

Neonatal rhabdomyosarcoma: an unusual presentation

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Abstract: Though rhabdomyosarcoma (RMS) is the most common soft tissue sarcoma in the paediatric age group, its occurrence in new born is rare. Often it presents as a large mass arising from anywhere in the body, most common site being head and neck region. Here we are describing a case of neonatal embryonal RMS with multiple subcutaneous nodules.

Keywords: Rhabdomyosarcoma (RMS); paraneonate; embryonal

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Introduction

Rhabdomyosarcoma (RMS) is a malignant mesenchymal neoplasm that exhibits striated muscle differentiation. It is the most common soft tissue sarcoma of childhood, accounting for 5-8% of all malignant solid tumors in children and third most common extracranial solid tumour of childhood after neuroblastoma and Wilms tumour (1). On the basis of histology RMS has five recognized types: embryonal, botryoid variant, spindle cell variant, alveolar and undifferentiated (2).

We are presenting a rare case of RMS with two very unusual features. First, the patient was a paraneonate and secondly presented as multiple subcutaneous nodules. Multiple skin nodules are present in many malignant and non-malignant conditions also, so it was very difficult to diagnose.

Case

A male baby was born of non-consanguineous marriage to a primigravida mother with normal birth weight. Perinatal period was uneventful. Mother was an unbooked case with no proper antenatal care. Since time of birth parents noticed a small neck swelling along with multiple tiny small swellings all over the body. As a baby was apparently asymptomatic, not much attention was paid to these swellings. After few days mother noticed progressive increase both in size and number of these swellings,

especially swelling over neck and eyes.

In our hospital, a patient was brought at the age of two months, with large neck swelling and multiple small swellings all over body and unilateral proptosis (*Figure 1A,B*). There was no history of hoarseness, dysphagia, epistaxis, unilateral nasal discharge, chronic suppurative otitis media, blood tinged ear discharge, hematuria, urinary retention, constipation, abdominal mass or jaundice. On examination, there was a very large 9 cm × 8 cm swelling in the right cervical region extending from right pinna to the midline in suprasternal region. There were also multiple small few mm to few cm sizes nodular, pinkish blue and purple color, shiny subcutaneous swellings all over body including eye swelling, giving the baby a blueberry muffin appearance.

Laboratory investigations like complete blood count, serum electrolytes, serum creatinine and liver function test were within normal limits. X-ray chest and ultrasonography of abdomen showed no organomegaly and no other intrathoracic or intra-abdominal mass respectively. Such disseminated disease was ruled out.

Biopsy from right para umbilical swelling was taken. Pathological findings in the hematoxylin and eosin sections showed a nodular malignant round cell tumor infiltrating the superficial and deep dermis. The tumor cells showed round nuclei with vesicular nuclear chromatin, prominent nucleoli and eosinophilic cytoplasm, which in some of the cells imparted a distinct rhabdoid look. Intermixed with these tumor cells were seen 'wreath-like' giant cells (*Figure 1C-F*).

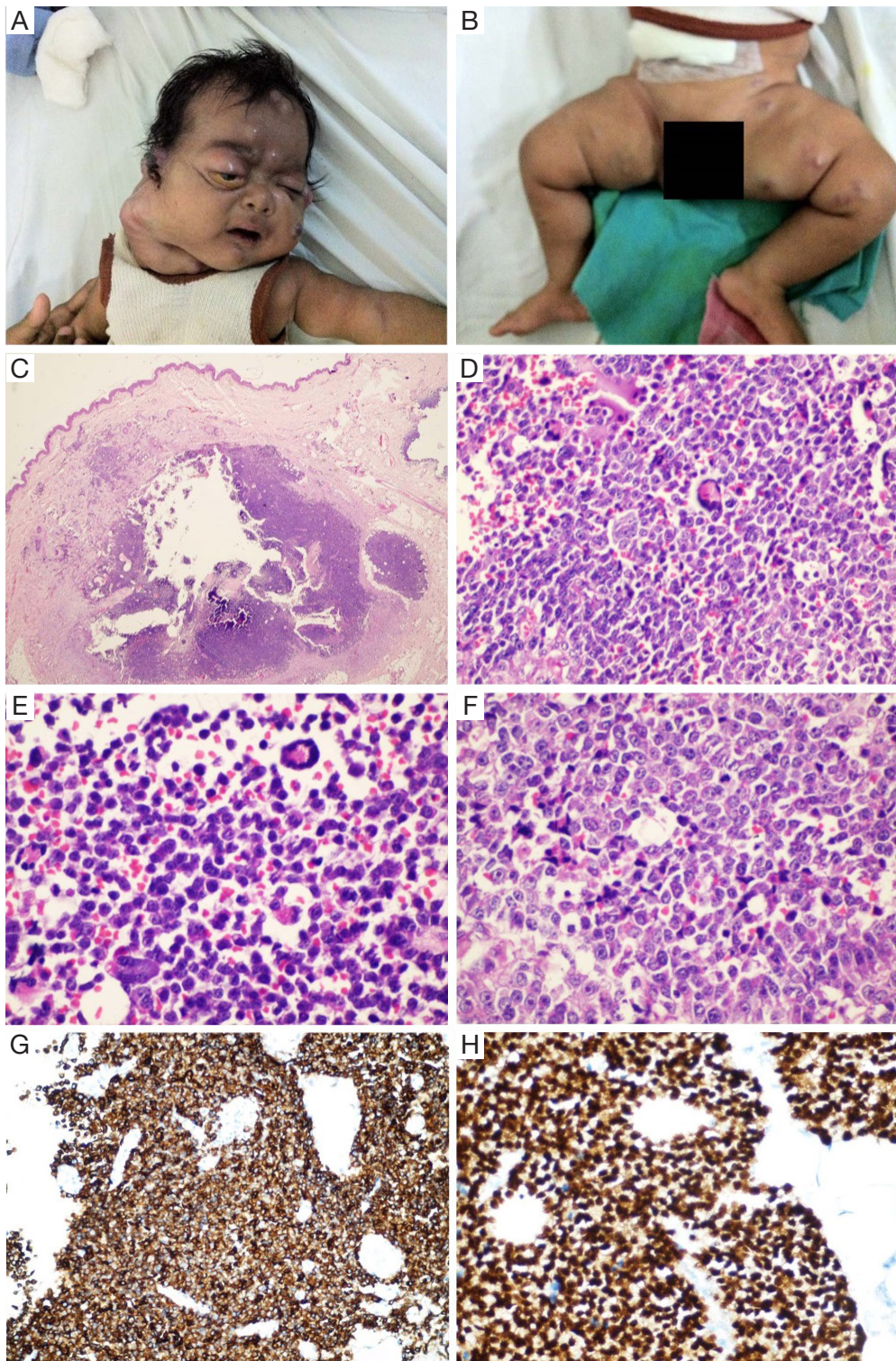


Figure 1 Clinical picture of case showing the subcutaneous swellings on faces and lower limbs (A&B); H&E (4×) nodular malignant round cell tumor in skin (C&D); H&E (40×) tumor cells are round, have prominent nucleoli & eosinophilic cytoplasm (E); H&E (40×) wreath like giant cells (F); IHC (40×) tumor cells express desmin (G); IHC (40×) tumor cells express myogenin (H).

On immunohistochemistry studies, the tumor cells showed strong cytoplasmic reactivity with Desmin (*Figure 1G*) and nuclear reactivity with Myogenin (*Figure 1H*) which confirmed the diagnosis as embryonal RMS.

We managed the patient with chemotherapy, VAC (vincristine, actinomycin D, cyclophosphamide) regime. After two cycles of chemotherapy nodules regressed both in size and number. Unfortunately patient was lost to follow up before completing the treatment.

Discussion

RMS is a common, highly malignant, childhood tumour, which is extremely rare in the neonates and paraneonates; constituting only 1-2% of all childhood RMS (3). There are only a few reports about this tumor in this age group. Of more than three thousand patients registered in the Intergroup Rhabdomyosarcoma Study (IRS) I-IV, only 14 were in the neonatal period at the time of diagnosis, showing rarity at this age group (4). In the first three IRS trials, about 35% to 40% of all tumors originated from the head or neck area, slightly less than 25% from the genitourinary tract, approximately 20% from extremity, 10% from trunk and the remaining 10% from other rare sites (5).

Primary cutaneous alveolar RMS accounts for 0.7% of RMS and tends to arise in the skin of the face (6). Rare cases of congenital alveolar RMS either presenting with multiple rashes or multiple cutaneous metastases have been reported, most with dismal outcome (1). Embryonal RMS presenting as multiple subcutaneous nodule is still a rare entity; even rarer than alveolar RMS. We report herein a case of embryonal RMS, possibly congenital in nature that presented in an unusual fashion and caused a clinical dilemma. Dilemma being to differentiate it from non-malignant causes of blueberry muffin like extramedullary hematopoiesis, cutaneous vascular anomalies, langerhans histiocytosis and from other round cell tumors. Thorough literature search could reveal only few case reports mentioning RMS metastasizing to skin at multiple sites (7,8).

The management of childhood RMS includes all the three modalities of cancer treatment that is surgery, chemotherapy and radiotherapy; depending on the risk group to which patient belongs.

According to a review done by Hazime *et al.* (9), ten-year survival rates for RMS is 86.3% for patients in low-risk subgroup A, 80.7% for low-risk subgroup B, 62.7% for intermediate-risk subgroup A, 61.7% for intermediate risk subgroup B, and 38.1% for the high-risk group.

The correct identification of RMS is a substantial challenge. Using a central review mechanism for patients enrolled in IRS, up to 20% of children enrolled are found to

be incorrectly diagnosed with RMS or have their histologic subtype reclassified.

This article emphasizes that in case of a round cell tumor, immunohistochemical studies are mandatory in order to rightly identify the tumor and differentiate amongst various malignancies like RMS, neuroblastoma, malignant lymphoma, small cell carcinoma and primitive neuroectodermal tumor which look similar on histopathological examination.

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None.

Footnote

Conflicts of Interest: The authors have no conflicts of interest to declare.

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