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CASE REPORT

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A patient with Eradheim-Chester disease presenting with progressive cystic lung lesions and confirmed pulmonary artery hypertension: a case report

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Abstract

Background Erdheim-Chester disease (ECD), a rare type of non-Langerhans cell histiocytosis, was classified as a haematopoietic tumour by the World Health Organization (WHO) in 2016. It involves multiple systems and is challenging to diagnose due to its broad spectrum of clinical manifestations. The pulmonary manifestations of ECD lack specificity. We present a case of ECD with pronounced cystic lung abnormalities to increase awareness of the disease among pulmonologists and expedite diagnosis and treatment.

Case presentation We report the case of a 44-year-old male who presented with intermittent fever, cough, bilateral leg pain, extensive xanthomas on his face, and extensive pulmonary cystic changes noted on imaging following a pulmonary stab wound incident. Thoracoabdominal enhanced computed tomography (CT) revealed progressive cystic changes in the lungs, notably in the upper lungs and subpleural areas; thickened interlobular septa; circumferential wall thickening of the left subclavian artery; uneven thickening of the aortic wall; and soft tissue shadows in the right atrium of the heart. Bone scintigraphy revealed bilateral symmetric long-bone uptake. Despite his advanced lung abnormalities, he exhibited no hypoxia. Notably, echocardiography indicated severe pulmonary artery hypertension, and right heart catheterization confirmed increased mean pulmonary artery pressure at 37 mmHg and elevated pulmonary vascular resistance. Pathology examination of transbronchial lung biopsy and the facial xanthomas confirmed the presence of ECD-characteristic histiocytes, and genetic testing revealed a BRAF V600E mutation. Treatment with dabrafenib improved respiratory symptoms and facial xanthomas, although some symptoms persisted. Follow-up CT showed reduced interstitial lesions but more pronounced cystic changes.

Conclusions This case of ECD illustrates rare pulmonary cystic changes alongside pulmonary arterial hypertension, challenging typical presentations of ECD. This is the first documented instance of pulmonary hypertension associated with ECD, broadening the understanding of its potential complications. These findings emphasize the need for considering ECD in the differential diagnosis of atypical cystic lung lesions, especially when accompanied by systemic symptoms such as xanthomas and bone pain.

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Keywords Erdheim-Chester disease, Pulmonary cystic disease, Pulmonary arterial hypertension

Background

Erdheim-Chester disease (ECD) is a rare, multisystemic non-Langerhans cell histiocytosis syndrome. Since Erdheim and Chester first reported ECD in 1930, there have been more than 1,500 cases documented in the medical literature [1]. With advances in molecular research, over 80% of patients with ECD exhibit activation of the mitogen-activated protein kinase (MAPK) pathway, primarily due to the BRAF-V600E mutation [1]. This led to the classification of ECD as a hematopoietic tumor in the 2016 World Health Organization (WHO) disease categorization [2]. Tissue biopsy is required for diagnosing ECD, but must be interpreted alongside clinical, radiographic, and recently, molecular findings. Typical immunohistochemical pattern of ECD is positive for the markers CD68, CD14, and CD163 and negative for CD1a and CD207 [3], which are positive in Langerhans cell histiocytosis (LCH). It is currently advised for all patients to have BRAF-V600E mutation testing performed on biopsy specimens to ascertain eligibility for first-line treatment with BRAF inhibitors such as vemurafenib or dabrafenib [2] in pursuit of enhanced treatment efficacy [4, 5].

The organs involved in ECD are extensive, including the long bones in approximately 90% of patients, as well as the lungs, kidneys, heart, skin, and brain, among others. Moreover, many patients do not exhibit overt clinical symptoms, resulting in a wide spectrum of clinical manifestations that significantly complicate the diagnosis of the disease. ECD is characterized by specific radiological features in various affected organs, such as bilateral symmetric long-bone osteosclerosis, a "coated aorta", and a "hairy kidney". However, in terms of pulmonary lesions, computed tomography (CT) findings lack distinctiveness compared to those of other interstitial lung diseases, often manifesting as interlobular septal thickening, ground-glass opacities, or pulmonary nodules [6]. We present a case of ECD with pronounced cystic lung abnormalities, intended to increase awareness of the disease among pulmonologists and to expedite the diagnostic and treatment process.

Case presentation

A 44-year-old male with no history of smoking presented with intermittent fevers and cough a year and a half ago after pulmonary stab wound. His fever, characterized by daily afternoon temperature peaks of 40 °C, was unresponsive to antibiotic therapy but subsided spontaneously six months later without treatment. Upon visiting our outpatient department one year ago, blood tests were roughly normal, but the hsCRP level and ESR were significantly elevated. Blood and bronchoalveolar lavage fluid (BALF) cultures, including mNGS, were negative for pathogens. CT scans of the thorax revealed ground-glass opacities in the lungs, thickened interlobular septa and pleural thickening (Fig. 1A). Pulmonary function tests revealed an forced expiratory volume in the first second (FEV1) of 2.25 L (63%), an FEV1/ forced vital capacity (FVC) ratio of 80.95%, a total lung capacity (TLC) of 67%, and a diffusing capacity for carbon monoxide (DLCO-SB) of 42%, indicating restrictive ventilatory dysfunction with reduced diffusion capacity and a negative bronchodilator test. Concurrently, the patient developed extensive xanthomas on his face. Recently, the patient was admitted to the hospital again due to an exacerbation of cough and new bilateral knee pain. Repeated chest CT revealed worsening bilateral lung lesions with multiple thin-walled cysts, predominantly in the upper lobes and subpleural areas (Fig. 1B). Despite these findings, the patient exhibited no symptoms of hypoxia and maintained a pulse oximetry reading of 100% in ambient air. The patient's chest CT demonstrated multiple pulmonary cystic lesions, accompanied by ground-glass nodules. From the differential diagnosis of pulmonary cystic disease, our initial clinical suspicion was pulmonary Langerhans cell histiocytosis (PLCH), followed by rare infections such as Pneumocystis jirovecii pneumonia (PCP) and Talaromyces marneffei infection. Given the patient's male gender, absence of skin or renal tumors, and lack of relevant family history, the likelihood of lymphangioleiomyomatosis (LAM) or Birt-Hogg-Dubé Syndrome (BHD) was deemed low. To exclude potential infection, bronchoscopy with repeat bronchoalveolar lavage was performed, yielding negative results for pathogenic organisms. A bone scan was subsequently conducted to evaluate LCH-related osseous involvement. 99mTc- Methylene Diphosphonate (MDP) single-photon emission computed tomography (SPECT)/CT revealed increased radioactive uptake in the nasal bones, maxilla and bilateral symmetric long bones of the limbs (Fig. 2A), indicating a potential diagnosis of ECD. 18 F- Fluorodeoxyglucose (FDG) positron emission tomography (PET)/CT also revealed bilateral maxillary sinus mucosal thickening with diffusely increased metabolism, nodular thickening of the mesentery with mildly increased metabolism, and multiple areas of increased metabolism in the long bones of the limbs (Fig. 2B). Further thoracoabdominal enhanced CT revealed circumferential wall thickening and lumen narrowing at the root of the left subclavian artery (Fig. 2C), uneven thickening of the aortic wall (Fig. 2D), soft tissue shadows in the right atrioventricular groove (Fig. 2E), an anterior descending branch area of the heart (Fig. 2F) and multiple soft tissue density nodules in the abdomen, some with significant

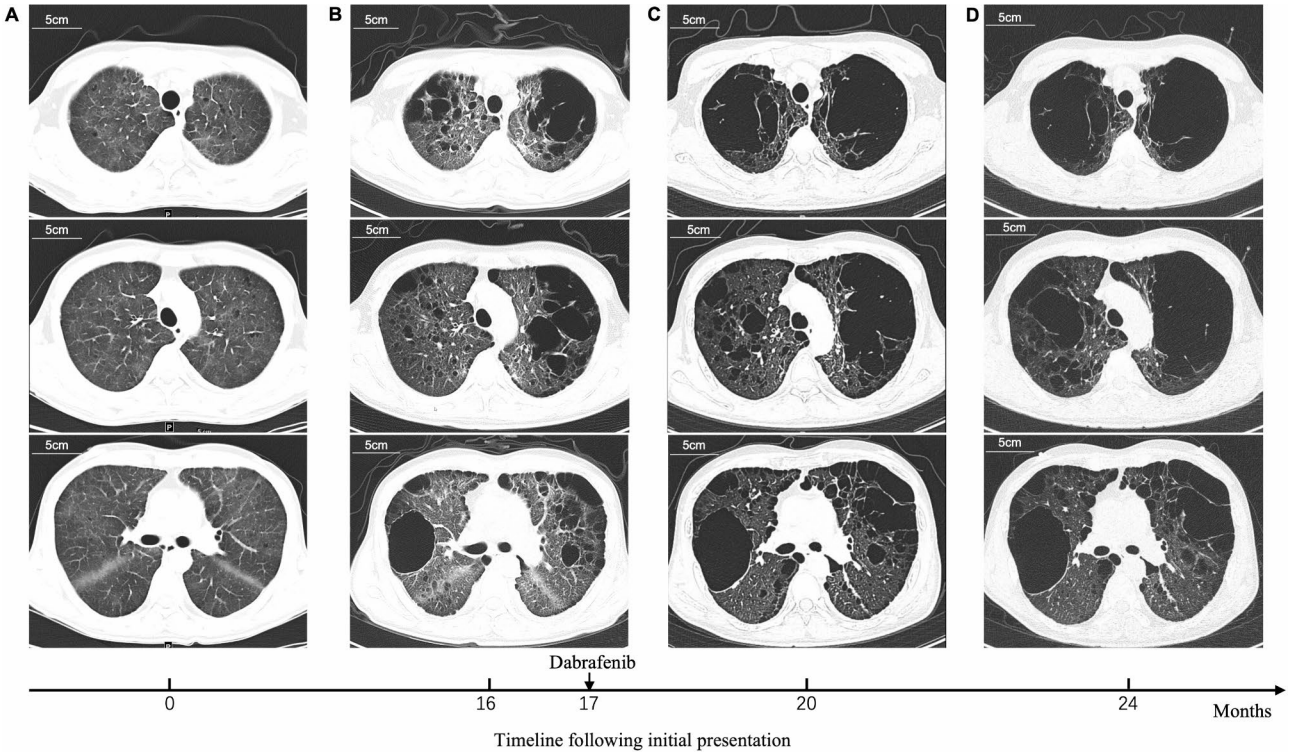

Fig. 1 Sequential computed tomography (CT) imaging. (**A**) CT scans of the thorax 1 year prior to admission revealed bilateral multiple ground-glass opacities in the lungs, thickened interlobular septa and pleural thickening. (**B**) A repeated chest CT after admission showed marked progression of interstitial changes with multiple thin-walled cysts and thickening of the bilateral interlobular pleura. (**C**) After three months of treatment, high-resolution computed tomography (HRCT) revealed a significant alleviation of interstitial lesions in both lungs, with reductions in ground-glass opacities and interlobular septal thickening and significantly more pronounced cystic alterations. (**D**) After seven months of treatment, HRCT showed no significant changes in the interstitial lesions, with slightly progressed cystic changes in both lungs

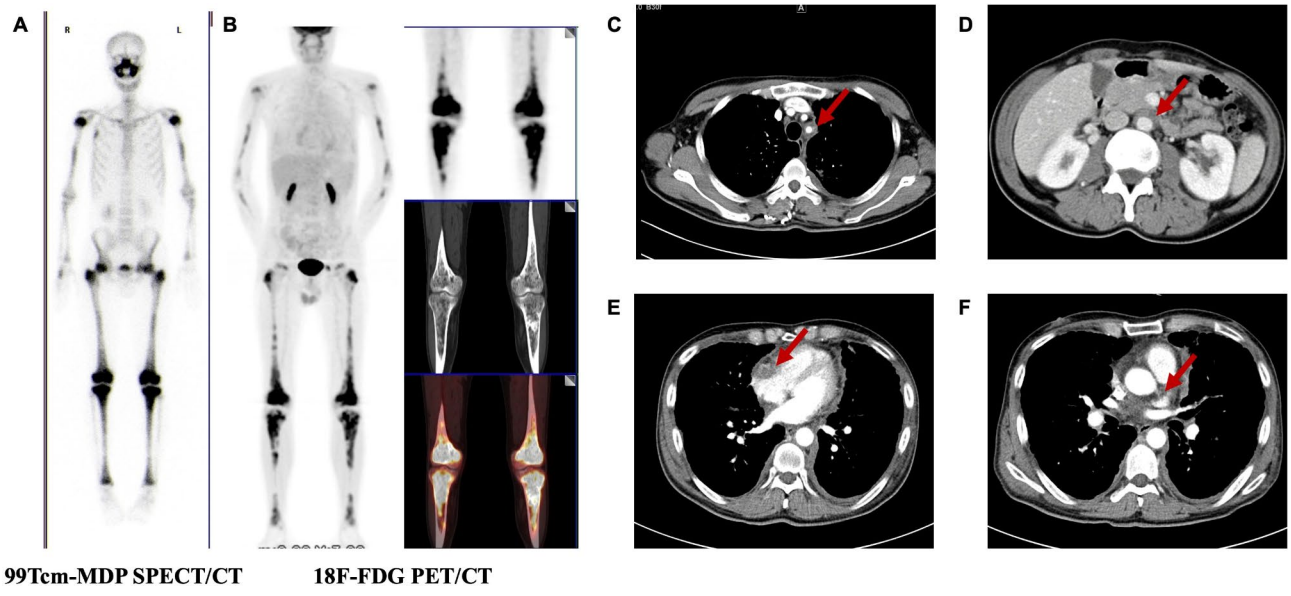

Fig. 2 Comprehensive Imaging Profile of the Patient. (**A**) Bone scintigraphy showed increased radioactive uptake in the nasal bones, maxilla and bilateral symmetric long bones of the limbs. (**B**) 18 F-FDG PET/CT revealed multiple areas of increased metabolism in the long bones of the limbs. Thoracoabdominal enhanced computed tomography (CT) showed (**C**) circumferential wall thickening and lumen narrowing at the root of the left subclavian artery, (**D**) uneven thickening of the aortic wall, (**E**) soft tissue shadows in the right atrioventricular groove and (**F**) soft tissue shadows in the anterior descending branch area of the heart

Table 1 Clinical information of previously reported cases of ECD patients with pulmonary cystic changes

Case No.	Age/sex	Ethnicity or nationality	Symptoms	BRAF V600E status	Treatment	Treatment efficacy	Publication Year
1	63/M	Caucasian	dyspnea, dry cough	-	none*	-	2000 [11]
2	50/M	Japanese	dyspnea	-	systemic prednisolone	deteriorated	2011 [12]
3	42/F	Chinese	fever, dyspnea	mutated	interferonα	stable	2022 [13]
4	18/M	Chinese	dyspnea, blurred vision#	mutated	vemurafenib	improved	2024 [14]
5	50/M	Ethiopian	bilateral leg pain	-	analgesic	-	2024 [15]

*, The patient passed away due to pulmonary infection and renal failure prior to treatment; #, The patient had blurred vision due to lesions involving the orbit. F, female; M, male

enhancement, all of which are suggestive of ECD lesions. An echocardiogram revealed a pulmonary artery systolic pressure of 70 mmHg, and the left ventricle was "D" shaped. The right heart catheterization findings demonstrated that the mean pulmonary artery pressure (mPAP) was 37 mmHg, the mean pulmonary arterial wedge pressure (mPAWP) was 6 mmHg, the pulmonary vascular resistance (PVR) was 5.68 Wood units, and the acute pulmonary vasodilator testing (APVT) test was positive. Pathology of transbronchial lung biopsy (TBLB) and yellowish tumors on the cheeks both revealed numerous histiocytes consistent with ECD, and immunohistochemistry (IHC) assays revealed CD163+, CD68+, CD1a (-), B-RAF (IHC) (-) and Langerin (-). Furthermore, subsequent skin pathology MAPK pathway mutation analysis by next-generation sequencing (NGS) confirmed a BRAF V600E mutation. The patient started dabrafenib 150 mg twice daily and reported improvement in his facial xanthomas and cough after one month of treatment. With intermittent low-grade fever, ongoing bilateral leg pain and fatigue persisted. After three months of treatment, high-resolution computed tomography (HRCT) revealed significant alleviation of the interstitial lesions in both lungs, with reductions in ground-glass opacities and interlobular septal thickening; the cystic alterations in the lungs appeared more pronounced than before (Fig. 1C). Simultaneously, there was a marked reduction in the serum interleukin-6 (IL-6) concentration and the N-terminal pro-brain natriuretic peptide (NT-proBNP) level returned to normal. Approximately seven months after initiating treatment, follow-up HRCT showed no significant changes in the interstitial lesions, while the cystic changes in both lungs had slightly progressed (Fig. 1D). Simultaneously, increased alkaline phosphatase (ALP) concentration compared to the pretreatment values, alongside decreased beta collagen degradation product (β-CTX) may suggest a reduction in bone turnover or diminished bone resorption activity. Treatment with dabrafenib was still ongoing at the time of the last follow-up.

Discussion and conclusions

ECD is essentially a type of histiocytic neoplasm. Primarily, ECD, like LCH, was classified as an inflammatory disease. However, with the confirmation of recurrent activating mutations in the MAPK/extracellular signal-regulated kinase (ERK) pathway's role in both ECD and LCH, ECD was reclassified as a hematopoietic and lymphoid tumor in the 2016 WHO classification [7]. Current reports indicate that the rate of pulmonary involvement in ECD ranges from 25 to 50% [8–10], with a prevalence second only to bone involvement [9] and is not related to the presence of the BRAF V600E mutation [11]. The most common pulmonary manifestation of ECD is pleural involvement, followed by interstitial lung disease (ILD), characterized predominantly by interlobular septal thickening and pulmonary nodular lesions. Thin-walled cystic lung lesions are less commonly observed [9].

Table 1 summarizes the clinical information of reported ECD patients with pulmonary cystic changes, though survival data remain scarce. Another retrospective study reported 8 cases of ECD patients with pulmonary cystic changes, while corresponding clinical characteristics was lacked [9]. Cystic lung lesions are more prevalent in LCH compared to ECD. LCH patients often have a history of smoking and frequently present with pneumothorax, whereas pneumothorax is rarely seen in ECD patients [9].

The diagnosis of ECD is a multifaceted process that goes beyond pathology or chest imaging alone, requiring the integration of histopathologic features with clinical, radiographic, and molecular findings. Evaluation of tumour tissue for molecular alterations, where feasible, provides crucial insights that not only confirm the diagnosis but also guide treatment decisions as recommended by national comprehensive cancer network (NCCN) guidelines for histiocytic neoplasms [16]. The BRAF V600E mutation is found in 55–71% of ECD patients [9, 17]. Reported response rates for BRAF V600E inhibitors in the treatment of ECD range from 43% [18] (6 of 14), to 100% [19] (10 of 10), with a median progression-free survival (mPFS) of 25.9 months. Although data on overall survival are still immature [20], it has already

been observed that patients with pulmonary involvement have significantly poorer survival outcomes [10].

The present case is an example of such severe pulmonary cystic disease being relatively rare in ECD, where the largest cystic lesion in the lung reached a diameter of 10 cm. The mechanisms underlying lung cyst formation in histiocytosis disorders such as PLCH or ECD remain poorly understood at present. Tracking imaging studies in PLCH proposed that the process begins with Langerhans cells infiltrating the small airways, forming granulomas that damage the bronchiolar walls. As this evolves, fibrosis develops over time, and traction on the surrounding lung tissue leads to the formation of cystic spaces [21]. In this case, the initial chest CT revealed ground-glass nodules, along with thickening of the interlobular septa and pleura, while pulmonary cystic changes emerged mainly in the later stages of the disease. Imaging improvements after targeted therapy were mainly reflected in the resolution of the ground-glass nodules and interlobular septal thickening. Additionally, the patient's pulmonary function tests indicated a nearly normal FEV1/FVC ratio but significantly impaired diffusion capacity, suggesting that interstitial involvement was more prominent than airway involvement, which from an additional perspective, corroborates the hypothesis. However further research is definitely required to elucidate the pathological processes that drive cyst development in these rare histiocytic proliferative disorders. The current consensus on the pathogenesis of ECD posits that activation of the MAPK pathway, including BRAF and MEK, results in clonal proliferation of histiocytes. Additionally, immune-mediated proinflammatory processes may also be involved in this disease mechanism [22]. Previous studies have shown that IL-6 levels in patients with ECD are elevated up to 16-fold compared to those in healthy controls, with notable reductions following interferon-alpha therapy [23]. In this case, the decrease in IL-6 after treatment was especially pronounced, suggesting potential novel avenues for ECD treatment approaches in the future.

This case is also the first to report pulmonary arterial hypertension in a patient with ECD, confirmed via right heart catheterization, suggesting that pulmonary hypertension could be a complication of ECD. However, pulmonary hypertension is more common in patients with PLCH [24]. Pathology of the lung in ECD patients revealed peribronchial and perivascular histiocytic infiltrates with fibrosis [11], and periarterial interstitial thickening might be the main cause of pulmonary hypertension in ECD, with subsequent cystic changes possibly altering vascular structures. Positive APVT might indicate that the changes in the vascular wall differ from the vascular remodelling observed in idiopathic pulmonary hypertension and have a certain degree of reversibility, as previous cases have reported significant reductions in echocardiographic estimates of pulmonary artery systolic pressure after targeted therapy for ECD [14]. The pulmonary manifestations of ECD are varied. The presence of multiple irregular cystic lesions, especially predominant in the upper lungs, should raise suspicion for a proliferative histiocytic disorder. Greater suspicion for ECD is warranted when imaging findings do not align with clinical indications of hypoxia or when the patient presents with concurrent symptoms such as xanthomas and bone pain.

Abbreviations

ECD	Erdheim-Chester disease
MAPK	Mitogen-activated protein kinase
WHO	World Health Organization
LCH	Langerhans cell histiocytosis
CT	Computed tomography
BALF	Bronchoalveolar lavage fluid
FEV1	Forced expiratory volume in the first second
FVC	Forced vital capacity
TLC	Total lung capacity
DLCO-SB	Diffusing capacity for carbon monoxide
PLCH	Pulmonary Langerhans cell histiocytosis
PCP	Pneumocystis jirovecii pneumonia
LAM	Lymphangioleiomyomatosis
BHD	Birt-Hogg-Dubé Syndrome
MDP	Methylene diphosphonate
SPECT	Single-photon emission computed tomography
FDG	Fluorodeoxyglucose
PET	Positron emission tomography
mPAP	Mean pulmonary artery pressure
mPAWP	Mean pulmonary arterial wedge pressure
PVR	Pulmonary vascular resistance
APVT	Acute pulmonary vasodilator testing
TBLB	Transbronchial lung biopsy
IHC	Immunohistochemistry
NGS	Next-generation sequencing
HRCT	High-resolution computed tomography
IL-6	Interleukin-6
NT-proBNP	N-terminal pro-brain natriuretic peptide
ALP	Alkaline phosphatase
β-CTX	Beta collagen degradation product
ERK	Extracellular signal-regulated kinase
ILD	Interstitial lung disease
NCCN	National comprehensive cancer network
mPFS	Median progression-free survival

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None.

Author contributions
YXW completed the writing. HWW was responsible for the analysis of pathological data. JWG assisted in investigation. XFS contributed to the conceptual framework of the research and supervised the project. All authors reviewed and approved the final manuscript.

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Data availability
The data that support the findings of this study are not openly available to protect study participant privacy. To obtain access to the raw data, reasonable requests should be directed to the corresponding author via email at sunxfer@sina.com.

Declarations

Ethics approval and consent to participate
This case report was approved by the Medical Ethics Committee of PUMCH and clinical trial number was not applicable.

Consent for publication
Informed consent for publication was obtained from the patient. The patient gave written informed consent for his clinical details along with images to be published in this study by signing the consent form.

Competing interests
The authors declare no competing interests.

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