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The Silent fibrous Legacy of Familial Adenomatous Polyposis: A Desmoid Tumor Unveiled in the Abdominal Wall

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Abstract **Case Report**

Desmoid tumors are rare fibroblastic proliferations with locally aggressive behavior and no metastatic potential. They are frequently associated with genetic mutations, particularly in the context of familial adenomatous polyposis (FAP). We report the case of a 29-year-old woman operated for colonic adenomatous polyposis in 2018, in whom an abdominal parietal mass revealed a desmoid tumor. We describe the clinical features, diagnostic means, including imaging, and therapeutic options, through a review of the literature.

Keywords: Desmoid tumor, Familial adenomatous polyposis (FAP), Abdominal wall mass, Fibroblastic proliferation, Imaging.

INTRODUCTION

Desmoid tumors or aggressive fibromatoses are rare fibroblastic neoplasms, accounting for less than 3% of all soft tissue tumors. Their occurrence is often sporadic, but in 10-20% of cases they are associated with familial adenomatous polyposis (FAP), particularly in its severe form known as Gardner's syndrome. They can affect a variety of sites, but the abdominal wall is a preferred location, particularly in young women, often multiparous or postpartum. Diagnosis is based essentially on imaging and histopathological analysis. Their management remains controversial due to their unpredictable evolution.

OBSERVATION

A 29-year-old female patient with familial adenomatous polyposis coli underwent prophylactic total colectomy at the age of 22. She presented with a painless swelling of the right abdominal wall, which had appeared progressively after the operation. Clinical examination revealed a firm, immobile mass measuring approximately 14 cm long, with no inflammatory signs.

Imaging Resources and Results

Initial ultrasound showed a hypoechoic, heterogeneous, poorly limited mass infiltrating the abdominal musculature *(figure 1)*.

Abdominal-pelvic CT scan revealed a subumbilical, subcutaneous, aponeurotic soft tissue mass resting on the rectus abdominis muscle, oval in shape, circumscribed, regular in outline, spontaneously isodense, enhanced after injection of contrast medium *(figure 2)*.

Abdominal-pelvic MRI revealed a well-individualized mass, isointense on T1, heterogeneously hyperintense on T2 with areas of moderate enhancement after gadolinium injection, with no invasion of intra-abdominal organs.

An ultrasound-guided biopsy confirmed the desmoid nature *(figure 3)*.

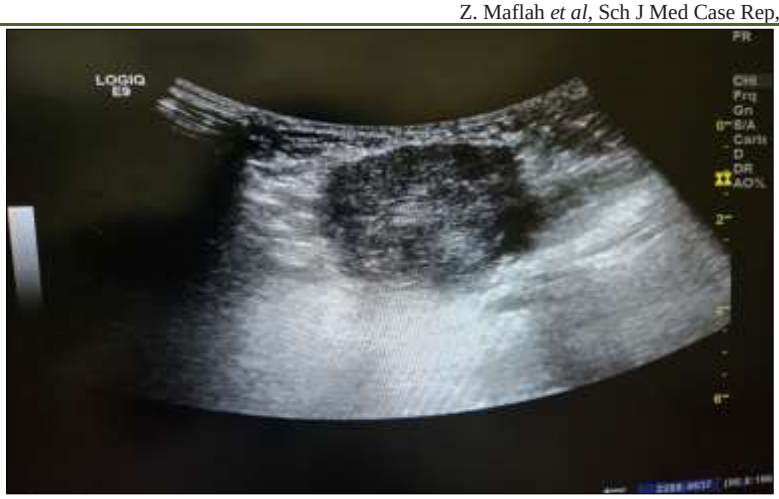


Figure 1: B-mode ultrasound cross-section

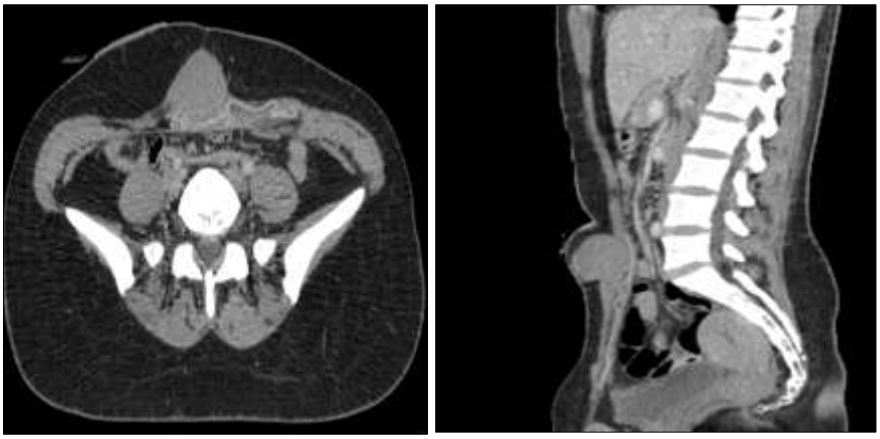
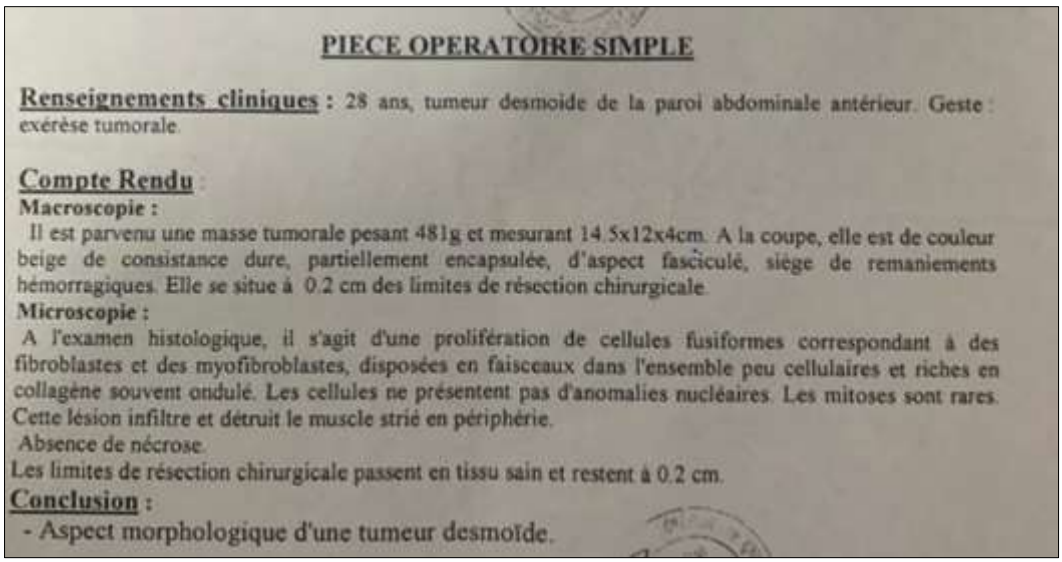


Figure 2: Scannographic image after PDC injection: axial and coronal: Mass of soft tissue below the umbilicus, subcutaneous, supra-aponeurotic, resting on the rectus abdominis muscle on the right side there is also a wrinkled plaque opposite



PIECE OPERATOIRE SIMPLE

Renseignements cliniques : 28 ans, tumeur desmoide de la paroi abdominale antérieur. Geste : exérèse tumorale.

Compte Rendu :

Macroscopie :

Il est parvenu une masse tumorale pesant 481g et mesurant 14.5x12x4cm. A la coupe, elle est de couleur beige de consistance dure, partiellement encapsulée, d'aspect fasciculé, siège de remaniements hémorragiques. Elle se situe à 0.2 cm des limites de résection chirurgicale.

Microscopie :

A l'examen histologique, il s'agit d'une prolifération de cellules fusiformes correspondant à des fibroblastes et des myofibroblastes, disposées en faisceaux dans l'ensemble peu cellulaires et riches en collagène souvent ondulé. Les cellules ne présentent pas d'anomalies nucléaires. Les mitoses sont rares. Cette lésion infiltre et détruit le muscle strié en périphérie.

Absence de nécrose.

Les limites de résection chirurgicale passent en tissu sain et restent à 0.2 cm.

Conclusion :

- Aspect morphologique d'une tumeur desmoïde.

Figure 3: Pathological findings: Morphological appearance of a desmoid tumor

DISCUSSION

Desmoid tumors, also known as aggressive fibromatoses, are benign fibroblastic proliferations with no metastatic potential, but with marked local infiltrative power and a high rate of local recurrence. Their link with familial adenomatous polyposis (FAP) is well established, particularly in Gardner's syndrome, where

mutations in the *APC* gene play a central role in the constitutive activation of the Wnt/β-catenin pathway, leading to uncontrolled fibroblastic proliferation.

In our case, the occurrence of a desmoid tumor in a patient with PAF, several years after prophylactic surgery, is in line with the literature. Indeed, according to Church *et al*., (2003), up to 10-15% of patients with PAF develop desmoid tumours, often within 2-3 years of abdominal surgery. Surgical trauma, hormonal changes and underlying genetic mutations are recognized triggering or aggravating factors.

Radiologically, magnetic resonance imaging (MRI) is currently considered the tool of choice for the diagnosis and follow-up of desmoid tumors, due to its ability to accurately assess locoregional extension and muscular or fascial infiltration. Typical semiological features include:

- T1: iso- or hypointense signal in relation to the muscle;
- T2: variable signal, often heterogeneous, with areas of high intensity reflecting myxoid components, but sometimes also more hypointense fibrous patches
- Sequences after gadolinium: moderate to intense enhancement, often heterogeneous, sometimes in radial or linear bands corresponding to poorly vascularized fibrous trabeculae;
- STIR or fat-sat T2 sequences: very useful for assessing active or inflammatory areas, showing a clear hypersignal in progressive forms.

According to Desai *et al*., (2016), MRI can also be used to classify tumors according to their evolutionary behavior: some remain stable or even regress spontaneously, while others progress rapidly. This justifies the current "wait and see" approach proposed by several reference centers (e.g. Bonvalot *et al*., 2013), which advocate initial active surveillance in the absence of symptomatology or rapid progression.

Abdominal CT is also used, especially preoperatively, to assess topography in relation to adjacent organs, but is less effective than MRI in assessing invasion of muscular or fascial structures.

Ultrasound, although limited for deep or complex lesions, is a good follow-up tool, particularly for superficial masses. It typically shows a hypoechoic, poorly limited mass with irregular contours, sometimes with Doppler vascularization if the lesion is active.

From a therapeutic point of view, the current options are multiple:

- Surgery is no longer systematically performed as first-line treatment, except in the case of a symptomatic or compressive mass, due to the high risk of local recurrence (up to 30-50% according to Mankin *et al*.,).
- Medical treatments include anti-estrogens (tamoxifen), non-steroidal anti-inflammatory drugs (NSAIDs), tyrosine kinase inhibitors (sorafenib, pazopanib) and low-dose chemotherapy (methotrexate + vinblastine).
- Radiotherapy is reserved for inoperable and progressive forms, although it does carry a risk of long-term complications.

Progression is unpredictable, and each case requires discussion at a multidisciplinary consultation meeting. Emphasis is placed on the importance of follow-up imaging, particularly MRI, to detect changes in size, signal or biological behavior (vascularization, areas of necrosis).

Finally, a biopsy is always essential to confirm the diagnosis and rule out a sarcoma or other soft-tissue tumour. Immunohistochemical evidence of nuclear β-catenin positivity is a strong argument in favor of the diagnosis of desmoid tumor, particularly in sporadic forms. In PAF-related forms, a truncated mutation of the *APC* gene is usually found.

CONCLUSION

Desmoid tumors are a rare but important entity to be aware of in patients with PAF. Diagnosis is based mainly on imaging and histology. Management, which is often multidisciplinary, increasingly favors conservative strategies, given the unpredictable evolution of these tumors.

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