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Congenital Primary Cutaneous Rhabdomyosarcoma of the Perineum with Mixed Histopathological Features and Unusual Expression of Smooth Muscle Actin: Report of a Case

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Case Report

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Abstract

Rhabdomyosarcoma (RMS) affecting the soft tissue of the perianal and perineal regions in children is uncommon and account for 2% of all rhabdomyosarcomas. Primary cutaneous presentation in this anatomical location is extremely rare. We report a 9-month-old female infant with a congenital primary cutaneous RMS presenting as a polypoidal exophytic mass in the perineum mimicking giant condyloma acuminatum (Buschke and Löwenstein tumor). Histopathological examination revealed a predominantly alveolar RMS with areas showing embryonal and pleomorphic features. In addition to expressing desmin and myogenin, the tumor cells also expressed smooth muscle actin.

Keywords: Rhabdomyosarcoma; Soft Tissue; Visceral Organs; Tumors; Embryonal

Introduction

Rhabdomyosarcoma (RMS) is a malignant mesenchymal neoplasm that arises from mesenchymal precursors of striated (skeletal) muscles [1]. It typically arises in deep soft tissue or in visceral organs and thus seldom presents to dermatologists. It represents 53% of soft tissue tumors in the pediatric population [2]. Histopathologically, RMS includes three main types: embryonal (including botryoid, spindle and anaplastic variants), alveolar and pleomorphic with the first two types being the most commonly encountered types [3-5]. While the embryonal type tends to involve the soft tissues or viscera of the head and neck or the genitourinary tract, the alveolar type tends to affect the deep soft tissue of extremities [6]. Rare histopathological types of RMS include a distinctive spindle cell variant [7] and an epithelioid variant [8].

Compared to conventional RMS, tumors presenting in the skin are extremely rare and were found to represent 0.7% of all RMS cases collected at two large specialized institutes [9]. On the other hand, among all cases of cutaneous malignant solid tumors (primary or metastatic) studied in a Spanish pediatric dermatology department over duration of 14 years, cutaneous RMS represented 32% of the cases [10]. The skin can be involved by RMS either as a primary or a metastatic event. Primary cutaneous cases are very rare with less than 50 cases reported so far in the literature [11]. Cases are classified as primary cutaneous only after clinical and radiological exclusion of metastasis from RMS elsewhere or extension to the skin from an underlying deep soft tissue lesion.

Primary cutaneous rhabdomyosarcomas (PC-RMS) tend to affect both children and adults with a bimodal age

distribution [12]. While conventional RMS has a slight male predominance, PC-RMS tends to be more common in females [11]. Among pediatrics, the face tends to be the most frequently affected site with PC-RMS [1,2,9,12-15]. Other rarely involved sites include the hand [16], chest [17], feet [12], anus [18,19], and the perineum [20]. Congenital cases are rare [2,20,21]. Among pediatrics, PC-RMS is usually of the alveolar or embryonal types while in adults most primary cutaneous cases are pleomorphic, epithelioid or not otherwise specified (RMS-NOS) [12].

Immunohistochemically, all types of RMS show skeletal muscle differentiation and are at least focally positive for desmin, muscle specific actin (HHF-35), myogenin and myo D1 with the last two being the more specific diagnostic markers [3,4].

Herein we report a female infant with congenital primary cutaneous rhabdomyosarcoma affecting the perineum and showing mixed histopathological features and unusual strong positive staining with smooth muscle actin.

Case Report

A 9-month-old female infant presented to the outpatient clinic of the Dermatology Department of Al-Zahraa Hospital (Faculty of Medicine for Girls, Al Azhar University) with a perineal mass. The mass was present at birth and underwent rapid progressive increase in size over time with history of spontaneous bleeding. Examination revealed a large (8X12 cm) polypoidal exophytic firm mass with a lobulated pink to dusky erythematous surface showing focal ulceration (Figure 1). Other than the mass, her medical history and clinical examination were unremarkable. A suggested clinical diagnosis of giant condyloma acuminatum (Buschke and Löwenstein tumor) was done and the patient was referred to the Pediatric Surgery Department for excision of the mass. Histopathological examination revealed: hyperplastic focally ulcerated epidermis and a dermal neoplastic infiltration. The tumor extended to the underlying subcutaneous fat and reached the lower margin of the specimen. In the majority of the lesion, the neoplastic growth was composed of discrete nests separated by fibrous septa (Figure 2a). Although most of the nests revealed solid growth pattern, some showed characteristic central cellular dissociation (Figure 2b).

The nests were made of large cells with pleomorphic vesicular nuclei and contained many easily recognizable rhabdomyoblasts with abundant esinophilic cytoplasm and characteristic 'strap" or "tadpole" appearance (Figure 3 a&b). Many multinucleated cells were detected within the nests (Figure 3c), some with wreath like morphology. In some parts of the tumor the growth appeared as small rounded to spindle-shaped undifferentiated cells arranged in loose myxoid stroma with scattered identifiable rhabdomyoblasts (Figure 4). Focally, aggregates of large number of bizarre spindle shaped cells admixed with readily identifiable polygonal and pleomorphic rhabdomyoblasts were identified (Figure 5). The morphological features were typical of RMS with a predominant alveolar morphology admixed with areas of embryonal and pleomorphic features. The diagnosis was confirmed by the diffuse positive staining of tumor cells with myogenin and desmin (Figure 6). The tumors cells showed also strong diffuse positivity with smooth muscle actin (SMA) (Figure 7). All photomicrographs presented are according to their original magnification.

Following diagnosis the patient was referred to the Cancer Institute for further assessment, re-excision and further treatment. Radiological workup revealed no lesions elsewhere and no extension in the perineal soft tissue.

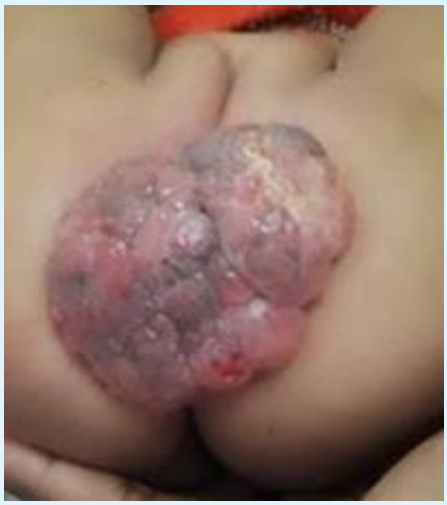

Figure 1: Large exophytic polypoidal mass with lobulated focally ulcerated surface affecting the perineum.

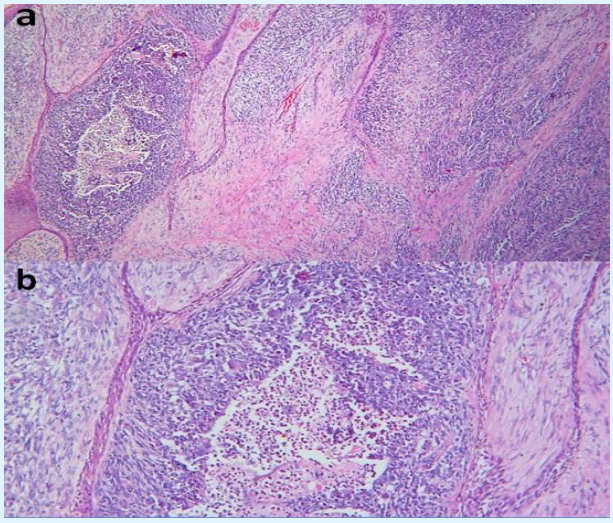

Figure 2: Photomicrograph showing alveolar RMS presenting with discrete nests of blue cells separated by fibrous septa (a, H&E X40). One of the nests shows cellular dissociation (b, H&E X100).

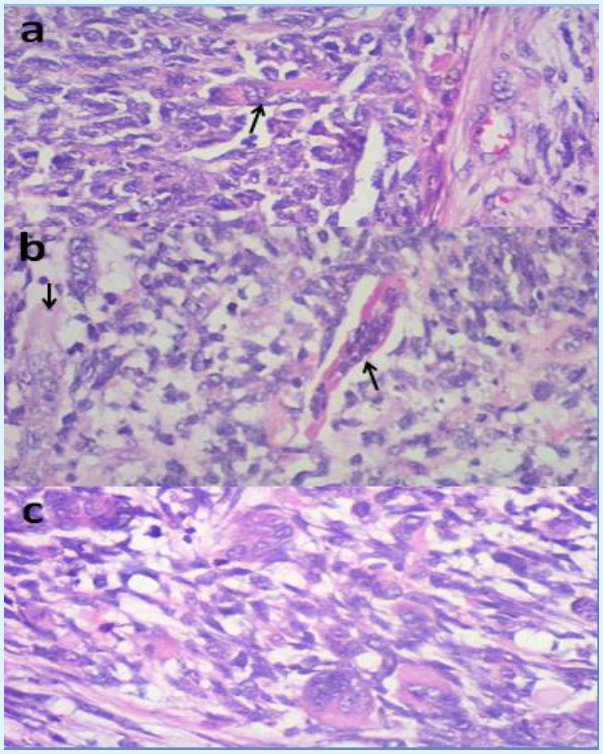

Figure 3: Photomicrograph showing cellular details of the alveolar areas: characteristic rhabdomyoblasts with "tadpole" and "strap cell" appearance (a & b respectively, H&E X400, black arrows) and many multinucleated giant cells (c, H&E X400).

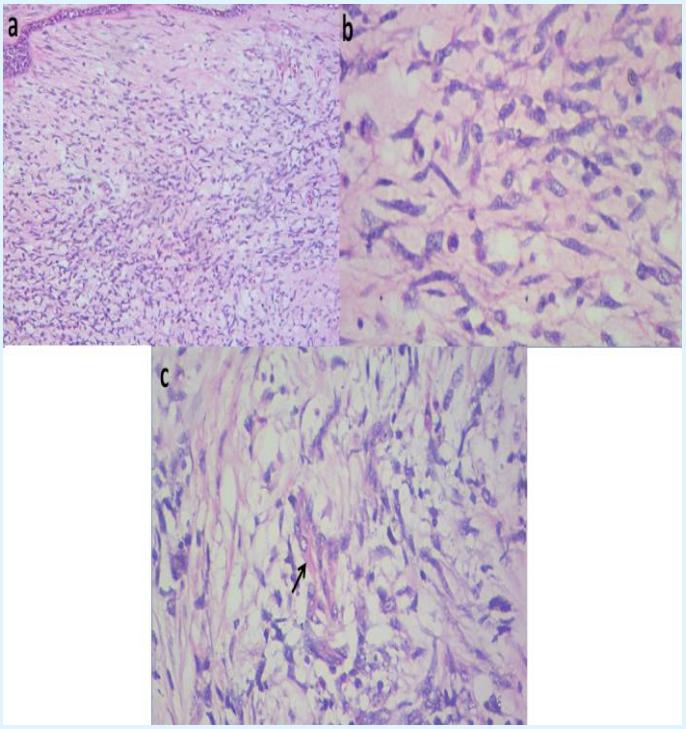

Figure 4: Photomicrograph showing areas with embryonal features: small round to spindle cells set in a myxoid stroma (a, X40; b, H&E X100). Rhabdomyoblasts are identified (c, H&E X100-black arrow).

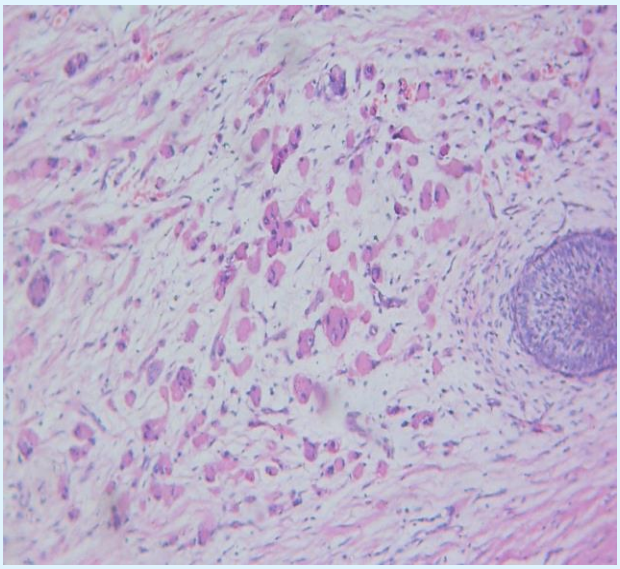

Figure 5: Photomicrograph showing an area with pleomorphic features: bizarre spindle shaped cells admixed with readily identifiable polygonal and pleomorphic rhabdomyoblasts (H&E X100).

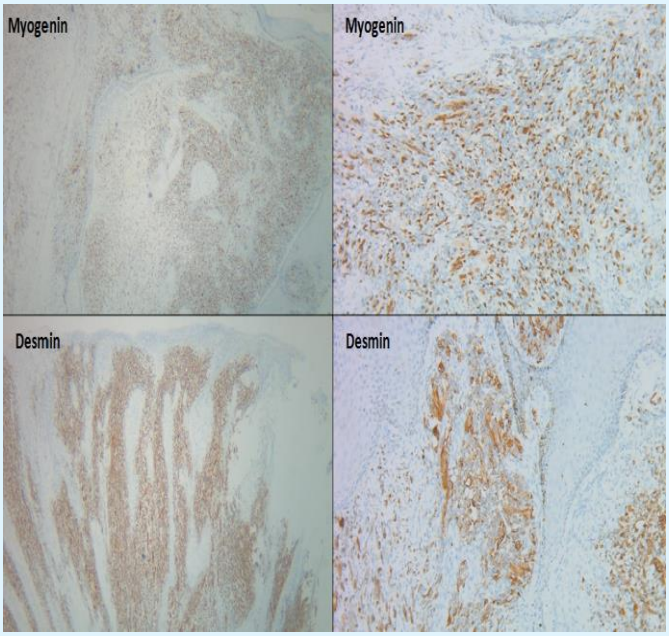

Figure 6: Photomicrograph showing immunohistochemical features: tumor cells were positively stained with myogenin and desmin (Myogenin and Desmin immunostain X40 and X100).

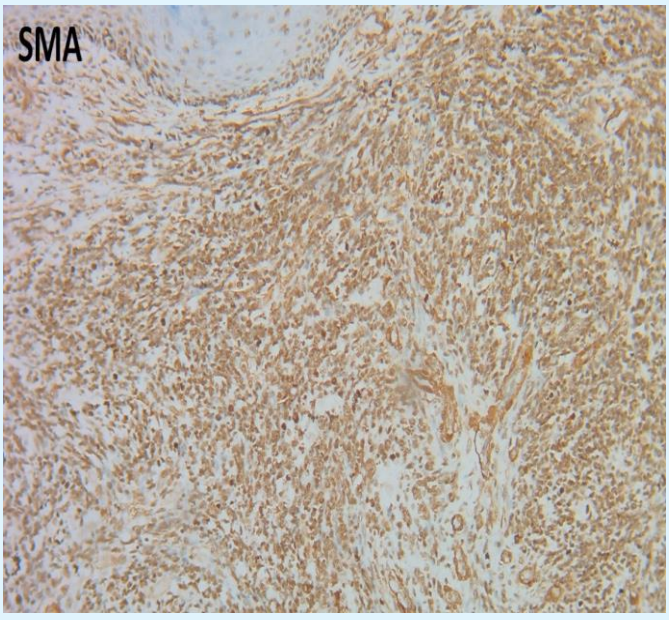

Figure 7: Photomicrograph showing immunohistochemical features: tumor cells were strongly diffusely positive with SMA (SMA immunostain X100).

Discussion

Rhabdomyosarcoma affecting the soft tissue of the perianal and perineal regions in children is uncommon and account for 2% of all rhabdomyosarcomas [22]. On the other hand, primary cutaneous presentation in this anatomical location is extremely rare. Primary cutaneous embryonal RMS presenting with polypoidal masses mimicking condyloma acuminatum have been reported in the anus [18,19]. Moreover, Gong et al. [20] reported a newborn with abnormal symmetrical perineal overgrowth causing ambiguous genital morphology and histologic examination revealed cutaneous alveolar RMS admixed with areas of embryonal and pleomorphic RMS features. Only a relatively small number of mixed-type RMS have been reported and are usually associated with poor prognosis. They usually affected soft tissue and visceral organs [23-25].

The presence of many readily identifiable rhabdomyoblasts with characteristic 'strap cell' or 'tadpole' morphology in our case was typical of RMS on morphological level. Further immunohistochemical studies confirmed the diagnosis. However, in undifferentiated cases with undetectable rhabdomyoblasts, the diagnosis RMS can be difficult and requires extensive immunohistochemical workup. In children, the alveolar type can be mistaken with other tumors of the "small round blue cell" category such as Ewing sarcoma/primitive neuroectodermal tumors, metastatic neuroblastoma, hematopoeitic malignancies and extra renal Wilm's tumor, while in adults Merkel cell carcinoma and small cell carcinoma of the lung can also be a differential diagnosis. The embryonal type can be mistaken with leukemia cutis and the pleomorphic variants of RMS should be differentiated from other pleomorphic tumors with rhabdoid features such as: atypical fibroxanthoma, superficial pleomorphic undifferentiated sarcoma, proximal type epithelioid sarcoma, epithelioid angiosarcoma, high grade synovial sarcoma, melanoma, metastatic carcinoma and sarcomatoid squamous cell carcinoma [12,26-30].

An unusual finding in our case was the presence of strong diffuse SMA expression by tumor cells. Although this finding has been previously reported in literature in pleomorphic rhabdomyosarcomas [31-34], the positivity with SMA in our case was diffuse throughout the tumor and not restricted only to the pleomorphic areas. The exact significance of this SMA expression is unknown and whether it indicates a true smooth muscle differentiation

or just an anomalous immune reactivity could not be verified as ultrastructural studies were not feasible.

Similar to conventional soft tissue RMS, primary cutaneous forms are also aggressive and can lead to distant metastases, usually to the lung. According to the largest available series of PC-RMS, the mortality rate was estimated to be 36% [12]. This necessitates rapid diagnosis and evaluation for metastasis. The treatment of PC-RMS usually includes a multidisciplinary approach with a combination of surgical excision, chemotherapy and radiotherapy [12]. Awareness of this rare tumor and its variable presentations is important to allow for early diagnosis and proper treatment.

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