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Case report

Unilateral proptosis in a child-need for prompt diagnosis

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ABSTRACT

Proptosis in a child often presents as a diagnostic dilemma. Proptosis can be secondary to infection or childhood malignancies. It warrants urgent and relevant investigations to facilitate correct diagnosis and treatment. The common causes of proptosis include infection and malignant lesions. Any delay in intervention in either of the causes can lead to significant morbidity or can impair the vision of the child. An orbital neoplasm in the pediatric age group is an uncommon clinical finding which can initially manifest as proptosis. Here, we report a case of a 2-year-old girl, presenting with progressive swelling of the right eye. An incisional biopsy confirmed the diagnosis of embryonal rhabdomyosarcoma (RMS). RMS is an aggressive tumor; hence, early diagnosis and prompt treatment are highly essential to prevent significant morbidity and can save the vision of the child.

Keywords: Child, Proptosis, Malignancy, Rhabdomyosarcoma

INTRODUCTION

In children, ocular proptosis is the main clinical sign of a spectrum of orbital pathologies. The most common cause of proptosis in children is orbital cellulitis followed by malignant conditions like rhabdomyosarcoma, retinoblastoma, lymphoma, ewings sarcoma and metastasis.[1] Other causes include inflammatory,vascular malformations and developmental anomalies.[1]

Rhabdomyosarcoma is considered the most common soft tissue sarcoma of the head and neck region in children which comprises of 4% of all childhood malignancies and 10% of all cases occurring in the orbit.[2,3] The most characteristic presentation of primary orbital rhabdomyosarcoma is the rapidly growing unilateral painless proptosis.[4]

We report a case of 2 years old girl child who presented with rapidly progressive swelling of right eye preceded by a history of trauma and was diagnosed as embryonal rhabdomyosarcoma after the biopsy of the lesion.

CASE REPORT

A 2-year-old girl child was brought with the complaints of rapidly progressing the right eye swelling for 2 months. The family gave a history of trivial fall before the onset of eye swelling. There was no history of fever, abdominal distention, generalized lymphadenopathy, bony pain, bleeding manifestations, or leukocoria. On examination, there was a diffuse swelling of the right eye noted, measuring 5 × 5 cm with erythema of the upper eyelid [Figure 1]. Rest of the systemic examination was normal.

Baseline investigations revealed hemoglobin of 12 gm/dl, total count of 5300 cells/cu. Mm, and platelet count of 1.64 lakhs/cu. mm. Magnetic resonance imaging of the orbit showed a large, ill-defined mass measuring 4.5 × 3.9 × 3.1 cm, in the superomedial, medial and inferior quadrant, iso-hypointense on T1, and hyperintense on T2 imaging. The medial rectus and inferior rectus muscle could not be identified separately [Figure 2].

Biopsy of the lesion was taken. Histopathological examination showed sheets of small blue cells with focal areas of necrosis (20%) [Figure 3]. Mitosis was 23/10 high power fields. Immunohistochemistry (IHC) was done which showed positive staining for Vimentin, Desmin, and Myogenin [Figure 4a-c]. CD45, CD99, and FLI-1 were negative on IHC, thus confirming embryonal rhabdomyosarcoma (RMS) and ruling out the differential diagnosis of other round blue cell tumors such as lymphoma and Ewing's sarcoma. Bone marrow aspirate

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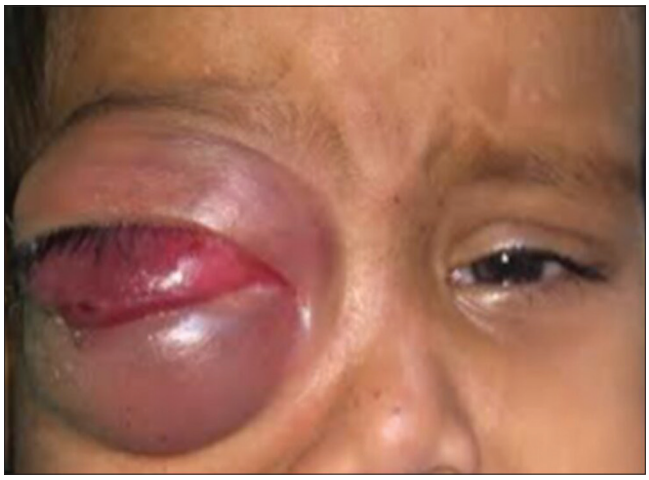

Figure 1: Right eye swelling at the time of presentation.

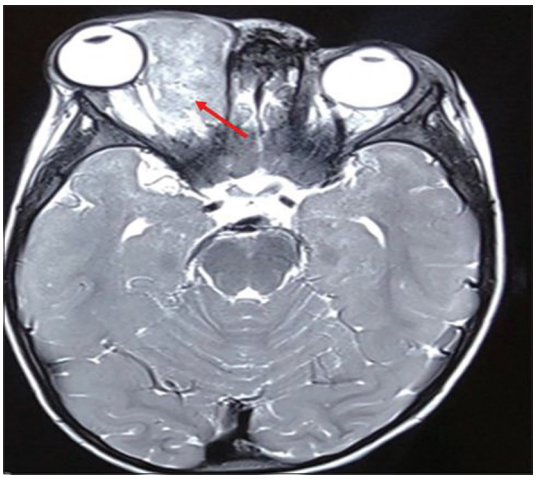

Figure 2: Magnetic resonance imaging showing T2 hyperintense mass lesion in the medial aspect of orbit.

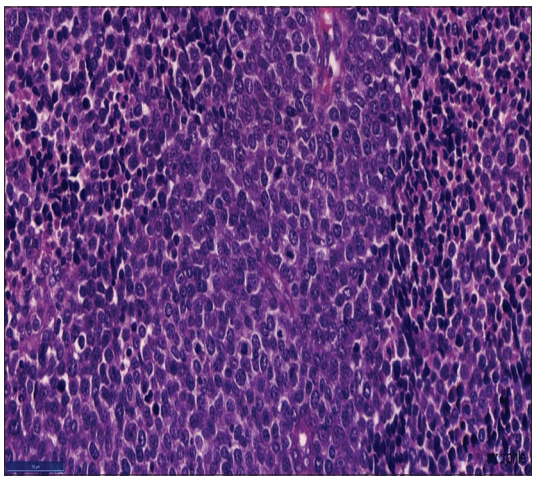

Figure 3: Hematoxylin and eosin ×40 - Sheets of small round blue cells with mitotic figures.

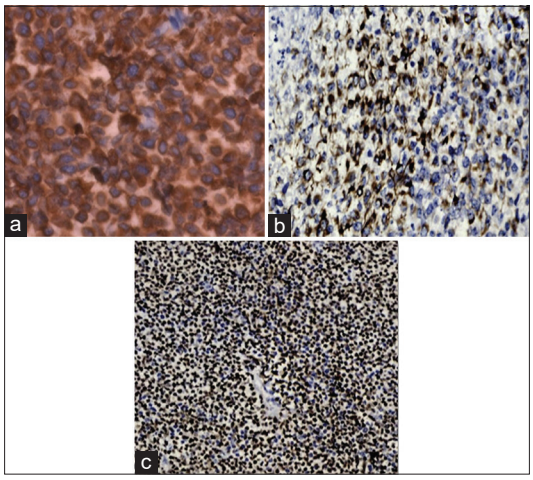

Figure 4: (a) Immunohistochemistry ×40 for Vimentin shows cytoplasmic positivity. (b) Immunohistochemistry ×40 - Desmin shows cytoplasmic positivity. (c) Immunohistochemistry ×40-Myogenin shows nuclear positivity.

Figure 5: Post-chemotherapy image showing significant improvement.

and bone marrow biopsy were done as part of staging investigations and were reported to be normal. She was started on Children's oncology group ARST 0331 protocol which comprises of Vancomycin, Dactinomycin, and Cyclophosphamide and she improved symptomatically after 3 weeks of chemotherapy [Figure 5].

DISCUSSION

In children, ocular proptosis is the main clinical sign of a spectrum of orbital pathologies. The most common cause of

proptosis in children is orbital cellulitis followed by malignant conditions such as RMS, retinoblastoma, lymphoma, Ewing's sarcoma, and metastasis.[1] Other causes include inflammatory, vascular malformations, and developmental anomalies.[1]

RMS is considered the most common soft-tissue sarcoma of the head and neck region in children which comprises 4% of all childhood malignancies and 10% of all cases occurring in the orbit.[2,3] The most characteristic presentation of primary orbital RMS is the rapidly growing unilateral painless proptosis.[4]

Although orbital cellulitis has been considered as the most common cause of proptosis in children, recent studies have quoted malignant lesions as the most common cause, since increased use of antibiotics has reduced the progression of preseptal cellulitis to orbital cellulitis. Masud *et al*., Bajaj *et al*., and Chakraborti *et al*. have all reported malignancy as the primary cause of proptosis in children.[5-7] RMS, retinoblastoma, lymphomas, optic glioma, and Ewing sarcoma are the primary orbital tumors. Metastatic or secondary tumors are neuroblastoma, leukemia, and bone tumors.[8] Opthalmic RMS involves mainly the orbit and other structures such as conjunctiva, uveal tract, and rarely the eyelid. Orbital RMS accounts for 10% of all RMS cases and is the most common orbital malignancy in children.[1] The tumor is derived from the undifferentiated mesoderm and shows phenotypic and biological features of skeletal muscle differentiation. Although the tumor arises *de novo*, history of trauma may be associated with the clinical presentation.[3,9] This condition should be suspected in a child with rapidly evolving unilateral proptosis of sudden onset with a downward and outward displacement of the globe as 2/3rd of these tumors are superonasal.[3] They can also present as lid masses or masses in other parts of orbit with worsening eyelid edema, erythema, chemosis, ophthalmoplegia, and ptosis.[10] Proptosis associated with signs of inflammation has been reported by Kaliaperumal *et al*.[11] Thus, orbital RMS mimicking as orbital cellulitis should also be considered.

Orbital embryonal RMS has a good clinical outcome due to its favorable anatomic site and histologic subtype. Hence, early diagnosis is very essential as the rapid growth and aggressive nature of the tumor results in frequent invasion of adjacent bone, soft tissues, and intracranial extension. Metastatic orbital RMS carries an unfavorable prognosis compared to localized disease.[1,12]

Embryonal and alveolar are the two major histological subtypes which affect the orbit, embryonal being more common (89%).[3] Tumor location within the orbit correlates with histology to a certain extent.[9] Embryonal RMS more commonly arises in the superior nasal quadrants, whereas the alveolar type is usually noted within the inferior orbit. Other small round blue tumors that should be considered as differential diagnosis include Ewing's sarcoma and lymphoma and these can be ruled out by IHC markers. Desmin positivity suggests myogenic origin while Myogenin positivity confirms skeletal muscle origin. To rule out the differentials for other small round blue cell tumors, IHC markers such as CD45 for lymphoma, CD99, and FLI1 for Ewing's sarcoma can be done.

Surgical excision or debulking procedure can be done depending on the extent of tumor involvement. When surgery is not feasible, systemic chemotherapy and radiotherapy are the mainstay of treatment. Multimodality approach and prompt initiation of treatment of RMSs of the head and neck in pediatric patients may lead to significant improvement in the survival rate. Overall 10 year survival rate of orbital RMS is 87%.[13]

CONCLUSION

In a child with proptosis, a thorough general examination, complete blood count with peripheral smear and local imaging study is mandatory to rule out malignant conditions. Histopathology aids in the precise diagnosis, thus playing a major role to confirm the condition. Adequate knowledge of orbital presentation of pediatric malignancies is essential for family physicians and pediatricians to aid in correct diagnosis and timely referral for saving vision.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent.

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Conflicts of interest

There are no con.flicts of interest.

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