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Case report

Supravalvular pulmonary stenosis as a manifestation of a mediastinal germ cell tumor: a case report

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ABSTRACT

Extrinsic compression of the pulmonary artery by a mediastinal mass is an exceedingly rare cause of pulmonary stenosis. We report the case of a 22-year-old man with no relevant medical history who, during a pre-surgical evaluation, presented with a three-month history of exertional dyspnoea. Echocardiography demonstrated supravalvular pulmonary stenosis, and chest computed tomography revealed a giant mediastinal mass encasing the great vessels and causing collapse of the left pulmonary artery branch. Mediastinal biopsy and immunohistochemical analysis were consistent with a primary mediastinal germ cell tumour of seminomatous morphology, accompanied by serum alpha-fetoprotein elevation, leading to its classification as a high-risk extragonadal germ cell tumour. This case highlights the importance of multimodal imaging in the differential diagnosis of mediastinal masses with compressive cardiac involvement, as well as the value of histopathological correlation and tumour marker assessment in guiding prognosis and multidisciplinary management.

Keywords: Stenosis Pulmonary Artery; Neoplasms, Germ Cell and Embryonal; Seminoma;; Multimodal Imaging (Source: MeSH-NLM).

RESUMEN

Estenosis pulmonar supravalvular como manifestación de un tumor mediastinal de células germinales: reporte de caso

La compresión extrínseca de la arteria pulmonar por una masa mediastinal es una causa extremadamente rara de estenosis pulmonar. Presentamos el caso de un varón de 22 años, sin antecedentes médicos relevantes, que durante una evaluación prequirúrgica refirió disnea de esfuerzo de tres meses de evolución. El ecocardiograma mostró estenosis pulmonar supravalvular y la tomografía de tórax reveló una masa mediastinal gigante que encasillaba grandes vasos y colapsaba la rama pulmonar izquierda. La biopsia mediastinal y el estudio inmunohistoquímico fueron compatibles con un tumor de células germinales primario mediastinal con morfología de seminoma y la elevación sérica de alfafetoproteína, por lo que se le catalogó como un tumor de células germinales extragonadales de alto riesgo. Este caso resalta la importancia de la imagen multimodal en el diagnóstico diferencial de masas mediastinales con compromiso cardíaco compresivo, así como la correlación histopatológica y de marcadores tumorales para orientar el pronóstico y el manejo multidisciplinario.

Palabras clave: Estenosis de Arteria Pulmonar; Neoplasias de Células Germinales y Embrionarias; Seminoma; Imagen Multimodal (Fuente: DeCS-BIREME).

Introduction

Mediastinal masses are uncommon and usually do not cause symptoms in their early stages. However, when large, they can cause compression of the heart and great vessels, presenting with dyspnoea, chest pain, and even superior vena cava syndrome. Supravalvular pulmonary stenosis secondary to a mediastinal tumour is an even rarer manifestation, with only a few cases reported in the literature [1-3].

The masses that most commonly occupy the anterior mediastinum are malignant thymomas, lymphomas, and thyroid tumours; malignant germ-cell tumours are less frequent, accounting for only 10% of cases [4]. Among the latter, mediastinal seminomas are rare and have a favourable prognosis; however, concomitant elevation of any tumour marker suggests non-seminomatous biological behaviour and may worsen the prognosis [5].

Case report

We present the case of a 22-year-old male patient with no relevant medical history who, during a preoperative evaluation for acute appendicitis, was found to have a grade III/VI multifocal systolic murmur on auscultation. The patient reported a 3-month history of exertional dyspnoea associated with dry cough and unquantified weight loss. Baseline electrocardiographic findings are shown in **Figure 1**.

The presence of a grade III/VI multifocal murmur of considerable intensity in a young patient raised the possibility of acyanotic congenital heart disease, such as a ventricular septal defect or patent ductus arteriosus. However, the finding of increased P-wave amplitude on the electrocardiogram broadened the differential diagnosis to include congenital pulmonary stenosis.

Transthoracic echocardiography (TTE) incidentally showed a large mass with irregular borders and heterogeneous content lateral to the left ventricle, causing compression and displacement towards the contralateral hemithorax **(Video 1)**. A narrowing of the pulmonary artery at the supravalvular level was noted, with an obstructive gradient (maximum velocity 3.2 m/s; maximum gradient 40.9 mmHg) **(Figure 2)**. However, right-sided chamber dimensions were normal, as were right ventricular systolic function (TAPSE: 2 cm, S´ wave: 13 cm/s, fractional area change: 49%) and right ventricular-pulmonary arterial coupling (TAPSE/PASP: 0.64) **(Video 2)**. Left ventricular systolic function was normal, with a biplane ejection fraction of 55%, although cardiac output was reduced at 3.6 L/min. In addition, a mild pericardial effusion was found, with a maximum pocket of 6 mm.

Contrast-enhanced computed tomography (CT) of the chest, abdomen, and pelvis showed a giant anterior mediastinal mass measuring 144 x 181 x 160 mm, with lobulated borders and heterogeneous density, areas of central necrosis, and punctate calcifications. The mass extended into the left hemithorax and displaced the heart to the right. It also encased the ascending aorta, aortic arch, part of the thoracic aorta, and carotid arteries, and compressed the pulmonary trunk, with a cross-sectional area of 1.49 cm^2 at the point of greatest narrowing, causing collapse of the left pulmonary artery branch, without evidence of direct infiltration **(Figure 3)**. No metastatic lesions or regional or extraregional lymphadenopathy were observed. On cardiac magnetic resonance imaging (MRI), the mass showed heterogeneous signal, isointense on T1-weighted images and hyperintense on T2-weighted images, with diffusion restriction and central necrotic areas, progressive enhancement after contrast administration, and collapse of the left pulmonary artery branch without myocardial invasion **(Figure 3)**.

Two CT-guided percutaneous biopsies were performed. The first showed proliferation of atypical round cells with fibrous septa, with negative immunohistochemistry for CD3, CD20, pancytokeratin, and BCL-2, but a Ki-67 index of 70%, indicating very high cellular proliferation. The second biopsy, indicated because the first did not allow a definitive diagnosis, confirmed a round-cell neoplasm and ruled out lymphoma; it was also negative for synaptophysin and CD30. Finally, biopsy of the mediastinal surgical specimen showed a malignant round-cell neoplasm with extensive necrosis, increased mitotic activity, and positivity for SALL4 and CD117, compatible with mediastinal seminoma **(Figures 4 and 5)**.

Laboratory tests showed microcytic hypochromic anaemia, with haemoglobin of 10.2 g/dL (normal range: 14-

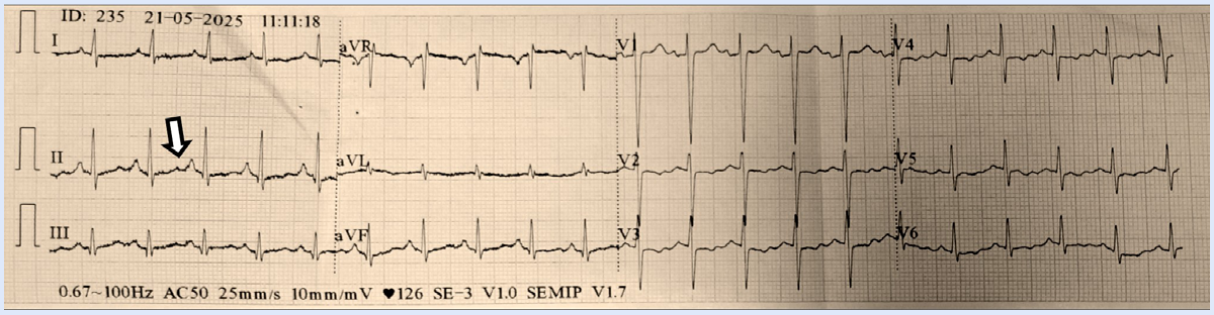

Figure 1. Twelve-lead electrocardiogram. Sinus tachycardia at 112 bpm and pulmonary P waves in lead II, with an amplitude of 0.3 mV and a duration of 0.04 ms, are noted. Non-specific repolarisation abnormalities are also observed in the precordial and lateral leads.

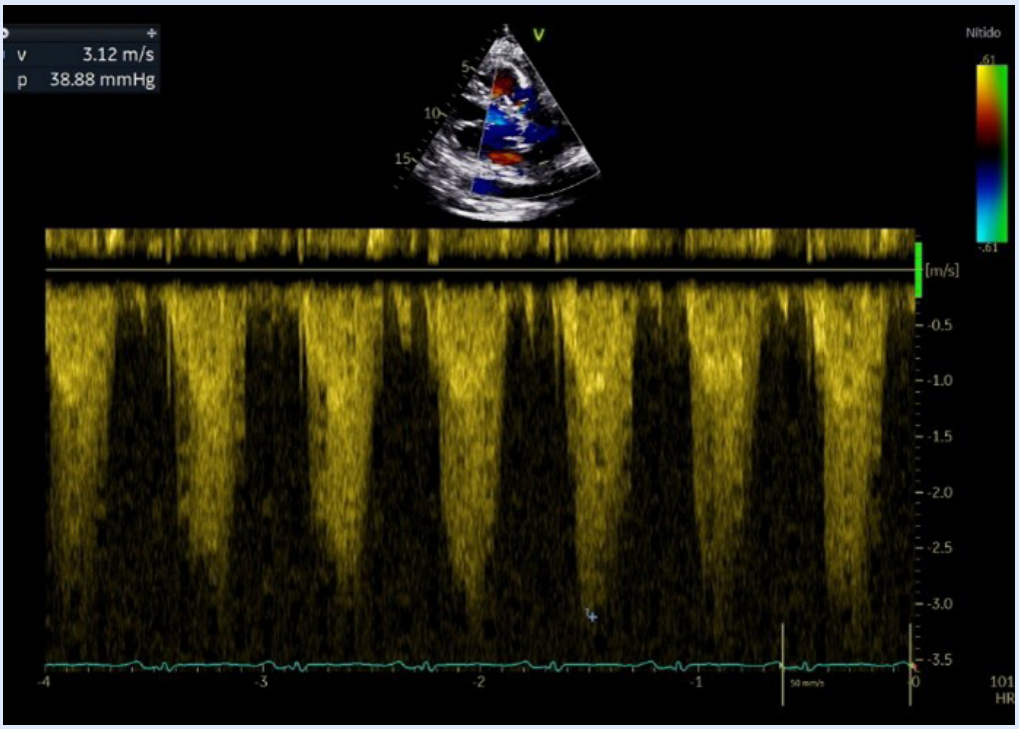

Figure 2. Supravalvular pulmonary stenosis on transthoracic echocardiography. Continuous-wave Doppler showed increased peak velocity and maximum gradient, consistent with pulmonary stenosis.

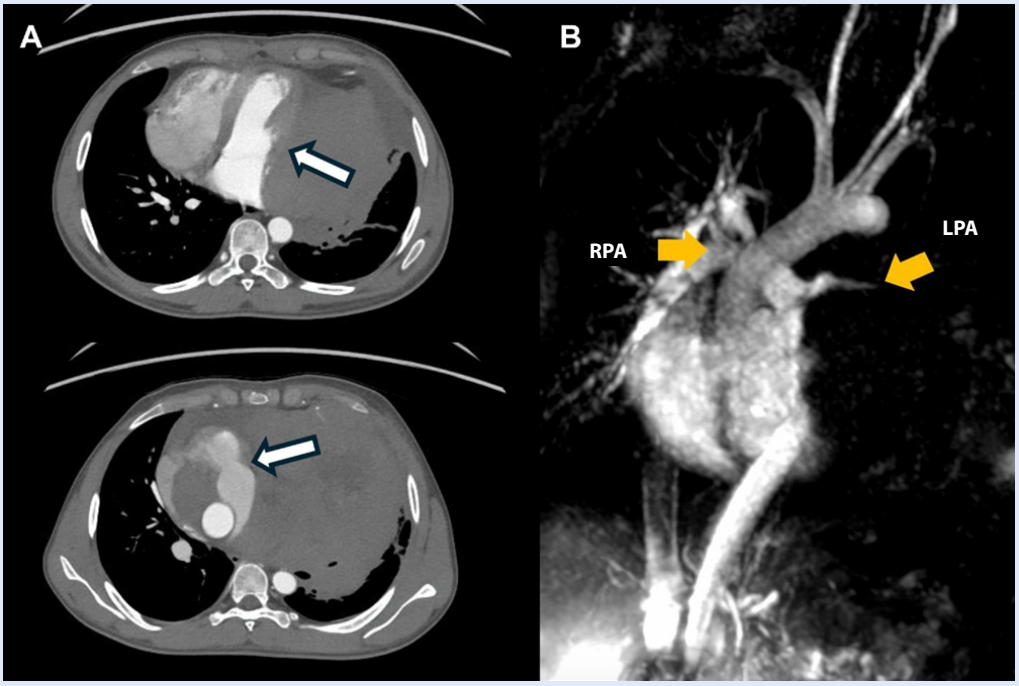

Figure 3. **(A)** Contrast-enhanced chest computed tomography. In the upper image, the arrow indicates the compressive effect on the left-sided cardiac chambers and their displacement towards the contralateral hemithorax; in the lower image, the arrow indicates narrowing of the main pulmonary artery, producing an obstructive effect with increased velocities and gradients on ultrasound. **(B)** Thoracic magnetic resonance imaging showing collapse of the left pulmonary artery branch.

RPA: right pulmonary artery, LPA: left pulmonary artery

17), mean corpuscular volume of 73.9 fL (normal range: 80-96), and mean corpuscular haemoglobin of 23.9 pg (normal range: 28-33), as well as lymphopenia, with lymphocytes accounting for 7.1% (normal range: 25-40). NT-proBNP was 398 pg/mL (normal range: <100). Other biochemical findings included alkaline phosphatase of 480 U/L (normal range: 40-129). Tumour markers were elevated, with LDH 2599 U/L (normal range: 135-225), alpha-fetoprotein (AFP) 169 ng/mL (normal range: 0-7), and β-HCG 71 mIU/mL (normal range: <2).

During the clinical course, the patient developed superior vena cava syndrome and underwent emergency surgical excision of the mediastinal tumour. During the procedure, he developed cardiorespiratory arrest that did not respond to advanced cardiopulmonary resuscitation manoeuvres, resulting in intraoperative death.

Discussion

Supravalvular pulmonary stenosis is a rare form of pulmonary stenosis and represents a diagnostic challenge. Congenital causes are the most frequent; however, it may also develop after surgical intervention on the pulmonary artery, rubella infection, and, even more rarely, mediastinal tumours causing compression of the pulmonary trunk or its branches [6]. In this case, progressive dyspnoea, a multifocal systolic murmur, and pulmonary P waves initially suggested acyanotic congenital heart disease. However, the incidental finding of a large mass redirected the diagnosis towards an extrinsic cause with haemodynamic and systemic repercussions.

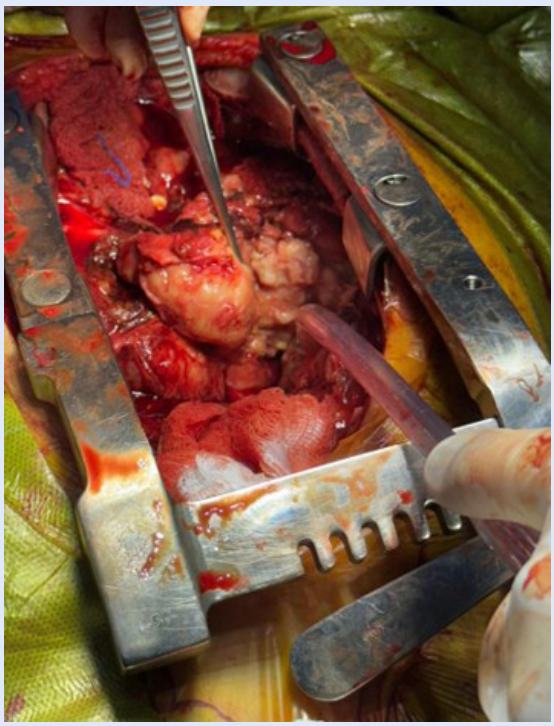

Figure 4. Intraoperative finding of the mediastinal mass. Whitish tissue with a heterogeneous appearance and nodular yellowish areas can be observed.

TTE allowed assessment of the obstructive haemodynamic effect of the mediastinal mass on the pulmonary artery. CT and MRI precisely identified the point of narrowing of the pulmonary trunk above the valvular plane and the collapse of its left branch, while histopathological examination confirmed the obstructive neoplastic cause. In addition, significant local extension was observed, but without myocardial invasion, an important finding because cardiac infiltration confers a worse prognosis and influences the therapeutic approach. This highlights the usefulness of multimodality imaging in guiding the diagnostic and therapeutic process and even in establishing the prognosis of a mediastinal tumour [4,7].

Anterior mediastinal tumours in young patients are classically summarised as the "four Ts": thymoma, teratoma and other germ-cell tumours, thyroid lesions including ectopic goitre or carcinoma, and "terrible" lymphoma [4]. Primary mediastinal seminomas are uncommon germ-cell tumours, typically located in the mediastinum and less frequently in the retroperitoneum. Unlike non-seminomatous germ-cell tumours, seminomas usually show expansive but less aggressive growth, with a lower initial propensity to metastasise and a frequently non-specific clinical presentation, which may delay diagnosis [8,9]. In a study of 120 patients diagnosed with mediastinal seminoma, the most frequent symptoms were chest pain (32.4%), dyspnoea (12.9%), and superior vena cava syndrome (9%); however, up to one-third of patients have been reported to be asymptomatic at diagnosis (32.4%) [10].

The true incidence of mediastinal masses is difficult to establish because of heterogeneity in reports, classifications, and the inclusion of benign or malignant conditions with diverse prognoses [4]. This underscores the importance of considering these neoplasms in the differential diagnosis, even when the initial presentation is uncommon and involves cardiovascular symptoms.

The diagnostic approach requires a combination of advanced imaging techniques, including CT and MRI, to define the extent of disease and vascular compression, together with biopsy and immunohistochemical analysis to characterise the histology and tumour component [11]. In our case, biopsy revealed a round-cell neoplasm with a high Ki-67 index, extensive necrosis, and positivity for SALL4 and CD117, together with negativity for lymphoid markers (CD3, CD20) and neuroendocrine markers (synaptophysin, CD30). These findings confirmed the morphology of mediastinal seminoma; however, AFP elevation, which is uncommon in pure seminomas, suggested the presence of a non-seminomatous component, such as yolk sac or embryonal tumour, and a more aggressive biological behaviour [8].

According to the International Germ Cell Cancer Collaborative Group (IGCCCG) classification, regardless of tumour marker levels or distant metastases, this case falls

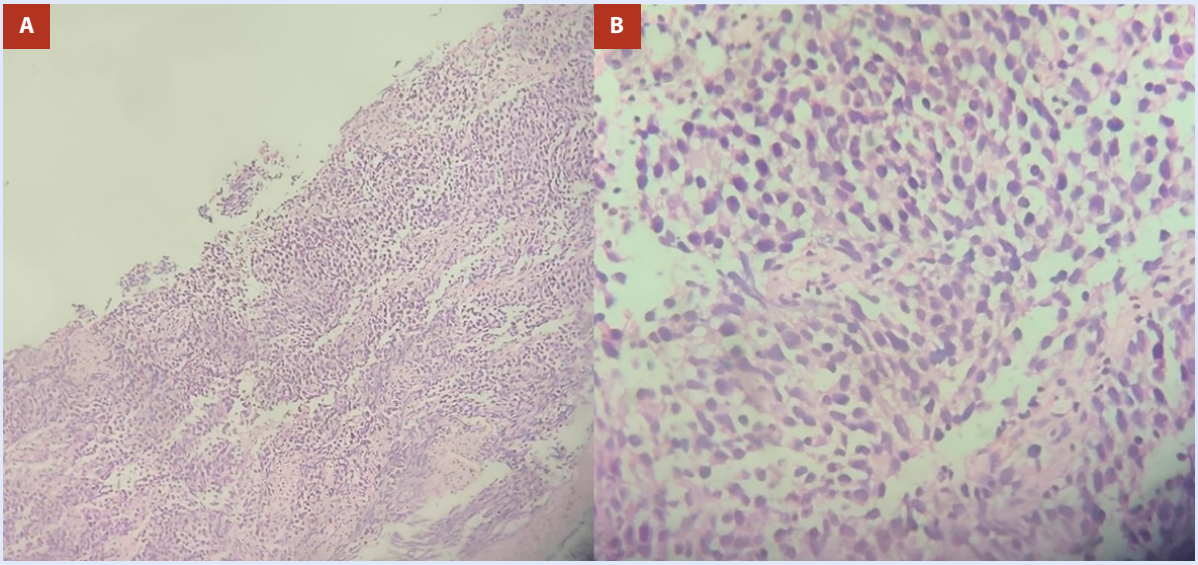

Figure 5. **(A)** Cells with atypical hyperchromatic nuclei and prominent nucleoli (4×). **(B)** Tissue specimen view: malignant round-cell neoplasm with extensive necrosis and increased mitotic activity (40×). Haematoxylin and eosin stain in both panels.

into the poor-risk category. Although the neoplasm showed seminomatous morphology histologically, AFP elevation means that, for therapeutic purposes, this tumour should be considered a high-risk non-seminomatous tumour, with an indication for combination chemotherapy according to international clinical practice guidelines such as NCCN, ESMO, and EAU guidelines [8,9,12].

From a pathophysiological perspective, the mass caused mediastinal compression syndrome, a potentially life-threatening complication requiring urgent management. Critical compression of the great vessels and progressive respiratory difficulty illustrate how large mediastinal masses can mimic or exacerbate cardiovascular disease, affecting haemodynamics and increasing perioperative risk.

In conclusion, mediastinal masses, although uncommon, should be considered in the differential diagnosis of new cardiac murmurs and progressive dyspnoea in young patients, especially when accompanied by systemic symptoms or signs of mediastinal compression. In this case, the mass corresponded to a mediastinal germ-cell tumour with seminomatous morphology and AFP elevation, which determined management as a non-seminomatous tumour requiring chemotherapy. Multimodality imaging allowed staging of the mass and definition of great-vessel and airway involvement, anticipating potentially severe complications such as superior vena cava syndrome or haemodynamic collapse. This underscores the importance of an integrated diagnostic approach that includes clinical assessment, multimodality imaging, and biomarkers to enable timely diagnosis and treatment.

Ethical aspects

Informed consent was obtained from a first-degree relative of the patient for data collection and publication of this case. Confidentiality of the patient's data was ensured at all times.

Author contributions

BGS: conceptualisation, writing—original draft, editing, review, and approval of the final version. **HDC:** conceptualisation, investigation, review, and approval of the final version. **JAG:** writing—original draft, investigation, review, and approval of the final version. **CACP and BIPA:** investigation, review, and approval of the final version. **GV and PRV:** writing—original draft, review, and approval of the final version. **AZC:** review and approval of the final version.

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