CASE REPORT

Suspected hydatid cyst of liver harbouring an aggressive desmoplastic small round cell tumour

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SUMMARY

Among the group of small round cell tumours, there is a distinct and rare tumour known as desmoplastic small round cell tumour (DSRCT). DSRCT presents as multiple, widespread masses in the abdomen and pelvis and may be accompanied by extensive tumour implants throughout the peritoneum as the tumour is known to spread diffusely along serosal surfaces. We discuss a case of DSRCT in a 16-year-old boy who presented with abdominal pain since 2 years, a non-tender mass was palpable on the right upper quadrant of the abdomen, ultrasonographic and CT findings suggested hydatid cyst of liver. Laparotomy revealed multiple small peritoneal deposits along with a single mass in the liver. On histopathology, the lesion was found to be neoplastic and composed of predominantly clusters of small round blue cells, in a desmoplastic stroma; tumour cells were diffusely positive for cytokeratin, vimentin and neuron-specific enolase, thus confirming the diagnosis of DSRCT.

BACKGROUND

Desmoplastic small round cell tumour (DSRCT) was first described by Sesterhenn *et al*¹ only two decades ago. The tumour is found predominantly in adolescent and young adult males.² Although the tumour is known to spread widely, the organ of its origin is usually unknown.³ The biological behaviour, histogenesis and genetic characteristics of the tumour are yet to be elucidated. The tumour shows immunohistochemical evidence of epithelial, mesenchymal and neural differentiation.⁴ A specific chromosomal translocation t(11,22)(p13,q12) involving two genes: EWS gene which is characteristic of Ewing's sarcoma and WT1 gene which is Wilms tumour 1 gene, has been documented in DSRCT.⁵

We are discussing here the case of DSRCT of liver, because of the rarity of this tumour and the widespread involvement presents as a diagnostic difficulty, emphasising a multidisciplinary approach in its diagnosis and management involving clinical suspicion, radiological, histopathological, immunohistochemical and cytogenetic studies.

CASE PRESENTATION

A 16-year-old boy of average build presented with pain in the right upper quadrant, weight loss and anorexia for past 2 years. There was no significant medical history, and he did not have any family history of cancer. On palpation, a non-tender mass was palpable in the right upper quadrant of the abdomen.

On account of the radiological investigations, a hydatid cyst of liver was suspected, and the patient underwent laparotomy which revealed widespread multiple small peritoneal deposits of tumour along with a single mass lesion in the liver. The histopathological and immunohistochemical findings guided us to correctly diagnose the tumour as DSRCT.

INVESTIGATIONS

On abdominal ultrasonography, a multiloculated cyst within cyst was identified in liver. CT of the abdomen revealed hepatomegaly with a heterogeneous, predominantly hypodense superficial lesion with multiple internal septations and dense matrix, involving segment 2 of the left lobe of the liver (figure 1).

The mass from the liver was resected and submitted for histopathological examination. Histopathology of the specimen showed the lesion to be neoplastic and composed of predominantly clusters of small round cells with scant cytoplasm. Nuclei were of small size and pleomorphic, hyperchromatic with inconspicuous nucleoli. The tumour stroma showed marked desmoplastic response around the tumour cell clusters (figure 2A, B). Immunohistochemical studies showed positivity for pan-cytokeratin (figure 3), vimentin (figure 4) and neuron-specific enolase (NSE; figure 5).

DIFFERENTIAL DIAGNOSIS

DSRCT belongs to the group of small round cell tumours of infancy and childhood. The group includes Ewing's sarcoma, primitive neuroectodermal tumour, embryonal or alveolar rhabdomyosarcoma, neuroblastoma, malignant lymphoma, Askin's tumour and rhabdoid tumour. Almost all

Figure 1 Contrast-enhanced CT scan showing involvement of the segment 2 of the left lobe of the liver.

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Figure 2 (A) Small round tumour cells in a desmoplastic stroma, few cords of hepatocytes are also seen (H&E, $\times 100$). (B) Clusters of small round blue tumour cell with scant cytoplasm surrounded by desmoplastic stroma (H&E, $\times 400$).

these tumours are characterised by small uniform cells with sparse cytoplasm, diffuse growth pattern and high cellularity. Widespread involvement, specific histopathological and immunohistochemical features are helpful in distinguishing DSRCT from other tumours.

Figure 3 Pan-cytokeratin positivity by tumor cells (pan-cytokeratin, $\times 100$).

Figure 4 Vimentin positivity by tumour cells (vimentin, $\times 100$).

TREATMENT

The patient underwent laparotomy, with the resection of liver mass.

OUTCOME AND FOLLOW-UP

Postoperative period was uneventful. Currently, the patient is in follow-up and doing well after 3 months of surgery.

DISCUSSION

DSRCT is a distinct and rare tumour which belongs to the group of small round cell tumours of infancy and childhood. The tumour presents with a mean age of 18–20 years (range 3–48),⁶ and >70% of patients present before the age of 30.⁷ The most common presenting symptoms of DSRCT are abdominal pain and distention, related to tumour mass effect and sometimes due to ascites also. The tumour has a predilection for the serosal surfaces without an obvious visceral primary site. An omental or pelvic mass is another common presentation along with multiple smaller satellite nodules adherent to the peritoneum.

Grossly, the tumour is firm, smooth and bosselated, with gray-white cut surface and focal necrotic and haemorrhagic areas.⁸ Histologically, DSRCT is characterised by well-defined groups or strands of uniform small round tumour cells surrounded by an abundant stroma showing desmoplastic reaction. Usually the

Figure 5 Neuron-specific enolase (NSE) positivity by tumour cells (NSE, $\times 100$).

Learning points

- ▶ Desmoplastic small round cell tumour is a rare tumour having specific histological and immunocytochemical features; it usually involves the peritoneal cavity of adolescent and young adult males with widespread peritoneal tumour implants.
- ▶ The characteristic cytological and histological features along with the polyphenotypic immunostaining pattern are helpful in diagnosing this rare aggressive tumour so that optimal therapeutic intervention is started at the earliest.
- ▶ Radiological investigations may sometimes be misleading, as in our case and a high index of suspicion by the pathologist is necessary while reviewing cases of abdominal mass, specially having multiple satellite lesions in young and adult males for correct diagnosis of this rare tumour.
- ▶ Cytogenetic karyotypic analysis and molecular studies are helpful additional investigations for arriving at the correct diagnosis in problematic cases.

cellular and stromal components in a particular tumour are nearly equal but the amount of stroma varies greatly with some tumours being predominantly cellular and others with a predominantly desmoplastic stroma. Rarely myxoid areas, cystic degeneration and calcification may be seen in the stroma. On immunohistochemistry, DSRCT express simultaneously markers of epithelial (keratin and epithelial membrane antigen), mesenchymal (vimentin), myogenic (desmin) and neural (NSE and CD56) tissues. Simultaneous expression of epithelial, mesenchymal and neural antigens suggests that tumour cells may have a pluripotent stem cell origin. Cytogenetic and molecular analysis have revealed a typical t(11;22)(p13;q12) translocation. This translocation involves fusion of the EWS gene on chromosome 22 with the WT1 on chromosome 11. EWS/WT1 transcript is diagnostic of this tumour and codes for a protein that acts as a transcriptional activator that fails to suppress tumour growth.⁹

The optimal therapeutic strategy for this aggressive neoplasm is still unclear due to its rarity and hence limited experience for treating this tumour. Most reports suggest total resection of the mass and

postoperative combinatorial multiagent chemotherapy and radiotherapy as the best therapeutic option. Practically, total resection is often not possible because of multiple satellite peritoneal implants. Some authors also suggest preoperative chemotherapy to shrink the multiple satellite lesions. Intraperitoneal hyperthermic chemoperfusion has also been administered to reduce the bulk of the tumour. The experience with pseudomyxoma peritonei is behind this concept. Despite the combined therapeutic approach, including surgery and chemoradiation therapy, the prognosis for these patients remains very poor, especially for those with metastatic disease—29% for a 3-year survival and just 18% for a 5-year survival.¹⁰

Contributors KA and FA diagnosed the case. SQ and WMA collected the relevant literature. FA and SQ prepared the manuscript.

Competing interests None.

Patient consent Obtained.

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