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Orbital Rhabdomyosarcoma with anaplastic features

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Abstract

Orbital rhabdomyosarcoma (RMS) is the most common orbital malignancy of childhood. It is a highly malignant tumour and is one of the few life threatening diseases, if its diagnosis is delayed. Among the entire group of extraconal intraorbital tumours RMS poses a significant challenge for the pathologist and oncologist in terms of its diagnosis and treatment. This case report highlights the general overview of primary orbital RMS, its histopathological and immunohistochemical features and management.

Keywords: Rhabdomyosarcoma, Orbit, Paediatric, Malignant, Histopathology.

Introduction

RMS is a rare, highly malignant tumor, tissue of origin is pluripotent mesenchyme.(1) It is the most common soft tissue sarcoma of head & neck in childhood accounting for 4% of all paediatric malignancies.(1) The orbit is the primary site.(2) Most of tumors occur in the first decade of life.(3) Boys are affected more often than girls. It presents as rapidly evolving unilateral proptosis(4) but clinical manifestations depend on location of the tumor & its rate of growth. The tumor has predilection for superior nasal quadrant of orbit & radiology is important in evaluation of this tumor. The common histopathologic types are embryonal & alveolar varieties. Treatment usually consists of excisional biopsy followed by combination of radiotherapy and chemotherapy. Orbital RMS usually presents as space occupying lesion & may mimic other neoplastic, inflammatory masses occurring in the orbit. It is one of few life threatening diseases therefore prompt diagnosis and treatment is life-saving issue.(2) Hence knowledge of clinical, histopathological, radiological features as well as the more recent advances in the management of this entity is essential.

Case Report

A 5 year male child was brought to the ophthalmology department with swelling in right eye since 15 days. On external examination right eye had non tender, non-mobile, firm mass at medial canthus with proptosis & chemosis of right eye. There was no history of recent trauma, systemic illness, family history of malignancy. Other part of ophthalmic examination including visual acuity, pupillary reflex, biomicroscopic, fundoscopic examination were normal. The left eye was unremarkable. Magnetic Resonant Imaging (MRI) of brain and orbit was done which showed well-defined, homogeneous enhancing mass lesion in the intraorbital extraconal compartment of right eye on its medial & superior aspects. There was no infiltration or adjacent bony erosion (Fig. 1). In view of short clinical history & likely possibility of neoplasm, histopathological correlation was advised. A complete excision of mass was performed which was 3x3x2cm firm, fleshy, homogeneous grayish white (Fig. 2).

Histopathological examination showed highly cellular tumor, nuclei of tumour cells were round to spindly & hyperchromatic with moderate eosinophilic cytoplasm (Fig. 3). Also seen were giant anaplastic cells showing nuclear enlargement, hyperchromasia, abnormal mitosis (Fig. 4). Immunohistochemical

staining for myogenin & desmin was diffusely positive (Fig. 5). The final diagnosis was Embryonal Rhabdomyosarcoma with Anaplastic (Pleomorphic) features. Computed Tomography (CT) scans of abdomen & chest & bone scan demonstrated no evidence of lymphnode involvement of metastasis. On repeat MRI brain there was residual tumour and chemotherapy was advised for completeness of treatment.

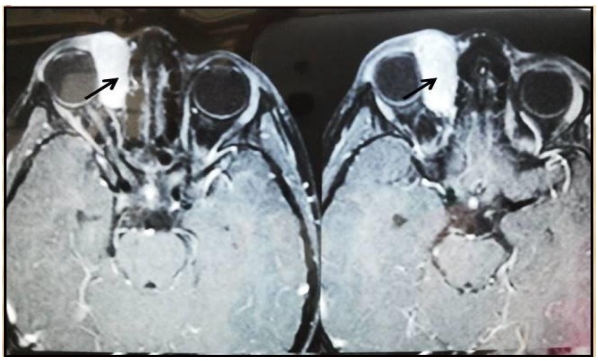

Fig. 1: MRI of brain and orbit which showed a well-defined, homogeneous enhancing mass lesion in the intraorbital extraconal compartment of right eye in the medial and superior aspects with no inflitration or adjacent bony erosion

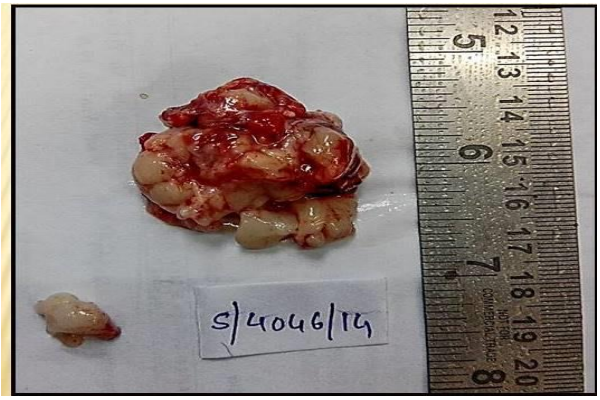

Fig. 2: Gross photo shows firm, fleshy, homogeneous and grayish white mass

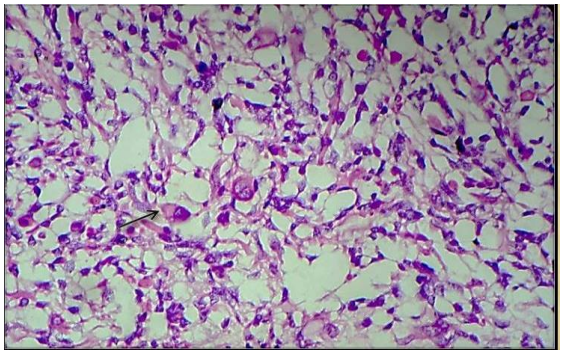

Fig. 3: Histology shows a highly cellular tumor with round to oval to spindle shaped cells in sheets with scant to moderate eosinophilic cytoplasm against a myxoid background

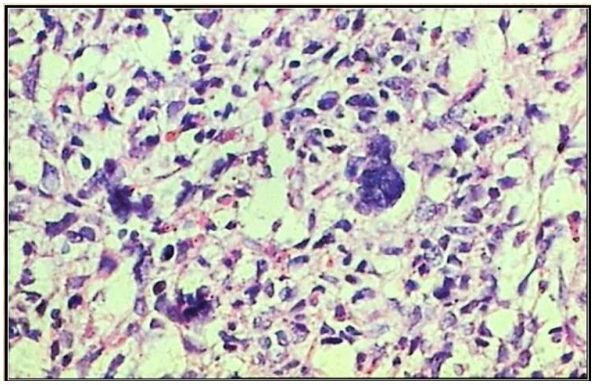

Fig. 4: Shows giant anaplastic cells with nuclear enlargement, hyperchromasia and abnormal mitosis

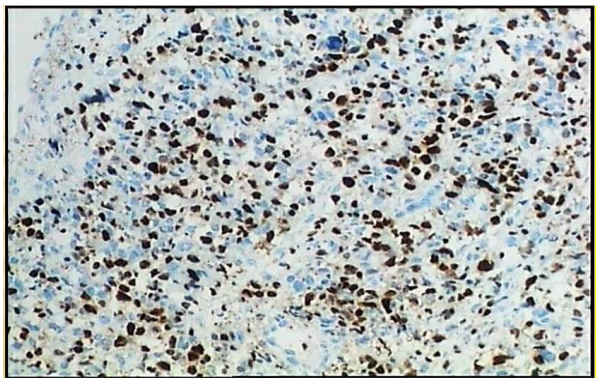

Fig. 5: Shows Immunohistochemical staining for myogenin is diffusely positive

Discussion

Rhabdomyosarcoma is most common primary orbital malignancy of childhood. It is usually seen in first decade of life with male predominance[5] as was seen in our case. Classic clinical picture is sudden onset with

proptosis & without history of previous trauma or signs of upper respiratory tract infections[6] which was seen in our patient. On imaging studies orbital RMS is extraconal (37-87%) and superonasal in location especially for embryonal RMS.[7] It appears as a well circumscribed, homogeneous, soft tissue mass with postcontrast enhancement.[7] Intracranial extension and invasion of paranasal sinuses are uncommon, while changes in adjacent bone may frequently occur. In our patient all characteristic features of orbital RMS on imaging were present. Detailed clinical history & imaging studies helps to exclude other differential diagnosis which include benign & malignant conditions.[2] Biopsy should be performed if malignant orbital tumor is suspected. But there are practical difficulties while performing biopsy or fine needle aspiration cytology in these patients because of vascular nature of the tumour.[5] However histopathology is important in terms of establishing diagnosis and determining prognosis for orbital RMS.[2]

RMS was once believed to arise from extraocular muscles, but currently orbital RMS develops from pluripotent mesenchymal cells that have ability to differentiate into striated muscle.[2] Orbital RMS is divided into three histological subtypes: embryonal, alveolar & pleomorphic. The embryonal type majorly comprising 50-70% of orbital RMS[5] shows round to spindle cells with highly eosinophilic cytoplasm & hyperchromatic nuclei. The alveolar type is characterised by loosely arranged poorly differentiated malignant cells separated by thin fibrovascular septa. The cells are large round to polygonal in shape, vesicular nuclei with abundant eosinophilic cytoplasm. The pleomorphic subtype occurs almost exclusively in adults[2] but is rarely diagnosed in children.

In our case histopathological features were characteristic of embryonal RMS with anaplastic features. This subtype forms only a small group of cases (less than 1%).[8] These tumours show presence of varying numbers of anaplastic cells with giant lobated nuclei & variable amount of cytoplasm. These cells are usually 3 times larger than surrounding neoplastic cells. Such tumours with anaplastic features have an unfavourable prognosis compared with non-anaplastic tumours.[8] According to Inter- group Rhabdomyosarcoma Study (IRS I,II,III) our case falls in group I- tumours contain only scattered anaplastic cells.[8]

The differential diagnosis of RMS from other poorly differentiated pediatric round cell tumours including the Ewing's sarcoma, neuroblastomas and hematopoetic neoplasms can be difficult only on routine H&E staining. Therefore immunohistochemical studies constitute the main approach to accurate diagnosis. The markers found in RMS include antibodies against vimentin, desmin, muscle specific actin, myogenin, myoD1 & myoglobin.[2,7] The expression of markers specific for rhabdomyoblastic differentiation such as Myogenin and MyoD1 are particularly useful for the diagnosis of RMS.[7,10] The diagnosis of RMS is confirmed if atleast one of these proteins is positive.[10] In our case, there was diffuse positivity for myogenin which confirmed diagnosis of RMS.

In early 1970s, The North American Intergroup Rhabdomyosarcoma Study Group (IRSG) introduced guidelines regarding management which is based on histopathologic confirmation & staging of orbital RMS. Our patient falls into Group III i.e. gross residual disease. Current management includes surgery, irradiation and chemotherapy depending on the stage.[2] Our patient underwent chemotherapy treatment.

Conclusion

Orbital RMS has relatively good prognosis. However on histology thorough search of anaplastic cells in embryonal type should be carefully looked for as it has unfavourable prognosis. Thus knowledge of clinical, histopathological, radiology & more recent advances in the management of this entity is necessary.

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