CASE REPORT

Deep angiomyxoma of retroperitoneum presenting as a perineal lump in a young woman

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SUMMARY

Aggressive angiomyxoma (AAM) is a rare locally aggressive neoplasm of mesenchymal origin which occurs mainly in adult women. High local recurrence rate after surgery is due to incomplete surgical excision and most of the reported cases originate in the pelvis. We present a case of massive angiomyxoma arising from the abdomen and extending into the pelvis and perineum. A 23-year-old woman presented with a swelling of the left lower abdomen with discomfort and pain for 4 years. The swelling increased in size progressively and involved the perineum. At surgery the mass was abutting the urinary bladder and rectum. On microscopy there were typical features of AAM. Our case is being reported due to the rarity of its presentation. Close follow-up is necessary as there is high chance of recurrence.

BACKGROUND

The abdomen, with a host of organs and numerous structures inside, remains a puzzle for physicians. Owing to multiple facial planes, hernia orifices and huge peritoneal spaces, abdominal lumps can grow into an enormous size or project into extra abdominal sites. We encountered a case of a large deep (aggressive) angiomyxoma of retroperitoneum which could not be diagnosed until the final histopathology. In 1983 Steeper and Rosai first coined the term 'aggressive angiomyxoma' which was later changed to 'deep angiomyxoma' by WHO.¹ It is a locally aggressive neoplasm of mesenchymal origin occurring mainly in elderly adults. The vulva in women and spermatic cord and inguinal region in men are the most commonly affected organs.² The histogenesis of the tumour is poorly understood. Only a small number of cases have been reported in literature. We therefore present such a case where the tumour occurred in a young woman. It arose from the abdomen (unlike the sites reported) and presented as a perineal lump.

CASE PRESENTATION

A 23-year-old unmarried woman presented to us with a history of dragging pain and discomfort in the left lower abdomen for 4 years. For the past 1 year she had noticed a gradually increasing swelling in the perineum to the left of the vulva. She had also noticed frequency of micturition, oligomenorrhoea and constipation for the past 2 months.

INVESTIGATIONS

On general physical examination no abnormality was detected. Abdominal examination revealed an ill-defined firm, fixed, retroperitoneal lump in the

left iliac fossa extending to the pelvis. On perineal examination, there was a 20×10 cm solitary, lobulated, soft swelling in the perineum and left vulva (figure 1A). This increased in size on squatting. Cross-fluctuation between the abdominal and perineal lumps could not be elicited. An ultrasonogram, contrast-enhanced CT (CECT) and MRI of the abdomen and pelvis showed a large soft tissue mass extending from the left iliac fossa to the pelvis extending into the perineum, displacing the urinary bladder and the bicornuate uterus (figure 2A–D). The descending and sigmoid colon and rectum appeared to be uninvolved but the left iliac vessels appeared to be encased. A fine-needle cytology (FNAC) and fine-needle biopsy (FNAB) were inconclusive. A surgical removal of the lump was planned.

Pathological finding

Grossly the tumour comprised of many pieces of an ill-circumscribed soft tissue mass with focal areas of haemorrhage. The tumour was 40×30×10 cm in size. The cut surface of the tumour was grey-white and gelatinous with focal areas of haemorrhage (figure 1C,D).

H&E-stained slides on light microscopy showed a hypocellular tumour comprising of stellate cells in a myxomatous background, showing evidence of old haemorrhage and minimal fibrosis. These cells had scant to moderate amount of eosinophilic cytoplasm and elongated to star-shaped nuclei. Interspersed in these cells were variable-sized thick-walled and thin-walled vessels. There was perivascular hypercellularity with scattered lymphoid cells mostly around vessels (figure 1E,F). Immunohistochemically the tumour showed diffuse positivity for smooth muscle actin (SMA), desmin, estrogen receptor and progesterone receptors (figure 3A–D).

DIFFERENTIAL DIAGNOSIS

- Angiomyofibroblastoma
- Cutaneous myxoma
- Myxoid neurofibroma

TREATMENT

Hence, surgical removal of the lump was planned. At laparotomy, there was a large, jelly-like translucent mass, shaped like a cockscomb, arising from the retroperitoneum occupying the left iliac fossa, filling the pelvis, adherent to the urinary bladder, the broad ligament of the uterus and rectum. The mass was also involving the left pelvic wall, encasing the left iliac vessels and was then projecting through the perineum. A combined abdominal

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Figure 1 Clinical photograph showing solitary, lobulated, soft swelling (20×10 cm) in the perineum (A). Intraoperative photograph showing jelly like mass (b). Gross photograph showing a gelatinous mass (C and D). Photomicrograph showing variable sized vessels in a myxoid background, at areas showing perivascular hypercellularity and lymphoid cell around vessels (E, H&E ×200). Photomicrograph also showing features of an aggressive angiomyxoma with stellate cells with ill-defined cytoplasm in a myxoid background (F, H&E ×400).

Figure 2 Axial T1-weighted (A), axial T2-weighted (B), coronal T2-weighted (C) image showing ill-defined soft tissue mass which is displacing bladder and uterus anteriorly. The mass is hypointense on T1 and heterogeneously hyperintense on T2. Fat plane with bladder and sacrum is maintained. Divergent endometrium suggestive of bicornuate uterus. Axial contrast-enhanced CT showings homogenous filling mass (D).

Figure 3 Photomicrograph of immunohistochemistry. Microphotograph showing the stellate cells positivity for estrogen (A, IHC (ER) $\times 400$), progesterone (B, (PR) $\times 400$), smooth muscle antigen (C, IHC (SMA) $\times 200$) and desmin (D, IHC (DES) $\times 200$).

and perineal dissection was performed and the mass was removed in piece meal. No visible tumour was left behind. However, residual microscopic tumour especially over the iliac vessels could have remained. Her postoperative period was uneventful.

OUTCOME AND FOLLOW-UP

The patient did not attend follow-up until 6 months later following which she presented with a lump in the previous perineal site. CECT showed a recurrence almost the size and extension of the initial one. It was explained to the patient about the possibility of sacrificing adjacent viscera for a complete removal of the tumour. At the time of writing this report, the patient has been considering further surgery for 2 months.

DISCUSSION

This uncommon soft tissue neoplasm has a female predominance (6:1) but occurs in all age groups; most commonly in the third to fifth decade of life.² The tumour occurs mostly in the perineal area. The relative incidence in scrotum, spermatic cord, perineum and pelvis are 38%, 33%, 13% and 8%, respectively.³

Its erstwhile name 'aggressive angiomyxoma' is due to the high local recurrence and typical histological findings of myxoid stroma, rich in collagen fibre with stellate-shaped cells. There are numerous thick-walled blood vessels with perivascular hypercellularity with focal areas showing lymphoid aggregates. Literature reveals no pathological difference between male and female patients, despite hormonal sensitivity.²

The tumour has a divergent differentiation as there is no definitive tumour marker. Immunohistochemically the tumour cells are positive for CD34, desmin, estrogen, progesterone and SMA focally. Most tumours are idiopathic though a few have been reported to have t(12:21) and HMGA2 rearrangement.⁴ Estrogen and progesterone positivity indicates hormonal influence on the growth of the tumour.⁵ However, its occurrence in

both sexes and in all age groups suggests that hormonal influence may not have a major role to play.

Histologically, the closest differential diagnosis is the superficial angiomyxoma, which usually occurs in the subcutaneous tissue in the anogenital region. It resembles deep angiomyxoma, but lacks thick-walled blood vessels and hormonal expression. Angiomyofibroblastoma is another close mimicker which occurs in a similar location and is composed of plump epithelioid cells in a myxoid background with numerous thin-walled blood vessels but lacks hormonal expression. Multinucleate giant cells and smaller tumour size favour an angiomyofibroblastoma. It should be differentiated from deep angiomyxoma, as the treatment and prognosis of both are different. The other conditions that could be confused with deep angiomyxoma include fibromatosis and intramuscular myxoma.

Ultrastructural and immunohistochemical analysis showed a divergent differentiation of tumour cells—fibroblastic, myofibroblastic and smooth muscle cell lines. This may be due to the origin of the tumour from multipotent primitive mesenchymal cells.

Wide local excision of these tumours is required but recurrence occurs in 30–70% of cases.⁶ The recurrence can occur as early as within 1 month.⁷ The type of surgical approach depends on the epicentre of the tumour. For tumours mainly confined to pelvis or perineum, which is in most of the cases, perineal approach or trans-sacral approach through inverted Y incision can avoid unnecessary complications as described by Hong *et al.*⁸ In our case, the main bulk of the tumour was in the lower abdomen and pelvis with dense adhesion to viscera, hence a combined abdominal and perineal approach was performed. The recurrence is usually due to its local infiltration and the inability to excise the tumour completely. Only an occasional case of metastasis has been reported.⁹ In view of hormonal sensitivity, some researchers have advocated the use of hormonal therapy in those who have a recurrence.

Learning points

- ▶ Deep angiomyxoma is a rare benign neoplasm that can mimic many clinical and histological entities.
- ▶ Its aggressive but indolent course leads to late diagnosis.
- ▶ Surgery is the only form of treatment available albeit with a high risk of recurrence, thus necessitating a close follow-up.

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