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Post Surgical Desmoid Tumors in Familial Adenomatous Polyposis: A Case Report

Literature review indicates that desmoid tumors are exceedingly common in familial adenomatous polyposis (FAP), where the comparative risk is 852 times that of the general population. Prior abdominal surgery has been found in as many as 68 % of FAP patients with abdominal desmoid. Fifty-five percent develop these lesions within 5 years of surgery. We describe a 45-year-old patient with Gardner's syndrome complicated by a desmoid tumor 2 years after he had a prophylactic colectomy.

Keywords: surgery, fibromatosis, aggressive, gardner syndrome, adenomatous polyposis coli

Introduction

There are two major etiologic classification categories for desmoid tumors, namely, sporadic versus hereditary (those occur in familial adenomatous polyposis (FAP), particularly the so-called Gardner's variant). Sporadic desmoid tumors occur primarily in the abdominal wall and in extra-abdominal sites, in contrast to desmoid tumors in FAP, that occur in the bowel mesentery as well as in the abdominal wall. Desmoid tumors are sometimes difficult to distinguish from low-grade fibrosarcomas; however, they lack sarcomatous nuclear and cytoplasmic features. Nevertheless, some authors consider desmoid tumors the same as low-grade fibrosarcomas.¹ Attempts at palliative or curative resection of the tumors result in high rates of morbidity because of the lesions recur.²⁻⁴

Incidence of desmoid tumors in FAP

Berk et al. in a review of 498 patients registered in the Steven Atanas Stavro FAP Registry at the Mount Sinai Hospital in Toronto, Canada, found that 53 (11 %) had desmoids, 40 of which were mesenteric desmoids.⁵ The mean age was 31 years and most of the tumors were diagnosed after colectomy for FAP. Of these 40 individuals, 7 (18%) died as a result of desmoid-related complications. The authors conclude "... surgical excision of desmoids is hazardous and was associated with a recurrence rate of up to 85% in this study."

Jarvinen studied 71 FAP families with 168 verified FAP patients and found that 19 (11%) were complicated by desmoid tumors,⁶ while only six of these FAP patients (3.6%) had a desmoid before colectomy. The cumulative risk of postoperative desmoid tumors was 16% at 10 years after colectomy. The median interval between the surgery and the diagnosis of desmoid tumor was 2.3 years. The mean age of desmoid occurrence was 28.5 years.

Gurbuz et al. investigated the risk of desmoid in FAP, as well as the relationship between specific APC gene mutations and desmoid tumor formation.⁷ The absolute risk of a desmoid in FAP patients was 2.56/1000 person-years, whereas

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the comparative risk was 852 times that of the general population.

Our purpose is to describe a patient with Gardner's syndrome who manifested recurrent desmoid tumors in all the three-trocar sites within a year of laparoscopic examination.

Case Report

The patient was a 46-years-old Iranian male who displayed the full phenotypic triad of Gardner's syndrome, namely colonic adenomatous polyps, osteomas, and epidermoid_cysts. His father and uncle each had Gardner's syndrome and died of colorectal carcinoma at ages 45 and 50, respectively. He underwent proctosigmoidoscopic examination in 2002 at age 43, when the characteristic colonic phenotype of multiple colonic adenomas was diagnosed. He then underwent a prophylactic colectomy with ileorectal anastomosis. Because of leakage at the site of surgery, ileostomy was performed.

Two years after the surgery, the patient had a complication of multiple abdominal masses. On ultrasound, multiple large masses with heterogeneous echogenicity were seen that extended from the epigastric area down to the pelvis.

On CT scan, a large soft tissue mass with heterogeneous density was seen extending from the upper abdomen to the pelvis, with irregular enhancement in



Fig 1. IVP film shows multiple soft tissue masses in LUQ, mid abdomen with extension to pelvis, which deviates and compresses the small bowel loops. Also medial deviation of left ureter in upper right and lateral deviation in pelvic is noted.

the small bowel mesentery, irregular mucosal thickening, involvement of the rectus muscles, upward

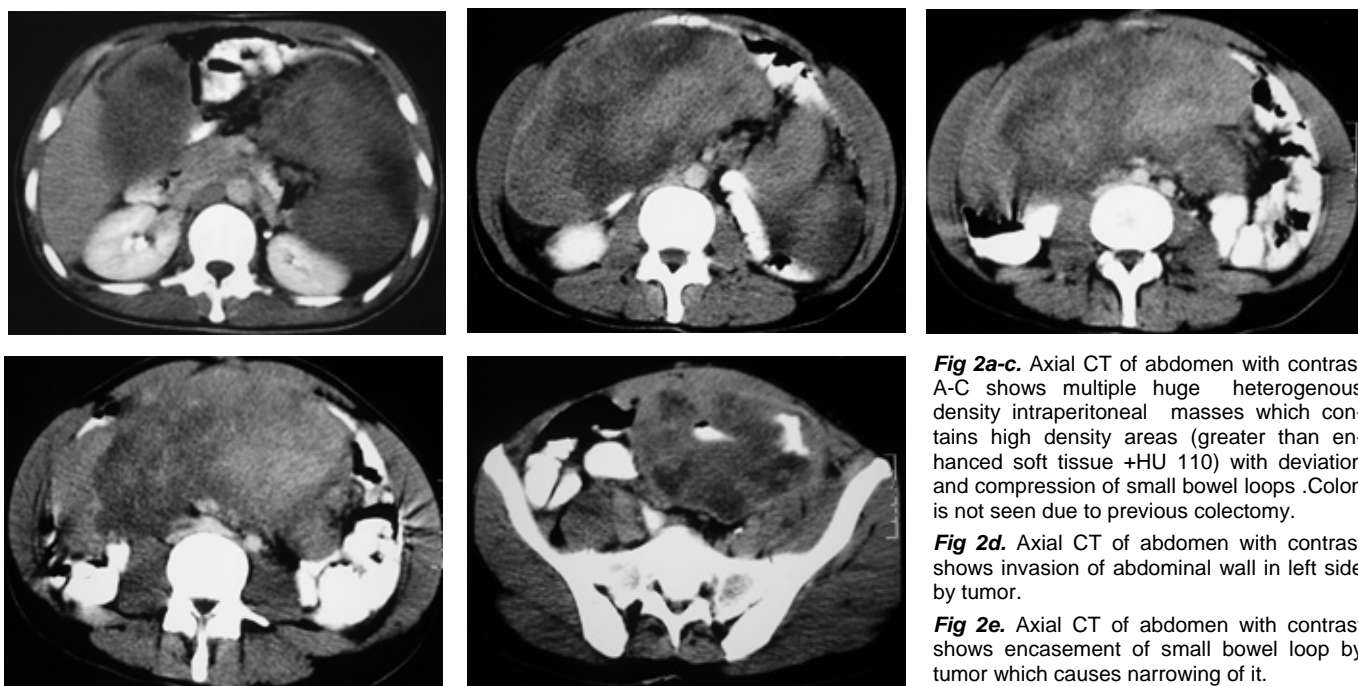


Fig 2a-c. Axial CT of abdomen with contrast A-C shows multiple huge heterogeneous density intraperitoneal masses which contains high density areas (greater than enhanced soft tissue +HU 110) with deviation and compression of small bowel loops. Colon is not seen due to previous colectomy.

Fig 2d. Axial CT of abdomen with contrast shows invasion of abdominal wall in left side by tumor.

Fig 2e. Axial CT of abdomen with contrast shows encasement of small bowel loop by tumor which causes narrowing of it.

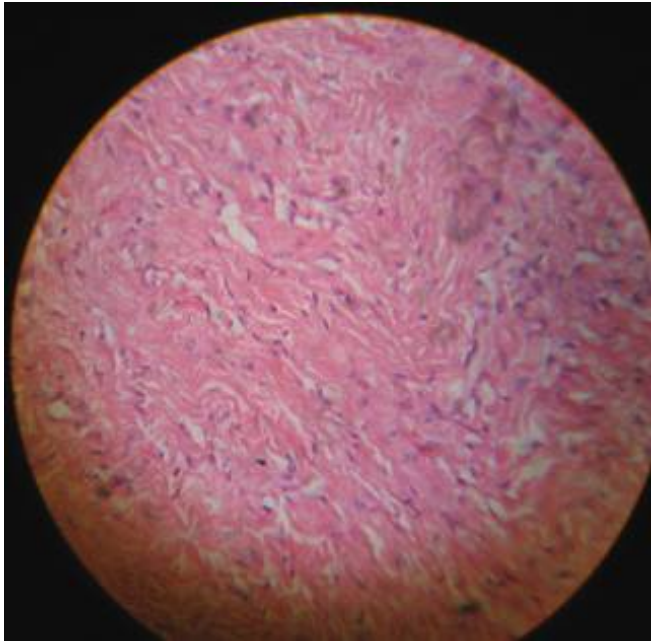


Fig 3. In pathologic examination, proliferation of myofibroblasts composed of spindle large cells without mitosis and cellular pleomorphism are seen with vascular and collagen fibers in stroma with thick membrane which is compatible with fibromatosis

displacement of the left kidney, and pressure effect on the bladder (Figure 1 and 2).

At laparotomy, a large intraperitoneal mass with fibrosis and adhesions to small bowel were seen, which was inoperable.

On pathologic examination, proliferation of myofibroblasts was evident that consisted of large spindle cells without mitosis and cellular polymorphism, with vascular and collagen fibers in the stroma, and thick membranes, compatible with fibromatosis (Figure 3).

Desmoid tumors and APC gene mutations

The subclassification of desmoids in FAP/Gardner's syndrome may be elucidated by identifying mutations in the APC gene. This identification may be of clinical/genetic importance, because knowing whether a FAP patient is or is not at increased risk for desmoid tumors developing could impact the surgical management of FAP.

CT scans of desmoid

The role of CT in the assessment of desmoid tumors in FAP patients is limited. Kawashima et al. reviewed 101 abdominal CT-scans from 23 patients over a 13-year time frame.⁸ Among the desmoids, 40 were intra-abdominal, with 30 of them being associated with

Gardner's syndrome in 13 patients, whereas 10 desmoids in 10 patients were sporadic. An attempt was made to define the desmoid location, whether they were single or multiple, and whether there were any specific CT characteristics relevant to their margins, attenuation numbers, or contrast enhancement. Findings disclosed that there were no CT characteristics, such as attenuation values, margins, or contrast enhancement findings that could allow differentiation between isolated intra-abdominal desmoids and those associated with Gardner's syndrome. However, it was found that desmoids associated with Gardner's syndrome occurred more often in the mesenteric abdominal wall, whereas those in isolated cases involved the retroperitoneum and pelvis. Following the CT-scans over time, it was observed that new lesions appeared to develop more often in the Gardner's syndrome (3 of 13), whereas no new lesion was observed in those individuals with isolated desmoids.

Because of the diagnostic limitations of CT-scan, we chose laparoscopy, which unfortunately precipitated a desmoid tumor at each trocar site. We believe this experience is the most significant observation ever reported indicating surgical intervention in the initiation of desmoids in FAP patients.

Secular cancer mortality changes in FAP

Belchetz et al. studied 461 individuals with FAP from 158 families, in which 140 deaths had been recorded.⁹ The results showed that when stratified by decade, from the 1930s to the 1990s, increasingly more deaths had occurred from extracolonic manifestations of FAP than those caused by colorectal cancer."

These results clearly indicate the need for careful monitoring of patients with FAP for extracolonic manifestations once the disease has been diagnosed in the family, and careful attention to the causes of prophylactic colectomy. Herein particular attention must be given to periampullary neoplasms, papillary thyroid carcinoma, and desmoid. Unfortunately, with respect to periampullary tumors and desmoids, there are no known cost-effective surveillance protocols or therapeutic interactions for preventing morbidity and mortality from these lesions.^{10,11} However, certain FAP families appear to show familial aggregation and segregation of desmoids and periampullary cancer;

therefore, this knowledge might be used prudently in the management of patients from these families.^{7, 12}

In conclusion, molecular genetic information in FAP may provide clues as to which patients are most susceptible to desmoid formation. Avoidance of surgery whenever possible should be exercised in these genetically susceptible patients.

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