

Ewing's Sarcoma of Rectum – A Rare Case Report

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ABSTRACT

A 16-year-old male presented with constipation, acute retention of urine and abdominal swelling associated with pain. Per-rectal examination revealed painful growth in lateral and anterior wall of rectum 4cm from anal verge. X-ray erect abdomen was done which showed Indistinct bowel gas pattern suggestive of Intestinal obstruction. Diversion transverse colostomy was done in view of acute intestinal obstruction. Investigations revealed large well defined exophytic heterogenous lesion arising from lateral wall of rectum with normal CEA levels. Biopsy revealed small round blue cell tumour which was positive for Pan CK and CD99. He was planned for 6 cycles of Neoadjuvant chemotherapy (NACT) with VAC IE followed by reassessment for local therapy followed by adjuvant chemotherapy with 8 cycles of VAC IE. He received 6 cycles of VAC IE and Definitive radiation therapy 45Gy in 25 fractions by 3DCRT. After completion of radiation therapy, he received 8 more cycles of VAC IE in TATA memorial hospital. We present this case report, due to the rarity of Extraosseous Ewing's sarcoma (EES) of the rectum.

KEYWORDS: scabies, wound, wound healing, inflammation, cytokines, interleukins, IL-1 α , IL-1 β , neutrophil extracellular trap, NET, oxidative stress, reactive oxygen species, ROS.

ARTICLE DETAILS

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CLINICAL PRESENTATION

A 16-year-old male presented with 1 month history of constipation and lower abdominal swelling associated with pain. He also had history of acute retention of urine for which he was catheterized. Per-rectal examination revealed tender exophytic growth in anterior and left lateral wall of rectum 4cm from anal verge, extending from 12-4 'o' clock position, upper extent could not be felt, rest of the rectal mucosa was normal with preserved sphincter tone.

INVESTIGATIONS

X-ray erect abdomen (Fig 1) was done which showed indistinct bowel gas pattern suggestive of Intestinal obstruction. Diversion transverse colostomy was done in

view of acute intestinal obstruction. Once the patient was stabilised, he was evaluated further.

CECT abdomen and PET CT both revealed large well defined exophytic heterogenous lesion measuring 13*10.7*13.5cm (max SUV 12.4) arising from left anterior-lateral wall of distal rectum at 1-4 o'clock position obscuring the rectal passage. Few distended sigmoid bowel loop seen above the lesion with faecal impaction. CT guided biopsy (Fig 2) revealed small round blue cell tumour (Fig 3a & 3b). Immunohistochemistry (IHC) was positive for Pan CK with ki67- 50% and uniform membrane staining for CD99, negative for CK7, desmin, chromogranin and synaptophysin (Fig 4a & 4b). CEA was 4.12. Bone marrow biopsy was negative for malignant cells.

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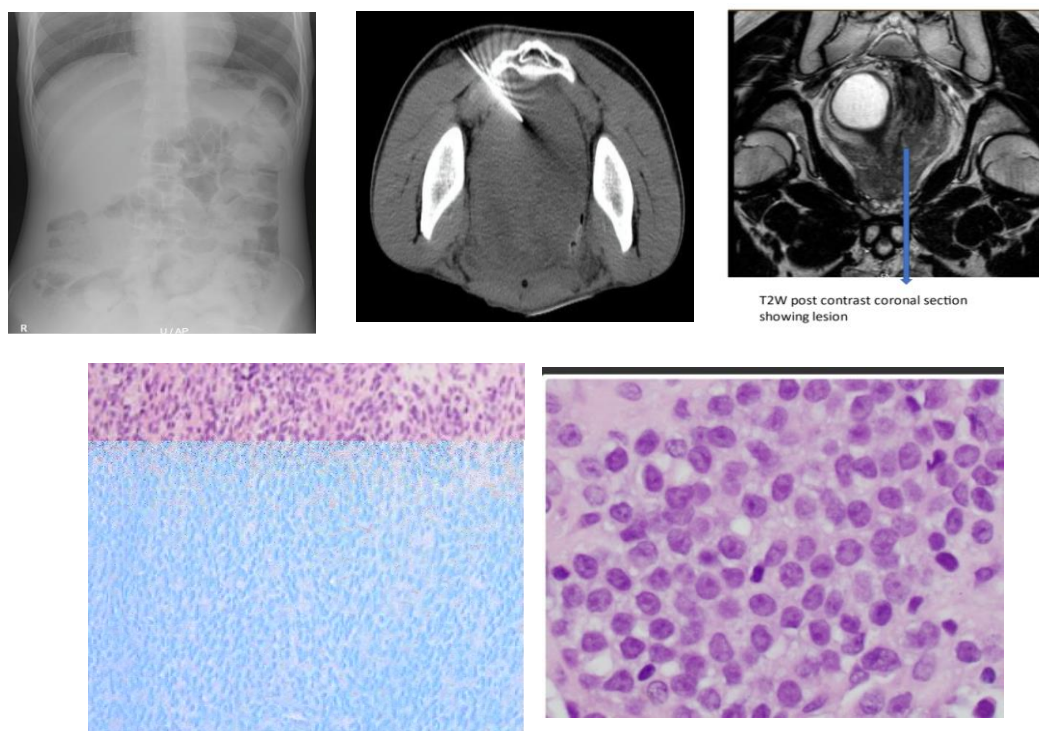
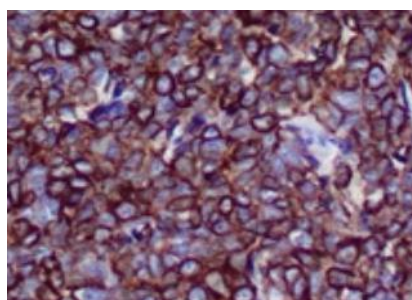
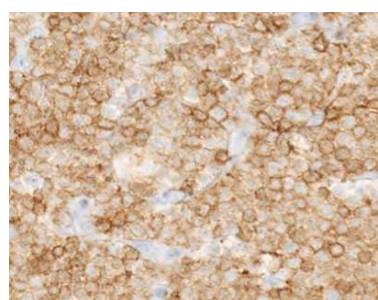


Fig 3a. Histology showing small cells with round heterochromatic nuclei, with scanty cytoplasm arranged in nests, sheets, dispersed in fibromuscular stroma with few blood vessels and foci of necrosis- 40x magnification. Fig 3b: 100x magnification



4a) PAN CK positive staining



4b) CD99 membrane staining

Subsequent histological review and IHC indicated that diagnosis was consistent with a PNET/Ewing's sarcoma arising from anterior-lateral wall of rectum. He was planned for NACT by VAC IE regimen for 6 cycles followed by reassessment for radical treatment by radiation therapy or surgery, followed by adjuvant chemotherapy by VAC IE regimen for 8 cycles (7 cycles of VAC + 7 cycles of IE)[3]. The NACT regimen included VAC (Vincristine 2mg IV on day 1, Adriamycin 60mg/m² IV on days 1-3, Cyclophosphamide 1200mg IV on day 1) and IE (Ifosfamide 2300mg IV on days 1-5, Etoposide 130mg IV on days 1-5) with Mesna. Granulocyte colony stimulating factor injection was given to minimise bone marrow suppression.

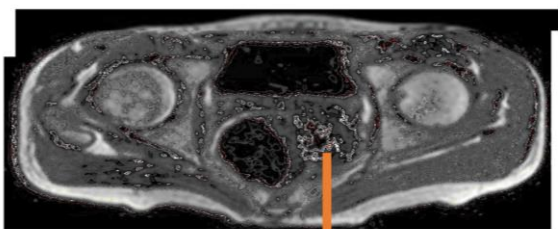
Post 6 cycles of VAC IE, per rectal assessment revealed exophytic firm mass palpable 4cm from anal verge, mainly felt in left lateral wall of rectum and partly in anterior wall, upper extent could not be made out, rest of the rectal mucosa was normal with preserved sphincter tone.

MRI abdomen and pelvis with contrast showed well-

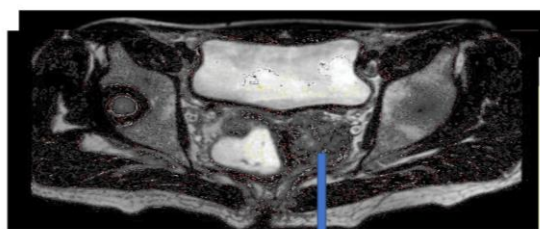
defined exophytic heterogenous lesion noted arising from the left anterior-lateral wall of the distal rectum at 1-4'o clock position, approximately measuring 3.5 x 4.5 x 10 cm (Ap x Tr x Cc). The lesion appeared T1W hypointense (Fig 5a & 5b), T2W heterogeneously hypointense (Fig 5a & 5b & 5c) and restriction on diffusion images, with heterogenous enhancement on post GAD images with non-enhancing cystic areas (s/o necrosis/cystic areas).

The lesion showed focal loss of fat planes with the prostate at its origin at 1-2' o clock position and left seminal vesicle anteriorly. Posteriorly, the lesion was confined to the mesorectal fascia with focal extension into the presacral space to involve the left lateral pelvic wall. The lesion was seen extending from S2 vertebra superiorly (Fig 5b) to 4.2cm from anorectal junction. The lesion is also seen involving left obturator internus, levator ani and pubococcygeus muscles. MRI showed partial/poor response to neoadjuvant chemotherapy.

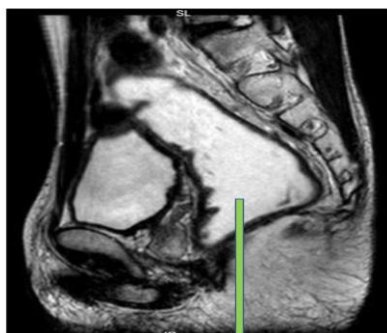
Ewing's Sarcoma of Rectum – A Rare Case Report



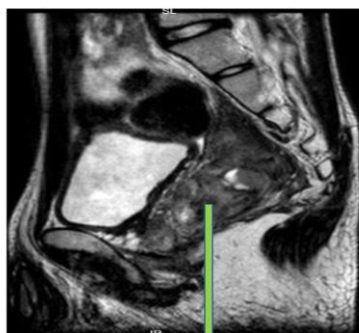
T1 W axial section image visualising lesion



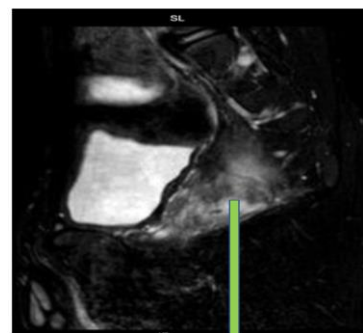
T2 W post contrast axial section image visualising lesion



Distended sigmoid colon, impacted faeces



T2 W sagittal section visualising the lesion

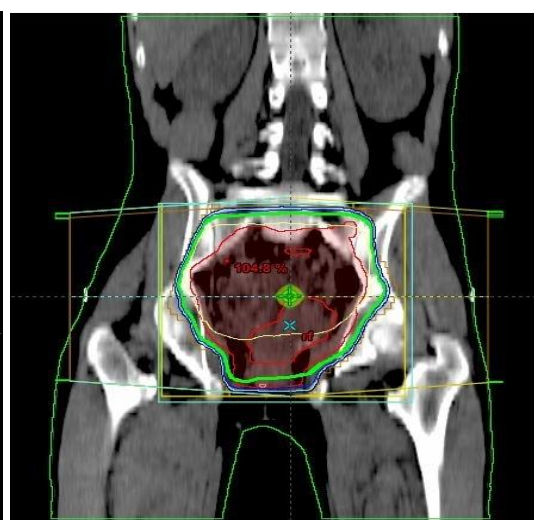
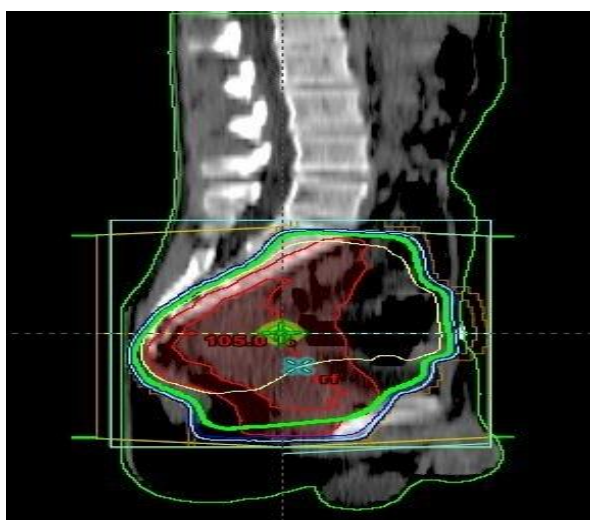


T2 W SPAIR image showing lesion

5b) Saggital section showing distended sigmoid colon with impacted faeces, T2W and T2W SPAIR images showing the lesion

Surgery was deferred in view of partial response with indistinct planes for surgery. Hence, he was planned for Definitive Radiation therapy 45Gy in 25 fractions followed by boost to 59.4Gy to the primary by 3DCRT[4] .(Fig 6a & 6b & 6c). After 25 fractions patient defaulted treatment and

was lost to follow up. When contacted last, patient's attender informed that he received 8 cycles of Adjuvant VAC IE in TATA memorial hospital, Mumbai, during which the patient's condition worsened. He expired after 40 days of last cycle of VAC IE.



Extrasosseous Ewing's sarcoma is a rare type of tumour that belongs to the Ewing's Sarcoma Family of Tumours (ESFT)^[2]. It is the second most common primitive bone cancer in children and adolescents. Approximately 95% of patients with these mesenchymal neoplasms have chromosomal translocations that lead to gene fusion^[2,3]. This triggers membrane IGF-1 receptor that increases tumour

proliferation. Due to this, these tumours show aggressive behaviour and high recurrence rate.

EES is a rare entity, accounting for only 1.1% of malignant soft tissue tumours^[5] and 20-30% of all ESFT. It was first described by Tefft *et al*^[1] in 1969. EES typically originates from the soft tissues of trunk and extremities, and most of these cases are reported in 10-30 years of age group. Here we

Ewing's Sarcoma of Rectum – A Rare Case Report

highlight the first rare case of Ewing's sarcoma of the rectum in our institute.

The clinical manifestations of EES are non-specific such as localized pain and/or swelling.

Tissue diagnosis is difficult due to resemblance of cells to other tumour types, including carcinoma, lymphoma, melanoma, synovial sarcoma, and rhabdomyosarcoma. Immunohistochemically, cellular membranes mostly stain in a strong Olympic-ring-like pattern due to high expression of the cell surface membrane protein p30/32 or CD99 antigen (encoded by the MIC2 gene). Tumour cells also show positive for Vimentin, CD99, Cyclin D1, and Bc12 and were negative for CD45 (LCA), PanCK, and Desmin on IHC.^[8]

Imaging modalities such as CT, MRI, and PET/CT aid in evaluating metastatic EES and assessing local tumour resectability.

Recent evidence suggests that about 75% of patients present with localised disease and that 70–80% are curable with combined treatment modalities of chemotherapy, radiotherapy, and surgery.^[6]

Aboumarzouk et al.^[2] reported a case of 34-year-old female presenting with rectal pain and bleeding which on biopsy showed poorly differentiated cloacogenic carcinoma. Imaging revealed liver metastases. IHC favoured PNET. Chemotherapy regimen using IVAD (Ifosfamide, Vincristine, Adriamycin) resulted in complete resolution of primary tumour and liver metastases. Follow up of 7 years showed no recurrence, then she presented with malignant anal fissure. Imaging revealed no metastases. Biopsy showed T1N0M0 basaloid squamous carcinoma.

Athary et al.^[7] reported a case of 31-year-old female presenting with rectal bleeding. Examination revealed a painful friable rectal mass. Imaging showed no metastases elsewhere. Surgical excision of mass was done. Histopathologic and IHC reporting favoured diagnosis of Ewing's sarcoma. Adjuvant therapy was given.

ES of rectum is a rare entity. It needs careful evaluation and multi-disciplinary approach for achieving good response rates. It is also important to follow up with the patient closely as this tumour is associated with a high recurrence rate which may warrant further intervention.

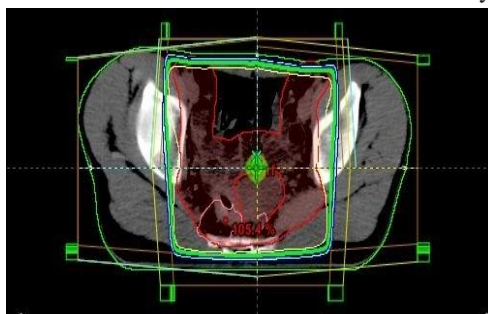


Fig 6a & 6b & 6c. Contouring and external beam planning showing beam arrangement and isodose curves.6c)

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