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Case Series

Beyond the commons: diagnosis and management of rare intra-abdominal masses-a case series

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ABSTRACT

Rare intra-abdominal masses pose significant challenges in diagnosis and management due to their uncommon presentation and varied etiology. This retrospective case series evaluates four unique cases, including primary retroperitoneal melanoma (PRM), small bowel leiomyosarcoma, ruptured desmoid tumor in familial adenomatous polyposis (FAP), and small intestinal Ewing's sarcoma. Data were collected from the government Stanley medical college between 2019 and 2023. The study highlights the role of advanced imaging, immunohistochemistry, and multidisciplinary collaboration in improving diagnostic accuracy and treatment outcomes. Prognoses varied, with some cases showing poor outcomes despite aggressive management. This study underscores the necessity of individualized treatment protocols for such rare conditions.

Keywords: Primary small bowel leiomyosarcoma, Primary retroperitoneal melanoma, Ruptured desmoid tumour, Small intestinal lymphoma, Rare intra-abdominal masses, Immunohistochemistry, Multidisciplinary management

INTRODUCTION

Intra-abdominal masses can arise from any abdominal structure, including the gastrointestinal tract, liver, and peritoneum, and vary from benign to malignant. While common masses are well-documented, rare types remain poorly understood due to their low incidence and diagnostic complexity.[1] Misdiagnosis and treatment delays are common, impacting patient outcomes.[2]

This study focuses on rare intra-abdominal masses, emphasizing the need for specialized diagnostic approaches such as immunohistochemistry and high-resolution imaging. Due to the limited clinical experience with these conditions, establishing effective treatment guidelines remains a challenge.[3]

The rarity of cases discussed here may provide valuable insights into their presentation, diagnosis, and treatment.

CASE SERIES

Case 1

A 53-year-old, post-menopausal female presented with a history of abdominal pain for 1 week. Past and medical histories were insignificant. Abdominal examination revealed fullness in the hypogastrium. General physical and chest examination was normal. CECT abdomen and pelvis showed a well-defined heterogeneously enhancing lesion of size 10×15×15 cm in the pelvis with internal necrosis encasing branches of the inferior mesenteric artery and compressing left iliac vessels (Figure 1).

Laparotomy revealed a black coloured mass occupying the lower abdomen and pelvis, with omentum showing multiple tiny dark spots. Since the mass was adherent to major vessels, complete dissection of the mass was not possible; a biopsy was taken from the mass wall and omentum. Solid organs appeared normal. Intraoperative

image of the mass occupying the lower abdomen and pelvis (Figure 2).

Omentum showed sheets of atypical cells with melanin pigments. Given the retroperitoneal location of the mass, it was suspected to be a secondary deposit, and a search for primary melanoma was made.

HPE showed a cyst wall with malignant cells with eosinophilic cytoplasm and round vesicular nuclei with intra- and extracytoplasmic melanin pigmentation. IHC staining showed positivity for HMB45, Pan CK, and CD117 (Figure 3).

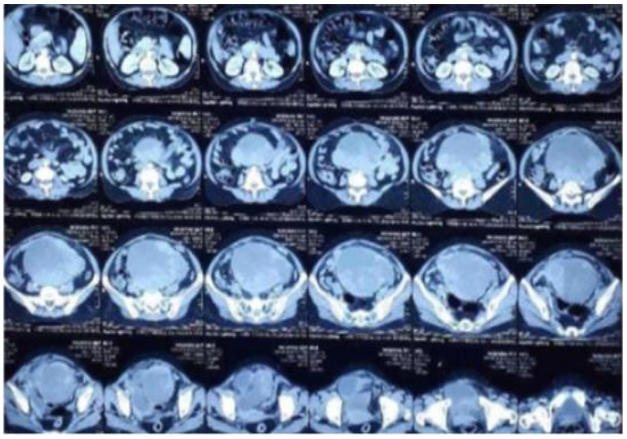

Figure 1: CECT abdomen and pelvis showed a well-defined heterogeneously enhancing lesion of size 10x15x15 cm in the pelvis with internal necrosis encasing branches of the inferior mesenteric artery and compressing left iliac vessels.

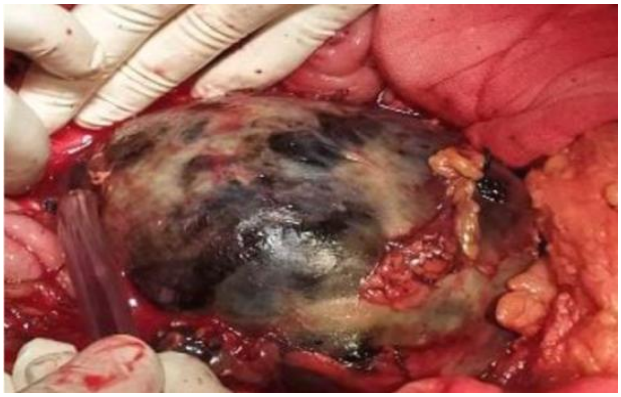

Figure 2: Intraoperative image of the mass occupying the lower abdomen and pelvis.

The patient did not have any pre-existing skin lesions nor a family history suggestive of malignant melanoma. Detailed dermatological, ocular, and mucosal evaluation failed to identify lesions that can be considered as primary, leading to the diagnosis of this case as a PRM. The patient was started on imatinib chemotherapy and kept on regular follow-up, and was unremarkable until three months back, when she succumbed to the disease after developing corneal metastasis resistant to radiotherapy.

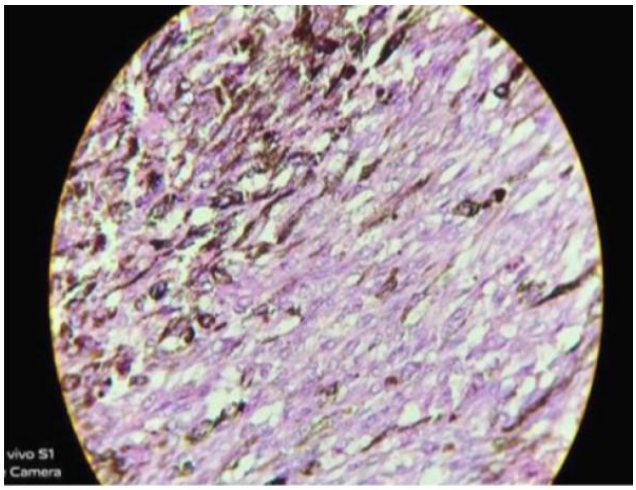

Figure 3: HPE showed a cyst wall with malignant cells with eosinophilic cytoplasm and round vesicular nuclei with intra- and extracytoplasmic melanin pigmentation.

Case 2

A 70-year-old female presented with early satiety and fullness of the upper abdomen for 1 month. The patient had no history of bowel disturbance, loss of weight loss, or loss of appetite. On examination patient was pale, the abdomen was distended, and a vague mass of size 15×10 cm was palpable occupying the epigastric, left hypochondrium, and left iliac fossa, which was firm in consistency and dull on percussion.

CECT abdomen showed a well-defined lobulated enhancing lesion with central non-enhancing areas arising from the DJ junction and proximal jejunum, measuring approximately 9.8×14.4×18.3 cm, with a possibility of GIST (Figure 4).

After explaining the disease and prognosis, the patient was planned for resection of the primary tumor.

Intraoperative images showing an 18×10×7cm mass adherent to the 4th part of the duodenum, ascending colon, and transverse mesocolon with high vascularity. The rest of the small bowel, large bowel, omentum, mesentery, solid organs, and peritoneum appeared normal. The intraoperative and postoperative period was uneventful (Figure 5).

HPE report was leiomyosarcoma with IHC positive for SMA, negative for CD117, DOG1, Myogenin, and Ki67=7-10% (Figure 6).

The patient was diagnosed with small bowel leiomyosarcoma and started on adjuvant chemotherapy with doxorubicin and gemcitabine.

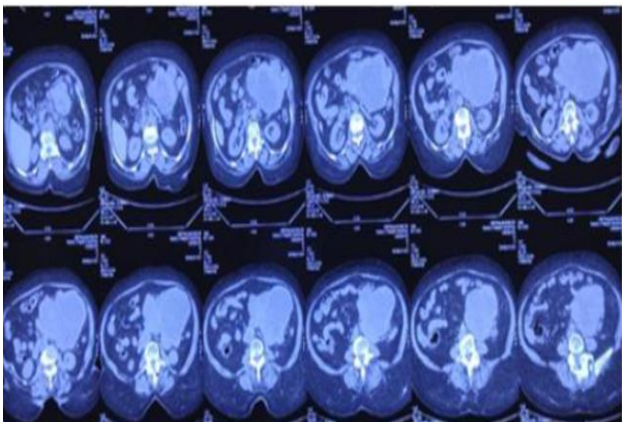

Figure 4: CECT abdomen showed a well-defined lobulated enhancing lesion with central non-enhancing areas arising from the DJ junction and proximal jejunum, measuring approximately 9.8×14.4×18.3 cm with a possibility of GIST.

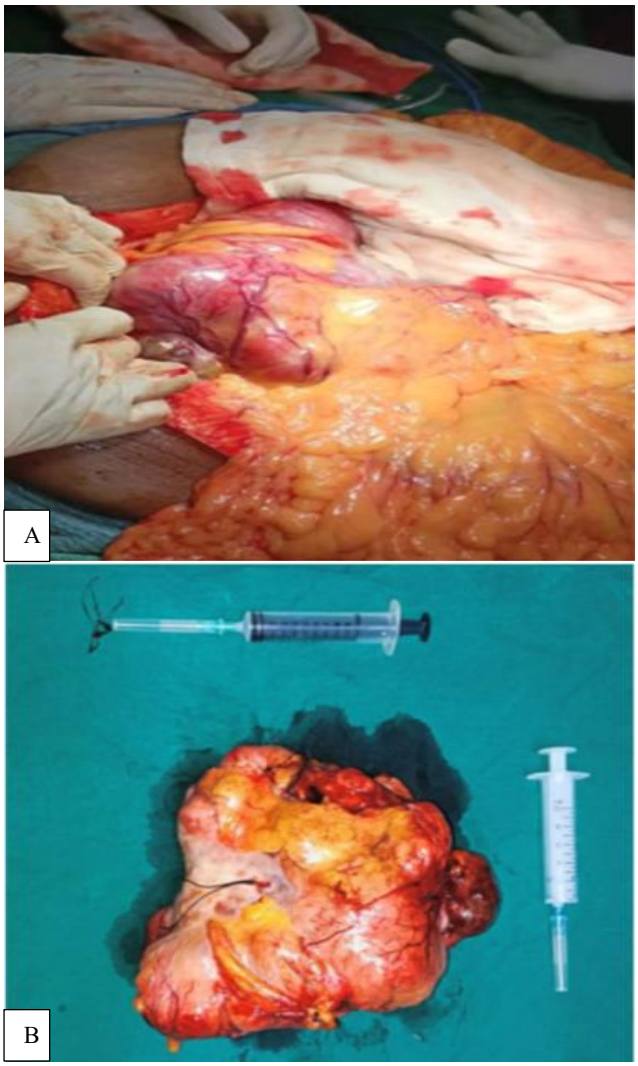

Figure 5 (A and B): Intraoperative images showing an 18×10×7cm mass adherent to the 4th part of the duodenum, ascending colon, and transverse mesocolon with high vascularity.

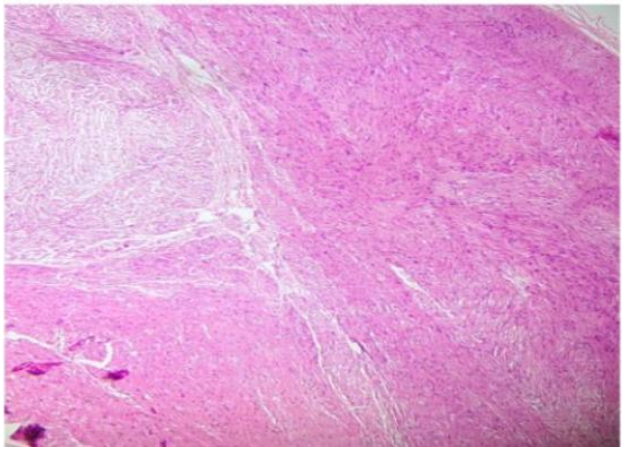

Figure 6: HPE report was leiomyosarcoma with IHC positive for SMA, negative for CD117, DOG1, Myogenin, and Ki 67=7-10%.

Case 3

A 34-year-old male patient presented to the emergency room with 20 days of vague abdominal pain, acutely exacerbating for the past 12 hours, with episodes of vomiting. Apart from a maternal history of gastric carcinoma in her fourth decade, the patient's history was unremarkable.

On examination, the patient was tachypneic and tachycardic with abdominal distention and diffuse guarding. Per rectal examination revealed a polypoidal lesion 2×2 cm, 6 cm from the anal verge. Emergency CECT abdomen and pelvis showed a heterogeneously enhancing lesion of size 12×9×8 cm communicating with ileal loops with pneumoperitoneum and free fluid in the pelvis.

Emergency laparotomy revealed hemoperitoneum with the above-described mass with a perforation (Figure 7).

En masse resection of the lesion and bowel with a proximal jejunostomy and distal ileum as a mucous fistula was performed along with biopsy of the rectal polyp. HPE revealed an intra-abdominal desmoid and a tubulo-villous rectal adenoma with low-grade dysplasia (Figure 7).

A colonoscopy revealed multiple sessile rectal polyps. The patient opted against proctocolectomy and was placed under surveillance. At the 6th month follow-up, a colonoscopy revealed multiple colonic pseudopolyps and pancolitis with ulceroproliferative growth in the lower 1/3rd rectum. Further workup revealed elevated CEA levels with presacral nodes, and HPE ultimately confirmed rectal adenocarcinoma grade 2.

The patient was put on neoadjuvant capacitabine-based chemoradiotherapy, and after 4 cycles, proctocolectomy with jejunoileal anastomosis with permanent end

ileostomy was done. The patient is on 6-monthly follow-ups.

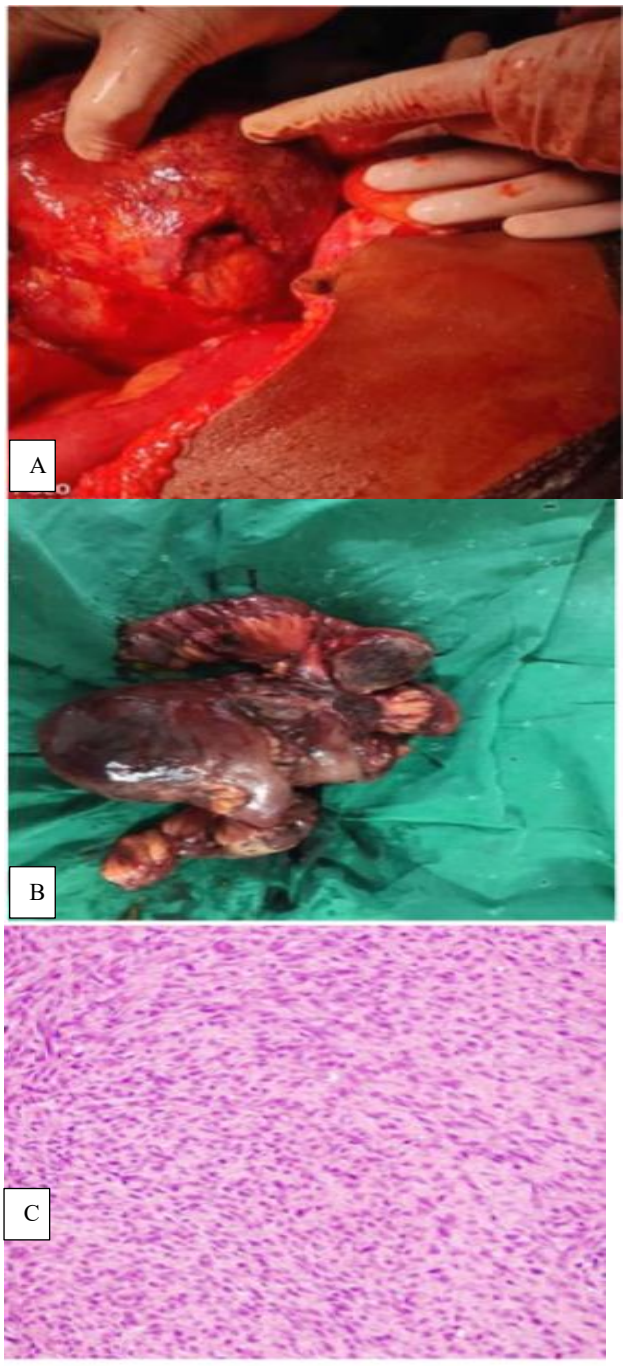

Figure 7 (A-C): Emergency laparotomy revealed hemoperitoneum with the above-described mass with a perforation; and HPE turned out as Intrabdominal Desmoid and Tubulo-villous rectal adenoma with low-grade dysplasia.

Case 4

A 26-year-old male was admitted with complaints of abdominal pain and distension for a week. On examination, the patient had tachycardia with a distended abdomen. CECT abdomen and pelvis showed a heterogeneous mass lesion of 9×6 cm arising from the ileum, and an inconclusive USG-guided biopsy of the same was inconclusive.

During evaluation, the patient developed a sudden, spontaneous bowel perforation. Emergency laparotomy with extended right hemicolectomy and ileal resection along with tumor, with end ileostomy done (Figure 8).

The postoperative period was eventful with multiple episodes of flushing ,diaphoresis and hemodynamic instability. HPE showed a diffuse small cell tumor with IHC in favor of Ewing's sarcoma (Figure 9).

Further PET CT showed peritoneal and omental metastasis, serosal deposits, and epiphrenic lymph nodal metastasis. The patient was started on imatinib and discharged later.

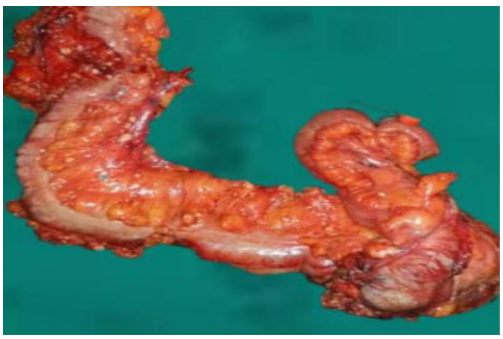

Figure 8: Resected distal ileum, cecum, ascending, and proximal transverse colon.

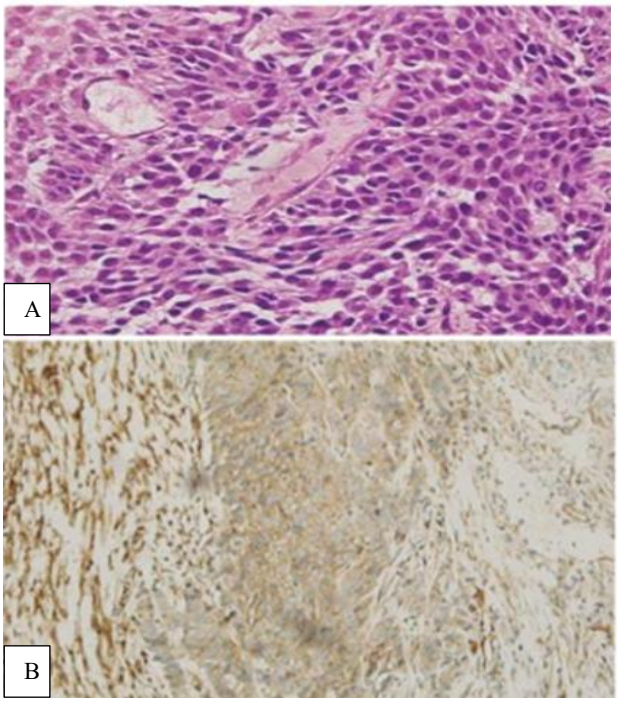

Figure 9 (A and B): HPE- diffuse small cell tumor with IHC in favor of Ewing's sarcoma.

DISCUSSION

This case series investigates rare intra-abdominal masses, emphasizing their diagnostic complexity, treatment challenges, and varied clinical outcomes. The study sheds light on four distinct cases, each highlighting rare conditions: PRM, small bowel leiomyosarcoma, ruptured desmoid tumour in FAP, and Ewing's sarcoma of the small bowel. These cases underscore the critical need for multidisciplinary approaches and advanced diagnostic tools such as immunohistochemical (IHC) staining, high-resolution imaging, and genetic testing.

PRM

Primary extracutaneous melanoma constitutes 4-5% of melanoma cases, commonly involving eyes and mucosa. Usually an incidental finding with nonspecific signs and symptoms. These lesions lack association with sun damage, family history, or precursor nevi.[4] Definitive diagnosis requires HPE and IHC studies. HPE cannot determine whether the retroperitoneal mass is PRM or metastasis from cutaneous or visceral melanoma, thus necessitating further radiological evaluation. The initial symptoms of PRM can be quite nonspecific and include fatigue, weight loss, anorexia, and leg swelling. To date, only 8 cases of PRM have been reported in the literature. Among the eight reported cases, certain specific symptoms have been noted. Half of the patients experienced abdominal pain, while 38% (3 out of 8) had abdominal distension or fullness.[5] Imaging is crucial for diagnosing PRM as it helps locate the tumor. Nevertheless, immunohistochemical stains are essential for a definitive diagnosis.

Both cutaneous and non-cutaneous melanomas share the same immunohistochemical and structural characteristics. S100 is the most sensitive marker for melanoma, while SOX10, a transcription factor, is also highly sensitive. Additionally, the BRAF gene mutation was found in about 50% of advanced melanoma. Negative immunostaining for CK7, CK20, GATA3, and PAX8 was crucial in excluding other malignancies and confirming the melanoma diagnosis.[6] Due to the rarity of primary retroperitoneal malignant melanoma, clear treatment guidelines are not established, and therapy is based on evidence from cutaneous melanoma treatment. Surgical resection is the primary treatment, but for unresectable or advanced tumors, systemic therapy is recommended. Vemurafenib and dabrafenib, both BRAF kinase inhibitors, are first-line treatments for BRAF mutation-positive melanoma, with dabrafenib having less toxicity. Additionally, PD-1 and PD-L1 inhibitors like pembrolizumab and nivolumab are proving effective for metastatic melanoma.[7]

PRM is a rare entity among the extracutaneous melanomas with an aggressive nature and grave prognosis. This case has been highlighted for its rarity. A thorough search for a primary melanoma should be made before planning the management. The case also emphasizes the role of IHC in diagnosis (HMB45, S100, SOX10) and the potential benefit of BRAF-targeted therapies in advanced stages.

Small bowel leiomyosarcoma

Tumors of the small bowel account for less than 5% of all gastrointestinal malignancies. Of these, the majority are histologically carcinoid and adenocarcinomas, with sarcomas ranking 5th (~1.2%). Leiomyosarcomas of the small bowel are extremely rare, with an incidence of 22.7 cases per million. Due to their rarity, limited demographic or clinical data are available. Since being differentiated from gastrointestinal stromal tumors (GISTs), only 26 cases of small bowel leiomyosarcomas have been reported, underscoring the importance of documenting these cases.[8]

Diagnosing small bowel tumors is challenging, often delayed until metastasis occurs. Common symptoms like weight loss, constipation, and rectal bleeding typically lead to esophagogastroduodenoscopy (OGD) and colonoscopy, but these can miss small bowel tumors. Advanced imaging techniques such as magnetic resonance enterography (MRE), CT colonography (CTC), and wireless capsule endoscopy (WCE) are more effective. MRE and CTC detect mucosal and metastatic changes, while WCE is superior for visualizing small superficial lesions.[9,10]

Histologically, leiomyosarcomas lack CD117, DOG1, and CD34 markers, which are present in GISTs, but show positive results for smooth muscle actin (SMA) and desmin. They are graded for aggressiveness using the Trojani or French systems.

Treatment primarily involves radical surgical resection. Unfortunately, most cases present late with metastases, limiting prognosis. Chemotherapy, effective in GISTs and uterine leiomyosarcomas, has not been proven beneficial for small bowel leiomyosarcomas.

Prognosis is influenced by tumor size and histological stage, and although it is somewhat better than that for small bowel adenocarcinomas, survival rates remain poor. Advanced imaging plays a critical role in early diagnosis and timely surgical intervention in these rare tumors. Our case contributes to the limited pool of knowledge regarding small bowel sarcomas.

Ruptured desmoid tumour in FAP

Desmoid tumors are rare, generally benign, and often associated with FAP. Intra-abdominal desmoids, particularly in the ileum, occur in approximately 10-25% of FAP patients, with familial history increasing the risk.[11] Desmoid development in FAP is linked to the loss of both copies of the adenomatous polyposis coli gene. Their growth rate is unpredictable, with some cases of

spontaneous regression reported. Surgical trauma is a recognized trigger, with desmoid appearing between 3 months to 5 years post-surgery.

Recurrence is common after incomplete resection, suggesting that surgical trauma might lead to new mutations. Hormonal factors, including estrogen and prostaglandins, have been implicated in the growth of desmoid. Desmoid tumors are the second leading cause of death in FAP patient's post-colectomy. Sporadic intra-abdominal desmoids are extremely rare and can present as long-standing abdominal or suprapubic masses. Symptoms may include anemia, weight loss, malnutrition, abdominal pain, ureteric obstruction, nausea, and vomiting.

Surgery, aiming for tumor-free margins, remains the primary treatment, though the proximity to vital organs complicates achieving this. Positive margins are a major cause of recurrence, though the optimal margin width is not well established.[12] Wide-margin excision, including resection of surrounding bowel, is preferred, as incomplete excision increases recurrence risk.

Adjuvant therapies such as sulindac, toremifene, radiotherapy, NSAIDs, chemotherapy (e.g., vinblastine, methotrexate), and antiestrogens (e.g., tamoxifen) are also used, particularly when full resection is not possible. Differential diagnoses for extrinsic small bowel tumors include gastrointestinal stromal tumors (GIST), lipomas, hamartomas, carcinoids, lymphomas, and metastatic adenocarcinomas.[13]

Our case was a ruptured desmoid tumour in a patient with FAP, particularly challenging due to the patient's refusal of early colectomy, which allowed the progression to adenocarcinoma. This case, being next of the only three documented instances of ruptured intra-abdominal desmoid tumors, provides invaluable insights into the management of FAP-related tumors, emphasizing the importance of regular surveillance and timely surgical intervention.[14]

The classical adenoma-carcinoma sequence was noted in our case. Periodical follow-up of the patient and relatives was provided. This case is presented for its rarity of presentation and various diagnostic and therapeutic challenges posed along its way.

Ewing's sarcoma of the small bowel

Small round-cell tumors with specific recurrent translocations, commonly EWSR1/FLI1, include Ewing Sarcoma (EWS) and primitive neuroectodermal tumors (PNET). These tumors primarily affect the bone, particularly in childhood and adolescence. Pathologically, differentiating EWS from PNET is challenging due to overlapping genetic abnormalities, leading to their classification as a spectrum of the same disease. PNET is now considered a less differentiated form of EWS, and both are treated similarly.[15] Batsakis et al categorized PNET tumors into three groups: CNS PNETs, neuroblastoma, and peripheral PNETs (pPNET). Most pPNETs occur in individuals aged 10-20, but our case involved a 42-year-old patient, highlighting the disease's potential occurrence in older individuals.[16] Recent studies suggest that age might not significantly affect prognosis, as improved chemotherapy and surgical techniques may enhance outcomes for older patients.

While pPNET typically arises in the chest, extremities, or retroperitoneum, small bowel involvement is rare. Since the first documented case in 2000, only 30 small bowel E-EWS/pPNET cases have been reported, with the most common tumor location being the ileum (61.29%), followed by the jejunum (22.58%) and duodenum (6.45%). The most frequent presentation was an abdominal mass, while gastrointestinal perforation, like in our case, was rare.[17]

Histologically, pPNETs are characterized by small, round cells, often forming rosettes. CD99 and FLI1 are commonly used markers for diagnosis, with CD99 positive in 93.1% of cases. Vimentin is also frequently positive, while markers like S100, chromogranin A, and synaptophysin show variable sensitivity.

Treatment for small bowel pPNET typically involves En bloc surgical resection followed by systemic chemotherapy. Surgery alone often results in poor outcomes, with metastatic disease being the most common cause of death. Although molecular testing, such as EWS-FLI1 fusion identification, can help confirm diagnoses, there is no standard protocol for managing small bowel pPNET, and survival times vary significantly.[18]

Perforation of the digestive tract caused by pPNET is uncommon, and the small intestine is the most common site of involvement, followed by the stomach, colon, and rectum. Given its rarity and low incidence rate, intestinal perforation caused by pPNET is often neglected and is difficult to diagnose, with only two cases reported to date, including our case, which is the third.[19]

CONCLUSION

This case series highlights the critical importance of early and accurate diagnosis of rare intra-abdominal masses. While surgical resection remains the cornerstone of treatment, the use of advanced diagnostic techniques like IHC and genetic testing is vital for confirming rare entities such as PRM or leiomyosarcoma. Furthermore, the study highlights the need for a tailored, multidisciplinary approach in managing these complex cases. The rarity of these tumors makes establishing clear treatment guidelines difficult; however, such cases contribute to better understanding and management of similar conditions in the future.

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