Refractory ulcerative chronic pouchitis in a patient with Gardner syndrome following restorative proctocolectomy with ileal pouch-anal anastomosis

Dear Editor,

Gardner syndrome (GS) is a rare autosomal dominant inherited disorder with a high degree of penetrance characterized by intestinal polyposis, bone and soft-tissue tumors, including osteoma, epidermal inclusion cyst, lipoma, fibroma, gastric and duodenal polyposis and desmoid fibromatosis. In 100% of patients with GS untreated, cancer develops in the large intestine before the age of 40 years. Hence, prophylactic colectomy is indicated (1).

The restorative proctocolectomy in combination with ileal pouch-anal anastomosis (RPC/IPAA) is the best operation for GS patients because it not only resects all large intestine mucous membranes to avoid carcinogenesis, but also preserves the intestine function and sex ability and avoids colostomy, thus improving the quality of life of patients (2).

We present the case of a patient with GS treated with restorative proctocolectomy with ileal pouch-anal anastomosis with poor post-operative outcome, who developed severe ulcerative chronic pouchitis refractory to treatment.

Case report

Woman aged 51 years, with a family history of colon cancer, diagnosed with Gardner syndrome (colonic adenomas detected at the age of 28, gastric polyposis, cranial osteomas, suprarenal adenomas and muscular fibrolipomas). The genetic study, performed on two occasions, did not detect mutations in the APC gene.

In 2001, at the age of 41, given the rapid increase in number and size of the colonic adenomas, a total proctocolectomy and ileal J-pouch-anal anastomosis were performed in two stages.

Following radical surgery of the rectum and colon, the patient began presenting periodic and intermittent episodes of hypogastric pain, blood-streaked diarrhea, pain and severe anal itching with recurrent fissures, at times coinciding with the menstrual cycle.

The various ileoscopies performed showed signs of inflammation and small ulcerations in the pouch, which were increasingly more extensive and confluent, until becoming a giant ulcer (Fig. 1); biopsies and serology ruled out CMV infection. Endoscopic lesions and clinical manifestations did not respond appropriately to the various treatments tested (rofecoxib; metronidazole + ciprofloxacin; rifaximin; VSL#3 probiotic; oral and topical 5-ASA; topical steroids). Notwithstanding our advice, the patient refused an ileostomy on several occasions. None of the check-ups performed by means of a conventional endoscopy (we do not have a pancho-moscopy at our disposal) detected new adenomas in the pouch.

Following surgery, every two years, a duodenoscopy by means of a side-viewing video endoscope was performed.

In addition, the gastric polyposis significantly increased in number and size. The histology corresponds to hyperplastic lesions.

Discussion

Endoscopies constitute a fundamental tool in the monitoring and management of patients with Gardner syndrome and ileal
pouch outcomes following surgery. They also allow for resection and monitoring of both gastric polyps and periampullary lesions.

The incidence of pouchitis after ileal J-pouch anal anastomosis is approximately 50%, with two thirds of these patients having multiple episodes. Chronic pouchitis occurs in a minority of patients, 5.7% in some studies (3).

Recent reports have suggested the mucosa of an ileal reservoir could be at risk of neoplasia. Risk factors may include the age of the pouch, chronic pouchitis, and previous colonic neoplasia. The development of dysplasia in ileal pouches performed for ulcerative colitis is probably a rare (1.7%) event within 15-20 years of pouch surgery (4); however, in the case of Gardner syndrome it could be much higher.

The exact etiology of pouchitis and its pathogenesis remain an enigma. Proposed etiological factors include stasis, mucosal ischemia, bacterial dysbiosis and immune deficiency. It has been postulated that some immunological, metabolic or genetic defect is the common underlying cause for all three pathological conditions. Chronic pouchitis occurs mainly in an intrinsically abnormal pouch and/or a systematically susceptible host (5).

In the case of our patient, we believe the most probable cause of the pouchitis and chronic ulcerations presented may be mucosal hypoperfusion in the pouch, as hypothesized by other authors (6). This would be associated with a significant reduction in the intramucosal pH, which could very well justify the symptoms presented by the patient.

To date, no case similar to that presented has been described in the literature, which concerns ulcerative chronic pouchitis, refractory to every therapy tested, in a patient with Gardner syndrome subjected to a restorative proctocolectomy.

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References