



Desmoplastic Small Round Cell Tumor

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ABSTRACT

Desmoplastic small round cell tumors (DSRCT) are rare and highly aggressive sarcomas.¹ The World Health Organization defines DSRCT as a malignant mesenchymal neoplasm composed of small round tumor cells with polyphenotypic differentiation embedded within a prominent desmoplastic stroma.² They present mainly in young adult Caucasian males developing as intrabdominal or pelvic masses.¹ The masses develop along serosal surfaces which can cause serous effusions. The molecular hallmark is the t(11;22)(p13;q12) translocation EWS-WT1 fusion protein.^{1,3,4} There is no standard treatment plan for patients with DSRCT and the rate of survival is very poor.

CLINICAL HISTORY

An 18-year-old male presents with abdominal pain. He received a CT which shows diffuse abdominal masses and carcinomatosis. A biopsy was performed on one of the omental masses and preliminary pathology results showed a desmoplastic small round cell tumor. Additionally, ascites fluid was examined and reported to have malignant desmoplastic small round cells. The patient went through a round of systemic chemotherapy (Vincristine, Adriamycin, Ifosfamide) and had a good response with some tumor regression before having the tumor debulking surgery.

GROSS DESCRIPTION

Part H is received fresh labeled "low anterior resection with en bloc right ureter." It consists of a low anterior resection specimen which includes a segment of bowel which is 27.0 cm in length and 3.0 cm in luminal diameter with up to 4.5 cm of attached adipose tissue, a 6.0 x 0.3 cm right ureter, 4.5 x 0.4 cm right vas deferens, and 7.5 x 1.5 x 0.7 cm right seminal vesicle (figure 1-2). Two masses are identified measuring 9.5 x 7.5 x 5.0 cm and 4.5 x 3.5 x 2.3 cm which are attached to the distal end of the bowel. The serosa is tan-pink, hemorrhagic, and smooth. A portion of the bowel, the right ureter, right vas deferens, and right seminal vesicle are adherent to the larger mass. The anterior surface of the mass is inked blue and the posterior surface is inked black. Sectioning the larger mass reveals a semi cystic, firm, white-yellow cut surface with pushing borders (figure 3). The larger mass surrounds the ureter, is at, but does not appear to be extending into, the muscularis propria of the bowel wall and extends to, but does not invade, the vas deferens and seminal vesicle (figure 3-4). Sectioning reveals the smaller mass is firm, well-demarcated, and has a smooth, white-yellow cut surface. The smaller mass is at the anterior and posterior margins and is 1.5 cm from the larger mass. The remaining bowel mucosa is tan with the usual folds. There are multiple, tan-yellow, rubbery nodules on the bowel serosa ranging from 1.3 x 1.0 x 0.5 cm to 3.0 x 1.8 x 1.5 cm which are 0.1 cm from the underlying muscularis propria.

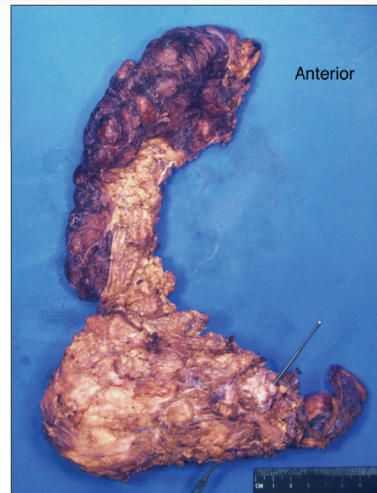


Figure 1: Overall specimen, anterior aspect

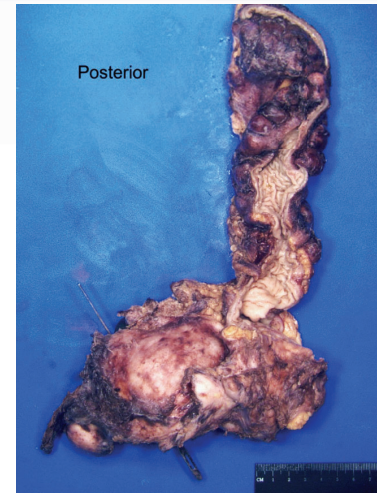


Figure 2: Overall specimen, posterior aspect

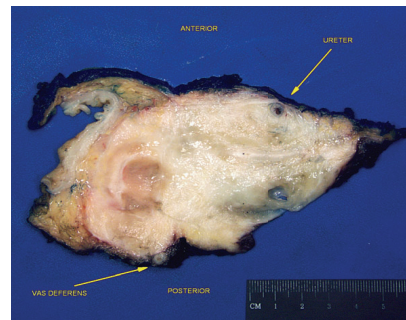


Figure 3: Cross-section of mass with attached ureter and vas deferens

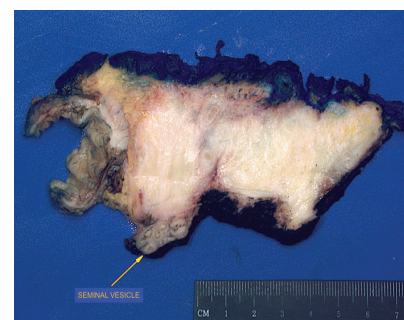


Figure 4: Cross-section of mass with attached seminal vesicle

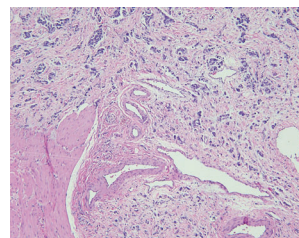


Figure 5: Tumor invading bowel wall

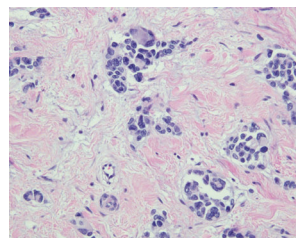


Figure 6: Tumor cells

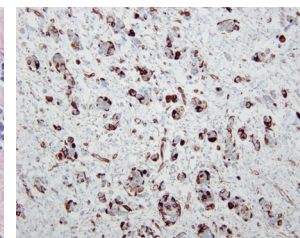


Figure 7: WT-1 IHC stain (positive)

FINAL DIAGNOSIS

The final report showed the tumor involved the pericolic, periureteral, and peritesticular tissue. The tumor also invaded into the muscularis propria of the bowel (figure 5). Immunohistochemical (IHC) stains and molecular tests were completed to confirm the diagnosis of desmoplastic small round cell tumor. Molecular studies showed the patient has the EWS-WT1 fusion protein which further supports the diagnosis of desmoplastic small cells tumor. IHC done on the tumor was positive for WT-1 (figure 7), vimentin, desmin, CAM5.2, and CD99.

DISCUSSION

DSRCT have a poor prognosis with only a 15%, 5-year survival rate.⁵ There is no standard treatment for patients with DSRCT because there are so few cases to be studied. Some treatments used are chemotherapy, radiation therapy, surgical excision, and bone marrow transplant.⁵ Even with the most aggressive treatment plans, relapse is common, and survival is still dismal. Histologically, DSRCT have clusters of tumor cells distributed within a cellular stroma (figure 6).^{3,5} The tumor cells are round to oval with hyperchromatic nuclei, a scant amount of cytoplasm, and indistinct cell borders.³ The IHC stains performed on these tumors demonstrate the multilineage differentiation, which can be used to distinguish them from other small round blue cell tumors.^{3,4} The different lineages within the tumor can include: epithelial, muscle, and neural markers.⁵ Below are the positive tumor markers performed for this case:

- WT-1: kidney and ovarian epithelium (positive in Wilms' tumor)
- CD99: soft tissue sarcomas (positive in Ewing sarcomas)
- CAM5.2: epithelial derived tissues
- Desmin: muscle cells (Z-band)
- Vimentin: mesenchymal derived tissues

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