Vacuolar internal anal sphincter myophaty as a rare cause of proctalgia fugax and constipation

Key words: Proctalgia fugax. Constipation. Myopathy. Internal anal sphincter.

Dear Editor,

A 62-year-old female patient with a history of arterial hypertension treated with the combination of trandolapril and verapamil, an anxiety-depressive disorder and a left mastectomy for breast cancer years ago, currently in remission, presented complaining of intense proctalgia mainly at night, with an obstructive defecation syndrome and expulsive constipation of 5 years of evolution. Physical examination revealed a hypertonic anal sphincter and intense pain during rectal palpation. Three-dimensional endoanal ultrasonography showed an enlarged internal anal sphincter (IAS) with more than 4.5 mm of thickness (Fig. 1A). Anorectal manometry showed a hypotonic external anal sphincter (EAS), elevated resting pressure and deficient relaxation of the IAS (with ultraslow waves), and rectal hyposensitivity (Fig. 1B). Initial medical treatment was provided with calcium antagonists of retarded action and β2 adrenergic agonists, with no significant clinical improvement. Surgery consisting of a 1 cm transanal internal sphincter myomectomy and a mucosal plasty was carried out, with satisfactory postoperative recovering. During the following medical visits the patient reported significant clinical improvement. Histological study informed of a hypertrophic IAS myopathy with vacuoles and polyglucosan inclusion bodies or amyloid-like PAS positive bodies (Fig. 1 C and D).

Discussion

Anorectal and perianal pain has been described related to different diseases, most of which are easily recognized (hemorrhoids, perianal sepsis or tumors); however, in many cases no organic cause can be attributed even after performing a meticulous physical examination and the mandatory complementary studies (1).
Proctalgia fugax is one of the entities that may cause chronic anorectal pain. It is an infrequent benign pathology characterized by intense perianal pain, of sudden onset, that is transitory, of short duration presenting in frequent intervals, without a demonstrable organic cause (2). The etiology is uncertain, but it has been associated with muscular spasms of the pelvic floor (3), and structural and functional alterations of the IAS (4,5).

The association between proctalgia fugax and constipation has been well described (6), which is known as the hypertrophic myopathy of the IAS. Patients with hypertrophic myopathy of the IAS have intense, self-limiting anal pain with sudden onset, mostly at night, due to spasmodic contractions of the IAS, which has an increased thickness. Functional constipation is another characteristic symptom (7). This syndrome presents more often as sporadic cases, although it has also been rarely described as a hereditary form (8,9). During the physical exploration the IAS is thickened, which may simulate an anal tumor with no other positive findings. The endoanal ultrasonography shows thickening of the IAS, and the anorectal manometry evidences an elevation of the resting pressure (anal hypertonia), with rhythmic increasing pressures at different levels of the anal canal during the episodes of proctalgia, and also with deficient muscular relaxation. The goal of treatment is to achieve anal sphincter relaxation with topical or systemic muscular relaxing agents, such as nitrates, calcium antagonists, β2 adrenergic agonists or botulinum toxin (10). Cases refractory to medical treatment can benefit from surgical intervention, which consists of a myomectomy with complete section of the fibers of the IAS. The histological findings of hypertrophic smooth muscle fibers with numerous vacuoles that may contain polyglucosan inclusion bodies (6,8) are characteristic of this myopathy.

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References