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CASE REPORT

Primary desmoplastic small round cell tumor in the left orbit: a case report and literature review

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Abstract
Purpose Desmoplastic small round cell tumor is a rare malignant neoplasm that most often occurs in the abdomen or pelvis of young men. We herein describe a rare case of desmoplastic small round cell tumor arising from the left orbit in a 16-year-old male.
Methods and results A biopsy was performed and the histology showed the nests of tumor cells with small round cell morphology. The tumor cells showed immunopositivity for desmin, CD99, CD56, SMA, NSE, CgA, SYN, Ki67 and vimentin. Fluorescence in situ hybridization study using EWSR1 break-apart probe was positive for EWSR1 gene rearrangement. After complete surgical resection of the tumor, we did not find tumor recurrence or metastasis with one-year follow-up. Furthermore, a review of the relevant English literature has been discussed.
Conclusions In the present study, for the first time, we report a case of desmoplastic small round cell tumor which is located in the orbital region.

Keywords Desmoplastic small round cell tumor · Left orbit · Histology · Immunohistochemistry · Fluorescence in situ hybridization

Introduction

Desmoplastic small round cell tumor (DSRCT) is found predominantly in adolescents and young adults and is much more common in males than in females [1]. Histologically, DSRCT is typified by variably sized, well-defined invasive tumor islands separated by a desmoplastic stroma [2]. Immunohistochemically, the tumor cells express epithelial, mesenchymal and neural markers [3]. The characteristic translocations t(11;22)(p13;q12) with resultant EWSR1-WT1 fusions can be detected by fluorescence in situ hybridization (FISH) [4]. In previous reports, DSRCT has been frequently found in the abdominal cavity and pelvis. In the present report, we for the first time report a case of DSRCT which is located in the orbital region.

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Case presentation

A 16-year-old male was admitted to the Department of Ophthalmology of 3201 Hospital, with a 6-month history of pain swelling and a palpable nodule in his left orbit. He had no relevant personal or family history of malignancy. On physical examination, a palpable, slightly mobile, solid nodule, was found in the superior nasal region of the left orbit. A computed tomography scan revealed a solitary mass of high intensity, 1.5 × 1 cm in size (Fig. 1). An incisional biopsy was performed and tested for tumor cells markers, which were positive for desmin, CD99, CD56, SMA, NSE, CgA, SYN, Ki67 and vimentin (Fig. 2). Microscopically, the tumors consisted of nests of 'small cells,' with scant cytoplasm embedded in a densely fibrotic stroma (Fig. 3a). Furthermore, FISH analysis has identified a recurrent translocation, t(11; 22)(p13; q12) in 15 cells of 50 cells (Fig. 3b). The histological, immunohistochemical and cytological findings supported the diagnosis of a DSRCT. A tumor resection was performed on this patient (Fig. 4), and he was recovered with no recurrence tumor or metastasis after one-year follow-up. A formal informed consent for this case study was obtained from the participant.

Discussion

Desmoplastic small round cell tumor is a rare and aggressive, malignant tumor. To date, more than 200 cases of DSRCT have been reported in English-language publications, while only 15 cases were women [5, 6]. In some previous reports, it has found that 95% of DSRCTs were located in the abdominal cavity and pelvis, while < 5% of cases may affect other organs [7]. Some reports have shown that most common site of DSRCT occurrence is the pelvis (62%) [8]. In the present report, for the first time, we reported a DSRCT arising in the orbital region in the English literature.

Contrast-enhanced CT and magnetic resonance imaging reveal that DSRCT is a mass of heterogeneous density and intensity [9]. A diagnosis of DSRCT is usually made by a combination of the histologic appearance and immunohistochemical staining results. Most DSRCTs coexpress epithelial markers, mesenchymal markers and neural markers [3, 10]. In our case, immunohistochemical staining disclosed negativity for PGP9.5, S-100, EMA, etc. On the other hand, the tumor cells were showing positive marker for desmin, CD99, CD56, SMA, NSE, CgA, SYN, Ki67, and vimentin. Ordonez assessed a variety of epithelial, mesenchymal and neuronal markers in 39 tumors. Cytokeratin has been detected in 94.9% tumors, EMA in 96% tumors, desmin in 100% tumors, vimentin in 81.5% tumors, NSE in 72% tumors, SYN in 15.8% tumors, CgA in 4.5% tumors, WT1 protein in 88.9% tumors, MSA in 15.8% tumors and α-SMA in 18.8% tumors [11]. However, it is unclear the relationship between these markers and the diagnosis, prognosis and treatment of DRSCT. Histologically, DRSCT in our patient shown a nest of small round tumor cells embedded in a dense desmoplastic fibrous stroma. In cytology, DSRCT is associated with a unique chromosomal translocation, t(11;22)(p13;q12), that fuses the N terminus of the EWS gene to the C terminus of the

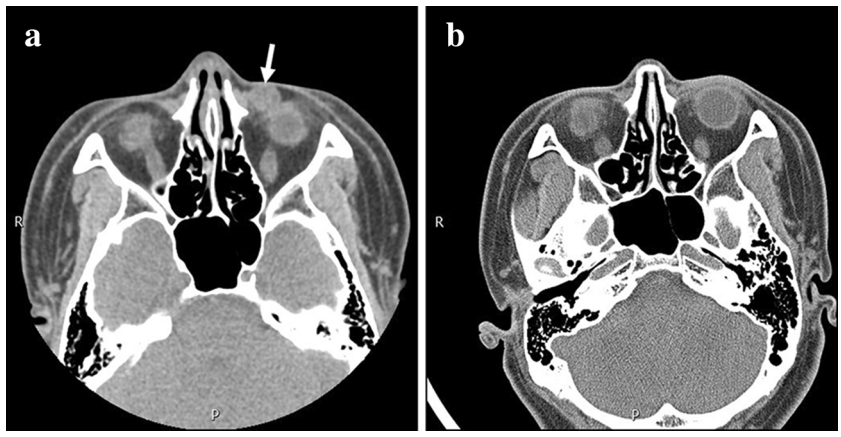

Fig. 1 Computed tomography images of the head at initial presentation (**a**) and 12 months after surgical resection (**b**). Arrows indicate preoperative masses

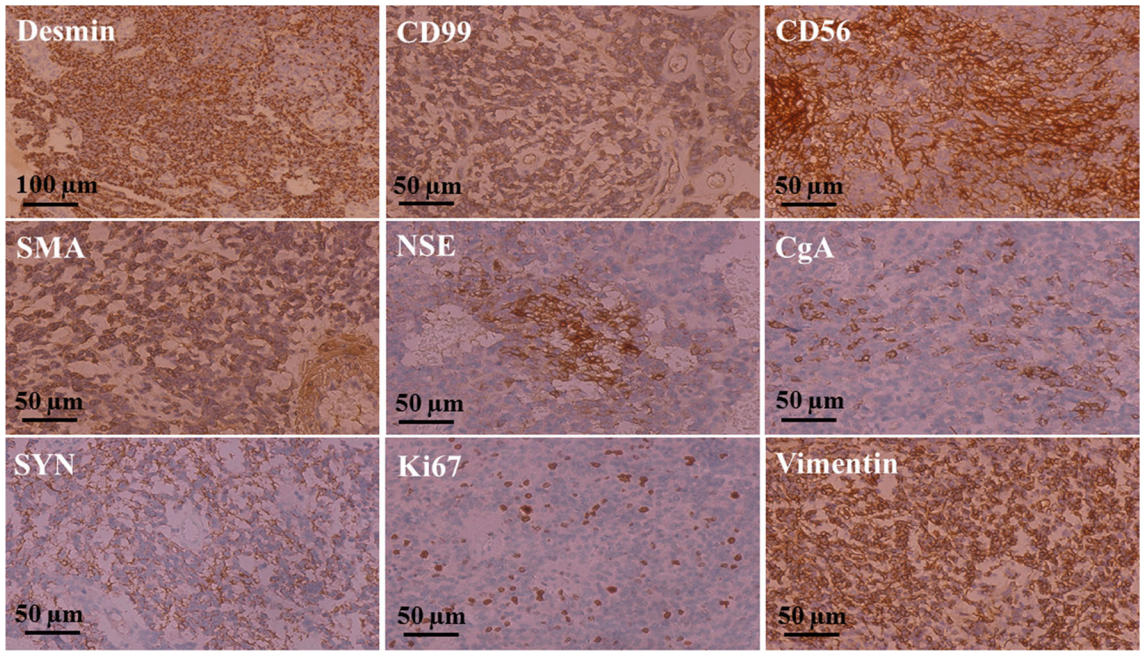

Fig. 2 Immunohistochemical staining shows that the cells were positive for desmin, CD99, CD56, SMA, NSE, CgA, SYN, Ki67 and vimentin

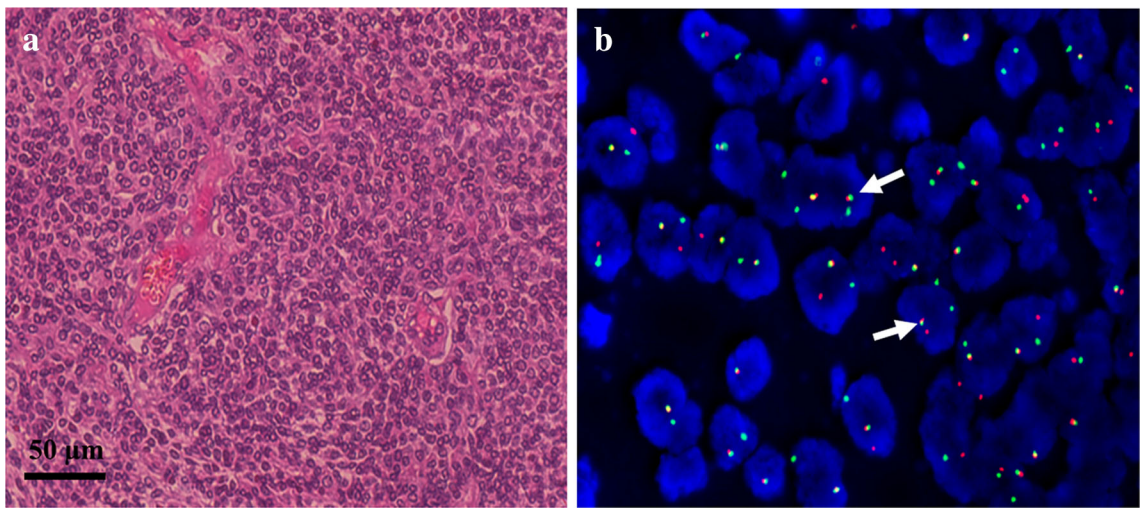

Fig. 3 Nests and clusters of small round cells (**a**) (hematoxylin–eosin × 200). Fluorescent in situ hybridization assay showing positivity for EWSR1 gene rearrange (**b**)

WT-1 gene [12]. In a study by Lae et al., this translocation was present in 96.7% examined tumors [13].

Since this type of tumor is rare, the treatment of DSRCT remains clinically challenge. At present, there is no standard treatment strategy for patients with DSRCT, particularly in cases involving metastatic DSRCT. In recent years, some reports have shown an effective treatment strategy by combining the intense alkylator therapy (P6 protocol), surgical resection and radiotherapy. In 1996, Kushner for the first time suggested to use P6 protocol for control of DSRCT [14]. In 2000, Quaglia et al. reported that there is a significant correlation between the use of P6 protocol,

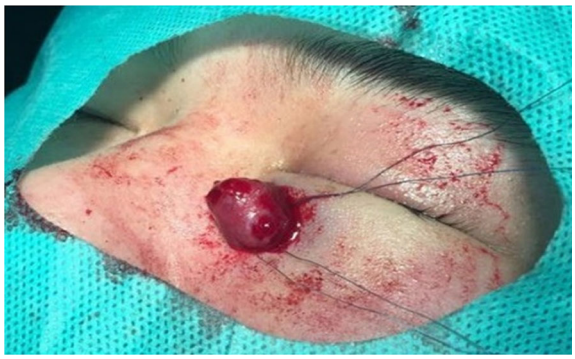

Fig. 4 Patient was in the surgical resection of desmoplastic small round cell tumor

and gross total resection with improved overall survival of DSRCT [15]. In 2005, Lal et al. reported that three- and 5-year survivals were 44 and 15%, respectively, after treatment with a combination of the P6 regimen, surgical debulking and 30 Gy of radiotherapy [16]. A recent report reported a trend for improved survival in patients treated with a P6 protocol comprised of vincristine, doxorubicin, cyclophosphamide, ifosfamide and etoposide [17]. Last year, Zhang et al. found that synchronous chemotherapy with cyclophosphamide, adriamycin and cisplatin combined with radiotherapy (a dose of 40 Gy plus a 20-Gy boost) significantly improved locoregional control of DSRCT [18]. Ikeue et al. reported that a 32-year-old man with pleura DSRCT received the chemotherapy regimen (P6 protocol), surgery and radiotherapy (50 Gy in 25 fractions) for two months and had an optimistic outcome [19]. Xie et al. reported that a case of 30-year-old female with ovarian DSRCT accompanied with lymph node and lung metastase received chemotherapy (intravenous administration of 2 mg vincristine, 50 mg/m^2 doxorubicin and 500 mg/m^2 cyclophosphamide on day 1, repeated every 3 weeks) for three months, and tumor nodule size was decreased [6]. However, this therapeutic strategy exhibits the greatest efficacy which remains to be elucidated in the future by increasing the sample size.

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Compliance with ethical standards

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

Ethical approval All procedures performed in studies involving human participants were in accordance with the ethical standards of the institutional and/or national research committee and with the 1964 Helsinki Declaration and its later amendments or comparable ethical standards.

References

1. Leuschner I, Radig K, Harms D (1996) Desmoplastic small round cell tumor. Semin Diagn Pathol 13:204–212
2. Cao Y, Chen Y, Yang LI, Qian ZH, Han SG, Li QH, Yu RS (2015) Desmoplastic small round cell tumor: a case report of a rare differential diagnosis of solid tumors of the pleura. Oncol Lett 10:2991–2995
3. Gerald WL, Miller HK, Battifora H, Miettinen M, Silva EG, Rosai J (1991) Intra-abdominal desmoplastic small round-cell tumor. Report of 19 cases of a distinctive type of high-grade polyphenotypic malignancy affecting young individuals. Am J Surg Pathol 15:499–513
4. Ud Din N, Pekmezci M, Javed G, Horvai AE, Ahmad Z, Faheem M, Navarro AL, López-Terrada D, Perry A (2015) Low-grade small round cell tumor of the cauda equina with EWSR1-WT1 fusion and indolent clinical course. Hum Pathol 46:153–158
5. Shen XZ, Zhao JG, Wu JJ, Liu F (2014) Clinical and computed tomography features of adult abdominopelvic desmoplastic small round cell tumor. World J Gastroenterol 20:5157–5164
6. Xie YP, Shen YM (2016) Ovarian involvement of a desmoplastic small round cell tumor of unknown primary origin with lymph node and lung metastases: a case report. Oncol Lett 11:1125–1129
7. Bismar TA, Basturk O, Gerald WL, Schwarz K, Adsay NV (2004) Desmoplastic small cell tumor in the pancreas. Am J Surg Pathol 28:808–812
8. Gil A, Gomez Portilla A, Brun EA, Sugarbaker PH (2004) Clinical perspective on desmoplastic small round-cell tumor. Oncology 67:231–242
9. Zhang WD, Li CX, Liu QY, Hu YY, Cao Y, Huang JH (2011) CT, MRI, and FDG-PET/CT imaging findings of abdominopelvic desmoplastic small round cell tumors: correlation with histopathologic findings. Eur J Radiol 80:269–273
10. Ordóñez NG (1998) Desmoplastic small round cell tumor: II—an ultrastructural and immunohistochemical study with emphasis on new immunohistochemical markers. Am J Surg Pathol 22:1314–1327
11. Ordóñez NG (1998) Desmoplastic small round cell tumor: I—a histopathologic study of 39 cases with emphasis on unusual histological patterns. Am J Surg Pathol 22:1303–1313
12. Wang L, Bhargava R, Zheng T, Wexler L, Collins MH, Roulston D, Ladanyi M (2007) Undifferentiated small round cell sarcomas with rare EWS gene fusions:

identification of a novel EWS-SP3 fusion and of additional cases with the EWS-ETV1 and EWS-FEV fusions. J Mol Diagn 9:498–509
13. Lae ME, Roche PC, Jin L, Lloyd RV, Nascimento AG (2002) Desmoplastic small round cell tumor: a clinicopathologic, immunohistochemical, and molecular study of 32 tumors. Am J Surg Pathol 26:823–835
14. Kushner BH, LaQuaglia MP, Wollner N, Meyers PA, Lindsley KL, Ghavimi F, Merchant TE, Boulad F, Cheung NK et al (1996) Desmoplastic small round-cell tumor: prolonged progression-free survival with aggressive multimodality therapy. J Clin Oncol 14:1526–1531
15. Quaglia MP, Brennan MF (2000) The clinical approach to desmoplastic small round cell tumor. Surg Oncol 9:77–81
16. Lal DR, Su WT, Wolden SL, Loh KC, Modak S, La Quaglia MP (2005) Results of multimodal treatment for desmoplastic small round cell tumors. J Pediatr Surg 40:251–255
17. Zhang J, Xu H, Ren F, Yang Y, Chen B, Zhang F (2014) Analysis of clinicopathological features and prognostic factors of desmoplastic small round cell tumor. Pathol Oncol Res 20:161–168
18. Zhang S, Zhang Y, Yu YH, Li J (2016) Complete response of giant desmoplastic small round cell tumor treated with chemoradiotherapy: a case report. Oncol Lett 11:1069–1072
19. Ikeue T, Ohi I, Noguchi S, Fukao A, Terashita S, Horikawa S, Sugita T (2016) Desmoplastic small round cell tumor of the pleura successfully treated with a lower dose of pazopanib. Intern Med 55:2463–2467