**Vulvar sarcoma in a young patient with neurofibromatosis syndrome with missed opportunities: A case report**

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**Abstract**

**Background:** Vulvar malignancies are the fourth commonest of genital malignancies. The sarcoma type is associated with neurofibromatosis disease.

**Case presentation:** A 29-year-old nulliparous presented to the outpatient gynecological clinic with a five-year history of a recurrent right vulvar mass. The mass was initially excised without histological analysis. Histological examination of a reexcised mass diagnosed a spindle cell sarcoma. She was then lost to follow-up for one year. The mass recurred and she received external beam radiation. On admission, she was in good general status with generalised café-au-lait spots and neurofibromas. Perineal examination revealed an ulcerated right vulvar mass. Toilet vulvectomy and diversion colostomy were performed followed by chemotherapy. She was discharged after five months with a well-granulated wound. Colostomy was reversed six months later.

**Conclusion:** Vulvar sarcoma is a rare vulvar malignancy type usually associated with neurofibromatosis. This case highlights the missed opportunities in its management. A high suspicion threshold is critical for early diagnosis and management to curb the morbidity and mortality rate.

**Keywords:** loss to follow-up, neurofibromatosis, vulvar sarcoma, vulvectomy

**Introduction**

Vulvar carcinoma is the fourth most common genital malignancy representing 3-5% of genital cancers (1,2). Squamous cell carcinoma is the commonest histological type followed by melanoma, basal cell carcinoma, and vulvar sarcoma. Vulvar malignancies commonly occur in postmenopausal women in association with chronic vulvar epithelial disorders (1). Occurrence in the younger population is commonly associated with human papillomavirus (HPV) infection and immune deficiency states. Vulvar sarcoma has been reported in patients with neurofibromatosis disease (3,4). Vulvar malignancies may present with a vulvar mass, pruritus vulvae, offensive vaginal discharge, and pain but can also be asymptomatic (5). Vulvar malignancies are mostly misdiagnosed as candidiasis or vaginitis. Misdiagnosis coupled with clinicians having a low index of suspicion, particularly in the younger population, may delay early management and disease advancement before diagnosis (6,7). Treatment entails surgical excision, chemoradiation, or chemotherapy (1). This is a case of recurrent vulvar sarcoma in a 29-year-old nulliparous with neurofibromatosis disease with missed management opportunities.

**Case presentation**
A 29-year-old nulliparous presented to the outpatient gynecological clinic at the Kenyatta National Hospital with a five-year history of right vulvar mass as a referral. The mass first appeared in 2017 as a small pimple that progressively developed into a painless mass with a foul-smelling discharge. She sought medical assistance during which excision was performed without histological analysis of the mass. The mass recurred in 2020 and was excised at the same peripheral facility. The histological analysis reported a spindle cell sarcoma. She was referred to a gynecologist but was lost to follow-up until 2021 when the mass recurred. She was reviewed by a medical oncologist and scheduled for 33 sessions of external beam radiation. She was HIV-seronegative, gave no history of prior sexual contact, and had never used contraceptives.

On admission, she was in good nutritional status, not pale, and with stable vital signs. Physical examination revealed Café-au-lait spots and neurofibromas distributed throughout the body (Figure 1). Perineal examination revealed a mass on the right side of the labia majora that measured 15 x 10 cm. The mass was ulcerated, foul-smelling, fixed to the underlying tissue, and extended close to but did not involve the anus (Figure 2). The left side of the vulva, vagina, urethra meatus, and clitoris were not involved. A right inguinal node that measured 3 x 2 cm was palpable. There were no neurofibromas in the perineal region. A multidisciplinary team approach was employed. Vulvectom and colostomy were performed one day apart (Figure 3). Her postoperative period was uneventful. Histological analysis confirmed a spindle cell sarcoma with clear margins. One month postoperative, she was started on gemcitabine/doxorubicin adjuvant chemotherapy and received six cycles. A postsurgical pelvic ultrasound revealed multiple enlarged pelvic floor lymphadenopathy and grossly normal pelvic viscera. After five months, she was discharged in a stable condition with a well-granulated wound. Colostomy was reversed by the surgical team six months following the vulvectomy. The patient was satisfied with the cosmetic and functional outcome of the procedures.

**Figure 1:** Café-au-lait spots on the abdomen (A), left arm (B), and the right thigh (C).

**Figure 2:** A: Ulcerating and foul-smelling right vulvar mass before surgery; B: Excised mass; C: Immediate postexcision wound.
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Figure 3: Wound appearance five months postexcision and chemotherapy with a colostomy bag in situ.

Discussion
Spindle cell sarcomas are extremely rare histological subtypes that are underreported (8). Vulvar malignancies commonly occur in postmenopausal women in association with chronic vulvar epithelial disorders, but may occur in younger women in association with immunocompromised states or HPV infection (2). Patients affected with type 1 neurofibromatosis (NF1) are susceptible to developing both benign and malignant tumors (3). Neurofibromin-1 gene mutations cause NF1. Neurofibromin-1 regulates cellular proliferation by activating the intrinsic GTPase of p21-ras. The loss of function in NF1 leads to a loss of control over cell proliferation. As a result, patients with NF1 have a fourfold increased risk of developing benign and malignant tumors. Neurofibromas can develop on any body part. However, the involvement of the female vaginal system is not typical (3,9).

Clinical presentation of vulvar sarcoma may be asymptomatic and when symptomatic, it may present with vulvar mass, pruritus vulvae, offensive vaginal discharge, and vulvar pain (5). Misdiagnosis of these symptoms as vaginitis and a low suspicion index of vulvar cancer, especially in the younger population leads to late diagnosis with delayed management initiation, which may result in increased morbidity and mortality (6,7). Here, the patient had a recurrent vulvar mass, which prompted her to seek care relatively early. However, there were gaps and missed opportunities, for instance, lack of initial histological evaluation and the loss to follow-up, that contributed to the delayed management. Management of vulvar cancer entails mass excision, and chemoradiation with or without neoadjuvant chemotherapy, depending on the size and depth of stromal invasion of the lesion, nodal involvement, and lymphovascular invasion (1). In case the anus is involved or close to being involved, there might be a need to include the surgical team for temporary diversion colostomy to allow wound healing, as in this case. Vulvar sarcomas are rare, and few cases of patients with a diagnosis of neurofibromatosis have been described (3).

Conclusion
Vulvar sarcoma is a rare vulvar malignancy type usually associated with neurofibromatosis. This case highlights the missed opportunities in its management. A high suspicion threshold is critical for early diagnosis and management to curb the morbidity and mortality rate.

Consent for publication
Informed consent for publication was obtained from the patient.

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