CLINICAL CASE

Paratesticular Desmoplastic Tumor: Case Report and Review of Literature

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KEYWORDS

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Abstract Desmoplastic small round blue cell tumor is a rare neoplasm of the mesenchymal tissue. It occurs in adolescents and young adults with a median age of 22 years. Characteristically, it affects the intra-abdominal area, although other locations, including the paratesticular area, have been described in recent years. Usually, it has an aggressive clinical evolution with multiple recurrences, invasion to adjacent structures, and distant metastasis. **Case presentation:** We report the case of a 23-year-old male with symptoms of pain and testicular volume enlargement who underwent radical right orchiectomy with pathology report of desmoplastic small round blue cell tumor. He received intravenous chemotherapy with the vincristine, Adriamycin, cyclophosphamide/ifosfamide and etoposide regimen for 17 courses, with subsequent recurrence at the inguinal and retroperitoneal level, and surgery plus post-operative radiotherapy was therefore carried out. At the conclusion of radiotherapy, he progressed at the systemic, pulmonary, and hepatic levels and died due to multiple organ failure. (creativecommons.org/licenses/by-nc-nd/4.0/).

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INTRODUCTION

Desmoplastic small round blue cell tumor is a rare and highly aggressive neoplasm of the mesenchymal tissue. It occurs in adolescents and young adults with a median age of 22 years¹. Typically, it develops in the abdominal cavity (95%), whereas only 5% occurs in other sites, including the paratesticular region². It is a highly aggressive neoplasm, with clinical evolution characterized by multiple recurrences, invasion to adjacent structures, and distant metastasis, mainly to the lymph nodes and the lung¹.

We present the case of a patient with a desmoplastic paratesticular tumor treated with multimodal therapy where an aggressive course of disease was observed.

CASE PRESENTATION

This is the case of a 23-year-old male patient with a history of smoking since 16 years of age at a rate of three cigarettes per day and social alcoholism. He had a one-month history of progressive right testicular enlargement associated with pain. Testicular ultrasound revealed a tumor of solid appearance on the right hemiscrotum, separated from the testis (paratesticular). Laboratory tests reported Hb: 17.6, WBC: 6,300, platelets: 301,000, LDH: 312 IU/L, AFP: 1.6 ng/ml, HCG: 0 mIU/L.

The patient underwent right radical orchiectomy and inguinal exploration, with findings of a paratesticular tumor of approximately 7 x 5 cm, rock-hard in consistency, and with irregular borders, involving the spermatic cord and its elements, in addition to right testicle upper pole. The histopathological report corresponded to a desmoplastic small round blue cell tumor of the spermatic cord; surgical margins were negative. Immunohistochemistry showed positivity to vimentin and WT-1; positivity to NSE, desmin, CD56 and Bcl-2; and negativity to ALK-1, chromogranin, myeloperoxidase and S100 (Figs. 1 and 2).

Chest, abdomen and pelvic CT scan showed no distant disease.

Adjuvant treatment was then started with intravenous chemotherapy based on Ewing's sarcoma-extrapolated VAC/IE

Figure 1. Light microscopy.

Figure 2. Immunohistochemistry study.

regimen, with vincristine 2 mg/m² (maximum dose, 2 mg), doxorubicin 75 mg/m², cyclophosphamide 1,200 mg/m², actinomycin D 1.25 mg/m² (when the Adriamycin dose of 375 mg/m² was reached) and ifosfamide at 1,800 mg/m² plus etoposide at 100 mg/m² every three weeks for a total of 17 courses (49 weeks).

Disease control studies after treatment were negative. At three months' follow up, there was recurrence at the retroperitoneum and right groin (Figs. 3 A and B).

Right inguinal tumor resection plus right iliac vein grafting was performed, with findings of a 15 x 8 x 6 cm tumor infiltrating the superficial tissue and fascia and spreading towards the Retzius space, with infiltration to the right external iliac artery and vein: unresectable retroperitoneal tumor. The pathology report classified it as a desmoplastic small round blue cell tumor. The patient then underwent radiotherapy (RT) to the inguinal and retroperitoneal region at 50 Gy in 25 sessions. At the conclusion of RT, a subcutaneous nodule was discovered in the mesogastrium; the control computed tomography (CT) scan showed multiple lung and liver metastases (Figs. 4 A and B). The patient evolved with multiple organ failure and died.

REVIEW OF LITERATURE

Among intrascrotal masses, paratesticular tumors account for barely 2%, as opposed to testicular tumors, which account for 98% of cases. The vast majority of paratesticular tumors (70%) are of benign etiology, among which lipoma (66%), adenomatoid tumor, leiomyoma, and fibroma are predominant. Thirty percent are of malignant etiology, out of which 90% correspond to sarcomas, with the most common being leiomyosarcoma (32%), followed by rhabdomyosarcoma (24%), and liposarcoma (20%)^{3,4}. Paratesticular tumors are characterized by slow and painless growth. This neoplasm is more common in male adults between the second and fifth decades of life. It can affect the testicular tunics, the spermatic cord and the epididymis, with the latter being the site of higher occurrence⁵. Desmoplastic small round blue cell tumor is a rare and highly aggressive malignant ne-

Figure 3. A: The arrow shows retroperitoneal lymph node cluster.

Figure 4. A: Lung metastases are observed.

Figure 3. B: The arrow shows right groin lymph node cluster.

Figure 4. B: Liver metastases.

oplasm of the mesenchymal tissue. It was first described in 1989 by Gerald and Rosai. It occurs in adolescents and young adults with a median age of 22 years. The male-to-female ratio is 4:1^{6,7}.

It typically develops in the abdominal cavity (95%), while only 5% occur in other sites, including the paratesticular region, pancreas, retro-orbital region, cranial cavity, lung, head and neck, and salivary glands⁸.

Desmoplastic tumor is considered to be a member of the childhood small round blue cell tumors family together with primitive neuroectodermal tumor (PNET), alveolar and embryonic rhabdomyosarcoma, poorly-differentiated synovial sarcoma, and rhabdoid tumors. Therefore, there is no appropriate classification system for it².

These tumors are typically characterized by an association with the t(11;22) (p13;q12) translocation, which involves WT1 and EWSR1 genes^{4,6}.

Most of these tumors remain asymptomatic until diagnosis. In advanced cases, clinical presentation involves ascites, pain, vomiting, and weight loss.

These are highly aggressive tumors with median survival of less than 12 months without treatment. They have a clinical course characterized by multiple recurrences, invasion to adjacent structures, and distant metastases, mainly to lymph nodes and lungs¹. Differential diagnosis of this en-

tity is mainly with Ewing's sarcoma/PNET, which can show positivity to keratins in frozen sections, but in paraffin-embedded sections, Ewing's sarcoma/PNET is characterized by positivity to CD 99 and vimentin, and negativity to cytokeratins and muscle markers. The t(11;22)(q24;q12) chromosomal reciprocal translocation in Ewing's sarcoma/PNET involves chromosome 11 long arm, unlike the translocation observed in desmoplastic tumor, which involves chromosome 11 short arm¹.

With regard to treatment, the literature mentions that, owing to the t(11;22) translocation, these tumors can be treated as Ewing's sarcoma, with multimodal therapy with surgery, radiotherapy, and chemotherapy being recommended, and median survival of 17-25 months being reached with this treatment, with less than 20% survival at five years^{9,10}.

In an extensive literature review, only few cases of small round blue cell tumors with paratesticular localization have been reported. The largest series reports only 13 cases, two of them with lung metastases at diagnosis, and four with retroperitoneal and inguinal and cervical chain lymph node metastases, with median survival of 16 months with multimodal treatment^{6,11-13}.

In a retrospective analysis of patients treated at the Memorial Sloan Kettering Cancer Center from July 1972 to July

2003, 66 patients with abdomen- or pelvis-located tumors (96%) and with other tumor sites (2% testis, 2% thoracic cavity), 50% with regional lymph node metastasis and 41% with distant metastasis, were treated with multimodal therapy: seven courses of VAC/IE, followed by surgery and consolidation RT at a dose of 30 Gy. Myeloablative chemotherapy with carboplatin followed by autologous transplantation was carried out in 16 patients who failed to respond to the above-mentioned therapy⁵. The results of this retrospective analysis show that a three-year overall survival of 44% is reached with multimodal therapy⁵.

The presented case is worth mentioning owing to the extreme rarity of presentation and torpid evolution, which ended in the patient dying even after aggressive multimodal therapy.

CONCLUSION

Desmoplastic tumor is an extremely rare and highly aggressive neoplasm. The low frequency of its occurrence makes for treatment guidelines to be based on generally retrospective experiences of several groups with individualized treatments, and the results are therefore not comparable, with multiple controversies being generated.

DECLARATION OF INTEREST

The authors did not receive any funding to carry out this article. The authors declare not having any conflicts of interests.

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