

Intraabdominal desmoplastic small round cell tumour mimicking metastatic adenocarcinoma

Introduction

Desmoplastic small round cell tumour (DSRCT) is a rare, extremely uncommon and highly aggressive variety of malignant neoplasm. Males are more commonly affected than females, with a male to female¹ ratio of 3:1, predominantly affecting adolescents and young adults particularly in their second to third decade of life. The cell of origin is thought to be a progenitor cell with multi phenotypic expression. This tumour has a predilection for serosal surfaces. Multiple primary sites have been described however, DSRCT most commonly presents as a multicentric abdominal masses and can resemble metastatic adenocarcinoma. Clinical signs and symptoms are nonspecific so as the radiological and histopathological findings which are only suggestive of but not specific of DSRCT and the diagnosis is usually asserted by Immunohistochemistry.² We here by describe a rare case of DSRCT of abdomen mimicking metastatic adenocarcinoma.

Case presentation

Clinical history and examination

A 38years old male presented to our hospital OPD with complaints of something moving in the abdomen for the past two months. On physical examination, there was a palpable lump in the right periumbilical area about 5x4cm in size, which was non tender, mobile, firm in consistency and smooth margins. Rest of abdominal examination was normal. The routine haematological and biochemistry investigations were within normal limits.

Imaging

On ultrasound abdomen, there was an irregular hypoechoic lesion of size 6.8x4.8cm in the right para aortic region with an evidence of calcification within it. On contrast enhanced computerized tomography (CECT) abdomen, there was a heterogeneously enhancing soft tissue mass arising from mesentery of about 9x5.3x5.6cm with evidence of calcification within it.

Surgical treatment

Patient underwent exploratory laparotomy. Intraoperatively, there were multiple nodules of different sizes present in mesentery of transverse colon, ileocaecal region and right side of parities suggestive of metastatic disease but no primary origin was seen. Largest mass of approximately 8x6x6cm size present in the mesentery of transverse colon was excised and sent for frozen section intraoperatively. Liver and rest of the viscera were normal. There was no ascites. A diagnosis of metastatic adenocarcinoma was made initially on frozen imprints. Postoperatively, patient was discharged in satisfactory condition awaiting histopathology examination report.

Pathological findings

Gross examination of the specimen revealed multiple variably sized nodules studded in the mesenteric fat. The nodules varied in diameter from 0.4cm to 7.0cm. Cut-section of these nodules was solid, grey white to grey brown and firm in consistency (Figure 1). On

frozen section examination, a diagnosis of metastatic adenocarcinoma was rendered based on clinical and gross findings, clustering of tumour cells, atypical nuclei and myxoid material in the background that was misinterpreted as mucin. Hematoxylin and eosin (H&E) staining on paraffin embedded tissue showed these nodules to be composed of a tumour showing sharply circumscribed aggregates of tumour cells. These aggregates varied in size and shape from being singly scattered to tiny clusters to irregularly shaped islands. Tumour stroma was desmoplastic and showed areas of myxoid change. The tumour cells were small with round to oval hyperchromatic nuclei, scanty cytoplasm, indistinct cell borders and showed high mitotic activity (Figure 2). Immunohistochemistry showed the tumour cells to have strong cytoplasmic positivity for desmin, vimentin and cytokeratin (Figure 3). Thus, the final histopathological diagnosis made was intra-abdominal desmoplastic small round cell tumour which mimicked metastatic adenocarcinoma.



Figure 1 Gross showing cut section of the mass.

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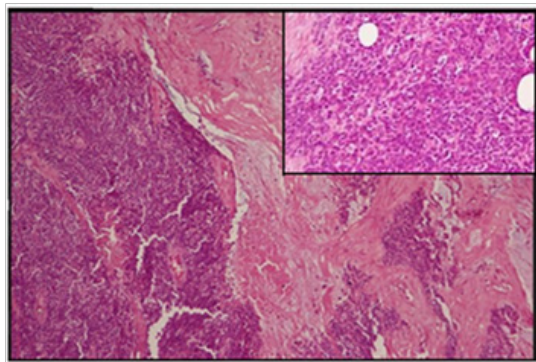


Figure 2 Photomicrograph showing nests of small round tumour cells in desmoplastic stroma (H&E x40) Inset, small round tumour cells with hyperchromatic nuclei (H&E x400).

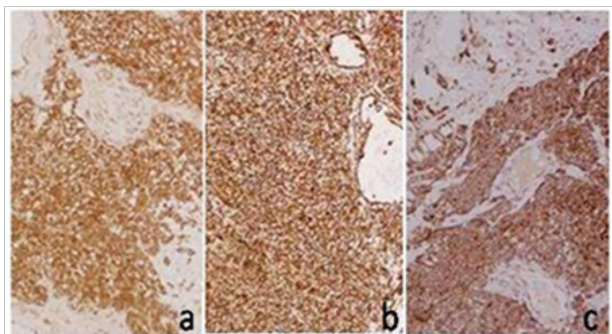


Figure 3 Photomicrograph immunohistochemistry DSRCT.

Figure 3A Tumour cells positive for vimentin.

Figure 3B Tumour cells positive for desmin.

Figure 3C Tumour cells positive for cytokeratin.

Discussion

DSRCT is a rare, aggressive tumour having predilection towards the peritoneum and mesothelium lined surfaces and primarily occurs as masses in the abdomen. The tumour generally presents as extensive intra abdominal or less often endopelvic mass with widespread peritoneal and lymphatic dissemination, without an apparent organ of origin.² The usual clinical features are nonspecific abdominal complaints and patient generally presents late in the course of the disease, with many people already harbouring metastatic disease on presentation. Presentation may include abdominal pain, distension, lack of appetite, palpable abdominal lump which are nonspecific and non-diagnostic.³ The most important imaging tool is CECT scan which may show heterogeneous bulky peritoneal masses without any apparent organ of origin with mesenteric lymphadenopathy.² Fine needle aspiration cytology has also been used for the diagnosis but still it's not the most preferable way, since molecular cytogenetics require larger biopsies.^{4,5} Macroscopically, the tumour presents as boss elated grey surfaces with areas of necrosis. Pathology reveals clusters of small to medium sized cells with hyperchromatic nuclei and increased nuclear/cytoplasmic ratio, surrounded by a dense desmoplastic stroma.⁶ Hattori et al suggested that prominent cytoplasmic vacuoles and cohesion in serous effusions of DSRCT may resemble metastatic adenocarcinoma.⁷ DSRCT occurs rarely in females and can masquerade ovarian cancer⁸ Zeeshan-ud-din et al.⁹ reported two cases of intra-abdominal desmoplastic small cell tumour

resembling adenocarcinoma of colon.⁹ By virtue of its resemblance both clinically and radiologically with metastatic adenocarcinoma, the final diagnosis is only based on immunohistochemistry which demonstrate epithelial markers like cytokeratin, mesenchymal markers like vimentin, neuronal elements like neuron specific enolase and myogenic elements like desmin and cytogenetic abnormality which in most cases is a reciprocal translocation involving t(11;22) (p13;q12), whose product is a EWS/WT1 (Ewing sarcoma/Wilm's Tumour) transcript, that mainly act through ENT4(Equilibrium nucleoside transporter).

The ideal therapeutic strategy is the debulking surgery to reduce the majority of the tumour bulk. However complete removal cannot be done because of extensive peritoneal deposits regarding the aggressiveness of the disease, treatment is based on multi-modal therapy. It has been reported that the combination of surgery, chemotherapy with or without additional radiotherapy have shown best results leading to an overall response rate of 39% and a 3-year survival rate of approximately 50%^{6,10} as compared to each modality used separately. Despite these strategies, the overall survival remains dismal.

Conclusion

DSRCT is an aggressive neoplasm, despite disseminated disease at the presentation there are very few warning signs. Since these tumour mimics metastatic adenocarcinoma of abdominopelvic organs so we must keep a high degree of suspicion to diagnose these tumours early. So that early debulking surgery with multimodal treatment can be started as soon as possible in order to improve the prognosis and final outcome.

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Conflict of interest

The author declares no conflict of interest.

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