Surgical Appearance of Aortic Involvement in Erdheim–Chester Disease: A Report of Two Cases

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SOUHRN

Protože Erdheimova–Chesterova nemoc (Erdheim–Chester disease, ECD), známá i pod označením polyostotická sklerotická histiocytóza, působí i na cévní struktury, může být mylně diagnostikována jako ischemická choroba dolních končetin. Postižení aorty při ECD obvykle odhalí rentgenové vyšetření. Dosud však nemáme k dispozici dostatek informací o vzhledu postižení během chirurgického výkonu ani o léčbě postižení aorty při ECD. V tomto článku popisujeme případ pacienta s postižením vzestupné aorty zjištěným náhodně při provádění koronárního bypassu a dalšího pacienta s postižením břišní aorty připomínajícím aortoiliackou okluzivní nemoc.

ABSTRACT

Erdheim–Chester disease (ECD), known as histiocytic neoplastic disease, can be diagnosed as a peripheral arterial disease because of its effects on vascular structures. Aortic involvement in ECD is usually recognised radiologically. However, there is insufficient information about the surgical appearance and treatment of aortic involvement in ECD. In this report, we presented a patient with ascending aortic involvement detected incidentally during coronary bypass and a patient with abdominal aortic involvement mimicking aortoiliac disease.

Introduction

Erdheim–Chester disease (ECD), which was classified as a haematopoietic neoplasm of histiocytic origin by the World Health Organisation, is defined as Langerhans (L) group histiocytosis characterised by foamy macrophage accumulation, chronic inflammation, and fibrosis.[1,2] Although ECD is diagnosed histologically, it is increasingly reported in the literature, thanks to radiological advances, but its global incidence is still unknown. ECD can be asymptomatic or life-threatening by causing complications such as myocardial ischaemia, organ perfusion damage, and renovascular hypertension.[3,4] Unlike other system involvement, it is rare that patients present to the hospital with vascular involvement.[5] Aortic involvement has been mostly reported in the literature.[6]

Herein, we present a patient with ascending aortic involvement detected incidentally during coronary bypass and a patient with abdominal aortic involvement mimicking aortoiliac disease.

Case description

Case 1

A 57-year-old male patient was admitted to the cardiology clinic with chest pain. Coronary angiography was performed after the effort test was positive. The patient had no complaints, except for stable angina pectoris. He had used hypertension treatment (beta blocker) for 5 years, and a stent had been applied to the left anterior descending artery (LAD) 3 years ago. He did not have diabetes, any systemic disease, and did not smoke. In his family, only patient's mother had coronary artery disease. Physical examination, biochemical parameters, and complete blood count were normal. Echocardiography showed that ejection fraction was normal (60%). Because the LAD proximal lesion was not suitable for percutaneous intervention, coronary artery bypass graft (CABG) operation was selected after coronary angiography (Figs 1A, 1B). After the pericardium had been opened, the ascending aorta was very stiff and had a marble-like appearance surroun-

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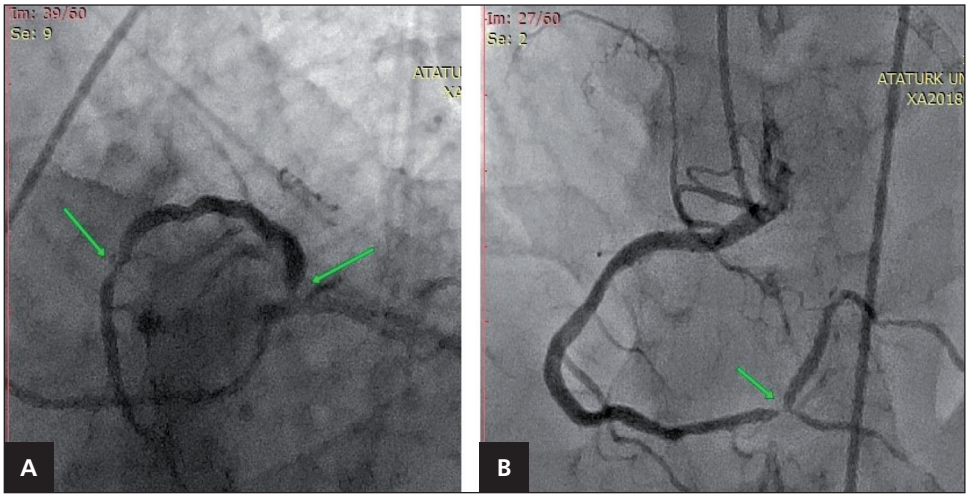

Fig. 1 – Coronary angiography image: (A) LAD stenosis and (B) RCA stenosis (arrow).

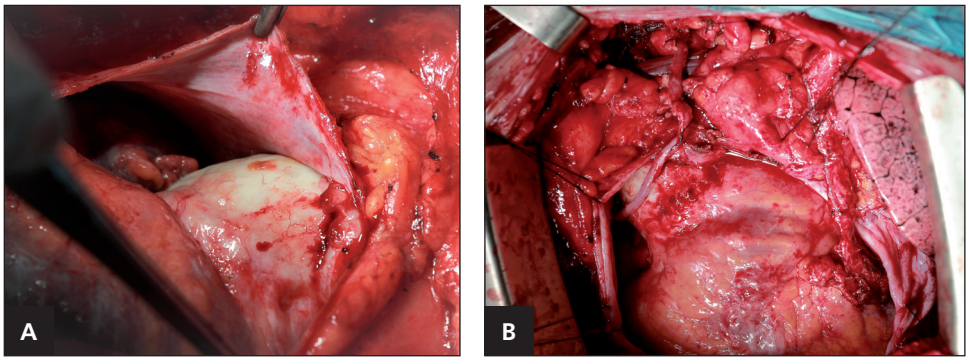

Fig. 2 – (A) Intraoperative view of the ascending aorta. (B) Left internal mammary artery and saphenous vein anastomosis.

ded by grey-white fibrous tissue (Figs 2A, 2B). Therefore, off-pump CABG was performed. After the left internal mammary artery–LAD anastomosis was completed, the proximal part of the right coronary artery (RCA) saphenous anastomosis was applied to the truncus brachiocephalic artery (Figs 2A, 2B). The image of the aorta was compatible with ECD. Therefore, in the biopsy taken from the ascending aorta, a mononuclear infiltrate of lipid-loaded foamy histiocytes stained positive for CD68 and negative for CD1a (Figs 3A, 3B). No complications such as mediastinitis developed during the follow-up. Since the patient was asymptomatic for ECD, he was treated for coronary artery disease and was referred to the oncology clinic after discharge. Since he had undergone surgery, corticosteroid tablets were prescribed by the oncology department, and he was followed up closely.

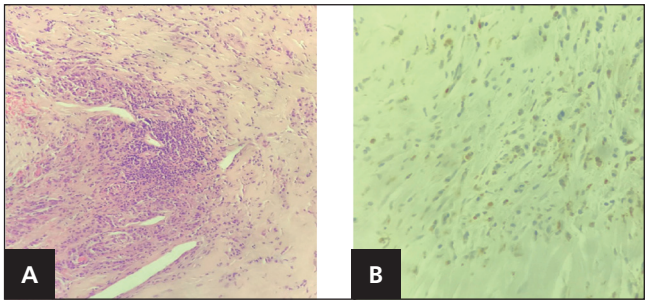

Fig. 3 – Pathological view of the biopsy taken from the ascending aorta for the first patient: (A) fibrotic background with mononuclear inflammatory cells; (B) CD-68 + histiocytic cells.

Case 2

A 54-year-old male patient was admitted to our clinic with the complaint of intermittent claudication at 150 m and walking distance decreased compared with the last year, and the pain was usually localised at hip level. He had not smoked for 5 years, and he had no other disease in his medical history and family. Pulse was weakly palpable on the femoral and distal artery bed, and the ankle–brachial index value was 0.8. All laboratory values were within the normal limits. Abdominal aortic colour Doppler ultrasonography (USG) was performed for the prediagnosis of peripheral artery disease. Abdominal computed tomography (ACT) was performed because the abdominal aortic diameter measured 4 cm on USG. On ACT, the abdominal aorta was completely covered with fibrous tissue, and the diameter at the bifurcation level was <1.5 cm. Since the image made us think of ECD, we wanted to perform periaortic biopsy guided by CT, it was suggested to the patient; however, the patient refused to undergo the procedure. He was referred to oncology with a prediagnosis of ECD. The patient was followed because he did not have any

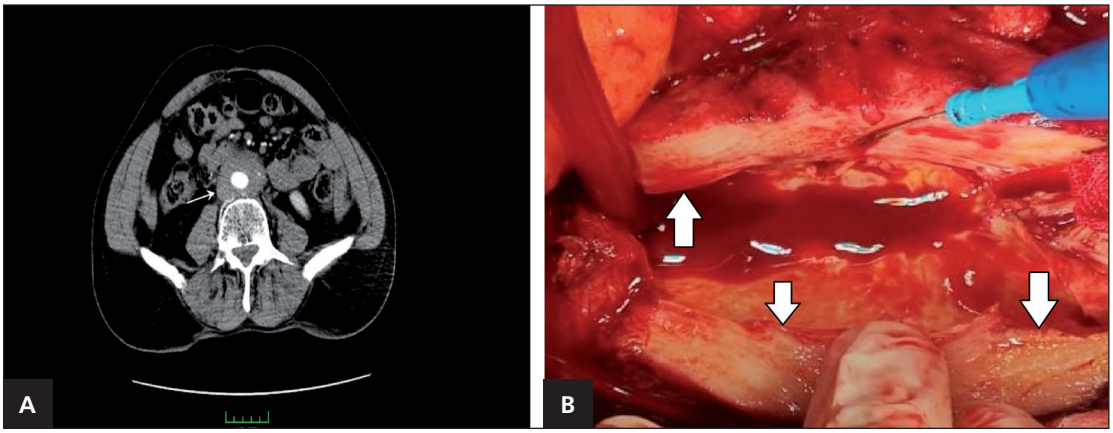

Fig. 4 – (A) Muscle-isodense tissue coated the abdominal aorta (arrow) and (B) thick fibrotic wall of the abdominal aorta intraoperatively (arrow).

other organ involvement and was negative for *BRAF* mutation.

Nearly 6 months later, the patient was hospitalised for severe right lower-extremity pain at rest, and surgery was decided because the right femoral artery pulse was not palpable. The abdominal aorta was exposed by median laparotomy. The aorta had a firm texture with a thick fibrous tissue narrowing the lumen, and iliac artery stenosis was prominent at the level of the bifurcation (Figs 4A, 4B) Aortobifemoral bypass with 16-8 mm gelatin sealed, knitted Dacron polyester was performed, and a large-scale biopsy was taken from the abdominal aorta. Biopsy showed CD1a-negative histiocytes and CD68-positive histiocytic multinuclear cells and diffuse fibrous stroma including foamy histiocyte infiltration and mononuclear cells (Figs 5A, 5B). After the operation, the patient's complaints improved. The treatment with sirolimus and prednisone tablets was initiated by the oncology department, as the patient was followed up for pseudoaneurysm. No complications developed in the first year of follow-up.

Discussion

Given its rarity, vascular surgeons seldom encountered patients with ECD. Our knowledge of ECD is limited to the approximately 1500 patients identified since the first case of ECD was reported in 1930. Multisystem involvement and potentially fatal nature make ECD an important disease, and in the clinic, the disease may be asymptomatic or symptomatic depending on the organs involved.[7,8] It is not easy to predict which organ will be affected by the ECD. Therefore, a patient with ECD should undergo whole-body scanning; however, cardiovascular system involvement is known to be associated with a poor prognosis.[9] Villatoro-Villar et al.[5] published the rates of cardiovascular system involvement in ECD. Our first patient did not have cardiac involvement, such as pericardial effusion, pericardial and myocardial thickening and non-atherosclerotic coronary artery thickening, as described in a retrospective cohort study.[5] This patient had coronary artery disease and had no complaints, except for stable angina pectoris, and had no symptoms of ECD. LAD and RCA lesions were atherosclerotic, and aortic involvement in ECD was observed during CABG. The right atrium was normal, and coronary arteries were normal enough to easily perform distal anastomoses. This may indicate that ECD has a very different clinical picture because of its heterogeneous nature.

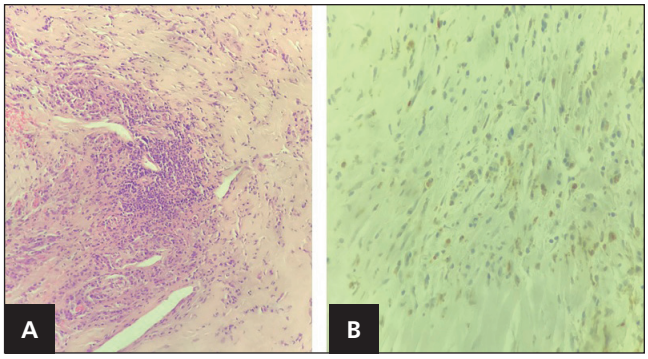

Fig. 5 – Pathological view of the biopsy taken from the abdominal aorta for the second patient. (A) Foamy histiocytic infiltration and diffuse fibrous stroma; (B) CD68+ histiocytic multinuclear cells.

The 'coated aorta' phenomenon was defined by Serratrice et al. as a new sign of ECD.[10] This pathognomonic sign, which is a radiological definition, should be differentiated from aortic aneurysms. However, we have macroscopically viewed aortic involvement of ECD. The ascending aorta was so hard that we described this as a marble aorta. The abdominal aorta also had a firm structure but not as much as the ascending aorta, and we did not experience any difficulty in placing the graft end-to-end proximal to the abdominal aorta. This may be due to the disease duration or its different behaviours depending on the organ involved. In the literature, various procedures are performed according to organ involvement, such as pericardial, intracranial, and orbital. However, we did not find any operative images related to aortic involvement, except for radiological imaging. Therefore, the images we obtained from our patients are valuable (Figs 2A, B and Figs 5A, 5B). Although ECD has been described radiologically for years, cohort studies are necessary to give a surgical name to the macroscopic image of ECD.

Aortic involvement in ECD is usually asymptomatic and is detected incidentally or becomes symptomatic by causing stenosis according to the affected anatomical region. Haroche et al.[11] reported the main aortic branch involvement in 13 of 66 patients. In the arterial map of Villatoro-Villar

et al.,[5] this rate was 10–30%. Since ECD is not a very common disease, these rates are likely to be higher. Our two cases were identified incidentally and radiologically. In the first patient, the ascending aorta was surrounded by fibrous tissue from the truncus to the brachiocephalic branch. The incision of the brachiocephalic trunk for the proximal anastomosis of the RCA showed that the lumen was completely open, and there was no non-ostial infiltration.[12]

In the second patient, intermittent claudication made us consider aortoiliac disease. In our clinic, our first approach for these patients is to perform non-invasive examinations. As an abdominal aortic diameter of 4 cm and stenosis extending to the iliac bifurcation level were detected on USG, we considered the prediagnosis of ECD with ACT. Therefore, we wanted to perform CT-guided periaortic biopsy; however, the patient declined the procedure. We performed surgical intervention because the patient complained of severe lower-extremity resting pain after approximately 6 months. The clinical picture of this case highlighted that patients with ECD should be followed closely and immunosuppressive therapy should be started immediately after diagnosis.

Surgical or endovascular treatment strategies may be considered when the aorta and its branches are involved.[5,11] However, there is no definite information about treatment interventions in patients with vascular involvement but renovascular stent. These interventions must be performed more carefully than traditional atherosclerotic vascular interventions. According to our experience, the fibrous tissue surrounding the vessel can damage the stent and even make its placement difficult. As the surgical experiences of aortic involvement in ECD become widespread, our surgical and interventional knowledge will increase.

Conclusions

Although ECD is extremely rare, it should be well recognised in terms of vascular involvement. Asymptomatic vascular involvement, and the lack of knowledge and experience may overlook many cases. Perhaps, cardiovascular surgeons who do not frequently encounter ECD may not have recognised ECD in most patients with aortic aneurysms. Therefore, we believe that a retrospective re-evaluation of patients with aneurysms is beneficial. Surgeons presenting their experiences on vascular involvement will provide us with more information about ECD.

Author contributions

Conceptualization: Ümit Arslan, Oktay Tüydeş
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Writing – original draft: Ümit Arslan
Writing – review & editing: Ümit Arslan, Bilgehan Erkut

Conflict of interest

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Ethical statement

Not applicable for case reports in Turkey.

Informed consent

Written informed consent was obtained from the patient's legal guardians/parents for publication of this case report and any accompanying images. A copy of the written consent is available for review by the Editor-in-Chief of this journal.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

Declaration of data availability

We accept that our name, e-mail address, affiliation, and other contact details the publication might require will be used for the regular operations of the publication. Data sharing is not applicable to this article as no new data were created or analyzed in this study. The data that support the findings of this study are available in the supplementary material of this article.

References

1. Estrada-Veras JI, O'Brien KJ, Boyd LC, et al. The clinical spectrum of Erdheim-Chester disease: an observational cohort study. Blood Advances 2017;1:357–366.
2. Emile JF, Abla O, Fraitag S, et al. Histiocyte Society. Revised classification of histiocytoses and neoplasms of the macrophage-dendritic cell lineages. Blood 2016;127:2672–2681.
3. Pegoraro F, Papo M, Maniscalco V, et al. Erdheim–Chester disease: a rapidly evolving disease model. Leukemia 2020;34:2840–2857.
4. Das JP, Xie L, Riedl CC, et al. Cardiothoracic manifestations of Erdheim-Chester disease. Br J Radiol 2019;92:20190473.
5. Villatoro-Villar M, Bold MS, Warrington KJ, et al.Arterial involvement in Erdheim–Chester disease, Medicine 2018;49:e13452.
6. Cohen-Aubart F, Emile JF, Carrat F, et al. Phenotypes and survival in Erdheim-Chester disease: Results from a 165-patient cohort. Am J Hematol 2018;93:E114–E117.
7. Campochiaro C, Tomelleri A, Cavalli G. Erdheim-Chester disease. Eur J Intern Med 2015;26:223–229.
8. Goyal G, Heaney ML, Collin M, et al. Erdheim-Chester disease:consensus recommendations for evaluation, diagnosis, and treatment in the molecular era. Blood 2020;135:1929–1945.
9. Ghotra AS, Thompson K, Lopez-Mattei J, et al. Cardiovascular manifestations of Erdheim–Chester disease. Echocardiography 2019;2:229-236.
10. Serratrice J, Granel B, De Roux C, et al. "Coated aorta": a new sign of Erdheim–Chester disease. J Rheumatol 2000;27:1550–1553.
11. 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 (Baltimore) 2004;83:371–392.
12. Dion E, Graef C, Haroche J, et al. Imaging of thoracoabdominal involvement in Erdheim-Chester disease. Am J Roentgenol 2004;183:1253–1260.