A previously well 29-year-old man presented with a 2-week history of progressive orthopnoea, dry cough, neck and facial swelling, and night sweats. On further questioning, he described 2 kg of weight loss and increasing swallowing difficulties over the previous year. An oesophagogastroduodenoscopy performed 1 year prior was unremarkable. A chest radiograph demonstrated a large mediastinal mass (figure 1A), and a subsequent CT scan revealed a large anterior mediastinal mass measuring 11 cm in diameter. There was central necrosis and complete occlusion of the superior vena cava (SVC; figure 1C,D). An urgent biopsy of the mass was performed, and immunohistochemistry showed malignant cells positive for OCT3/4, placental alkaline phosphatase, D2-40 and CD117 but negative for pancytokeratin, S100, CD30, human chorionic gonadotropin (HCG), alpha-foetoprotein (AFP), CD45, CD20 and CD3. There were no masses found elsewhere on CT imaging of the chest, abdomen, pelvis and head, or on testicular ultrasonography. Serum AFP and HCG levels were within normal limits, and lactate dehydrogenase was elevated at 559 U/L. Based on the immunohistochemical and radiological findings, a diagnosis of primary mediastinal seminoma was made. Around 5% of male germ cell tumours (GCT) arise extragonadally.1 2  Fifty to seventy per cent of extragonadal GCTs arise in the anterior mediastinum. 1 2  This patient was treated promptly with three cycles of
Primary mediastinal germ cell tumours (GCT) are very uncommon, almost always found in men, rarely metastasise to solid organs and have a median age at presentation of 33 years. The most common presenting symptom is dyspnoea (in 25% of cases), and superior vena cava (SVC) obstruction is present in 6% of cases. In such cases presenting with SVC obstruction, SVC stent insertion can usually be avoided by the commencement of definitive treatment. Given the potential for masking the competing differential diagnosis of lymphoma, steroids should be used with caution in the acute management of SVC obstruction unless the diagnosis is established. Tumour markers may not be elevated in seminomas, and diagnosis requires pathological and immunohistochemical analysis.

Three cycles of BEP (bleomycin, etoposide and cisplatin) chemotherapy is the accepted standard of care for primary mediastinal seminomas and, given the exquisite sensitivity of seminomas to platinum-based chemotherapy, they carry a favourable prognosis. The 5-year overall survival is ~90%, which compares favourably with the 40%–45% 5-year overall survival rate for non-seminomatous GCTs.

BEP (bleomycin, etoposide and cisplatin) chemotherapy. The symptoms and signs of SVC obstruction disappeared within 2 days of its commencement. A chest radiograph 18 days after treatment initiation showed a dramatic improvement (figure 1B). The patient remains asymptomatic.

Contributors MKT and DML wrote the case history and GJD wrote the case discussion and context. All authors contributed and agreed to the final version.

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References

Learning points
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