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Rectal Synovial Sarcoma: A Case Report And Review Of The Literature

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Abstract
Synovial Sarcoma (SS) is a rare and aggressive mesenchymal malignancy, representing 5-10% of all soft tissue sarcomas. Approximately 70% of patients experience local recurrence or distant metastasis. Primary SS in the gastrointestinal tract is exceedingly uncommon, with less than 1% being located in the retroperitoneal location. Morphology could be monophasic, biphasic, or poorly differentiated subtypes having translocation of SS18 with SSX1, SSX2, or SSX4 genes most typically. A 30-year-old lady presented to our outpatient department with symptoms of rectal bleeding and perineal discomfort. Clinical examination and imaging studies suggested the presence of rectal synovial sarcoma, which had extended into the right ischiorectal and ischioanal fossa. Subsequent biopsy and immunohistochemistry confirmed the diagnosis of biphasic synovial sarcoma. The patient was reviewed by a multidisciplinary tumor board, and a treatment plan involving neoadjuvant chemotherapy followed by surgery was established. After chemotherapy she underwent abdominoperineal resection, and her postoperative recovery was uneventful. Henceforth, rectal sarcoma is a very rare malignancy with a poor prognosis. However, surgery with curative intent with a negative margin may additionally confer better overall survival.

Keywords: *Sarcoma, Rectal synovial sarcoma, Biphasic synovial sarcoma, Abdominoperineal resection, ELAPE*

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I. Introduction

Synovial sarcoma (SS) is a malignant mesenchymal neoplasm of uncertain origin that accounts for 5 to 10% of all soft tissue sarcomas and can involve any anatomical site. [1] It can occur in various anatomical regions, although it is most frequently found in the lower extremity, followed by the trunk, head, and neck. Primary gastrointestinal (GIT) SS are exceptionally uncommon, with retroperitoneal involvement being present in less than 1% of cases. Lymph nodal metastasis occurs in 3-7%, and up to 70% of individuals with SS may experience local recurrence and distant metastasis.Morphologically categorised as monophasic (MSS), biphasic (BSS), or poorly differentiated (PDSS).[2] These different histologic subtypes can considerably overlap with one another and with other types of neoplasms. The monophasic type consists of uniform spindle cells; BSS is morphologically characterised via the coexistence of the spindle cell component and an epithelial element, which might be often arranged in glandular structures, nests, or cords that vary in quantity. In both types, the mitotic activity is limited. The poorly differentiated type includes primitive round or spindle cells that may have a rhabdoid appearance and exhibit a high mitotic count. The chromosomal translocation t(X;18) (p11.2;q11.2) fuses the SS18 (SYT) gene to the SSX gene and is seen as a founding event in the oncogenic development of synovial sarcoma.[3] However, the actual transformative event of the chimeric SYT-SSX gene product has not been absolutely elucidated. For GIT SS, most of the literature includes case reports and case series. This article discusses diagnosis, treatment, and literature review in synovial sarcoma.

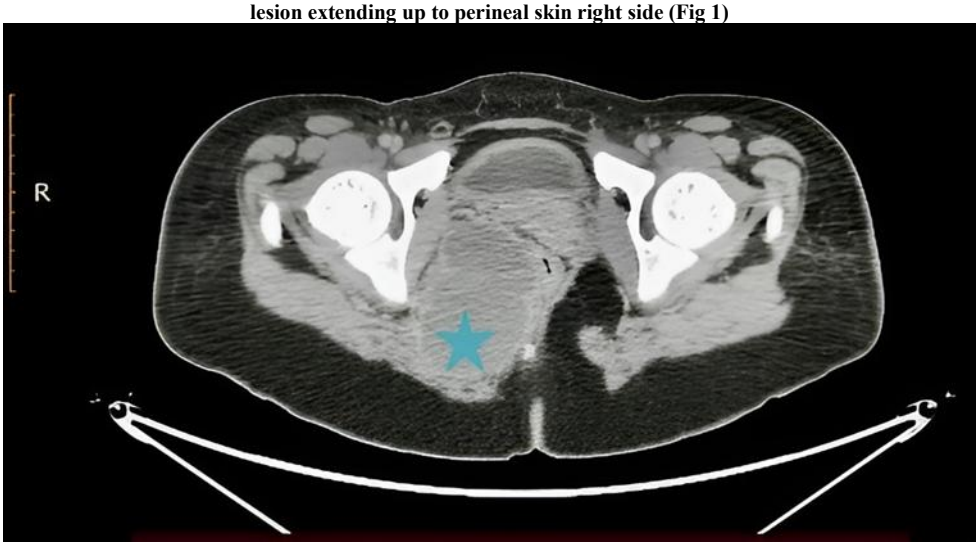
II. Case Report

A 30-year-old lady presented to our surgical oncology outpatient department with rectal bleeding and perineal discomfort, having no chronic illness. On general and local examination, the patient had a performance

status of ECOG 0; vitals had been within normal limits; the abdomen was soft without organomegaly; on rectal examination, the rectal mucosa was smooth with a mass palpable on the right lateral aspect 4 cm from the anal verge.

Laboratory tests revealed a CEA level of 1.39 ng/ml. Imaging studies, including CECT of the abdomen and pelvis, showed a 9.3* 8.3* 6 cm mass located in the right ischiorectal and ischioanal fossa, with no signs of distant metastasis (Fig 1). MRI pelvis demonstrated a 9.6* 6.2 cm lesion in the right ischiorectal and ischioanal fossa extending supralevator with mass effect on the anorectal junction, uterus, and bladder.

PRE NACT CECT ABDOMEN AND PELVIS 9.3*8.3*6 cm Right ischiorectal fossa, ischioanal fossa lesion extending up to perineal skin right side (Fig 1)

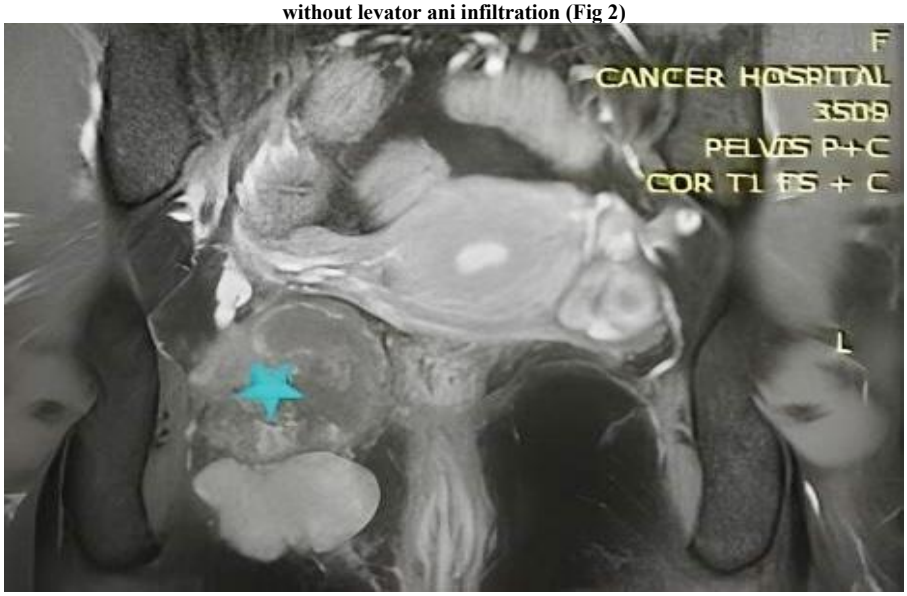


Further USG-guided biopsy of the lesion done was suggestive of differential diagnoses was between biphasic synovial sarcoma and carcinosarcoma with morphological and immunohistochemical features favouring the first diagnosis . Immunohistochemistry (IHC) tests were positive for CK7 and CK20 and negative for CDX2, CD10, PAX8, and Bcl2.

The case was discussed at an MDT (multidisciplinary tumor board) and planned for 3 cycles of neoadjuvant chemotherapy (NACT) with Doxorubicin, Ifosfamide, and Mesna followed by surgical treatment abdominopelvic resection (APR).

Post-NACT review MRI pelvis suggests reduced size to 6.3* 4.5* 7*5 cm in the right ischiorectal and ischioanal fossa without levator ani infiltration (Fig 2).

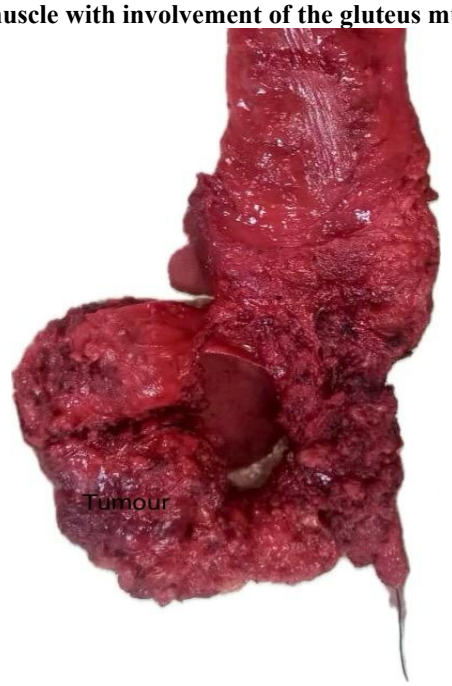
POST NACT MRI PELVIS 6.3*4.5*7.5 cm lobulated mass in right ischiorectal and ischioanal fossa without levator ani infiltration (Fig 2)



With prior consent and explaining the necessity and complications of surgery, the patient is posted for APR. In APR distal sigmoid is mobilised , ureter and gonadal vessels identified and preserved, superior rectal artery dissected from origin and ligated, colon is mobilised and mesocolon is divided , total mesorectal excision done , mobilisation of rectum with excision of mass with adjoining adherent structures done till levator muscle , colon transected, anus sutured with purse string suture , teardrop incision is made ,dissection continued till levator muscle insertion , pudendal and haemorrhoids vessel ligated , mesorectum separated from anterior presacral fascia , pubococcygeous and puborectalis muscle divided, sigmoid colon, rectum and mass removed with adherent gluteal muscle and sent for hpe, pelvic floor closed with interrupted suture and skin closure done with drain insitu . Intraoperatively a 8*5*6 cm bilobed mass adherent to the right lateral wall of the rectum extended up to the lateral pelvic wall and levator ani muscle with involvement of the gluteus muscle(Fig. 3).

SS subtypes
Differential diagnosis
Salient features

A specimen of APR with the finding of an 8*5*5 cm bilobed mass adherent to the right lateral wall of the rectum reaching up to the lateral pelvic wall and levator ani muscle with involvement of the gluteus muscle (Fig. 3).



Postoperative perineal wound infection with serous fatty discharge was present and managed conservatively; the rest was uneventful.

III. Discussion

Synovial sarcoma is a rare soft tissue malignancy, often affecting adolescents and young adults. Morphologically categorised into MSS, BSS, and PDSS subtypes. The defining genetic feature of SS is chromosomal translocation t(X;18) (p11;q12), which fuses the SS18 gene with SSX family genes (SSX1, SSX2, or SSX4), ensuring in SYT-SSX fusion, initiating tumorigenesis. Current research have explored the molecular genetics of SS, focusing on SYT-SSX fusion and identifying biomarkers such as PAX8 and estrogen receptor expression [4].

IHC markers in SS typically show positivity for CK, EMA, Bcl-2, CD99, TLE 1, and the SS18-SSX fusion protein, while markers such as CD34, S100, desmin, myogenin, and Sox10 are generally negative [5]. In comparison, carcinosarcoma normally expresses AE1/AE2, vimentin, desmin, CD10, p16, p53, and PAX8 in IHC testing.

Morphologically, many differentials are present for MSS, BSS, and PDSS.(Table1)

MSS	Malignant peripheral nerve sheath tumor (MPNST)	SOX10, molecular studies and clinical history are useful in making the diagnosis; shows focal keratin positivity and TLE1+ in up to 30%.
	Cellular schwannoma Dilated vessels with hyalinized walls	S100 and SOX10 are diffusely positive, TLE1+ in most cases.

	Solitary fibrous tumor	CD34+ with a characteristic patternless pattern STAT6+ ,TLE1 can be positive in up to 40%
	Leiomyosarcoma	May be desmin+, SMA+, calponin+, hcaldesmon+
	Spindle cell rhabdomyosarcoma	Desmin+, myogenin+, MyoD1+
	Adult fibrosarcoma	Diagnosis of exclusion
	Dermatofibrosarcoma protuberans	Usually CD34+
	Epithelioid sarcoma	Can be keratin+, TLE1+ (30%) and have spindle cell morphology CD34+ (50%) and INI1 lost in majority.
	Biphenotypic sinonasal sarcoma	Has neural and myogenic IHC markers, including S100 and SMA, calponin, desmin or myogenin
	Sarcomatoid carcinoma	Keratin+ TLE1- and lacks the SSX translocation
BSS	Adenocarcinoma	Lack spindle cell areas and are typically TLE1-
	Biphasic mesothelioma	Can show sarcomatoid features and cytokeratin positivity but WT1+ and lacks the SSX translocation
	Glandular nerve sheath tumors	Very rare
	Branchial anlage mixed tumor	Composed of spindle cells and epithelial tissue, should also see mature adipose tissue, myoepithelial expression (CK5/6+, CK14+, SMA+, CD10+,calponin
PDSS	Small round blue cell tumors	SS18-SSX translocation studies and lineage specific markers, such as myogenin, MyoD1, desmin, FLI1 are useful

The primary concern for differentiating synovial sarcoma from other mesenchymal tumour like GIST (Gastrointestinal stromal tumours), leiomyomas, leiomyosarcomas, schwannomas, solitary fibrous tumours, as they share common morphological features making accurate diagnosis reliant on immunohistochemical staining and molecular analysis.

Biphasic synovial sarcoma though more common in extremity can occasionally present in GIT in less then 1%, which include rectum. Among GIT most common location is stomach followed by colorectal, small intestine, gastroesophageal junction and esophagus.

Rectal SS often present with non specific symptoms like rectal bleeding, abdominal pain mimicking other GIT disorder. Diagnosis generally entails imaging like CECT or MRI Abdomen and pelvis accompanied through image guided core needle biopsy requiring IHC.

In imaging USG is non specific shows heterogeneous hypoechoic mass. CT scan along with soft tissue heterogeneous density mass additionally suggests calcification if present.The MRI is modality of choice showing the triple sign in T2WI observed as imaging characteristics due to area of necrosis, cystic degeneration, dystrophic calcification and fibrotic band, T1WI shows lesion isointense to muscle and T1 with contrast enhancement is prominent and may be diffuse (40%), heterogeneous (40%) or peripheral (20%).[6]

The selection between APR and ELAPE (Extralevator abdominopelvic resection) remains a subject of discussion.[7] While APR is the standard procedure for advanced lower rectal cancer, it carries a higher risk of positive circumferential resection margins (CRM+), and intraoperative perforation (IOP), due to complex anatomy of rectum. These factors can increase the chance of local recurrence of the tumor.

After the introduction of cylindrical APR ,European multicenter showed that with the use of cylindrical APR, the rate of CRM+ reduced from 49.6% to 20.3%, and that the incidence of IOP fell from 28.2% to 8.2%, and this study recommended adoption of extralevator abdominoperineal excision (ELAPE) instead of cylindrical APR.[8] But ELAPE is seen to increase the perineal wound infection, perineal hernia compared to APR and also large pelvic defect requires reconstruction of pelvic floor using myocutaneous flap (gluteal rotation/advancement

flaps, inferior gluteal artery myocutaneous island transposition flaps, transverse rectus/vertical rectus abdominis, and gracilis) or human acellular dermal matrix or biological mesh closure.

For neoadjuvant therapy, neoadjuvant chemotherapy or chemoradiation are evaluated in singleor multicenter studies in high-grade tumors; however, most data speaks about extremity sarcoma management. A randomized study that compared surgery alone versus neoadjuvant chemotherapy followed by surgery in 134 evaluable patients with high-risk tumors (tumors ≥8 cm of any grade, grade II/III tumors <8 cm, grade II/III locally recurrent tumors, or tumors with inadequate surgery) did not show a major survival benefit for patients receiving chemotherapy. The estimated 5-year disease-free survival (DFS) rate was 52% for the no-chemotherapy arm and 56% for the chemotherapy arm. The corresponding 5-year OS rate for both arms was 64% and 65%, respectively.[9] Another single-institution study involving 48 patients with high-grade extremity STS (≥8 cm), the outcome of patients treated with neoadjuvant chemoradiation with the MAID (mesna, doxorubicin, ifosfamide, and dacarbazine) regimen followed by surgery and adjuvant chemotherapy with the same regimen was superior.[10] RTOG9514 study followed the same protocol .Long-term follow-up data of these studies confirmed that neoadjuvant chemoradiation followed by resection and adjuvant chemotherapy with a doxorubicin-based regimen improves local control and OS and DFS rates in patients with high-grade STS of extremity and body wall. Another ongoing trial, STRASS II, is evaluating the role of neoadjuvant chemotherapy in high-risk retroperitoneal STS.

Available evidence from meta-analyses [11, 12] and randomized clinical trials [13, 14] suggests that adjuvant chemotherapy improves RFS in patients with STS of extremities. However, it is not clear if the conclusions from these trials are applicable to retroperitoneal/intra-abdominal sarcomas, and thus care should be individualized.

Chemotherapy with single agents (dacarbazine, doxorubicin, epirubicin, or ifosfamide) or anthracycline-based combination regimens (doxorubicin or epirubicin with ifosfamide and/or dacarbazine) has been widely used for patients with advanced, unresectable, or metastatic disease.

There are case reports and case series for GIT SS; most of them emphasize clinical-pathological and IHC features.

Another study by Junlin Zhang et al. summarized 68 cases of SS in GIT; they concluded that with higher age and large size tumors (>5 cm), there may be a high risk of progression to demise after diagnosis. In author case report SS stained + with vimentin , CD99 , Bcl2 EMA and MiB-1 suggest MSS.[15]

Marc Ladanyi and team has retrospectively analysed the clinical behaviour of SS in SYT-SSX Fusion type in which they observed 25% are BSS and 74% are MSS, median and 5yr OS were 6.1 yr and 13.7 year respectively OS better in SYT-SSX2 cases and size <5 cm tumour .Age, sex, histological type, and axial versus peripheral primary site had no impact on overall survival.SYT-SSX fusion type appears to exert impact on prognosis.[16]

Ho Xuan Tuan published a rare case of perineal SS in 17 year male , MRI allows for identification of triple sign and core needle biopsy done for tumour grade and IHC , post wide local excision hpe reveals monophasic SS and anthracycline based CT given post operative, patient died after 1 year.[17]

Silvia Stacchiotti and Brian Andrew Van Tine mentioned about the SS treatment prospect they focused on the pharmacologic management of SS, both in the curative setting, where the standard approach is wide surgical excision combined with radiotherapy and/or (neo)adjuvant chemotherapy as appropriate, and in the palliative setting. In advanced disease, chemotherapy with anthracyclines and/or ifosfamide, trabectedin, or pazopanib has been demonstrated to be more active compared with other soft tissue sarcomas. There include targeted agents, immunotherapy, and metabolic therapies.[18]

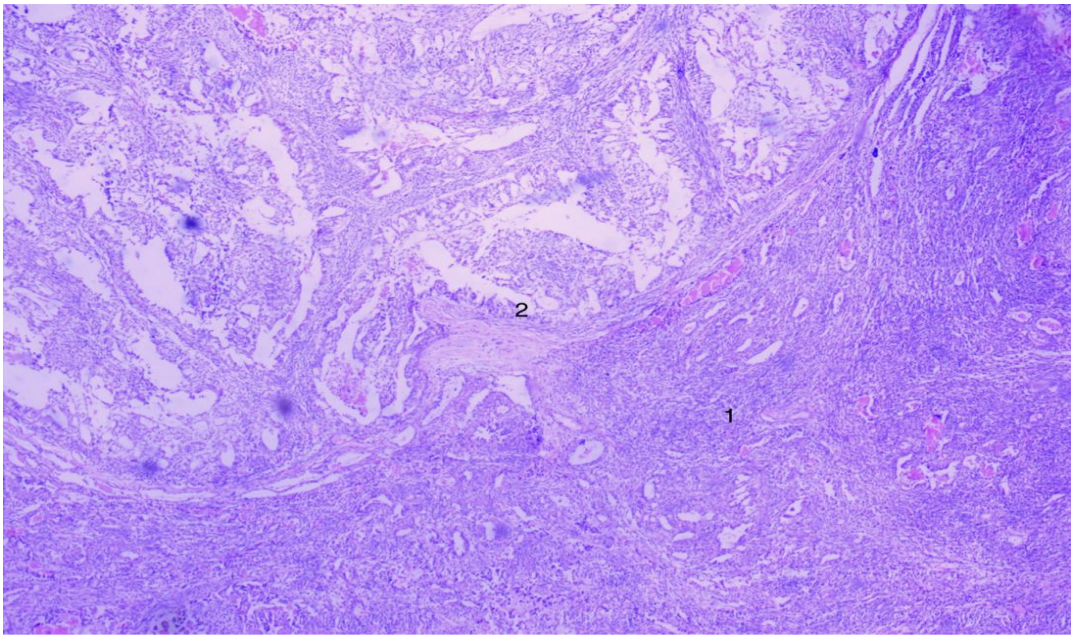
A Khan et al reported a case of SS of descending colon in which they noted a polyp in descending colon subjected to polypectomy , HPE exhibits SS , IHC stains SS18-SSX fusion protein.SS is considered aggressive and high grade with metastatic ability, so prompt diagnosis and early management is importantly for better patient outcome.[19]

Michael R. Freund and team assessed the significance of resection in rectal sarcoma in 133 patient representing only 0.03% of whole rectal cancer NCDB database over a period of 16 years, they observed the mean age of 65 years with mean size of tumour 6.1 cm having OS 22.5% . They concluded rectal malignancy is very rare with a poorer prognosis, and undergoing surgery with curative intent with a negative margin may confer better OS [20].

In our case we worked up a 30-year-old lady with rectal biphasic SS as per our institute protocol and discussed the case with the MDT, where 3 cycles of NACT (Doxorubicin, Ifosfamide, and Mesna) were planned. Post-NACT there is a moderate reduction in size of the tumor from 9.3*8.3*6 cm to 6.3*7.5*4.5 cm. Further the patient was subjected to APR.

Histopathology examination (HPE) suggests a poorly differentiated neoplasm with D/D of carcinosarcoma and biphasic synovial sarcoma. The tumor is 12*9.7*3.4 cm, showing glandular structure and spindle cells. (Fig 4). The tumor is infiltrating through the muscularis propria into pericolonic tissue, and 5 out of

6 LNs are positive without ENE. IHC positive for CK7, CK20, SS18-SSX/SS18, and IHC negative for Bcl2, CD10, CDX2, PAX8, PR, and TLE, favoring biphasic synovial sarcoma.



Biphasic Synovial Sarcoma histopathology having spindle cells 1 and gland-like epithelial structures 2. (Fig 4)

So comparing from previous literature, this case has a younger age of presentation (30 years vs. >50 years), size >5 cm, poorly differentiated neoplasm, and biphasic SS on HPE and IHC, without metastasis, and underwent APR for a negative margin (in preference to ELAPE), favoring the aggressive nature of the disease and high risk. There are not any established recommendations; however, the best mode is extensive surgical resection with tumor-free margins followed by chemoradiation/chemotherapy or neoadjuvant chemotherapy/radiation followed by surgical resection and adjuvant chemotherapy/radiation.

Prognosis for SS varies according to tumor size, location, grade, morphology, age at diagnosis, mitosis activity, IHC, and presence of metastasis at diagnosis. Generally, SS is aggressive with a poor prognosis, but the biphasic variant has a good outcome compared to MSS and PSS [6]. The 5- and 10-year survival rates for adults with SS are 62% and 52%, respectively [21].

IV. Conclusion

Rectal biphasic synovial sarcoma is an exceedingly rare malignancy with a generally poor prognosis; however, surgical resection with a negative margin may confer better overall survival. Our review and evaluation of SS in the rectum and other GIT found that patients with aging or a large tumor size (≥ 5 cm) have a higher risk and poor prognosis. We advocate that SS be considered inside the differential for any spindle cell lesion in the digestive tract. IHC is to be considered in making the differential for SS.

Statement of Ethics: Ethical approval is not required for this case report in accordance with national guidelines. Written informed consent was obtained from patient for publication of the details of their case and any accompanying images (available upon request).

Conflict of interest: The authors has no conflicts of interest to declare.

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