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Congenital Anaplastic Rhabdomyosarcoma Presenting As Abdominal Wall Mass

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KEY WORDS

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ABSTRACT

Rhabdomyosarcoma encompasses a group of malignant myogenic neoplasms expressing a multitude of clinical and pathological diversities. It is the commonest soft tissue sarcoma of childhood but neonates are rarely affected. Embryonal subtype is the most frequent. Head-neck and genitourinary tracts are predominant sites, while trunk is considered among the unusual sites of rhabdomyosarcoma. Herein we report a case of anaplastic rhabdomyosarcoma in a newborn girl presenting, at the Pediatric Surgery Outpatient Department of North Bengal Medical College and Hospital, India in 2013 with a large tumor mass in the left flank region, arising from abdominal wall muscles.

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Introduction

Rhabdomyosarcoma (RMS) is the most prevalent malignant soft tissue tumor among children. The embryonal subtype is more common during the first decade of life, whereas alveolar RMS (ARMS) frequently arises between 10 to 25 yr of age (1). Only 6% of all RMS cases occur in infancy (2, 3). RMS arising before the age of one month is termed as congenital RMS (4). Congenital RMS represented 0.4% of all RMS cases in Intergroup Rhabdomyosarcoma Study (IRS) documents (5). In German and French series, its incidence prevailed at 1.7% and 2.9% respectively (3, 6). RMS has a special predilection for head and neck region, genitourinary tract and extremities. Truncal involvement is relatively rare, observed in 12% cases only (7).

Of the various subtypes of RMS, the anaplastic variant is extremely rare and prognostically unfavourable as well. Histologically, in addition to classic embryonal or alveolar histology, presence of bizarre rhabdomyoblasts and atypical mitoses characterizes this variant (8). Congenital experience of this unusual variant could not be retrieved from previous literatures, even after diligent search.

Here we present a rare case of congenital anaplastic rhabdomyosarcoma originating from abdominal wall in a three day old child, who

succumbed to death following surgical removal of the mass.

Case Report

A full-term newborn girl in the third day of her life was brought by her parents at the Pediatric Surgery Outpatient Department of North Bengal Medical College and Hospital, India in 2013 with a large 7cm ˣ 7cm ˣ 5cm mass involving the left flank and extending towards the posterior aspect of abdomen. Skin over the mass appeared tense and inflamed (Fig. 1). The mass was firm to hard in consistency, tethering to abdominal wall. No other morphological abnormalities could be elicited on her physical examination. Abdominal skiagram revealed a homogeneous mass density involving the part, without a definite ascertainment towards its site of origin (Fig. 2A). Magnetic resonance imaging (MRI) of her abdomen depicted the mass to be related with the skin, subcutaneous tissue and abdominal wall muscles without any evidence of visceral invasion (Fig. 2B & 2C).

Fine needle aspiration cytology (FNAC) from the mass demonstrated the presence

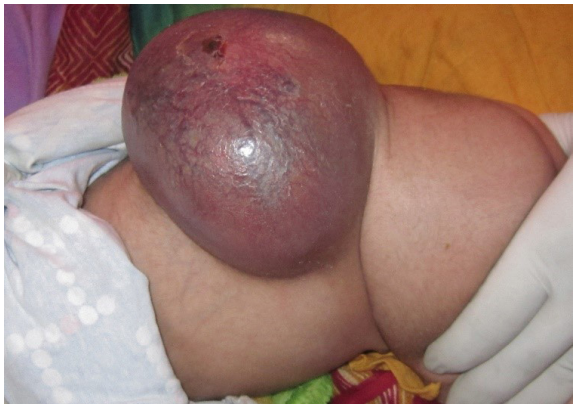

Fig. 1
Congenital rhabdomyosarcoma: large globular mass arising from left flank of abdomen

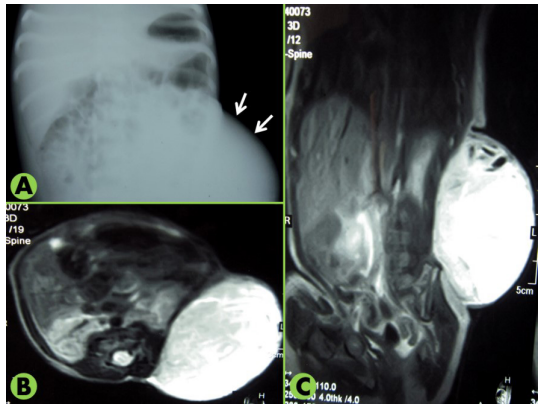

Fig. 2
Congenital rhabdomyosarcoma: abdominal X-ray revealed a homogeneously dense mass (arrows) involving left flank of abdomen (A); MRI confirmed the mass to be arising from parietal abdominal wall constituents (B & C)

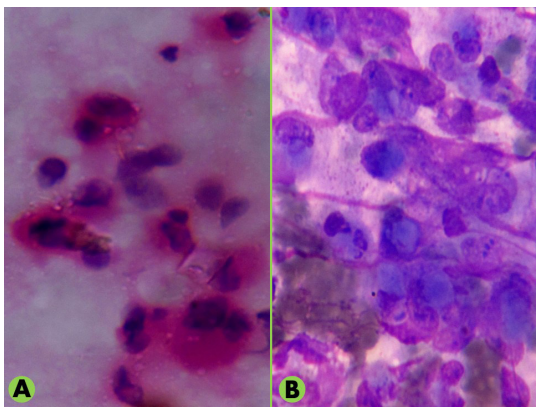

Fig. 3
Rhabdomyosarcoma: cytologically, mono-nucleated and bi-nucleated characteristic rhabdomyoblasts in dispersed population admixed with undifferentiated small round cells [H&E stain, 400x (A); Leishman stain, 400x (B)]

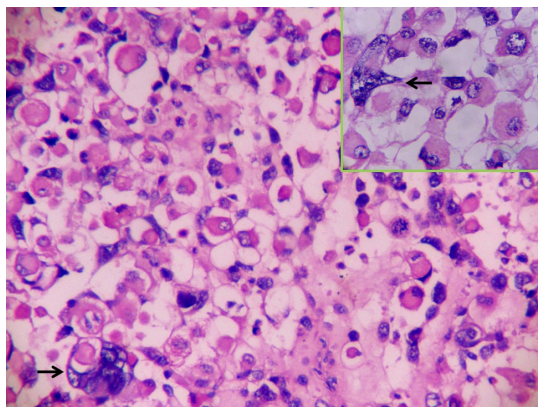

Fig. 4
Anaplastic rhabdomyosarcoma: histologically, few pleomorphic, bizarre rhabdomyoblasts (arrows) interspersed between classical ERMS morphology [H&E stain, 100x; inset: H&E stain, 400x]

of characteristic well-differentiated rhabdomyoblasts, having round-shaped one-to-two eccentric nuclei and abundant dense inclusion-like cytoplasm; admixed with few undifferentiated small round tumor cells (Fig. 3). Considering these cytological evidences the neoplasm was confidently diagnosed as embryonal RMS (ERMS).

Subsequently the mass was completely excised under high-risk prerequisites. Histopathologically, the tumor was relatively circumscribed and separated from the overlying thinned-out epidermis by a band of fibroconnective tissue, infiltrated with lymphoplasmacytic cells. The neoplasm was composed of uniformly distributed, characteristic, round-to-oval, eosinophilic rhabdomyoblasts in diffuse sheets; featuring cytoplasmic dense granularity or deep eosinophilic stringy material concentrically juxtaposed to its regular round nuclei, having vesicular chromatin and prominent nucleoli. Few excessively large, pleomorphic rhabdomyoblasts with markedly irregular, hyperchromatic and often complexly lobulated nuclei alongside distinctive rhabdomyoblastic cytoplasm, were conspicuous (Fig. 4). The pre-operative cytological diagnosis was modified into anaplastic RMS. All the margins of the mass were found to be free from any neoplastic involvement.

Despite the hastened attempt to improve the child's morbid condition by resecting the mass, she ultimately breathed her last on third post-operative day.

An informed consent form was taken from the patient's parents.

Discussion

RMS is a malignant soft tissue tumor, expressing skeletal muscle differentiation (7). It represents 4-8% of all malignant solid neoplasms among children. Median age at its diagnosis is 5 yr. Approximately two-thirds of all cases manifest before the first decade of life. Adults are rarely affected and only 2% of these patients present at birth (9). In a large study of 3217 RMS patients; the IRS encountered only 0.4% of neonates in their first 30 days of life, while males were the dominant cohort amongst them (5).

RMS most often arises in head and neck, genitourinary tract, retroperitoneum, and extremities. Trunk is involved in only 12% instances of RMS (7). Two rare instances of thoracic wall presentation were described in congenital RMS (10, 11). In contrast, the discussed newborn girl in this illustration presented sharp at the age of three days with a large mass in the left flank of abdomen.

MRI is the best imaging modality in RMS due to its excellent ability to determine soft-tissue changes. CT scan is useful in assessing any metastatic manifestation from RMS (10). Considering the current patient, MRI confirmed the abdominal wall constituents as origin of the mass, ruling out any visceral involvement. Corresponding CT scan further excluded any visceral metastasis.

Histopathologically, RMS is primarily classified into embryonal, alveolar and pleomorphic variants (12). Anaplastic RMS is a special entity, presents at a median age of 40 months (13). It is characterized by focal presence of pleomorphic rhabdomyoblasts intertwined commonly between classic embryonal or rarely with alveolar histomorphology; whereas pleomorphic RMS predominantly affects adults and is comprised almost exclusively of bizarre rhabdomyoblasts without any tendency towards better differentiation (1, 8). Previously anaplastic RMS was stratified as ARMS or ERMS with anaplastic features (1); however, because of the unfavorable prognostication compared to classic ARMS or ERMS, these cases are now separately grouped as anaplastic RMS (8).

Classic cytomorphology of ERMS has been described as predominantly dissociated population of undifferentiated rounded or spindle cells with variable admixture of triangular,

strap-shaped or ribbon-like well-differentiated rhabdomyoblasts having eccentric nuclei and dense cytoplasm (14). Because of the varying cellular distribution in different regions of a RMS mass, the pleomorphic rhabdomyoblasts from anaplastic RMS often fail to be represented in the aspirated smears (15). Recapitulating similar phenomenon, the aspirates in present case yielded equivocal distribution of undifferentiated and well-differentiated rhabdomyoblasts in absolute absence of any pleomorphic rhabdomyoblasts. Under these cytomorphological considerations the mass was diagnosed as ERMS; however, histopathology revealed the presence of pleomorphic multinucleated rhabdomyoblasts in conjugation to classic ERMS morphology. Finally, the resected mass from the discussed patient was rendered the diagnosis of anaplastic RMS.

Before the immunohistochemical (IHC) era, phosphotungstic acid hematoxylin (PTAH) and iron-hematoxylin special stains were frequently implicated to complement the striated muscle derivation of RMS. Subsequently, numerous adequately sensitive IHC markers; like desmin, muscle-specific actin, and myoglobin; were isolated. However, their diagnostic value was soon undermined by substantial lack in specificity (1).

In this context, Wang et al. demonstrated nuclear expression of MyoD1 and myogenin with comprehensive sensitivity and specificity, which are presently the most widely accepted IHC markers for RMS (16). Ultrastructural visualization of cytoplasmic myofilaments also supports its myogenicity (1). However, the presence of characteristic well-differentiated rhabdomyoblasts, in examined smears and sections, clinched the diagnosis of RMS in reported case, nullifying the necessity of any additional diagnostic work-ups.

Differential considerations of present case, exhibiting exuberant dominance of well-differentiated rhabdomyoblasts, include an array of malignancies with focal heterologous rhabdomyoblastic differentiation: epithelial neoplasms like carcinosarcoma; sarcomas like dedifferentiated liposarcoma; or neuroectodermal tumors like malignant peripheral nerve sheath tumor (malignant Triton tumor) (17). Difficult instances often demand prompt deployment of specific myogenic IHC markers (1). The diffuse distribution of well-defined rhabdomyoblasts in the discussed neoplasm readily depicted its rhabdomyoblastic phenomena, dismissing the need of any further ancillary tests, and virtually ruled out the possibility of any divergent neoplasms.

Depending upon the size of tumor, various treatment modalities have been described as combined brachytherapy and chemotherapy without surgery, only surgery, chemotherapy with surgery and triple therapy comprising of chemotherapy, and surgery as well as radiotherapy (18). Peripheral stem cell support is associated with longest survival among RMS patients (19). However due to her extreme age, the current patient failed to cope with the stress exerted by extensive surgical procedure and succumbed to it.

Conclusion

The present case exemplifies that abdominal wall is an extremely rare site for congenital RMS. Of the various histological variants, anaplastic RMS is an uncommon subtype. Diagnostic dilemma frequently occurs with cytological preparations. In this setting, radiological findings must be corroborated with representative histopathological findings, to substantiate definitive diagnosis.

Conflict of interest

The authors declare that there is no conflict of interests.

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