CASE REPORT

Devil within: an incidental and rare finding of an extragonadal retroperitoneal germ cell teratoma carcinoid tumour

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SUMMARY

Extragenadal germ cell tumours (GCTs) are uncommon neoplasms comprising approximately 2%–5% of all GCTs. Of these, approximately 30%–40% occurs as primary retroperitoneal GCTs. This case report details the presentation, diagnosis and management of a 53-year-old man presenting with an incidental finding of a primary extragonadal retroperitoneal teratoma with carcinoid transformation.

BACKGROUND

This case represents an unusual presentation of a primary extragonadal retroperitoneal germ cell teratoma with a carcinoid component. Only three cases of such a tumour are documented in the literature to date.

CASE PRESENTATION

A 53-year-old man, and long-term psychiatric inpatient, attending a follow-up appointment at an ear–nose–throat clinic for sinus symptoms reported intermittent constipation. Opportunistic examination revealed a palpable abdominal mass in the right lower quadrant. The patient had no prior history of gastrointestinal or genitourinary pathologies. Medical history was significant for a right parietal lobe haemorrhage, C₂H₅OH excess, Korsakoff's psychosis and refractory depression. Regular medications included significant antipsychotic medications including risperidone, aripiprazole, olanzapine and haloperidol. Subsequent CT imaging was performed to delineate the underlying cause, and the patient was referred to general surgical services for further evaluation.

INVESTIGATIONS

CT of the abdomen and pelvis confirmed the presence of a spherical right-sided retroperitoneal mass, with a cystic component (figure 1). The mass was visualised immediately anterior to the right psoas muscle and the solid component of the mass measuring approximately 5.5×5.3 cm possessed an arterial supply originating from the right common iliac artery. No pathological lymphadenopathy or calcification was observed. The remainder of the study did not demonstrate any abnormalities of the left kidney, spleen and pancreas or prostate. An interval CT of the abdomen and pelvis was performed and this demonstrated the mass to

be stable in size but that small and subtle hepatic metastases had developed, although no metastatic adenopathy was noted. A laparotomy with resection of the retroperitoneal tumour was undertaken. The tumour, although closely applied to the right kidney, is not arising from it (figure 2). There is a definite plane between them, and with careful dissection, the tumour was excised in its entirety (figure 3). Histopathological analysis of the mass demonstrated eosinophilic cells with large oval nuclei and prominent nucleoli in a predominantly reticulated arrangement (figure 4). Immunohistochemistry was performed and lesional cells demonstrated positivity for AE1/AE3, CD56 and synaptophysin while there was focal weak positivity for epithelial membrane antigen. CD117, placental alkaline phosphatase, alpha fetoprotein (AFP), CD30, CD20, cytokeratin 7, TTF-1, thyroglobulin, HepPar-1, inhibin and calretinin were all negative. A proliferation index (Ki67) was reported as <1%. Laboratory investigations revealed elevated catecholamines, chromogranin A and a mildly elevated CA19-9.

DIFFERENTIAL DIAGNOSIS

A wide differential diagnosis was considered including nerve sheath tumour, a small bowel gastrointestinal stromal tumour and sarcoma. Based on the radiological, clinical and histopathological features, the patient was diagnosed with a metastatic germ cell tumour (GCT), comprising teratoma with a large carcinoid tumour arising from the teratoma.

OUTCOME AND FOLLOW-UP

Following diagnosis, laboratory investigations revealed elevated catecholamines, chromogranin A and a mildly elevated CA19-9. A CT of the thorax performed to complete staging did not reveal any obvious pulmonary mass secondary to deposits, but confirmed that hepatic metastases were stable. A ultrasound scan testis did not reveal evidence of malignant testicular involvement. The testes and epididymis were of normal size and morphology. The patient's postoperative course was complicated. He required prolonged high-dependency care for the management of aspiration pneumonias, and three respiratory arrests one of which necessitated two cycles of cardiopulmonary resuscitation. Clinical examination and radiological investigations excluded a cerebrovascular accident as precipitating

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Figure 1 CT image of the abdomen and pelvis demonstrating spherical right-sided retroperitoneal mass with a cystic component. The solid component of the mass measured 5.3 cmx5.5 cm approximately, and possessed an arterial supply from the right common iliac artery.

factor. It was concluded that his recurrent aspirations were due to generalised deconditioning. Antipsychotic medications were held indefinitely, and a percutaneous endoscopic gastrostomy (PEG) tube was sited. He responded well to conservative medical management, intense physiotherapy and speech and language therapy. The patient was discharged to a step-down unit once clinically stable, and although improved, he remained exclusively on PEG feeding. The patient's prolonged postoperative course delayed adjuvant oncological therapy. Further investigations and treatment planning could occur now that he was clinically

Figure 2 This magnified view shows that the tumour, although closely applied to the right kidney, is not arising from it. There is a definite plane between them, and with careful dissection, the tumour was excised in its entirety.

Figure 3 The ex vivo specimen demonstrating the cystic and solid nature, and its size.

stable. He was subsequently referred to the oncology services and somatostatin receptor scintigraphy demonstrated interval development of metastatic carcinoma involving the liver, bone, retroperitoneum and neck. A treatment regime of somatostatin analogues was commenced as the standard therapy for functioning neuroendocrine tumours. The patient was discharged back to a community setting and he continues to make progress and is responding to treatment.

DISCUSSION

Teratomas have been described since antiquity and have attracted interest and wild speculations about their origin.^{1 2} Extragenadal GCTs account for 1%–5% of all GCTs, and some controversy remains as to whether such tumours are metastatic deposits of a viable or burned-out primary gonadal tumour.^{3 4} Indeed, the pathogenesis of extragenadal GCTs remains poorly understood and offered hypotheses suggest that transformed germ cells undergo reverse migration or that GCTs are derived from germ cells that fail to complete normal embryological migration.^{5 6} Teratomas classically appear along the sagittal midline in the sacrococcygeal region and gonadal sites.⁴ Primary

Figure 4 Microscopic appearance of the mass demonstrating a well-circumscribed lesion consisting mostly of a reticulated arrangement of eosinophilic cells containing large oval nuclei with prominent nucleoli. In small areas, these cells are arranged in sheets. In small areas, these cells are arranged in sheets with small foci components of various epithelial components. Immunohistochemistry demonstrated CD56 and synaptophysin positivity.

retroperitoneal tumours represent approximately 1%–11% of all primary extragonadal teratomas, with a bimodal incidence peaking within the first 6 months and third or fourth decade of life.^{1 2 7} Retroperitoneal GCTs typically present once their size exerts a mass effect in vivo, as exemplified by this case. In prior cases, patients presented once the mass effect generated obstructive symptoms, most commonly abdominal pain.^{1 2} Of these prior cases, tumours were most commonly found at the superior pole of the left kidney and approximately 25.8% underwent malignant transformation into carcinoma and sarcoma.^{1 2 6} The appearance of these tumours may be either solid to cystic or a combination of both. Radiological imaging studies demonstrate that calcifications within the tissue mass may be present in approximately 50% of cases, and the most characteristic radiological features of these tumours is the presence of a complex mass with a well-circumscribed fluid volume and fat fluid level.^{1 2 8} Certain radiological features may suggest a diagnosis of GCT; however, histopathological analysis provides a definitive answer. Additionally biochemical markers AFP, CEA, CA 19–9 are occasionally elevated in the presence of retroperitoneal GCTs. Although the clinical utility of these tumour markers is uncertain, elevated AFP levels are known to fall following resection of a tumour and

can reappear with recurrence.^{1 2} Teratomas are largely considered a surgical disease and treatment for these entities involves surgical resection. Subsequent therapy is determined by the findings of histopathological analysis, but chemotherapy and radiotherapy are secondary to surgical intervention. Overall prognosis is variable and dependent on the histopathological features of each tumour, and response to adjuvant chemotherapy or radiotherapy. Following treatment, approximately 50%–69% of patients remain disease-free. Tumours with malignant transformation have a poorer prognosis, particularly if associated with metastasis and all patients need to be closely monitored for malignancy.^{1 2 9}

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Learning points

- ▶ Primary extragonadal retroperitoneal tumours are rare tumours that often present incidentally or when obstructive symptoms provoke investigation.
- ▶ The most characteristic radiological features of these tumours is the presence of a complex mass with a well-circumscribed fluid volume and fat fluid level on CT imaging.
- ▶ Primary surgical resection is considered to be the most appropriate treatment, and any adjuvant chemotherapy or radiotherapy is dependent on histopathological subtype.
- ▶ Prognosis can vary and up to 69% of patients remain disease-free. Tumours with malignant transformation often require chemotherapy and response to treatment can be monitored with tumour markers. Tumours with metastatic presentations are associated with poorer outcomes.

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