A RARER CASE OF A RARE DISEASE:
AGGRESSIVE ANGIOMYXOMA (AAM) IN A SIXTEEN YEAR OLD.

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ABSTRACT

A 16 year old female presented with a two month history of an uncomfortable mass in her upper inner thigh. The mass was thought to be cystic in nature and removed as a day case procedure. However, on histological examination the mass was found to be composed of abundant myxoid stroma, and a diagnosis of aggressive angiomyxoma (AAM) was diagnosed.1 Following the histology report and given AAM’s propensity to recur5,7, further resection of the margins were carried out, which were found to contain two small patches of tumour. AAM is a very rare benign mesenchymal tumour found mainly in the pelvis and perineum. The patient remains recurrence free one year on. Up until now most cases have been described in patients in their 3rd and 4th decade.3, 11 This case report highlights the need to consider AAM in all women of reproductive age who present with a mass of unknown cause in the vaginal surrounding area.

BACKGROUND

- Haldar et al reported in 2009 that there were less than 250 reported cases of AAM, therefore it is important to report all cases and variations in presentation to those already written up.2
- This case focuses on a patient who was sixteen years old at diagnosis, with the average reported age of AAM around the fourth decade it is important to report cases seen in younger or older patients so that further research can take into account these differing demographics.3
- Misdiagnosis is common; therefore raising awareness in clinicians to the presence of this condition is important.

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CASE PRESENTATION:

A 16 year old female, first presented with an uncomfortable, small lump growing on her inner-upper thigh. She presented to her GP complaining of discomfort with the lump, especially when seated. On examination the patient had a 2cm x 2cm, tender, lump (noted to be the size of a fifty pence piece) in her left medial gluteal region. The surrounding area was slightly erythematous, but she reported no discharge or broken skin. The patient had been aware of the lump two months prior to presentation, and she had felt that it had only grown slightly during this period.

The patient had no past medical history of note. She was later diagnosed with Iron Deficiency Anaemia, but this was thought to have no connection to her AAM. She was on no regular medication and reported no known allergies. She has a regular 28 day cycle for which she bleeds for seven days. She had received the HSV 16 & 18 vaccine. The only relevant family history to note was that her paternal aunt had suffered from skin cancer but no further details were given.

The patient was initially referred for an excision of the lump that was considered to be cystic in nature. Removal of the lump was performed as a day case, under general anaesthetic and was uneventful. Histology showed that the lump had been caused by a benign neoplasm of the connective tissue, known formally as an aggressive angiomyxoma. Further surgery was swiftly undertaken to further resect margins to ensure there had been complete removal.

INVESTIGATIONS:

At the time of original presentation, no specific investigations were undertaken. Routine histopathological tests were performed after the first surgery on the excised tissue, which flagged the unexpected results. Macroscopically two irregular pieces of soft mucoid tissue measuring around 30 x 15 x 10mm were seen. The biopsy showed benign tumour composed of abundant myxoid stroma, rich in sizeable blood vessels. Numerous spindle cells with elongated cytoplasm and irregular elongated nuclei were seen. Inflammatory cells were present. No associated normal tissue was seen. Immunostaining of the spindle cells, were positive for vimentin, smooth muscle actin and CD34, they were negative for desmin, S-100 protein and oestrogen and progesterone receptors. These findings contradicted the original suspicion of a simple cystic lesion, and instead suggested the more serious diagnosis of aggressive angiomyxoma.

An MRI pelvis was organised for the patient, after the unexpected histological findings were reported, to look for any further involved areas. A
single 10mm lymph node was seen, but no other residual or recurrent disease was seen.

**TREATMENT:**

It is known that AAM can reoccur locally if not completely excised, so as previously described a second surgery was arranged for further re-excision of the lesion. The re-excision showed two very small areas measuring 0.5mm of residual angiomyxoma, which were thought to represent part of the original lesion, that were now separate lesions due to the previous excision. No medical therapies were used to treat this case of AAM.

**OUTCOME AND FOLLOW-UP:**

Although, aggressive angiomyxomas have a strong history of recurrence, after the second excision it was felt, by the MDT coordinating her care, that no further active treatment was required but that the patient should be followed up at a regional cancer centre. Follow up appointments for the patient were scheduled at three monthly intervals over a period of two years to examine the affected area for any signs of further growth. One year following the second surgery, the scar is well healed, with no current indications of further growth or change in the area. The patient has been referred back to her district general for all further follow up.

**DISCUSSION:**

AAM was first reported by Steeper and Rosai in 1983, describing nine cases of a mesenchymal neoplasm.(1) In 2009 there were less than two hundred and fifty reported cases of AAM, highlighting the rarity of this condition.² Most reported cases, have been in females of reproductive age in their fourth decade, with a female to male ratio of 6:1.(3) The case we have reported is at the younger boundary of reported cases, with our patient aged only sixteen.(3&4) Literature searches have highlighted very few reported cases of or below this age, with the lowest age reported being of a six year old.³

This condition is misdiagnosed in 80% of cases, commonly as a cystic lesion, as was the case with our patient.(5) The classic presentation is of a slow growing painless lump in the vulval, perineal or gluteal region. Patients often report the lumps to be asymptomatic though they can show signs of slight erythema. Histology is the gold standard for diagnosis, which generally is reported as showing thick walled vessels of medium size, surrounded by a layer of stromal cells of myxoid appearance.(6) When stained this stromal cells can show the presence of vimentin, desmin, smooth muscle actin and CD
34, oestrogen and progesterone receptors. (2) In our case the stromal cells were vimentin, smooth muscle actin and CD34 receptor positive.

AAM is well known for its high recurrence rate of up to 70% after excision. There is disagreement as to whether this is due to poorly demarcated borders leading to inadequate resection, or another unknown means of recurrence. (4,5,6&7) Excision is still the regarded as the best treatment but GnRH agonists have been reported to prevent recurrence in isolated cases. (8) This is mainly used in patients with progesterone or oestrogen receptor positive masses, which was not true for our patient. (11) No role for chemotherapy nor radiotherapy has been found in the treatment of AAM.

There are only two reported cases of AAM metastising and it normally presents as a localised, benign neoplasm. (9,10) Therefore recurrence, despite the doubts in the mechanism by which it occurs, is not a sign of poor prognosis and can be dealt with simply by excision.

The case we have reported is a classical presentation of AAM, and like many others was classically misdiagnosed. Yet what stands this case apart from those previously reported is the age of the patient. She is one of the youngest reported cases, highlighting the importance of not overlooking AAM as a possible diagnosis in a younger population.

LEARNING POINTS:

- Aggressive angiomyxoma should always be in the differential for gynaecological lumps without a clear cause in women of reproductive age.
- Histology is the only way of making a definite diagnosis
- Treatment should be to excise the tumour with the option of gonadotrophin-realising hormone agonist used as an adjunct

REFERENCES:


The patient was not involved in any clinical trials. She has consented to the writing of this article.