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CASE REPORT

A Case Report of Abdominal Desmoplastic Small Round Cell Tumor in a Young Tunisian Woman

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Introduction

Desmoplastic small round cell tumour (DSRCT) is a rare and highly aggressive variety of abdominal sarcoma [1]. It was recently described, in 1989, by Gerald and Rosai [2]. It affects young males, especially adolescents with an annual incidence of 0.1 cases per million [3]. The cell of origin is not yet known. But, a peritoneal origin is commonly admitted [4]. It is postulated to originate from the serosal lining also because it is frequently found in the mesothelial-lined surface, but an epithelial, neurogenic and blastomeric origin are reported [5]. Due to the small number of patients, treatment guidelines have been difficult to define. In this article, we report a case of an intra-abdominal DSRCT in a young adult Tunisian woman and we proceed to clarify the major characters of this rare tumour.

Case Report

A 46-year-old diabetic and hypertensive woman was complaining, for 6 months, about continuous epigastralgia without any particular irradiation or other associated signs. On physical examination, reveal a non-distended abdomen with an oval well-limited epigastric mass of 20 cm in its longest diameter that was painful, fixed to the deep abdominal structures and mobile according to the abdominal wall, with no local inflammatory signs. There were no biological abnormalities and tumour markers (CA19-9, ACE and CA 125) were also normal. Abdominal ultrasound showed a heterogeneous and badly vascularized mass of 13 × 11 × 9 cm with hypoechoic structure and poly-lobed edges (Fig. 1). Esophagogastroduodenoscopy was normal. Abdominal CT-scan showed a large irregular gastric mass of 18 × 17 × 11 cm with supra-mesocolic development. This mass was heterogeneous with central necrosis. The greater omentum was thickened with many heterogeneous adenopathy and ascites of low abundance (Fig. 2). Intraoperative exploration showed an enormous mass of 20 cm developed from the greater omentum with many nodules all around, under diaphragm and colon. This mass was situated between transverse colon and posterior side of stomach. Surgical debulking (reducing as much as possible of a tumour) was performed. Tumour and great omentum were resected without tumoral effusion (Fig. 3). The histopathological examination concluded to desmoplastic small round cell tumour that weight 1.7 kg with many tumour nodules. There was no postoperative morbidity. But after 5 weeks, the tumor has enhanced according to a second abdominal CT-scan showing multiple peritoneal nodules. The patient was healthy, so she underwent palliative chemotherapy based on cyclophosphamide, doxorubicin and vincristine (HD-CAV) for 3 cycles. Abdominal CT-scan has shown tumour necrosis with a progression of tumour size (Fig. 4). A second chemotherapy regimen consisting of ifosfamide and etoposide was indicated.

Discussion

DSRCT is a recently rare tumour. It predominates in pediatric age and young adults with male predominance. In 73% of cases, the age ranged from 10 to 16 years [6]. Diagnosis of intra-abdominal DSRCT is difficult and it is usually done at an advanced stage [7]. Typically, the tumour produces few symptoms until it is large enough to compress or invade

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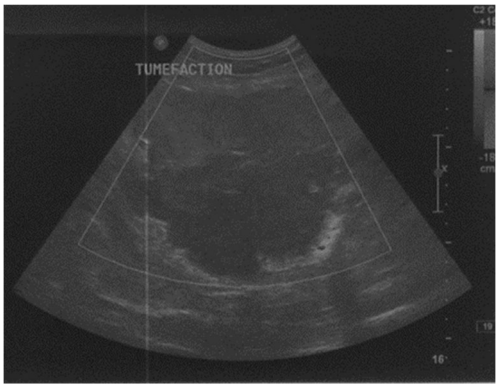

Fig. 1 Abdominal ultrasound echography showed a heterogeneous hypoechoic and badly vascularized mass of 13 × 11 × 9 cm with polylobed edges

surrounding structures. Symptoms include abdominal discomfort/distension, abdominal pain or change in bowel habits associated with weight loss. It is rare for a patient to present with one or two masses. Even in specialized sarcoma centre, it is difficult to diagnose this rare tumour. About 15% of patients referred to specialized sarcoma centre had started treatment for another type of tumour [8]. Imaging features are non-specific that can overlap the appearance of DSRCT. Then gastrointestinal and intravenous contrast is useful. Abdominal CT often shows a large abdominopelvic mass, heterogeneous, a seat of haemorrhage, necrosis and/or calcification. This mass is usually associated with multiple peritoneal implants and metastatic lesions essentially hepatic and lung localization [6]. A magnetic resonance imaging (MRI), the tumour has a heterogeneous high signal intensity and for a signal sequence T2 and hypointense in T1 sequence [9]. MRI is also helpful in cases of pelvic and hepatic lesions. Therefore, positon emission tomography (PET) can better evaluate distant metastasis at the time of staging [10]. However, only histological analysis with an immunohistochemical and cytogenetic study may confirm the diagnoses [11]. The cytological study shows small separated blue cells, round, oval or speculated with little cytoplasm, irregular nuclear membrane, granular chromatin and inconspicuous nucleolus associated to desmoplastic stroma [12]. DSRCT have a complex immunohistochemical and ultrastructural profile reflecting with divergent and simultaneous multidirectional phenotypic [2]. They express simultaneously antigens of epithelial tissues (cytokeratin and EMA), mesenchymal (vimentin), muscle (desmin) and neuroendocrine (NSE). Gerald and Rosai are the first to identify a specific translocation t(11;22)(p13;q12), which generate an active fusion protein involving the Ewing sarcoma (EWS) and Wilms tumour (WT-1) genes. If this fusion protein cannot be identified, the diagnosis of DSRCT cannot be made [2]. The most frequently raised biological tumour marker is CA125, due to the serosal tumour infiltration [5] but in our case it was negative. Due to its rarity, therapeutic options of abdominal DSRCT are not well documented. Regarding its aggressiveness, treatment is based on multimodal therapy including an extensive surgery if it is feasible, chemotherapy-associated or not to abdominal radiotherapy [11]. Most patients consulted when the tumour is disseminated due to its clinical latency. Typically, omental disease, peritoneal studding on the diaphragm, spleen, Morrison's pouch, abdominal wall peritoneum, small bowel and colonic diseases are found at initial presentation. Therefore, curative treatment is extremely difficult. In these cases, for complete resection and cytoreduction, peritonectomy is required.

The impact of cytoreductive surgery on survival remains unclear. La Quaglia et al. found a 3-year overall survival of 58% with complete resection, and it was of 0% when patients are treated only with chemotherapy and radiotherapy without resection [1]. Some authors suggest an improvement in survival with surgery with a median survival of 34 months compared to 14 months for an operable and inoperable tumour, respectively [13]. Shown Schwartz has found a complete response, in 13 patients among a total of 15 for which a resection of 90% of a tumour has been possibly associated with chemotherapy and radiotherapy [14].

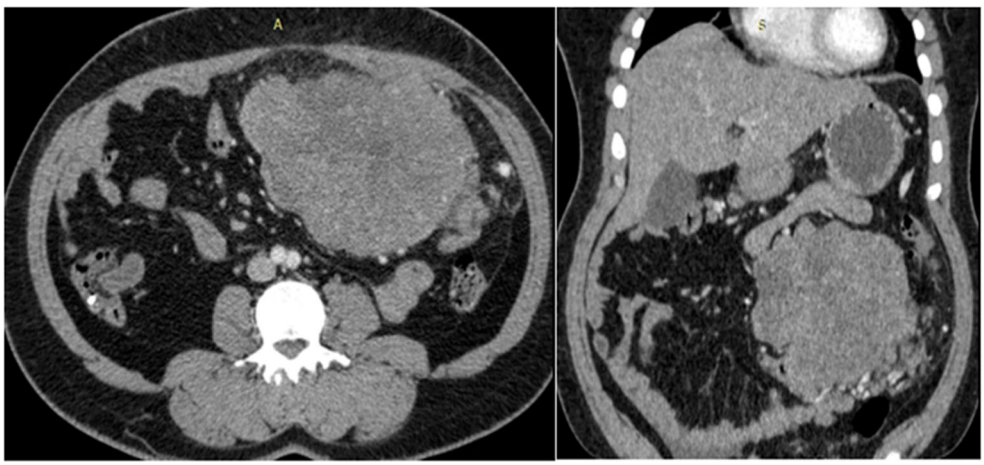

Fig. 2 Axial and coronal abdominal CT-scan showed a large irregular gastric mass of 18 × 17 × 11 cm. This mass was heterogeneous with a minimal central necrosis. It was associated to an all-around densification of the greater omentum with ascites and many heterogeneous adenopathies

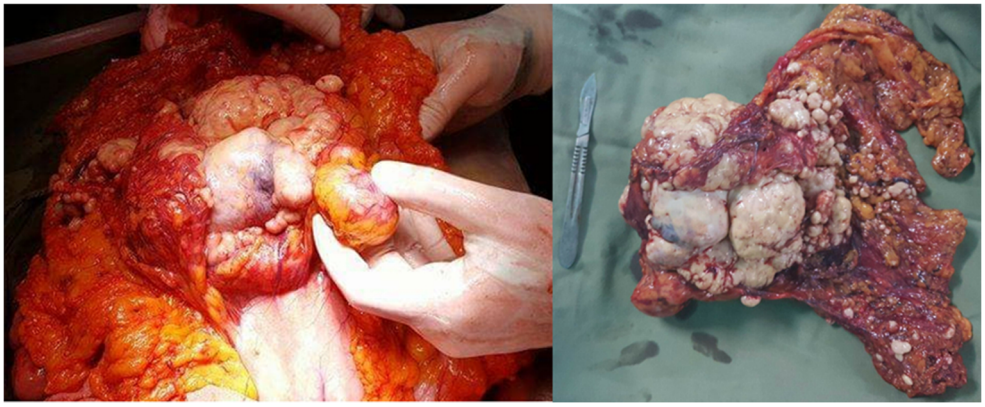

Fig. 3 Per-operative picture showed an enormous mass of 20 cm depending on the great omentum

DSRCT are sensitive to chemotherapy. Indeed, control of tumour progression can be achieved with an aggressive multifaceted treatment approach, consisting of chemotherapy (P6 protocol: cyclophosphamide, doxorubicin, vincristine, ifosfamide and etoposide) followed by surgery [14]. Kushner at Memorial Sloan Kettering Cancer Center has used P6 protocol followed by local radiotherapy or by the new cure of chemotherapy based on thiotepa to myelosuppressive dose with conservation of stem cells. In that series of 12 patients, 7 were alive and free of disease with recurrence between 21 and 60 months. The 5 other patients have presented a progression of their disease in a period of 13 to 26 months [15]. In another study, Lippe suggests carboplatin, etoposide and doxorubicin as the first-line chemotherapy drugs followed by CD34+ stem cell mobilisation with high-dose cyclophosphamide (7 g/m^2) or with cytoreductive surgery [16]. In our case, we had to deal with fast tumour enhancement after cytoreductive surgery by high-dose chemotherapy. But we had obtained tumour progression.

Radiotherapy in DSRCT is controversial; some researchers conclude that a whole abdominopelvic irradiation (WAPI) in DSRCT has a certain effect [17]. It can be used for unresectable DSRCT. Its impact on overall survival remains to be determined. Our patient was not treated with radiation therapy. Its prognosis is quite poor with five-year overall survival estimated at only 15 to 30% [14, 18, 19]. A complete surgical resection remains the major prognosis factor.

The treatment of DSRCT is still a clinical challenge taking into account the absence of management strategies to evaluate the effects of current treatment for such rare tumour. Majority of research concluded that for patients with desmoplastic tumours with small round cells without extraperitoneal metastases, there was one multimodal approach combining perioperative chemotherapy, macroscopically complete resection and radiotherapy, and that there was no benefit to combining intraperitoneal chemotherapy (CHIP or CIPPI) at surgery. But for patients with extraperitoneal metastases, surgery does not seem to bring any benefit and an exclusive chemotherapy is then recommended. But bad results of all these treatments modalities reclaim the development of novel treatment agents in order to improve prognosis.

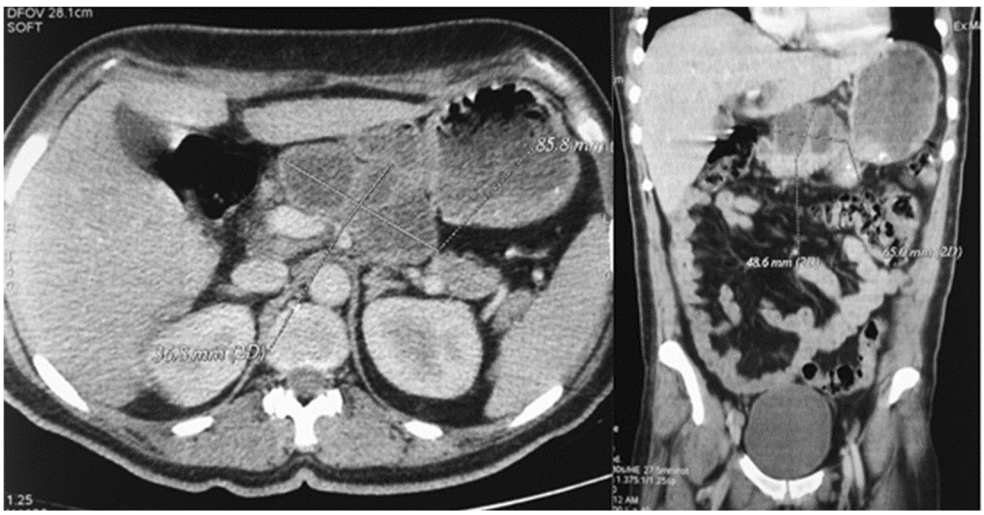

Fig. 4 Axial and coronal abdominal CT-scan after 3 cycles of (HD-CAV) chemotherapy showing multiple abdominal nodules and masses: The lesser omental sac mass is the largest with large central necrosis. It has enhanced considering pre-chemotherapy CT-scan, from 54 × 38 mm to 86 × 37 mm

Compliance with Ethical Standards

Conflict of interest The authors declare that they have no conflict of interest.

References

1. La Quaglia MP, Brennan MF. The clinical approach to desmoplastic small round cell tumor. Surg Oncol. 2000;9(2):77–81.
2. Gerald WL, Rosai J. Case 2 desmoplastic small cell tumor with divergent differentiation. Pediatr Pathol. 1989;9(2):177–83.
3. Worch J, et al. Racial differences in the incidence of mesenchymal tumors associated with EWSR1 translocation. Cancer Epidemiol Prev Biomark. 2011;20(3):449–53.
4. Kim J-W, et al. A case of desmoplastic small round cell tumor diagnosed in a young female patient. Cancer Res Treat. 2009;41(4):233.
5. Al Bouzidi A, et al. Tumeur desmoplastique intra-abdominale à petites cellules rondes (TDPCR): une nouvelle entité. Med Maghreb. 1998;68.
6. Jellouli M, et al. Tumeur desmoplastique à petites cellules rondes intra-abdominale de l 'enfant: à propos d 'une observation et revue de la littérature. Bull Cancer. 2003;90(12):1063–6.
7. Hayes-Jordan A, Anderson PM. The diagnosis and management of desmoplastic small round cell tumor: a review. Curr Opin Oncol. 2011;23(4):385–9.
8. Angarita F, et al. Clinical features and outcomes of 20 patients with abdominopelvic desmoplastic small round cell tumor. Eur J Surg Oncol. 2017;43(2):423–31.
9. Chouli M, et al. Intra-abdominal desmoplastic small round cell tumors: CT findings and clinicopathological correlations in 13 cases. Eur J Radiol. 2005;54(3):438–42.
10. Zhang W, et al. FDG-PET/CT imaging findings of abdominopelvic desmoplastic small round cell tumors: correlation with histopathological findings. Eur J Radiol. 2010;20(2257):e2264.
11. Stuart-Buttle C, et al. Desmoplastic small round cell tumour: a review of literature and treatment options. Surg Oncol. 2008;17(2):107–12.
12. Presley AE, et al. Cytology of desmoplastic small round-cell tumor. Cancer Cytopathol. 2007;111(1):41–6.
13. Biswas G, et al. Desmoplastic small round cell tumor: extra abdominal and abdominal presentations and the results of treatment. Indian J Cancer. 2005;42(2):78.
14. Gerald WL, et al. Clinical, pathologic, and molecular spectrum of tumors associated with t (11; 22)(p13; q12): desmoplastic small round-cell tumor and its variants. J Clin Oncol. 1998;16(9):3028–36.
15. Kushner BH, et al. Desmoplastic small round-cell tumor: prolonged progression-free survival with aggressive multimodality therapy. J Clin Oncol. 1996;14(5):1526–31.
16. Lippe P, et al. Desmoplastic small round cell tumour: a description of two cases and review of the literature. Oncology. 2003;64(1):14–7.
17. Goodman KA, et al. Whole abdominopelvic radiotherapy for desmoplastic small round-cell tumor. Int J Radiat Oncol Biol Phys. 2002;54(1):170–6.
18. Park BJ, et al. Treatment of primary peritoneal mesothelioma by continuous hyperthermic peritoneal perfusion (CHPP). Ann Surg Oncol. 1999;6(6):582–90.
19. Ladanyi M, Gerald W. Fusion of the EWS and WT1 genes in the desmoplastic small round cell tumor. Cancer Res. 1994;54(11):2837–40.