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Case Report

Pathology Section

Rare Presentation of Metastatic Dedifferentiated Liposarcoma of Paratesticular Origin to the Colon Mimicking Gastrointestinal Mesenchymal Tumours: A Case Report

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ABSTRACT

Mesenchymal tumours of the Gastrointestinal (GI) tract are relatively uncommon, with Gastrointestinal Stromal Tumours (GISTs) representing the most frequent subtype. Other mesenchymal neoplasms of the GI tract are far rarer and encompass a heterogeneous group of lesions that resemble their soft-tissue counterparts in other anatomical sites. These include lipoma, liposarcoma, leiomyoma, leiomyosarcoma, desmoid tumours, schwannoma, inflammatory fibroid polyps, and fibromatosis. Because of overlapping clinical and morphological features, distinguishing these tumours can be challenging. We report the case of a 67-year-old male who presented with an abdominal mass and diarrhoea. Imaging studies demonstrated retroperitoneal lesions with extension to the colon. A right hemicolectomy revealed two serosal-based tumours. Histopathological evaluation showed a spindle-cell neoplasm exhibiting marked atypia, brisk mitotic activity, and occasional lipoblasts. Given the broad differential diagnosis- including GIST and other spindle-cell sarcomas- immunohistochemistry was crucial. The tumour showed strong nuclear positivity for MDM2, confirming the diagnosis of metastatic Dedifferentiated Liposarcoma (DDLPS). Correlation with previous medical history revealed an earlier diagnosis of paratesticular DDLPS, establishing these colonic lesions as metastatic deposits. The DDLPS is a rare, high-grade mesenchymal tumour, and its occurrence in the paratesticular region is particularly uncommon. Metastasis to the colon is exceedingly rare, with only isolated cases described in the literature. This case underscores the importance of comprehensive histopathological analysis, clinical correlation, and the use of immunohistochemical markers such as MDM2 in reaching an accurate diagnosis. Awareness of these rare metastatic patterns is essential to avoid misinterpretation as more common GI mesenchymal tumours and to guide appropriate therapeutic management.

Keywords: Gastrointestinal stromal tumor, Immunohistochemistry, Spindle-cell neoplasm

CASE REPORT

A 67-year-old male patient was referred to our hospital with the chief complaint of a palpable abdominal mass, which had been gradually increasing in size over the past two months. He had a recent history of diarrhoea for two days, occurring 4-5 times per day. There was no history of weight loss, constipation, rectal bleeding, melena, abdominal pain, loss of appetite, or fever. The patient's past medical history revealed a high inguinal orchidectomy for a paratesticular mass two years prior, was diagnosed as DDLPS (high-grade). The patient had not undergone adjuvant chemotherapy or radiotherapy.

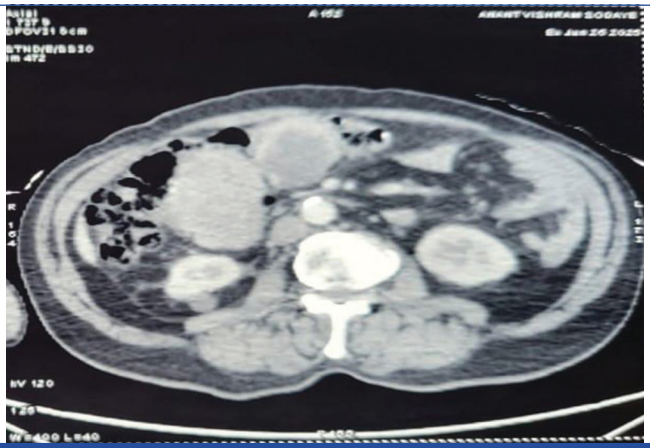
Abdominal examination revealed a non-tender, globular, smooth lump. Vital signs and routine haematological investigations were within normal limits.

Contrast-Enhanced Computed Tomography (CECT) showed two retroperitoneal lesions. The first lesion measured 10.6×6.5×7.6 cm in the right anterior pararenal space. The second lesion measured 6.3×6.2×5.1 cm, located superoanterior to the first mass in the transverse mesocolon, abutting the proximal colon with loss of fat planes [Table/Fig-1].

Differential diagnosis on the basis of CECT was:

1. Retroperitoneal metastases with mesenteric intraperitoneal extension;
2. Ascending colon GIST.

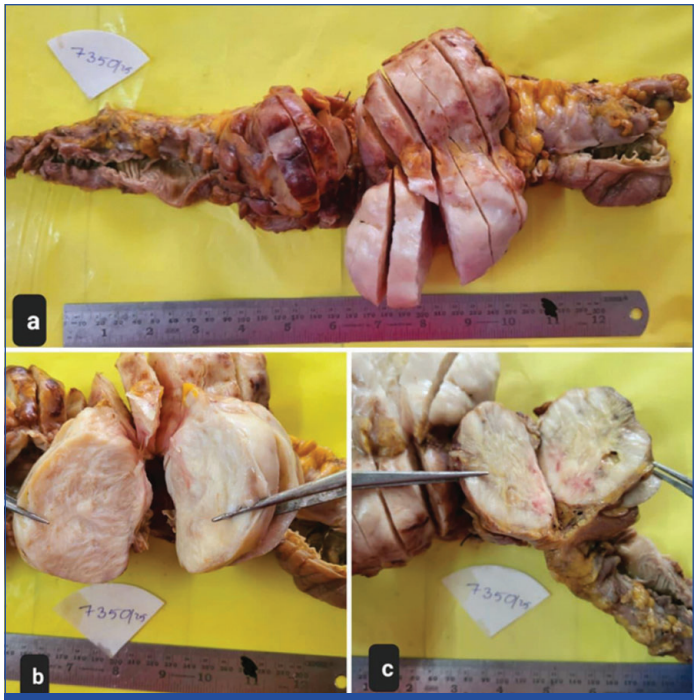
The patient underwent exploratory laparotomy for right hemicolectomy with excision of the abdominal lumps. Ileocolic anastomosis was performed.



[Table/Fig-1]: Contrast-Enhanced Computed Tomography (CECT) showed two retroperitoneal lesions. The first lesion measured 10.6×6.5×7.6 cm in the right anterior pararenal space. The second lesion measured 6.3×6.2×5.1 cm, located superoanterior to the first mass in the transverse mesocolon.

The resected specimen of the right hemicolectomy showed two well-circumscribed serosal surface tumour masses. The larger (proximal) tumour measured 11×9×6 cm. The smaller (distal) tumour measured 6×6×5 cm. The distance between the two masses was 3.5 cm.

On gross examination, both tumours were solid, firm, grey-white, with a whorled appearance. The larger mass grossly resembled a smooth muscle tumour, prompting an initial impression of leiomyomatous tumour. The smaller mass showed focal myxoid areas with a few yellow streaks [Table/Fig-2].



[Table/Fig-2]: a) On gross right hemicolectomy, specimen showing two tumour on serosal surface. On cut surface; b) Larger tumour shows solid grey white, whorled appearance; and c) Smaller tumour shows myxoid areas and few fatty streaks.

Microscopic examination from both tumour masses revealed circumscribed spindle cell tumour arranged in interlacing fascicles and bundles infiltrating up to the mucosa. The tumour cells exhibited pleomorphic, plump ovoid nuclei; coarse chromatin with prominent eosinophilic nucleoli; ill-defined cytoplasm; bizarre tumour cells and multinucleated tumour giant cells. Mitotic activity was 10-20/10 HPF.

Sections from the smaller tumour showed similar histology. However, areas with myxoid change, occasional lipoblasts, and mature adipose tissue with variable-sized adipocytes were also seen. The colonic mucosa overlying both tumours showed ulceration.

The histopathological differential diagnosis was GIST, leiomyosarcoma and liposarcoma.

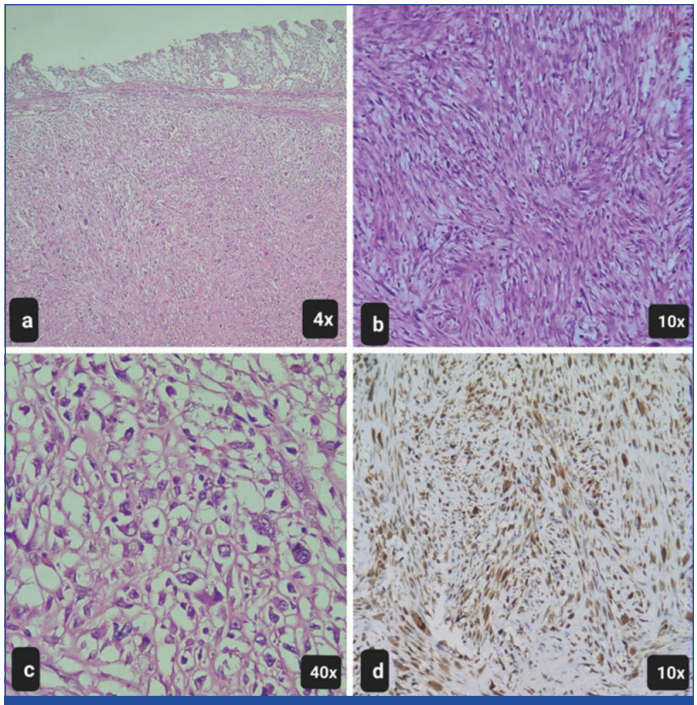
Immunohistochemistry was performed taking into consideration of CECT findings of retroperitoneal mass and histopathological findings of spindle cell morphology. CD117 and DOG-1 were negative ruling out GIST. S100 was negative, ruling out neurogenic origin. MDM2 was strongly positive thus confirmed the diagnosis of DDLPS [Table/Fig-3]. On follow-up, the patient was continuing chemotherapy at the time of writing this report.

DISCUSSION

Liposarcoma is the most common soft-tissue sarcomas in adults, with the deep soft-tissues of extremities and retroperitoneum being the most frequent sites [1]. DDLPS is approximately 18% of all liposarcomas and demonstrates a strong predilection for the retroperitoneum. However, primary GI or intraperitoneal involvement is rare; when present, it is usually secondary, resulting from extension of a retroperitoneal mass. Among the few reported GI-involving cases, the ascending colon, mesocolon, small-bowel mesentery, omentum or peritoneum have been described [2-5].

DDLPS often manifests macroscopically as a large, firm, grey-white solid mass representing the high-grade, non-lipogenic component, together with soft, yellowish areas characteristic of the well-differentiated lipogenic portion. The dedifferentiated and well-differentiated components are frequently well demarcated, although sampling only the solid areas may lead to missed diagnosis of the lipogenic component [6].

Intraperitoneal liposarcoma involving omentum, mesentery or peritoneum has also been reported, although such occurrences



[Table/Fig-3]: a) Spindle cell tumour arranged in interlacing fascicles and bundles, the colonic mucosa overlying tumours showed ulceration (4x); b) The tumour cells exhibited pleomorphic, plump ovoid nuclei; coarse chromatin with prominent eosinophilic nucleoli; ill-defined cytoplasm (10x); c) Areas with myxoid change, occasional lipoblasts, and mature adipose tissue with variable-sized adipocytes were also seen (40x); d) On immunohistochemistry MDM2 was strongly positive (10x).

remain extremely uncommon [3,7]. Taken together, the literature underscores that although the retroperitoneum remains the most common site of origin, DDLPS can rarely manifest within the peritoneal cavity either primarily (very rarely) or more commonly by extension with potential for transmural bowel invasion.

Although rare, several cases of mesenteric DDLPS with direct bowel involvement have been documented. A small-bowel mesenteric DDLPS with transmural invasion into the bowel wall and submucosa was described by Dhakal S and Prajapati I a presentation remarkably similar to the present case [8]. Other series have described giant mesenteric DDLPS masses displacing bowel loops peripherally, sometimes with multiple synchronous mesenteric/extraperitoneal nodules that preclude curative excision [4,5]. In such cases, clinical presentation and imaging (CECT) often suggest alternate diagnoses such as GIST or leiomyomatous tumour, making histopathology and immunohistochemistry (especially demonstration of MDM2 and/or CDK4 overexpression) essential for correct diagnosis [Table/Fig-4].

Feature	Present case	Dhakal S and Prajapati I [8]	Meher S et al., [2]
GI involvement	Yes- transmural invasion of bowel wall	Yes- transmural invasion	Mass abutting bowel; mass effect
Typical imaging differential	GIST/leiomyomatous tumour	GIST	GIST/sarcoma
Gross morphology	Firm grey-white mass with yellow fatty areas	Large solid mass with heterogeneous cut surface	Large mass with solid and fatty components
IHC/Molecular	MDM2+	MDM2+, CDK4+	MDM2+, CDK4+
Local invasion	Present—bowel wall invasion	Present- transmural	Variable
Recurrence risk	High due to site and dedifferentiation	High (literature-based)	High (literature-based)
Prognosis	Guarded; high-grade retroperitoneal tumour	Guarded	Guarded

[Table/Fig-4]: Comparison of present case vs representative published cases [2,8].

DDLPS is universally regarded as a high-grade sarcoma, with a substantial risk of local recurrence following resection. Published

series report local recurrence rates in the order of 40% or higher, particularly in retroperitoneal tumours [9].

Moreover, DDLPS exhibits a non-negligible propensity for distant metastasis overall metastatic rates are reported in the range of 15-30%. Among these, the lung is the most common site, followed by soft-tissue/subcutaneous tissue, lymph nodes, liver, and bone [10]. In one large review, the median time to metastasis was as short as eight months [11].

Retroperitoneal DDLPS tends to have especially poor outcomes because of its deep anatomical location, its typically large size at diagnosis, and its common extension into neighboring organs. When the tumour shows transmural involvement of the bowel, as in this case, the likelihood of leaving behind residual disease and the risk of local recurrence are further increased [2,4].

CONCLUSION(S)

The DDLPS is a rare, high-grade mesenchymal tumour, especially when arising from the paratesticular region. Even rarer is its metastasis to the colon. This case highlights the importance of detailed histopathological analysis and immunohistochemistry in reaching an accurate diagnosis, especially when clinical, radiological and morphological findings mimic other common entities such as GIST or leiomyomatous tumour.

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