

Desmoplastic small-round-cell rectal tumor

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A 22-year-old male presented with abdominal and back pain and significant weight loss. On examination he was cachectic with a large palpable mass arising from the pelvis. A digital rectal exam revealed an obstructed rectal mass 5 cms from the anal verge. A contrast-enhanced computed tomography (CT) scan of the abdomen revealed a rectal mass with peritoneal metastasis in the form of extensive pelvic deposits with surface liver deposits (Fig. 1 A-D). Rectal biopsy revealed nests of small round cells with

scanty cytoplasm, round to oval nuclei and visible mitotic figures, embedded in a desmoplastic stroma seen in the lamina propria and muscularis mucosa of the rectum. On immunohistochemistry, these cells were immunopositive for cytokeratins AE1/AE3 (focally positive) and desmin (strongly positive), while they were negative for leukocyte common antigen (LCA), synaptophysin, Wilm's tumour-1 protein (WT-1) and neuron-specific enolase (NSE). MIC2 protein showed dot-like positivity (Fig. 2 A-D). A diagnosis of desmoplastic small-round-cell rectal tumor was made. No cytogenetic analysis was performed. The patient had a poor general condition and did not opt for further treatment.

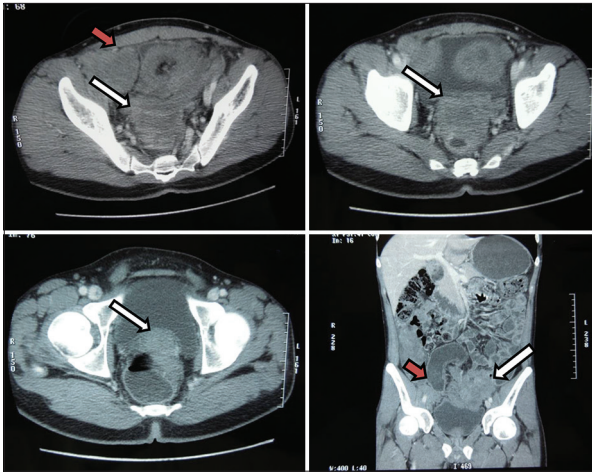


Figure 1 Pelvic sections of contrast-enhanced computed tomography scan of the abdomen and pelvis showing a low attenuation rectosigmoid upper rectal growth (white arrow) with peritoneal metastasis in the form of multiple, low attenuation soft tissue masses in the pelvis (red arrow) with surface liver deposits (section not shown)

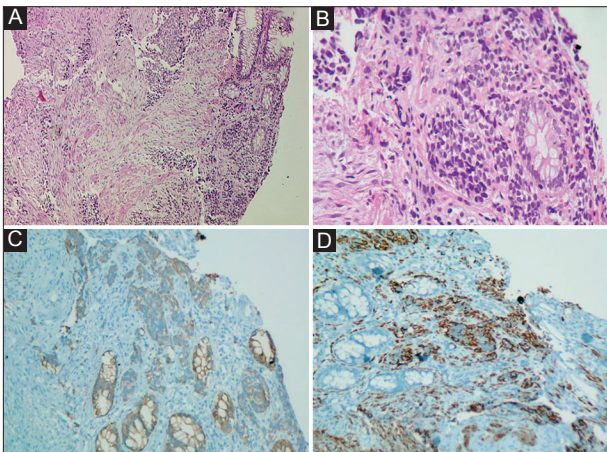


Figure 2 Histopathological examination of rectal biopsy showed malignant round cell tumor nests of small round cells with scanty cytoplasm, round to oval nuclei and visible mitotic figures, embedded in a desmoplastic stroma seen involving the lamina propria and muscularis mucosa of the rectum [A; H&E (40X) & B; H&E (200X)], intervening colonic mucosal glands are unremarkable. The tumor cells revealed weak and focal immune-positivity for cytokeratin [C; DAB (100X)] and strong immune-positivity for desmin [D;DAB (100X)]

Desmoplastic round-cell-tumor was first described as a pathological entity by Gerald and Rosai in 1989 [1]. It is a rare, highly aggressive tumor associated with poor prognosis, predominantly affecting young males. It is a sarcoma belonging to the family of “small round blue cell tumors” of the pediatric population [2]. It is associated with a unique chromosomal translocation $t(11:22)(p13;q12)$ that involves the *EWSR1* and *WT1* genes.

This tumor is usually large and widespread at presentation and it is impossible to identify the organ of origin in many instances. Hence it is speculated that the tumor most probably arises from mesothelial cells explaining its occurrence in the peritoneum and occasionally in the pleura, lung, tunica vaginalis, sinus cavity and the posterior cranial fossa [5,6]. However, it has also been reported to arise from organs like the stomach, ovary, liver, pancreas, kidneys, bone etc. One report described a localized origin from the sigmoid [3]. In our reported case, the rectum was the organ of origin with metastasis to the peritoneal cavity.

CT scan aids in the diagnosis of these tumors by demonstrating multiple, bulky low attenuation soft tissue masses in the omentum or mesentery or peritoneum mostly without a distinct organ of origin. Histological analysis typically shows small round blue cells in nests separated by an abundant desmoplastic stroma. Immunohistochemistry demonstrates a polyphenotypic antigen expression profile and is positive for desmin, vimentin, smooth muscle actin, neuron specific enolase, cytokeratin and epithelial membrane antigen. Diagnosis can be confirmed by cytogenetic studies.

The combination of Ewing-sarcoma-based induction chemotherapy followed by aggressive surgical debulking and external beam radiotherapy has been recommended for the treatment of these tumors. The impact of intensive chemotherapy regimens and new techniques such as HIPEC or IMRT needs to be clearly defined [4,5].

This report highlights a rare pathological diagnosis of desmoplastic small-round-cell tumour of the rectum, never reported previously.

References

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Conflict of Interest: None

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