Case Report ISSN 2835-6276

American Journal of Medical and Clinical Research & Reviews

Desmoplastic small round cell tumor a case report of a rare intra-abdominal tumor of the young adult

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Received: 15 March 2024; Accepted: 17 March 2023; Published: 25 March 2024

Citation: AZHARI Ilias. Desmoplastic small round cell tumor a case report of a rare intra-abdominal tumor of the young adult. AJMCRR 2024; 3(3): 1-5.

ABSTRACT

Desmoplastic small round cell tumor is a rare and highly aggressive mesenchymal tumor described as a distinct clinico-pathological entity in 1989 by Gerald et al (1) . These tumors occur mainly in the peritoneal cavity,although other primary sites, such as testicular, ovarian, thoracic, pulmonary, intracranial and head and neck regions have been reported.

Clinical symptoms are non-specific. Diagnosis is based on histological analysis combined with immunohistochemical study.(11)

Histological study typically shows small round blue cells in nests separated by abundant desmoplastic stroma associated with a single chromosomal translocation t(11:22) (p 13; q 12) involving the EWSR1 and WT1 genes.(11)

The prognosis is particularly poor, with median survival ranging from 17 to 25 months. Management of this tumor remains difficult, and current regimens do not achieve a significant cure rate despite the use of aggressive treatments such as polychemotherapy.

Introduction

Desmoplastic small round cell tumor is a rare malignant tumor usually affecting young, male subjects. It frequently presents as diffuse involvement of the abdominal serosa at the time of diagnosis. The histological appearance is characteristic, and the immunohistochemical profile reveals a tumor with polyphenotypic differentiation. The diagnosis is confirmed by the presence of the specific trans-

location t(11;22) (p13;q12). The prognosis is poor, despite multidisciplinary management including surgery, radiotherapy and high-dose chemotherapy.

Case presentation

We report the case of a 20-year-old male with no particular pathological history, who presented with left hypochondrium pain with no other associated signs.

The patient initially underwent an abdominal CT scan, which revealed an 11 cm long mass between the spleen and the left kidney (Figure 1) and a 14 cm hypogastric mass. (Figure 2)

The patient underwent exploratory laparoscopy with biopsy, revealing a mass in the left hypochondrium adherent to the left colonic angle and the greater omentum, and a hypogastric mass. Pathological examination revealed a tumoral process organized into irregularly rounded basophilic structures of variable size, presenting cytonuclear atypia with images of mitosis, evolving within a well individualized, dense, desmoplastic fibrous connective tissue.

On immunohistochemical study, WT-1 labelling was negative, pan-cytokeratin (AE1/AE3), desmin, anti-CD99, anti-Ki-67 were positive, concluding that the anatomopathological and immunohistochemical appearance was in favor of a desmoplastic small round cell tumor.

The patient subsequently received IE-VAC chemotherapy.

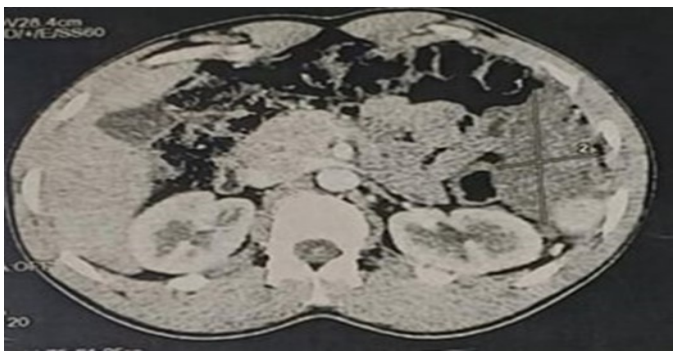

Figure 1: CT scan of the abdomen showing the mass of the left hypochondrium

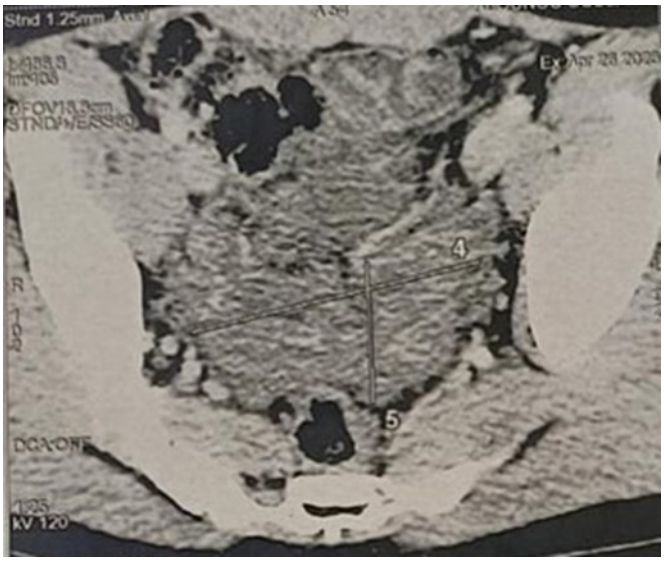

Figure 2: CT scan of the abdomen showing the hypogastric mass

The patient underwent laparotomy with tumor resection of two masses a mass in the left colonic angle and a hypogastric mass with cytoreduction of carcinosis's nodules.

Surgical exploration showed the absence of effusion, the absence of liver metastases, and the presence of nodules of carcinosis with a PCI score of 22, the presence of a 12 cm mass adherent to the left colonic angle and the greater omentum, coming into contact with the spleen, the stomach and the tail of the pancreas without invading them (figure 3), and the presence of a 7 cm mass between the bladder and the rectum (figure 4).

Postoperatively, follow-up care was unremarkable. Patient was declared discharged after 3 postoperative days.

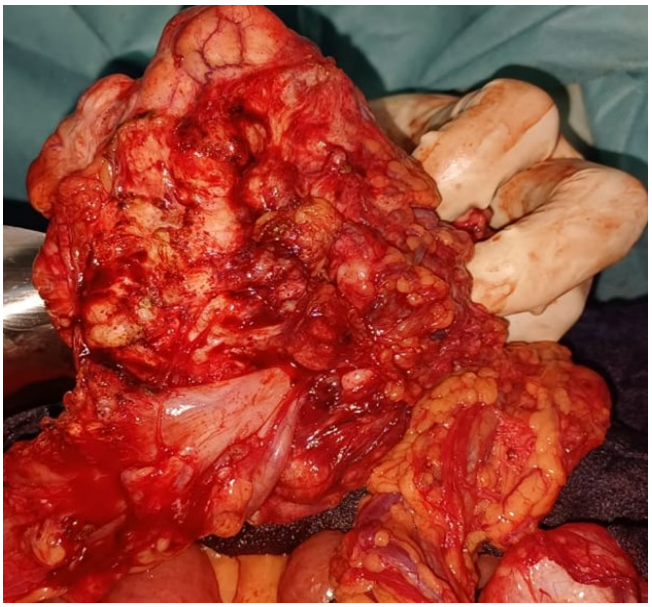

Figure 3 The mass of the left hypochondrium adherent to the left colonic angle and the greater omentum,

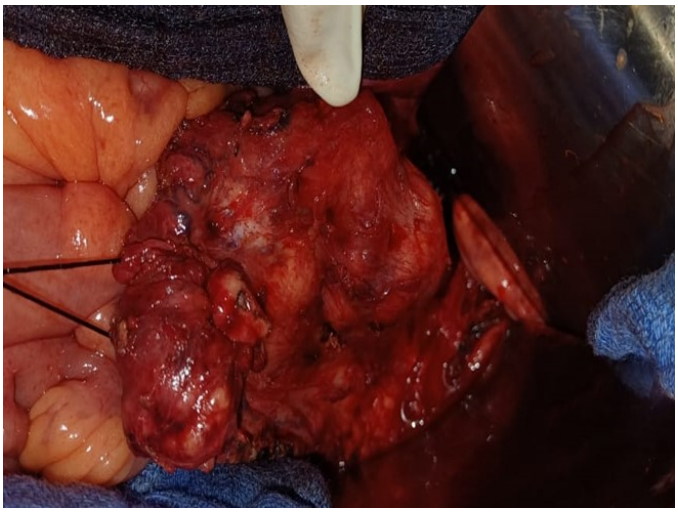

Figure 4 The hypogastric mass between the bladder and the rectum

Discussion

Desmoplastic small cell tumors are rare mesenchymal tumors first described in 1989, affecting adolescent and young adult males (1).

This cancer has a diffuse distribution in the peritoneal cavity and there is no evidence of a primary site. It develops in the abdomen, invades the greater omentum and forms several peritoneal implants (1) and may be metastatic from the outset; extra-abdominal localization is possible but rare (2).

The clinical picture is not specific, the tumor may remain asymptomatic for a long time, and the pathology presents as abdominal distension associated with abdominal pain.

The tumor is associated with a single t(11:22) chromosomal translocation (p 13; q 12) involving the EWSR1 and WT1 genes. The translocation leads to fusion of the two genes with expression of a chimeric EWSR1-WT1 oncogene protein that acts as a transcriptional regulator, modifying gene expression and ultimately allowing tumor growth.

Histologically, the tumor is composed of small, round, atypical, mitotic cells in clusters separated from a desmoplastic stroma (3). The differential diagnosis is made with other round cell tumors: Ewing's sarcoma, neuroblastoma, Wilms's tumor, rhabdomyosarcoma.

On computed tomography (CT), the tumor appears as a single or multiple lobulated peritoneal mass with no organ involvement (4), which may be associated with ascites, liver metastases, adenopathy, tumor calcification or diffuse nodular thickening of the peritoneum (5).Magnetic resonance imaging (MRI) is useful in cases where pelvic and hepatic lesions are present.

The most common site of initial organ metastases is usually the liver. The lungs, pleura and mediastinum may also be sites of metastases.

Treatment involves a multimodal approach with neoadjuvant chemotherapy, surgical resection and radiotherapy. (6)

There is currently no consensus on a standard chemotherapy protocol (6). This chemotherapy is generally based on alkylating agents and is used in combination with a complete surgical excision and postoperative complete abdominal radiotherapy, which has been shown to improve survival.

Several case series have demonstrated the crucial and decisive role of surgery(9)(10). A case study of 100 patients showed that R0 or R1 resection prolonged survival compared with R2 resection or no cytoreduction (7).

Complete surgical resection should not be performed until the response to neoadjuvant chemotherapy has reached a plateau after 4 to 6 months. It should be noted that many lesions will not decrease in size after chemotherapy, but there will be a reduction in tumor vascularization. (1) Intraperitoneal chemotherapy is used by some teams, but has not been shown to improve patient survival (8).

After the end of treatment, progression or recurrence of the disease is to be expected. Despite multimodal treatment with chemotherapy and surgery, median survival varies from 17 to 25 months, with less than 20% of patients achieving 5-year survival (11).

Conclusion

Desmoplastic Small Round Cell Tumor is a rare and aggressive cancer that predominantly occurs in a young male population, with poor diagnosis. The multimodal management is the most efficient approach to treat this cancer although the survival rate is poor.

Acknowledgements: None.

Conflicts of Interest: All the authors have no personal or financial conflicts of interest regard this case report

Funding: No sources of funding to declare.

Consent: Written informed consent was obtained from the patient for publication of this case report and accompanying.

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