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Malignant extrarenal rhabdoid tumour in the oral cavity: Report of a rare malignancy in an unusual location

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Abstract: *Malignant rhabdoid tumour (MRT) is a well established aggressive malignant neoplasm occurring in kidney particularly in infants and children. When this type of tumor involves any extrarenal site it is termed malignant extrarenal rhabdoid tumor (MERT). Here we report a case of MERT involving the oral cavity. To the best of our knowledge, apart from us only one oral MERT has been documented. Due to their scarcity, prompt and bold diagnosis is quite challenging for a histopathologist. Confirmed final diagnosis must be given after thorough immunohistochemical workup. Confusions and obscurities regarding a definitive treatment policy further complicate management also. We adopted a multimodality approach comprising of surgery, radiotherapy and chemotherapy for our patient. She is disease free for last 30 months on regular followup and maintaining her normal daily activities.*

Keywords: *malignant exrarenal rhabdoid, oral cavity, immunohistochemistry.*

I. Introduction

Malignant rhabdoid tumor (MRT) is a very aggressive neoplasm originally described in kidneys of infants and children. Beckwith et al. First described MRT as a sarcomatous variant of Wilm's tumor.[1] However it had its own distinct pathological characteristics which are different from Wilm's tumor. The word "rhabdoid" was endorsed because of its resemblance with rhabdomyoblast under light microscope. With progression of time mrts had been described over a broader array of locations like central nervous system, liver, soft tissue, mesentery, stomach and spine. They are termed malignant extrarenal rhabdoid tumors (MERT). Recurrent alterations of chromosome 22 in both renal and extrarenal mrts provided the preliminary perception that these tumors could share a common genetic basis.[2,3] Here we describe a girl of 13 year with MERT involving the buccal fold in the oral cavity. To the best of our knowledge, till date apart from us only one case report describes oral MERT.[4] Our case underwent multimodality treatment comprising of surgery, chemotherapy and radiotherapy.

II. Case report

A 13 year old girl presented with a progressively enlarging painless lump arising from the mucosal aspect of right cheek over last four months. She experienced few episodes of exsanguinations. On examination, the lump was 7 cm. x 4 cm. in size, firm, nontender, dark red in color and denuded at places. General systemic examination was unremarkable. All routine hematological and biochemical investigations were within normal limit.Fine Needle Aspiration Cytology (FNAC) was inconclusive. Computed Tomography (CT) scan revealed an irregular soft tissue mass at right cheek (29 mm. x 19 mm.) with involvement of underlying bone with no lymphadenopathy. Incisional biopsy was done. It revealed a malignant connective tissue tumor; but further categorization was not possible in that small bits.

The patient underwent right hemimandibulectomy with reconstruction along with bilateral modified radical neck dissection. Resected tumor with a part of mandible was sent for histopathological examination. A proliferative growth measuring 3.2 cm. x 2.5 cm. x 1.9 cm. was noted. The tumor was irregular, grayish in color. Microscopical examination revealed a tumor with relatively circumscribed margins. It was composed of spindle shaped cells with marked myxoid change. The cells had elongated plump hyperchromatic nuclei, prominent nucleoli and abundant eosinophilic cytoplasm. Focal areas of round cells composed of vesicular nuclei, prominent nucleoli and abundant amount of eosinophilic cytoplasm were also noted. Mitotic figures

were encountered. Areas of necrosis were noted. All resection margins including the deep one were free from the tumor involvement. Adjacent bone and lymph nodes also got rid of the tumor. Juxtanuclear hyaline inclusion was found in Periodic Acid Schiff (PAS) stained sections. Based on morphology a histological diagnosis of spindle cell sarcoma, most probably embryonal rhabdomyosarcoma (pleomorphic type) was given and immunohistochemistry was suggested to confirm the case. The tumor cells stained positively for Epithelial Membrane Antigen (EMA) and were immunonegative for cytokeratin, S-100, HMB-45, MELAN-A, CD30, Alk-1, CD43, CD20, CD34 & CD31. Most importantly the tumor cells lost expression of INI-1. Taking morphological and immunohistochemical findings into consideration a final diagnosis of malignant extrarenal rhabdoid tumor was given.

Our patient received external beam radiation to the tumor bed and bilateral neck upto a total dose of 20 Gray (Gy) in 10 equal fractions over 2 weeks. Thereafter she received systemic chemotherapy with 3 weekly alternate VDC/ICE regimen (VDC consisted of Vincristine [1.5mg/m^2 BSA; max. daily dose 2mg.] on DAY1 + Doxorubicin [50 mg/m^2 BSA] on DAY1 + Cyclophosphamide [1000 mg/m^2 BSA] on DAY1; ICE consisted of Ifosfamide 1800 mg/m^2 BSA on DAY1-DAY4 + Carboplatin 400 mg/m^2/BSA on DAY1 + Etoposide 100 mg/m^2 BSA on DAY1-DAY4 with mesna uroprotection and G-CSF support for both regimens.) which continued for 1 year. She is on monthly follow up since then and she is free from any loco-regional or systemic relapse for last 30 months.

III. Discussion

Scarcities of documented cases of MERT with full immunohistochemical workup and subsequent obscurity regarding a definite and established treatment regimen have complicated the scenario furthermore. Brennan *et al*. estimated a three-year survival of meager 9% for extracranial, extrarenal rhabdoid tumors. [5] Clinical presentation of such cases can be of diverse type according to the site of involvement. Among the unusual locations MERTs involving oesophagus, heel etc. have been documented. [6,7] In our case the tumor involved the buccal fold of the oral cavity. Patron *et al.* reported a case of MERT involving the tongue in a boy of only 10 days; the baby died only 17 days after the diagnosis again reminding us the fatal potentiality of the tumor.[4] To best of our knowledge apart from us this is the only reported case of MERT occurring in the oral cavity.

Histogenesis of MERT is controversial. In World Health Organization (WHO) classification it is placed in tumor with uncertain histogenesis. Amongst close histological diagnoses epithelioid sarcoma, synovial sarcoma, rhabdomyosarcoma, malignant melanoma, meningioma and lymphoma should be ruled out. PAS positive granules found in MERT are basically aggregates of intermediate filament which can be proved by ultrasatructural examination. It is now established that most cases of MERT share a common genetic deletion in 22q chromosome. As a result loss of INI-1 protein expression happens; which can be considered pathognomic for MERT. Our case also lost such expression.

We treated the patient with alternating cycles of VDC/ICE and she is free from disease for last 30 months. As there is immense obscurity regarding treatment regimen like Wagner *et al.* we also opine that it warrants investigative clinical trials.[8] Role of radiotherapy is not well defined in MERT. But we think that it forms a cornerstone of successful combined modality of treatment. In line with us Puri *et al.* advocated such multimodality treatment comprising of radiotherapy, chemotherapy and surgery MERT.[9] Though clear cut treatment guide line not available for this tumor but here we followed this multimodality approach and got satisfactory result till now. Finally, to conclude we can say that correct tissue diagnosis with relevant immunohistochemical panel in conjunction with multimodality treatment can finally change the outcome a lot.

Figures

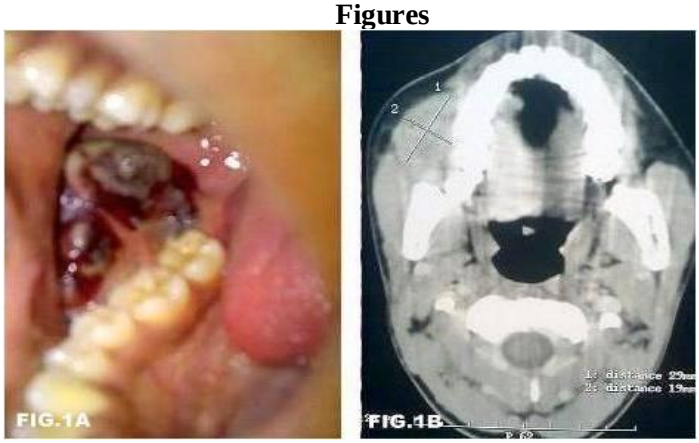

Fig. 1. 1A. Clinical photograph of the lesion. 1B. CT scan showing the tumor. (29 mm. x 19 mm.).

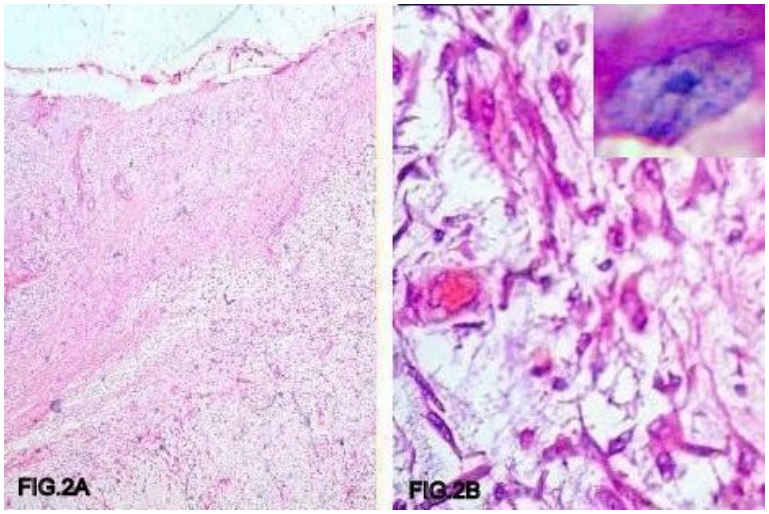

Fig. 2. 2A. Photomicrograph showing a well circumscribed tumor area surrounded by normal buccal mucosa tumor composed of monomorphic round cells arranged in diffuse sheets. (Hematoxylin & Eosin, X100). 2B. Photomicrograph showing tumor composed of spindle shaped rhabdoid cells with areas of myxoid degeneration. (Hematoxylin & Eosin, X400). 2B (Inset) Polygonal rhabdoid cells with eccentric vesicular nuclei, prominent nucleoli and juxtanuclear PAS (+ve) eosinophilic hyaline inclusion (globules). (X400).

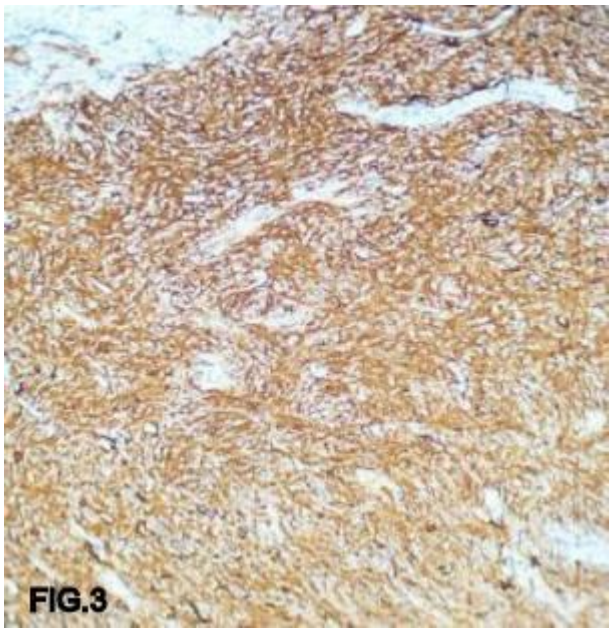

Fig. 3. *Tumour cells showing strong membrane positivity for EMA. (X100).*

III. CONCLUSION

In conclusion we emphasize that Malignant extra-renal rhabdoid tumor should be kept in mind while diagnosing the histological type of any tumor of oral cavity, particularly when it originates from buccal mucosa. Because correct histological diagnosis alongwith complete immunohistochemical workup will be of immense help in early institution of multimodal treatment approach. Finally more and more such rare tumors in such uncommon location is reported, we can determine a definitive immunohistochemical pannel for such grey zone malignant neoplasm where the ontogeny or tumor histogenesis is still unknown. Moreover, a clear cut and more definitive treatment protocol can be introduced which can save many lives.

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