CASE REPORT

A PROGRESSIVE EMBRYONAL RHABDOMYOSARCOMA OF THE UTERINE CERVIX WITH RIGHT BREAST LUMP IN A 43-YEAR-OLD ADULT: A CASE REPORT

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ABSTRACT

Introduction: Embryonal Rhabdomyosarcoma is an uncommon type of sarcoma often seen in pediatric and adolescent patients, with the botryoid subtype being the most common. The incidence of this disease in adult females is 0.4% to 1% with the affected age group being patients in the third to fourth decade of life. It is rare in patients above 40 years in Nigeria.

Case Presentation: We describe the case of a 43-year-old, para 4+1, 4 alive, premenopausal female health worker, with an ECOG performance status of 1, who presented with a protruding cauliflower, exophytic cervical polyp extending to the lower one-third of the vagina of eight months duration. Post-polypectomy histology revealed a huge, highly vascularized mass, with high histologic grade, and some cartilaginous components. Based on financial limitations and out-of-pocket payments, the patient could not afford standard concurrent chemo-radiation post-surgery. Adjuvant chemotherapy with intravenous (IV) Vincristine 2mg Day 1, Doxorubicin 100mg Day 1, and Cyclophosphamide 1000mg D1 (VAC) regimen, 3 weekly for 6 courses, was offered to the patient. However, a partial

response after the third course was noticed, with an incidental finding of a right breast mass noted. A modified regimen with the addition of IV Dacarbazine 1500mg on Day 1 was given due to poor response and tumour recurrence. Total abdominal hysterectomy (TAH) with bilateral salpingo-oophorectomy (BSO) was done, and immunohistochemistry for Desmin was strongly positive. The patient's performance status remains the same while further evaluation of the right breast is being carried out. She has been counseled to undergo radiotherapy as soon as possible. We discuss the disease in general, pathologic features of the disease, epidemiology, presentation patterns with prognostic factors, management modalities with consideration of standard radiation therapy management, and outcomes.

Keywords: Polypectomy, Cervical Polyp, Sarcoma, Radiotherapy, Cervical embryonal rhabdomyosarcoma, Nigeria

INTRODUCTION

Embryonal Rhabdomyosarcoma (ERMS) is a rare histological form of cancer with mesenchymal malignant cells in resembling the primitive developing embryonic skeletal muscle. The uterine cervix is a rare presentation site for ERMS.[1,2] It tends to present in the age range of 10-30 years, and the majority of cases are in individuals less than 10 years of age, with no preference in race or ethnicity. Found in mucosal-lined organs like the vagina, bladder, and the nasopharynx. About 60-70% of RMS is embryonal rhabdomyosarcoma.

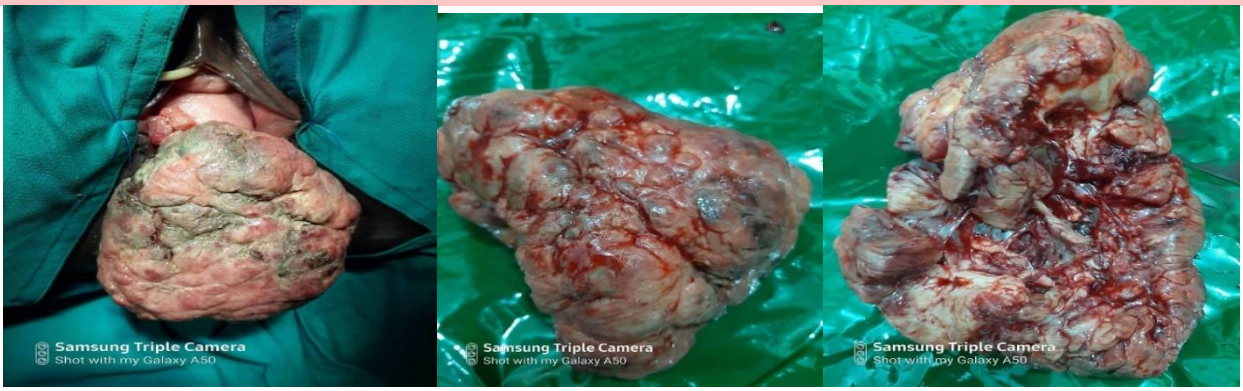
The treatment approach is multidisciplinary and multimodal, with a significant increase in good prognoses using Surgical intervention, Chemo-Radiation, and, to an extent, combination Chemotherapy with Vincristine, Adriamycin, Cyclophosphamide (VAC), and Ifosfamide (with Mesna), Vincristine, Adriamycin (IVA).

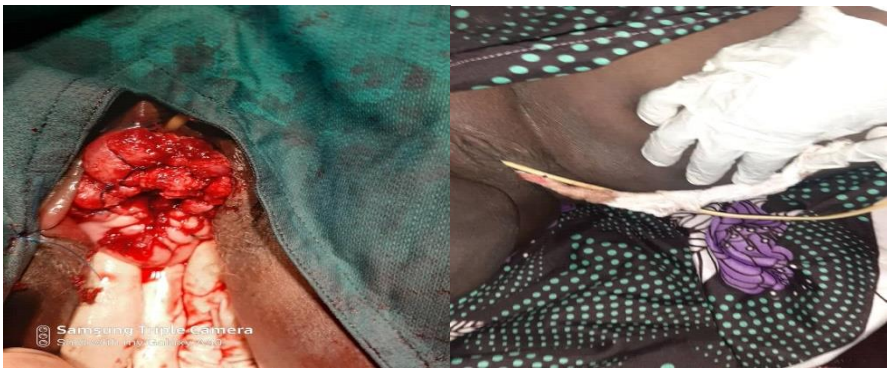
CASE PRESENTATION

A 43-year-old Para 4^{+1}, 4 alive, Human Immunodeficiency Virus (HIV) negative Nigerian woman. The last childbirth was fourteen months ago. She presented at Ahmadu Bello University Teaching Hospital (ABUTH), Zaria, Nigeria, with an eight-month history of a protruding, huge, fungating, cauliflower, exophytic, pinkish multi-nodular mass in her vaginal introitus measuring17cm×12cm× 8cm. The mass extended to the lower one-third of the vagina, was highly vascularized with multiple areas of necrosis.

On clinical examination, her performance status was 1 using the Eastern Cooperative Oncology Group (ECOG) score, with vaginal examination revealing a cervical polyp extending to both the anterior and posterior lips of an elongated cervix with satellite lesions on the anterior and lateral walls of the vagina abutting the bladder. There was a palpable nodular lesion in the left parametrium, and polypectomy was done.

Microscopic findings were those of embryonal rhabdomyosarcoma of botryroid subtype. She was staged using the International Federation of Gynecology and Obstetrics (FIGO), American Joint Committee on Cancer (AJCC 2019) as stage IIIA. No immunohistochemistry was done due to financial constraints. Other systems showed no abnormality with no evidence of metastasis.





MANAGEMENT AND OUTCOME

The patient was to have polypectomy with histology of the mass and then chemo-radiation using radiotherapy to achieve a total dose delivery of 70-75Gy with both components of external beam radiotherapy (EBRT) and brachytherapy. However, due to the patient's financial constraints and lack of insurance coverage, only polypectomy with histology was initially carried out (see Figures 1-5 above). Histology revealed small round to spindle primitive cells with occasional rhabdomyoblast and tadpole cells seen, and a high mitotic count within a fibromyxoid stroma containing foci of immature cartilage with areas of hemorrhage and congested thick-walled vascular channels. Immunohistochemistry, however, was not done.

Three weekly cyclical chemotherapy was administered using intravenous (IV) Vincristine 2mg on Day 1, IV Doxorubicin 100mg on Day 1, IV Cyclophosphamide 1000mg on Day 1 regimen for three courses. IV Dacarbazine 1500mg on Day 1 was added for an additional four courses due to poor response of the tumor on mid-cycle evaluation after the first three courses, and an incidental finding of a right breast lump. There was a partial response after seven courses of chemotherapy with no change in the performance status of the patient.

Hence, total abdominal hysterectomy (TAH) with bilateral salpingo-oophorectomy (BSO) was done and the histopathology report revealed a thickened myometrial wall with a gray-white intramural nodule that measures 2.5x1.5cm and a dark brown cervix with multiple polypoid growths which on cut section shows gray-white solid appearance with multiple focal cystic spaces containing dark gritty and hard material.

The microscopic section showed an infiltrative tumor showing small round blue cells some of which show rhabdoid differentiation and admixed with large and bizarre forms in a fibromyxoid stroma with numerous abnormal mitotic figures, extensive areas of skeletal muscle differentiation with abundant heterologous elements of chondroid and osteoid tissue, the tumor extends into the endometrial cavity and entraps unremarkable glands, the ectocervical epithelium shows cambium layer with hyper and hypocellular tumor nest with premature fairly uniform round blue cells. On immunohistochemistry, the cells were strongly positive for Desmin. The histological findings were in keeping with the diagnosis of embryonal rhabdomyosarcoma. The pathological stage was pT3a, i.e., the tumor has extended to the ectocervix with myometrial invasion and extending to the lower third of the vagina without the pelvic sidewall involvement. FIGO stage IIIA (T3a, any N, M0).

We are currently awaiting further evaluation of the right breast lump identified on ultrasound during treatment. She is currently being counseled for EBRT as soon as possible.

DISCUSSION

Rhabdomyosarcoma (RMS) is an uncommon malignant tumor with a mesenchymal tissue origin. It is most commonly seen in the head and neck and rarely in the genitourinary tract, accounting for 2-5% of all adult sarcoma cases,[2] just as in this patient presented. The major subtypes of RMS are the pleomorphic, alveolar, and embryonal RMS, with the latter being the most common subtype in pediatric patients. The ERMS typically has a loss of heterozygosity of the short arm of chromosome 1. It is further divided into sarcoma botryoid or spindle variants.[3] Histological characteristics of the tumor were dependent on the subtype, although all cases demonstrated a similar immunohistochemistry profile, with RMS of the uterus having a very poor prognosis, and data regarding treatment are limited.[4] However, the patient's presentation of a right breast lump triggered the suspicion of a synchronous tumor, fibroadenoma, or other types of RMS. Primary RMS arising from the breast is exceedingly rare in adults, as sarcomas of the breast constitute less than 1% of all malignant breast tumors. To date, nine cases of primary rhabdomyosarcoma of the breast have been presented in international journals.[5] Adult ERMS that originate from undifferentiated myogenic progenitor cells are predominantly a pediatric disease.[6]

The use of chemotherapy regimens has proven to destroy cancer cells as it goes throughout the body via the bloodstream to kill cancer cells that have spread to other parts of the body. In the treatment of RMS, chemotherapy plays an important part even after surgery.

Chemotherapy can destroy tiny deposits of RMS post-surgery. Chemotherapy is given 3weekly for 6-12 courses intravenously. The combination of drugs depends on the risk groups: for the low-risk group VAC or VA (vincristine and dactinomycin(actinomycin-D) are given, for intermediate-risk group VAC/VAC/VI (Vincristine and irinotecan) is administered and its being studied whether adding targeted drug Temsirolimus to the VAC/VI regimen may be more effective. For high-risk group patients, the VAC regimen is most commonly used with the use of more intense drugs such as doxorubicin, ifosfamide, and etoposide. The use of stem cell transplant is still on trial as it has shown results for more aggressive management in pediatric cancers following chemotherapy, but its role in the management of RMS is yet to be clear as it can cause more side effects.[7]

Fertility-sparing surgery (trachelectomy/knife cone biopsy) and chemotherapy in well-selected patients with ERMS of the cervix who are premenopausal can be considered, which results in a low complication rate and excellent oncologic outcome, depending on the tumor size. Such paediatric patients should be managed by both pediatric oncologists and gynecologic oncologists.[8] However, some drugs might affect fertility or increase the risk of developing a second type of cancer, usually a form of leukemia, and years after RMS is cured. Though this is very rare, the significant benefit of chemotherapy in the treatment of RMS outweighs this risk.[7]

Our patient was not a candidate for fertility-sparing as her tumor size was large and chemoresistant, so TAH + BSO was carried out. According to the Intergroup Rhabdomyosarcoma Study Group (IRSG), fertility-sparing surgery should not be considered if a patient presents with uterine involvement and/or metastasis.[9] The histopathology report of our patient, done post-operatively, revealed endometrial cavity extension, which disqualified our patient for fertility-sparing surgery.

A study of patients found that the cases they examined were positive for myoglobin and desmin if the patient was younger than 30 years, but patients older than 40 years rarely showed such characteristics. Positive desmin, actin, and myoglobin also depend on the tumor's differentiation, having low sensitivity and specificity in the absence of good differentiation. By contrast, myogenin is considered the most sensitive and specific marker in 70-100% of cases.[10] Our patient's immunohistochemistry was strongly positive for desmin, which is uncommon, and desmin has a lower sensitivity and specificity as far as the study is concerned.

A recent study observed that DICER1-associated sarcomas exhibit recurring and characteristic histologic features, irrespective of the site of origin, with marked morphologic similarity to pleuropulmonary blastoma. The characteristic features include undifferentiated small round blue cells, spindle, foci of anaplasia (large, bizarre pleomorphic cells), and foci of rhabdomyoblastic differentiation with an expression of skeletal markers. Among the sarcoma types associated with DICER1 alteration is the ERMS of the uterine cervix.[11] These histologic features were almost similar to those of our patient. However, genetic testing was not done due to the lack of such genetic testing facilities in our environment.

Adult RMS should be generally treated aggressively with a multidisciplinary approach due to its poor prognosis as compared to children.[12] Considering that our patient presented with a rare tumor site and an uncommon presentation of a breast mass post 3 courses of chemotherapy, however, with good performance status, aggressive multidisciplinary management was an option. The treatment options depend on the stage of disease, age, patient's performance status, comorbidity, financial capability, and equipment availability. Stage IA1 disease represents micro-invasive disease, and simple hysterectomy is considered for postmenopausal women with the addition of BSO. While for premenopausal women, a knife cone biopsy performed in selective patients who want to retain fertility. Lymph node involvement has been reported in 7.4% of stage IA2 patients, so modified radical hysterectomy with lymph node dissection is offered to them. However, fertility-sparing options for these patients include radical trachelectomy if the tumor size is 2cm with no lymphovascular space involvement. Radical radiotherapy is done for unfit patients or patients who decline surgery. For Stage IB1-IIA (tumor < 4cm) disease, surgery + radical radiotherapy, with post-operative cisplatin-based chemotherapy, is given to high-risk patients. For stage IB2-IVA patients, the standard treatment is concurrent chemo-radiation (EBRT, cisplatin-based chemotherapy, and brachytherapy). For Stage IVB disease, palliative treatment is offered, which is aimed at improving the quality of life of the patients. Our patient, being diagnosed with stage IIIA disease, with surgical intervention, her standard management should have been cisplatin-based concurrent chemoradiation if no challenges were encountered.

Radiotherapy plays a key role in eradicating residual disease following surgery. Modern radiation techniques like 3D-conformal radiation therapy (3D-CRT) and intensity-modulated radiation therapy (IMRT) offer precise tumor targeting, minimizing dose to surrounding normal tissues.[7] A total dose of 45–50.4 Gy (1.8–2 Gy per fraction over 25–28 fractions) is administered over 5 to 5.5 weeks using a four-field box technique. When lymph node involvement is confirmed radiologically or surgically, radiation should include the internal iliac, external iliac, and obturator regions.

Brachytherapy provides high-dose internal radiation focused on the tumor, enhancing local control while sparing nearby organs (bladder, rectum, small intestines). The additional 20–30 Gy from brachytherapy boosts the total dose to 70–75 Gy in combination with EBRT. Radiation doses in brachytherapy are defined using the Manchester system of implant dosimetry, with reference points A and B. These doses may be delivered using low-dose rate (LDR: 0.4–2 Gy/hr), medium-dose rate (MDR: 2–12 Gy/hr), or high-dose rate (HDR: >12 Gy/hr) techniques. The Manchester system applies the inverse square law to distribute radiation, utilizing gynecological applicators composed of a tandem (inserted through the uterine canal) and two ovoids or colpostats (placed in the right and left vaginal fornices) Point A is defined as 2 cm superior to the external

cervical os and 2 cm lateral to the central uterine canal (tandem)—it typically receives about 80% of the prescribed radiation dose. Point B is located 2 cm superior to the cervical os and 5 cm lateral to the tandem, i.e., 3 cm lateral to point A, lying at the same horizontal level. These definitions are based on the work of Tod and Meredith (1953). Additional reference points, Point C and Point D are positioned 2 cm above points A and B, respectively, along the median and lateral lines of reference.[14] The bladder point is determined after a Foley catheter is inserted and inflated with 7 mL of contrast medium, then secured against the urethra. It is identified as the center point on the anteroposterior (AP) radiograph and the most posterior point on the lateral radiograph. The rectal point is located 0.5 cm posterior to the most distal face of the ovoid on the lateral film, and at the midpoint between the ovoids along the tandem on the AP film. Our patient would have benefited from this treatment either palliatively or to prevent disease progression.

Unfortunately, widespread metastases usually appear early in the course of the disease and lead to a rapid death; this makes accurate evaluation of any therapeutic regime difficult.[15] Favorable prognostic parameters, such as localized disease without myometrial invasion, a single polyp, and embryonal histologic subtype, are effectively treated by surgery, but unfavorable prognostic parameters benefit from surgery, adjuvant chemotherapy, and radiotherapy.[16] Multimodal therapy in cervical rhabdomyosarcoma appears to be associated with a good prognosis. Favorable prognostic factors such as early-stage at diagnosis and favorable histological subtype may contribute to excellent observed survival.[17] Despite our patient's ECOG-1 status, younger age (43 years), and access to functional radiotherapy, negative factors, including chemoresistance, tumor progression, financial constraints, and limited access to radiotherapy centers, undermine her prognosis.

CONCLUSION

Although ERMS typically presents in pediatric patients, its occurrence in adults—especially with classic features—raises significant clinical concerns due to limited treatment options. The appearance of a cervical polyp at any age warrants careful evaluation to exclude RMS. Early diagnosis significantly improves prognosis, especially in younger patients. Aggressive, multimodal treatment—including surgery, chemotherapy, and radiotherapy—should be considered for adult ERMS. While outcomes vary depending on favorable and unfavorable prognostic factors, clinical trials investigating novel therapies may offer additional hope for affected patients.

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