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CASE REPORT

Colonic Xanthomatosis
Relationship to Disordered Motility and Review of the Literature

JAMES SCHEIMAN, MD, GRACE ELTA, MD, THOMAS COLTURI, MD, and TIMOTHY NOSTRANT, MD

KEY WORDS: xanthomatosis; colonic dysmotility.

Although the presence of xanthomas in the stomach has been increasingly recognized through the widespread use of gastrointestinal endoscopy (1), similar lesions in the colonic mucosa have been rarely observed. Romeu and Rybak (2) reported a case of "lipid proctitis" in a patient with diarrhea, tenesmus, and rectal pain who had multiple yellow mucosal papules on sigmoidoscopic examination. They hypothesized that these lesions, which microscopically demonstrated many lipid-laden histiocytes in the lamina propria, led to disordered mucosal function, although direct confirmation was lacking. Beutler et al (3) described a patient with multiple myeloma and disseminated gastrointestinal xanthomatosis who had dysphagia associated with esophageal aperistalsis, nondistensibility and thickening of the gastric mucosa, and rigidity and narrowing of the colon. These lesions, combined with cutaneous sclerosis, produced a syndrome resembling scleroderma. The clinical similarity to gastrointestinal amyloidosis led to the connection between lipid infiltration and disordered gastrointestinal motility.

We report two additional cases of colonic xanthomatosis with confirmation of associated colonic dysmotility.

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From the Section of Gastroenterology, Department of Internal Medicine, University of Michigan Medical Center and Ann Arbor VA Medical Center, Ann Arbor, Michigan.

Address for reprint requests: Dr. Timothy T. Nostrant, University of Michigan Medical Center, 3912 Taubman Center, Ann Arbor, Michigan 48109-0362.

MATERIALS AND METHODS

Case 1. J.V., a 41-year-old woman, was admitted to the University of Michigan Medical Center for evaluation of a 30-lb weight loss and uncontrolled hypertension. She complained of postprandial abdominal bloating, rectal pressure and urgency, flatulence, and frequent small-caliber stools. She had a long history of xanthomatous lesions of the trunk, flexural surfaces, and eyelids. Measurements of cholesterol and triglycerides were normal. She also had a longstanding elevation of serum gamma globulins and a monoclonal spike in serum IgG. A diagnosis of benign monoclonal gammopathy was made on the basis of her long clinical history and a normal bone-marrow examination.

Physical examination was unremarkable except for hypertension, multiple large cutaneous xanthomas and positive stool examinations for occult blood.

A diagnosis of benign normolipemic xanthomatosis was confirmed by normal blood lipid levels and skin biopsy showing planar xanthomas. Because of persistent symptoms and blood in the stools, colonoscopy and upper endoscopy were performed. In the rectosigmoid, multiple yellowish brown plaques were observed (Figure 1), while the duodenum did not have any observed abnormality. Biopsies of both the rectosigmoid and small bowel revealed lipid-laden macrophages within the lamina propria (Figure 2).

Because of uncontrolled hypertension, bilateral renal endarterectomies were performed. Enlarged lymph nodes filled with lipid were seen at laparotomy.

Postoperatively, persistent rectal urgency and tenesmus prompted motility studies of the rectum and left colon. After an overnight fast, rigid sigmoidoscopy was performed for placement of recording clips which were attached to the colonic mucosa at 5 cm and 20 cm from the anal verge. Motor activity was monitored in the left lateral decubitus position. Motor response was recorded with two strain-gauge transducers (Millar Instruments, Houston Texas; model PC 350). Each transducer was mounted on the side of a plastic alligator clip similar to that described by Snape et al (4). Recordings were made on a Gould pen recorder (Gould Inc., Instruments Divi-

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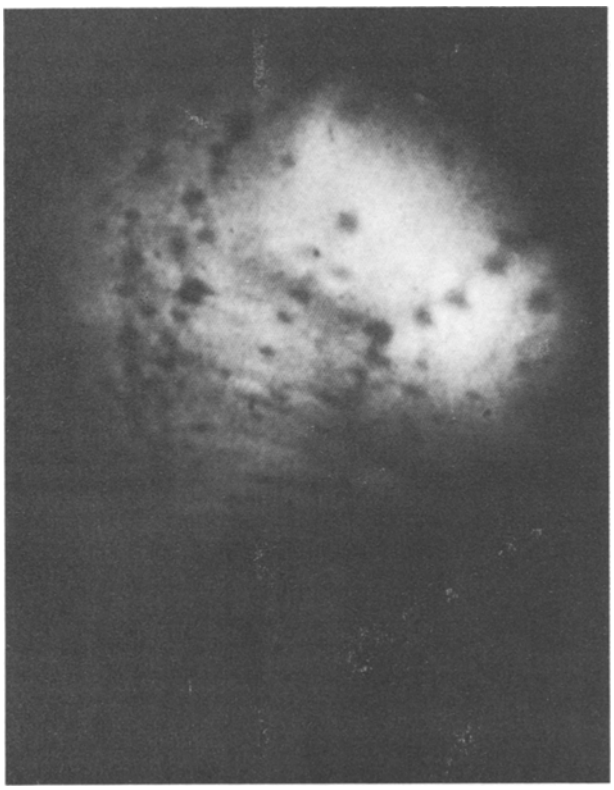

Fig 1. Endoscopic photograph of the rectosigmoid region in case 1. Numerous yellowish-brown plaques are seen distorting the normal mucosal appearance.

sion, Cleveland, Ohio; model 2400, eight channel) and calibrated using a transducer control unit (Millar Instruments; model TC 100). The lower and upper cutoff frequencies for motor activity were 0 and 30 Hz. Basal activity was recorded for 120 min; the patient was given a meal, and contractile activity was recorded for an additional 120 min. The motor recordings was significantly abnormal, with profound baseline hypomotility (quiescense noted 80–90% of basal 2-hr recording) which failed to increase after ingestion of a meal (Figure 3).

Case 2. S.M. is a 29-year-old woman with a 12-year history of chronic active hepatitis.

At the age of 17 she developed fatigue and jaundice. Because of persistent symptoms and elevated transaminases, liver biopsy was performed six months after symptom onset and revealed cirrhosis with mildly active chronic hepatitis. No specific viral or autoimmune markers were found. Prednisone treatment was begun with rapid improvement in symptoms and return of bilirubin and transaminases to normal levels. Repeat liver biopsy six months after initiation of prednisone treatment showed quiescent cirrhosis.

During regular clinic visits, the patient complained of rectal pain, urgency, and blood in the stools. Colonoscopy revealed a 2.5-cm colon polyp at 10 cm, which was removed. In addition, the rectum and rectosigmoid regions were carpeted with yellowish nodules and plaques (Figure 4). Biopsy revealed lipid-filled macrophages in the lamina propria. Serum cholesterol and triglycerides were normal. Symptoms have persisted postpolypectomy and lower gastrointestinal endoscopy continues to show xanthomas. Before rectosigmoid motility studies could be performed, the patient underwent an unsuccessful liver transplant and died.

DISCUSSION

The association of lipid infiltration in the colon and rectal symptoms has been previously reported (2). We have presented two additional cases with confirmation of disordered colonic motility.

The first case is a patient with benign normolipemic xanthomatosis or xanthoma disseminatum. This is a rare dermatologic disorder of unknown cause (5). It is occasionally accompanied by diabetes insipidus, osseous lesions, and lipid infiltration of multiple tissues (5). Abnormal histiocytic proliferation is felt to be the cause of clinical manifestations, based upon the diffuse nature of the lesions and the frequent association with other reticuloendothelial neoplasms, particularly multiple myeloma (6). The pattern of motility seen in this case is similar to that documented in patients with progressive systemic sclerosis where the basal colonic contractile activity is markedly decreased and there is no postprandial activity increase when compared to normal subjects (7). This activity pattern is also different from that seen in patients with irritable bowel syndrome who demonstrate increased 3 cycle/minute slow-wave activity and have increased postprandial contractile activity (6). The location of the plaques and the disordered motility makes lipid infiltration the most likely cause for the rectal symptoms. This is the first case of colonic lipid infiltration with coexistent dysmotility.

Our second patient had extensive distal colonic xanthomatosis and rectal symptoms. Since these symptoms persisted despite removal of the colon polyp, the lipid infiltration appears to be the most likely cause for her symptoms. We have previously reported cases of gastric xanthomatosis associated with liver disease, but in these cases, no colonic xanthomas were found (1). Gastrointestinal xanthomatosis has also been reported in patients with hypercholesterolemia and hypertriglyceridemia (1). Patients in this report had normal serum levels of both cholesterol and triglycerides.

Previous reports of gastrointestinal xanthomatosis have hypothesized an association between disordered gastrointestinal function and the extensive lipid infiltration (2, 3). Motility studies were not performed in

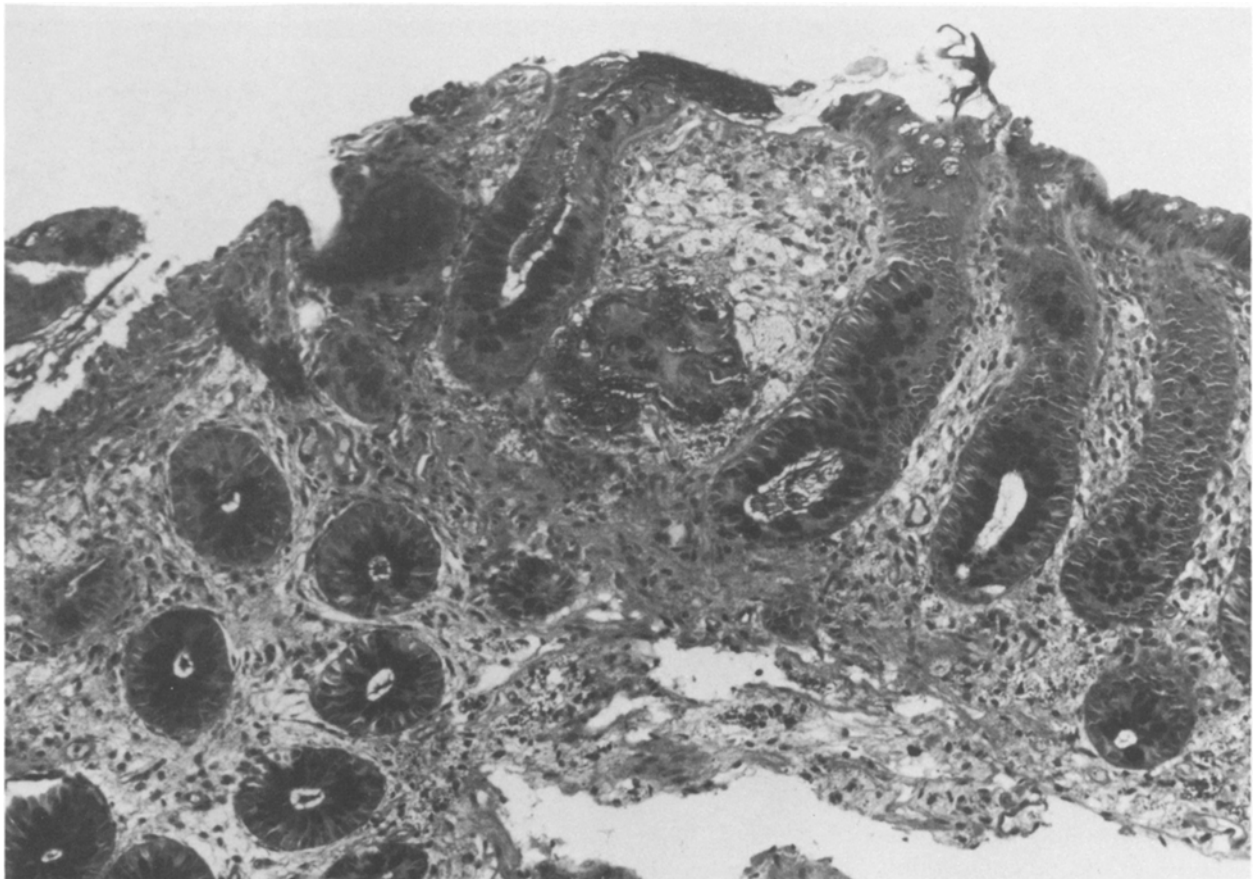

Fig 2. Histology of the lesions observed in case 1. Numerous lipid-laden macrophages are seen within the lamina propria. A large aggregate is evident near the center of the field (PAS, ×200).

these patients. Radiographic documentation of esophageal dysmotility and narrowing and nondistensibility of the stomach and colon in the patient with multiple myeloma (3) lends support to the contention that lipid-laden macrophages infiltrating the gastrointestinal tract can produce intestinal dysmotility. Similar to other infiltrative disorders such as amyloidosis and scleroderma, these lesions may disrupt normal motility through interference with nerve conduction, muscle contraction, or both. Patients with upper or lower abdominal complaints and associated colonic or gastric xanthomatosis should have gastric and intestinal motility studies to further elucidate the cause for their symptoms.

SUMMARY

We report two additional cases of colonic xanthomatosis associated with persistent rectal symptoms.

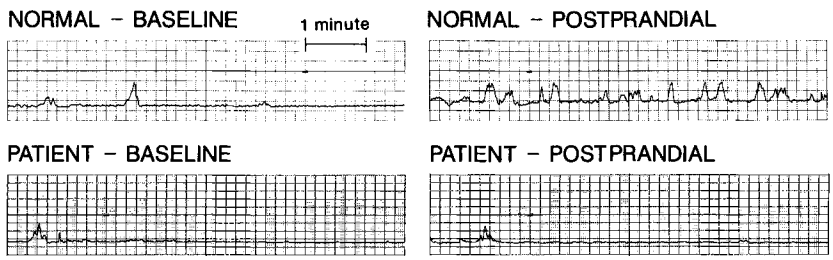

Fig 3. Rectosigmoid motility tracing from case 1. A normal tracing is provided for comparison. The patient demonstrates profound baseline hypomotility that failed to increase postprandially.

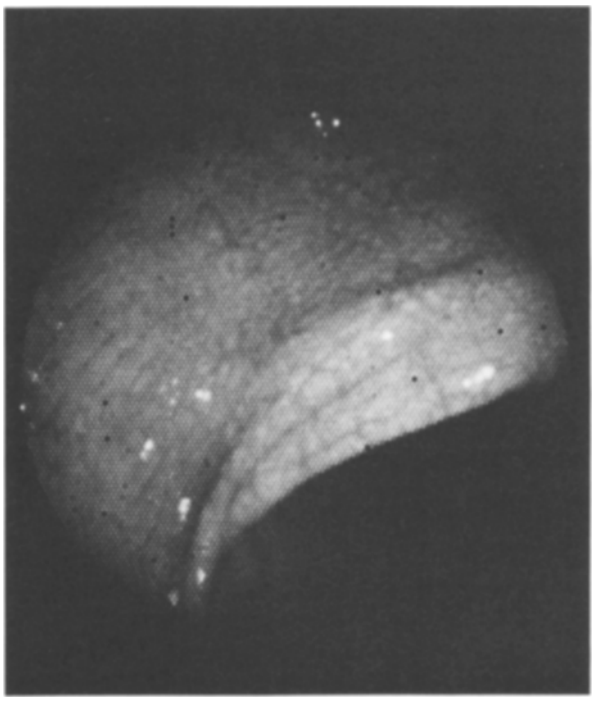

Fig 4. Endoscopic photograph of the rectosigmoid region in case 2. The normal mucosa is carpeted with yellowish nodules and plaques.

Disordered colonic motility in the areas of lipid infiltration was documented in one patient. We conclude these lesions may have a pathophysiologic role in the alteration of intestinal motility which appears to be the cause of our patients' symptoms.

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REFERENCES

1. Coates AG, Nostrant TT, Wilson JAP, Dobbins, WO, Agha F: Gastric xantho matosis and cholestasis: A causal relationship. Dig Dis Sci 31:925–928, 1986
2. Romeu J, Rybak B: Lipid proctitis. N Engl J Med 301:1099–1100, 1979
3. Beutler SM, Fretzin DF, Jao J, Desser R: Xanthomatosis resembling scleroderma in multiple myeloma. Arch Pathol Lab Med 102:567–571, 1978
4. Snape WJ, Carlson GM, Cohen S: Colonic myoelectric activity in the irritable bowel syndrome. Gastroenterology 70:326–330, 1976
5. Altman J, Winkelmann RK: Xanthoma disseminatum. Arch Dermatol 86:62–76, 1962
6. Lynch PJ, Winkelmann RK: Generalized plane xanthoma and systemic disease. Arch Dermatol 93:639–646, 1966
7. Snape WJ: Colonic motility in health and disease. *In* Functional Disorders of the Digestive Tract. WY Chey (ed). New York, Raven Press, 1983, pp 287–291