

World Journal of *Clinical Cases*

World J Clin Cases 2020 February 6; 8(3): 487-651



REVIEW

- 487 Comprehensive review into the challenges of gastrointestinal tumors in the Gulf and Levant countries
Rare Tumors GI Group, Farhat F, Farsi AA, Mohieldin A, Bahrani BA, Sbaity E, Jaffar H, Kattan J, Rasul K, Saad K, Assi T, Morsi WE, Abood RA

CLINICAL AND TRANSLATIONAL RESEARCH

- 504 Novel zinc alloys for biodegradable surgical staples
Amano H, Miyake K, Hinoki A, Yokota K, Kinoshita F, Nakazawa A, Tanaka Y, Seto Y, Uchida H

CASE REPORT

- 517 Can cyclin-dependent kinase 4/6 inhibitors convert inoperable breast cancer relapse to operability? A case report
Palleschi M, Maltoni R, Barzotti E, Melegari E, Curcio A, Ceconetto L, Sarti S, Manunta S, Rocca A
- 522 Radiation recall dermatitis with dabrafenib and trametinib: A case report
Yilmaz M, Celik U, Hascicek S
- 527 Isolated vaginal metastasis from stage I colon cancer: A case report
Kwon SK, Yu CS, Lee SW, Kim J, Song I, Lee JL, Kim CW, Yoon YS, Park IJ, Lim SB, Kim JC
- 535 Ruptured splenic peliosis in a patient with no comorbidity: A case report
Rhu J, Cho J
- 540 Successful kidney transplantation from an expanded criteria donor with long-term extracorporeal membrane oxygenation treatment: A case report
Seo HW, Lee S, Lee HY, Park SC, Chung BH, Yang CW, Ban TH
- 546 Boarding issue in a commercial flight for patients with cavitary pulmonary tuberculosis: A case report
Jo W, Pak C, Jegal Y, Seo KW
- 552 Cytomegalovirus ileo-pancolitis presenting as toxic megacolon in an immunocompetent patient: A case report
Cho JH, Choi JH
- 560 Successful treatment of adult-onset still disease caused by pulmonary infection-associated hemophagocytic lymphohistiocytosis: A case report
Wang G, Jin XR, Jiang DX

- 568** Complex liver retransplantation to treat graft loss due to long-term biliary tract complication after liver transplantation: A case report
Li J, Guo QJ, Jiang WT, Zheng H, Shen ZY
- 577** Peutz-Jeghers syndrome with mesenteric fibromatosis: A case report and review of literature
Cai HJ, Wang H, Cao N, Wang W, Sun XX, Huang B
- 587** Cutaneous nodules and a novel GNAS mutation in a Chinese boy with pseudohypoparathyroidism type Ia: A case report and review of literature
Li YL, Han T, Hong F
- 594** Complete response to trastuzumab and chemotherapy in recurrent urothelial bladder carcinoma with HER2 gene amplification: A case report
Jiang Q, Xie MX, Zhang XC
- 600** Large cutaneous epithelioid angiomatous nodules in a patient with nephrotic syndrome: A case report
Cheng DJ, Zheng XY, Tang SF
- 606** Clinicopathologic characteristics of prostatic stromal sarcoma with rhabdoid features: A case report
Li RG, Huang J
- 614** Erdheim-Chester disease with asymmetric talus involvement: A case report
Xia Q, Tao C, Zhu KW, Zhong WY, Li PL, Jiang Y, Mao MZ
- 624** Camrelizumab (SHR-1210) leading to reactive capillary hemangioma in the gingiva: A case report
Yu Q, Wang WX
- 630** Combined surgical and interventional treatment of tandem carotid artery and middle cerebral artery embolus: A case report
Zhang M, Hao JH, Lin K, Cui QK, Zhang LY
- 638** Sternal Hodgkin's lymphoma: A case report and review of literature
Yin YY, Zhao N, Yang B, Xin H
- 645** Esophageal tuberculosis complicated with intestinal tuberculosis: A case report
Mao L, Zhou XT, Li JP, Li J, Wang F, Ma HM, Su XL, Wang X

ABOUT COVER

Editorial Board Member of *World Journal of Clinical Cases*, Valerio D'Orazi, MD, PhD, Professor, Department of Surgical Sciences, Sapienza University of Rome, Rome 00161, Italy

AIMS AND SCOPE

The primary aim of *World Journal of Clinical Cases (WJCC, World J Clin Cases)* is to provide scholars and readers from various fields of clinical medicine with a platform to publish high-quality clinical research articles and communicate their research findings online.

WJCC mainly publishes articles reporting research results and findings obtained in the field of clinical medicine and covering a wide range of topics, including case control studies, retrospective cohort studies, retrospective studies, clinical trials studies, observational studies, prospective studies, randomized controlled trials, randomized clinical trials, systematic reviews, meta-analysis, and case reports.

INDEXING/ABSTRACTING

The *WJCC* is now indexed in PubMed, PubMed Central, Science Citation Index Expanded (also known as SciSearch®), and Journal Citation Reports/Science Edition. The 2019 Edition of Journal Citation Reports cites the 2018 impact factor for *WJCC* as 1.153 (5-year impact factor: N/A), ranking *WJCC* as 99 among 160 journals in Medicine, General and Internal (quartile in category Q3).

RESPONSIBLE EDITORS FOR THIS ISSUE

Responsible Electronic Editor: *Yan-Xia Xing*
 Proofing Production Department Director: *Xiang Li*

NAME OF JOURNAL <i>World Journal of Clinical Cases</i>
ISSN ISSN 2307-8960 (online)
LAUNCH DATE April 16, 2013
FREQUENCY Semimonthly
EDITORS-IN-CHIEF Dennis A Bloomfield, Bao-Gan Peng, Sandro Vento
EDITORIAL BOARD MEMBERS https://www.wjnet.com/2307-8960/editorialboard.htm
EDITORIAL OFFICE Jin-Lei Wang, Director
PUBLICATION DATE February 6, 2020

COPYRIGHT © 2020 Baishideng Publishing Group Inc
INSTRUCTIONS TO AUTHORS https://www.wjnet.com/bpg/gerinfo/204
GUIDELINES FOR ETHICS DOCUMENTS https://www.wjnet.com/bpg/GerInfo/287
GUIDELINES FOR NON-NATIVE SPEAKERS OF ENGLISH https://www.wjnet.com/bpg/gerinfo/240
PUBLICATION MISCONDUCT https://www.wjnet.com/bpg/gerinfo/208
ARTICLE PROCESSING CHARGE https://www.wjnet.com/bpg/gerinfo/242
STEPS FOR SUBMITTING MANUSCRIPTS https://www.wjnet.com/bpg/GerInfo/239
ONLINE SUBMISSION https://www.f6publishing.com

Erdheim-Chester disease with asymmetric talus involvement: A case report

Qin Xia, Cheng Tao, Ke-Wei Zhu, Wei-Ye Zhong, Peng-Lin Li, Yi Jiang, Min-Zhi Mao

ORCID number: Qin Xia (0000-0002-5578-9438); Cheng Tao (0000-0002-2635-2691); Ke-Wei Zhu (0000-0002-1955-5116); Wei-Ye Zhong (0000-0002-3739-642X); Peng-Lin Li (0000-0002-1607-1968); Yi Jiang (0000-0002-6773-4338); Min-Zhi Mao (0000-0002-0392-1652).

Author contributions: Mao MZ designed the case report and drafted the manuscript; Xia Q and Li PL analyzed the patient data and reviewed the literature; Tao C and Zhu KW provided treatment advice and performed the surgery; Jiang Y and Zhong WY performed the pathological and immunohistochemical examinations.

Informed consent statement: Informed written consent was obtained from the patient for publication of this report and any accompanying images.

Conflict-of-interest statement: The authors declare that they have no conflict of interest.

CARE Checklist (2016) statement: The authors have read the CARE Checklist (2016), and the manuscript was prepared and revised according to the CARE Checklist (2016).

Open-Access: This article is an open-access article that was selected by an in-house editor and fully peer-reviewed by external reviewers. It is distributed in accordance with the Creative Commons Attribution NonCommercial (CC BY-NC 4.0) license, which permits others to distribute, remix, adapt, build

Qin Xia, Cheng Tao, Ke-Wei Zhu, Wei-Ye Zhong, Peng-Lin Li, Min-Zhi Mao, Department of Orthopedics, The Second Xiangya Hospital, Central South University, Changsha 410011, Hunan Province, China

Yi Jiang, Department of Pathology, The Second Xiangya Hospital, Central South University, Changsha 410011, Hunan Province, China

Corresponding author: Min-Zhi Mao, PhD, Doctor, Department of Orthopedics, The Second Xiangya Hospital, Central South University, No. 139 Middle Renmin Road, Changsha 410011, Hunan Province, China. maominzhi@csu.edu.cn

Abstract

BACKGROUND

Erdheim-Chester disease (ECD) is a rare multi-system or multi-organ histiocytic proliferative disease with diverse clinical manifestations, and the development of the disease is complex, which makes clinical diagnosis and treatment difficult. The characteristic clinical manifestations include multi-organ involvement, especially in the symmetrical diaphysis and metaphysis of the bilateral extremities. ECD with a unilateral talus lesion is extremely rare. Here, we report an unusual case of ECD invading the asymmetric talus and tibia without involving other organs. The patient had good outcome after surgery.

CASE SUMMARY

We report a case of a 67-year-old man who was referred to our outpatient department because of left ankle chronic pain for 5 years, which exacerbated after a foot sprain 6 mo previously. We discovered multiple sclerotic lesions of the tibia and talus on his previous X-ray films, which were initially missed in a local hospital. Therefore, enhanced computer computed tomography (CT) and magnetic resonance imaging were performed. These examinations showed multiple lesions in the bone marrow cavity of the left tibia, and cortical sclerosis and osteonecrosis of the left talus. Specimens were collected *via* bone puncture from the two lesions, and a final diagnosis of ECD was confirmed by pathological and immunohistochemical examinations. In addition, other auxiliary examinations including head CT, pulmonary CT, spinal CT, abdominal CT, cardiac ultrasound and thyroid ultrasound showed no obvious abnormalities. The patient underwent surgery for the tibia lesion scraping and talus lesion scraping combined with cement casting. The patient started on a progressive rehabilitation at 4 wk, and felt no pain after surgery. During a 2-year follow-up period, the patient exercised normally without pain, and there were no signs of recurrence.

upon this work non-commercially, and license their derivative works on different terms, provided the original work is properly cited and the use is non-commercial. See: <http://creativecommons.org/licenses/by-nc/4.0/>

Manuscript source: Unsolicited manuscript

Received: November 21, 2019

Peer-review started: November 21, 2019

First decision: December 23, 2019

Revised: December 25, 2019

Accepted: January 2, 2020

Article in press: January 2, 2020

Published online: February 6, 2020

P-Reviewer: Alimehmeti RH,

Ghoch ME, Lee SH

S-Editor: Zhang L

L-Editor: Filipodia

E-Editor: Liu MY



CONCLUSION

This study shows that surgery treatment may also achieve good results for ECD patients with only bone involvement.

Key words: Erdheim-Chester disease; Talus; Tibia; Osteosclerosis; Surgery; Case report

©The Author(s) 2020. Published by Baishideng Publishing Group Inc. All rights reserved.

Core tip: Erdheim-Chester disease (ECD) is a subtype of non-Langerhans's cell histiocytosis, for which diagnosis is difficult and no treatment guidelines have been available. In addition, ECD is a systemic disease with multi-organ involvement, and especially affects the symmetrical diaphysis and metaphysis of the bilateral extremities. We report the first case of ECD invading the unilateral tibia and talus without involvement of any other organs. The patient was treated with surgery, and had a good prognosis, which added evidence to the diagnosis and treatment of this disease.

Citation: Xia Q, Tao C, Zhu KW, Zhong WY, Li PL, Jiang Y, Mao MZ. Erdheim-Chester disease with asymmetric talus involvement: A case report. *World J Clin Cases* 2020; 8(3): 614-623

URL: <https://www.wjnet.com/2307-8960/full/v8/i3/614.htm>

DOI: <https://dx.doi.org/10.12998/wjcc.v8.i3.614>

INTRODUCTION

Erdheim-Chester disease (ECD) is a subtype of non-Langerhans's cell histiocytosis with multi-system invasion and poor prognosis, which is now thought to be an inflammatory histiocytic disease caused by oncogene mutations^[1-5]. Studies have shown that almost half of the patients with ECD have the BRAF^{V600E} mutation, and this oncogene mutation activates the mitogen-activated protein kinase pathway, which is involved in the histiocytosis pathogenesis of inflammation and fibrosis^[3,4,6,7]. In addition, typical histopathology revealed activated fibro-inflammatory histiocytes and Touton giant cells. This disease mainly occurs in adults between the fourth and seventh decades of life, with a slight male predominance^[6,8].

Due to its multiple clinical manifestations and lack of specificity, diagnosis of ECD is difficult. ECD can potentially occur in all organs of the body with multi-system involvement, most commonly affecting bones, the central nervous system, cardiovascular system, lungs and retroperitoneal cavities^[9]. Bone pain is the most common clinical presentation, and other frequent extra-skeletal presentations include periaortic infiltration as "coated aorta" in the cardiovascular system, and perinephric fat infiltration as "hairy kidney" in the urinary system. Therefore, it may become a fatal illness when extensive central nervous, pulmonary and other systems are affected^[10].

The characteristic X-ray presentation of ECD is the symmetrical osteosclerosis of the extremities, and generally on the diaphysis of appendicular long bones. In addition to typical osteosclerosis, it may also be associated with partial epiphysis sclerosis, periostitis, and even bone infarction^[11]. The disease rarely involves only bone or unilateral limbs, and most patients with bone involvement are accompanied by extra-bone lesions. In addition, asymmetric talus and tibia involvement without other organs affected has not been reported in the literature.

There are few prospective studies or randomized controlled clinical studies on ECD, so the treatments are mainly based on case reports or empirical summaries, and there is still a lack of high-level evidence. In general, except for a small number of asymptomatic patients, all patients should start treatment immediately after the disease is found rather than observe, and many therapies have been used for this disease. Currently, through in-depth studies of the pathogenesis of ECD, treatments such as interferon- α recommended as the first-line therapy, anakinra (IL-1 receptor antagonist), vemurafenib (specific BCRF inhibitor) and cobimetinib (specific MEK inhibitor) have been proven to have various efficacies^[4,12-18]. Besides, surgical treatment can also achieve good results^[19-21]. However, there is still a lack of standard guidelines for the surgical treatment of focal bone lesions with ECD involvement. We aim to share our experience in managing a case of ECD with asymmetric bone involvement,

and suggest the feasibility of surgery.

CASE PRESENTATION

Chief complaints

A 67-year-old man complained of left ankle pain with slight limitation of motion.

History of present illness

The patient had a 5-year history of left ankle chronic pain, which exacerbated after a foot sprain 6 mo previously. The patient suffered from pain in his left ankle during walking. He noted some swelling in the left medial malleolus region but no fever, chills or loss of weight.

History of past illness

His past history was unremarkable.

Personal and family history

His family history was unremarkable.

Physical examination upon admission

Physical examination showed swelling in his left medial malleolus region with tenderness. He presented with pain and weakness in eversion.

Laboratory examinations

The laboratory tests showed a white blood cell count of $6.09 \times 10^9/L$, C-reactive protein of 1.64 mg/L and erythrocyte sedimentation rate of 2.00 mm/h. Biopsies were obtained from the affected tibia and talus for pathological and immunohistochemical examinations. Microscopy revealed foamy histiocytes filled with lipids and granulomatous inflammation containing fibrous tissue, lymphoid tissue, plasma cells and Touton giant cells. Immunohistochemical examination showed positive expression of CD68 and AACT, but negative expression of CD34, S100 and CD1a (Figure 1). In addition, molecular pathologic examination was negative for the BRAF^{V600E} mutation.

Imaging examinations

We discovered osteosclerosis of the talus and tibia on the previous plain X-ray film taken by the patient 5 years ago, which was initially missed at a local hospital (Figure 2). His recent left tibiofibular X-ray showed irregular osteolytic lesions in the talus, and focal osteosclerosis in the distal tibia (Figure 3). The computer tomography (CT) revealed left talus osteolytic destruction and cortical discontinuity of the anterior articular surface (Figure 4). Magnetic resonance imaging (MRI) revealed lesions of the talus and tibia hypointense on T1-weighted images (T1WI) and hyperintense on T2-weighted images (T2WI) (Figures 5 and 6). The whole-body technetium bone scan showed increased asymmetric uptake at the left talus and tibia (Figure 7).

FINAL DIAGNOSIS

Erdheim-Chester disease with a wild-type mutation in the BRAF^{V600E} gene.

TREATMENT

Based on the X-ray findings over a 5-year period, the patient was found to have relatively slow progress of the disease, and the other organs were not involved. Moreover, side effects of IFN and other drugs were frequently reported, so surgery was performed after consent was obtained from the patient. The patient was placed in a supine position with a thigh tourniquet. An anterior medial incision was made to avoid osteotomy of the medial malleolus. Intraoperatively, synovial hyperplasia was found in the ankle joint. A tawny and soft osteochondral lesion without obvious envelope was found in the neck and body of the talus. Complete resection of the astragalar lesion was performed, and specimens were sent for pathological examination. As there was a large area of defective bones in the internal side of the talus, combined with a small area of defective cartilage, bone cement was cast into this defective area without extending beyond the borders and damaging the surrounding tissues. A small incision was made in the distal tibia, and the diseased tissue in the

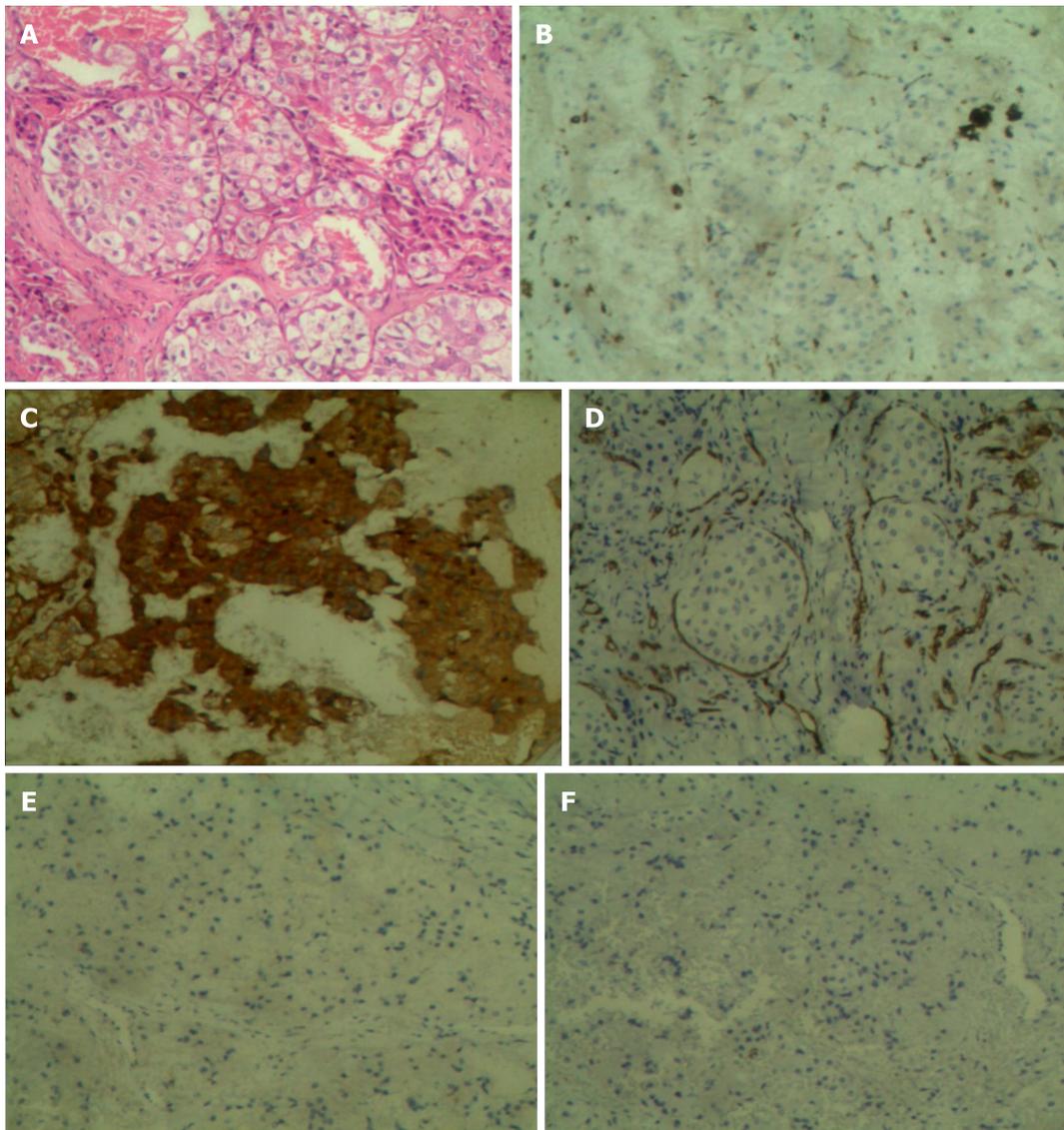


Figure 1 Histopathological findings of the lesions (100 ×). A: Hematoxylin-eosin stains of the specimen; B: Immunohistochemistry showing positive staining for CD68; C: Immunohistochemistry showing positive staining for ACTT; D: Immunohistochemistry showing negative staining for CD34; E: Immunohistochemistry showing negative staining for S100; F: Immunohistochemistry showing negative staining for CD1a.

medullary cavity of the tibia was completely scraped away. After sufficient hemostasis, the incision was covered with a dressing.

OUTCOME AND FOLLOW-UP

Close follow-up was conducted after operation. At 1 mo after surgery, the patient was examined in our outpatient department. No postoperative incision infections or complications were found. At 6 mo post-operatively, the patient could walk normally without pain or limited mobility, and CT showed that the lesion in the talus was completely removed and the bone was filled with cement (Figure 8). During a 2-year follow-up period after the surgery, X-ray and whole-body bone scans showed no abnormalities, and no evidence of recurrence or metastasis was recorded (Figures 7 and 9).

DISCUSSION

The diagnosis of ECD is based on characteristic pathological findings, clinical manifestations and unique imaging features^[5,9]. The most common pathological manifestation is diffuse sclerosis characterized by foam-like adipose tissue infiltration



Figure 2 Plain X-ray film taken 5 years ago showed slight osteosclerosis in the tibia and the talus (white arrow).

and granulomatous inflammation containing fibrous tissue, lymphoid tissue, plasma cells and Touton giant cells. Histologically, CD68(+), CD34(-), CD1a(-) and S100(-) were observed in immunostaining^[22]. The clinical manifestations range from mild asymptomatic focal lesions to severe life-threatening multiple organ dysfunction syndrome, which are especially common in the diaphysis and metaphysis of long bones^[23]. Affected extra-osseous organs include cardiovascular, respiratory, central and neuroendocrine, orbital, retroperitoneal and other rare sites^[22].

In recent years, with the deepening of imaging research, diagnosis, evaluation, monitoring and follow-up of ECD become increasingly important. X-ray is the primary method of examining changes in bone structure; CT, MRI and radionuclide bone scans are more sensitive to early and hidden lesions. The unique imaginary feature is the symmetrical osteosclerosis of the long bone, and it may also be associated with partial epiphysis sclerosis, periostitis, and even bone infarction^[11]. MRI is highly sensitive to bone marrow infiltration, which shows that normal intramedullary fat is replaced by abnormal hyperplasia tissue. In addition to assessing the extent of bone marrow infiltration, MRI can clearly show changes in the periosteum and cartilage.

ECD affecting the unilateral talus is rare, and may be easily missed in diagnosis. Up to date, there are only about 1000 cases reported in the literature. The disease may be ignored by inexperienced clinicians, leading to delayed diagnosis. In general, ECD can occur in any site of the body, but almost all patients have symmetrical osteosclerosis of the limb long bone, such as the femur, tibia and fibula. Our patient only had unilateral talus and tibia involvement, while no lesions were found in the rest of the body. According to the literature review, all the affected bones are above the ankle plate. Epiphysis, articular cartilage and talus involvement has been scarcely reported previously^[9-11,22]. Anatomically, there is no muscular attachment to the talus, all the bones are surrounded by the articular surface of cartilage, and there is also cartilage continuation at the edge of the upper articular surface; in addition, the main blood supply comes from the dorsal foot artery, which passes into the body from the talus anterolateral neck^[24]. In the case of trauma, destruction of the talus is likely to occur due to cartilage damage or ischemic necrosis caused by blood supply interruption. Extensive synovial hyperplasia, articular cartilage damage and severe bone destruction of the affected talus were discovered during surgery. Upon reaching this point, we are faced with two possibilities: One would be the disease itself, which exacerbates bone and cartilage destruction. Considering that the patient had a history of trauma, the other possibility is that trauma as an inducement would lead to bone destruction and articular cartilage damage of the affected talus, which further exacerbates the deterioration of ECD.

To date, many different drugs for ECD have been explored, but few prospective studies or randomized controlled clinical studies are being conducted. Currently, the best ECD treatment is IFN- α and PEG-IFN- α therapy. A prospective, non-randomized, observational cohort analysis of 53 ECD patients revealed that 46 patients with ECD who received IFN- α - or PEG-IFN- α -based treatment had increased overall survival^[12]. Numerous adverse events such as fever, fatigue, gastrointestinal symptoms and depression were observed. Anakinra has also been available as an IL-1 receptor antagonist^[15,16]. In addition, based on the pathogenesis that almost half ECD patients have the BRAF^{V600E} mutation and almost all ECD tissue samples are active in extracellular signal-regulated kinase phosphorylation, the use of vemurafenib as a specific BRAF inhibitor and cobimetinib as a specific MEK inhibitor resulted in good



Figure 3 Plain anteroposterior radiography of the tibiofibula indicated osteosclerosis in the tibia and osteolytic lesions in the talus (arrow).

therapeutic effects as well^[13,14,25]. However, the most frequent side effects with vemurafenib or cobimetinib were skin complications, gastrointestinal symptoms and rhabdomyolysis. There are very few studies on the surgical treatment of ECD. Alfieri *et al*^[19] have confirmed that surgical resection and radiation therapy can be a good treatment choice for cerebral lesions; Wimpissinger *et al*^[20] have reported that renal complications in ECD should be considered as part of the surgical strategy; and Mahoozi *et al*^[21] have concluded that surgical treatment is an option in the management of isolated cardiac ECD. Here, we describe a unique case of ECD that only affected the focal talus and tibia, but no other organs. The lesions were completely removed by surgery, and there was no recurrence or metastasis during the 2-year follow-up period.

CONCLUSION

Although different drug treatments for ECD have been reported, surgery for bone involvement alone should also be considered to avoid side effects caused by drugs. Given its rarity, further research on ECD, as well as a comparison of the clinical efficacy of surgery and medication, will undoubtedly be vital and difficult.



Figure 4 Computed tomography clearly suggested the left talus osteolytic destruction and cortical discontinuity of the anterior articular surface (white arrow).



Figure 5 Magnetic resonance imaging showed osteolytic lesions in the talus hypointense on T1WI and hyperintense on T2WI (white arrow).

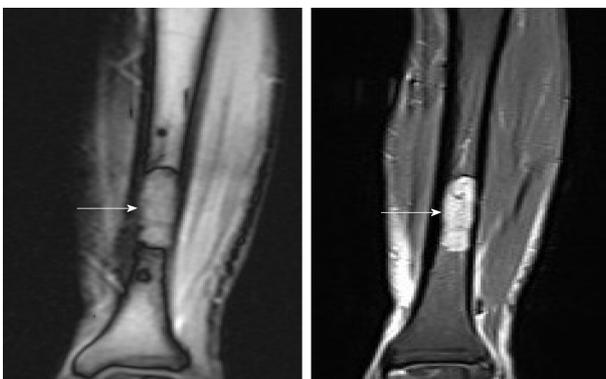


Figure 6 Magnetic resonance imaging suggested a marrow-replacing infiltrative lesion of the tibia hypointense on T1WI and hyperintense on T2WI.

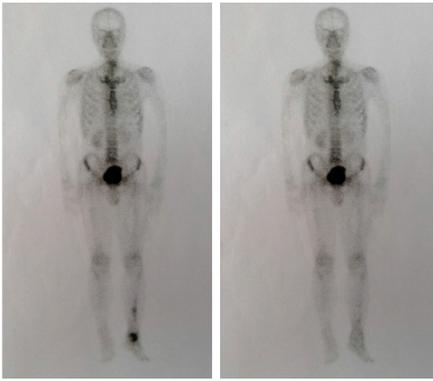


Figure 7 Comparison of preoperative and postoperative whole-body bone scans.

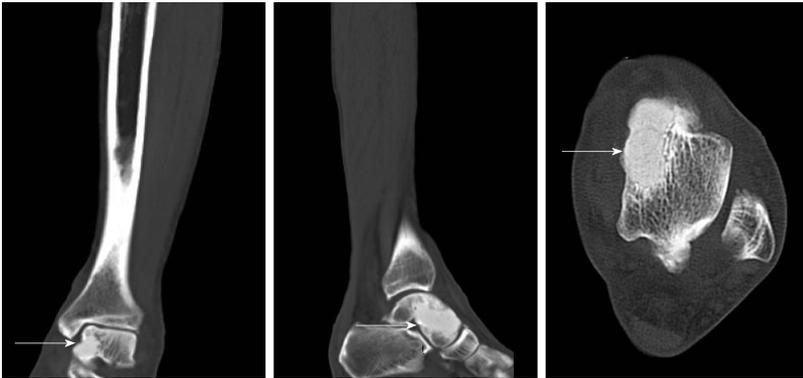


Figure 8 Computed tomography showed that lesions in the talus were completely removed and the bone was filled with cement casting.

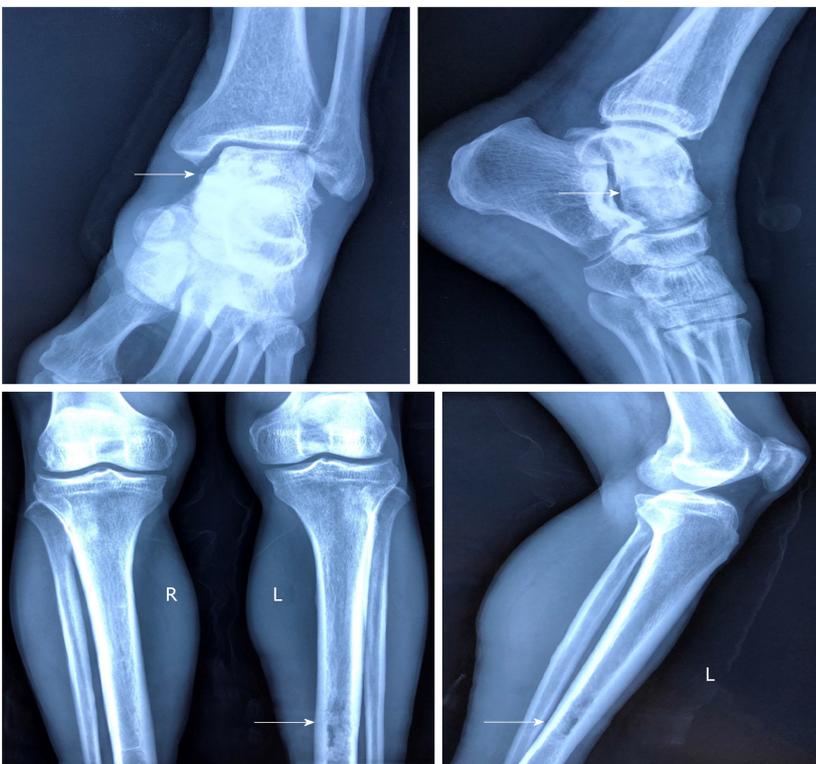


Figure 9 Postoperative plain X-rays indicated complete lesion removal and no signs of recurrence.

REFERENCES

- 1 **Badalian-Very G**, Vergilio JA, Degar BA, MacConaill LE, Brandner B, Calicchio ML, Kuo FC, Ligon AH, Stevenson KE, Kehoe SM, Garraway LA, Hahn WC, Meyerson M, Fleming MD, Rollins BJ. Recurrent BRAF mutations in Langerhans cell histiocytosis. *Blood* 2010; **116**: 1919-1923 [PMID: 20519626 DOI: 10.1182/blood-2010-04-279083]
- 2 **Chapman PB**, Hauschild A, Robert C, Haanen JB, Ascierto P, Larkin J, Dummer R, Garbe C, Testori A, Maio M, Hogg D, Lorigan P, Lebbe C, Jouary T, Schadendorf D, Ribas A, O'Day SJ, Sosman JA, Kirkwood JM, Eggermont AM, Dreno B, Nolop K, Li J, Nelson B, Hou J, Lee RJ, Flaherty KT, McArthur GA; BRIM-3 Study Group. Improved survival with vemurafenib in melanoma with BRAF V600E mutation. *N Engl J Med* 2011; **364**: 2507-2516 [PMID: 21639808 DOI: 10.1056/NEJMoa1103782]
- 3 **Haroche J**, Charlotte F, Arnaud L, von Deimling A, Hélias-Rodzewicz Z, Hervier B, Cohen-Aubart F, Launay D, Lesot A, Mokhtari K, Canioni D, Galmiche L, Rose C, Schmalzing M, Croockewit S, Kambouchner M, Copin MC, Fraïtag S, Sahn F, Brousse N, Amoura Z, Donadieu J, Emile JF. High prevalence of BRAF V600E mutations in Erdheim-Chester disease but not in other non-Langerhans cell histiocytoses. *Blood* 2012; **120**: 2700-2703 [PMID: 22879539 DOI: 10.1182/blood-2012-05-430140]
- 4 **Cohen-Aubart F**, Emile JF, Carrat F, Hélias-Rodzewicz Z, Taly V, Charlotte F, Cluzel P, Donadieu J, Idhah A, Barete S, Amoura Z, Haroche J. Phenotypes and survival in Erdheim-Chester disease: Results from a 165-patient cohort. *Am J Hematol* 2018; **93**: E114-E117 [PMID: 29396850 DOI: 10.1002/ajh.25055]
- 5 **Papo M**, Cohen-Aubart F, Trefond L, Bauvois A, Amoura Z, Emile JF, Haroche J. Systemic Histiocytosis (Langerhans Cell Histiocytosis, Erdheim-Chester Disease, Destombes-Rosai-Dorfman Disease): from Oncogenic Mutations to Inflammatory Disorders. *Curr Oncol Rep* 2019; **21**: 62 [PMID: 31115724 DOI: 10.1007/s11912-019-0810-6]
- 6 **Davies H**, Bignell GR, Cox C, Stephens P, Edkins S, Clegg S, Teague J, Woffendin H, Garnett MJ, Bottomley W, Davis N, Dicks E, Ewing R, Floyd Y, Gray K, Hall S, Hawes R, Hughes J, Kosmidou V, Menzies A, Mould C, Parker A, Stevens C, Watt S, Hooper S, Wilson R, Jayatilake H, Gusterson BA, Cooper C, Shipley J, Hargrave D, Pritchard-Jones K, Maitland N, Chenevix-Trench G, Riggins GJ, Bigner DD, Palmieri G, Cossu A, Flanagan A, Nicholson A, Ho JW, Leung SY, Yuen ST, Weber BL, Seigler HF, Darrow TL, Paterson H, Marais R, Marshall CJ, Wooster R, Stratton MR, Futreal PA. Mutations of the BRAF gene in human cancer. *Nature* 2002; **417**: 949-954 [PMID: 12068308 DOI: 10.1038/nature00766]
- 7 **Hervier B**, Haroche J, Arnaud L, Charlotte F, Donadieu J, Néel A, Lifermann F, Villabona C, Graffin B, Hermine O, Rigolet A, Roubille C, Hachulla E, Carmoi T, Bézier M, Meignin V, Conrad M, Marie L, Kostrzewa E, Michot JM, Barete S, Taly V, Cury K, Emile JF, Amoura Z; French Histiocytoses Study Group. Association of both Langerhans cell histiocytosis and Erdheim-Chester disease linked to the BRAFV600E mutation. *Blood* 2014; **124**: 1119-1126 [PMID: 24894769 DOI: 10.1182/blood-2013-12-543793]
- 8 **Nadjiri J**, Woertler K, Specht K, Harrasser N, Toepfer A. Erdheim-Chester disease with bilateral Achilles tendon involvement. *Skeletal Radiol* 2016; **45**: 1437-1442 [PMID: 27506209 DOI: 10.1007/s00256-016-2447-y]
- 9 **Munoz J**, Janku F, Cohen PR, Kurzrock R. Erdheim-Chester disease: characteristics and management. *Mayo Clin Proc* 2014; **89**: 985-996 [PMID: 24814521 DOI: 10.1016/j.mayocp.2014.01.023]
- 10 **Haroche J**, Amoura Z, Dion E, Wechsler B, Costedoat-Chalumeau N, Cacoub P, Isnard R, Gènereau T, Wechsler J, Weber N, Graef C, Cluzel P, Grenier P, Piette JC. Cardiovascular involvement, an overlooked feature of Erdheim-Chester disease: report of 6 new cases and a literature review. *Medicine (Baltimore)* 2004; **83**: 371-392 [PMID: 15525849 DOI: 10.1097/01.md.0000145368.17934.91]
- 11 **Perez A**, Crahes M, Laquerrière A, Proust F, Derrey S. Neurological form of Erdheim-Chester disease: Case report and review of the literature. *Neurochirurgie* 2014; **60**: 316-320 [PMID: 25441710 DOI: 10.1016/j.neuchi.2014.06.012]
- 12 **Arnaud L**, Hervier B, Néel A, Hamidou MA, Kahn JE, Wechsler B, Pérez-Pastor G, Blomberg B, Fuzibet JG, Dubourguet F, Marinho A, Magnette C, Noel V, Pavic M, Casper J, Beucher AB, Costedoat-Chalumeau N, Aaron L, Salvatierra J, Graux C, Cacoub P, Delcey V, Dechant C, Bindi P, Herbat C, Graziani G, Amoura Z, Haroche J. CNS involvement and treatment with interferon- α are independent prognostic factors in Erdheim-Chester disease: a multicenter survival analysis of 53 patients. *Blood* 2011; **117**: 2778-2782 [PMID: 21239701 DOI: 10.1182/blood-2010-06-294108]
- 13 **Haroche J**, Cohen-Aubart F, Emile JF, Maksud P, Drier A, Tolédano D, Barete S, Charlotte F, Cluzel P, Donadieu J, Benameur N, Grenier PA, Besnard S, Ory JP, Lifermann F, Idhah A, Granel B, Graffin B, Hervier B, Arnaud L, Amoura Z. Reproducible and sustained efficacy of targeted therapy with vemurafenib in patients with BRAF(V600E)-mutated Erdheim-Chester disease. *J Clin Oncol* 2015; **33**: 411-418 [PMID: 25422482 DOI: 10.1200/JCO.2014.57.1950]
- 14 **Diamond EL**, Subbiah V, Lockhart AC, Blay JY, Puzanov I, Chau I, Raju NS, Wolf J, Erinjeri JP, Torrisi J, Lacouture M, Elez E, Martínez-Valle F, Durham B, Arcila ME, Ulaner G, Abdel-Wahab O, Pitcher B, Makrutzki M, Riehl T, Baselga J, Hyman DM. Vemurafenib for BRAF V600-Mutant Erdheim-Chester Disease and Langerhans Cell Histiocytosis: Analysis of Data From the Histology-Independent, Phase 2, Open-label VE-BASKET Study. *JAMA Oncol* 2018; **4**: 384-388 [PMID: 29188284 DOI: 10.1001/jamaoncol.2017.5029]
- 15 **Aouba A**, Geogin-Lavialle S, Pagnoux C, Martin Silva N, Renand A, Galateau-Salle F, Le Toquin S, Bensadoun H, Larousserie F, Silvera S, Provost N, Candon S, Seror R, de Menthon M, Hermine O, Guillemin L, Bienvenu B. Rationale and efficacy of interleukin-1 targeting in Erdheim-Chester disease. *Blood* 2010; **116**: 4070-4076 [PMID: 20724540 DOI: 10.1182/blood-2010-04-279240]
- 16 **Tran TA**, Pariente D, Lecron JC, Delwail A, Taoufik Y, Meinzer U. Treatment of pediatric Erdheim-Chester disease with interleukin-1-targeting drugs. *Arthritis Rheum* 2011; **63**: 4031-4032 [PMID: 21898344 DOI: 10.1002/art.30638]
- 17 **Cohen Aubart F**, Emile JF, Maksud P, Galanaud D, Cluzel P, Benameur N, Aumaitre O, Amoura Z, Haroche J. Efficacy of the MEK inhibitor cobimetinib for wild-type BRAF Erdheim-Chester disease. *Br J Haematol* 2018; **180**: 150-153 [PMID: 27711968 DOI: 10.1111/bjh.14284]
- 18 **Al Bayati A**, Plate T, Al Bayati M, Yan Y, Lavi ES, Rosenblatt JD. Dabrafenib and Trametinib Treatment for Erdheim-Chester Disease with Brain Stem Involvement. *Mayo Clin Proc Innov Qual Outcomes* 2018; **2**: 303-308 [PMID: 30225465 DOI: 10.1016/j.mayocpiqo.2018.05.001]
- 19 **Alfieri A**, Gazeri R, Galarza M, Neroni M. Surgical treatment of intracranial Erdheim-Chester disease. *J Clin Neurosci* 2010; **17**: 1489-1492 [PMID: 20843693 DOI: 10.1016/j.jocn.2010.03.062]

- 20 **Wimpissinger TF**, Schernthaner G, Feichtinger H, Stackl W. Compression of kidneys in Erdheim-Chester disease of retroperitoneum: Open surgical approach. *Urology* 2005; **65**: 798 [PMID: [15833540](#) DOI: [10.1016/j.urology.2004.10.051](#)]
- 21 **Mahoozi HR**, Zittermann A, Hakim Meibodi K, Burchert W, Gummert JF, Mirow N. Erdheim-Chester disease in a female cardiac surgery patient. *Thorac Cardiovasc Surg* 2012; **60**: 428-430 [PMID: [21594819](#) DOI: [10.1055/s-0030-1271017](#)]
- 22 **Campochiaro C**, Tomelleri A, Cavalli G, Berti A, Dagna L. Erdheim-Chester disease. *Eur J Intern Med* 2015; **26**: 223-229 [PMID: [25865950](#) DOI: [10.1016/j.ejim.2015.03.004](#)]
- 23 **Carpinteri R**, Patelli I, Casanueva FF, Giustina A. Pituitary tumours: inflammatory and granulomatous expansive lesions of the pituitary. *Best Pract Res Clin Endocrinol Metab* 2009; **23**: 639-650 [PMID: [19945028](#) DOI: [10.1016/j.beem.2009.05.009](#)]
- 24 **Trovato A**, El-Rich M, Adeeb S, Dhillon S, Jomha N. Geometric analysis of the talus and development of a generic talar prosthetic. *Foot Ankle Surg* 2017; **23**: 89-94 [PMID: [28578800](#) DOI: [10.1016/j.fas.2016.12.002](#)]
- 25 **Nelson DS**, Quispel W, Badalian-Very G, van Halteren AG, van den Bos C, Bovée JV, Tian SY, Van Hummelen P, Ducar M, MacConaill LE, Egeler RM, Rollins BJ. Somatic activating ARAF mutations in Langerhans cell histiocytosis. *Blood* 2014; **123**: 3152-3155 [PMID: [24652991](#) DOI: [10.1182/blood-2013-06-511139](#)]



Published By Baishideng Publishing Group Inc
7041 Koll Center Parkway, Suite 160, Pleasanton, CA 94566, USA
Telephone: +1-925-3991568
E-mail: bpgoffice@wjgnet.com
Help Desk: <https://www.f6publishing.com/helpdesk>
<https://www.wjgnet.com>

