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CASE REPORT

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Imaging features of an advanced embryonal orbital Rhabdomyosarcoma in a pediatric case

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ABSTRACT

Rhabdomyosarcoma is a soft tissue highly malignant neoplasm. The most common sites in childhood are the head and neck with only 10% of all cases occurring in the orbit. Clinical manifestations are not specific which makes pre-operative diagnosis challenging. Orbital tumors are mostly detected at early stages, due to their obvious symptoms but they are characterized by rapid growth. Imaging techniques are very contributive in the diagnosis process. The purpose of this review is to provide a general overview of primary orbital Rhabdomyosarcoma through a representative case of a patient that has been managed at our institute as well as a review of the literature.

ARTICLE HISTORY

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Rhabdomyosarcoma; orbit; MRI; pediatrics

Introduction

Rhabdomyosarcoma is a highly malignant soft tissue neoplasm seen in children. It is noted in head and neck including the orbit [1]. In this specific location three sub-types are defined, orbital, parameningeal (nasal cavity, paranasal sinuses, ear, pterygopalatine fossa, mastoid, infratemporal fossa) and non-parameningeal (parotid, palate, tongue, etc.). Orbital Rhabdomyosarcoma comprises tumors that occur in the orbit, ocular adnexal structures or within the eye. Clinically patients usually present with proptosis developing rapidly, globe displacement and vision loss. Metastatic spread includes the lung and bones; however it's not very common and has a very poor prognosis [2,3].

Rhabdomyosarcoma can be divided into histological subgroups, the most common are embryonal and alveolar varieties. Embryonal rhbadomyosarcoma is usually associated with a better prognosis [3].

Clinical and pathological features of this tumor are well described in the literature however, little is known about the radiological appearance of this tumor. The main objective of this work is to review CT-scan and magnetic resonance imaging features of orbital rhabdomyosarcoma.

Case Report

Case presentation

A 2 year-old boy was seen at the pediatric clinic because of a rapidly progressive proptosis of the right eye (less than 1 month). The patient was seen by an ophthalmologist and upon clinical examination, the tumor caused very significant proptosis and supero-medial displacement of the right eye along with blepharoptosis, diffuse chemosis and eyelid edema (Fig. 1). MRI was performed at our institute and demonstrated a 70 × 40 × 51 mm soft tissue mass on the lateral side of the right orbit with well-defined border and irregular lobulated margins, enlarging the orbital area. The mass was isointense to muscle on T1 weighted images (Fig. 2), heterogeneously hyperintense to muscle on T2 weighted images without restriction of diffusion (Figs. 3 and 4). It showed early, persistent and heterogeneous enhancement post contrast (Fig. 5). The margin between the tumor and the lateral rectus muscle was masked, which implied its origin. The mass occupied both the intra- and extra-conal regions with infero-medial displacement of the globe. It also involved and displaced the intraorbital and intracanalicular segments of the optic nerve medially, without any signs of invasion. There were

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Figure 1. Appearance of the patient at presentation.

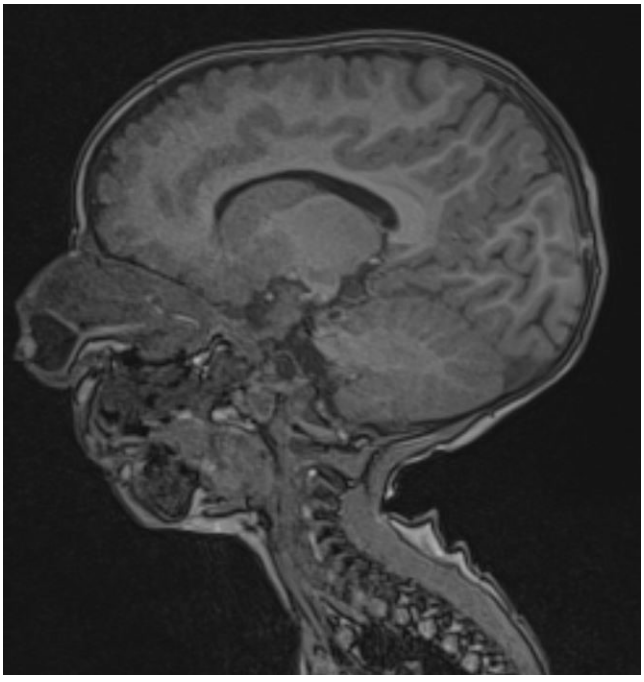


Figure 2. Sagittal T1WI demonstrating an intraorbital soft tissue mass isointense to extraocular muscle, displacing the globe and the optic nerve.

no signs of intracranial extension. CT images using bone windows demonstrated the osseous invasion of the superior and inferior orbital walls and the lateral wall was slightly thinned by the mass (Fig. 6). A CT scan of the chest was performed and no evidence of distant metastases was found.

Histopathology

A biopsy was performed and the histopathological diagnosis of stage II orbital embryonal rhabdomyosarcoma was achieved. The biopsy demonstrated a highly mitotically and active population of primitive looking small round malignant cells. Immuno-histochemical analysis showed that the tumor cells were positive for markers of rhabdomyoblastic differentiation. Fluorescence in situ hybridization (FISH) test for FKHR showed negative results for gene fusion, excluding an alveolar subtype (Figs. 7 and 8).

Treatment, out come and follow-up

He was treated with chemo according to standard protocol (VAC regimen: vincristine, actinomycin D, and cyclophosphamide), currently under course.

Discussion

Rhabdomyosarcoma is a high-grade malignant mesenchymal tumor, it's the most frequent pediatric soft tissue sarcoma, constituting 3%–5% of all malignancies with a predilection for the head and neck [2,4]. The true cause of Rhabdomyosarcoma is unclear. Certain factors can be incriminated such as genetic predisposition and environmental exposures (parental smoking, use of antibiotic…) [2]. Microscopically, the four main histological types are: embryonal, pleomorphic, alveolar and botryoid. Embryonal being the most frequent variant found in the head and neck region whereas the alveolar and pleomorphic types are rare in the orbit.

Clinically patients usually present with proptosis developing rapidly or globe displacement which is usually downward and outward because two-thirds of these tumors are supero-nasal. Metastatic spread of orbital Rhabdomyosarcoma is not very common, if left untreated it can spread to the lung and bone. It can also extend intracranially through invading the orbital bones. Metastatic cases of orbital Rhabdomyosarcoma have a poor prognosis compared to localized forms [1,3,5]. The diagnosis of orbital Rhabdomyosarcoma can be suspected based on the clinical and radiological features. Patients might experience pain, visual loss, signs of sinusitis and alterations of the conjunctiva, lid, or caruncle. The diagnosis is most suspected when the presentation is unilateral and rapidly progressive

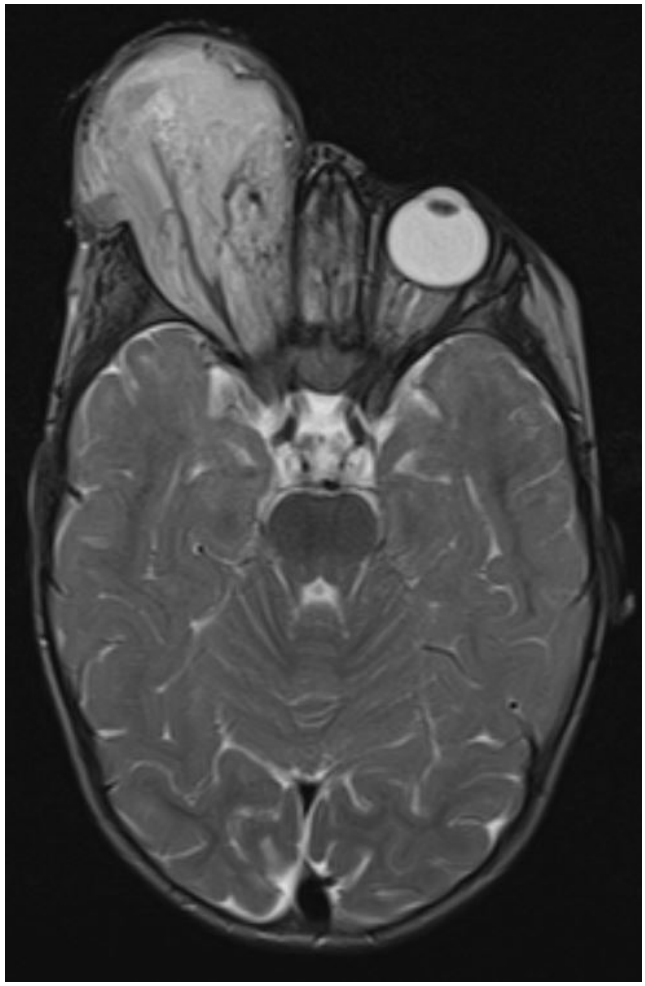


Figure 3. Axial T2WI showing an intraorbital soft tissue mass, with well-defined border and irregular margins, enlarging the orbital area, hyperintense to extraocular muscle.

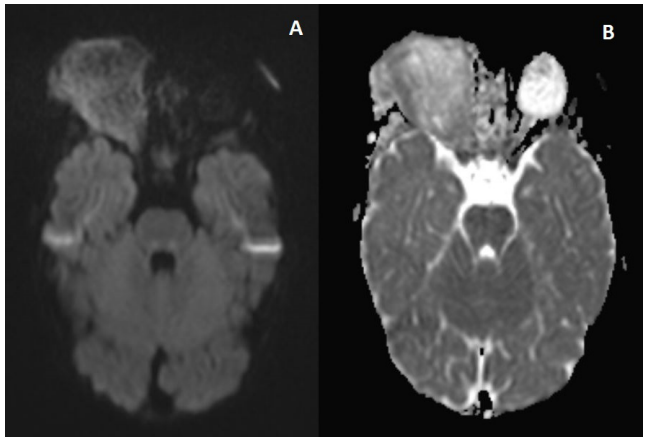


Figure 4. (A) Axial DWI MRI showing hypointense signal. (B) Axial ADC MRI showing predominant hyperintense signal.

and symptoms depend mostly on the origin the lesion [3,6]. Clinically differential diagnoses include

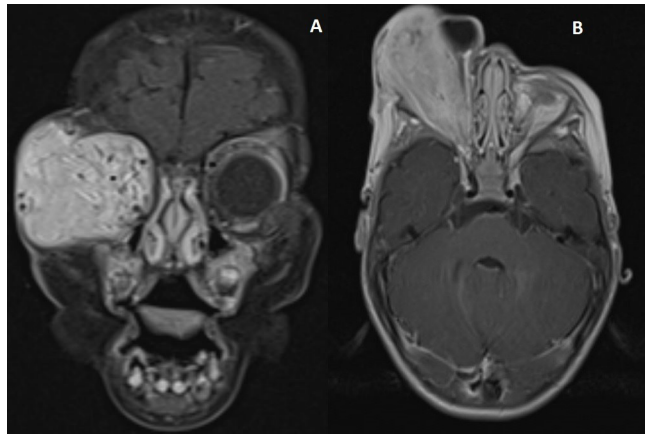


Figure 5. (A) coronal T1WI (B) Axial T1WI with fat saturation after administration of contrast showing heterogeneous enhancement.



Figure 6. CT image using bone window, demonstrating the osseous invasion of the right lateral orbital wall.

orbital cellulitis, lymphangioma, Langerhans cell histiocytosis dermoid cyst, hemangioma, metastatic neuroblastoma, and lymphoma [3].

CT scan and MRI are essential they help eliminate some differential diagnoses, determine the location, size and extension. Imaging techniques are also fundamental in treatment evaluation to look for any residual tumor or signs of recurrence.

CT generally shows a moderately well defined, homogeneous, round to ovoid orbital mass that is isodense to the extraocular muscles, with moderate to marked enhancement post contrast administration. A heterogeneous appearance can indicate the presence of hemorrhage and necrosis. CT is especially used to determine bone involvement. In the early stages, there is not bone destruction. In more advanced stages, we note invasion of adjacent structures, the presence of calcifications, destruction

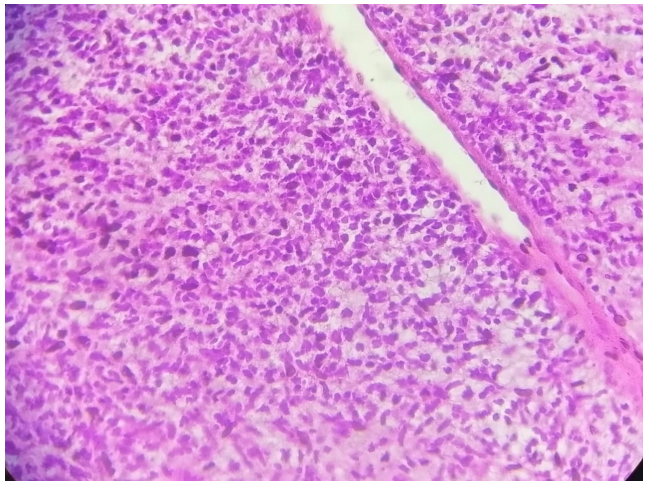


Figure 7. Rhabdomyosarcoma embryonal (H&E, ×40): round and spindle-shaped cells in sheets disposition, non-abundant cytoplasm and high mitotic activity.

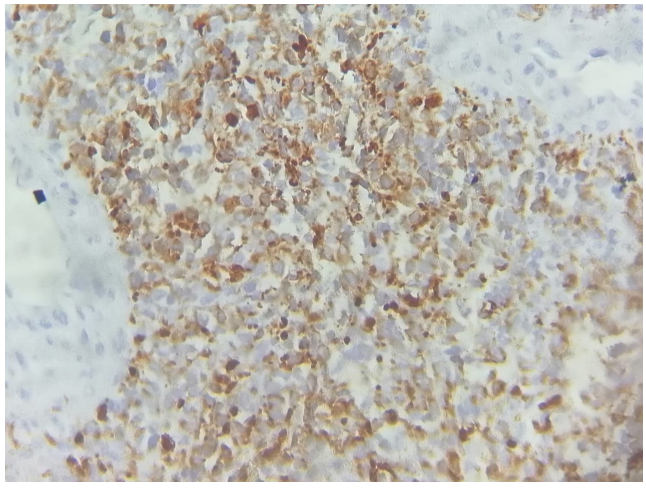


Figure 8. Small malignant tumoral cells (H&E, ×40) with positive immunohistochemistry staining for desmin.

of adjacent bone and sometimes extension to the sinuses or nasopharynx. Extra ocular muscles might be displaced by the tumor, however the belly of the muscle is not enlarged [3,6]. In our case CT scan demonstrated orbital walls invasion by the mass. CT is also used to depict metastatic spread, especially lung metastasis in order to stage the tumor. MRI is the imaging modality of choice to assess Rhabdomyosarcoma due to its superior soft-tissue resolution, it's very efficient to determine the site of origin of the tumor. It appears as a large mass isointense to extraocular muscles and hypointense to orbit fat on T1 weighted images and hyperintense to extraocular muscles and orbit fat on T2 weighted images with moderate to marked enhancement post gadolinium administration. Increased signal areas on T1 and T2 weighted images indicates focal hemorrhaging. The globe may be distorted or displaced but rarely invaded [5–7]. In our case we noted distortion and displacement of the globe without signs of invasion. Diffusion weighted imaging can be very useful. A study has been published by Lope et al. [8] in the journal of AAPOS, demonstrated that Rhabdomyosarcoma generally presents restricted diffusion. Hemangioma and Rhabdomyosarcoma present similar signal intensity in T1 and T2 weighted images, DWI/ADC can be used to make the difference, since Hemangioma has increased DWI/ADC [8]. The alveolar subtype is usually partly necrotic with lobulated margins and presents heterogeneous signal on T2 weighted images and heterogeneous enhancement after contrast administration. The Pleomorphic subtype also presents necrosis and it appears isointense to hyperintense to muscle on T1 weighted images, hyperintense on T2 weighted images with marked heterogeneous enhancement post contrast administration. The tumor heterogeneity and necrosis in the pleomorphic subtype is probably due to the irregular arrangement of cells and necrosis whereas in the alveolar type there is a loose arrangement of central cells, which explains the appearance. The presence of hyperintensities on T1 weighted images indicates the presence of areas of high protein or hemorrhage. The embryonal subtype is usually parameningeal (rarely orbital), presents as a homogenous, ill-defined mass with minimal necrosis and destruction of adjacent bones. It usually displays moderate enhancement after contrast administration [9–11]. CT and MR imaging are complimentary in both the evaluation and staging of orbital Rhabdomyosarcoma. MRI on the other hand is the examination of choice for long term follow-up.

Treatment consists of chemotherapy, but sometimes radiotherapy and excision are indicated depending on the age. Complete resection of the orbital mass is generally difficult due to its proximity to delicate orbital structures and high risk of complications. Despite the initial stage or group, all patients should be treated with chemotherapy. The standard chemotherapy protocol is VAC regimen (vincristine, actinomycin D, and cyclophosphamide). Radiotherapy helps present local recurrence. Overall orbital Rhabdomyosarcoma has a good prognosis especially the embryonal subtype. Risk factors for poor prognosis include age under 12 months, size of tumor >5 cm, alveolar subtype, undifferentiated types, lymph node involvement and presence of metastasis [12,13].

Conclusion

Knowledge of the clinical, histopathological, and imaging features of Rhabdomyosarcoma is essential to establish prompt diagnosis and better management of this tumor. A multidisciplinary approach is crucial for a better prognosis.

References

[1] Rootman J. (Ed.) Neoplasia. Diseases of the orbit: a multidisciplinary approach. Lippincott Williams and Wilkins, Philadelphia, PA, Vol. 54, pp 262–8, 2003.

[2] Turner JH, Richmon JD. Head and neck rhabdomyosarcoma: a critical analysis of population-based incidence and survival data. Otolaryngol Head Neck Surg 2011; 145:967–73.

[3] Shields JA, Shields CL. Rhabdomyosarcoma: review for the ophthalmologist. Surv Ophthalmol 2003; 48:39–57.

[4] Miller RW, Young JL Jr, Novakovic B. Childhood cancer. Cancer 1995; 75:395–405.

[5] Conneely MF, Mafee MF. Orbital rhabdomyosarcoma and simulating lesions. Neuroimaging Clin N Am 2005; 15:121–36.

[6] Freling NJ, Merks JH, Saeed P, Balm AJ, Bras J, Pieters BR, et al. Imaging findings in craniofacial childhood rhabdomyosarcoma. Pediatr Radiol 2010; 40:1723–38.

[7] Sohaib SA, Moseley I, Wright JE. Orbital rhabdomyosarcoma-the radiological characteristics. Clin Radiol 1998; 53:357–62.

[8] Lope LA, Hutcheson KA, Khademian ZP. Magnetic resonance imaging in the analysis of pediatric orbital tumors: utility of diffusion-weighted imaging. J AAPOS 2010; 14:257–62.

[9] Allen SD, Moskovic EC, Fisher C, Thomas JM. Adult rhabdomyosarcoma: cross-sectional imaging findings including histopathologic correlation. AJR 2007; 189:371–7.

[10] Hawkins WG, Hoos A, Antonescu CR, Urist MJ, Leung DH, Gold JS, et al. Clinicopathologic analysis of patients with adult rhabdomyosarcoma. Cancer 2001; 91:794–803.

[11] Komdeur R, Klunder J, van der Graaf WT, van den Berg E, de Bont ES, Hoekstra HJ, et al. Multidrug resistance proteins in rhabdomyosarcomas: comparison between children and adults. Cancer 2003; 97:1999–2005.

[12] Arndt CAS, Stoner JA, Hawkins DS, Rodeberg DA, Hayes-Jordan AA, Paidas CN, et al. Vincristine, actinomycin, and cyclophosphamide compared with vincristine, actinomycin, and cyclophosphamide alternating with vincristine, topotecan, and cyclophosphamide for intermediate-risk rhabdomyosarcoma: Children's oncology group study. J Clin Oncol 2009; 27:5182–8.

[13] Oberlin O, Rey A, Anderson J, Carli M, Raney RB, Treuner J, et al. Treatment of orbital rhabdomyosarcoma: survival and late effects of treatment - results of an international workshop. J Clin Oncol 2001; 19:197–204.