International Journal of Reproduction, Contraception, Obstetrics and Gynecology
Mukherjee S et al. Int J Reprod Contracept Obstet Gynecol. 2023 Mar;12(3):747-749
www.ijrcog.org
pISSN 2320-1770 | eISSN 2320-1789

DOI: https://dx.doi.org/10.18203/2320-1770.ijrcog20230549

Case Report

A giant aggressive angiomyxoma of the vulva in a dwarf: a rare case report

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Received: 13 January 2023
Accepted: 03 February 2023

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ABSTRACT

Aggressive angiomyxoma is a rare soft tissue tumour that carries a high risk of local relapse. The tumour usually arises from the pelvic and perineal regions. We presented a case of a young girl with dwarfism, mental retardation and thyroid agenesis who presented with aggressive angiomyxoma of the vulva. As it is almost exclusively seen in women of reproductive age group, this is the first case reported in a teenage girl, to the best of our knowledge. A mentally retarded girl, aged fifteen years, presented with a large mass hanging from the vulva, with ulceration over the mass. She had short stature, anasarca, mental retardation, primary amenorrhoea, under-developed secondary sexual characters .and thyroid agenesis. Local examination revealed a pedunculated mass of 15 by 4 cm size arising from the mons pubis closely resembling a giant penis. Full hormonal and radiological work-up was done. Her condition was optimized and the mass was removed with wide excision of margins followed by cystoscopy under general anaesthesia. Histopathology and immunohistochemistry were suggestive of aggressive angiomyxoma. She has no recurrence till date. Despite its rarity, angiomyxoma should be considered in the differential diagnosis of any painless swelling located in the genitofemoral region. The principal treatment is surgical excision. Long term follow-up is needed due to its high tendency of local recurrence.

Keywords: Dwarf, Angiomyxoma, Thyroid agenesis

INTRODUCTION

Aggressive angiomyxoma is an uncommon mesenchymal tumour which is predominantly seen in adult females of reproductive age group.[1] The tumour usually arises from the pelvic and perineal regions. Macroscopically, the tumour has a gelatinous appearance, and it is microscopically characterized by a myxoid stroma and abundant thick- walled vascular channels.[2] It is noteworthy that there is still a lack of knowledge about the clinical presentation, the management options and the follow-up results in the current literature. Aggressive angiomyxoma may initially be misdiagnosed as a gynaecological malignancy, clitoromegaly or even a hernia that leads to unnecessary surgical interventions.[3]

Since aggressive angiomyxoma was first described in 1983 by Steeper and Rosai, about 350 cases have been reported in medical literature.[4,5] To the best of our knowledge, we are reporting the first ever case of aggressive angiomyxoma in a teenage dwarf with co-existing thyroid agenesis.

CASE REPORT

A girl, aged 15 years, presented in out-patient department, with a big mass in the genital area. She had generalised swelling, hoarse voice, subnormal mentation and primary amenorrhoea (Figure 1). Her father informed that the mass was slow growing, painless but causes discomfort to the child. On examination, the child was conscious to place

and person but unable to communicate verbally. Her height was 120 cm and she weighed 50 kg. She had significant bradycardia (pulse rate 48/min). Her secondary sexual characters were under developed. Local examination revealed a 15 cm long pedunculated firm non tender mass arising from the mons pubis (Figure 2). The surface was smooth, pigmented with superficial ulcerations. The labia majora and minora was developed normally for age. The girl was admitted and a complete work-up was done. Hemogram revealed moderate iron deficiency anaemia. Her thyroid profile was severely deranged, suggestive of thyroid agenesis which was confirmed by ultrasound. The biochemistry panel was normal. Whole abdomen ultrasound revealed a hypoplastic uterus with normal sized ovaries. Rest of the hormonal assay was within normal limits. The echocardiography revealed global hypokinesia of the heart.

Thyroid replacement therapy was started. Iron and protein supplementation was given. The mass had superficial ulcers due to rough handling by the child. Daily dressing was done. Her thyroid level was optimized by the end of a month. Her haemoglobin improved. She was given surgical fitness finally by endocrinologist and cardiologist. She was planned for cystoscopy followed by wide excision of the mass under general anaesthesia. The surgical procedure was uneventful and the mass was sent for histopathological examination. The gross specimen was around 15 cm by 4 cm in size, pigmented with superficial ulcers. Necrosis was absent. Microscopic examination revealed spindle shaped tumour cells in myxoid stroma with haphazard dilated capillaries suggestive of aggressive angiomyxoma (Figure 3). Immunohistochemistry was positive for desmin, ER and PR.

The post-operative period was uneventful and there is no recurrence on 6 monthly follow up. She is, however, non-compliant and takes thyroxine replacement irregularly. She is taking treatment under endocrinologist and cardiologist.

Figure 1: The patient.

Figure 2: The mass.

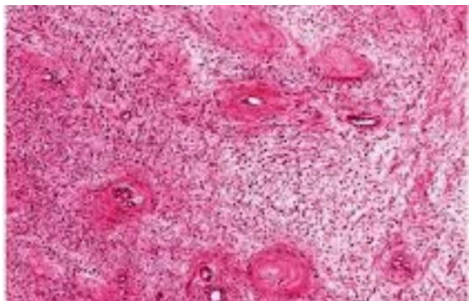

Figure 3: The micrograph of the histopathology.

DISCUSSION

Aggressive angiomyxoma is a rare, slow growing soft tissue tumour that usually arises from pelvis and perineal regions of women in reproductive age group.[6] Pre-operative diagnosis is difficult due to rarity and absence of diagnostic features, but it should be considered in every mass in genital, perianal and pelvic region. Aggressive angiomyxoma occurs almost exclusively in women of reproductive age, with a peak incidence in the third to fifth decades of life.[7] Other rare sites include lung, liver, larynx, and orbit.[5] Occasionally, they are seen in male patients in related sites like the scrotum and inguinal region. However, the ratio of occurrence in females to males is 6:1.[8] Aggressive angiomyxoma may be commonly misdiagnosed as angiomyofibroblastoma. However, the latter is well circumscribed, more cellular, and more vascular.

Many treatment modalities have been tried with varying success. Radical surgical excision with negative margins is the conventional treatment of choice. However, it is not always possible to achieve negative resection margins as the tumour is locally infiltrative, leading to high operative morbidity. Therefore, less radical surgery is recommended

nowadays. AA is known for multiple local recurrences to the extent of 36-72% and may occur as early as 2 months to as late as 20 years.[9] Begin et al described nine AA cases with local recurrence, all due to incomplete excision.[10] Siassi et al reported a death due to multiorgan metastasis invading the peritoneum, lungs, and lymph nodes.[11]

No evidence-based recommendations are available for post-surgery management of AA, but due to the high rate of local recurrences and possible metastasis, patients should be advised to undergo long-term follow-up uptil 15 years after the primary excision.

To the best of our knowledge, we are reporting the first ever case of aggressive angiomyxoma in a teenage girl with dwarfism and thyroid agenesis. There is paucity of literature in regards to the association of angiomyxoma with thyroid disease. The association of any syndrome with aggressive angiomyxoma is also not known.

CONCLUSION

Despite its rarity, aggressive angiomyxoma should be considered in the differential diagnosis of any painless swelling located in the genitofemoral region. The principal treatment should be complete surgical excision with tumour-free margins. The patient should be informed about the high morbidity of the surgical intervention. Long-term follow-up and careful monitoring are essential due to its high tendency of local recurrence in spite of wide excision of the tumour.

Funding: No funding sources
Conflict of interest: None declared
Ethical approval: Not required

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Cite this article as: Mukherjee S, Prakash A, Sodhiya H, Jaiswal S. A giant aggressive angiomyxoma of the vulva in a dwarf: a rare case report. Int J Reprod Contracept Obstet Gynecol 2023;12:747-9.