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CASE REPORT

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A complex clinical case of intracardiac leiomyomatosis

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Abstract

Background Intracardiac leiomyomatosis (ICL) is an uncommon condition characterized by the proliferation of intravascular tissue within the veins, leading to the development of tumor emboli. This can pose a significant threat to life when the tumor invades cardiac structures. The diagnostic process for this condition is complex and presents considerable challenges.

Case presentation We report a case of a 38-year-old female patient whose pulmonary artery computed tomography (CT) revealed low density structure in the branches of the pulmonary artery. Echocardiography revealed a mobile tumor within the right heart chambers and pulmonary trunks as well as characteristic thickening of the ventricular septum consistent with hypertrophic cardiomyopathy (HCM). Magnetic resonance imaging (MRI) revealed a mass in the right anterior uterine wall, extending to the inferior vena cava (IVC) and right iliac vein. Post-surgery histopathological analysis confirmed a diagnosis of intravenous leiomyomatosis (IVL).

Conclusions When IVL affects the heart, echocardiography is the best diagnostic tool for detecting the disease. CT and MRI are essential in identifying the location and extent of the tumor, as well as in evaluating prognosis.

Keywords Intracardiac leiomyomatosis, Echocardiography, Hypertrophic cardiomyopathy

Introduction

Intravenous leiomyomatosis (IVL) is a rare disease characterized by the proliferation of intravascular tissue within the veins, leading to the formation of masses that can obstruct blood flow. This condition is observed in approximately 10% of patients with uterine leiomyomas [1, 2]. In certain cases, tumor tissue may proliferate within the uterine vein, extend into the inferior vena cava (IVC), and potentially reach the right heart and pulmonary artery. According to reported cases, intracardiac extension was only 6-30% [3]. Such progression can obstruct the tricuspid and pulmonary valves, posing a significant risk to life and resulting in a poor prognosis. Intracardiac leiomyomatosis (ICL) is exceedingly rare and is often misdiagnosed as thrombosis or a primary cardiac tumor due to the atypical and infrequent clinical manifestations associated with ICL. This misdiagnosis can lead to inappropriate treatment strategies. Therefore, an accurate diagnosis of ICL is critically important for effective management of the condition. Computed tomography (CT) and magnetic resonance imaging (MRI) of the pelvis can reveal the primary tumor. A CT scan of the chest and abdomen reveals the extent of involvement in vascular structures. Furthermore, echocardiography has the advantage of assessing the mobility and morphological features of intracardiac and pulmonary masses. Based

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on the above findings, an appropriate surgical approach can be selected for the patient.

We report a complex clinical case of ICL, characterized by the proliferation of tumor tissue from the uterus along the IVC to the right cardiac chambers and the pulmonary artery.

Case presentation

A 38-year-old woman presented with mild bilateral lower extremity edema of two weeks duration following a long-distance train journey, which has progressively intensified since its onset. She had experienced amenorrhea for 3 months. She did not experience chest pain or cough, and had no history of allergic reactions. The patient denied taking any medications prior to admission. She had no family history of thrombosis or cancer. On physical examination, the skin was pale and peripheral edema was revealed. Additionally, a mass was revealed during abdominal palpation. Ultrasound examination did not reveal any deep venous thrombosis in the lower extremities. CT angiogram of the pulmonary artery revealed a filling defect in the main pulmonary artery and its branches (Fig. 1). Therefore, the patient was diagnosed with pulmonary embolism and she was transferred to the emergency room. A series of examinations were arranged instantly. Her laboratory work-up: on admission revealed elevated erythrocyte, hemoglobin, hematocrit, D-Dimer and NT-proBNP levels, along with a low platelet count (Table 1).

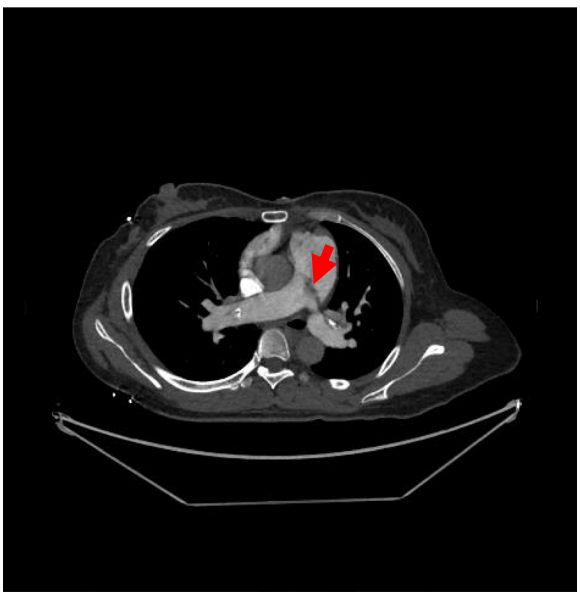

Fig. 1 The computed tomography angiography (CTA). Axial image of the chest at the level of the bifurcation showing a filling defect

Table 1 Laboratory indicators

Test	Results	Reference range
Erythrocyte	5.75*10^12/L	3.8—5.1*10^12/L
Hemoglobin	177.00 g/L	115—150 g/L
Hematocrit	53.90%	35—45%
Platelet	21*10^9/L	125—350*10^9/L
N-terminal pro-brain natriuretic peptide	5294 pg/mL	< 125 pg/mL
D-Dimer	1.44 mg/L	≤ 0.55 mg/L

*multiply

Transthoracic echocardiography revealed a mobile solid tumor extending from the IVC to the right heart and left proximal pulmonary artery. This structure oscillated in synchrony with cardiac cycles, resulting in obstruction of both the tricuspid and pulmonary valves (Fig. 2, Video S1-S3). The echocardiographic evaluation indicated the right heart enlargement and pulmonary hypertension, with a calculated pulmonary artery pressure of 93 mmHg. Contrast-enhanced pulmonary artery magnetic resonance angiography revealed a filling defect at the tricuspid orifice and within the right ventricle, originating from the IVC, consistent with echocardiographic findings. It is important to highlight that the patient exhibited a distinctive thickening of the interventricular septum, measuring approximately 20 mm. This finding aligns with the clinical presentation of asymmetric hypertrophic cardiomyopathy. However, no obstruction was observed in the left ventricular outflow tract at rest. This was an incidental finding and the patient was not previously diagnosed or managed for hypertrophic cardiomyopathy (HCM). This finding was corroborated by MRI (Fig. 3, Video S4). Pelvic MRI revealed a mass in the right anterior uterine wall, extending to the uterine contour (Fig. 4). Furthermore, a tumor or thrombus was identified in the right iliac vein (Fig. 5). Of note, the patient had previously undergone surgery for uterine leiomyomas twelve years prior. The patient ultimately received a working diagnosis of uterine leiomyomatosis with intracardiac involvement.

Due to the patient's critically low platelet count, which precluded surgery, a platelet transfusion was administered prior to the procedure. A successful one-stage procedure was conducted, which included total hysterectomy, bilateral oophorectomy, and excision of intracardiac lesions. Excision of an intravenous, intravascular, and intracardiac mass was achieved through a right atriotomy and longitudinal venotomy (Fig. 6). Histological analysis of the post-surgical specimen revealed proliferating smooth muscle fibers, without evidence of abnormal mitotic activity. This finding was

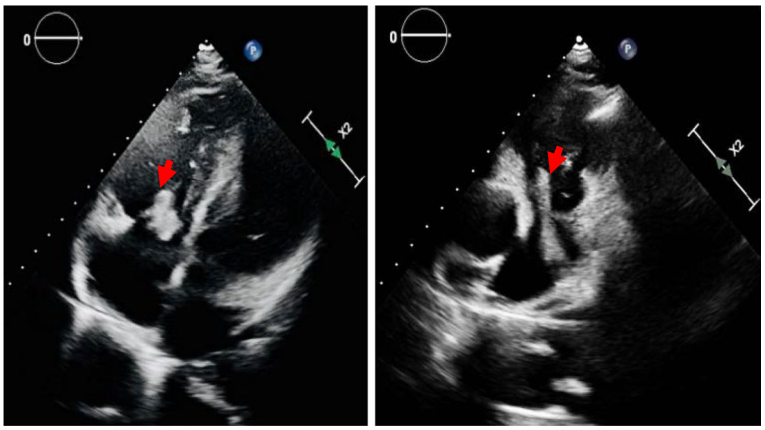

Fig. 2 Echocardiography showed the oscillating echogenic mass in right cardiac chambers (**A**) and pulmonary artery (**B**) in synchrony with the cardiac cycle

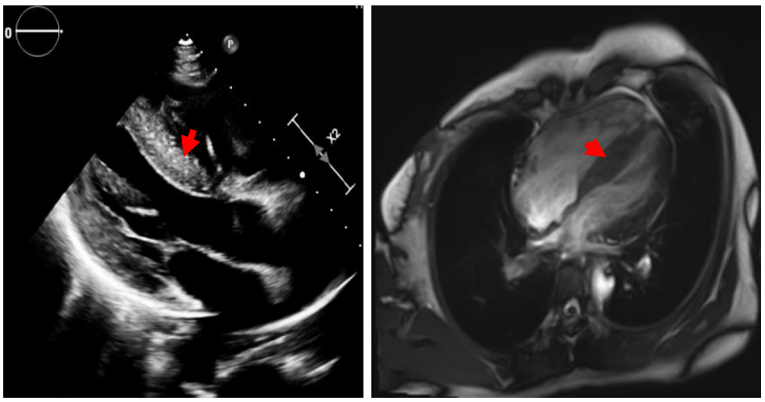

Fig. 3 Thick interventricular septum (red arrow) in echocardiography (**A**) and MRI (**B**)

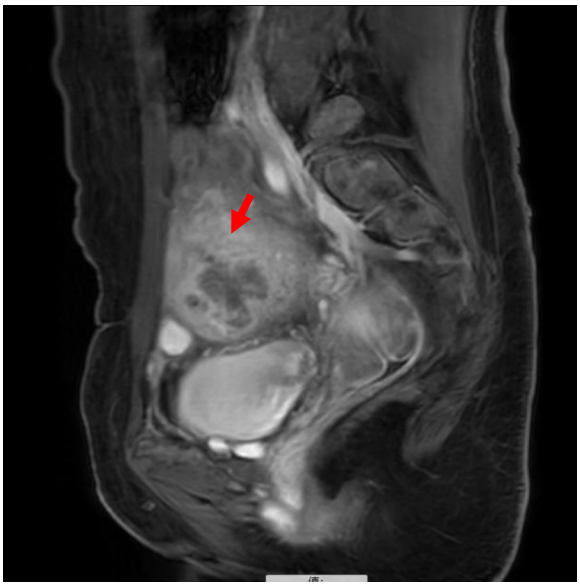

Fig. 4 Mass in pelvic MRI (red arrow)

consistent with a diagnosis of intravenous leiomyomatosis. The patient was discharged from the hospital on post-surgery day 25. At the 5-month follow-up, she displayed ongoing recovery, with no recurrence of cardiac symptoms seen on echocardiography.

Discussion

IVL is an uncommon uterine neoplasm encountered in clinical practice. Intracardiac extension was first described in 1907 by Durck H [4]. The World Health Organization (WHO) classifies IVL as a benign lesion within the category of tumors of the female reproductive system, characterized by the atypical proliferation of leiomyoma [5]. Due to its proliferative growth characteristics and propensity for recurrence, IVL has the potential to extend from the uterine vein along the IVC to the right cardiac chambers and the pulmonary artery. ICL represents a severe and potentially fatal manifestation of IVL. Nevertheless, the underlying pathophysiology remains unclear at this time. Two main proposed theories aim to

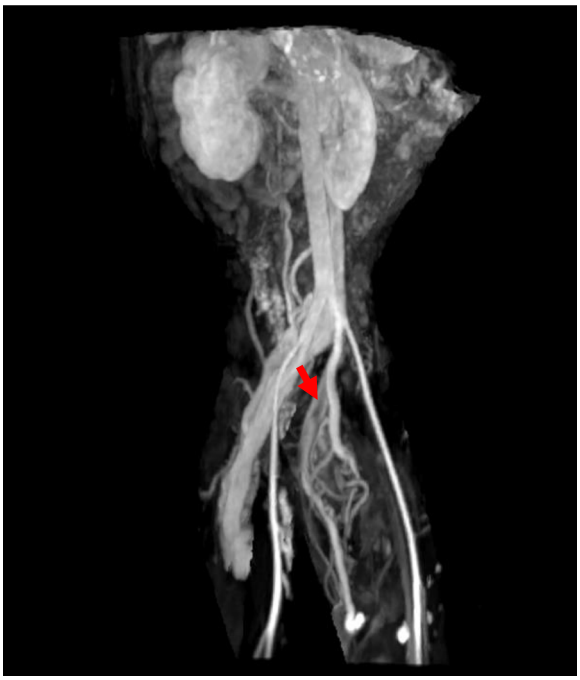

Fig. 5 3D MRI of right iliac vein showed a filling defect

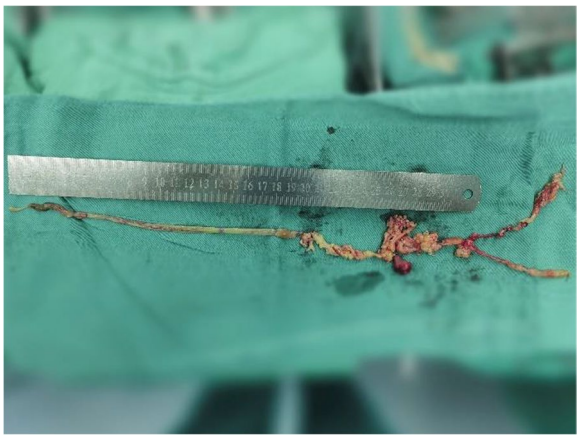

Fig. 6 A lengthy mass is observed following its removal from the IVC and the right heart

elucidate the pathogenesis of the disease. One hypothesis suggests that tumor cells originate from venous walls, while another proposes that vascular invasion is a result of primary uterine leiomyoma extension [6, 7]. Our case is consistent with a uterine origin.

Patients with IVL may present without symptoms in the initial stages of the disease. The onset of symptoms such as dyspnea, syncope, lower extremity edema, or even sudden death suggests that the neoplasm has extended to the cardiovascular system [8, 9]. Related pulmonary embolism is particularly rare, with reported cases less than 5% [10]. It is worth noting that pulmonary embolism is present in our case. Several potential explanations could account for this complication. Firstly, tumor cells possess specific surface receptors that can interact with platelets, resulting in platelet aggregation and the development of a hypercoagulable state in the blood. This hypercoagulable state is a contributing factor to the decreased platelet count in the patient. Secondly, tumor emboli proliferating within vessels continuously exert friction and cause erosion of the endothelial cells lining their surfaces. This process results in endothelial injury. Mechanical damage facilitates thrombus formation. Thirdly, intravascular tumors obstruct blood flow, leading to stasis of blood and increasing the risk of thrombus formation. When thrombi dislodge, they may move through the bloodstream to the pulmonary artery, resulting in a pulmonary embolism. However, the infrequency of ICL, coupled with its unusual clinical manifestations, often results in misdiagnosis as a cardiac or pulmonary thrombus. This misdiagnosis can lead to the administration of ineffective thrombolytic therapy. Thrombosis does not last for several days. It is completed during a brief infusion period. The underlying cause of the lower extremity edema in this patient was identified as obstruction of the IVC. Moreover, when ICL and HCM occur concurrently, it is imperative to conduct a more thorough assessment of the hemodynamic status and search for potential obstruction of the left ventricular (LV) outflow tract or mid-ventricular obstruction during examination. It is vital to clarify the underlying factors contributing to the related clinical manifestations.

The diagnosis of ICL is challenging due to its nonspecific clinical manifestations and imaging features. Various imaging techniques are critical for determining the precise location and extent of the tumor, including ultrasonography, MRI, and CT [11]. Transthoracic echocardiography is considered the most effective modality for evaluating intracardiac and associated vascular masses. An experienced operator is needed to mitigate the risk of misdiagnosis. An oscillating mass of neoplastic tissue inside the IVC, right cardiac chambers and pulmonary artery may be detected by real-time ultrasound imaging. The combination of uterine myoma revealed by pelvic ultrasound and previous surgical history led to a proper diagnosis in our case. CT and MRI provide comprehensive insights into the location, morphology, and extent of tumors throughout the entire body. MRI can demonstrate delayed enhancement of intravenous tumors, which is distinct from thrombus. In cases involving the lungs, lung nodules can be identified by chest CT [12].

A one-stage total hysterectomy, bilateral oophorectomy, and resection of intracardiac tumor represent a comprehensive approach and the most favorable option for managing ICL [13, 14]. The surgical approach for ICL presents significant challenges, encompassing procedures such as median sternotomy for chest access, total hysterectomy, bilateral oophorectomy, and excision of metastatic tumors in the internal iliac vein [14]. A thorough evaluation is necessary prior to the procedure, which should be conducted by a team of multidisciplinary surgeons. The perioperative period is associated with several potential risks, including intraoperative pulmonary artery rupture, laceration of the IVC, and postoperative retroperitoneal hemorrhage [15]. Although surgical procedures can provide significant benefits, the possibility of recurrence remains a concern [16]. This report has a primary limitation in the insufficient duration of the follow-up period. It remains essential to focus greater attention on the prognostic outcomes of patients with ICL who have undergone surgical interventions.

In conclusion, various imaging modalities are vitally important in the diagnosis and follow-up of ICL patients. The presence of a mass in the right cardiac chambers and IVC should raise suspicion for ICL, especially in patients with a history of uterine myomectomy or those with a uterine leiomyoma. Early identification, accurate diagnosis and appropriate treatment are beneficial for patients.

Abbreviations

IVL	Intravenous leiomyomatosis
ICL	Intracardiac leiomyomatosis
CT	Computed tomography
MRI	Magnetic resonance imaging

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Authors' contributions
Y.L., M.L. and X.L. designed the case report, collected the data, drafted the initial manuscript, and revised the manuscript; Y.L., L.S., W.Z., L.W., X.D and D.G. collected the data and revised the manuscript; X.L. conceptualized and designed the study and reviewed and revised the manuscript; and all the authors approved the final manuscript as submitted and agreed to be accountable for all aspects of the work.

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Data availability
The datasets used and/or analyzed during the current report are available from the corresponding author upon reasonable request.

Declarations

Ethics approval and consent to participate
The study was approved by the ethics committee of Beijing Chaoyang Hospital affiliated to Capital Medical University. The patient provided informed consent for this report.

Consent for publication
Written informed consent was obtained from the patient for all the manuscripts that included images and details. The authors have deidentified patient-specific information.

Competing interests
The authors declare no competing interests.

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