Abdominal mass with significant family history

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Clinical protocol

A 29-year-old man was admitted in December 2006 for abdominal distension and swelling of both legs. The distension was not associated with flank fullness or decreased urine output. There were no associated cardiac or respiratory complaints. There was no history of jaundice, gastrointestinal bleed, or bladder and bowel disturbances. There was no symptom to suggest chronic liver disease in the past.

Past history

The patient was involved in a road traffic accident in August 2003 and sustained fracture of the mandible. There was no obvious injury to the abdomen then.

In October 2003 he was admitted elsewhere with a history of abdominal pain, nausea, vomiting and fever of 1-week duration. The pain was in the right iliac fossa, mild, dull and continuous. There was no history of abdominal distension, weight loss, altered bowel habits or mass in the abdomen at that time.

Family history

The patient’s mother had a swelling in the neck and was operated on thrice; his sister had swelling in the anterior abdominal wall extending up to the groin and underwent surgery. The details of both surgeries were not available.

Examination findings

Clinical examination showed a malnourished, anemic individual with bilateral pedal edema, with no jaundice, cyanosis or lymphadenopathy. A huge mass(es) was felt extending from the epigastrium to the right iliac fossa. The mass(es) was firm to hard in consistency, immobile, with no bruit; there was no ascites, no visible peristalsis. Per rectal examination was normal.

Investigations (October 2003)

Hemoglobin 13.2 g/dL, TLC 15,400/cmm (P88, L12), ESR 73 mm in 1st hour; liver and renal profiles were within normal limits.

Ultrasonography showed diffuse fatty changes in the liver; gall bladder and pancreas were normal. Spleen appeared enlarged and measured 13.5 cm. A well-defined, hypoechoic mass with central echogenic areas was seen in the right iliac fossa. CT scan showed a mass in the right iliac fossa with luminal air along its medial aspect, with centrally placed distended bowel loops with air-fluid levels within, and soft tissue density along its walls. In view of the history of trauma, mesenteric injury with possible internal herniation of bowel loops with wall hematoma and secondary obstruction was considered.

At laparotomy a hard cecal mass was noted with mesenteric abscess, fecal peritonitis, and inflamed terminal ileum which was adherent to the mesentery. The mesenteric abscess was opened and pus drained. The free side of the ileum was anastomosed to the ascending colon.

Histology of the cecal mass showed only scanty material with a rare granuloma. The mesenteric node showed necrosis, hemorrhage and inflammation, consistent with an abscess. The omental tissue showed congestion, inflammation and focal reactive mesothelial hyperplasia. Culture of the pus showed heavy growth of E. coli.

Anti-TB IgM serology was positive in low titers in the postoperative period. The patient was therefore treated with anti-tubercular drugs for one year but had no response. Fever, vomiting, loss of appetite and back pain continued intermittently from 2004 to the current admission. He took symptomatic treatment from a local doctor, with no relief.

Investigations (December 2006)

Hemoglobin 7.8 g/dL, TLC 7200 (P 70, L 28, E2), hypochromic and microcytic anemia. Liver profile normal; serum creatinine 1.7 mg/dL. Electrocardiogram: sinus tachycardia. Chest radiograph: prominent bronchovascular markings.

Ultrasonography showed a hypoechoic mass in the right hypochondrium / epigastric region and right iliac fossa, measuring 12.7 cm x 9.0 cm in the right hypochondrium / epigastric region and 7.0 cm x 7.4 cm in the right iliac fossa, and a suspected intraperitoneal mass lesion, 10.6 cm x 6.0 cm, involving small bowel loops (?matted small bowel loops ?cocoon formation). CT scan showed a centrally placed large soft tissue intraperitoneal mass.
lesion with displacement of small and large bowel loops. A mesenteric mass (Figure 1). Small bowel enema showed soft tissue masses occupying the mid-abdomen causing external compression and displacement of ileal and jejunal loops, cecum and ascending colon, with dilatation of jejunal loops, suggestive of lymph nodal mass; lymphoma was suspected.

The patient was stabilized with blood transfusion, fluid and electrolyte replacement and albumin infusion. Subsequently he was taken up for laparotomy.

**Unit’s clinical diagnosis**

Huge abdominal mass – retroperitoneal / mesenteric in origin. Significance of family history and temporal relationship with trauma, not clear.

At laparotomy a midline mass was seen, firm to hard in consistency, with dense fibrous adhesions of all loops of the intestine to the anterior and lateral abdominal wall and totally enclosing the mass. The tumor was unresectable due to encasement of the superior mesenteric artery (Figure 2). The mass was biopsied.

The patient had a stormy postoperative period and succumbed to sepsis after 7 days. Consent for autopsy was denied.

**Discussion (P Abraham)**

The presenting symptoms of this patient suggest that he had solid (or localized fluid) mass or masses in the abdomen / pelvis. Swelling of the legs may be due to pressure effect from the pelvic mass(es) and / or hypoalbuminemia of chronic disease. He is unlikely to have had ascites or any luminal disorder.

He had a significant family history. His mother had swelling in the neck and was operated on thrice; the nature of the swelling was not known. His sister was also operated on for a swelling in the anterior abdominal wall (extending to the groin). However, details of the surgery were not available.

Considering his examination findings as well, the overall clinical impression is of a large solid (intra)abdominal mass(es) not extending deep into pelvis (rectal examination normal) and not compromising gut lumen; there was no vascular compromise, hypervascularity or peritoneal exudation clinically. The pathology in the mother and sister is probably similar lesions elsewhere. The description in the sister suggests an abdominal parietal mass.

Together with the laboratory findings, we see a sick patient with poor nutrition possibly due to a chronic disease, with abdominal mass(es), with low-grade or no active inflammation. The picture at admission 3 years ago was suggestive of inflammatory, possibly infective process in the ileo-cecal / appendicular region.

After laparotomy the patient received anti-tubercular treatment for a year with no response. Since then he has been getting recurrent episodes of fever, anorexia, vomiting and back pain and was treated by the family physician.

Analyzing and interpreting the findings so far, it appears that the patient had a chronic disease, which was an ongoing indolent abdominal inflammation, predominantly extraluminal but with no significant exudation, involving gut lumen with adhesion, low-grade obstruction, and one episode of perforation, and no lasting
response to antibacterials and standard anti-tubercular treatment. At the present the patient was in poor nutritional state (immune status not known).

The possibilities are

- Non-infective inflammation ?etiology
- Multi-drug resistant tuberculosis
- Low-grade lymphoma with desmoplasia

The imaging findings at the present admission suggest fibrosing mesenteric / peritoneal disease with encasement ("cocoon") of, or adhesions to, intestinal loops; or a lymph nodal mass.

With these possibilities the hard cecal mass, mesenteric abscess and perforative peritonitis cannot be explained. But mesenteric abscess and perforative peritonitis can be triggering factors for peritoneal fibromatosis.

My final diagnosis (prior to surgery) is

1. Mesenteric fibromatosis – desmoid – primary or secondary (fibromatosis in neck in mother and on abdominal wall in sister), or its variant (sclerosing mesenteritis [b-catenin negative])
2. Desmoplasia associated with low-grade lymphoma (MALToma) / neuroendocrine tumor (non-secreting)
3. Gastrointestinal stromal tumor (GIST)

Less likely possibilities are:

- Foreign-body granulomatous disease
- Other granulomatous disease (multi-drug resistant tuberculosis, Crohn’s disease, fungal) with organized abscess and adhesions.

Comments

Ashok Chacko: Possibility of GIST needs to be considered. This possibility can explain the large extraluminal mass lesion, slow and indolent course without ascites. The cecal perforation noticed three years before may be due to extrinsic compression of bowel by the mass, leading to ischemia and perforation.

K Raghuram: In view of slow-growing large abdominal tumor without ascites, low-grade lymphoma may be considered.

VS Sankaranarayanan: A possibility of fungal infection like actinomycosis should be included, since the mass originated in the ileocecal region.

VG Mohan Prasad: Neurilemmoma and schwannoma may be considered in this patient with large abdominal mass and no bowel involvement.

Pokli: An immunocompromised state like HIV infection, and related low-grade malignancy needs to be considered.

TS Chandrasekar: The HIV status of this patient was negative and there was no evidence of oral thrush or features of either focal or systemic fungal infection.

Pathology protocol (R Lawrence)

Multiple firm grey-white tissue fragments were submitted, in aggregate measuring 8-9 mm. Histologically, the lesions showed low to intermediate cellularity. The cells were uniform and spindle-shaped, in a dense bed of collagen (Figures 3, 4). Mitosis was not present in several sections studied. The margins were infiltrative. There was no pleomorphism and no tumor giant cells were present. The microscopic features were those of aggressive fibromatosis.

Figure 3: Histology of biopsy from mass showing slender spindle shaped cells in dense fibrous stroma (10 X)

Figure 4: Histology showing uniform spindle-shaped cells. No mitosis or nuclear atypia are seen (40 X)
**Final histopathology diagnosis**

Aggressive intra-abdominal deep fibromatosis (desmoid tumor)

**Discussion on pathology**

The term fibromatosis was proposed by Stout et al for a broad group of benign fibrous proliferative conditions of similar microscopic appearance. They are intermediate in their biological behavior between benign fibrous lesions and fibrosarcoma. Broadly, there are two main groups: superficial and deep fibromatosis.

The term desmoid tumor was originally applied to lesions that arise during pregnancy or in the postpartum period, as abdominal mass, independent of the uterus. Presently this term refers to deep fibromatosis, a group of clinically diverse, deep-seated fibrous neoplasms belonging to the family of myofibroblastic tumors. They have bland histology, slow growth and lack metastatic potential. They have hormone receptors and have binding sites for estrogen and anti-estrogen in some cases. They may respond to hormonal manipulations, e.g., tamoxifen, aromatase inhibitors and gonadotropin-releasing hormone agonist.

These lesions are divided into three main biological groups – sporadic, those associated with familial adenomatous polyposis (FAP), and those that are multicentric or familial.

Desmoids are classified by their location into three main subtypes:

1. Extra abdominal – 60%
2. Abdominal wall – 25%
3. Intra abdominal wall – 15%

This patient had familial diseases presenting as multicentric intra-abdominal tumors. Probably his mother had a lesion at an extra-abdominal site (neck) and sister had similar lesions on the abdominal parietal wall.

The term aggressive fibromatosis is often applied to these tumors, especially when they occur in the retroperitoneum. This refers to their potential for invasion and progressive growth. These tumors form large, infiltrative masses, which will recur if not excised widely but they do not metastasize.

Abdominal and retroperitoneal desmoids, along with fibromas, osteomas and epidermal cysts, are extra-colonic manifestation of FAP, associated with Gardner’s syndrome. A familial form of desmoid, not associated with FAP, is linked to mutations in the APC gene on the long arm of chromosome 5. Genetic, endocrine and physical factors play a role. Often, a history of prior surgery is the triggering mechanism. In this case prior abdominal surgery probably played a role.

**References**


The session was chaired by Professor Gourdas Choudhuri, Department of Gastroenterology, Sanjay Gandhi Post Graduate Medical Institute, Lucknow. Panel included Professor Ashok Chacko, Christian Medical College, Vellore; Professor K Raghuram (Retd), Madras Medical College, Chennai; VG Mohan Prasad, Gastroenterologist, Coimbatore; VS Sankaranarayanan, Pediatric Gastroenterologist, MIMS, Chennai; Pokli, Physician, Haryana.

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