Desmoplastic Small Round Cell Tumor: A Case Report

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Abstract

Desmoplastic small round cell tumor (DSRCT) is a rare and highly malignant mesenchymal tumor found in the abdominal cavity. It mainly affects young male patients. We report a case of DSRCT that occurred in the abdominal cavity of a 50-year-old man. The tumor was characterized by small round tumor cells with irregular nests in the prominent desmoplastic stroma. The tumor cells showed immunoreactivity for epithelial membrane antigen, desmin, vimentin, and neuron specific enolase.

Key Words: Desmoplastic small round cell tumor, Abdominal cavity, Soft tissue, Immunohistochemical stain

INTRODUCTION

Desmoplastic small round cell tumor (DSRCT) is composed of small round tumor cells of uncertain histogenesis, associated with prominent stromal desmoplasia and polyphenotypic differentiation.¹ It was first described by Geral and Rosai² in 1989, and has most commonly been reported in children and young adults, with a male-to-female ratio of 4:1. The location for this tumor has primarily been in the abdominal cavity. It mostly occurs in the abdominal cavity. Other primary sites have been rarely reported, have included the paratesticular region,³ the pleural serosa,⁴ the posterior cranial fossa,⁵ soft tissue and bone,⁶ ovary,⁷ and kidney.⁸

We report a case of desmoplastic small round cell tumor that occurred in the abdominal cavity of a 50-year-old man, and review the medical literature.
CASE REPORT

A 50-year-old man presented with abdominal discomfort for two months. He had a history of chronic hepatitis B and cirrhosis. Physical examination revealed an abdominal mass. The abdominal computed tomographic scan showed an intra-abdominal mass that appeared malignant on the right side of the abdomen (Fig. 1). A right hemicolectomy with excision of the mass was performed. A well circumscribed, gray-white, rubbery firm mass, measuring 14.0 x 12.5 x 8.5 cm in size, was attached to the transverse colon (Fig. 2). Multiple tumor nodules were present. Microscopic examination revealed variably sized nests in the desmoplastic stroma (Fig. 3). Tumor cells were uniform, small, and round.

Fig. 1. The abdominal contrast-enhanced computed tomographic scan shows a large tumor in the right abdominal cavity.

Fig. 2. Gross photograph shows a 14.0 x 12.5 x 8.5 cm, circumscribed, nodular, gray-white tumor attached to the serosa of the right colon.

Fig. 3. Tumor shows variably sized nests in the desmoplastic stroma (Hematoxylin-eosin stain, x40).

Fig. 4. Tumor cells are uniform, small, and round (Hematoxylin-eosin stain, x200).
(Fig. 4). Some tumor cells showed rosette formation (Fig. 5). Mitotic figures were seen. Tumor cell necrosis was also present (Fig. 6). Proliferation of endothelial cells was noted (Fig. 7). Lymphovascular invasion was found. The tumor showed direct invasion into the colon wall. Eight out of twenty nine regional lymph nodes showed metastasis. On the immunohistochemical stain, the tumor cells showed positivity for epithelial membrane antigen (Fig. 8), desmin, vimentin, and neuron specific enolase and negativity for alpha-fetoprotein, carcinoembryonic antigen, chromogranin, cytokeratin 7 and cytokeratin 20. A pathologic diagnosis of desmoplastic small round cell tumor was made. The patient died 40 months after surgery.

**DISCUSSION**

Desmoplastic small round cell tumor
(DSRCT) is a rare and aggressive malignant tumor. It usually present with widespread abdominal serosal involvement. There is a striking male predominance, with a peak incidence in the third decade of life (with a wide range from first to the fifth decade). The present case showed positivity for epithelial membrane antigen (epithelial marker), desmin (muscle marker), and neuron specific enolase (neural marker). These findings suggest that this tumor has a capacity for epithelial, muscular, and neural differentiation. The present case showed the morphological and immunohistochemical characteristics consistent with the diagnosis of a DSRCT. The presence of the t(11;22)(p13;q12) translocation is a common cytogenetic feature.\textsuperscript{20} EWS/WT1 gene fusion is specific for DSRCT.\textsuperscript{21}

The differential diagnosis for DSRCT includes Ewing’s sarcoma/PNET, neuroblastoma, embryonal rhabdomyosarcoma, malignant mesothelioma, poorly differentiated carcinoma or neuroendocrine carcinoma.\textsuperscript{22} Ewing’s sarcoma/PNET, neuroblastoma and embryonal rhabdomyosarcoma lack prominent desmoplastic stroma. Poorly differentiated carcinoma and neuroendocrine carcinoma are desmin-negative. Malignant mesotheliomas are CD15 and CD57 negative, whereas most DSRCT are positive for CD15 and CD57. Furthermore, malignant mesothelioma usually does not resemble DSRCT cytologically. The distinct immunohistochemical pattern of DSRCT is diagnostic.

DSRCT has a poor overall prognosis. In
our case, the patient died 40 months after surgery. Treatment consists of a combination of surgery, chemotherapy and radiotherapy. Typically, an initial partial response to tumor debulking and chemotherapy is followed by uncontrollable tumor relapse. Despite multidrug chemotherapy, 75% of the patients die within four to 72 months (average, 26 months) after presentation and only about five percent live beyond five years without evidence of tumor.\(^2\) Intensive alkylator-based therapy with aggressive surgery and radiotherapy to high risk sites has resulted in prolonged progression-free survival.\(^3\)

REFERENCES