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A painful vulvar mass: Bartholin gland cyst or aggressive angiomyxoma of the vulva?

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ABSTRACT

Aggressive angiomyxoma (AAM) is a rare invasive mesenchymal tumor of the pelvis and vulvar region, frequently occurring in women. We present a report in a 44 years old woman with a persistent vulvar mass on the right *labium majus* misdiagnosed as a Bartholin gland cyst.

SOMMARIO

L'angiomixoma aggressivo (AAM) è un raro tumore mesenchimale invasivo del bacino e della regione vulvare, che si verifica frequentemente nelle donne. Presentiamo il caso di una donna di 44 anni con una massa vulvare persistente sul *labium majus* destro diagnosticata erroneamente come una cisti della ghiandola di Bartolini.

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Key words

Aggressive angiomyxoma; Bartholin cyst; mesenchymal tumor; vulvar mass; tumors of uncertain differentiation.

INTRODUCTION

Aggressive angiomyxoma (AAM) is a rare type of soft tissue benign neoplasia, typically arising on the perineal region, which was first described in 1983 by Steeper and Rosai (1).

The term aggressive is used to emphasize the locally aggressive behaviour and the high frequency (40%) of local recurrence (2-4), although it does not reflect a high probability for metastasis, as only 2 cases with metastatic disease have been reported (5, 6). The name angiomyxoma derives from its analogy with myxoma and its vascular component. The etiopathogenesis is unclear, molecular mechanism is unknown. In some cases of AAM chromosomal abnormalities has been reported. The most involved chromosome is 12. Several analyses have shown how abnormalities of chromosomal 12 are associated with rearrangement of HMGIC gene, which is already implicated in the pathogenesis of others mesenchymal tumors (breast fibroadenomas, cutaneous lipomas, endometrial polyps and uterine leiomyoma) (7).

The AAM is commonly immunoreactive for desmine, smooth muscle actin, muscle specific actin, vimentin, estrogen and progesterone receptor, and hyaluronate receptor CD44. AAM generally reveals negativity for S100 protein, MUC4 and CD34. This tumor is distinguished from other mesodermal lesions by these histopathological features (8). The growth is slow and asymptomatic, and it can often be mistaken for a Bartholin Cyst, thus early diagnosis is rarely achieved (9). Most symptoms (feeling of local pressure, pain, dyspareunia) occur when the mass reaches dimensions higher than 8-10 cm (3, 10, 11).

Case report

We hereby present the case of a nulliparous, 44-year-old South-American woman, with no specific medical history, referring to our Emergency Room (ER) because of a painful mass in vulvar region. The patient reported a progressive and asymptomatic enlargement of the vulvar mass during the last 6 years, which has worsened in the last few days causing intolerable pain, difficult deambulation and dyspareunia.

Physical examination showed a bulky subcutaneous nodular lesion, of around 8 cm in diameter, painful at palpation involving the right labium majus. Overlying skin was normal, with no sign of in-

flammation and no bulky inguinal node. Intra-vaginal ultrasonography proved unremarkable. Pain relievers were administered unsuccessfully.

Diagnosis of a Bartholin cyst was then made. In a few hours, the patient underwent surgical excision because of the symptoms.

After surgical incision of the skin, a tubular, cul-de-sac cavity, was evident, macroscopically similar to intestinal serosa (**figure 1**). Thus, a herniated Meckel's diverticulum was suspected, surgical conversion to laparotomy was decided and general surgeon was involved.

Through bi-manual intra- and extra-abdominal palpation, hernias were excluded. The mass appeared completely independent from adjacent tissues and was then excised (**figure 2**).

Histology showed a 13 × 8 cm cystic mass, with smooth and translucent surface. The mass was capsulated and comprised lardaceous, translucent, homogenous tissue. Immunohistochemistry was positive for desmine, ER/PGR receptors and actin-MS, and the final diagnosis of aggressive angi-

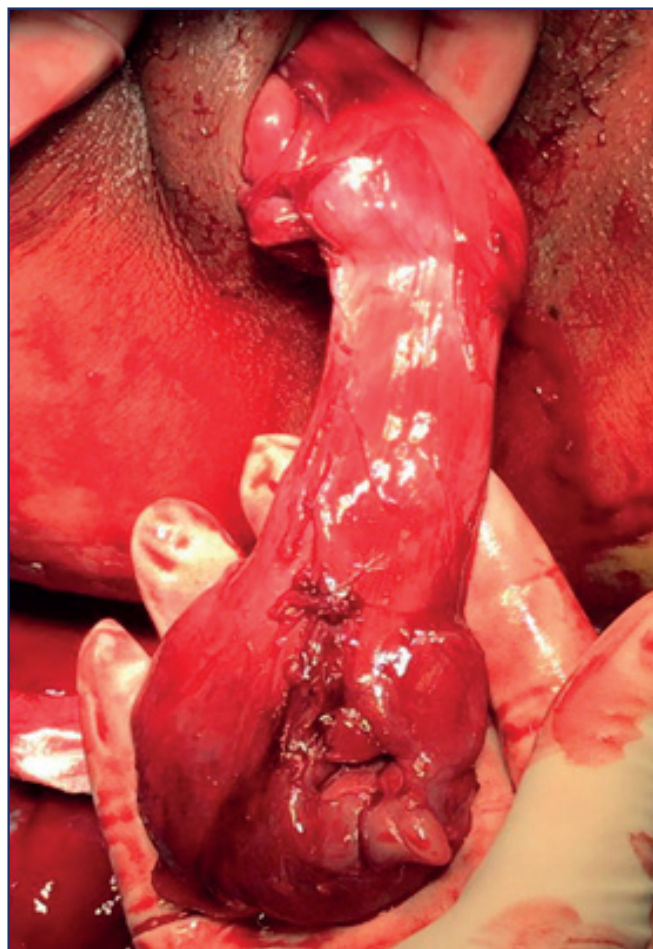


Figure 1 Approximately 10 cm mass macroscopically similar to intestinal serosa.

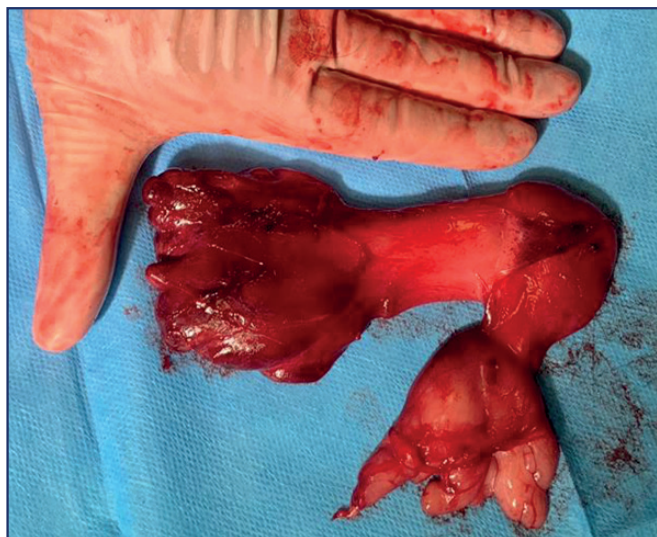


Figure 2 The mass appeared completely independent from adjacent tissues after the wide excision.

omyxoma was reported. The patient was referred to a tertiary care center and she is currently in good clinical condition with no sign of recurrence.

DISCUSSION

AAM is a rare mesenchymal tumor arising from connective tissue of the pelvis and the perineal region, and is typical of women about 95%, usually in the reproductive age with a female to male ratio of 6:1 (1).

The peak incidence of AAM is at 35-40 years of age like our case.

Steeper and Rosai (1) first described the histological aspects and highlighted its tendency to local infiltration and recurrence in 1983. Around one hundred and fifty cases have been reported in the world medical literature since then (12).

The cases described in literature were characterized by typical features including slow growth, gelatinous appearance and locally infiltrative, but non-metastasizing nature without evidence of nuclear atypia or mitosis; distant metastasis (to the lung) has been reported in only two cases (3, 5, 6). It is classified under "Tumors of uncertain differentiation" in the latest WHO classification.

Diagnosis is frequently difficult, because it is often asymptomatic until it become a bulky mass.

For this reason it is frequently misdiagnosed as a Bartholin's gland cyst, hydroadenoma, angiomyofibroblastoma, leiomyoma, inguinal hernias, vaginal prolapse and vulvar abscess. (9).

The presentation size is extremely variable ranging from 1 up to 60 cm, but most of them are > 10 cm (3). Only histology evaluation and specific immunohistochemistry allows definitive diagnosis. Magnetic Resonance Imaging (MRI) can be useful in order to evaluate the relationship with surrounding tissues and to plan surgical excision (13). In our case no biopsy or imaging of the lesion were performed as a diagnostic study because of the clinical conditions of the patient requiring prompt surgery.

Chemo- or radiotherapy have not proven efficient, but hormone therapy might be used as adjuvant treatment, because of the frequent expression of ER/PGT receptors by the AAM tissue (8). Hormonal therapy (GnRH, SERM or combinations of them) may be used as neoadjuvant therapy in case of enlarged masses, relapse or when surgery is contraindicated (14).

Treatment of choice is still a wide surgical excision with tumor free margins (1). However, retrospective studies have demonstrated that positive resection margins after surgery are not associated with higher rate recurrence (3), confirming the benign nature of the neoplasia, which is associated with very good prognosis.

Notwithstanding, patients should be informed that recurrence is not rare, even after decades after first surgical excision, thus, a long clinical follow-up is strongly recommended.

CONTRIBUTIONS

VM and SE did the project development. BC, BV, ZPL and PR did data collection and manuscript writing. All authors read and approved the final manuscript.

CONFLICT OF INTERESTS

The authors declare that they have no conflict of interests.

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