Hong Kong J Radiol. 2013;16:224-6 | DOI: 10.12809/hkjr1312120

CASE REPORT

Atypical Presentation of Gardner's Syndrome

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ABSTRACT

Gardner's syndrome is a subtype of familial adenomatous polyposis. Gardner's syndrome is a rare disorder involving polyps in the colon and tumours in other parts of the body. We report on a 38-year-old woman who presented with acute abdomen and fever. Subsequent diagnostic evaluation revealed multiple intra-abdominal desmoid tumours and rectal polyposis, meeting the diagnostic criteria of Gardner's syndrome.

Key Words: *Fibromatosis, aggressive; Gardner syndrome*

中文摘要

Gardner綜合徵的非典型表現

林卓恆、楊芷珩、范維洲、馬嘉輝

Gardner綜合徵是家族性腺瘤性息肉病的一個亞型。Gardner綜合徵是一種罕見的疾病，病徵包括結腸息肉和其他身體部位的腫瘤。本文報告一名38歲女性病例，因急性腹痛和發熱就診。隨後的診斷性檢查顯示腹腔內多個硬纖維瘤及直腸息肉，合乎Gardner綜合徵的診斷標準。

INTRODUCTION

Gardner's syndrome is a subtype of familial adenomatous polyposis. The condition is an autosomal dominant form of polyposis, characterised by multiple intestinal polyps and soft tissue tumours. Gardner's syndrome is caused by mutation in the *APC* gene located in chromosome 5q21, of which 20% are de novo in nature.[1] We report on a 38-year-old woman with multiple intra-abdominal desmoid tumours and rectal polyposis, suggestive of Gardner's syndrome.

CASE REPORT

A 38-year-old woman presented with abdominal pain and fever in November 2010. She had a history of benign intestinal tumour and underwent an operation in China in 1998. On physical examination, her abdomen was distended. There was a 10-cm periumbilical mass, which was firm and tender. Computed tomography (CT) revealed a pneumoperitoneum, multiple intra-abdominal abscesses, and soft tissue masses at the retroperitoneal region and anterior abdominal wall (Figures 1 to 3). Urgent laparotomy and drainage was performed. The abdominal pain and fever disappeared completely and the patient was discharged.

After 3 months, the patient returned with abdominal

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Submitted: 16 Jul 2012; Accepted: 18 Sep 2012.

pain and fever. CT again showed multiple abscesses. Ultrasound- / CT-guided drainage was performed. Biopsies of the abdominal wall and retroperitoneal lesions were taken. Histology and immunohistochemical stains demonstrated positive nuclear staining for beta catenin, and negative staining for CD34, calponin, smooth muscle actin, and CD117. The features were compatible with mesenteric fibromatosis-desmoid tumour. Follow-up CT revealed resolved abscesses with residual soft tissue masses, right hydronephrosis, and 30 to 40 enhancing rectal polypoid lesions, which were later confirmed by colonoscopy (Figure 4). The clinical signs, imaging findings, and pathology results were suggestive of Gardner's syndrome.

DISCUSSION

Gardner's syndrome was first reported by Gardner and Smith in 1958.[2,3] Extra-intestinal manifestations

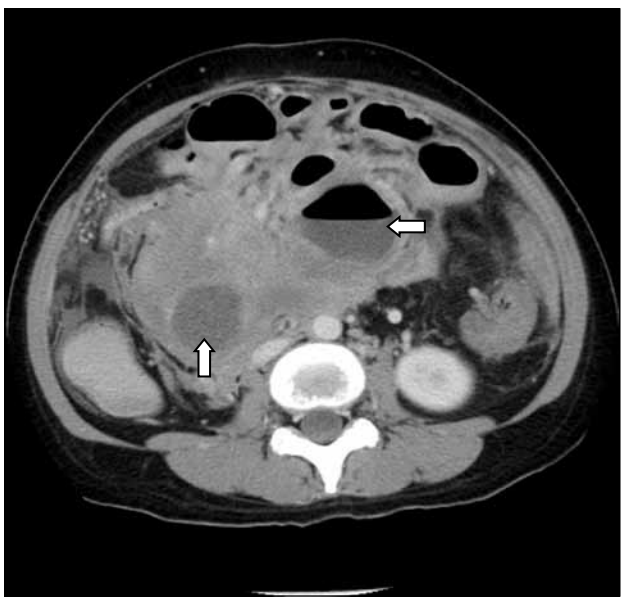

Figure 1. A computed tomography image shows multiple intra-abdominal abscesses (arrows).

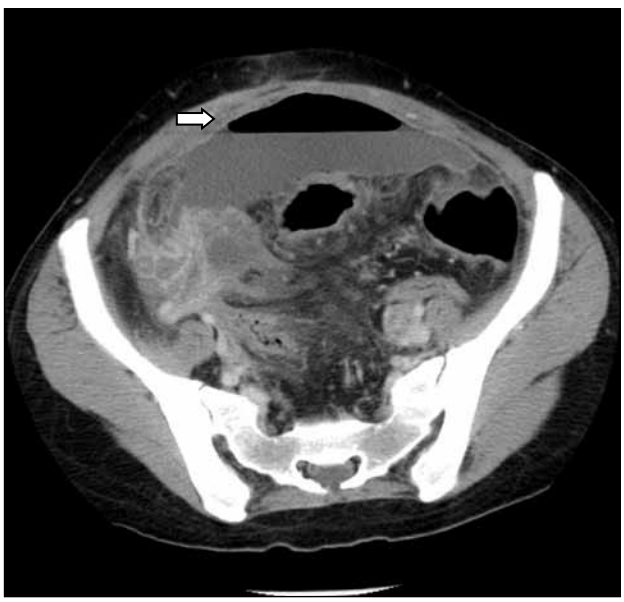

Figure 3. A computed tomography image shows gross pneumoperitoneum (arrow).

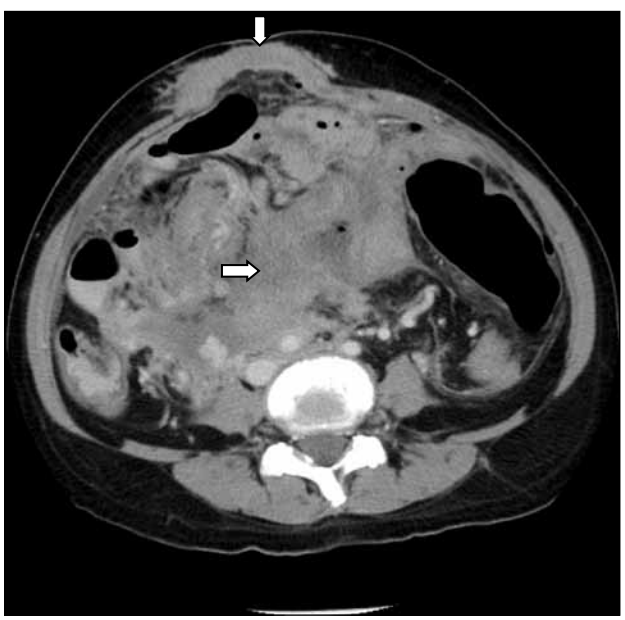

Figure 2. A computed tomography image shows mesenteric and abdominal wall soft tissue mass (arrows).

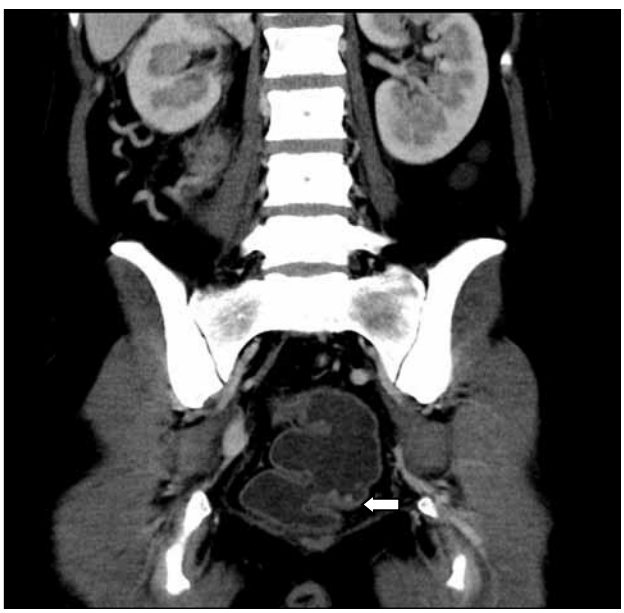

Figure 4. A computed tomography image shows multiple enhancing polypoid lesions (arrow).

usually occur earlier than in intestinal polyposis. If polyps are present, the colon is usually almost completely affected. Multiple colonic polyps start to appear during puberty and increase in number during the third and fourth decades of life, probably in response to oestrogen. The polyps are adenomatous and will undergo malignant transformation. Therefore, prophylactic total colectomy is recommended. Polyps may also grow in the stomach, duodenum, and small bowel.

The extra-intestinal tumours include osteoma of the skull, epidermoid cyst, fibroma, neurofibroma, sebaceous cyst, and desmoid tumours in approximately 15% of affected individuals.[4] Approximately 2% of desmoid tumours are associated with familial adenomatous polyposis, and patients with familial adenomatous polyposis are 852 times more likely to develop desmoid tumours than the general population without familial adenomatous polyposis.[5] Desmoid tumour is a benign tumour characterised by local invasiveness. The tumour often recurs after surgical resection. Desmoid tumour can present in peripheral or intra-abdominal forms. The tumour often arises in the abdominal wall, frequently from the aponeurosis of the rectus abdominis muscle and mesentery. Desmoid tumour is also known as mesenteric fibromatosis. Due to its desmoplastic tendency, desmoid tumour often leads to obstruction of the intestine and urinary tract.[6] These tumours cannot be differentiated from other soft tissue tumours by CT or magnetic resonance imaging. Histology is the only means of obtaining a definitive diagnosis. There is an association between intra-abdominal desmoid tumour with recurrent intratumour abscess formation, which has been documented in the literature.[7,8] Percutaneous drainage is usually the initial management.[8] There has been one report of pneumoperitoneum with intra-abdominal desmoid tumour and a 0.5-cm perforation was noted at the distal ileum intraoperatively.[9] Although preoperative CT showed gross pneumoperitoneum in this patient, no intestinal perforation was identified during surgery, possibly due to rupture of the intra-abdominal abscesses.

CONCLUSION

Abdominal abscesses with local recurrence should raise the possibility of intra-abdominal desmoid tumour, especially in the presence of pneumoperitoneum. Active search for evidence of intestinal polyposis may lead to a diagnosis of Gardner's syndrome so that prophylactic colectomy can be offered.

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