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Case Report

A Case of Primary Squamous Cell Carcinoma of the Small Intestine: A Case Report

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Keywords

Squamous cell carcinoma · Abdominal computed tomography scan · PET-CT · Case report

Abstract

Introduction: Primary squamous cell carcinoma (SCC) of the small intestine is an exceptionally rare malignancy, with limited cases reported in the literature. The rarity of this condition, combined with nonspecific clinical manifestations, poses significant diagnostic and therapeutic challenges. ***Case Presentation:*** We report the case of a 47-year-old woman who presented with a 2-month history of left lower abdominal pain and discomfort. Imaging studies, including enhanced abdominal CT and PET-CT, revealed a suspicious mass in the jejunum. Comprehensive diagnostic evaluations excluded metastatic origins, and a diagnosis of primary small intestinal SCC was confirmed by histopathology and immunohistochemistry. The patient underwent radical surgical resection, which revealed a poorly differentiated SCC invading the serosa and regional lymph nodes. Postoperative management included infection prevention and fluid rehydration, with recommendations for adjuvant chemotherapy and immunotherapy based on multidisciplinary consultation. Despite the advanced disease stage, the patient recovered well post-surgery and is undergoing regular follow-up. ***Conclusion:*** This case underscores the importance of thorough diagnostic evaluation to distinguish primary SCC from metastatic lesions. Early surgical intervention is critical for improving prognosis of this rare malignancy. The findings contribute to the limited knowledge of the primary SCC of the small intestine and emphasize the need for further research to guide optimal management strategies.

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Introduction

While the digestive tract is a common site for tumor development, primary malignancies of the small intestine remain exceptionally rare, comprising only a small fraction of all gastrointestinal neoplasms [1, 2]. Among benign tumors, leiomyomas are the most frequently encountered, whereas malignant small intestinal tumors primarily include adenocarcinomas, neuroendocrine tumors (carcinoids), lymphomas, and gastrointestinal stromal tumors [3]. Adenocarcinoma represents the predominant histological subtype, while mucinous adenocarcinoma, signet-ring cell carcinoma, and poorly differentiated carcinomas are also observed [4]. In contrast, primary squamous cell carcinoma (SCC) of the small intestine is exceedingly rare, with only sporadic cases documented in the literature [5].

The pathogenesis of primary small intestinal SCC remains poorly understood but may involve the malignant transformation of undifferentiated epithelial cells within the basal layer of the small intestinal mucosa [6]. The clinical manifestations are nonspecific and often include features of intestinal obstruction, such as vomiting, abdominal distension, and abdominal pain, as well as hematochezia or intestinal perforation. Some patients may present with atypical symptoms, including unexplained periumbilical pain, weight loss, or recurrent gastrointestinal bleeding.

The rarity of this malignancy, coupled with its nonspecific clinical presentation, frequently leads to delayed diagnosis and suboptimal treatment outcomes. Heightened clinical vigilance for persistent digestive symptoms is essential to facilitate early diagnosis, timely intervention, and improved survival outcomes in affected patients.

Case Report

Patient Information

Chief Complaints

The patient's chief complaint was a 2-month history of pain and discomfort in the lower left abdomen.

History of Present Illness

The patient presented to the hospital with a 2-month history of pain and discomfort localized in the left lower abdomen. The pain exhibited a dull and intermittent nature, exacerbated by eating. The pain did not radiate to other regions, and there were no accompanying symptoms such as nausea, vomiting, abdominal distension, diarrhea, black or bloody stool. The patient was initially admitted to a local hospital. An abdominal CT scan revealed a space-occupying lesion in the left lower abdomen. The patient underwent a full abdominal CT scan at an external hospital, which revealed a mass in the lower left abdomen with loss of interintestinal space and surrounding soft tissue density. The mass was lobulated with rough edges and poorly defined boundaries with the adjacent descending colon. Enlarged lymph nodes and slight thickening of the adjacent peritoneum were also observed. We have supplemented the radiological description with subsequent intraoperative measurements and histopathological findings, which confirmed a tumor size of 7.5 × 7.0 × 6.0 cm, with serosal invasion and regional lymph node metastases (shown in Fig. 1). Subsequently, the patient was transferred to our hospital for further diagnostic evaluation and treatment.

Medical, Family, and Psycho-Social History

The patient had a prior history of right-sided lung cancer treated with surgical resection 8 years ago. Pathological findings from the lung cancer indicated well-differentiated adenocarcinoma, with no recurrence on follow-up. No family history of gastrointestinal or

squamous cell malignancies was noted. The patient did not receive postoperative chemotherapy but underwent regular follow-up evaluations and remained disease-free prior to her current presentation.

Clinical Findings

Physical examination revealed a flat abdomen with no swelling or gastrointestinal type. Tenderness was noted in the left lower abdomen without rebound pain. A tough mass approximately 5 × 5 cm in size was palpable, with clear boundaries, limited mobility, and no tenderness. Vital signs were as follows: height 162 cm, weight 61 kg, BMI 23.24 kg/m^2, blood pressure 125/75 mm Hg, pulse 17 bpm, and respiration 18 bpm.

Timeline

Two Months prior to Presentation

The patient experienced intermittent dull abdominal pain localized to the left lower abdomen, worsened by eating, without associated symptoms such as nausea, vomiting, or changes in bowel habits.

Initial Medical Consultation

Abdominal CT at a local hospital revealed a space-occupying lesion in the left lower abdomen, prompting referral for further evaluation.

Hospital Presentation

The patient underwent diagnostic imaging, including enhanced CT and PET-CT and biopsy of abdominal masses.

Diagnostic Assessment

Laboratory Findings

Labs showed hemoglobin level of 78.00 g/L (anemia), stool occult blood strongly positive, non-small cell carcinoma antigen level of 3.50 ng/mL, and ferritin level of 6.58 ng/mL; there were no abnormalities in SCC antigen or infectious disease markers.

Imaging

Gastroscopy revealed esophageal submucosal eminence, non-atrophic gastritis with erosion, and diffuse polypoid lesions in the stomach and duodenum. Colonoscopy identified multiple polypoid lesions in the colorectal region with chronic mucosal inflammation and polypoid hyperplasia. Enhanced CT and PET-CT (shown in Fig. 2) revealed thickened intestinal walls, space-occupying lesions in the left abdomen, and multiple enlarged lymph nodes with increased FDG metabolism (SUVmax up to 40.7).

Histopathology

Biopsy of abdominal masses confirmed poorly differentiated carcinoma. Immunohistochemistry supported SCC with markers CK pan (3+), P63 (+), P40 (+), and Ki-67 (+80%). Excluding metastatic disease posed a challenge due to the patient's previous history of lung adenocarcinoma and imaging studies revealed multiple masses in the left abdominal cavity, multiple lymph nodes in the abdominal cavity, retroperitoneum, and left pelvic wall, and multiple nodules in both lungs.

Final Diagnosis

The postoperative pathological analysis revealed the presence of an infiltrating tumor measuring 7.5 cm × 7 cm × 6 cm in the jejunum. This tumor invaded the serosa layer and extended into the deep muscle layer of the descending colon. The tumor cells exhibited poor

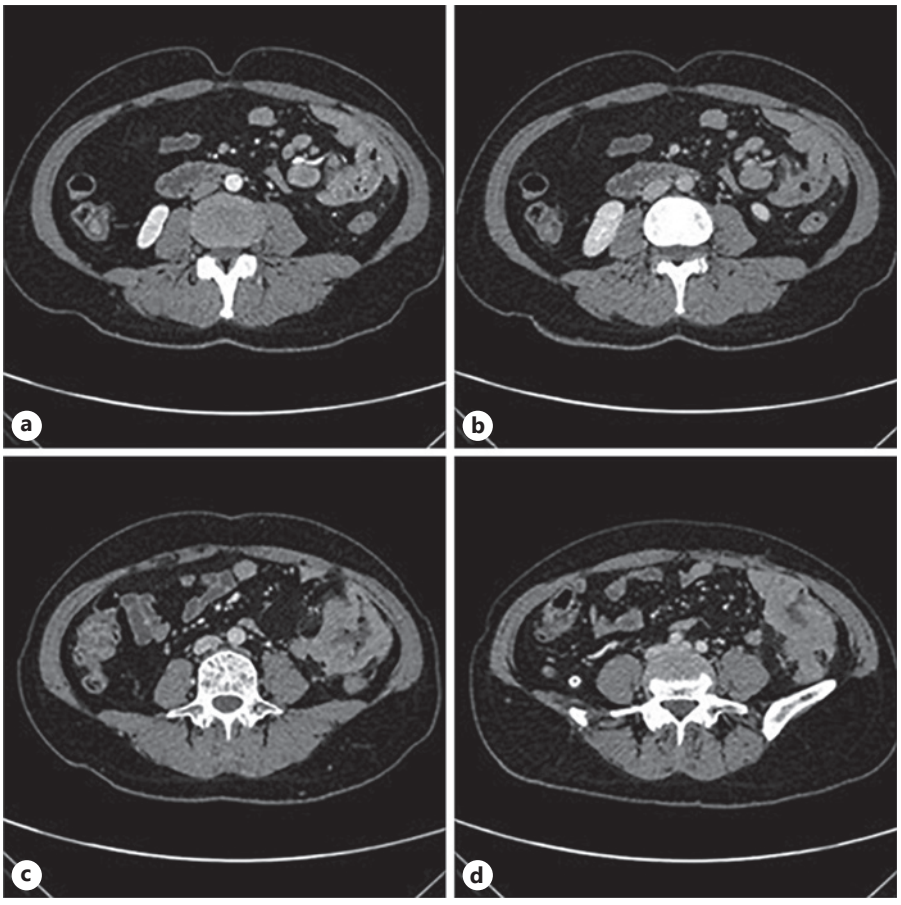

Fig. 1. a, **b** In arterial phase and venous phase, multiple lymph nodes can be seen in the abdominal cavity. **c**, **d** The local boundary between the tumor and the descending colon is not clear.

differentiation (G3: II grade/low differentiation). Immunohistochemistry confirmed the diagnosis of SCC. Histopathological examination of the resected jejunal tumor and associated lymph nodes revealed that a total of 21 lymph nodes were surgically excised and histologically analyzed. Among these, 1 lymph node (4.8%) was found to be positive for metastatic SCC. The primary tumor exhibited poorly differentiated SCC, with evidence of serosal invasion and extension into adjacent soft tissues, indicative of locally advanced disease. The nodal metastases were characterized by irregular nests of malignant squamous cells, consistent with the aggressive nature of the tumor. Furthermore, lymphovascular invasion was observed, reinforcing the aggressive behavior of the carcinoma, while perineural invasion was absent. Based on these findings, the tumor was staged as pT4N1M0, indicating locally advanced disease with regional lymph node metastasis but no evidence of distant spread. Polypoid hyperplasia of lymphoid tissue was detected in a different region of the intestinal mucosa. Additional tissue findings included a mesosalpinx cyst on the left side and fibrotic nodules exhibiting hyaline degeneration (small nodules in the intestinal wall). Immunohistochemical (IHC) analysis showed the following results: CK5/6 (+), P40 (+), P63 (3+), GATA-3 (−), WT1 (−), Ki-67 (+80%), Calretinin (weak+), MSH2 (+), MSH6 (+), MLH1 (+), PMS2 (+), D2-40 (−), S-100 (−), INSM1 (−), chromogranin A (CgA) (−), synaptophysin (Syn) (−), CD56 (−), INI 1 (+), BRG1 (shown in Fig. 3). Based on the patient's medical history, relevant imaging examinations, postoperative pathology, and IHC results, the final diagnosis for the patient was primary SCC of the small intestine.

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Therapeutic Intervention

The patient underwent radical surgical intervention, which included the resection of the jejunal tumor and associated lymph nodes. The surgical procedure also involved end-to-end anastomosis of the jejunum and descending colon. Postoperative care focused on infection prevention, fluid rehydration, and monitoring for complications.

The surgery was performed under general anesthesia. The resected jejunal mass measured approximately 10 × 8 cm, with adjacent gel-like deposits removed to minimize residual disease (shown in Fig. 4). Drainage tubes were inserted into the left abdominal and right pelvic cavities for postoperative fluid management. Post-surgery, the patient was closely monitored, and recommendations were made for adjuvant chemotherapy and immunotherapy based on multidisciplinary consultation.

One month after surgery, the patient visited our oncology department and completed one cycle of chemotherapy (nab-paclitaxel and cisplatin) following the esophageal SCC regimen. Due to personal reasons, the patient did not continue with further chemotherapy. Regular outpatient follow-ups were conducted, including tumor marker tests and abdominal CT/MRI scans with contrast, with no signs of recurrence.

Follow-Up and Outcomes

The surgery was successful. Based on the postoperative pathological findings, the oncology department consultation was enhanced. According to expert opinions, it is recommended to undergo combined chemotherapy and immunotherapy. The patient was discharged without complications and subsequently underwent multidisciplinary consultation at Beijing Cancer Hospital. Given the lack of significant association between intestinal tumors and lung malignancies, it is advisable to optimize the PD-L1 examination and manage SCC based on the adjuvant chemotherapy plan. The patient's latest PD-L1 test results revealed PD-L1 expression (SP263) at a rate of 50–60%, with no EGFR mutations detected. Based on immunohistochemistry results, the patient completed one cycle of chemotherapy combined with immunotherapy following the esophageal SCC regimen. Due to personal reasons, the patient did not continue with further chemotherapy. Regular outpatient follow-ups were conducted; as of August 23, 2024, the patient has been followed up for 15 months postoperatively, with no signs of recurrence on tumor marker tests and abdominal CT/MRI scans with contrast.

Discussion

The small intestine, which constitutes approximately 75% of the total length of the gastrointestinal tract and over 90% of its mucosal surface, is the longest segment of the digestive system [4]. Despite its anatomical dominance, malignant tumors of the small intestine are rare, with an estimated global incidence of less than 1 per 100,000 population [7]. This rarity is attributed to its unique anatomical and physiological features, including rapid peristalsis, which limits mucosal exposure to carcinogens, and its weakly alkaline pH, which reduces the conversion of pathogenic substances into carcinogenic compounds. Additionally, the enzymatic activity of small intestinal mucosal cells neutralizes exogenous carcinogens, while its abundant lymphoid tissue provides an immune barrier, further mitigating the risk of malignancy [8].

The duodenum is the most commonly affected site for small intestinal malignancies, accounting for over half of all cases. However, primary tumors in the jejunum and ileum are exceedingly rare, and SCC of the small intestine, whether primary or metastatic, is an exceptionally uncommon malignancy [9, 10].

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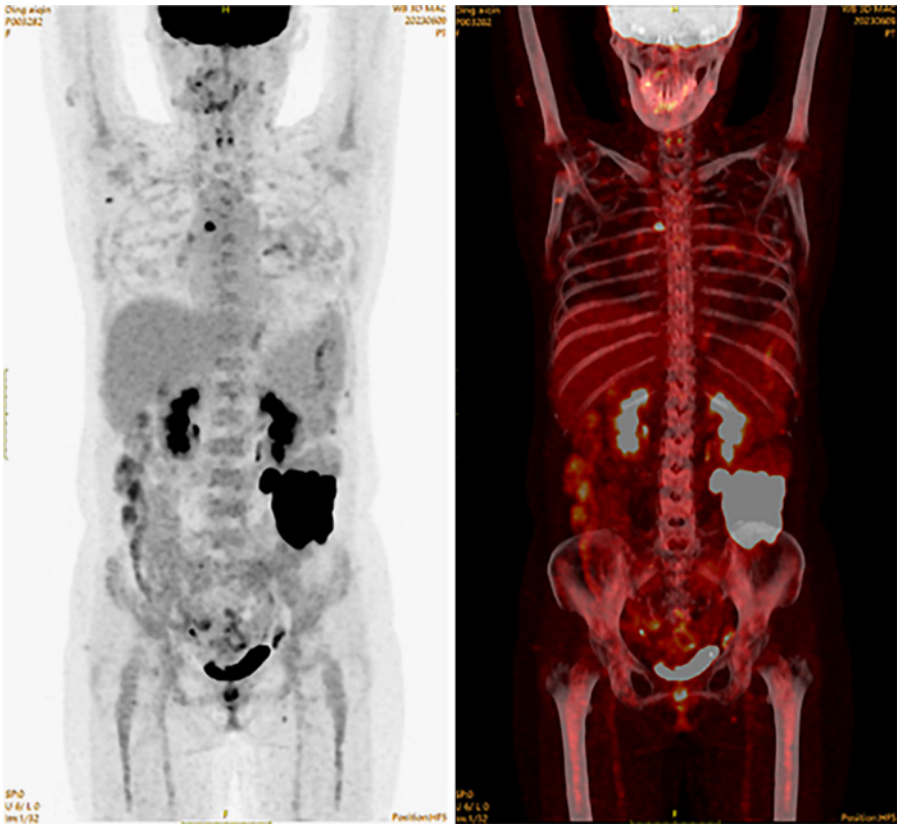

Fig. 2. PET/CT illustrated that the lesion surrounding the intestinal wall had highly active FDG uptake with an SUV max of up to 40.7. There were multiple enlarged lymph nodes of varying sizes around the lesion, increased FDG metabolism, and SUV max was 35.3, which was considered the possibility of lymph node metastasis. Multiple small lymph nodes in the left pelvic wall, increased FDG metabolism, SUV max was 6.7, not excluding the possibility of metastasis.

Metastatic SCC in the small intestine is more common than primary cases. Typical origins of metastatic SCC include anatomical sites covered by squamous epithelium, such as the skin, oral cavity, esophagus, and cervix [11, 12]. The small intestine can also serve as a site of metastasis for cancers such as pulmonary SCC, mandibular gingival carcinoma, esophageal carcinoma, and cervical carcinoma. Interestingly, squamous metaplasia in organs lacking squamous epithelium, such as the bronchi or gallbladder, may give rise to SCC [13].

The diagnosis of primary SCC of the small intestine is particularly challenging. While advanced imaging techniques such as multidetector spiral CT provide high-resolution visualization of tumors, adjacent organs, and lymph nodes, these modalities are often insufficient to distinguish between primary and metastatic disease [4]. PET-CT is a valuable diagnostic tool for evaluating FDG uptake patterns and can aid in determining tumor origin [14]. Histopathological evaluation, including IHC staining, is essential. Well-differentiated SCC is characterized by large keratinizing squamous cells, while poorly differentiated SCC predominantly features basal-type cells. Immunohistochemistry in this case revealed CK5/6, P40, and P63 positivity, with GATA-3 negativity, supporting the diagnosis of SCC. Importantly, metastasis was excluded based on the absence of primary SCC at other common anatomical sites, the involvement of mucosa rather than submucosa, and the lack of abnormal FDG uptake in other areas on PET-CT.

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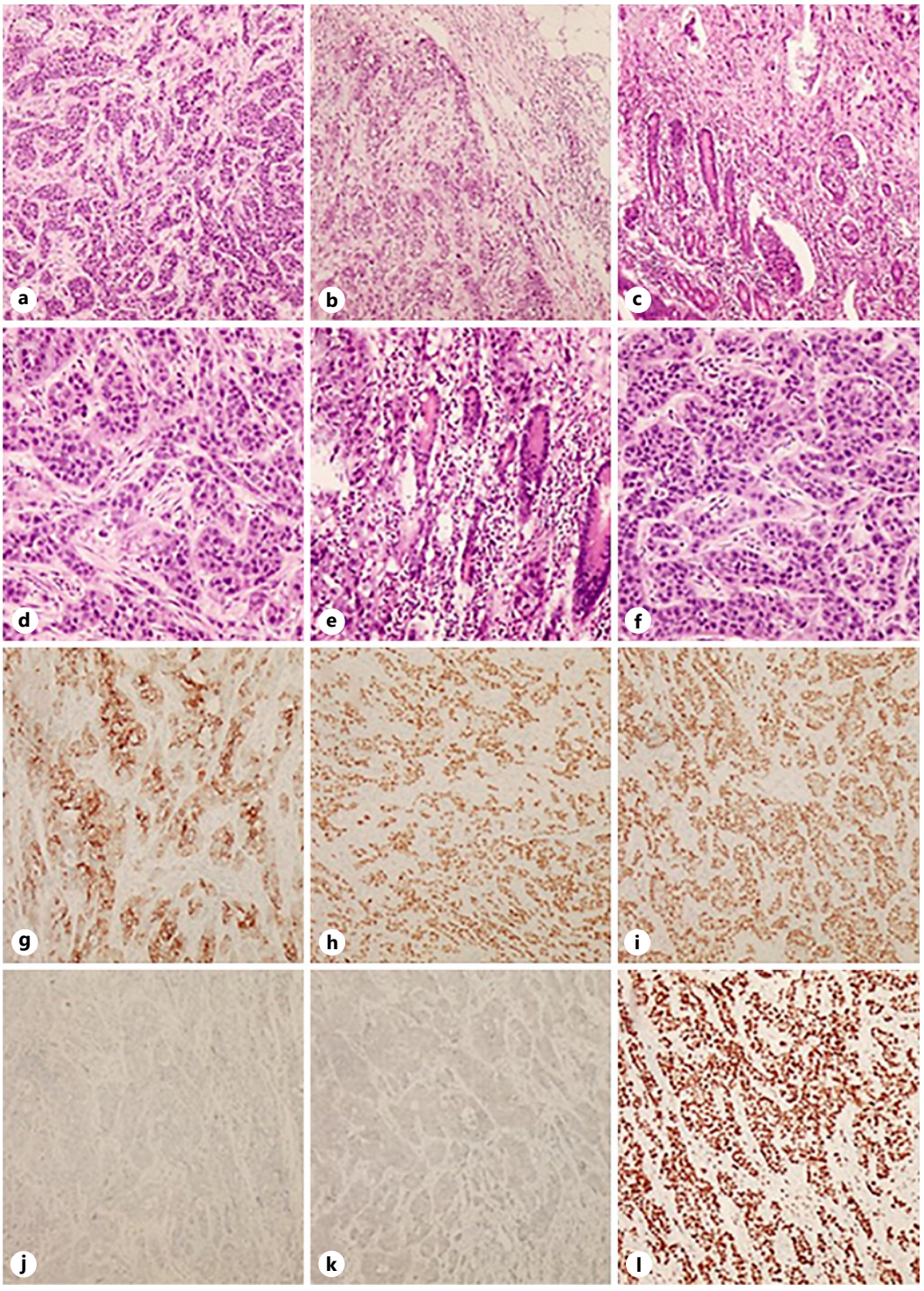

Fig. 3. Pathological and IHC findings. **a**, **b**, **c**, **e**, **f** Pathological findings from surgical specimen. **a** The lesion showed a serosal penetration. **b** Tumor cells are poorly differentiated. **c** Showing the boundary between normal mucosal cells and squamous cells. **h**, **i** Immunochemistry demonstrated that the staining for cytokeratin-5/6 and antioncogene P40 was positive. **g**, **k** Immunochemistry demonstrated that the staining for GATA-3 and INSM1 was negative. **d** Squamous tumor cells infiltrating into the muscular layer of the intestine. **j** Negative staining for GATA-3, ruling out breast or urothelial origin. **l** Ki-67 staining shows a high proliferation index, with approximately 80% of tumor cells positive.

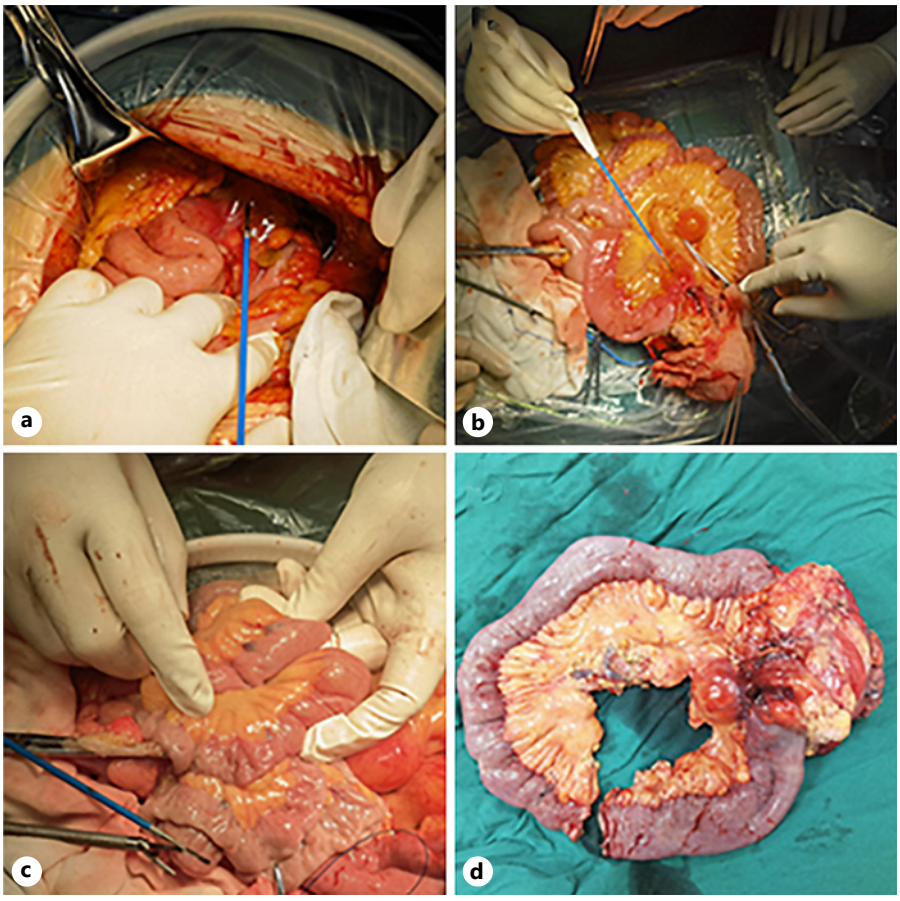

Fig. 4. a A jelly-like yellow substance of varying sizes can be seen around the mass. The mass penetrated the serosal layer and invaded part of the descending colon. **b** The mesangium showed masses of different sizes. **c** Multiple dark red nodules of varying sizes are seen in the rest of the small intestine. **d** Massive mass approximately 80 cm from the Treitz ligament.

Recent literature indicates that the 5-year survival rates for small intestinal malignancies, including SCC, range from 21% to 42%, with the prognosis further deteriorating in cases where regional lymph node involvement or distant metastases are present [15]. These survival rates are consistent with the generally poor prognosis associated with advanced-stage SCC of the gastrointestinal tract. Radical resection combined with regional lymph node dissection remains the mainstay of treatment for primary small intestinal malignancies, with the goal of improving survival outcomes [16, 17]. In cases where radical resection is not feasible, palliative surgery may reduce tumor burden and prevent complications such as obstruction or perforation, thereby enhancing the patient's quality of life. Due to the rarity of small intestinal SCC, MDT can provide a comprehensive and integrated treatment plan by combining the expertise of multiple disciplines. Considering the biological characteristics of SCC, there is a risk of obstruction or perforation, so radical surgery is recommended. After obtaining complete pathology, further tests such as EGFR, Her-2, PD-L1, and mismatch repair protein should be performed to guide subsequent adjuvant chemotherapy or immunotherapy. Although chemotherapy (e.g., taxanes and platinum) combined with immunotherapy is commonly used for SCCs of the esophagus or lung, there is currently no robust evidence to support its efficacy in primary SCC of the small intestine [7]. Further research is warranted to

establish effective postoperative treatment protocols and optimize therapeutic strategies for this rare malignancy.

This case report highlights the clinical and diagnostic challenges of primary SCC of the small intestine, an exceptionally rare malignancy. The strength of this report lies in its comprehensive diagnostic approach, which combined advanced imaging, histopathology, and immunohistochemistry to establish the diagnosis. The use of PET-CT to exclude metastasis and confirm the primary nature of the tumor is particularly noteworthy. However, this case report is limited by its focus on a single patient, which precludes broader generalizability. Additionally, the absence of follow-up data on adjuvant therapies limits the ability to evaluate long-term outcomes.

Conclusion

Primary SCC of the small intestine is a rare and aggressive malignancy with significant diagnostic and therapeutic challenges. Early diagnosis, radical surgical resection, and careful exclusion of metastatic disease are critical for improving patient outcomes. This case underscores the importance of collaborative care and highlights the need for further research to advance our understanding and management of this rare entity.

Patient Perspective

The patient expressed gratitude for the timely diagnosis and comprehensive care provided by the medical team. She noted that the persistent abdominal pain she experienced was initially concerning but was relieved to learn that surgical intervention was successful in removing the tumor. Despite the complexity of the procedure and the uncertainty surrounding her condition, she felt reassured by the clear communication and collaborative approach taken by her doctors.

While the prospect of undergoing additional treatment, such as chemotherapy and immunotherapy, was daunting, the patient acknowledged the importance of these recommendations for her long-term prognosis. She appreciated the multidisciplinary discussions and the explanation of her test results, which helped her understand the rationale behind the treatment plan. The patient remains committed to attending follow-up appointments and is optimistic about her recovery and future health.

Acknowledgments

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Statement of Ethics

Ethical approval to report this case (or case series) was obtained from the Medical Ethics Committee of Weihai Municipal Hospital (Approval No. 2023090). Written informed consent was obtained from the patient for publication of this case report and any accompanying images.

Conflict of Interest Statement

The authors have no conflicts of interest to declare.

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Author Contributions

Guangxing Li contributed to the conception and design of the study, performed the literature review, and drafted the initial manuscript. Chuanji Han conducted the patient diagnosis and managed the clinical treatment, including surgery. Jin Cao and Yongcun Zhu were responsible for data acquisition and analysis. Shengbo Sun contributed to final approval of the manuscript, overall supervision. All authors contributed to the interpretation of the data, provided critical revisions to the manuscript for important intellectual content, and approved the final version to be submitted.

Data Availability Statement

The datasets generated and analyzed during this study are available from the corresponding author upon reasonable request. Due to ethical and legal considerations, individual patient data are not publicly accessible to ensure confidentiality and comply with privacy regulations. Access to the data may be granted upon submission of a formal request and approval by the Medical Ethics Committee of Weihai Municipal Hospital. The CARE Checklist has been completed by the authors for this case report, attached as online supplementary material (for all online suppl. material, see https://doi.org/10.1159/000545101).

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