Diffuse Filiform Polyposis With Unique Histology Mimicking Familial Adenomatous Polyposis in a Patient Without Inflammatory Bowel Disease

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- Filiform polyposis is an uncommon entity that is most often encountered in the colon of patients with a history of inflammatory bowel disease (IBD). Filiform polyposis is characterized by a large number of “wormlike” polyps lined by histologically normal colonic mucosa. These polyps can mimic adenomatous polyps. Only rare cases without a history or evidence of IBD have been reported. Neuromuscular and vascular hamartoma of the small bowel is a rare, focal disorder characterized by disorganized smooth muscle fascicles throughout the submucosa accompanied by fibrosis, nerve fibers, ganglion cells, and vessels. To our knowledge, there is only one report of this lesion in the large bowel (cecum), where it presented as a mass. Here we report the case of a 50-year-old man with no known history or symptoms of IBD presenting with filiform polyposis involving the entire colon, clinically mimicking familial adenomatous polyposis, and showing histologic features similar to neuromuscular and vascular hamartoma of the small bowel.

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REPORT OF A CASE

A 50-year-old white man presented for a routine checkup with a medical history significant for obese body habitus, hypercholesterolemia, and sleep apnea. His surgical history was significant for appendectomy at age 8 years and arthroscopic repair of a torn meniscus a year prior. There was a questionable family history of colon cancer in a paternal aunt at age 89 years. His mother had “a few polyps,” but he reported recent normal colonoscopies of his son, age 25 years, and sister, age 54 years. His 2 other children, ages 15 and 12 years, are healthy. He had no personal history of colon polyps, colon cancer, or inflammatory bowel disease (IBD). His appetite had not changed; indeed, he had recently gained a few pounds. He reported no nausea, vomiting, abdominal pain, changes in bowel habits, or alterations in stool size, shape, or consistency. He denied melena and hema-tochezia. His social history was significant for drinking 12 beers on the weekend; he quit smoking tobacco 25 years prior. It was decided that he should undergo a routine screening colonoscopy, which discovered diffuse involvement of the entire length of his colon with numerous polyps of an adenomatous, filiform appearance. Biopsies were not obtained. The patient was diagnosed clinically with an adenomatous polyposis syndrome and was referred to our institution for genetic counseling. Genetic screening for APC gene mutations (for familial adenomatous polyposis) and MYH gene mutations (for MYH-related adenomatous polyposis, a condition with a phenotype similar to familial adenomatous polyposis that inherited in an autosomal recessive manner) revealed no evidence of the mutations most commonly associated with these disorders. Unable to completely exclude malignant transformation of these polyps, the decision was made to undergo a total colectomy. During the operation, it was noted that the entire colon was redundant. The postoperative course was uneventful, and the patient was discharged on the sixth postoperative day.

A 143-cm length of colon, including the anus and a 2.6-cm segment of terminal ileum, was submitted for pathologic examination. Macroscopically, the entire length of the large intestine was involved with more than 100 filiform polyps ranging in size from 3.5 × 1.0 cm to 0.2 × 0.1 cm (Figure, A). The mucosa was otherwise tan, with normal folds and without apparent defects or diverticula; the maximum wall thickness was 0.5 cm. The appendix was surgically absent, and the serosa, adventitia, terminal ileum, and mesentery appeared grossly unremarkable.

The polyps were liberally sampled for histologic evaluation, along with representative sections of grossly normal-appearing large intestine. Microscopically, the filiform polyps were lined by normal mucosa, including normal lamina propria and epithelium. The submucosal cores of some of the polyps were composed of fibrovascular tissue (Figure, B), but there were also several cores where the fibrovascular tissue was admixed with disorganized smooth muscle fascicles, nerves, and ganglion cells (Figure, C and D). No epithelial dysplasia was identified (there was no evidence of tubular adenomas or villous adenomas in any of the submitted sections). In addition, there was no evidence of arborizing smooth muscle or hyperplastic mucosa to suggest Peutz-Jeghers polyps (nor was there clinical evidence of mucocutaneous pigmentation), and there was no evidence of cystically dilated glands or expansion of the lamina propria to suggest juvenile polyps. The uninvolved (nonpolypoid and grossly normal) colon also showed submucosal abnormalities histologically. In random areas throughout the entire colon, a disordered and focally thickened muscularis mucosae merged with a submucosa that was fibrotic and had an increase in disorganized smooth muscle bundles and dilated vessels (Figure, E). Little to none of the loose connective tissue comprising normal submucosa was observed, and the muscularis mucosae often appeared in close approximation to the muscularis propria, with regions of possible direct continuity between the two smooth muscle layers (Figure, F).
A. Gross appearance of the patient's large intestine, showing numerous slender, wormlike filiform polyps. B through F, Microscopic features of the filiform polyps and nonpolypoid colon (all stained with hematoxylin-eosin). B, Tip of a filiform polyp showing normal colonic epithelium overlying a fibrovascular core without significant inflammation (original magnification ×40). C, The tip of another filiform polyp showing submucosal fibrovascular tissue with dilated vessels admixed with disordered smooth muscle fascicles, nerves, and a lymphoid aggregate (original magnification ×40). D, At higher magnification, the disordered smooth muscle bundles (*), nerves (arrowheads), and ganglion cells (arrows) are more evident and are interspersed with fibrous tissue and dilated vessels (original magnification ×100). E, In areas, even the grossly normal, nonpolypoid colon showed abnormal submucosal tissue with a lack of a defined muscularis mucosae and increased fibrosis with interspersed dilated vessels and disorganized smooth muscle bundles (original magnification ×40). MP indicates muscularis propria. F, There were areas where there was marked fibrosis of the submucosa with smooth muscle disarray, making it difficult to determine the location of the normal muscularis mucosae (original magnification ×100). MP indicates muscularis propria.
There was no evidence of acute inflammation, granulomatous inflammation, or chronic mucosal injury, features that would suggest active or inactive IBD. All of the submitted lymph nodes were benign, without granulomatous inflammation or lymphoid hyperplasia.

COMMENT

This is a case of so-called filiform polyposis that endoscopically mimicked an adenomatous polyposis syndrome, since it occurred in a patient with no history or evidence of IBD. In addition, the submucosal cores of these polyps, as well as the submucosa in nonpolyoid areas of the colon, demonstrated an interesting finding of neuromuscular and fibrovascular hyperplasia and disarray. Filiform polyps of the colon are typically considered to be variants of inflammatory pseudopolyps occurring in the setting of IBD.\(^1\) Filiform polyps are long, slender, wormlike or fingerlike projections that can extend up to 9 cm in length and can have occasional “bridging” between adjacent polyps.\(^2\) The polyps can be localized, or they can diffusely involve the colon, and they have been associated with benign strictures.\(^2\) Filiform polyposis has a slight preponderance in men, affecting a range of ages from 6 to 77 years, with an average of 38 years.\(^4\) In some cases, the polyps are difficult to distinguish from villous adenomas, and biopsies are needed to make the diagnosis.\(^2\) Microscopically, the mucosa overlying the filiform polyps ranges from normal to containing nonspecific acute or chronic inflammation,\(^2\) but always without dysplasia. The pathogenesis of filiform polyposis is uncertain, but since the vast majority of cases occur in the setting of IBD, the polyps are generally believed to form as a result of mucosal repair.\(^1,3\)

Since filiform polyposis typically occurs in the setting of IBD (ulcerative colitis and Crohn disease), it is important to realize that rare cases, including ours, have occurred with no prior history or evidence of IBD. There are 2 reports of filiform polyposis occurring in the setting of unusual inflammatory conditions (colonic tuberculosis and histiocytosis X)\(^5,6\) but, to our knowledge, only 3 other reports have noted filiform polyposis without evidence of an underlying inflammatory condition.\(^7-9\) One case, reported in 1989, was that of a 69-year-old woman with no reported history of IBD or colitis who presented with filiform polyps 1 to 2 cm in length within the transverse colon, histologically consisting of normal colon mucosa with “nonspecific inflammatory changes.”\(^7,9,148\) The second case, reported as a letter to the editor in 1989, was that of a 47-year-old Chinese woman who was in good health and presented with rectal bleeding; she was found to have multiple filiform polyps within the cecum and proximal ascending colon.\(^8\) The third case, reported in 1996, was that of a 74-year-old man who was found to have multiple filiform polyps in the distal transverse colon that, histologically, were polypoid ganglioneuromas.\(^9\) There was 1 additional case, in which a single filiform polyp located in the distal colon was described in a 77-year-old man without a previous or subsequent diagnosis of IBD.\(^7\) Our case differs from these reported cases because the filiform polyposis involves the entire length of the colon. As demonstrated by our case, the occurrence of diffuse colonic filiform polyposis in a patient without IBD and with a family history of colon polyps/cancer led the clinicians to determine that this was a case of adenomatous polyposis. Since adenomatous transformation could not be excluded, the patient underwent a total colectomy.

The clinical management of patients with filiform polyposis is variable and should be assessed on a case-by-case basis. Filiform polyps themselves are not considered precancerous.\(^1,2\) However, tight collections of filiform polyps or “giant inflammatory polyps” can mimic adenomatous polyps and even malignancy.\(^4\) While some authors believe that filiform polyposis in and of itself is not an indication to operate\(^2,10\) others, such as Bauknecht et al, have argued that “where there are broad findings, a malignant degeneration cannot be definitely ruled out by means of clinical methods” and “spontaneous involution of the finding cannot be expected.”\(^4,145\) Given the nondysplastic nature of the disorder, the majority of authors have advocated surgical intervention only in those cases in which symptomatic relief is desired, frequently for abdominal pain or severe hemorrhage and anemia.\(^2,9,10\)

In those cases in which filiform polyposis arises in association with active IBD, preemptive surgical resection would seem reasonable, particularly given the increased risk of colon cancer in ulcerative colitis and Crohn disease. Less certain, however, is the management of an asymptomatic patient without this history who is found to have filiform polyposis. The preponderance of cases with associated IBD suggests that these patients may, in fact, have occult IBD. Thus, if a biopsy of the polyps demonstrates a lack of dysplasia, additional biopsies from intervening nonpolypoid mucosa may reveal inactive or minimally active chronic colitis and establish an underlying diagnosis of IBD. However, some cases of filiform polyposis may arise instead from a separate etiology without a known risk for colon cancer; in these cases, filiform polyposis itself may not warrant a preemptive colectomy. Given the dearth of data on the actual rate of malignant transformation, if any, of the filiform polyp, it is difficult to say for certain. However, monitoring for concurrent or subsequent development of adenomatous polyps in a colon afflicted by filiform polyposis would be nearly impossible, and it might put the patient at risk for occult colon cancer arising in an adenoma “camouflaged” in the background polyposis. Ultimately, the onus may be upon the physician and the patient to weigh the risks of preventative colectomy versus the difficulty in predicting malignant transformation based on the available data and/or monitoring for a concurrent or subsequent adenomatous neoplasm if the segment of colon involved by filiform polyposis is not removed.

Since rare cases of filiform polyposis without evidence of IBD or other inflammatory conditions do exist, the pathogenesis of the filiform polyps in this setting (and possibly even filiform polyps occurring in IBD) may not be related to a postinflammatory reparative process. The neuromuscular and fibrovascular hyperplasia/disarray observed in our case is strongly reminiscent of the changes first described by Fernando and McGovern\(^11\) in a disorder they termed neuromuscular and vascular hamartoma of the small bowel. Since that time, there have been additional descriptions of small intestines afflicted with the appearance of bundled smooth muscle seemingly derived from the muscularis mucosae admixed with unmyelinated nerve fibers and ectatic blood vessels in the submucosa, often with some degree of fibrosis.\(^2,12,13\) Some have since challenged the hamartomatous nature of these changes, as several cases have been reported with clear or suggestive changes
of Crohn disease. While the same pattern of neuromuscular and fibrovascular disarray was evident in our case, these changes have been described mainly as localized strictures causing obstruction in the small intestine, with only one reported case of a masslike lesion in the cecum. It is difficult to compare our observation to those in the literature, since many of the microscopic descriptions of filiform polyps focused on the normality of the overlying colonic mucosa, with brief mention of a “fibrovascular core.” From the photomicrographs provided in those reports, it appears that the submucosa beneath the filiform polyps may have been fibrotic, which was confirmed by the microscopic description in one such case. It is interesting to postulate that the case of colonic ganglioneuroma presenting as filiform polyposis may in fact represent a similar hamartomatous process of the colon that was not apparent in those biopsies of the polyps. Since we observed submucosal neuromuscular and fibrovascular disarray in non-polypoid (grossly normal) areas, we postulate that the filiform polyps, at least in our case, represent diffuse hamartomatous replacement of much of the colonic submucosa, producing at first sessile polypoid projections into the lumen that are subsequently pulled to create the elongated filiform polyp. The hamartomatous process may be related to underlying colonic redundancy, as was noted in our patient’s colon. The redundant colon may have caused chronic irritation and mucosal prolapse, which then produced “hamartomatous” changes as a response to the chronic tugging on the mucosa. Although we cannot exclude the possibility that an unknown inflammatory condition led to the production of the polyps and then resolved, an inflammatory etiology seems unlikely in our patient. In light of this patient’s family history of colon polyps and possible colorectal cancer, we also cannot exclude the possibility that a heretofore-undescribed genetic alteration resulted in an inherited form of hamartomatous filiform polyposis.

In summary, this is a case of diffuse colonic filiform polyposis with submucosal neuromuscular and fibrovascular hyperplasia/disarray that mimicked an adenomatous polyposis syndrome in an asymptomatic 50-year-old man. The neuromuscular and fibrovascular disarray noted within the cores of the polyps as well as the submucosa in grossly “normal” colon is reminiscent of neuromuscular and vascular hamartoma of the small bowel. We hypothesize that neuromuscular and fibrovascular hamartomas can form within the submucosa of the colon in some patients, possibly as a response to chronic mucosal prolapse, and can then lead to the formation of filiform polyps.

References