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Yield of colonoscopy in identification of newly diagnosed desmoid-type fibromatosis with underlying familial adenomatous polyposis

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Abstract

Background: Desmoid-type fibromatosis can arise in patients with familial adenomatous polyposis (FAP), so patients with desmoids often undergo colonoscopy to rule out FAP. Because finding FAP is uncommon, we sought to define subsets of desmoid patients in whom colonoscopy frequently identified FAP.

Methods: Patients with desmoid-type fibromatosis were identified from surgery and pathology databases at a single institution, and information on colonoscopy and FAP diagnosis was collected retrospectively. *CTNNB1* mutation status was defined by Sanger sequencing and digital PCR of archived specimens.

Results: Among 626 patients with desmoids, 26 were diagnosed with FAP. In 20 patients, FAP diagnosis predated the desmoid diagnosis. Among patients without prior FAP diagnosis, 161 underwent colonoscopy, which identified only 6 cases of FAP (diagnostic yield 3.7%). Yields were substantially higher among patients with 4 characteristics: age <40 years (11% yield), intraabdominal or retroperitoneal tumors (5.4%), multifocal disease (29%), and family history (8%) (all $p<0.001$). All cases of FAP were detected in patients younger than 40 years and with at least 1 of the other 3 characteristics. *CTNNB1* mutation status was available in 82 patients with

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known FAP status. None of the 61 patients with *CTNNB1* mutations were diagnosed with FAP, while 7 of the 21 patients with no *CTNNB1* mutation detected (24%) were FAP patients.

Conclusion: Patients with desmoid-type fibromatosis and undiagnosed FAP generally have multiple risk factors, which may be used to selectively recommend colonoscopic screening. Routine *CTNNB1* sequencing may also rule out FAP and allow for deferral of colonoscopy.

Keywords

Desmoid-type fibromatosis; familial adenomatous polyposis; colonoscopy

Introduction

Desmoid-type fibromatosis, also called desmoid tumor, is a rare, mesenchymal tumor diagnosed in approximately 1200 patients in the US each year.[1, 2] The lesions lack metastatic potential, but they can be locally aggressive causing substantial morbidity. Over 90% of desmoids bear mutations in the *CTNNB1* gene, which encodes the β-catenin signaling molecule.[3, 4] These mutations can confirm diagnosis, and are widely screened for in mutation panels employed at cancer centers in the US. *CTNNB1* mutations prevent degradation of the protein, enhancing its transport to the nucleus, where it promotes transcription of oncogenic targets. A minority of desmoids lack the *CTNNB1* mutation but instead have inactivation of APC, a protein that promotes degradation of β-catenin; APC inactivation similarly results in nuclear localization of β-catenin and transcription of oncogenes.[3, 5–7]

While most desmoids are sporadic, a small proportion arise in the context of familial adenomatous polyposis (FAP), caused by germline *APC* mutation.[2, 3, 8] Patients with FAP can develop hundreds to thousands of adenomatous polyps, leading to a nearly 100% lifetime risk of colorectal cancer.[9] Furthermore, desmoid patients with FAP often have a spectrum of extra-colonic manifestations, including duodenal cancer, osteoma, and sebaceous cysts.[10] This constellation of findings is referred to as Gardner’s syndrome. In FAP and Gardner’s syndrome, prophylactic colectomy and upper gastrointestinal tract evaluation can minimize the risk of death from FAP-associated cancers; therefore, patients with underlying heritable disease should be identified.[10, 11]

Current NCCN guidelines suggest that colonoscopy should be considered for patients with a newly diagnosed desmoid-type fibromatosis; however, while 7.5–15% of all desmoids occur in the context of FAP, most often FAP is diagnosed before the desmoid.[2] More than 75% of all FAP patients are diagnosed early due to family history, and many desmoids in FAP patients arise following—and may be induced by—a prophylactic colectomy.[10, 12, 13] Therefore, we sought to test the hypothesis that routine screening in patients with a new desmoid diagnosis would infrequently identify patients with undiagnosed FAP. In addition, patients with FAP-associated desmoids are more commonly male and present with intraabdominal disease;[2, 14] therefore, we analyzed these and other possible risk factors to define subgroups in which colonoscopy would have higher yield. Our third goal was to determine whether routine mutational analysis of desmoid tumors could rule out underlying FAP and obviate colonoscopy in low-risk patients.

Methods

Patient cohort

This retrospective review was approved by the Memorial Sloan Kettering Cancer Center institutional review board. Patients with desmoid-type fibromatosis undergoing surgical resection between July 1, 1982, and June 1, 2015, were identified from a prospectively maintained soft tissue sarcoma database that includes clinicopathologic information. Patients treated non-operatively (with systemic therapy or active observation) were identified from an institutional database of pathologic specimens evaluated between July 1, 2007, and July 1, 2015, and clinicopathologic information was collected by retrospective chart review. For all patients, the retrospective review included colonoscopic reports, family history, history of previous pregnancy, and history of surgery or trauma in the region of the desmoid. Tumors in the shoulder, axilla, and buttock were considered to be localized to the extremity. Abdominal wall lesions were considered distinct from GI/intraabdominal tumors. Age was defined at the time of desmoid diagnosis. Size was defined at the time of presentation with primary disease (largest diameter on radiographic evaluation). Prior pregnancy was defined as having occurred within the 5 years prior to initial presentation, and prior trauma as any trauma in the patient's medical history localized to the region of the tumor. First degree family members were considered parents, children or siblings of the index patient, second degree as first cousins, aunts or uncles, or grandparents. Positive medical history was defined as diagnosis of colorectal cancer, FAP, or colon polyps.

Mutation status

Resected tumors, flash frozen at the time of surgery or recovered from FFPE blocks, were evaluated using cryomolds or H&E staining to ensure specimens contained >90% tumor tissue.[4] *CTNNB1* mutation status was determined using bidirectional Sanger sequencing with exon 3-directed primers, whole exome sequencing, and Illumina miSeq, as previously described,[4] or droplet digital PCR (ddPCR). To complete PCR analysis, custom assays specific for the detection of *CTNNB1* mutations T41A, S45F, and S45P were designed (Biorad). All reactions were performed on a QX200 ddPCR system (Biorad) in technical duplicates. PCR reactions containing primers and probes, DNA, and digital PCR Supermix for probes (no dUTP) were partitioned into a median of ~16,000 droplets per well using the QX200 droplet generator. Emulsified PCRs were run on a 96-well thermal cycler at 95 °C for 10 minutes; 40 cycles of 94 °C each 30 min, 55 °C for 1 min and 98 °C for 10 min. Plates were analyzed using QuantaSoft software. The assay threshold sensitivity was set at 2 mutant droplets.

Statistical analysis

Statistical differences in clinicopathologic characteristics between groups were determined using Fisher's test or Pearson's chi-squared test. Odds ratios were calculated using logistic regression.

Results

Patient and tumor characteristics

The study cohort included 652 patients treated at MSKCC (Table 1). Five hundred twenty (80%) were treated with surgical resection, and 132 (20%) were managed non-operatively with active observation, medical management, and/or radiation. Median follow-up for all patients after desmoid diagnosis was 63 months (range 0–421 months). As in prior reports, desmoid patients were commonly female (n=432; 66%) and young (median age 37 years, range 8–83). Tumors were most commonly diagnosed in the extremity (n=222; 34%), followed by the intraabdominal (including retroperitoneal) compartments (n=145; 22%), abdominal wall (n=134; 21%), and chest wall (n=101; 15%). Twenty-nine percent of all desmoids were identified after prior surgery or documented trauma affecting the site (46% of intra-abdominal desmoids and 10% of extremity desmoids). One hundred six tumors (16%) were diagnosed in patients with documented pregnancy within the 5 preceding years; among women with abdominal wall desmoids, recent pregnancy was noted in 61 of 111 (55%).

Clinicopathologic characteristics of FAP-associated desmoids

Of the 652 patients, 26 (4%) patients had a diagnosis of FAP. Patients with FAP, compared to those with sporadic desmoids, were significantly younger (median age 26 vs. 38 years; $p<0.001$) and had larger desmoids at diagnosis (mean 14 vs. 8.6 cm; $p=0.041$). Patients with FAP also had significantly higher rates of intraabdominal tumors (54% vs. 21%), multifocal disease (62% vs. 2.6%), family history of colorectal cancer in first-degree relatives (46% vs. 5.6%), and prior trauma at the site of the desmoid (65% vs. 28%) ($p<0.001$ for all 4 characteristics; Table 1). The prior traumas among patients with FAP consisted of 16 colectomies and 1 Caesarean section. Patients with FAP, unlike those with sporadic disease, were balanced in gender (50% were male vs. 33% of patients with sporadic desmoids; $p=0.09$; Table 1). The groups with and without FAP did not differ significantly in the rate of recent pregnancy, even when considering female patients alone ($p=0.52$).

Diagnostic yield for FAP of colonoscopy in patients with desmoid-type fibromatosis

Of 26 cases of FAP in this cohort, only 6 were diagnosed after primary presentation with desmoid (0.9% of the 632 patients without prior FAP diagnosis) (Supplementary Figure 1). Of all 632 patients who presented with desmoid fibromatosis and no personal history of FAP, 161 (25%) had documented colonoscopy to test for FAP. Patients with no prior history of FAP were more likely to have documentation of colonoscopy if they had any of several clinicopathologic characteristics associated with FAP (as defined above) (Table 2). Specifically, patients who underwent colonoscopy were more like to be male ($p=0.015$) and to have intraabdominal tumors ($p<0.001$), multifocal tumors ($p=0.001$), and family history of colonic neoplasms ($p<0.001$). Interestingly, however, older patients underwent colonoscopy more frequently than those younger than 40 years old (37% vs. 16%; $p<0.001$).

Despite the fact that patients selected for colonoscopy tended to have risk factors for FAP, the overall diagnostic yield of colonoscopy was low; only 6 patients of 161 (3.7%) with documented colonoscopy were found to have underlying FAP. We reviewed records of all other patients, including those who did not undergo documented colonoscopy, to identify a

later diagnosis of FAP (e.g., upon presentation of second neoplasm) but found none. The diagnostic yield differed strongly among groups stratified by clinicopathologic characteristics (Table 3). Colonoscopy resulted in FAP diagnosis in 6 (11%) of the 55 patients younger than 40 years at desmoid diagnosis versus 0% of the 106 patients over 40 years old (odds ratio 2.8×10^8). Colonoscopy resulted in FAP diagnosis in 29% of the 14 patients with multifocal primaries (odds ratio 29) and in 8% of the 38 patients with unknown family history or first-degree relatives with colorectal neoplasms (odds ratio 3.3). When considering these 4 characteristics (age <40, intra-abdominal location, multiple primaries, and unknown family history or primary relative with colorectal neoplasm) as risk factors for underlying FAP, none of the 109 patients with zero or one of these factors had FAP diagnosed on colonoscopy. Risk correlated with increasing number of these factors, with FAP diagnosed in 2 of 44 patients (4.5%) with 2 risk factors, 3 of 7 (43%) with 3 factors, and 1 of 1 (100%) with 4 factors ($p<0.001$).

Potential risk factors of the 6 patients diagnosed with FAP after colonoscopy are shown in Table 4. All patients were under age 40 at diagnosis, 4 had multiple primaries, 2 had a family history of colon cancer, and one had an unknown paternal family history.

Role of CTNNB1 mutation status in determining the risk of FAP

Since FAP is caused by a germline *APC* mutation, which has the same effect as a *CTNNB1* activating mutation, we hypothesized that the presence of a *CTNNB1* mutation in a desmoid would indicate absence of underlying FAP. Mutation status was available for 82 patients whose FAP status was known, 7 of them with FAP. Mutations in *CTNNB1* were detected in 61 of these patients (74%). Forty-four tumors had T41A mutations (59%), and 13% S45F, 8% S45P, and 1.3% had other mutations (Table 5). No patients with an identified *CTNNB1* mutation were found to have FAP, but 7 of the 21 patients (33%) without detected mutation were found to have FAP (Table 5).

Discussion

Though colonoscopy is often recommended to rule out FAP in patients with newly diagnosed desmoid tumors, FAP rarely presents initially with a desmoid, suggesting that the diagnostic yield of screening colonoscopy in this population is low. The goal of this study was to determine which patients presenting *de novo* with a desmoid tumor require endoscopic evaluation. We identified several characteristics associated with greater likelihood of subsequent FAP diagnosis upon colonoscopy: age <40 years, multifocal disease, and unknown family history or first-degree relatives with colorectal neoplasms. Risk correlated with an increasing number of these factors and with GI/intraabdominal tumors.

We observed a diagnostic yield of 3.7% for colonoscopy among the 161 patients who underwent screening for FAP. This is in line with other, smaller studies of 96 and 63 patients screened for FAP by colonoscopy and sigmoidoscopy/barium enema, respectively, in which 4.8 and 1.3% of patients were diagnosed with FAP.[1, 15] These percentages are lower than the proportion of all desmoids associated with FAP (7.5–15%),[2, 3, 16] likely because most patients with FAP-related desmoids are diagnosed with colorectal disease before presenting with a desmoid tumor or are members of a known FAP kindred.[13, 17, 18]

This study is subject to the limitations of retrospective analysis, including the fact that variable follow-up could have masked later diagnosis of underlying FAP. In addition, colonoscopy could fail to identify extracolonic manifestations of Gardner's syndrome or the limited manifestations of attenuated FAP in younger patients.[19] However, inevitable selection biases may also strengthen our conclusions. Patients with multifocal and intraabdominal disease or with family history of colorectal cancer were more likely to undergo colonoscopy. Both of these factors are associated with FAP, so the true prevalence of FAP in the whole patient population given the population was potentially enriched for high-risk patients at baseline, may be lower than 3.7%.

Clinical risk factors for FAP are easily identified during initial work-up of a desmoid. Increasingly, however, evaluation of the tumors also includes routine *CTNNB1* testing as part of next generation sequencing protocols employed at many major sarcoma centers or as a means to provide diagnostic or prognostic information. [20] *CTNNB1* mutations and *APC* abnormalities associated with FAP should be mutually exclusive and the presence of a mutation has the potential to provide further reassurance, in addition to absence of clinical risk factors, that a patient does not have the hereditary condition. In fact, we found that in addition to the clinicopathologic factors listed above, *CTNNB1* mutation excluded diagnosis of FAP. This finding has not been rigorously analyzed in the literature to date, and suggests that in settings where mutation screening is routine, detection of a *CTNNB1* alteration may obviate the need to perform the more invasive colonoscopy in many patients. Because we found a greater incidence of FAP among patients without a *CTNNB1* mutation, the absence of *CTNNB1* defect on next-generation sequencing in addition to multiple risk factors, could be considered a clear indication for colonoscopy (or germline testing) to rule out a germline defect. At treatment centers without access to routine sequencing or other means of *CTNNB1* mutation testing, selectively recommending colonoscopy to patients with more than one risk factor (age <40 years, intraabdominal or multifocal lesions, and positive or unknown family history of colorectal cancer) could eliminate the need for early colonoscopy in patients at low risk of FAP.

Supplementary Material

Refer to Web version on PubMed Central for supplementary material.

Acknowledgments

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Synopsis

Patients newly diagnosed with desmoids routinely undergo colonoscopy to screen for underlying familial adenomatous polyposis (FAP), but we found the diagnostic yield to be low. However, clinical characteristics and *CTNNB1* mutation status can identify patients at elevated risk of FAP.

Table 1.
Characteristics of patients with sporadic desmoid and FAP-associated desmoid.

Values are number (% within column).

	All patients n=652	Sporadic disease n=626	FAP n=26	p-value[a]
Gender				0.09
Female	432 (66)	419 (67)	13 (50)	
Male	220 (34)	207 (33)	13 (50)	
Age				<0.001
<40	362 (56)	338 (54)	24 (92)	
≥40	290 (44)	288 (46)	2 (7.7)	
Location				<0.001
Intra-abdominal	145 (22)	131 (21)	14 (54)	
Other	507 (78)	495 (79)	12 (46)	
Size (primary)				0.041
≤5 cm	199 (31)	195 (31)	4 (15)	
6–10 cm	276 (42)	267 (43)	12 (46)	
>10 cm	154 (24)	142 (23)	9 (35)	
Unknown	23 (4)	22 (2.3)	1 (3.8)	
Prior trauma or surgery	192 (29)	175 (28)	17 (65)	<0.001
Pregnancy <5 years prior	106 (16)	102 (16)	4 (15)	0.178
Multiple primaries				<0.001
No	603 (92)	593 (95)	10 (38)	
Yes	32 (5)	16 (2.6)	16 (62)	
Unknown	17 (3)	17 (2.7)	0 (0)	
Positive family history[b]				<0.001
1st degree	46 (7)	35 (5.6)	12 (46)	
2nd degree	28 (4)	28 (4.5)	0 (0)	
none	427 (65)	420 (67)	10 (38)	
Unknown	151 (23)	143 (23)	4 (15)	

FAP, familial adenomatous polyposis; GI, gastrointestinal

[a] p-value from Fisher's test or Pearson's chi-square test

[b] History of colorectal cancer

Table 2.
Characteristics of desmoid patients with vs. without colonoscopy.

Values are number (% within row).

	All patients n=632	No colonoscopy n=471	Colonoscopy n=161	p-value[a]
Gender				0.015
Female	423	328 (78)	95 (22)	
Male	209	143 (68)	66 (32)	
Age				<0.001
<40	344	289 (84)	55 (16)	
≥40	288	182 (63)	106 (37)	
Location				<0.001
Intra-abdominal	135	61 (45)	74 (55)	
Other	497	410 (82)	87 (18)	
Size (primary)				0.48
≤5 cm	196	140 (71)	56 (29)	
6–10 cm	268	206 (77)	62 (23)	
>10 cm	146	107 (73)	39 (27)	
Unknown	22	18 (82)	4 (18)	
Prior trauma or surgery	176	115 (65)	61 (35)	<0.001
Pregnancy <5 years prior	101	82 (81)	19 (19)	0.029
Multiple primaries				0.001
No	595	448 (75)	147 (25)	
Yes	20	6 (30)	14 (70)	
Unknown	17	17 (100)	0 (0)	
Positive family history				<0.001
1st degree	37	12 (32)	25 (68)	
2nd degree	28	19 (68)	9 (32)	
None	423	309 (73)	114 (27)	
Unknown	144	131 (91)	13 (9.0)	

[a] p-value from Fisher's test or Pearson's chi-square test

Table 3:
Yield of colonoscopy according to risk factors.

Values are number (% within row).

	Total	No FAP (n=155)	FAP (n=6)	p-value
Age				<0.001
< 40	55	49 (89)	6 (11)	
≥ 40	106	106 (100)	0 (0)	
Location (primary)				0.41
Intra-abdominal	74	70 (95)	4 (5.4)	
Other	87	85 (98)	2 (2.3)	
Multiple primaries				<0.001
No	147	145 (99)	2 (1.4)	
Yes	14	10 (71)	4 (29)	
Positive family history				0.31
1st degree	25	23 (92)	2 (8.0)	
2nd degree	9	9 (100)	0 (0)	
None	114	111 (97)	3 (2.6)	
Unknown	13	12 (92)	1 (7.6)	
Number of risk factors[a]				<0.001
0	40	40 (100)	0 (0)	
1	69	69 (100)	0 (0)	
2	44	42 (95)	2 (4.5)	
3	7	4 (57)	3 (43)	
4	1	0 (0)	1 (100)	

[a]Risk factors were age <40 years, GI/intraabdominal tumor, multiple primaries, and 1st degree relative or unknown family history

Table 4:

Characteristics of patients diagnosed with FAP after desmoid presentation

Gender	Age	Family history	Multiple primaries	Location
Male	29	Mother, colon cancer (age 42)	Yes	Intra-abdominal
Female	18	No	Yes	Intra-abdominal
Male	13	No	Yes	Mandible
Female	28	Unknown on father's side	Yes	Abdominal wall
Female	35	No	No	Intra-abdominal
Female	36	Mother, colon cancer (age 28)	No	Intra-abdominal

Table 5:

Mutation status and yield of colonoscopy

		Documented FAP Status* (n=82)	
***CTNNB1* mutation**	**Total (n=286)**	**No FAP (n=75)**	**FAP (n=7)**
T41A	144 (50)	44 (59)	0
S45F	62 (23)	10 (13)	0
S45P	21 (7.3)	6 (8)	0
Other/multiple	3 (1.0)	1 (1.3)	0
Wild type	56 (20)	14 (19)	7 (100)

* Known history of FAP, documented colonoscopy, or pathologic evaluation of resected colon.