

Case Report

Labia Majora, an unusual site presentation of Alveolar Rhabdomyosarcoma in a young girl: A case report and review of literature

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Abstract

Rhabdomyosarcoma is the most common soft tissue sarcoma in children under the age of 15 years, constitutes 5% to 8% of all solid tumors in the pediatric population. The most common RMS is the embryonal subtype, followed by the alveolar subtype. The alveolar subtype typically affects older children and young adults and frequently involves the extremities and perineal sites with peak at 10-25 years. Alveolar RMS of labia majora is extremely rare.

Keywords: Labia Major, Rhabdomyosarcoma (RMS), Alveolar Rhabdomyosarcoma (ARMS), vulva

1. Introduction

Rhabdomyosarcoma (RMS) is the most common soft tissue sarcoma in children under the age of 15 years, constitutes 5% to 8% of all solid tumors in the pediatric population. It is the third most common extracranial childhood solid tumor after neuroblastoma and Wilms' tumor¹. The most common site is head neck (44%), followed by parastemal region, retroperitoneum (20.4%) and extremities (14.6%). The most common RMS is the embryonal subtype, followed by the alveolar subtype. The alveolar subtype typically affects older children and young adults and frequently involves the extremities and perineal sites² with peak age at 10-25 years. The vulvar site Alveolar Rhabdomyosarcoma (ARMS) is extremely rare and only a few cases have been reported in literature³. We are reporting a case of alveolar RMS of Labia Majora (Vulva) in a young girl.

2. Case Presentation

A 3 year female child presented with 2X2 cm, firm and tender swelling at left half of labia majora at 3 o'clock position of 12 months duration. It became painful and increase in size since last 2 months. The fine needle aspiration cytology was reported as poorly differentiated malignant neoplasm likely to be sarcoma. Excision biopsy with wide margin was done. Gross Examination, a single nodular 3X2X1 cm size, firm grey white with presence of hemorrhagic area (0.5X0.2) and histopathology shows malignant undifferentiated cells arranged in alveolar pattern (Figure 1), Desmin positive, immunonegative for 2100, SMA, CD34 diagnosed as Alveolar Rhabdomyosarcoma. ARMS was staged as stage 2 (unfavorable site, T1, < 5cm, N0, M0), Group I with Intermediate risk (stage2, group I, alveolar histology) as per classification of the Soft Tissue Sarcoma Committee of the Children's Oncology Group. Postoperatively USG abdomen & pelvis, CT scan abdomen & Pelvis were within normal limits. After discussion between Surgeon and Radiation Oncologist decided to offer adjuvant treatment i.e. chemotherapy followed by local radiotherapy. She received VID (vincristine, Ifosfamide and Actinomycin D) six cycles chemotherapy at 3 weeks intervals followed local radical interstitial brachytherapy. The interstitial brachytherapy was given as two layer implant with high Dose Rate (HDR) to dose 31.5 Gy in 9# over 4.5 days. The patient is doing well with disease free in 3 years follow up (Figure2).

Figure -1: H&E X 10x microphotograph shows malignant undifferentiated cells arranged in alveolar pattern suggestive of alveolar RMS

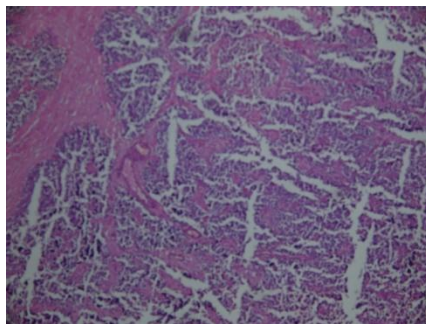


Figure - 2: Follow-up picture after 3 years showing scar mark of surgery



3. Discussion

The genitourinary tract is a common location for RMS in children and RMS usually originate deep to the introitus commonly in the vagina and less commonly in the vulva. Nearly 80% of genitourinary tracts RMS are embryonal in nature. The alveolar subtype is more prevalent among patients with less favorable clinical features (younger than 1 year or older than 10 years, extremity primary tumors, and metastatic disease at diagnosis) and Alveolar RMS in the perineal location is predominantly seen in the adolescent age group. Compared with the embryonal form, alveolar RMS carries a more aggressive clinical course and a poorer prognosis and significantly worse outcomes with regional lymph node involvement.⁴

ARMS characteristically presents as a painless or mildly tender mass and is frequently misdiagnosed as a cyst of the Bartholin duct.⁵ The present case presented as a painless swelling in Labia Majora. The histopathological diagnosis of ARMS is a delicate task and exhibit nonspecific characteristics similar to those of other neoplasm. It requires staining methods that are more specific than histopathological examinations of Hematoxylin / Eosin (H&E) stained specimen particularly when the tumor is poorly differentiated. In present case the diagnosis of ARMS was clearly made on staining with H & E.

Assessment of disease extent is critical, because therapy and prognosis depend on the degree to which the mass has spread beyond the primary site. The management of ARMS is very specific and requires multidisciplinary team approach to achieve an optimal survival and quality of life and as per the protocol based on Risk Group assignment⁶. Dramatic improvements in survival have been achieved for children and adolescent after a proper standard treatment⁷. The reported patient disease assessment was stage 2 (unfavorable site, T1, < 5cm, N0, M0), Group I with Intermediate risk (stage2, group I, alveolar histology).

RMS is mostly curable with localized disease who receives a combined modality treatment, with more than 70% surviving 5 years after diagnosis. Patients with smaller tumors (≤ 5 cm) have improved survival compared with children with larger tumors. Recurrence / relapses are very uncommon after 5 years of disease free survival, with a 9% late event rate at 10 years⁸. Outcome is primarily related to the use of multimodality therapy; all patients require chemotherapy and at least 85% also benefit from radiation therapy, with favorable outcome even for those patients with nonresectable disease. Initial radical surgery is not indicated for rhabdomyosarcoma of the vulva/vagina/uterus but the conservative surgical intervention for vaginal rhabdomyosarcoma, with primary chemotherapy and adjunctive radiation (often brachytherapy) for residual disease (Group II or III), results in excellent disease-free survival.⁹

The treatment and prognosis depend on the histology and molecular genetics of the tumor, it is necessary that the tumor tissue be reviewed by pathologists and cytogeneticists / molecular geneticists with experience in the evaluation and diagnosis of tumors in children. Desmin is the best single marker for RMS cells. ARMS have a specific chromosomal aberration demonstrable with molecular genetics methods and immunohistochemistry confirm the diagnosis with age and site predilection. Unique translocations between the FOXO1 (previously called FKHR) gene on chromosome 13 and either the PAX3 gene on chromosome 2 [t(2;13)(q35;q14)] or the PAX7 gene on chromosome 1 [t(1;13)(p36;q14)] are found in 70% to 80% of patients with alveolar histology tumors.¹⁰

Patients with localized disease who have an excellent chance of cure, the development of less toxic therapy has the potential for decreasing long-term morbidity and reducing the risk for secondary neoplasm so in our case we use the basic chemotherapy regimens i.e. VID followed by localized Radiotherapy i.e. Interstitial Brachytherapy as the patient was young.

In conclusion the present case illustrates the difficulties may occur in the diagnosis of RMS and emphasizes the importance of correct initial management for best prognosis of patient as per the standard guidelines.

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