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CASE REPORT Open Access

Low-grade mucinous neoplasm originating from intestinal duplication: a case report and review of the literature

Huihui Yin[1], Jie Yu[1] and Yunzhao Chen[1,2,3*]

Abstract

Background Low-grade mucinous neoplasms typically originate from the appendix and are characterized by a lining of low-grade mucus-secreting columnar epithelial cells and smooth muscle. However, atypical origins can occur, as demonstrated in this case report.

Case presentation We present a case involving a 33-year-old male who, upon physical examination, was found to have an abdominal mass. A computed tomography (CT) scan revealed a cystic mass located between the pancreatic tail and the adjacent bowel duct, with significant enhancement of the cyst wall observed on contrast-enhanced imaging. The patient subsequently underwent laparoscopic surgical resection of the mass, and the resected specimen was sent for pathological evaluation. The pathology results were consistent with the histological morphology and immunohistochemical characteristics of low-grade mucinous tumors arising from intestinal duplication. Three and a half years post-resection, the patient returned for a follow-up examination, during which abdominal CT and blood tumor markers indicated no signs of tumor recurrence.

Conclusions While low-grade mucinous tumors predominantly originate from the appendix, this case illustrates an unusual occurrence of such neoplasms arising from intestinal duplication. This report aims to enhance clinical awareness of low-grade mucinous tumors originating from intestinal duplication, thereby improving the rates of preoperative diagnosis and reducing instances of misdiagnosis.

Keywords Intestinal duplication, Low-grade mucinous tumors, Retroperitoneal, Pathological features

Introduction

Intestinal duplication is a rare congenital abnormality that can occur anywhere along the gastrointestinal tract, from the mouth to the anus, with the ileum being the most common site. This condition typically becomes evident in early childhood, with approximately 65.8% of cases identified before the age of 2, while occurrences in adults are rare [1]. The clinical presentations of intestinal duplication vary depending on the type and location of the duplication. In neonates and infants, the most common symptoms include vomiting and abdominal distension; however, many duplications remain asymptomatic

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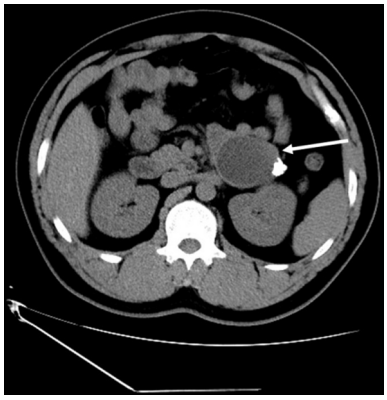

Fig. 1 Abdominal CT (flat scan) demonstrated a low-density shadow with well-defined boundaries and calcifications at the interface between the tail of the pancreas and the adjacent intestinal loop (white arrow)

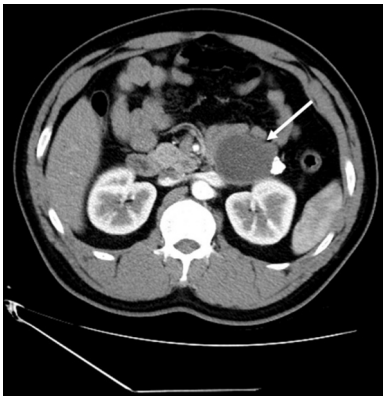

Fig. 2 Abdominal CT (enhanced) revealed partial enhancement of the cyst wall, indicating a cystic lesion with calcification in the left upper abdominal cavity (white arrow)

until complications arise, which may delay diagnosis until adulthood. Patients may experience chronic or acute abdominal pain, constipation, abdominal distension, or notice a palpable mass. The diagnostic challenges associated with intestinal duplication are exacerbated by its low incidence and the presence of nonspecific symptoms, often leading to confusion with other intestinal disorders. Notably, cases of pseudomyxoma peritonei originating from intestinal duplications have been reported [2–4]. While low-grade mucinous tumors are recognized as precancerous lesions that can lead to pseudomyxoma peritonei in the appendix, there is a scarcity of literature documenting the occurrence of low-grade mucinous tumors within intestinal duplications. This case report highlights a rare instance of low-grade mucinous tumors developing within an intestinal duplication in a 33-year-old male.

Case presentation

Chief complaints

The case involves a 33-year-old male patient who was admitted to the hospital following the discovery of a pancreatic mass during a physical examination conducted four days prior.

Laboratory examinations

Laboratory tests indicated that biochemical and tumor markers were within normal limits.

Imaging examinations

Abdominal CT (Fig. 1) revealed a low-density shadow with well-defined borders and calcifications at the interface between the tail of the pancreas and the adjacent intestinal loop. The cystic component within the lesion exhibited a CT value of approximately 13 HU and measured about 5.6 × 4.5 cm. Enhancement of the lesion demonstrated partial enhancement of the cyst wall, suggesting a cystic lesion with calcification located in the left upper abdominal cavity (Fig. 2).

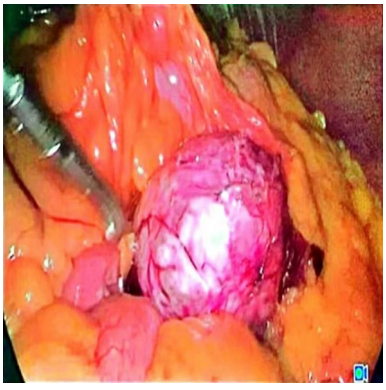

Fig. 3 Intraoperative photographs depicting a cystic mass

Final pathological diagnosis

The clinical diagnosis was a retroperitoneal mass, and the patient subsequently underwent laparoscopic resection of the mass (Fig. 3). Intraoperatively, it was observed that the mass was isolated and not connected to the intestine. Additionally, the liver, gallbladder, pancreas, spleen, small intestine, colon, and appendix exhibited no significant abnormalities. Pathological examination revealed a cyst measuring 7.0 × 5.0 × 3.5 cm, with a smooth wall thickness ranging from 0.1 to 0.6 cm, and localized hardness. The cyst contained a yellow-green viscous, mucus-like substance (Fig. 4).

Microscopically, the neoplastic cyst wall consisted of low-grade mucinous columnar epithelial cells lined with smooth muscle (Figs. 5, 6 and 7), resembling the intestinal wall but lacking mucosa and submucosa. The immunophenotype exhibited intestinal markers, including CK20 (Fig. 8), CDX-2 (Fig. 9), MUC2 (Fig. 10), while Villin, MUC5ac, and CK7 were partially expressed, indicating an origin from the gastrointestinal system. Based on

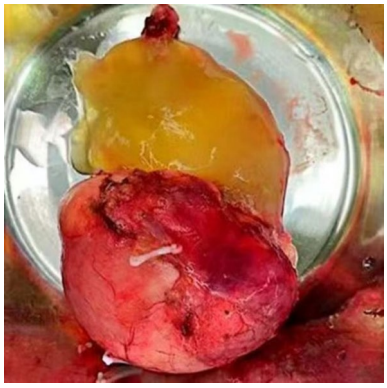

Fig. 4 A yellow-green, jelly-like mass was observed within the cystic structure

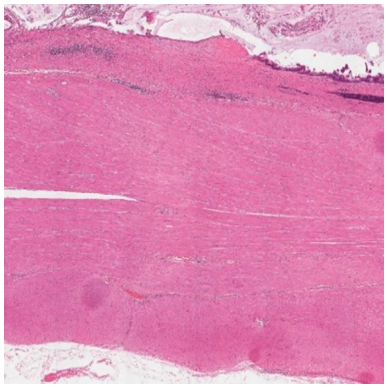

Fig. 5 Hematoxylin-eosin (HE) staining at 20X magnification displayed a low-power view of the cyst wall, which revealed an epithelial lining composed of double-layered smooth muscle containing mucus, originating from intestinal duplication

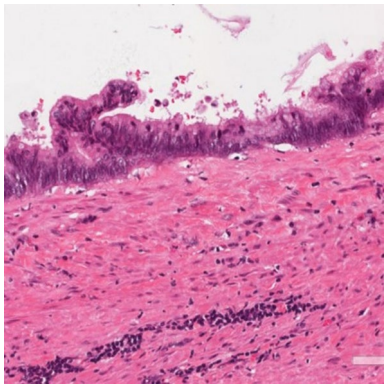

Fig. 6 Hematoxylin-eosin (HE) staining at 200X magnification illustrated pseudostratified low-grade mucinous epithelial cells lining double-layered smooth muscle, also originating from intestinal duplication

the characteristic jelly belly appearance, along with the microscopic morphology and immunophenotype, and after ruling out any abnormalities in the appendix and mucus leakage into the peritoneal cavity, the final diagnosis was determined to be a low-grade mucinous tumor originating from intestinal duplication, accompanied by local secondary dystrophic calcification and ossification.

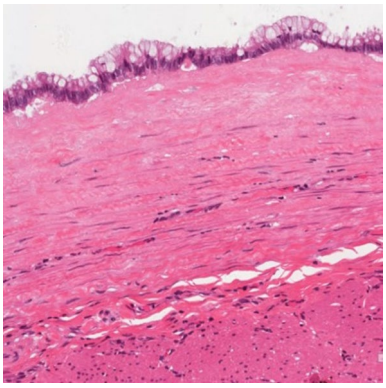

Fig. 7 Hematoxylin-eosin (HE) staining at 200X magnification showed a single layer of low-grade mucinous epithelial cells lining double-layered smooth muscle, again emanating from intestinal duplication

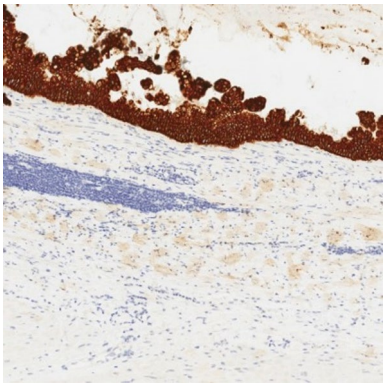

Fig. 8 Immunohistochemical staining demonstrated positivity for CK-20 at 100X magnification

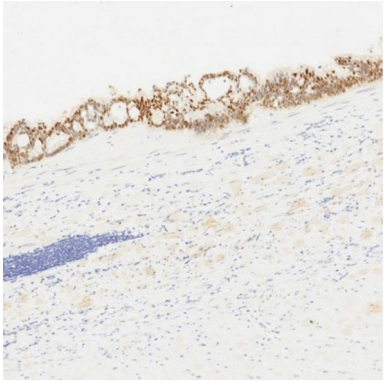

Fig. 9 Immunohistochemical staining indicated positivity for CDX-2 at 100X magnification

Treatment and follow-up

The patient underwent laparoscopic retroperitoneal tumor resection and was discharged 10 days after surgery without other treatment. He remained in good

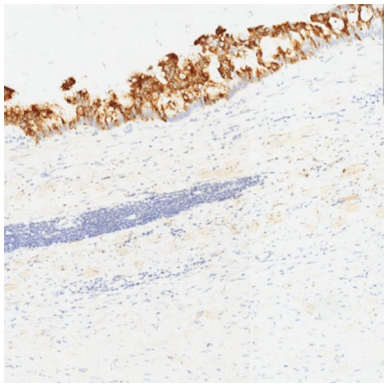

Fig. 10 Immunohistochemical staining revealed positivity for MUC-2 at 100X magnification

health during the 3.5 years follow-up by the inspection of abdominal CT and blood tumor markers.

Discussion and conclusions

Epidemiology

Digestive duplications are infrequent congenital anomalies, particularly in the first two years of life, and are rarely observed in adults [5]. Intestinal duplications can occur in any part of the digestive tract or adjacent organs, with the ileum being the most common site at 31.5%, followed by the ileocecal valve at 30.2%, the jejunum at 8%, the colon at 6-7%, and the rectum at 5% [1]. Additionally, intestinal duplications located in unexpected areas have only been described in scattered case reports, such as in the skin [6], the tongue [7], the liver [8]. In the early stages, the condition is often asymptomatic; however, patients may present with complications such as volvulus and intussusception, which can go undetected. More than 80% of cases present before the age of 2 years with acute abdominal symptoms or bowel obstruction, a presentation that is rarely seen in adults [1]. Due to the nonspecific nature of the symptoms, initial misdiagnoses of other conditions are common. In this study, the patient was a young adult who was incidentally found to have an abdominal mass during a physical examination.

Mechanisms

The mechanism of intestinal duplication remains controversial, with several hypotheses proposed; however, the etiology of this anomaly is largely unknown [9]. A compelling explanation, as articulated by Smith, involves the dorsal protrusion of the yolk sac due to herniation or adhesion to the ectoderm during the same phase of presomite development, leading to the formation of a 'dorsal remnants' structure. This structure can be categorized into several types: congenital dorsal enteric fistula, congenital dorsal enteric sinus, congenital dorsal enteric cyst, and congenital dorsal enteric diverticulum [10].

Differential diagnosis

Colorectal duplication can be categorized into two types: cystic and tubular duplication. Cystic duplication is the more prevalent form, accounting for approximately 86% of cases, while tubular duplication comprises only 14% of all instances [1]. Unlike cystic duplication, tubular colonic duplication establishes direct communication with the native tract [11]. Typically, cystic duplication originates solely within the abdominal cavity, as illustrated in the presented case. Cystic intestinal duplication is mainly identified with the following diseases. Firstly, intestinal diverticula establish direct communication with the native tract. Additionally, intestinal adhesions occur when adhered intestinal tubes maintain distinct anatomical structures and are disconnected from one another. Lastly, teratomas, characterized by the presence of epithelial and smooth muscle components within the digestive tract, should also be considered as potential diagnoses. Furthermore, the presence of epidermis, skin appendages, fat, cartilage, and other components can aid in the diagnosis of teratomas. As indicated in the literature, intestinal duplication may contain ectopic tissue [12], such as thyroid, pancreas, gastric mucosa, lungs, and cartilage, which necessitates careful differentiation from teratomas due to the complex and diverse pathological components involved.

Management

It is postulated that duplications of the large bowel possess malignant potential, associated with rare diseases and a relatively low incidence. Only a few cases of neoplasms arising from intestinal duplication have been reported, including tubular adenoma [13], pseudomyxoma peritonei (PMP) [2–4], adenocarcinoma [14–16], squamous carcinoma [17], and carcinoid tumors [18]. Low-grade mucinous neoplasms originating from intestinal duplication represent an extremely rare form of tumor disease. These duplications are lined by intestinal epithelial cells, and the occurrence of malignant degeneration, such as dysplasia or cancerization, is plausible, similar to the colonic mucosa. Given the low-grade mucinous neoplasm observed in our case and the documented instances of tubular adenoma [13], and adenocarcinoma arising from colonic duplication [14, 15], a comprehensive histological examination is essential to exclude abnormal epithelial lesions that may be obscured by the intestinal duplication and to ensure appropriate disease management. In our case, the entire tumor was sampled for histological observation, revealing that low-grade mucinous columnar epithelial cells lined smooth muscle and expressed intestinal markers CK20 (Fig. 8), CDX-2 (Fig. 9), and MUC2 (Fig. 10), indicating an intestinal origin.

Mucous tumors occurring in the appendix are classified into low-grade and high-grade mucinous tumors based on their histological structure and cellular atypia. Pseudomyxoma peritonei (PMP) develops when the tumor penetrates the cyst wall, resulting in the production of a substantial amount of mucus. The curative treatment for PMP involves complete cytoreductive surgery (CRS) combined with hyperthermic intraperitoneal chemotherapy (HIPEC), which includes 10 mg of mitomycin and 40 mg of cisplatin, along with concurrent intravenous chemotherapy using 5-FU (1 g) during a 90-minute thermal cycle. This approach has shown promising results for extra-appendiceal PMP [19]. Chua TC [20]reported on a cohort of 2,298 cases of pseudomyxoma peritonei originating from appendiceal mucinous neoplasms treated with CRS and HIPEC, noting a median survival rate of 196 months (16.3 years) and a median progression-free survival rate of 98 months (8.2 years), with 10- and 15-year survival rates of 63% and 59%, respectively. Additionally, a patient with pseudomyxoma peritonei arising from intestinal duplication who underwent CRS and HIPEC exhibited no tumor recurrence during follow-up visits five years after the initial operation [2]. This patient maintained good health throughout a 3.5-year follow-up period post-surgery. These findings underscore the importance of accurate diagnosis in determining the presence of mucus extravasation, which is crucial for understanding the disease and selecting appropriate treatment strategies.

The limitations of this report include a small sample size and restricted generalizability of the results, making it challenging to extrapolate findings to the broader population. Consequently, these limitations may lead to misinterpretations regarding specific treatments or disease characteristics, as individual variations can influence the applicability of the findings. Additionally, the detailed nature of case reports, which typically focus on a single patient or a small cohort, complicates the aggregation and integration of data across multiple reports. This inconsistency in the data hinders researchers' ability to conduct systematic reviews or meta-analyses, thereby limiting a comprehensive understanding of the topic. Due to the limited number of cases involving tumors originating from intestinal duplication, assessing their overall survival prognosis remains challenging. Taha reported that the RMH score [21]serves as a novel prognostic biomarker, showing promise as a readily available tool across various cancer types and clinical settings, thereby ensuring its optimal application in clinical practice and decision-making. Consequently, we aim to collaborate among hospitals to collect additional cases of low-grade mucinous tumors arising from intestinal duplication to evaluate survival outcomes and prognosis.

Intestinal duplication in adults is infrequent and poses diagnostic challenges without surgical intervention. Radiologic evaluations that reveal a tubular or cystic structure filled with large, jelly-like materials-whether communicating with the normal intestine or not-should prompt consideration of intestinal duplication, particularly among radiologists and surgeons prior to surgical procedures. Following surgery, meticulous gross and histological examinations are essential to identify the potential presence of associated neoplasms, such as low-grade mucinous tumors, tubular adenoma [13], pseudomyxoma peritonei (PMP) [2–4], adenocarcinoma [14–16], squamous carcinoma [17], and carcinoid tumors [18] arising within the intestinal duplication.

Abbreviations

CT	Computed tomography
CRS	Complete cytoreductive surgery
PMP	Pseudomyxoma peritonei
HE	Hematoxylin-eosin
CK	Cytokine
MUC-2	Mucin 2
HIPEC	Heated intraperitoneal chemotherapy

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Author contributions
Huihui Yin wrote the main manuscript text and Jie Yu prepared Figs. 1, 2, 3, 4, 5, 6, 7, 8, 9 and 10. All authors reviewed the manuscript and reviewing the case.

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Data availability
No datasets were generated or analysed during the current study.

Declarations

Ethics approval and consent to participate
Ethics approval and consent to participate Informed consent was obtained from the patient. For retrospective studies, ethical approval was waived by Institutional Review Board of Zhejiang Provincial People's Hospital.

Consent for publication
Consent for publication was obtained from the patient.

Competing interests
The authors declare no competing interests.

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