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CASE REPORT **OPEN ACCESS**

A Tale of Two Atypical Presentations of Desmoplastic Small Round Cell Tumors: A Rare Mesenchymal Tumor

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ABSTRACT

Desmoplastic Small Round Cell Tumor (DSRCT) is a rare malignant mesenchymal tumor that usually arises in the abdomen and pelvis and demonstrates a locally aggressive behavior with multiple peritoneal implants.The paucity of these cases and varied clinical presentations leads to a diagnostic dilemma, and there are noconsensus guidelines regarding management. We report 2 unusual presentations of DSRCT, one with testicular mass along with retroperitoneal lymphnodes and umbilical nodule, and another with left inguinal swelling and omental mass. These cases are being presented because of infrequent occurrences and unusual presentations that have been not reported in the literature so far.

Keywords: Desmoplastic Small Round Cell Tumor, mesenchymal tumor, rare, aggressive

INTRODUCTION

DSRCT is a recently identified malignant soft tissue sarcoma of mesenchymal origin with pathognomonic fusion protein EWS -WT1, described by Gerald and Rosai.[1,]It is a relatively rare lethal tumor arising in abdomen and pelvis, and often presenting as multiple implants in omentum, mesentery, diaphragm and peritoneum.Involvement of testes, ovaries and pleura is rare.[2]It is alocally aggressive tumor that spreads over serosal surface, but distant metastasis occurs late. It is common seen in young adults with predilection for males (M: F4:1).[3,4] There is no standard of care due to rarity of cases.Although these tumors arechemosensitive and radiosensitive, aggressive and complete surgical resection is the mainstay of treatment and a major determinant in patient's survival. However, complete resection is often not possible due to multicentric nature.[3]Despite multiple treatment modalities, the prognosis is dismal with a 5 years overall survival of 15%.[5]

CASE HISTORY

CASE 1

A 25 years old male with no comorbidities presented to the Department of Surgical Oncologywith a painless right testicular swelling of 1 year duration, progressively increasing in size.Local examination revealed a 15 x 10 cm hard, non-tender, right testicular mass with bosselated surface extending up to right inguinal region and on abdominal examination, there was a 1x1 cm hard nodule palpable at the umbilicus. [Figure 1]Serum tumor markers (Beta HCG, AFP and LDH) were within normal limits. USG guided FNAC of umbilical nodule was suggestive of metastatic disease. A contrast enhanced CT scan of abdomen and pelvis showed aright testicular/para-testicular mass with retroperitoneal lymph nodal mass and multiple soft tissue metastatic deposits in rectovesical pouch, bilateral iliac fossa and in subcapsular location in liver. He underwent Right High Inguinal Orchidectomy [Figure 2], histopathology report suggestive of Desmoplastic Small Round Cell Tumor. Immunohistochemical Studies showed CK, Desmin – Positive and MyoD1, Myogenin, SMA, CD99, OCT4, Calretinin -Negative [Figure 3,4]. He received 6 cycles of chemotherapy-

VAC-IE regimen. Post chemotherapy PET Scan done showed progressive disease with appearance of new lesions in the form of Left Supraclavicular and Mediastinal Lymphadenopathy. Patient was kept on palliative chemotherapy with tablet Pazopanib for 6 months, following which he succumbed to the disease.

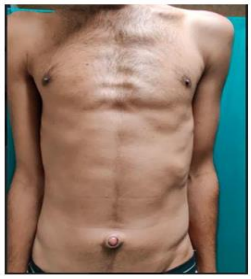

Figure 1: Umbilical Nodule

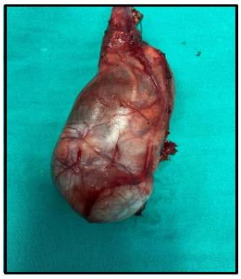

Figure 2: Right High Inguinal orchidectomy

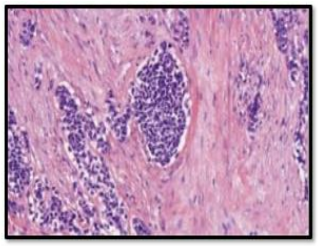

Figure 3: Nests of tumor cells surrounded by a characteristic desmoplastic stroma

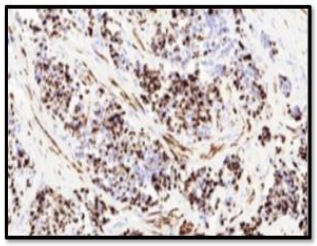

Figure 4: IHC showing diffuse positivity for Desmin

CASE 2

A 22 years old malewith history of Left Inguinal Hernia Repair 3 years back presented with swelling and pain in left groin for 2 months duration. Local USG done showed soft tissue lesions measuring 2.5cm x 3.8 cm and 1.8 cm x1.3 cm in the left inguinal region with excessive wavy appearance of mesh.CECT Abdomen done revealed awell-defined, encapsulated homogenously enhancing mass lesion measuring6.3 cm x 11cm x 8.1 cm in the pelvis likely arising in lower part of omentum [Figure 7] with extensive omental and peritoneal deposits and a mass in left antero-lateral abdominal wall extending into inguinal canal and perivesical region, surrounding the hernial mesh. A CT guided biopsy of omental mass was suggestive of Desmoplastic small round blue cell tumor. (IHC-Desmin, FL1, EMA positive. INI-1 retained).He received 5 cycles of chemotherapy (VAC regimen), following which he underwent Cytoreductive Surgery in the form of Excision of Omental Mass, Left Anterolateral parietal wall deposit, deposit over bladder, and pelvic and right diaphragmatic peritonectomy [Figure 5,6] along withHyperthermic Intraperitoneal Chemotherapy with Cisplatin (120 mg at 41 degree C). Postoperative period was complicated with a pelvic collection which was drained under USG guidance on POD-9. Final Histopathology report revealed multifocal desmoplastic small round cell tumor (Stage -pT4, LVI+, immunopositive for WT1, Desmin, EMA and immunonegative for CK, Myogenin, Myo-D1). He presented 1 month postoperatively with an Enterocutaneous fistula which was of low output, and he was managed conservatively for the same He has received two cycle of Adjuvant chemotherapy (VAC regimen) and developed recurrence while on adjuvant treatment. He succumbed to disease within 2 months of recurrence.

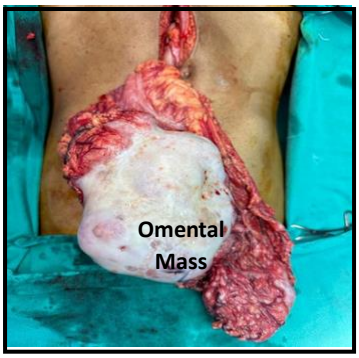

Figure 5 – Intraoperative Photo showing omental mass

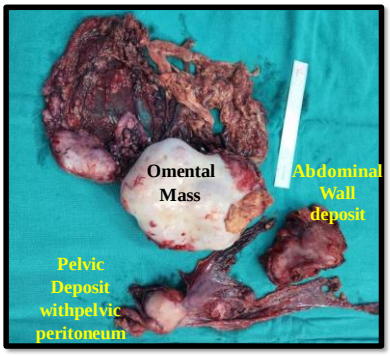

Figure 6 – Specimen showing omental mass, pelvic and abdominal wall deposit

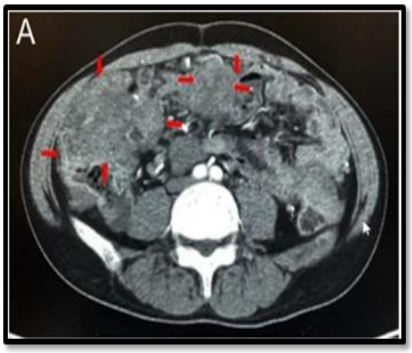

Figure 7- CECT showing homogenously enhancing mass lesions in pelvis

DISCUSSION

Mesenchymal tumors that arise within the abdominal cavity represent a heterogeneous group of entities that pose diagnostic difficulty to both surgeons and pathologists.Distinguishing between them is important as there are significant differences in their biologic potential and treatment. DSRCT are aggressive tumors with poor survival despite multimodal therapy (Chemotherapy, Debulking surgery +/- HIPEC, Whole abdominal radiation).[6]Due to rarity of this neoplasm, large population-based studies are lacking and evidence on management consist of small retrospective series. Studies have shown that aggressive and complete surgical resection is a major determinant inpatient survival. However, since DSRCT most commonly presents as a multicentric abdominal mass, completeupfront resection is not often possible.[3]Like Ewings Sarcoma, DSRCTs are chemosensitive and radiosensitive.[2] Although many strategies have been attempted, survival in patients with DSRCT remains dismal with a 15 % overall survival (OS) rate at 5 years. Cytoreductive surgery with orwithout HIPEC improves survival when compared medical therapy or radiation therapy

alone. Whole abdominopelvic (WAP) radiotherapy has also been proposed as an adjunct to surgery with aim to improve local control.[4]

CONCLUSION

DSRCT is a rare aggressive malignancy with dismal prognosis. Although intra-abdominal site is the most common location for DSRCT, it can present unusually as a testicular mass or inguinal mass. Our knowledge on the clinical and pathological nature of the disease is limited leading to diagnostic and therapeutic challenge. The standard management protocols are still evolving and multimodality treatment with NACT followed by Cytoreductive Surgery with HIPEC and adjuvantchemotherapy or whole abdominal radiotherapy offers the best shot.

REFERENCES

1. Hayes-Jordan, A., Green, H., Fitzgerald, N., & Xiao, L., & Anderson, P. (2010). Novel treatment for DSRCT: HIPEC. *J Pediatr Surg*, *45*(5), 1000-1006. doi: 10.1016/j.jpedsurg.2010.02.034. PMID: 20438942.
2. Wong, H. H., Hatcher, H. M., Benson, C., Al-Muderis, O., Horan, G., Fisher, C., ... & Judson, I. (2013). Desmoplastic small round cell tumour: characteristics and prognostic factors of 41 patients and review of the literature. *Clinical sarcoma research*, *3*, 1-9.
3. Lal, D. R., Su, W. T., Wolden, S. L., Loh, K. C., Modak, S., & La Quaglia, M. P. (2005). Results of multimodal treatment for desmoplastic small round cell tumors. *J Pediatr Surg*,*40*(1), 251-255. doi: 10.1016/j.jpedsurg. 2004.09.046. PMID: 15868593.
4. Subbiah, V., Viny, A. D., Anderson, P. M., Hayes-Jordan, A. A., Qiao, W., Benjamin, R. S., ... & Ludwig, J. A. (2012). Optimizing the therapy of desmoplastic small round cell tumor: Combined experience from the two major cancer centers. *Journal of Clinical*, *30*(15).
5. F.A. Angarita, S. Hassan, A.J. Cannell, B.C. Dickson, R.A. Gladdy, C.J. Swallow, A. Gupta, M.E. Blackstein, J.A. McCart,Clinical features and outcomes of 20 patients with abdominopelvic desmoplastic small round cell tumor,

European Journal of Surgical Oncology (EJSO), Volume 43, Issue 2,2017,Pages 423-431,ISSN 0748-7983.

6. Syed Hamza Bin Waqar et al.,Desmoplastic small round cell tumor: *Analysis of surveillance, epidemiology, and endresults (SEER) database* 1975 to 2018. *JCO* 40, e23540-e23540(2022).DOI:10.1200/JCO.2022.40.16.