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□ 증례 □

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 Rosai[2] 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.

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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

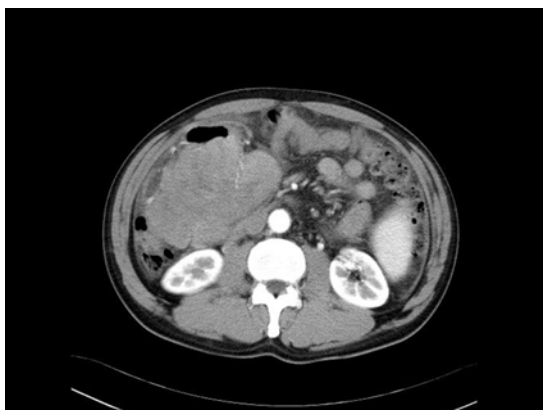

Fig. 1. The abdominal contrast-enhanced computed tomographic scan shows a large tumor in the right abdominal cavity.

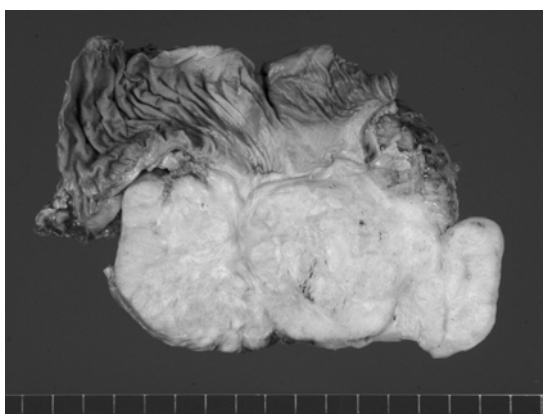

Fig. 2. Gross photograph shows a 14.0 x 12.5 x 8.5 cm, circumscribed, nodular, gray-white tumor attached to the serosa of the right colon.

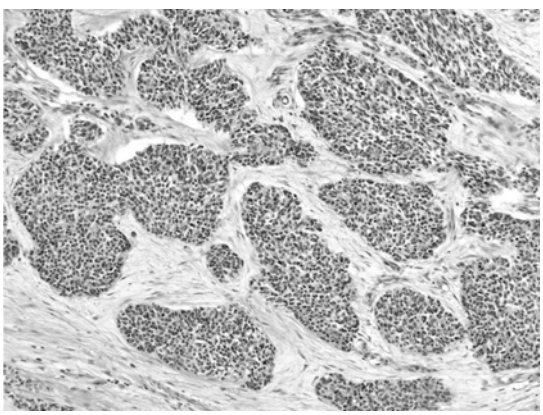

Fig. 3. Tumor shows variably sized nests in the desmoplastic stroma (Hematoxylin-eosin stain, x40).

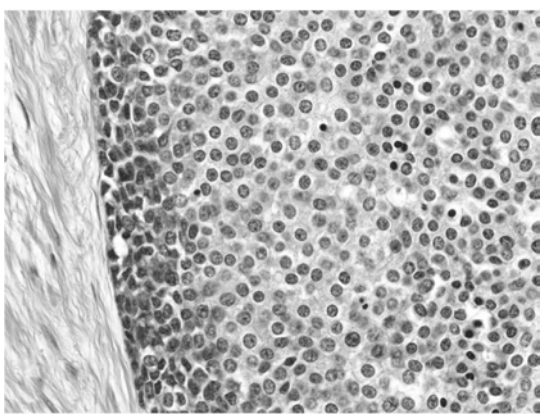

Fig. 4. Tumor cells are uniform, small, and round (Hematoxylin-eosin stain, x200).

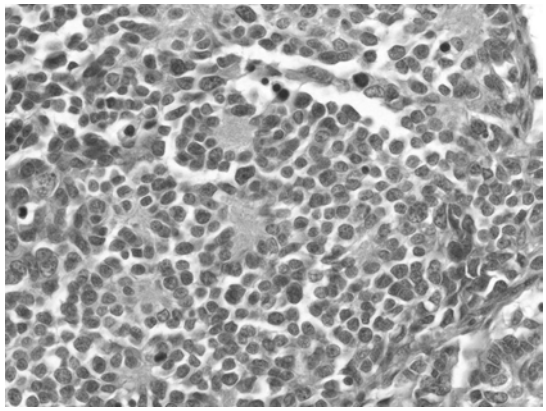

Fig. 5. Tumor cells show rosette formation (Hematoxylin-eosin stain, x200).

(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

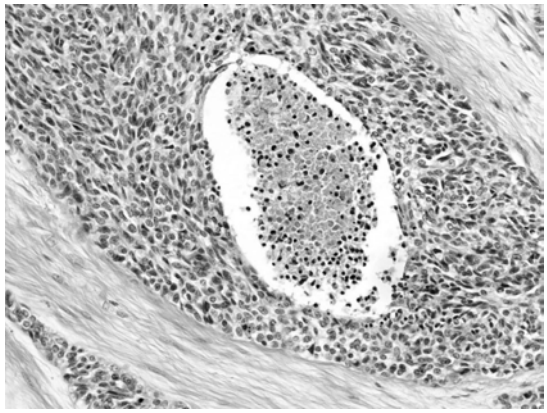

Fig. 6. Tumor cell necrosis is found (Hematoxylin-eosin stain, x100).

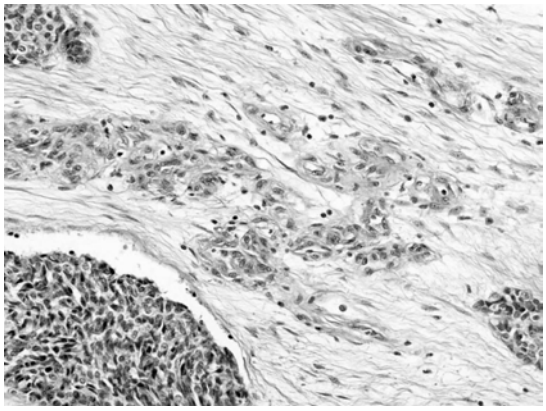

Fig. 7. The proliferation of endothelial cells is seen (Hematoxylin-eosin stain, x200).

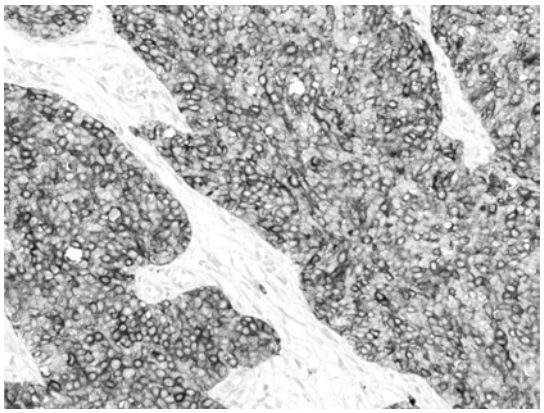

Fig. 8. Tumor cells show strong cytoplasmic positivity for epithelial membrane antigen (Immunohistochemical stain, x200).

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).[1] There have been 10 prior cases reported in Korea.[7-17]

The histogenesis of DSRCT is uncertain. Its predilection for serosal involvement suggests the possibility of a mesothelial origin. Presenting symptoms are usually related to the primary site and include: pain, abdominal distension, palpable mass, acute abdomen, ascites and organ obstruction.[1] The computed tomographic features of DSRCT include bulky intra-abdominal soft tissue masses that involve omental and serosal surfaces, without a distinct organ of origin.[18]

DSRCT shows multiple tumor nodules studding the peritoneal surface. The cut surface is firm, gray-white, with foci of hemorrhage and necrosis.[1] Histologically, DSRCT is characterized by variably size and shape as well as, sharply outlined nests of small neoplastic cells surrounded by a prominent desmoplastic stroma. Some tumors exhibit focal epithelial differentiation, with glands or a rosette growth pattern. The tumor cells are typically uniform with small hyperchromatic nuclei and scant cytoplasm. The desmoplastic stroma is composed of fibroblasts or myofibroblasts embedded in a loose extracellular material or collagen.

The immunoprofile of DSRCT is consistent and distinctive, showing a complex pattern of simultaneous multi-phenotypic differentiation, expressing proteins associated with epithelial, muscular and neural differentiation.[19] 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.[20] *EWS/WT1* gene fusion is specific for DSRCT.[21]

The differential diagnosis for DSRCT includes Ewing's sarcoma/PNET, neuroblastoma, embryonal rhabdomyosarcoma, malignant mesothelioma, poorly differentiated carcinoma or neuroendocrine carcinoma.[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. Futhermore, 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.[22] Intensive alkylator-based therapy with aggressive surgery and radiotherapy to high risk sites has resulted in prolonged progression-free survival.[23]

요 약

결합조직형성소원형세포종양은 매우 드문 악성 연부조직 종양으로 소아와 청소년기에 복강과 골반강 내에 주로 발생한다. 저자들은 복강에 발생한 결합조직형성소원형세포종양 1예를 경험하였기에 문헌 고찰과 함께 보고한다. 50세 남자가 2개월간의 복부 불쾌를 주소로 내원하였다. 방사선 소견에서 오른쪽 복강 내에 종괴가 관찰되었다. 절제된 종괴는 크기가 14.0 x 12.5 x 8.5 cm 이며, 회백색을 띠었으며 가로 잘록창자의 장막에 붙어 있었다. 조직학적으로 종괴는 결합조직을 형성하는 기질 내에 작고 둥근 핵을 가진 종양세포들의 증식이 보였다.

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