Case report

Recurrent and rapidly occurring pericardial tamponade in Erdheim Chester disease

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KEYWORDS

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ABSTRACT

Erdheim Chester disease is a very rare histiocytic disorder characterised by tissue infiltration by lipid laden histiocytes. The most common presentation is bone pains typically involving the long bones. Over time almost 50% of the patients develop extraosseous involvement. The prognosis depends on the extent and distribution of the extraskeletal manifestations. Cardiovascular involvement is seen in up to 40% of the patients and the most common manifestations are periaortic fibrosis and pericardial involvement. Respiratory distress, extensive pulmonary fibrosis, and cardiac failure are the most common causes of death in these patients. Cardiac tamponade has also been documented to cause death in these patients. We describe a patient of Erdheim Chester disease who presented with recurrent and very rapidly occurring cardiac tamponade in a short duration of time and benefited from timely recognition and management.

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Introduction

Erdheim Chester disease is a rare histiocytic disorder involving multiple systems with <400 cases been reported worldwide. Bony involvement is most common manifestation, although cardiovascular involvement is also frequently seen and is one of the common causes of death. Patients can present with recurrent, rapid, and life-threatening pericardial tamponade. Timely recognition of the disease and appropriate therapy can be lifesaving in these patients.

Case history

A 52-year-old man was seen in the emergency department with complaints of steadily progressing dyspnoea on exertion of 5–6 days duration. Clinical examination revealed tachycardia with a heart rate of 122 bpm, blood pressure of 100/64 mmHg, and muffled heart sounds. Chest radiography

showed cardiomegaly and an urgent echocardiography done showed a large pericardial effusion (PE) with echocardiographic features of cardiac tamponade (Figures 1 and 2). An immediate pericardiocentesis was done and 2500 mL of fluid was drained. Fluid analysis was suggestive of an exudative effusion and cytological examination showed the presence of mesothelial cells and more lymphocytes than neutrophils. The past history of the patient revealed that he was diagnosed to have diabetes insipidus and then subsequently found to have Erdheim Chester disease one year back. He had presented then with polyuria, polydipsia, and bilateral proptosis and had subsequently developed bone pains involving both the legs. A bone scintigraphy showed areas of increased tracer uptake. Diagnosis was confirmed from a tissue specimen taken from the retro-orbital space which showed the typical histological picture and immunohistochemistry.

He gave history of dyspnoea a month back and was also found to have a large PE which needed pericardiocentesis. He underwent three pericardiocentesis during this period because of rapid reaccumulation. Each pericardial aspiration had yielded 2000–2500 mL of fluid. Following this he was referred to our centre. By day 4 of the present admission the

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Figure 1 Chest radiograph taken in the emergency department showed a large cardiac shadow without any significant lung pathology.

patient had developed another large effusion needing drainage. A catheter was positioned in the pericardial cavity for frequent fluid aspiration. Almost 1000–1500 mL of fluid was aspirated every day. Investigations carried out showed no other apparent cause of PE and the features suggested that the recurrent PEs were due to Erdheim Chester disease involving the pericardium. The patient was already on interferon therapy but that did not prevent the pericardial involvement. Considering the life-threatening, recurrent, and rapid reaccumulation of fluid in the pericardial cavity, we decided that the patient should undergo a pericardiectomy. He underwent a surgery on day 10 of admission. Postoperative period was uneventful and the patient was discharged from hospital with an advice to continue interferon therapy. The patient came back for review after 6 months. He was asymptomatic with New York Heart Association (NYHA) functional class I.

Discussion

Erdheim Chester disease is a very rare form of histiocytosis with no specific aetiology. Histiocytosis is a group of rare disorders characterised by tissue infiltration by macrophages, monocytes or dendritic cells. It has further been broadly divided into Langerhan cell histiocytosis (LCH) and non-langerhan cell histiocytosis.^{1,2} The LCH is characterised by the infiltration of the tissues by the characteristic dendritic cells seen in the epidermis. The disease has a wide range of presentation and was earlier described as separate entities: (1) Letterer-Siwe disease; (2) Hand-Schüller-Christian disease; and (3) eosinophilic granuloma. They were thought to be different disorders but have now been considered to be various forms of LCH presentation.^{2,3} Although LCH involves multiple organ systems, the most common involvement is that of the bony system which is characterised by osteolytic lesions. The other organ systems involved are the skin, posterior pituitary gland, lymph nodes, rarely the gastrointestinal tract, nervous system, lungs, and the liver. The diagnosis of LCH is confirmed by the characteristic histologic picture and the presence of CD1a or CD207 positive histiocytic cells.¹ Electron microscopy shows

Figure 2 Urgent echocardiogram showed a very large pericardial effusion with typical features of tamponade. Patient had similar echocardiographic features 5 times within a span of 30–40 days necessitating urgent pericardiocentesis.

Figure 3 Xanthogranulomatous reaction with foamy histiocyte (arrow head).

the presence of Birbeck granules in the cells but is rarely used currently to diagnose the disease.

Non-LCH is an even more uncommon disease which has been termed as Erdheim Chester disease after the two people who described it first. It is a very rare disorder with a total of <400 cases reported worldwide. This disease is characterised by a mononuclear infiltrate consisting of lipid laden, foamy histiocytes with surrounding fibrosis (Figure 3). The histiocytes are constantly positive for CD68 and negative for CD1a (Figure 4) and S100, and ultrastructural studies show no Birbeck granules in contrast to the findings in LCH.^{4,5} The clinical manifestations range from localised form to a disseminated and life-threatening disease. Bone pain is the most frequent symptom. The disease is characterised by the involvement of the long bones, unlike the flat bone involvement in LCH. The bony involvement is in the form of osteosclerotic lesions involving the metaphyseal and diaphyseal region and sparing the epiphyses. This pattern of involvement represents the almost pathognomonic radiologic picture seen in this disorder.⁶ The extraskeletal manifestations include exophthalmos, diabetes insipidus, interstitial lung disease, bilateral adrenal enlargement, retroperitoneal fibrosis, renal impairment,

Figure 4 Immunohistochemistry is negative for CD1a.

testis infiltration, central nervous system, and cardiovascular involvement. The diagnosis is based on the almost pathognomonic radiological picture mentioned and the typical histologic picture with typical immunohistochemical staining. There is also a characteristically abnormal increased uptake of technetium-99 in the long bones of the lower limbs.⁷

Cardiovascular involvement is one of the most common causes of death and morbidity in these patients. Periaortic fibrosis and pericardial involvement are the two most common cardiovascular pathologies. Periaortic fibrosis leads to the so called “coated aorta” which is due to extensive fibrosis in the adventitial layer of the aorta which may vary from short segment to the involvement of the whole aorta. Pericardial involvement varies from only pericardial thickening to PEs with occasional cardiac tamponade. In a large series of 184 cases, 72 (39%) had cardiovascular involvement and of these 74 patients, 54 (75%) had involvement of the heart. Pericardial infiltration was seen in 32 (44%) of the patients (leading to tamponade in 5 cases), myocardial infiltration in 22 cases (31%), 40 (56%) patients had a periaortic fibrosis, and 19 (26%) patients had heart failure. There were also a small number of patients who had valvular lesions, right atrial thickening, and even acute myocardial infarction. Among the 58 patients (81%) with follow-up data, 35 (60%) died. Death was due to the cardiovascular involvement in 31% of the cases.⁸

Since, this is a very rare disorder, there is no consensus on the definitive therapy. Various modalities used include corticosteroids, immunomodulators, radiotherapy, and interferon- α . Our patient was also started on interferon- α ; however, that obviously did not preclude cardiovascular involvement, although the duration since the initiation of therapy was less than a year. Importantly the reaccumulation of effusions was very rapid causing tamponade, and was life-threatening. Patient had to undergo repeated pericardiocentesis necessitating pericardiectomy, which proved to be lifesaving in this case.

Conclusion

Erdheim Chester disease is a rare disorder with frequent cardiovascular involvement. Pericardial involvement is frequently seen and has caused death by pericardial tamponade in these patients. While a definitive therapeutic approach is still not defined, various modalities include corticosteroids, radiotherapy, and interferon- α . However, these therapies may not prevent a cardiovascular involvement. Rapidly accumulating PEs may be life-threatening. Prompt recognition of the aetiology and appropriate management can be lifesaving. A surgical pericardiectomy can be an option in such cases and the patients seem to do well after surgery.

References

1. Windebank K, Nanduri V. Langerhans cell histiocytosis. *Arch Dis Child* 2009;94:904–8.
2. Azouz EM, Saigal G, Rodriguez MM, et al. Langerhans' cell histiocytosis: pathology, imaging and treatment of skeletal involvement. *Pediatr Radiol* 2005;35:103–15.
3. Egeler RM, D'Angio GJ. Langerhans cell histiocytosis. *J Pediatr* 1995;127:1–11.
4. Simiele N, Novoa F, Rodriguez N. Erdheim-Chester disease and Langerhans histiocytosis. A fortuitous association? *Ann Med Int* 2004;21:27–30.
5. Sheu SY, Wenzel RR, Kersting C, et al. Erdheim-Chester disease: case report with multisystemic manifestations including testes, thyroid, and lymph nodes, and a review of literature. *J Clin Pathol* 2004;57:1225–8.
6. Breuil V, Brocq O, Pellegrino C, et al. Erdheim-Chester disease: typical radiological bone features for a rare xanthogranulomatosis. *Ann Rheum Dis* 2002;61:199–200.
7. Spyridonidis TJ, Giannakenas C, Barla P, et al. Erdheim-Chester disease: a rare syndrome with a characteristic bone scintigraphy pattern. *Ann Nucl Med* 2008;22:323–6.
8. Haroche J, Amoura Z, Dion E, et al. Cardiovascular involvement, an overlooked feature of Erdheim-Chester Disease: report of 6 new cases and a literature review. *Medicine* 2004;83:371–92.