

Diffuse intestinal submucosal lipomatosis with incidental epidermal inclusion cyst of caecum clinically masquerading as carcinoma caecum

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Abstract

Symptomatic diffuse submucosal intestinal lipomatosis is a rare entity. Also few cases of epidermal inclusion cyst of caecum have been reported in literature. Here, we are presenting a rare case of intestinal submucosal lipomatosis with coincidence of epidermal inclusion cyst of caecum and presumptively diagnosed as carcinoma of ileocaecal region during surgery in a 55 years old male. Both are rare entity considering the location even they should be kept as a differential diagnosis in unusual cases of intestinal perforations with inconclusive radiological findings or clinical uncertainty.

Introduction

Diffuse intestinal submucosal lipomatosis is very rare condition with a few cases reported in the medical literature.¹ Usually these patients are asymptomatic but may present as perforation, obstruction, ulceration, intussusceptions or bleeding.^{2,3} Epidermal inclusion cyst of the caecum is also a rare lesion which is considered a sequestration cyst, either of congenital or acquired origin.⁴ Here, we are presenting a rare case of intestinal submucosal lipomatosis with coincidence of epidermal inclusion cyst of caecum and presumptively diagnosed as carcinoma of ileocaecal region during surgery in a 55 years old male.

Case Report

A 55 years old male presented to emergency department with abdominal pain for two days. He had the history of previous hospital admission about three months back with continuous periumbilical pain of progressively increased intensity and aggravating by straining. Clinically, diagnosis of perforation peritonitis

was considered however computed tomography scan film showed a mass in ileocaecal region (Figure 1A). By that time, on emergency exploratory laparotomy an ileal perforation of 1×1 cm was seen at 20 cm. proximal to ileocaecal junction with coincidental finding of a firm, ill defined and immobile caecal mass of approximately 10×8 cm. in size. At the site of perforation bowel was found to be dilated with thinned out wall preoperatively. A biopsy from the mass and loop ileostomy for symptomatic relief were performed. Biopsy report did not reveal significant pathology. Post operative period was uneventful and the patient was discharged with advice of proper follow up.

Now patient was readmitted and subjected to contrast enhanced computed tomography scan of abdomen revealing well defined mass near ileocaecal junction which was of 6.3×4.7×4.6 cm. with definite enhancing wall and 80 cc of volume (Figure 1B). There is evidence of mildly distended small bowel loops with multiple air fluid levels proximal to the ileostomy site with mild free fluid in peritoneal cavity. Image guided aspiration cytology of the lesion revealed small keratinous material with many nucleated squamous cells and was reported as epidermal inclusion cyst of ileocaecal region. Following this, patient was planned for right hemicolectomy.

Gross

An excised terminal ileum with attached caecum, part of right ascending colon and appendix measuring 21.0 cm. in length received. On cutting, lumen of terminal ileum with thickened wall was found to be obliterated. A cystic mass measuring 4.2 cm. in maximum diameter was seen in caecal wall which was filled with cheesy material (Figure 2A). No calcification, hair, teeth or bone elements were detected. However, mucosa appeared to be normal with focal loss of folds.

Histopathology

Multiple sections from terminal ileum, caecal mass and ascending part of colon showed diffuse expansion of submucosa by lobules of mature adipocytes with focal superficial denudation of mucosal epithelium and few scattered chronic inflammatory cells in lamina propria (Figure 2B). Serosal surface of caecum revealed edema and a cyst lined by stratified squamous epithelium with presence of granular layer. Cystic cavity was filled with lamellated keratin layer (Figure 2C). No dermal adnexal structure was seen. One section also showed a small keratinous cyst in the muscle layer. (Figure 2D) Other layers were unremarkable. Focal area showed foreign body giant cell reaction (Figure 2D, inset). Histological diagnosis of diffuse submucosal intestinal lipomatosis with epidermal inclusion cyst of caecum was considered. The patient had an uneventful post-operative follow up of six months.

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Key words: ileocaecal, submucosal lipomatosis, epidermal inclusion cyst, peritonitis.

Contributions: BT, concept, design, literature search, manuscript preparation and editing; SK, concept, design, definition of intellectual content, manuscript editing and review; AB, design, definition of intellectual content, literature search, manuscript editing and review; SK, design, definition of intellectual content, manuscript preparation, editing and review.

Conflict of interests: the authors declare no potential conflict of interests.

Received for publication: 3 April 2014.
Revision received: 16 July 2014.
Accepted for publication: 16 July 2014.

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Rare Tumors 2014; 6:5380
doi:10.4081/rt.2014.5380

Discussion and Conclusions

Symptomatic intestinal lipomatosis, first described by Hellstrom in 1906, is a rare entity with an incidence at autopsy ranging from 0.04 to 4.5%.² Lipomatous lesions of the intestine may be solitary or multiple, encapsulated lipomas or diffuse, discrete, unencapsulated lobules of adipose tissue, called lipomatosis. In 90% of cases, these are localized in submucosa but occasionally they extend into the muscularis propria, while up to 10% are subserosal.⁵ Sometimes only one segment of intestine may be involved, known as segmental lipomatosis. Age of presentation is highly variable, ranging from neonatal period to seventh decade of life.^{1,6} Mostly asymptomatic, but they may precipitate a surgical emergency such as intussusceptions, obstruction, bleeding or perforation peritonitis especially when the ileocecal valve and small intestine are affected.^{3,7}

The etiology of lipomatosis is yet to be established. Hypothetical etiological factors include embryonic displacement of adipose tissue, degenerative disease with disturbance of fat metabolism, post-chemotherapeutic fat deposition, chronic irritation such as chronic inflammatory bowel disease, low-grade infec-

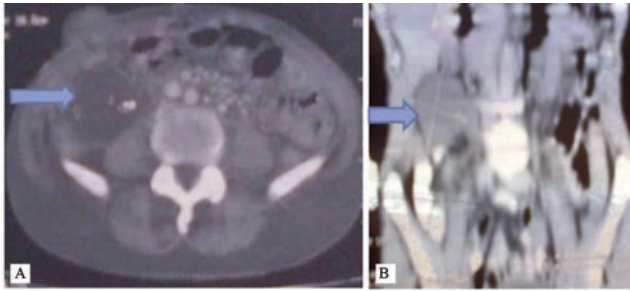


Figure 1. A) First preoperative computed tomography (CT) scan showing ileocaecal mass (arrow). B) Second preoperative CT scan revealing ileocaecal mass after loop ileostomy (arrow).

tion and hamartomatous syndromes.⁸ Catania *et al.* have reported association of segmental or diffuse lipomatosis with cutaneous, epilipoid, small intestinal, gastric or duodenal lipomas or with diverticula of colon or neurofibromatosis.³

Clinical diagnosis may be difficult but a preoperative computed scan may be of value to characterize large submucosal masses because it can show the specific nature of a mass and the extent of disease. Plain abdominal films are nonspecific and commonly demonstrate the presence of multiple air/fluid levels suggestive of mechanical obstruction. Barium enema may show filling defect in well circumscribed lesion. However, this appearance may resemble that of carcinomas and therefore diagnosis by this method is definitive only in some of the cases. In the majority of cases, biopsy cannot establish diagnosis as the lesion lies beneath the normal intestinal mucosa. The intra operative appearance of the lesion is confusing and it may be difficult to exclude carcinoma on gross appraisal during surgery.

Development of epidermal inclusion cyst in the intestine is very rare, considered to be sequestration or implantation cyst and not as neoplastic growth.⁴ Only a few cases of epidermoid cyst of the caecum have been reported in the literature.^{4,9} It could be acquired following implantation of epidermal fragments after trauma (*e.g.* post-surgical, spinal puncture), or can arise from congenital heterotopic cutaneous tissue either at the time of closure of the neural groove or of coalescence of other epithelial fusion lines.⁴ Other possible explanations exist like squamous metaplasia of enterogenous cysts and teratoma.¹⁰ These cysts may present with other complications including rupture, hemorrhage, necrosis, torsion, intussusception, obstruction, peritonitis, and malignant change. The differential diagnosis includes dermoid cyst and cystic teratoma that usually demonstrate other skin structures and embryonic tissue elements, respectively, which were absent in our case. To the best of our knowledge, the present

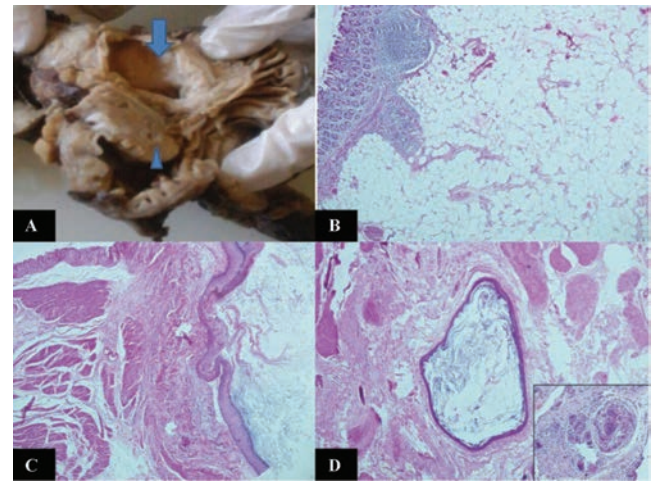


Figure 2. A) Gross specimen showing thickening of intestinal wall (arrowhead) along with cut open cystic cavity on serosal surface filled with cheesy material (arrow). B) Section showing diffuse expansion of intestinal submucosa by collection of mature adipocytes (Hematoxylin & Eosin, 40 \times). C) Section showing serosal cystic cavity lined by stratified squamous epithelium with lamellated keratin (Hematoxylin & Eosin, 100 \times). D) Section showing small epidermal inclusion cyst within the intestinal muscle layer (Hematoxylin & Eosin, 100 \times). Inset showing foreign body giant cell reaction (Hematoxylin & Eosin, 400 \times).

report is the first to describe a patient of diffuse intestinal submucosal lipomatosis with coexistence of epidermal inclusion cyst of caecum which is complicated due to perforation peritonitis initially. Later on, patient presented with obstructive symptoms. Before first laparotomy, there was no preceding iatrogenic cause, therefore it represent congenital sequestration cyst which is uncommon regarding the site of involvement. Perforation peritonitis might be due to spontaneous rupture of the cyst or relative avascularity of lipomatous lesion which makes the overlying mucosa prone to ischemia leading to perforation. Treatment always involved resection of the severely diseased bowel for symptomatic lipomatosis or in the cases where risk of malignancy can not be definitively excluded. Surgical approach should depend mainly on the presentation of the case as an elective or an emergency and must be established through the features of each case on an individual basis. Although, both are rare entity considering the location even they should be kept as a differential diagnosis in unusual cases of intestinal perforations with inconclusive radiological findings or clinical uncertainty.

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