CASE REPORT

Bifocal Adult Embryonal Rhabdomyosarcoma

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ABSTRACT

Embryonal rhabdomyosarcoma is commonly seen in young children, its occurrence in the adult population is rare. Here, we describe a 36-year-old male with the diagnosis of adult embryonal rhabdomyosarcoma who was admitted with two large masses protruding from the left side of the neck and lower trunk. Diagnosis was established through biopsy and immunohistochemical studies of the masses. Treatment included surgical resection along with chemotherapy and radiotherapy with complete resolution of masses. Furthermore, a follow-up was scheduled 12 months post-treatment.

Key Words: *Adult embryonal rhabdomyosarcoma. Metastasis. Lymph nodes.*

INTRODUCTION

An embryonal rhabdomyosarcoma is a primitive, malignant, soft tissue sarcoma that mimics the phenotypic and biological features of embryonic skeletal muscles. It is predominantly appreciated in the pediatric population and occurrence in adults is extremely rare. These tumors are generally localized on the neck, trunk, extremities and genitourinary tract region.[1] Unlike embryonal rhabdomyosarcomas in children, the prognosis of this malignancy in adults is poor.

Here, we report a unique case of a 36-year-old anemic male patient with the presenting complaint of pain in the upper left quadrant of the abdomen along with two protruding masses located on the left side of the neck and trunk region.

CASE REPORT

A 36-year-old male patient presented with two large masses protruding simultaneously from the left side of the neck and lower trunk (Figure 1A and 1B), which he noticed 2 months prior to his initial visit. The patient had a history of weakness, normochromic anisocytosis and blood transfusion. On examination, the patient displayed signs of pallor/anemia, cervical lymphadenopathy and splenomegaly. Local examination revealed that the tumors were warm, red, non-tender with no sign of superficial vessel engorgement. Both the masses were non-mobile and firm. A review of the patient's system revealed no sign of fever, chills, sweats, bone pain, headache dyspnea or cough. Moreover, he denied the use of tobacco products and alcohol. His family history revealed no incidence of cancer.

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Our initial step was to perform a complete metastatic workup including a Liver Function Test (LFT), Urinalysis (US) Complete Blood Count (CBC) and blood electrolytes. All tests were within normal limits except the CBC which showed a decreased number of Red Blood Cells (RBCs) respectively. Subsequently, the patient underwent Computed Tomography (CT) and Magnetic Resonance Imaging (MRI) of the head, neck, chest and abdomen. The images showed few, large, well-defined rounded lesions with marked internal necrosis and thick enhancing walls on the left side of the neck (Figure 2) and lower trunk. The largest lesion was located in the supraclavicular region measuring 8.0 x 7.0 x 6.6 cm (TS x AP x CC). This led to displacement of adjacent muscles and vessels without invasion. X-ray and ultrasound were not utilized since CT and MRI scans had already displayed conclusive evidence regarding the extent and position of both masses.

To conclude a diagnosis, biopsy of the regional lymph nodes was completed. The sections examined from the largest encapsulated nodular piece and separately lying multiple soft tissue pieces exhibited a neoplastic lesion composed of diffuse sheets and fascicles of oval to spindle shaped neoplastic cells which contained scant amount of eosinophillic cytoplasm and markedly pleomorphic hyperchromatic nuclei, with variably prominent nucleoli and brisk mitotic activity (Figure 3). In addition, the intervening connective tissue stroma revealed diffuse infiltration of chronic inflammatory cells. Next, immunohistochemical staining was performed in which the neoplastic cells revealed Desmin, Myo-D1, BCL2 and CD99 (Mic-2) were focally positive. The biopsy and immunohistochemical findings were similar in both the neck and lower trunk protrusions. Thus, on the basis of all the above findings, the diagnosis was confirmed as primary adult embryonal rhabdomyosarcoma of the neck with distal metastasis to the lower trunk.

The patient's age, location of the masses, nodal status and histologic/immunohistochemical findings were taken

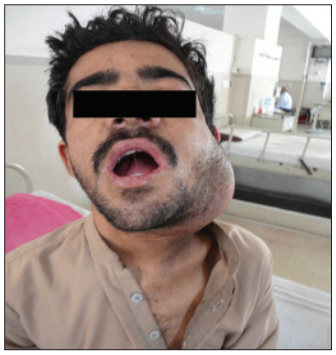

Figure 1a: Gross image displaying a mass protruding from the upper left side of the neck.

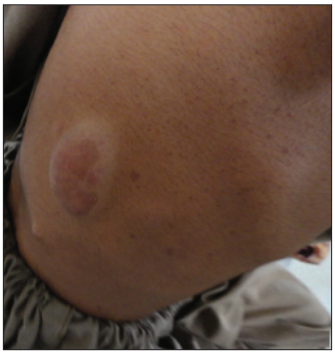

Figure 1b: Gross image showing an oval shaped mass protruding from the lower right trunk.

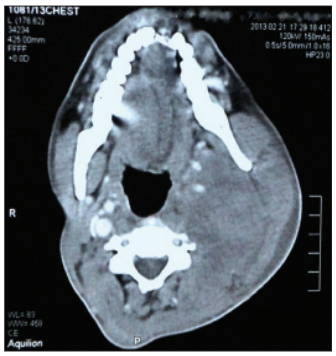

Figure 2: Computed tomography scan displaying a well-defined rounded lesions with marked internal necrosis and thick enhancing walls on the left side of the neck.

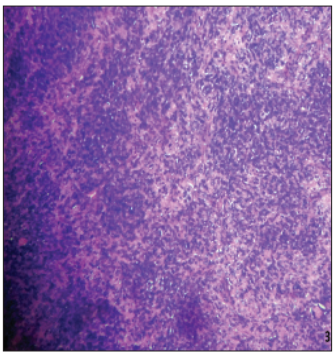

Figure 3: Histopathological image revealing a neoplastic lesion composed of diffuse sheets and fascicles of oval to spindle shaped neoplastic cells which contained scant amount of eosinophillic cytoplasm.

into account while planning for his treatment. Hence, a multimodality therapy integrating surgery, chemotherapy and radiotherapy was adopted. First, complete resection of both masses was done by using a wide excision technique with microscopically positive margins. Clear margins of both masses were assessed under ultrasound guidance during surgery. Extra precaution was taken since the mass in the neck was abutting the local muscles and vessels. The removed specimen was then handed over to the pathology laboratory for further evaluation.

Postoperatively, both chemotherapy and radiotherapy was administered. Chemotherapy agents included a three-drug regimen (vincristine, dactinomycin, cyclophosphamide) which was administered over a three week cycle. In addition, external beam radiation therapy was administered locally at the two sites of resection. Lastly, a 12-month follow-up was scheduled in which there were no signs of recurrence.

DISCUSSION

Embryonal rhabdomyosarcoma is a commonly occurring childhood tumor. However, its occurrence in adult individuals > 30 years old is a rare entity. Various studies have reviewed the cases of embryonal rhabdomyosarcoma in adults, discussing their presentation, pattern and prognostic factors. Nevertheless, a thorough literature search found only a few presentation of an adult embryonal rhabdomyosarcoma on two distinct locations simultaneously. Furthermore, this is the first presentation of its kind in the Pakistani literature.

Rhabdomyosarcoma (RMS), a malignant mesenchymal tumor, arises from cells destined to form skeletal muscles. Due to differences in the histologic patterns, RMS is classified into three types: embryonal (including sarcoma botryoid), alveolar and pleomorphic, with the latter more common in adults. Interestingly, childhood RMS commonly arises from head and neck region and genitourinary areas whereas adult have predilection in the extremities with head and neck and trunk region being the less common sites.[1] RMS can develop in an individual at any age but, as mentioned by a study conducted on 32 RMS patients, the average age of the sample population was found to be 26.4 years,[2] similar to the age of presentation in this patient.

Although the first clinical observation made was an enlarging mass of the neck and trunk, this patient also complained of weakness mainly due to anemia, which led to a preliminary suspicion of bone marrow infiltration.

Multiple CT scans revealed well-defined soft tissue masses with hypodense areas. Both tumor sizes were relatively large, measuring more than 5 cm and 7 cm in the neck and trunk. The size of tumor is a major determinant of metastasis particularly if they are more than 5 cm with an extremely high metastatic rate of tumor more than 10 cm.[3] Esnaola and colleagues reported that the 5-year survival rate for patients with RMS of sizes less than 5 cm was 60%. However, this percentage drops to zero when the tumor grows more than 10 cm.[4] Within the neck, the tumor size was large enough to compress adjacent tissue influencing a mass effect on adjacent muscles and larynx and may be a cause of slight airway obstruction. In addition, the cervical lymph nodes of the patients were enlarged indicating regional metastasis. It is recognized that lymph node invasion is an unfavorable prognostic factor patients with embryonal RMS.

To confirm the diagnosis, biopsy of the regional lymph node was carried out. Presence of undifferentiated, small, round or spindle-shaped cells with eosinophillic cytoplasm indicated characteristics of rhabdomyoblasts, while presence of loose myxoid foci with dense spindle cells suggested embryonal type. Awareness of this newer classifications and their molecular basis is key to stratifying patients on modern therapeutic protocols and genetic therapies. Additionally, immunohistochemical studies are also indicated. Recent studies have highlighted the importance of immunohistochemistry for differentiating between different categories of sarcoma.

The expression of Myo-D1 and CD99 (Mic-2), desmin and myogenin aids in classification of RMS.

Treatment strategy involved surgical excision, chemo-therapeutic agents and radiation therapy. A recent study mentioned that 55% overall and 64% disease-free survival at 2 years is anticipated when both surgery and chemoradiotherapy are used therapeutically.[4]

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