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Original Research Paper

Oral Pathology

PLEOMORPHIC RHABDOMYOSARCOMA IN THE HEAD AND NECK REGION: A RARE CASE REPORT

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ABSTRACT Rhabdomyosarcoma (RMS) is a rare, aggressive, malignant mesenchymal tumor of skeletal muscle cells. The pleomorphic histological variant of RMS occurs in adults beyond 45years of age and represents the most aggressive subtype with an incidence of approximately 0.44/100,000. The diagnosis of RMS is difficult with the 5year overall survival rate less than 50%. It presents varied clinical and biological behavior and requires individualized management. The common region of metastasis includes lymph nodes, lungs and bone marrow. Here we report a case of pleomorphic rhabdomyosarcoma in 58year old female. The patient reported with a swelling in the neck region and had a history of surgical treatment for ovarian tumor. Histopathology findings revealed metastatic undifferentiated carcinoma. The case was positive for immunohistochemistry markers and their findings are diagnosed as pleomorphic rhabdomyosarcoma. This is a rare case of RMS which had metastasized to head and neck and this article emphasizes the importance of IHC in accurate and clear diagnosis of RMS.

KEYWORDS : Head and neck, Metastasis, Rhabdomyosarcoma

INTRODUCTION

Head and neck region sarcomas are rare tumors comprising of, approximately 1% of all malignancies.[1]

Rhabdomyosarcoma (RMS), was first described by Weber in 1854.[2]

An incidence rate showed approx. 0.44/100,000 per year in children and adults. 30% of paediatric RMS are found in the head and neck region, whereas in adults it occurs more frequently in the extremities and rarely in the head and neck region. Histological subtypes of RMS include embryonal (ERMS), alveolar (ARMS), pleomorphic (PRMS) and spindle cell/sclerosing (SC-RMS). ERMS and ARMS are the main types, seen in the paediatric population.[3]

In this report, we present a rare case of pleomorphic RMS in the head and neck region in a 58-year-old lady.

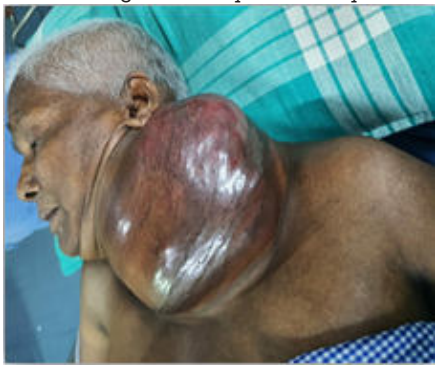

Figure 1: Clinical presentation of swelling

CASE REPORT

A 58-yr-old female was referred to our institution, for the investigation of painful swelling in the left side of the neck for the past 3 years. Patient revealed a history of surgical treatment for an ovarian tumor before 3 years.

Clinical examination showed a severe facial asymmetry. The swelling extended from the right side of 2/3rd of neck to the left side of posterior auricular region. The swelling was large in size, with a smooth surface and well- defined edges.

The skin over the swelling was stretched and inflamed. (Figure 1). There was also a presence of restriction in the neck movements and mouth opening. Tenderness on palpation with rise in temperature was noted. The consistency was firm along with the lymph nodes enlargement.

An excisional biopsy was done. The macroscopic features showed a globular soft tissue mass, measuring about 6.5x3.5x1.2cm.

External surface of the tissue appeared congested. The surface of cut section appears greyish white, firm with the vague nodularity.

Histological examination of the specimen showed clusters of small round cells with the hyperchromatic nuclei and the eosinophilic cytoplasm separated by fibrovascular septae (Figure 2 & 3).

Morphologic features of the cells indicated a differential diagnosis comprising of RMS, Ewing's sarcoma, malignant melanoma and an epithelial tumor.

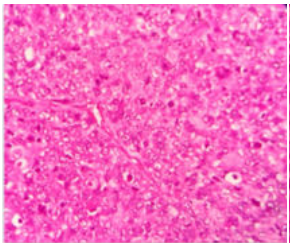

Figure 2: Histologic features showed clusters of small round cells with the hyperchromatic nuclei

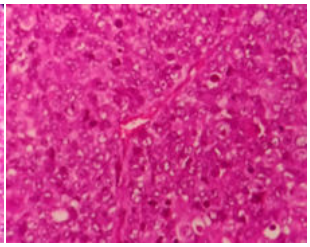

Figure 3: Histologic features showed eosinophilic cytoplasm separated by fibrovascular septae

Immunohistochemistry report revealed that neoplastic cells were strongly positive for vimentin (Figure 4), desmin (Figure 5), smooth- muscle actin (SMA) (Figure 6), myogenin (Figure 7)

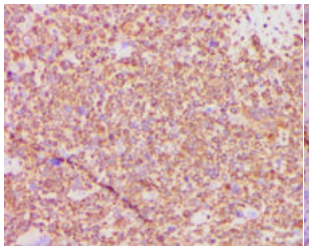

Figure 4: Vimentin - positive

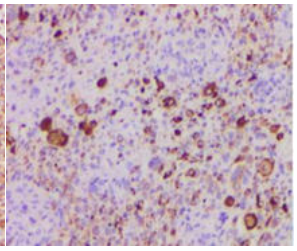

Figure 5: Desmin - positive

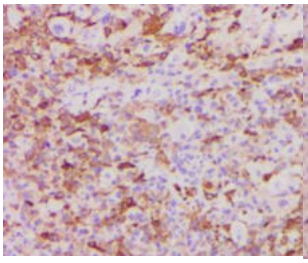

Figure 6: Smooth- Muscle Actin (SMA) - positive

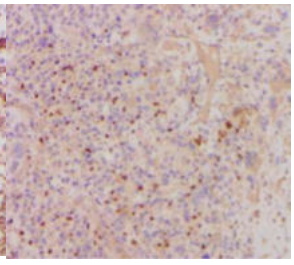

Figure 7: Myogenin - positive

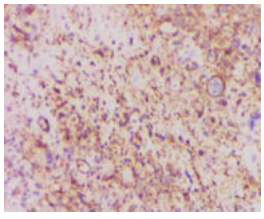

Figure 8: CD34- positive

and CD34 (Figure 8) and showed negative for CD45 (Figure 9), HMB45 (Figure 10), PanCK (Figure 11), CD68 (Figure 12) and S-100 (Figure 13).

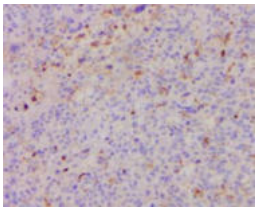

Figure 9: CD45- Negative

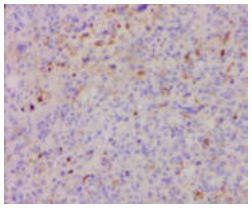

Figure 10: HMB45 - Negative

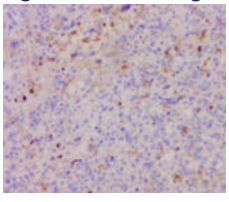

Figure 11: PanCK- Negative

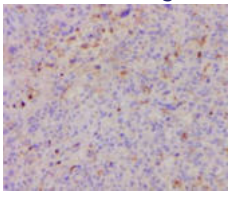

Figure 12: CD68- Negative

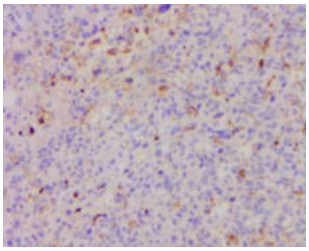

Figure 13: S-100 - Negative

After performing the standard diagnostic work-up, the tumor was diagnosed as the pleomorphic RMS. Then, the patient was referred to the oncology department and the proposed treatment was "Multimodal therapy", a combination of surgical excision, chemotherapy (Ifosfamid+ Vincristine+ Actinomycin D - IVA regimen) and radiotherapy including external beam and brachytherapy.

DISCUSSION

RMS occurs due to genetic mutations.[4]
Incidence rate was higher in children aged from 1-4years and it reduced as the age increases.[2]

It is 1.5 times more prevalent in males than in females.[5]

Generally, RMS can be seen in any anatomic site and it depends only on the histological subtypes. Identification of 'fusion positive' (FP) and 'fusion negative' (FN) helps to divide the subtypes. Childhood RMS have best fusion status when it's compared with adults RMS. Fusion genes are often detected in clinical biopsy material by RT-PCR and Fluorescent In-Situ Hybridization (FISH) assays.[6]

Pleomorphic rhabdomyosarcoma (PRMS) is a rare adult variant of RMS which occurs in adults more than 45 years.[4,6]

PRMS is often seen in the deep soft tissues of the extremities and rarely in the head and neck region.[8]

It may present clinically as extensive fast-growing facial swelling occasionally associated with pain, trismus, paraesthesia, facial palsy, and nasal discharge[2]

non-tender palpable mass in submandibular region,[8] maxillary sinus and palate.[3]

Other commonly involved sites are liver[9], pancreas[10], uterus[11] and abdomen, urethra, lumbar region, etc.[7]

Radiographic examination often reveals the size of the lesion, space relation, and the extent of bone destruction.[2]

CT scan reveals poorly defined, inhomogeneous soft-tissue masses destroying the adjacent bone structure.[12]

MR imaging showed that of a homogeneous mass, isointense or minimally hyperintense relative to muscle on T1-weighted images and hyperintense relative to both muscle and fat on T2- weighted images, with postcontrast images of the tumour.[12] The findings were reviewed retrospectively to evaluate points of tumor origin, homogeneity, marginal definition, density/signal intensity, bone destruction, calcification, hemorrhage, necrosis, and nodal extension. The densities/signal intensities of the tumors were compared with those of muscle. These findings are similar in children and adults.[12]

For diagnosing, it requires either an incisional or excisional biopsy or core needle biopsy for histology and molecular pathology studies. Morphologically, the RMS cells are of heterogenous shape and it ranges from undifferentiated &

round cells, ovoid cells, 'tadpole-like' cells, spindle- shaped cells & fully differentiated rhabdomyoblasts.[6]

Spindle cells are often seen in a haphazard arrangement. The nuclei are in oval shaped or elongated with packed chromatin.[4]

atypical and occasionally multi-nucleated cells with prominent eosinophilic cytoplasm.[3]

FNAC often reveals monomorphic cells with a blastoid-like appearance and rare rosette-like formations. It showed highly cellular and composed of large, pleomorphic cells admixed with lymphocytic infiltrate. The cells range from epithelioid or spindle-shaped to rhabdoid morphology usually with high nucleocytoplasmic ratio and prominent mitotic activity. Moreover, multinucleated giant cells and clusters of atypical cells with abundant eosinophilic cytoplasm were observed. Immunocytochemistry show positive staining for MyoD1 and myogenin and negative staining for S-100.[8]

Initial differential diagnosis based on the monomorphic pattern included blastoid- like lymphoma and small blue round cell tumors.[8]

Other differential diagnosis like squamous cell carcinomas, have same imaging features of RMS, but the age at diagnosis is elder than that of RMS, and that helps in differential diagnosis.[12]

IHC study helps to identify the pleomorphic RMS from malignant fibrous histiocytoma, leiomyosarcoma and liposarcoma.[7]

Median time for recurrence was 17.8 months. Most patients had local recurrences (72%). The 5-year overall survival (OS) rate was from 28.3% to 8.7%. Multivariate analysis identified 4 factors that were associated with poor survival: 1) alveolar subtype (relative risk [RR], 2.0), 2) parameningeal or "other" sites (RR, 2.6), 3) systemic recurrence (RR, 3.1), and 4) recurrence on therapy (RR, 2.3).[13]

In children, PRMS can be treated with multidisciplinary therapy. Surgical resection and radiotherapy are used for the management of first tumor site, while chemotherapy is employed to stop metastasis. Chemotherapeutic drugs include vincristine, actinomycin- D, cyclophosphamide doxorubicin and ifosfamide and the same treatment is applied for the adults.[9]

Hence ERMS occurs in children, have far better prognosis. While ARMS occurs more frequently in adults, and shows more aggressive biological behaviour. PRMS can occur in both children and adults, but the outcome is significantly worse in adults, with higher rates of recurrence and metastasis.[3]

Prognosis can be decided by the following variables such as tumour invasiveness, metastasis, age at diagnosis, regional node involvement and histopathologic subtypes.[14]

Overall survival is poor in adults when compared with children. Major sites of metastases are lung, lymph nodes, and bone marrow, followed by the heart, brain, and meninges. The frequency of nodal metastasis is 7% to 50% in general and less common for tumours located in the head and neck, but there is no evidence of a relationship between lymphatic spread and age, sex, or histologic subtype.[12]

CONCLUSION

Pleomorphic RMS is a rare tumour of skeletal muscle origin. Diagnostic criteria for pleomorphic RMS with combined histology and immunohistochemistry will help in early diagnosis and treatment.

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