Erdheim-Chester Disease - Multisystemic Radiological Manifestations

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INTRODUCTION

Erdheim-Chester disease (ECD) is a rare, multisystem disorder with a constellation of clinical and radiological findings. We present a case of ECD who came with acute obstructive uropathy and chronic bone pain. Radiological work up in this patient led to a host of findings in various systems, the correlation of which led to narrowing of the differential diagnoses.

PRESENTATION OF CASE

A 44-year-old male presented with right loin pain and nausea for 1-day duration. A provisional diagnosis of right ureteric colic was made. Routine lab investigations revealed normal total counts and urine analysis. Additional laboratory investigations revealed a raised erythrocyte sedimentation rate (ESR) and C-reactive protein (CRP).

Radiological Investigations

1. Patient was referred for ultrasound of abdomen which revealed mild to moderate right hydronephrosis with a suspicious hypoechoic area (~ 0.7 x 1.2 cm) in the renal pelvis region raising possibility of a lesion.
2. Contrast computed tomography (CT) study of abdomen was performed which revealed enhancing soft tissue infiltration (+30 to +45 HU) around bilateral kidneys (giving 'hairy kidney appearance') (Figures 1, 2 & 3) and adrenal glands with infiltration to renal sinus and constriction of bilateral renal pelves resulting in moderate right and mild left hydronephrosis. Thin streak of excreted contrast was traversing the narrowed segment of renal pelves (Figures 4 & 5).
3. Soft tissue infiltration covering the visualized thoracic and abdominal aorta (giving 'coated aorta appearance') (Figures 6 & 7), extending below renal arteries to aortic bifurcation and proximal bilateral common iliac arteries.
4. The adrenal glands were not separately visualized from the above mentioned perinephric soft tissue.
5. Both ureters were normal in course and caliber. No evidence of ureteric calculus.
6. No significant retroperitoneal lymphadenopathy.
7. Few tiny centriacinar nodules with patchy ground glass opacities in postero basal segment of both lungs (left > right) (Figure 8).

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Past Medical History

Additional clinical history from the patient revealed that the patient had presented with chronic pain in bilateral thighs, knees and legs, 13 months back from the date of admission. Prior radiographs were retrieved which showed mild sclerosis of bilateral distal femora and proximal tibiae & fibulae with multiple fairly defined lucent lesions and narrow zone of transition. The lesions were also noted involving the tibial condyles with relative sparing of patellae and femoral condyles. The lumbar spine, pelvic bones and proximal femora showed no significant abnormality. The patient was lost for follow up after these X rays and had presented with right loin pain later.

DIAGNOSIS

Clinical & Radiological Diagnosis

With the constellation of above imaging findings, a radiological diagnosis of Erdheim–Chester disease was made.

Differential Diagnosis

Other less likely differentials included lymphoma (for perinephric and skeletal manifestations) / retroperitoneal fibrosis / extramedullary haematopoiesis.

DISCUSSION OF MANAGEMENT

The patient was referred to higher centre for further workup (including pathological confirmation) and management.

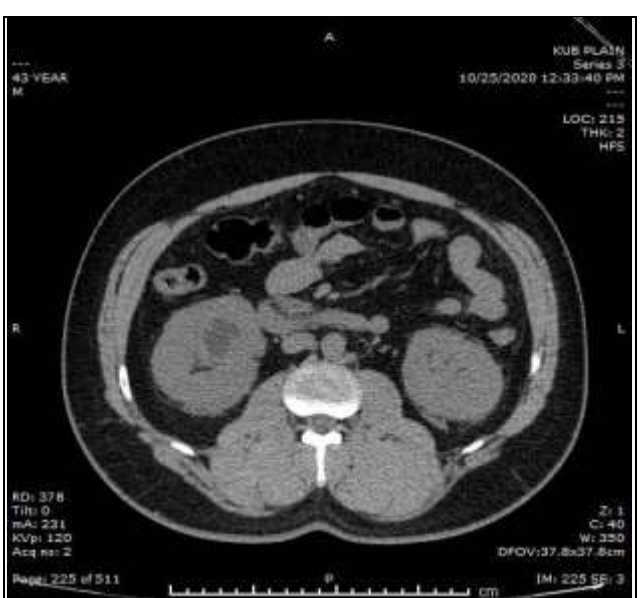

Figure 1. Axial Non-Contrast CT Abdomen at Renal Level – Bilateral Perinephric Stranding with Mild Prominence of Right Renal Pelvis

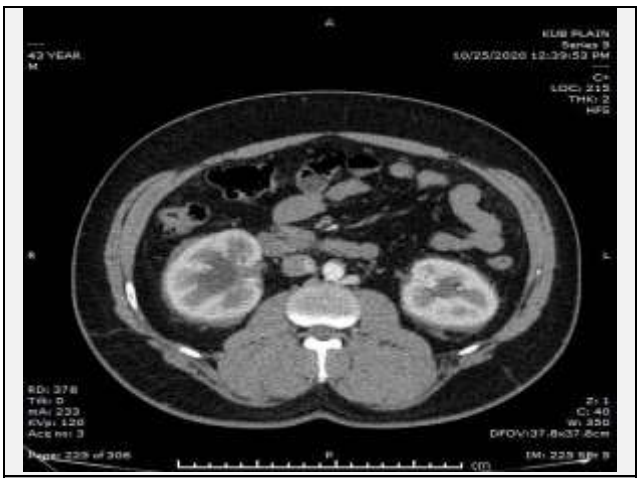

Figure 2. Axial Post-Contrast CT Abdomen (Corticomedullary Phase) at Renal Level – Enhancing Soft Tissue Infiltration around Bilateral Kidneys Giving 'Hairy Kidney Appearance', with Mild Prominence of Right Renal Pelvis

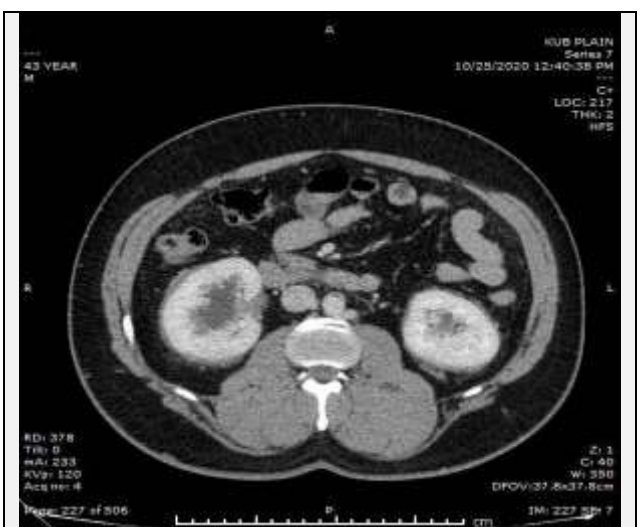

Figure 3. Axial Post-Contrast CT Abdomen (Nephrographic Phase) at Renal Level – Enhancing Soft Tissue Infiltration around Bilateral Kidneys Giving 'Hairy Kidney Appearance', with Mild Prominence of Right Renal Pelvis

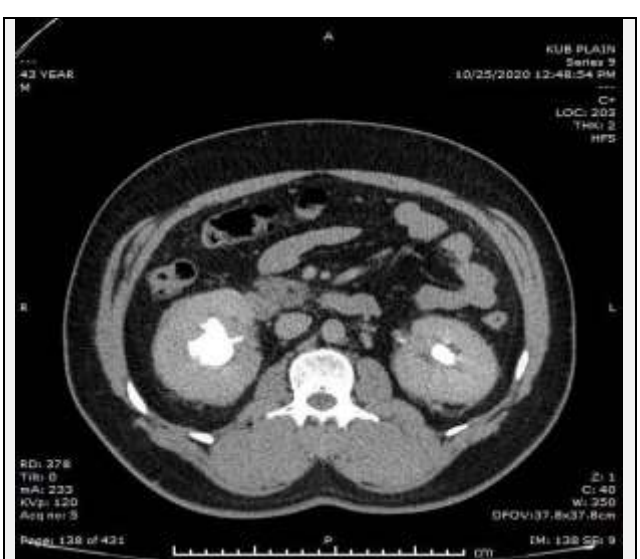

Figure 4. Axial Post-Contrast CT Abdomen (Excretory Phase) at Renal Level – Excreted Contrast in Bilateral Dilated Renal Pelves (Right > Left) with Thin Streak of Contrast Traversing the Narrowed Segment on Right Side

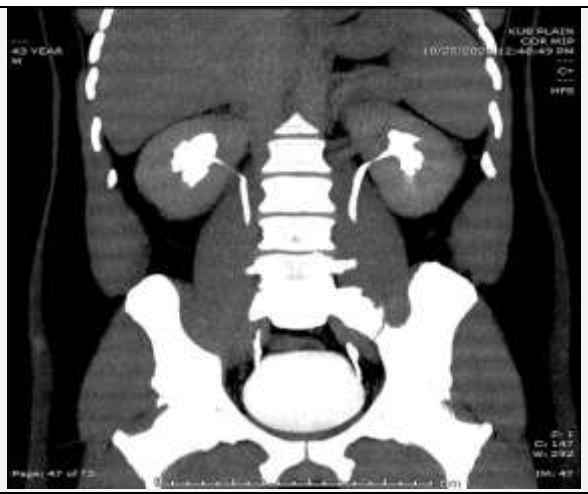

Figure 5. Coronal CT Abdomen (Urographic Image) – Excreted Contrast in Bilateral Dilated Renal Pelvic-Calyceal Systems (Right > Left) with Thin Streak of Contrast Traversing the Narrowed Segments of Bilateral Renal Pelves

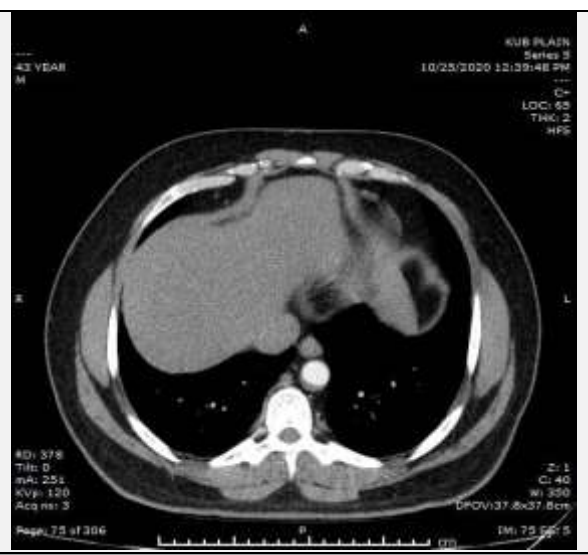

Figure 6. Axial Post-Contrast CT at Lower Chest Level - Soft Tissue Infiltration Covering the Distal Thoracic Aorta, Giving 'Coated Aorta Appearance'

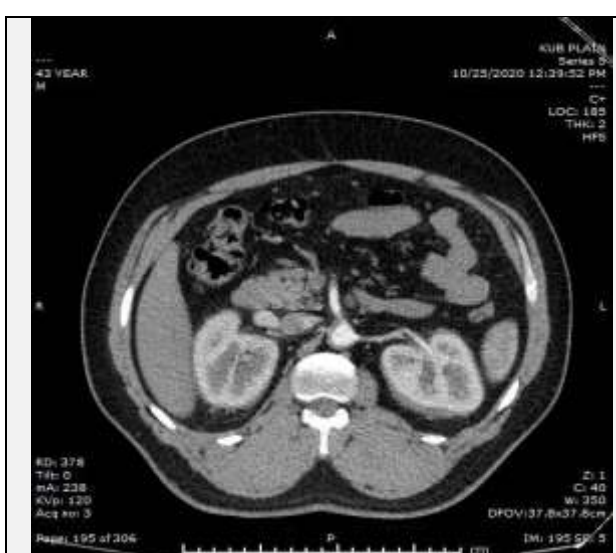

Figure 7. Axial Post-Contrast CT Abdomen at Renal Level - Soft Tissue Infiltration Covering the Abdominal Aorta, Giving 'Coated Aorta Appearance'

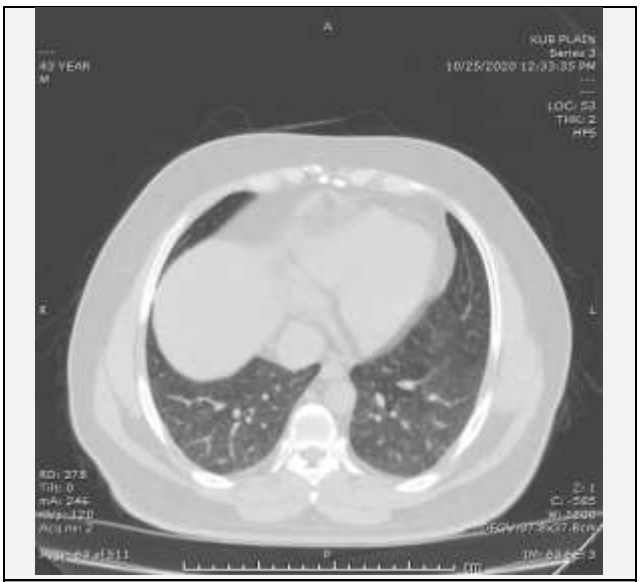

Figure 8. Axial CT (Lung Window) at Lower Chest Level - Few Tiny Centriacinar Nodules with Patchy Ground Glass Opacities in Postero basal Segments of Both Lungs

DISCUSSION

ECD is a rare, non-Langerhans cell histiocytosis with widespread manifestations and highly variable severity, originally described in 1930 by William Chester Tran et al.[1] It is a multisystemic disorder involving skeletal and extra skeletal systems (central nervous system, orbits, pulmonary, cardiovascular, vasculature, kidneys & retroperitoneum).[2]

ECD usually affects people above 40 years of age with slight male preponderance.[3] Patients may present with a variety of non-specific symptoms to multiorgan failure. The most common presenting symptom being bone pain. Patients may also present with focal neurological signs, cerebellar or pyramidal signs, diabetes insipidus, exophthalmos, retroperitoneal fibrosis, obstructive uropathy, cardiac dysfunction and interstitial lung disease due to extra skeletal involvement of these systems.[4] It is associated with significant morbidity and mortality due to infiltration of critical organs and hence an early diagnosis leads to a favourable outcome.[5] Establishing the diagnosis depends on correlation of the radiographic and pathologic findings, due to its non-specific findings.[2]

The aetiology is unknown. Pathologically there is multiorgan infiltration of lipid-laden macrophages, multinucleated giant cells, and inflammatory cells composed of lymphocytes and histiocytes.[6]

Skeletal Manifestations

Conventional radiography of EC disease typically shows a bilateral symmetric pattern of medullary sclerosis involving the diametaphyses of the lower extremities with sparing of the epiphysis. Patchy areas of increased density with coarse trabeculae and cortical thickening may also be seen.[7] Less frequently, mixed osteolytic and osteosclerotic lesions may develop. Involvement of the axial skeleton is rare.[4] On Tc99 m scintigraphy, there is an intense, bilaterally symmetric

uptake at the end of long bones with sparing of the epiphysis, which is characteristic of ECD.[8] Differential diagnoses for bone lesions include lymphoma, chronic osteomyelitis, Paget's disease, or metastases. However, amongst these, symmetrical meta diaphyseal uptake on nuclear scintigraphy is exclusive to ECD.[8,9]

Extra Skeletal Manifestations

CNS & Orbits

Central diabetes insipidus results from posterior pituitary involvement. Dural and meningeal involvement may be either diffusely infiltrating, or there may be formation of discrete masses. Orbital involvement leads to exophthalmos secondary to symmetrical infiltrating hypodense soft tissue usually in retro-orbital, intraconal location, encasing the optic nerve and extraocular muscles. Differential diagnoses for CNS involvement is Langerhans's cell histiocytosis (LCH) (skull lesions are usually lytic compared to sclerotic in ECD). Differentials for bilateral exophthalmos with multicompartmental involvement include Grave's disease, haematological malignancies (leukaemia, lymphoma) and sarcoidosis.[4,5,9,10]

Pulmonary

Pleuroparenchymal involvement leading to inter & intralobular septal thickening; other non-specific findings like centrilobular nodules, cysts, consolidation, ground glass attenuation, or pleural effusion. Findings are indistinguishable from other interstitial lung disease like LCH, sarcoidosis, pulmonary oedema, or veno occlusive disease.[4,5,8]

Cardiovascular and Vasculature

CVS involvement leads to a poor response to chemotherapy and leads to death in ~ 60 % of cases secondary to the compromise of either chamber filling or cardiac conduction defects resulting in myocardial infarction, arrhythmia, or valvular insufficiency. Plaque-like thickening of pericardium/myocardium is seen on imaging. There is plaque like perivascular soft tissue encircling the aorta and its branches, which may be segmental with both symmetric and asymmetric circumferential forms – giving "coated aorta sign" on CT. It may lead to extrinsic compression of the involved segment and consequent ischemic complications like cerebrovascular events, angina or mesenteric ischemia depending on the site affected. Vascular involvement in ECD may be indistinguishable from other vasculitides, especially Takayasu arteritis.[4,5,8]

Kidneys & Retroperitoneum

Soft tissue masses are present in bilateral perirenal and posterior pararenal spaces, causing mass effect on the kidney. Perirenal fat infiltration results in spiculated appearance, termed as the "hairy kidney sign". As the disease progresses, these soft tissue masses compress the pelvicalyceal system or ureters leading to obstructive uropathy. The major differential diagnosis with such pattern of involvement is retroperitoneal fibrosis, either primary or secondary.[8]

Unusual Sites

ECD may potentially involve any site including skin, breast, lymph nodes, thyroid gland, testis, and visceral organs.[4]

In our patient, the classical findings of skeletal and extraskeletal involvements in the given clinical setting led to a radiological diagnosis of ECD. This needed further confirmation by pathological correlation for which the patient was referred to higher centre.

TREATMENT

The treatment usually includes oral steroids and in more severe cases, chemotherapy or external radiation therapy, but with variable outcomes.[11] The most common cause of death include pulmonary and heart failure and the prognosis depends on the extent & distribution of extra skeletal disease.

CONCLUSIONS

Erdheim-Chester disease is a rare multisystem disorder with a spectrum of manifestations and varying severity. Proper radiological workup in patients presenting with specific symptoms leads to identification of various signs of the disease. When the signs are put together one can narrow the differential diagnoses and reach a final diagnosis. Due consideration to this rare diagnosis needs to be given, as the combination of radiological signs are specific in this condition. However, pathological confirmation is necessary to reach a definitive diagnosis, which was not possible in our case due to limited resources. Patient management requires multimodal approach and the outcome is variable.

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Disclosure forms provided by the authors are available with the full text of this article at jebmh.com.

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