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Mesenteric Desmoid Tumors in an Adolescent Girl with Familia Adenomatous Polyposis

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Abstract **Case Report**

Desmoid tumors (TD) are rare tumors that infiltrate tissues and have no metastatic potential. They are most often sporadic and in 5 to 10% of cases associated with familial adenomatous polyposes (PAF). They usually appear in the young adult, more rarely in the child. We report the case of a 21-year-old who had undergone total colectomy due to PAF with de novo APC mutation, usually not associated with TD. Three years later, she presented an occlusive sd with vomiting. The CT scan revealed a mesenteric TD, about 20 cm long axis invading the ileo rectal anastomosis. The procedure consisted of a resection of the tumor of the part of the rectum invaded with preparation of a low ileo rectal anastomosis. This observation recalls the importance of regular screening of patients with FAP, especially after surgery, regardless of age and type of mutation. Progress remains to be made to determine the risk factors for developing TD in these subjects and to treat them effectively. Current therapeutic management remains difficult because of the infiltrative and recurrent nature of these tumors and requires a multidisciplinary opinion.

Keywords: Desmoid tumor, adenomatous polyposis, surgery.

INTRODUCTION

Desmoid tumors (TD) are myofibroblastic proliferations developed from fasciae and aponeuroses. They are rare (3 to 4 / million inhabitants / year), slow growth, and can affect all organs [1]. These are aggressive tumors that compress and infiltrate the tissues, but are devoid of metastatic potential. The majority are associated with a mutation of the catenin gene (CTNNB1) and are sporadic (TDS), but in 5 to 10% of cases they remained in the context of familial adenomatous polyposis (PAF) by APC gene mutation [2]. Between 3.5 and 32% of patients with PAF develop TD. Since the prophylactic colectomy, these tumors have become one of the leading causes of death in these patients, their complications and their difficulty in treating [2-4]. We report the case of a young carrier with a complicated PAF of TD.

CASE REPORT

21-year-old patient, having as a history a brother operated in 2014 for PAF. The circumstances of discovery can be traced back to the screening of a PAF after the brother's surgery, the patient has benefited from a colonoscopy showing the presence from the rectum to the cecum, but especially at the level of the left colon of several polyps. sessile and certain planes (at least 40 polyps), whose histopathological examination is in favor of a tubular adenoma with low grade dysplasia. The patient was operated in 2016 having undergone a subtotal colectomy with mechanical ileo-rectal anastomosis. Anatomopathological examination of the piece in favor of a tubular adenoma in low grade dysplasia. After 2 years the patient presented abdominal pains especially hypogastric with recursions of average abundance. At colonoscopy presence at 15 cm from the anal margin of an ulcerated squeezing sessile lesion in the center, 1 cm long axis, presence of the anal margin until the anastomosis of 4 sessile milimetric polyps. Abdominal-pelvic CT showed irregular pelvic thickening of 4 cm * 11 cm in length, associated with irregular thickening of the ileo-rectal anastomosis extending 6 cm in length (Figure-1). The patient was operated on 05/18, in whom the surgical exploration revealed the presence of a large distal mesenteric mass about 20 cm in diameter, taking about 80 cm from the distal small bowel and at the mesenteric level at the anastomotic level. benefited from rectal resection (middle rectum) and hailectomy upstream of

the tumor with preparation of a gastro-rectal anastomosis with ileostomy of protection.

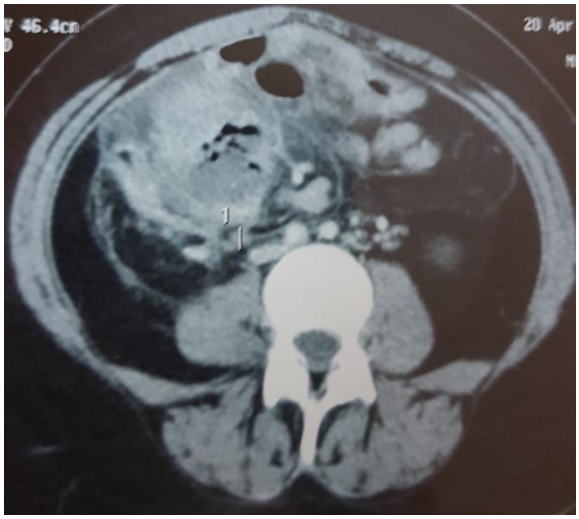

Fig-1: The Mesenteric Demoid Tumor

DISCUSSION

PAF has a prevalence in Europe of 1/11 300 to 37 600. It is an autosomal dominant disease, but 15 to 30% of de novo mutations are described. The APC gene, localized on chromosome 5 in q22, has more than 826 pathogenic variants [5]. Its penetrance, close to 100% for polyps, is variable for other diseases. The first polyps appear at an average age of 16 years (7-36 years) and screening is proposed from 12 years. In the absence of surgery, colonic cancer can appear 10 years after the appearance of the first polyps, and the risk of rectal cancer is 100% after 40 years [5]. Surgical indications include severe dysplasia and profuse PAF. A proctocolectomy is often performed because a surgical revision at the rectal level can be difficult if TD develops [2]. PAF can also be complicated by rarer cancers (duodenum, thyroid, liver, pancreas, bile ducts, brain) [5].

PAs associated with PAFs develop mainly in the mesentery, more rarely in the abdominal wall, thorax, trunk, neck, face and limbs [1, 2]. Gardner syndrome associates PAF, TD, epidermal cysts, osteomas, and dental abnormalities. In our observation, the girl had neither cysts nor dental anomalies. TD usually occurs in young adults (mean age 30 years). In children, PAs associated with PAF usually appear after 14 years of age [3], whereas TDS appear earlier (median age 8 years), but TDs have been observed in young children before PAF is not diagnosed [6]. The main risk factors for the development of PA associated with FAP are surgical trauma and genetic factors. More than 70% of the TD appear as in our observation 2 to 3 years after a colectomy: they could result from an excessive healing process of mesenchymal stem cells [1, 3]. A meta-analysis showed that an APC gene mutation and a family history of TD were independent risk factors [3]. The mutation of the APC or CTNNB1 genes prevents the degradation of a protein, b-catenin, within the Wnt signaling pathway, which activates its function as a target gene transcription factor, favoring the occurrence of tumors [1, 5, 7]. Studies have found a correlation between genotype and risk of TD: some mutations seem to involve a higher risk (codons 1399, 1444, 1310-2011, 1265-2035) but this remains to be clarified [2, 3, 8]. Our patient did not carry any of these mutations (codon 1068). The role of hormonal factors is controversial: women may be more at risk as for TDS [3].

The diagnosis of TD is generally evoked on an ultrasound and accurate on a CT scan or MRI (hyposignal in T2 sequence characteristic of the fibrous tissue). In FAPs, it is recommended to avoid an aggressive surgical biopsy that could accelerate TD growth. A biopsy remains recommended in TDS or in case of doubt with sarcoma [5, 7]. Without treatment, about 80% of TDs grow slowly and stabilize, 5% to 10% regress spontaneously and 10% evolve rapidly, which can lead to intestinal obstruction, mesenteric infarction or venous thrombosis, digestive fistulas, peritonitis, hydronephrosis, nerve compression [1, 7].

The therapeutic management of TD is difficult because of their invasive and recurrent nature: a multidisciplinary opinion is essential. A European consensus for adults in 2014 and the European group for studying soft tissue sarcomas in children in 2009 developed management algorithms [1, 7]. Because of their rarity, few prospective randomized controlled studies have been conducted. Mesenteric TD, like our patient's case, pose many more problems. Several types of surgery are feasible. First of all, in the case of resectable tumors, one can perform an isolated tumor excision or a wide excision most often associated with a visceral sacrifice. According to the studies, simple excision [9], resection margins of at least two centimeters [11] or larger excision with extensive resection of the small intestine are recommended due to the frequent proximity of mesenteric vessels [10]. At

the extreme, cases of total enterectomy with small bowel transplant have been described in the literature [11]. While for some authors, healthy resection margins do not influence the rate of recidivism [9, 12], others have emphasized their need [13]. In addition, partial exceriations seem to lead to an acceleration of tumor growth [14]. However, TD are sometimes infiltrating tumors extending well beyond the palpable tumor limits, making resection often incomplete. Most often tumor excision is delicate and associated with high morbidity. According to the series, the rates of major postoperative complications (occlusion, intra-abdominal abscess, herniae, digestive fistula, short bowel syndrome and postoperative deaths) are 22 to 60% of postoperative mortality [9]. The St Mark's Hospital team reported that the majority of deaths related to abdominal CT occurred postoperatively immediately [13]. Smith *et al.*, [16], in a prospective study of 70 cases showed that there was no significant difference in survival whether or not there was surgical treatment. In addition, the recurrence rate after surgical treatment is high, ranging from 37 to 88% of cases according to the series [10]. In the case of unresectable tumors, digestive bypass can be performed (gastroenteral, enteroenteral, enterocolic, ileostomy or colostomy).

Intra-abdominal TD sometimes complicates a prophylactic surgical procedure necessary for the therapeutic management of polypostes [17]. The existence of a family history of TD leads some authors to modify the management of patients with PAF. For some [18], the existence of cases of TD in a family of polyposis may encourage repelling as long as possible prophylactic colectomy to avoid the occurrence of TD. Church [18] even suggested using less invasive techniques such as laparoscopy to perform prophylactic colectomy. However, this technique has not demonstrated its superiority in preventing the development of TD compared to laparotomy. In families associating PAF and TD or for patients with TD discovered at the time of laparotomy, some authors recommend that a total coloproctectomy be performed immediately with ileoanal anastomosis on reservoir, given the potential difficulties to achieve in a second step resection of the rectum in the presence of a TD if it became necessary [15, 17].

CONCLUSION

This observation recalls the importance of regular screening of patients with FAP, particularly after surgery, regardless of age and type of mutation. Progress remains to be made to determine the risk factors for developing TD in these patients and to treat them effectively (prospective randomized studies). TDs are disabling chronic diseases and the long-term prognosis of our young patient remains uncertain.

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