ORIGINAL RESEARCH PAPER

Volume-6 | Issue-12 | December-2017 | ISSN No 2277 - 8179 | IF : 4.176 | IC Value : 93.98

INTERNATIONAL JOURNAL OF SCIENTIFIC RESEARCH

A RARE CASE OF ERDHEIM CHESTER DISEASE IN 46 YEAR OLD MALE PATIENT

Radiodiagnosis

Dr. Dhwani upadhya	Third year Resident
Dr.Monik Patel	second year resident
Dr. Lakshminarayanan Sankar	First year Resident
Dr.Purvi Desai	Associate professor Radiology

ABSTRACT

Erdheim Chester is a rare disorder of histiocytes affecting multiple organ systems. We discuss the clinical, radiological, and histopathological findings in 46 year old male who reported with bilateral shaft of femur fracture after fall down at home. He was thoroughly investigated and biopsy confirmed the diagnosis of Erdheim Chester disease.

KEYWORDS

Erdheim Chester disease, polyostotic sclerosing histiocytosis.

Introduction : Erdheim Chester disease is a rare disease characterised by abnormal multiplication of white blood cells called histiocytes / tissue macrophages. It is also termed as Non Langerhans cell Histiocytosis. It is characterised usually by symmetrical long bone involvement & extraskeletal organ involvement like kidney, skin, brain involvement.

Case report:

A 46 year old male came with h/o fall down in bathroom 4 days back. He was admitted in a local hospital and primary treatment was given after which he was referred to NCH Surat. Patient complained of pain over both thighs. On examinations the patients vitals were stable with inability to stand and severe pain over both thighs. Pelvic compression test and chest compressiom test were negative. Other examinatory findings were uneventful. Patient also gives a past history of trauma over both eyes at different times with loss of vision in both eyes.

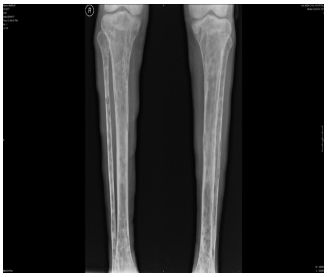
X rays were taken on both lower limbs which revealed spirally displaced fractures noted involving both mid shaft of femur noted. Based on the triviality of the trauma, patient was diagnosed as a case of bilateral pathological fracture mid shaft of femur without neurological deficits Screening USG abdomen revealed hepatosplenomegaly with (64x61x58)mm3 sized abscess in right lobe of liver.

Patient was operated and biopsied to identify the cause of pathological fractures. Patient was extensively evaluated including urine bence jones protein, Hb electrophoresis, protein electrophoresis, serum LDH, serum ALP, Bone marrow aspiration to identify the cause of these pathological fractures.

Positive findings after evaluation include slightly elevated ALP and Lactate Dehydrogenase, slightly Decreased Total protein.

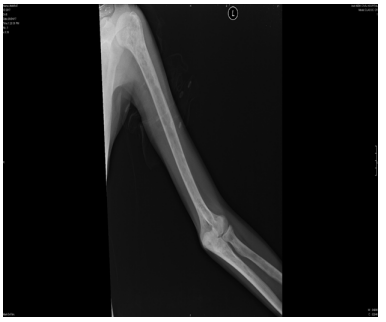
X ray features of long bones revealed Multiple Lytic sclerotic lesions noted involving metadiaphyseal region of Tibia and Fibula with Periosteal Reaction and Cortical Thickening. Relative sparing of subcortical epiphyseal region noted. Other long bones also showed similar picture with lesser severity of lytic sclerotic lesions.

X ray both tibia and fibula



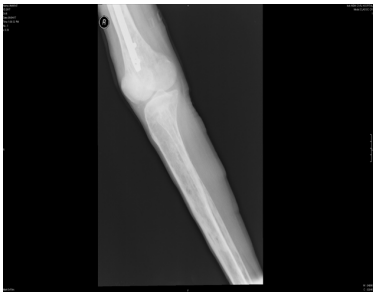
Multiple Lytic sclerotic lesions noted involving metadiaphyseal region of Tibia and Fibula with Periosteal Reaction and Cortical Thickening. Relative sparing of subcortical epiphyseal region noted

X Ray Humerus Left with left elbow joint



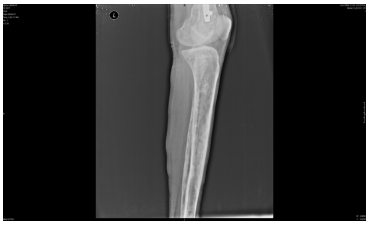
Multiple Lytic sclerotic lesions noted involving metadiaphyseal region of humerus with Periosteal Reaction and Cortical Thickening.

Right Knee joint lateral view



Multiple Lytic sclerotic lesions noted involving metadiaphyseal region of knee with Periosteal Reaction and Cortical Thickening.

Left Knee joint lateral view

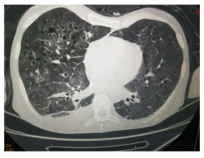
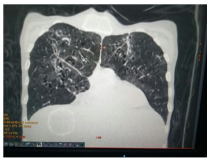
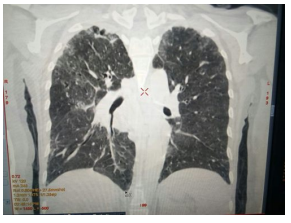


HRCT Thorax revealed multiple ill defined bizarre shaped small cysts with surrounding ground glass opacities and fissural thickening with paraseptal emphysema in right upper apical lobe and mild pleural effusion with collapse of posterobasal segment of right lower lobe. Few subcentimeter sized non necrotic lymphnodes noted in pretracheal, paratracheal, prevascular, precarinal, subcarinal space and aortopulmonary window.

Main pulmonary artery, right and left pulmonary artery measures

32mm, 24mm, 20mm respectively with normal contrast opacification.

HRCT Thorax

Few subcentimeters sized non-necrotic lymph nodes noted in pre/paratracheal, prevascular, pre/subcarinal regions and aortopulmonary window. MPA, RPA and LPA apperas dilated measuring 32 mm, 24 mm and 20 mm respectively and shows normal contrast opacification. Cardiac size is within normal limits.

Cardiac size appeared to be normal.

Histopathological report from the biopsied site revealed a cellular picture suggestive of "Erdheim Chester Disease" MRI spine was done which proved to be uneventful

Discussion

Erdheim Chester disease is a rare multiorgan disorder of specific white blood cells called histiocytes. It is known by variety of names like "polyostotic sclerosing histiocytosis", " Non Langerhans cell Histiocytosis", " Lipid Granulomatosis", " Non familial Multicystic granulomatosis", "S100 nnegative LCH". Clinical symptoms in Erdheim Chester disease include bone pain, dyspnea, diabetes inspidus, renal symptoms, pituitary involvement, exophthalmos etc.

Musculoskeletal symtoms are more common in Erdheim Chester disease which are more pronounced in lower limbs.

Diagnosis of Erdheim Chester disease is mainly by radiology and pathology. Patients with Erdheim Chester disease show bilateral symmetrical patchy osteosclerotic skeletal involvement with sparing of Epiphysis noted in long bones. Periaortic, Perirenal infiltrative involvement is also most common in Erdheim Chester disease.

Skeletal Involvement is a rule in Erdheim Chester disease with 50% showing extraskeletal features.

A mixed lytic osteosclerotic picture is also possible. Erdheim Chester disease is a disease of varied presentation and is a diagnosis of exclusion.

Symmetrical interstitial opacities, septal thickening, fissural thickening, ground glass opacities, Centrilobar nodular opacities, recurrent pleural effusion are all the features possible in lung

Cardiac features of Erdheim Chester disease is one of the most characteristic features. They include periaortic infiltration involving aorta, inferior vena cava,superior vena cava, coronary arteries, aortic branches, pericardial effusion. Periaortic infiltration noted in Erdheim Chester disease is called "Coated aorta". Mortality in Erdheim Chester disease is directly proportional to lung and cardiac involvement.

Renal and perirenal infiltration are also very characteristic of Erdheim Chester disease. Retroperitoneal infiltration on Erdheim Chester disease spares IVC and pelvic ureters which differentiates from retroperitoneal fibrosis. Perirenal infiltration in CT has a characteristic eponym called "Hairy appearance". Secondary obstructive uropathy may occur as a result of perirenal and retroperitoneal involvement. But renal symptoms are very much rare in Erdheim Chester disease.

Skeletal involvement with perirenal and peri aortic infiltration is almost pathognomonic of Erdheim Chester diseaseHypothalamic lesions are usually seen in Erdheim Chester disease. Symmetrical cerebellar and pontine altered signals on T2 weighted imaging is pathognomonic of Erdheim Chester disease. Lentiform Nucleus involvement and ependymal enhancement are other MRi Findings.

Histopathologically, tissue samples show xanthomatous and xanthogranulomatous infiltration by Lipid laden foamy macrophages with surrounding fibrosis Erdheim Chester is a disease with varying presentation and is a diagnosis of clinical exclusion and histopathological confirmation with radiological aid in diagnosis

Results

Erdheim Chester disease is a rare disease of varied presentation.
Our patient being histopathologically diagnosed as Erdheim Chester disease manifested only with skeletal and pulmonary involvement proves the clinical diversity of Erdheim Chester disease.

REFERENCES

1. Veyssier-Belot C, Cacoub P, Caparros-Lefebvre D, et al. Erdheim-Chester disease: clinical and radiologic characteristics of 59 cases. Medecine. 1996;75(3):157–69. [PubMed]
2. Gomez C, Diard F, Chateil JF, Moinard M. Imagerie de la maladie d'Erdheim-Chester. J Radiol. 1996;77:1213–21. [in French] [PubMed]
3. Cattin F, Runge M, Nagy N, et al. [Case #5. Erdheim-Chester disease] J Radiol. 2005;86:527–30. [in French] [PubMed]
4. Kenn W, Stäbler A, Zachoval R, et al. Erdheim-Chester disease: a case report and literature overview. Eur Radiol. 1999;9:153–58. [PubMed]
5. Breuil V, Brocq O, Pellegrino C, et al. Erdheim-Chester Disease: typical radiological bone features for a rare xanthogranulomatosis. Ann Rheum Dis. 2002;66:199–200. [PMC free article] [PubMed]
6. Evans S, Williams F. Case report: Erdheim-Chester disease: Polyostotic sclerosing histiocytosis. Clin Radiol. 1986;37:93–96. [PubMed]
7. Murray D, Marshall M, England E, et al. Erdheim-Chester disease. Clin Radiol. 2001;56:481–84. [PubMed]
8. Rozas Reyes P, Senaris Gonzales A, Gonzales Rodriguez CM. Orbit xanthogranul omatosis. Erdheim-Chester disease. Arch Soc Esp Oftalmol. 2 0 0 4 ; 7 9 : 5 1 5 – 1 8 . [PubMed]
9. Rosier RN, Rosenberg AE. Case 9–2000 – A 41-year-old man with multiple bony lesions and adjacent soft-tissue masses. N Engl J Med. 2000;342(12):875–83. [PubMed]
10. Waite RJ, Doherty PW, Liepmann M, Woda B. Langerhans cell histiocytosis with the radiographic findings of Erdheim-Chester disease. Am J Roentgenol. 1988;150:869–71. [PubMed]
11. Brower AC, Worsham GF, Dudley AH. Erdheim-Chester disease: a distinct lipoidosis or part of the spectrum of histiocytosis? Radiology. 1984;151(1):35–38. [PubMed]
12. Martinez R. Erdheim-Chester disease: MR of intraaxial and extraaxial brain stem lesions. Am J Neuroradiol. 1995;16:1787–90. [PubMed]
13. Weidauer S, von Stuckrad-Barre S, Dettman E, et al. Cerebral Erdheim-Chester disease: case report and review of the literature. Neuroradiology. 2003;45:241–45. [PubMed]
14. Tien RD, Brasch RC, Jackson DE, Dillon WP. Cerebral Erdheim-Chester disease: persistent enhancement with GD-DTPA on MR images. Radiology. 1989;172:791–92. [PubMed]
15. Dion E, Graef C, Haroche J, et al. Imaging of thoracoabdominal involvement in Erdheim-Chester disease. Am J Roentgenol. 2004;183:1253–60. [PubMed]
16. Abdelfattah AM, Arnaout K, Tabbara IA. Erdheim-Chester disease: A comprehensive review. Anticancer Res. 2014;34(7):3257–61. [PubMed]
17. Mazor RD, Manevich-Mazor M, Shoenfeld Y. Erdheim-Chester disease: a comprehensive review of the literature. Orphanet J Rare Dis. 2013;8:137. [PMC free article] [PubMed]