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CASE REPORT

Pseudomyxoma peritonei originating from intestinal duplication: A case report and review of the literature

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Abstract

BACKGROUND

Pseudomyxoma peritonei (PMP) is a rare mucinous neoplasm with a relatively low incidence of 1 to 2 per million individuals. It is typically characterized by a type of gelatinous ascites named "jelly belly". Most cases of PMP occur in association with ruptured primary mucinous tumors of the appendix (90%). Periodically, PMP can originate from mucinous carcinomas at other sites, including the colorectum, gallbladder, and pancreas. However, unusual origin can occur, as noted in this case report.

CASE SUMMARY

A 52-year-old woman had an unusual derivation of PMP from intestinal duplication. The patient complained of abdominal distension and increasing abdominal girth. Abdominal contrast-enhanced computed tomography showed a mass in the greater omentum located on the left side of the abdomen, likely to be a cystic mass of peritoneal origin. A PMP diagnosis was presumed based on the specific signs of the mass with flocculent and stripe-like echoes in ultrasound images. Ultrasound-guided percutaneous aspiration suggested a high likelihood of PMP. Once the PMP diagnosis was recognized, identification of the origin of the primary tumor was indicated. Thus, an exploratory laparoscopy was performed. In the absence of a primary tumor of appendix origin, the diagnosis of a low-grade mucinous neoplasm of intestinal duplication origin was finally confirmed by histopathology.

CONCLUSION

PMP is secondary to mucinous carcinomas of the appendix mostly. This case resulted from an unusual derivation from intestinal duplication.

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Core Tip: Pseudomyxoma peritonei (PMP) happens rarely. Most of the tumors that occur in PMP are not primary, but secondary to ruptured mucinous tumors of other organs. Existent medical literature shows that the appendix is the most common origin site. However, other origins such as intestinal duplication can occur. The present case report describes a 52-year-old patient with PMP that is derived from a primary intestinal duplication.

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INTRODUCTION

Pseudomyxoma peritonei (PMP) is an uncommon disease with a relatively low incidence of 1 to 2 per million individuals[1,2]. PMP is often misdiagnosed clinically due to the lack of specific clinical presentation[3].

Classically, most PMP tumors are not primary, but secondary to ruptured mucinous tumors of other organs, especially the appendix[4]. Occasionally, PMP arises from adenocarcinomas of other sites within the gastrointestinal tract[5,6]. Typically, this disorder is characterized by an abundant accumulation of mucinous ascites developing from mucin secretion by a primary tumour[7-9]. The primary tumour ruptures and tumor cells then spread to implant throughout the peritoneal cavity, which results in the typical "jelly belly" appearance. Considering the rarity of the primary lesion of intestinal duplication, we report the current case with PMP seen in our hospital. This is an extremely rare origin of tumor disease.

CASE PRESENTATION

Chief complaints

A 52-year-old woman presented with the symptoms of abdominal distension and increasing abdominal girth.

History of present illness

Because of decreased appetite, the patient was referred to our hospital for further evaluation.

History of past illness

The patient had a rheumatoid arthritis history for 18 years. She also had a diagnosis of superficial gastritis for 2 mo.

Personal and family history

There was no family history.

Physical examination

Physical examination revealed the patient had a distended abdomen with a hard and non-tender mass. The mass was approximately 15 cm in diameters with ill-defined margins. Shifting dullness could not be found.

Laboratory examinations

Blood examination was performed. Normal levels of the tumor markers carcinoembryonic antigen (CEA), carbohydrate antigen 12-5 (CA12-5), CA19-9, and CA724 were observed. However, an increased CA242 level (25.87 U/mL) was found. Other physical examination results were as follows: Body temperature 37.0 degrees, pulse 80 beats per minute, respiratory rate 16 breaths per minute, blood pressure 120/70 mmHg, and abdominal gurgling sounds approximately three times per minute. No abnormalities were seen on the ultrasonic cardiogram or gastroscopy. The patient had a relatively unremarkable previous medical history apart from rheumatoid arthritis. She denied any other relevant, specific past medical or family medical history. The urinary and bowel elimination functions were reported to be good. She denied weight loss.

Imaging examinations

Contrast-enhanced abdominal computed tomography (CECT) (Figure 1) showed a mass in the greater omentum located on the left side of the abdomen, likely to be a cystic mass of peritoneal origin.

Ultrasound indicated that the mass in the left upper and middle abdomen was flocculent, with an internal stripe-like echo (Figure 2A). The mass could not be deformed by the probe pressing technique[10,11]. The space occupying lesion looked like a mucinous mass, on the basis of the flocculent and stripe-like echoes observed in the scan. Subsequently, ultrasound-guided percutaneous aspiration of the cystic lesion revealed hallmark yellow gelatinous material characteristic (Figure 2B). These findings, combined with the clinical presentation, suggested a clinical diagnosis of PMP.

Surgical operation

Once the PMP diagnosis was recognized, identification of the origin was indicated. Thus, an exploratory laparoscopy was performed. It showed a mass with cystic characteristics and jelly-like content located inside the greater omental cavity. Considering the jelly in the greater omentum, the suspicion of PMP increased in possibility[12]. However, from macroscopic observation, the appendix had an elongated shape with no edema or tumor mass. Nonetheless, complete microscopic tissue examination of the appendix was needed. Considering that appendiceal origin was most likely, appendectomy was performed with the permission of the family. Frozen sections of the excised appendiceal tissue were immediately analyzed during the operation. Hematoxylin-eosin (HE) staining showed chronic appendicitis obliterans of the tissue (Figure 3).

Women with PMP often have mucinous tumors involving both the appendix and the ovary[13,14]. Hence, the ovaries were carefully inspected in the patient, which showed negative results. Exploratory laparotomy therefore continued, in the absence of primary tumors from appendix and ovary. The location of jelly in the omental cavity necessitated total omentum removal. During radical greater omentectomy, an extremely sticky mucoid material was observed proximal to the splenic flexure of colon, which was a helpful feature likely signaling the primary origin of the cystic tumor. Besides, it was consistent with the location of the lesion on preoperative CECT imaging. Therefore, the sticky mucoid material was separated. And a mucinous tumor was found located on the anterior lobe of mesocolon on the left part of the splenic flexure of the transverse colon (Figure 4). Omentectomy was performed as the preferred option under this set of conditions.

FINAL DIAGNOSIS

There was a subsequent finding of mucoid material in the anterior lobe of the transverse mesocolon. Immunohistochemistry identified intestinal duplication origin. Low-grade mucinous epithelial cells were lining in the capsule wall of the cystic mass in the focal area. Extensive fibrosis and calcifications were found in the cystic wall. The smooth muscle layer could also be seen at some sites (Figure 5).

As shown in Figure 6, immunohistochemical staining of the mucinous tumor lesion demonstrated negative expression of cytokeratin (CK)-7, but strongly positive expression of CK-20, Villin, CDX-2, and Mucin 2 (MUC-2). PMP typically originates from MUC-2 over-expression of goblet cells[15]. CDX-2 plays a crucial role in cell proliferation and differentiation[16]. The finding of CDX-2 positive expression indicated that the tumor originated from the gastrointestinal (alimentary) system[17].

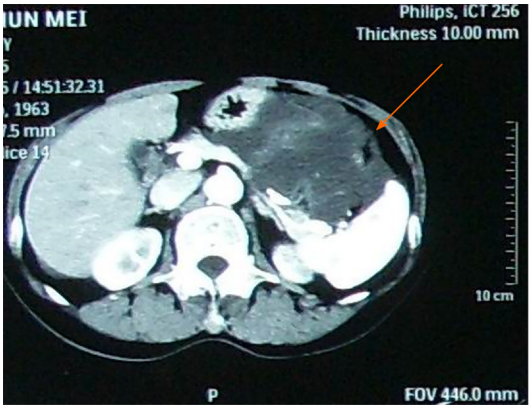

Figure 1 Abdominal contrast-enhanced computed tomography revealed a low density mass in the upper abdomen proximal to the spleen (arrow).

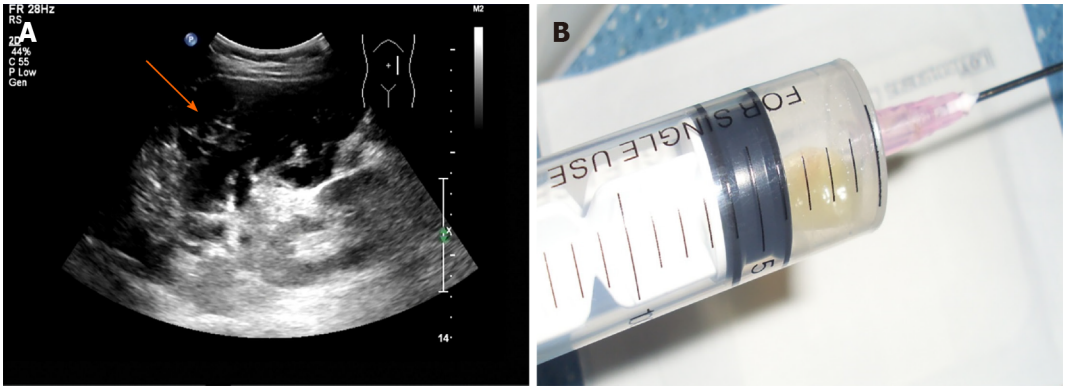

Figure 2 Ultrasound image and transabdominal ultrasound-guided percutaneous aspiration of the mass in the left upper abdomen. A: A large mass with flocculent and stripe-like echoes (arrow) was detected in the left middle and upper abdomen by ultrasound; B: Yellow gelatinous material was aspirated from the abdomen *via* transabdominal ultrasound-guided percutaneous aspiration.

Further pathology consultation with two other hospitals (Peking University Cancer Hospital and Peking Union Medical College Hospital) was performed to confirm the diagnosis. The two hospitals obtained the similar results that the presented case was PMP derived from intestinal duplication.

TREATMENT

Macroscopic tumor excision combined with heated intraperitoneal chemotherapy (HIPEC) has shown encouraging outcomes for extra-appendiceal PMP[2,18]. The patient was therefore treated with HIPEC, which consisted of 10 mg of mitomycin and 40 mg of cisplatin along with concurrent intravenous chemotherapy therapy of 5-FU (1 g). A 90-min thermal cycle was adopted.

OUTCOME AND FOLLOW-UP

The peritoneal cancer index[19] was estimated in the patient to assess the extent of PMP. The size of the lesion was scored: 0 = no tumor, 1 = tumor ≤ 0.5 cm, 2 = 0.5 cm < tumor ≤ 5.0 cm, and 3 = tumor > 5.0 cm. The cystic lesion was located behind the posterior wall of stomach, in the front of the pancreas, and on the inside of the spleen, which occupied regions of 3, 4, and 0. The scores of the three regions were all 3. The jelly like ascites in the uterus-rectum-fossa in region 6 was scored 1. Thus, the aggregative score of 13 abdominopelvic regions reached 10 in surgery. A complete cytoreduction was achieved after surgery. The degree of cytoreduction reached a grade of 0. Post-treatment CEA, CA12-5, CA19-9, CA724, and CA242 were all negative.

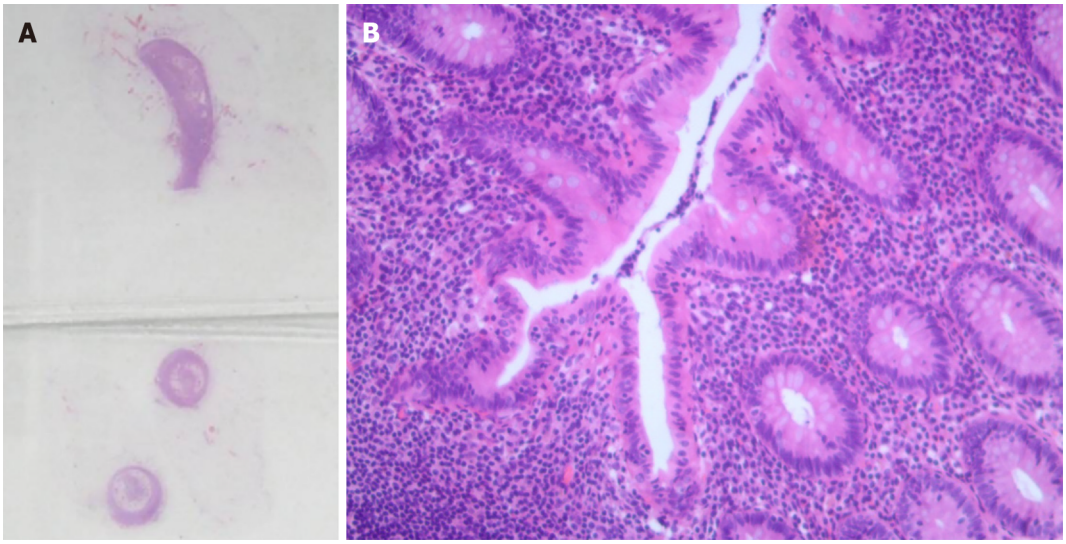

Figure 3 The obtained appendix specimen and its hematoxylin-eosin staining results. A: Gross pathology of appendix showed a length of 5.0 cm and width of 0.3-0.6 cm in diameter; B: Hematoxylin-eosin staining results of the specimen demonstrated appendicitis obliterans.

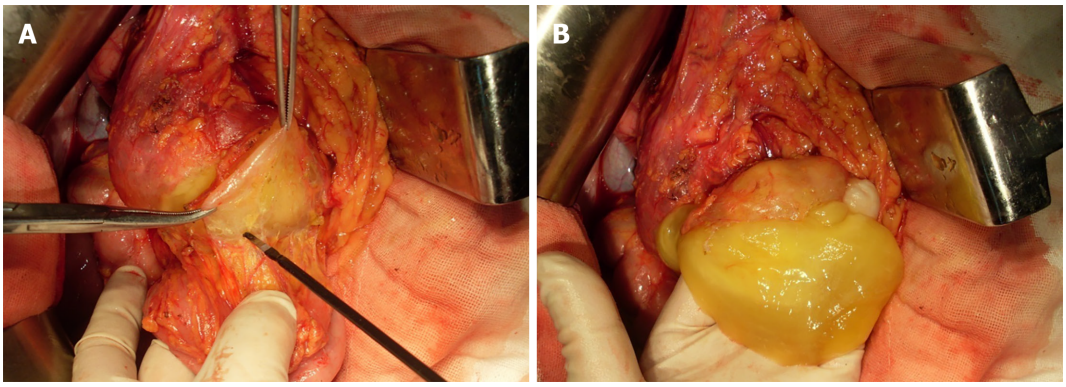

Figure 4 Intraoperative pictures. A: Characteristic cystic mass (arrow) presented in the anterior lobe of the transverse mesocolon in the left part of the splenic flexure; B: A yellow jelly-like mass existed inside.

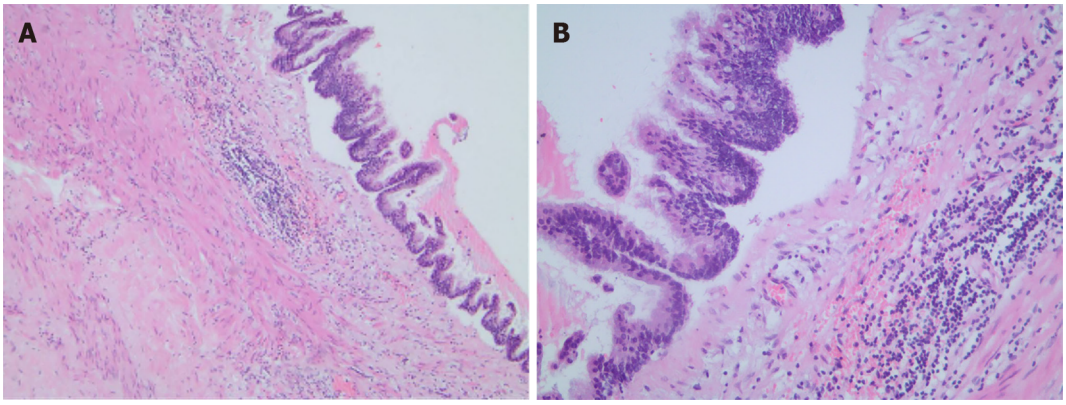

Figure 5 Hematoxylin-eosin staining of the specimen found in the splenic flexure of the colon revealed a cystic mass emanating from the intestinal duplication, with low-grade mucinous epithelial cells lining in the capsule wall. A: × 40; B: × 200.

Additionally, no obvious abnormalities were observed on repeat abdominal computed tomography (CT). The patient had no tumor recurrence in follow-up visits until May, 2020 (5 years after the initial operation).

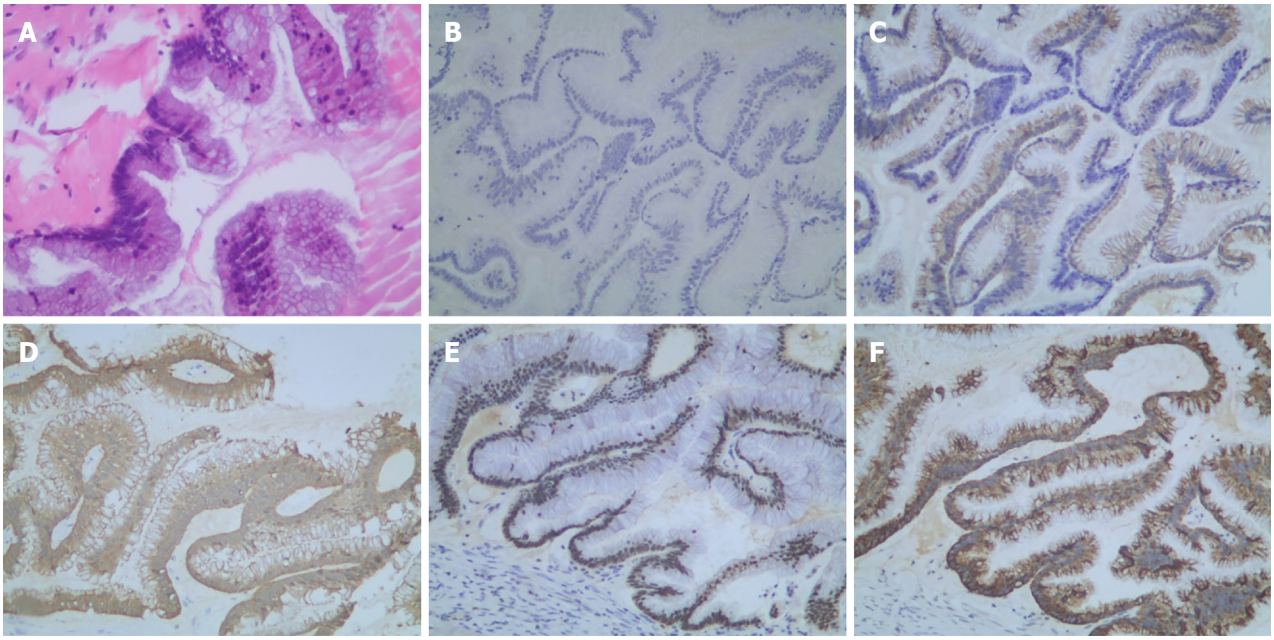

Figure 6 Histologic presentation of low-grade mucinous neoplasm. A: Hematoxylin-eosin staining of the primary tumor; B-F: Immunohistochemical staining found that the primary tumor was CK-7(–) (B), CK-20(+) (C), Villin(+) (D), CDX-2(+) (E), and MUC-2(+) (F).

DISCUSSION

The diagnosis of PMP, a rare clinical syndrome, is difficult[20,21]. Commonly, the presenting symptom is increasing abdominal girth. As symptoms are typically non-specific, an initial misdiagnosis of other conditions occurs frequently. Usually a suspected diagnosis may be made by ultrasonography. Ultrasonography, CT, and other examinations, followed by histopathologic verification of extensively sampled tumor, are the preferred ways to confirm a diagnosis of PMP[22]. The feature of flocculent and stripe-like echoes could be detected by an experienced observer[10,23], which is helpful for the diagnosis of PMP. Detection of yellow gelatinous material[24] *via* transabdominal ultrasound-guided percutaneous aspiration strengthens the probability of PMP diagnosis.

Once the PMP diagnosis is recognized, the source should be identified. The great majority of PMP cases are associated with the spread of a primary mucin-producing tumor of the appendix, accounting for approximately 90% of cases. According to the clinical experience of our center, more than 86% (904/1050) of the center's PMP cases originated from appendix. A primary PMP tumor can arise from elsewhere in the gastrointestinal tract as well. Since most PMP cases are due to appendiceal tumors, the appendiceal region should be closely inspected. Studies[25,26] have reported that it might be impossible to identify an appendiceal origin of PMP at surgery because the residual appendix may be small or fibrosed after rupture. Thus, it is preferred to perform an appendectomy. The appendix should be sent for serial sections for definitive histopathology examination before another primary site is considered[27]. The coexistence of ovarian and appendiceal mucinous tumors is commonly encountered[4]. Substantial research discusses long-held controversies regarding origin from either the appendix or the ovary in mucinous tumor cases of PMP in women. Several studies[28-31] have suggested that most cases of PMP in women are of intestinal origin with secondary ovarian involvement. The removal of the ovaries is routinely advised in patients with colonic origin of carcinomatosis and menopause, as there is a high chance of ovarian metastasis.

Since there was an abnormal omental mass indicated by preoperative CT in this case, careful inspection of the anatomic region of the peritoneal cavity where extremely sticky mucoid material occurred was required. Finally, a mucinous tumor in the anterior lobe of the transverse mesocolon was observed on exploratory laparoscopy, though peritoneal tumors were difficult to identify. The subsequent pathology test of the lesion revealed a low-grade mucinous adenocarcinoma, originating from intestinal duplication.

In terms of severity of the disease, PMP is classified into either low-grade or high-grade mucious adenocarcinomas. The distinction between low-grade and high-grade carcinomas is of prognostic significance. Patients with low-grade tumors generally have a good 5-year survival of 63%-86% comparatively, whereas high-grade tumors generally indicate a survival of only 28%-44%[2,30,31]. It is noteworthy that adult intestinal duplication is quite rare[32]. In the current case, the intestinal duplication was characterized by well-developed smooth muscle. Additionally, a low-grade mucinous epithelium and smooth muscular layers in the intestinal tumor focal area were present. Typically, intestinal duplication arises from the mesenteric border of the bowel[33]. But the abnormal changes in the mesentery or mesocolon could not be detected by ultrasound or CT due to the anatomical complexity of the region[34].

It is hypothesized that the case was caused by the metaplasia of mucous epithelial cells in duplication of the intestine. Mucinous tumor cells produced progressive amounts of mucinous materials and then penetrated through the intestinal wall, eventually spread to the peritoneal cavity in the form of gelatinous deposits. Increased abdominal girth then occurred, but the mechanism for this process needed further study. PMP tumors are mostly CK-20 positive and CK-7 negative. The positive expression of CDX-2 in this case indicated an origin from the gastrointestinal system [33-37]. High level of CA242 was also effective in diagnosis of gastrointestinal cancer [38].

CONCLUSION

In conclusion, PMP is a rare condition characterized by the deposition of mucinous material on peritoneal surfaces. Most of the tumors are not primary, but secondary to ruptured mucinous tumors of other organs[5]. The appendix is by far the most common primary site. It is noteworthy that intestinal duplication could also be the origin of PMP, which was also reported by Lemahieu *et al*[39] and Letarte *et al*[40].

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