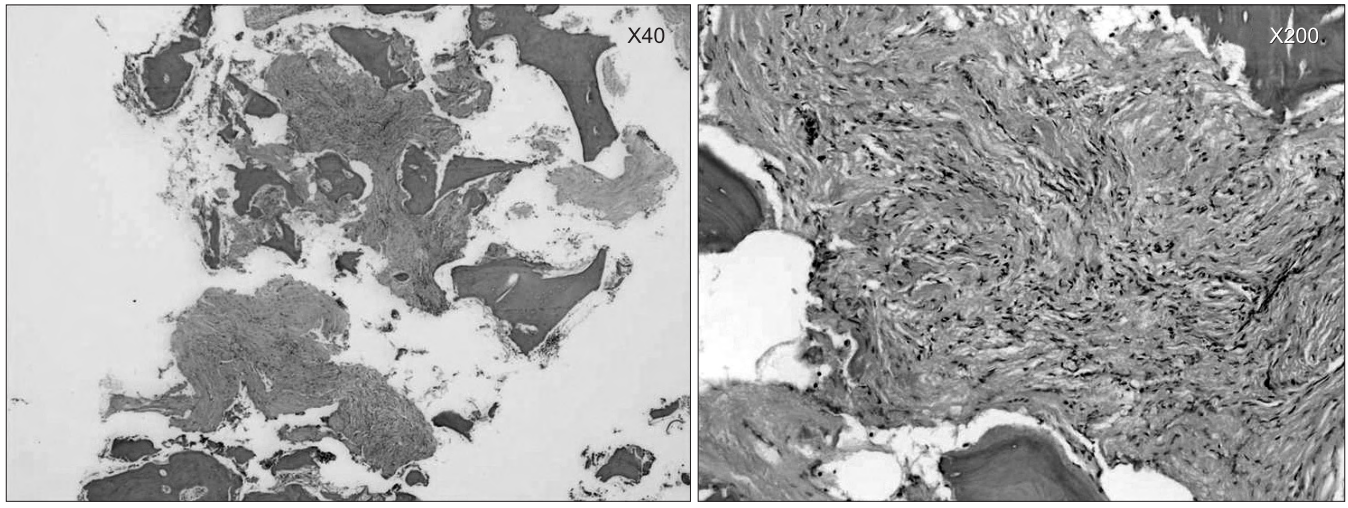


Figure 5. The bone biopsy revealed an intraosseous fibrosis. H & E (Hematoxylin & Eosin) staining.

the hospital with arthralgia or lower extremity pain caused by orthopedic osteoarthritis. Thus, if the disease is not recognized, it is impossible to prevent its progress and the condition will become worse.

Generally bony invasion is characterized by osteosclerotic lesions which are well marked by the bilateral symmetrical boundary of iliac metapre or are patch-shaped.[3] Our patient had mixed lesions of osteosclerotic and osteolytic lesions, resulting in authors' misdiagnosis with multiple bone metastasis.

The patient was positive for CD68 but negative for CDla and S-100 on the immunohistochemical stain. Based on this, LCH which shows similar clinical conditions could be excluded.

The patient could be diagnosed with ECD based on orthopedical bone biopsy and clinical characteristics. Then, she could be systemically treated and invasive diagnosis testing which is more dangerous could be prevented.

Although ECD is progressive and fetal disease and its prognosis is determined by invaded organs or its degree, the condition often becomes serious by patients often seen in the hospital with arthralgia who cannot recognize their disease. In other words, it is considered that clinical suspicion of ECD is needed based on the above radiological and clinical findings.

As ECD is very rare and fatal disease which is not easy to diagnose, correct diagnosis and treatment is essential. If middle-aged people seen with arthralgia and lower extremity pain show symmetric and bilateral bone lesions, the clinical suspicion of ECD is necessary. Thus, the authors reported the diagnosis and caution of ECD which has few orthopedic reports and is often misdiagnosed through our experience and literature review.

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# Erdheim Chester Disease (ECD): 증례 보고

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Erdheim chester disease (ECD)은 골격계 및 다발성 장기에 발생하는 매우 드문 비 랑거한스세포 조직구증(Langerhans cell histiocytosis, LCH)으로 구분되는 질환으로 진행성이며 때론 치명적인 결과를 야기한다. 하지만 이는 LCH 혹은 다발성 골 전이로 오인되기 쉬우며 진단 또한 극히 어렵다. 국내에서는 1999년에 10예가 처음으로 보고되었을 뿐이며 특히 정형외과적으로 접근 및 보고는 문헌상 영어권에서 극히 소수의 문헌만이 존재하며 국내에는 보고된 예가 없다. 저자들은 슬관절 및 하지통은 주소로 본과에 협진된 환자에 대해 골 조직검사 시행하였으며 이에 대한 진단적 경험을 문헌 고찰과 함께 보고하고자 한다.

**색인단어:** 랑거한스세포 조직구증

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