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Orbital Rhabdomyosarcoma in a Child

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Abstract
Rhabdomyosarcoma (RMS) is the most common soft tissue sarcoma in children and frequently involves the head and neck region with 10% of the cases primarily involving the orbit. It is a highly malignant tumor which can be life threatening. CT and MRI are important in the evaluation of this tumor. Histologically, embryonal subtype is the commonest in children. Complete resection followed by chemotherapy and radiotherapy is the preferred treatment of choice. We report a case of Rhabdomyosarcoma of Orbit presenting with rapid proptosis of left eye in a 10 year old male child.
Keywords: *RMS, child, orbit, histology, embryonal.*

Introduction

Orbital RMS is the most common primary soft tissue sarcoma in children[1]. It accounts for 4% of all the childhood orbital malignancies[2]. Most of these tumors occur in the first decade of life[3]. The average age of presentation is 7 to 8 years. However, the condition is also reported from birth to eighth decade[4]. The condition should be suspected in a child with rapidly evolving unilateral proptosis of sudden onset[5]. Clinical presentation combined with characteristic imaging features of the disease aid in the diagnosis. The common imaging modalities are USG, CT and MRI[6]. Myogenin is a specific and sensitive marker in the diagnosis of RMS.

Case Report

A 10 year old male patient presented at the ophthalmology OPD with the complaint of painless rapidly increasing protrusion of left eye ball since four months. There was no history of fever, trauma or any other specific complaints. On examination, the patient was of average built and apyrexic. There was no pallor, lymphadenopathy or organomegaly. Ocular examination revealed a left sided proptosis with deviation of eyeball upwards and outwards. There was marked

restriction of ocular movements in all positions of gaze. Right eye appeared normal.

Routine investigations including CBC, ESR and LFT were within normal limits. X- Ray Chest and Ultrasound abdomen revealed no abnormality. CT Scan showed an ill-defined soft tissue density lesion, iso attenuating to extra ocular muscles with moderate inhomogeneous post contrast enhancement involving extraconal and intraconal compartment of left orbit mainly in infero lateral compartment with no internal cystic component or calcification with extension into extraocular muscles and left maxillary sinus with bony erosions suggestive of primary neoplastic etiology/ pseudotumor involving orbit.

Under general anaesthesia, an incisional biopsy of the mass taken and submitted for histopathology. Microscopic examination revealed sheets of small round cells with hyperchromatic nuclei and scant to moderate amount of cytoplasm admixed with spindle shaped cells in a loose syncytial arrangement. Highly cellular areas were seen alternating with areas of low cellularity in a loose myxoid stroma. Increased mitotic activity was seen along with atypical mitoses. Based on the histological findings, a diagnosis of a small round cell tumour probably Embryonal Rhabdomyo-sarcoma was made. Immunohistochemistry showed positive nuclearstaining with antibodies to Desmin and Myogenin, confirming the diagnosis of RMS. The patient was referred to an Oncology Institute for further management.

Discussion

Rhabdomyosarcoma of the orbit is the most common orbital malignancy in childhood accounting for 10% of all RMS cases[6]. The mean age is 6 to 8 years [5,6]. Boys are affected more often than girls[3]. Though most often the tumors arise denovo, a history of trauma may be associated with the clinical presentation of this tumor[3]. Increased risk has been noted in children with rare inherited diseases like Li Fraumini syndrome, Neurofibromatosis type II and MEN IIA[6]. Patients with orbital Rhabdomyosarcoma usually present with unilateral proptosis progressing rapidly within few days to weeks and/or globe displacement which is generally downwards and outwards because two thirds of these tumors are superonasal. Though superonasal orbit is the most common location, RMS has also been reported as lid masses and as masses in other areas of orbit[4]. In our case, the tumor was located in the superonasal region of the left orbit. In the study by Kalai et al, proptosis associated with signs of inflammation was noticed in a few cases. Hence RMS mimicking as orbital cellulitis is an important consideration to be kept in mind [4]. Chemosis and edema of the lids were noted in our patient also. The rapid growth and aggressive nature of the tumor frequently result in involvement of adjacent orbital bone. Intracranial extension may occur. Regional lymph node metastasis is uncommon. Hematogenous metastasis occurs to the lungs and bones[6]. Metastatic RMS has an unfavourable prognosis[2]. Both the CT and MRI are useful diagnostic tools in determining the location and size and also in evaluating the residual or recurrent disease[3]. On CT scan, the tumor appeared as a well-defined homogenous mass isodense to the extraocular muscles[3,6]. Bone destruction is common[3,6]. Histologically, two major subtypes of RMS may involve the orbit, the more common subtype is the embryonal type (89%). Alveolar RMS is the less common and more aggressive subtype[2,6]. Histologically, Embryonal Rhabdomyosarcoma consists of primitive mesenchymal cells most of which are round to oval or sometimes spindle shaped dispersed in a myxoid background. Cytoplasm is scanty and eosinophilic. Cross striations visible sometimes on light microscopy. Nuclei are round to oval and hyperchromatic. Frequent and atypical mitotic figures are seen.. Following biopsy, staging for orbital RMS is uniformly done worldwide according to the IRS postsurgical staging system[2]. Our patient comes under Group III with gross residual disease remaining after biopsy. Immunohistochemistry markers which confirm the diagnosis include

Desmin (90%), Myogenin, Myo D1, MSA and Myoglobin[2]. Vimentin and desmin though usually positive are less specific because they can be positive with other tumors with muscle differentiation[2,3]. Desmin remains a reliable and specific marker for RMS according to a study by S Kumar et al.[7]. Desmin was positive in our case as well. Myogenin and Myo D1 are transcriptional regulatory factors expressed early in skeletal muscle differentiation, hence are more specific in diagnosing Embryona lRhabdomyosarcoma[2]. All paediatric Rhabdomyosarcomas show nuclear expression of myogenin. Myogenin was expressed in all RMS samples but in no other soft tissue tumours according to several studies[8]. In our case strong nuclear positivity for Myogenin was noted. However it should be kept in mind that Myogenin and MyoD1 may also be expressed in other tumours demonstrating skeletal muscle differentiation and in regenerating muscle fibres entrapped within any infiltrating tumour. If a complete resection is possible, surgical removal of tumor with subsequent chemoradiation is suggested[9]. As complete resection was not possible in our case, the patient received combined chemoradiation. Factors positively influencing survival are younger age (less than 10 years), female sex, embryonal histology and a more localized disease[2,4]. Orbital RMS has a good prognosis because of the favorable anatomic location, favorable histology and biology (80% embryonal) [2]. Embryonal RMS has a 94% 5-year survival[2]. Overall survival is excellent for groups I, II and III (92%) at 5 years[2].

Conclusion

RMS is the most common primary malignant orbital neoplasm in children. It should be suspected in a child presenting with rapidly progressing unilateral proptosis. It is a highly malignant and life threatening tumor. Due to the multimodal therapy, RMS patients have a better prognosis than before. Hence, early diagnosis is crucial. The immunohistochemical staining of paediatric rhabdomyosarcomas with antibodies to Myogenin provides sensitive and specific diagnostic information. Orbital RMS has a good prognosis because of favorable anatomic location and favorable histology.

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