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Intra-abdominal desmoid tumor on the background of lynch syndrome. An unusual association mimicking recurrent colon cancer

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Introduction

Desmoid tumors are rare benign tumors that tend to be locally invasive rather than metastatic, and have a high recurrence rate despite complete excision [1]. Intra-abdominal desmoids are sporadic in most cases with only 20% associated with familial syndromes, in particular Familial Adenomatous Polyposis (FAP). Sporadic cases seem to be less aggressive and associated with a lower mortality and recurrence rate than FAP-associated desmoids [2]. The etiology is unknown, however, a history of previous trauma is sometimes identified [3]. Desmoid tumor diagnosis in the context of Hereditary Nonpolyposis Colorectal Cancer (HNPCC- Lynch syndrome) has not been previously described. The authors report a case of intra-abdominal desmoid tumor in a patient with HNPCC mimicking recurrence of colon cancer, and review the literature.

Case report

A 46-year-old gentleman was referred to our institution with a 6-month history of recurrent rectal bleeding. He had a strong family history of cancer with two brothers having died of colon cancer before the age of 50, and a son of brain tumor at the age of 2.

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Colonoscopy showed a large hypervascular pedunculated polyp (2x2.5 cm) in the sigmoid colon that was excised. Histology revealed a tubulo-villous adenoma with no dysplasia. Follow-up colonoscopy a year later, showed no recurrence at the polypectomy site, but another sessile 2x1 cm polyp was found and excised in the ascending colon. Histology showed a focus of well differentiated adenocarcinoma (pT1a) arising within a tubular adenoma. Resection margins were negative. CEA level was within normal limits. A staging CT of the chest, abdomen and pelvis was unremarkable.

The case was discussed at the multi-disciplinary colorectal tumor board and the patient, decision was taken for the patient to undergo surgical resection. A laparoscopic right hemicolectomy with primary side-to-side ileo-transverse anastomosis was performed. Histology revealed no residual malignancy or dysplasia, and all 16 harvested lymph nodes were negative for cancer. Therefore, no adjuvant chemotherapy was administered.

Genetic testing confirmed the diagnosis of Hereditary Non-polyposis Colorectal Cancer (HNPCC-Lynch syndrome). The c.2131C>T (p.Arg711*) variant was detected in heterozygosity in the MSH2 gene. No mutations were detected in the MLH1 gene.

At 1-year follow-up, the patient was asymptomatic with no complaints. Blood tests including CEA were normal. A follow-up colonoscopy showed few ulcerations at the anastomotic site, biopsies of which revealed only non-specific inflammation. A follow-up CT scan of the abdomen and pelvis showed a speculated mesenteric mass of 2x1.7 cm in close proximity to the ileo-colic anastomosis. There was peri-anastomotic lymphadenopathy of up to 0.8 cm (Figure 1). A PET-CT scan was performed, and this showed the mass to have FDG uptake with SUV max of 4.67 (Mean Liver SUV based on body weight was 2.6) (Figure 2). Findings were suggestive of local recurrence and an exploratory laparotomy was carried out. The previous ileo-colic anastomosis with the adjacent mesenteric mass were excised. A primary side-to-side anastomosis was carried out. Histopathology showed no malignancy, but an ill-defined spindle cell proliferation within the pericolic fat, and mesentery arranged in short sweeping fascicles with long thin-walled blood vessels (Figure 3,4). Tumor cells were positive for beta catenin and negative for SMA, desmin and CD117, confirming the diagnosis of desmoid tumor and excluding other differential diagnoses of GIST, leiomyoma, neurofibroma and schwannoma.

The post-operative course was complicated by a right-sided intra-abdominal collection with no evidence of leak. An ultrasound-guided drainage of the collection was performed, and the patient made an uneventful recovery. Two years' surveillance workup showed no evidence of recurrence.

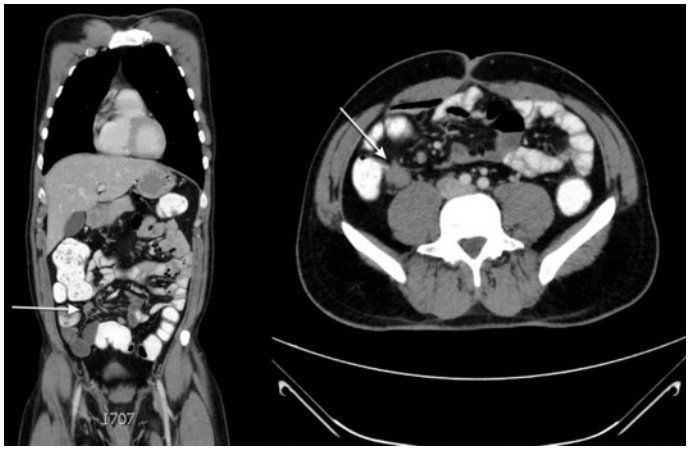

Figure 1: Coronal and transverse CT scan sections, showing a suspicious soft tissue mass at the surgical bed (arrow)

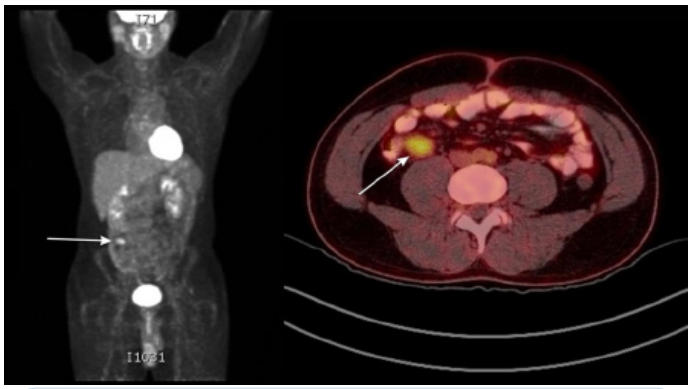

Figure 2: Coronal and transverse PET-CT scan sections showing FDG uptake of the mass (arrow).

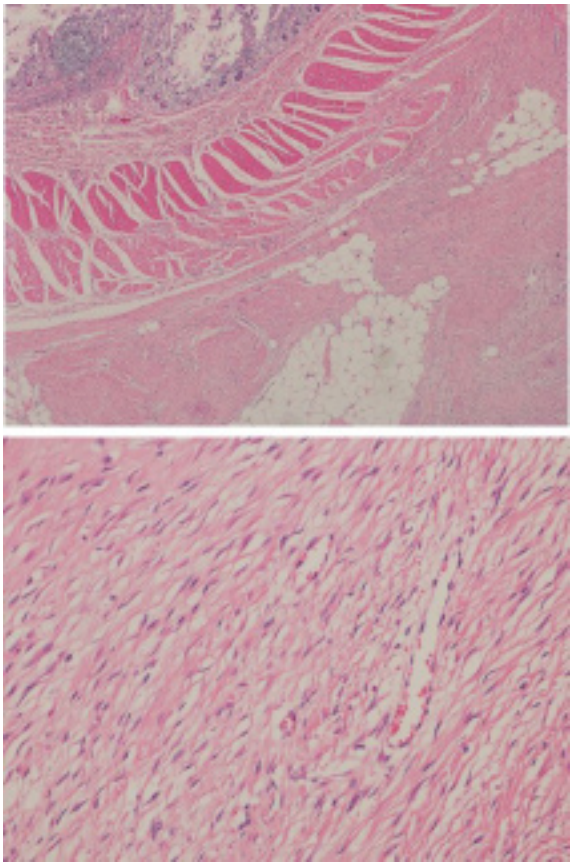

Figure 3: Microscopic sections showing spindle cell proliferation without significant atypia or pleomorphism. HE x10(b) HE x40.

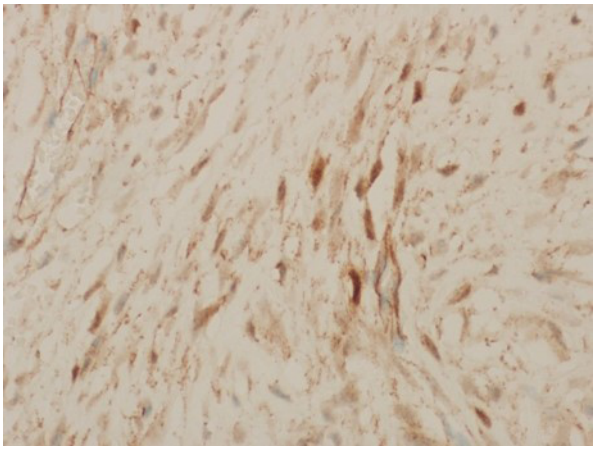

Figure 4: Positive nuclear staining with beta-catenin stain. HE x40.

Discussion

Hereditary Nonpolyposis Colorectal Cancer (HNPCC- Lynch syndrome) is one of the most common hereditary colonic polyposis syndromes along with Familial Adenomatous Polyposis (FAP) syndrome. They both account for about 5% of colorectal cancer cases.

Lynch syndrome is an autosomal dominant disease caused by a pathogenic mutation of mismatch repair (MMR) genes, with exclusion of BRAF mutation [4]. Clinical screening is based on the "revised Bethesda criteria"[5], the Amsterdam I or Amsterdam II criteria [6,7].

Desmoid tumors are rare benign tumors. WHO defines desmoid as "clonal fibroblastic proliferation that arises in the deep soft tissues and is characterized by infiltrative growth and a tendency toward local recurrence but an inability to metastasize". They account for about 0.03% of all neoplasms and <3% of all soft tissue tumors [1]. The incidence in USA general population is ~2-4 per million per year [1]. The peak age of diagnosis is 30-40 years with a female predominance [1].

The vast majority of cases are sporadic and found in the abdominal wall and in extra-abdominal sites, with less than 15% of cases being intra- abdominal. However, in patients with FAP the percentage of desmoids in the intra-abdominal location reaches 50 % [3].

The etiology of sporadic desmoid tumors is poorly understood, but there is an association with a history of trauma in 30 % of cases [9]. Family history of desmoid tumors, pregnancy, and steroids have also been implicated as etiological factors [10-12].

Radiological studies such as Computed Tomography (CT) scan and Magnetic Resonance Imaging (MRI) are useful for diagnosis, planning of surgery, monitoring response to radiotherapy or hormonal therapy, and surveillance to detect recurrence. On imaging studies, desmoids appear as well-circumscribed hypervascular soft tissue masses, without calcifications [13]. PET scan may show increased FDG uptake due to the high cellular and mitotic activity of desmoids [14-18]. Therefore, findings may be misleading, suggesting malignancy especially in patients with a previous history of cancer as in the reported case.

Diagnosis and grade of differentiation are established by biopsy showing monoclonal fibroblastic proliferation appearing as small bundles of spindle cells in an abundant fibrous stroma. Histologically, sporadic and FAP-associated desmoids are indistinguishable [19].

Historically, surgery was the management of choice for intra-abdominal desmoid tumors with multiple single center case series reporting a local recurrence of up to 20% at 5-year follow-up.

Surgical resection with negative margins has usually been the first choice of management. However, guidelines are changing into a more conservative approach with close observation in asymptomatic cases with no impaired or threatened function [19]. Non-surgical management includes non-cytotoxic treatment (e.g. NSAIDs and Tamoxifen), radiotherapy, targeted therapy (Imatinib), and/or cytotoxic chemotherapy. Due to the high recurrence rate of desmoids, close postoperative follow-up is recommended. NCCN guidelines suggest clinical and radiological follow-up every three to six months for two years then yearly [19]. Factors associated with recurrence are extremity location, young age, and large tumor size [20].

In the reported case, association with HNPCC may be coincidental and sporadic, however, a still unknown genetic predisposition may be a possibility.

In conclusion, desmoid tumors should be considered in the differential diagnosis of patients with HNPCC presenting with findings suggestive of local recurrence after surgery.

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