Case Report
Middle East Journal of Cancer; April 2016; 7(2): 93-95

Bilateral Intraocular Rhabdomyosarcoma: A Case Report

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Abstract

Here we report the case of a 1.5-year-old Iraqi boy who was referred for chemotherapy after left eye enucleation. The patient had a history of left eye leukocoria since 2 months of age. According to history, physical examination and paraclinical work up, he was first diagnosed as a case of retinoblastoma by an ophthalmologist. However, the pathology report favored embryonal rhabdomyosarcoma. In conclusion, a patient with leukocoria should be evaluated carefully for other underlying malignancies.

Keywords: Orbital tumor, Pediatric, Rhabdomyosarcoma

Introduction

Rhabdomyosarcoma (RMS) is a highly malignant tumor that usually manifests as an expanding mass. Superficial tumors may be palpable and detected early, but those in deep locations (e.g., the retroperitoneum) may increase to a large size before causing symptoms. The head and neck region and in particular, the orbit, represent a major anatomic site for RMS. However, RMS can primarily involve the eyelid, conjunctiva, and rarely, the uveal tract.[1] Orbital RMS should be considered in the differential diagnosis of any child with a progressive unilateral proptosis. Diagnosis consists of a detailed history, ocular examination, imaging studies that include computed tomography (CT) scan or magnetic resonance imaging (MRI), and biopsy. The differential diagnosis for orbital RMS includes a group of childhood inflammatory or infectious processes, vascular, and neoplastic conditions that result in proptosis.

Imaging is of particular importance in diagnosis of orbital RMS and its subsequent management. Imaging guides appropriate surgical planning for incisional or excisional biopsy of the orbital tumors.[2, 3] The ultimate diagnosis of RMS requires biopsy for histopathological evaluation.[3]

Until now, there have been a

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Received: May 10, 2015; Accepted: October 25, 2015

limited number of intraocular RMS case reports in the literature.[4] Bilateral intraocular RMS is very rare. Herein, we report the case of a 1.5-year-old boy with bilateral intra ocular RMS.

Case report

A 1.5-year-old boy was referred to the Pediatric Oncology Clinic after enucleation of his left eye for chemotherapy. The patient had history of left eye leukocoria for the past 13 months. According to the ophthalmologist note, physical examination was unremarkable except for left eye protrusion. Complete blood counts and coagulation profile were all within normal limits except for a minimal rise in lactate dehydrogenase (LDH: 535 IU/dl). Before enucleation, MRI of the brain and orbit revealed a heterogeneous markedly enhancing lesion within the left orbital cavity that involved the left eye globe with significant extension to the preseptal region. A sign of a small (approximately 3 mm), round, mildly enhancing lesion was also detected at the lateral aspect of the right eye globe which could be a focus of tumor. The findings were most consistent with a retinoblastoma. A whole body scan with 99mTc was normal with no evidence of involvement of the bones adjacent to the tumor. Chest CT scan and abdominopelvic sonography were unremarkable.

At first, according to history and physical examination, the patient was diagnosed with retinoblastoma by an ophthalmologist. He underwent left eye enucleation and was subsequently referred to an oncology hospital for further evaluation and consideration of chemotherapy.

Nonetheless, bone marrow aspiration and trephine biopsy revealed mild hypocellularity with patchy infiltration of small round cells.

The cut section of the orbital mass demonstrated an infiltrative gray white firm mass. The tumor cells were predominantly composed of small hyperchromatic malignant cells with no fibrous component without any tendency for resetting. The resected surgical margin was grossly involved by tumor.

Immunohistochemical (IHC) study on paraffin-embedded formalin-fixed enucleation tissue, bone marrow aspiration and biopsy showed tumor cells positive for desmin, MyoD1, and vimentin, and negative for LCA, CD20, CD23, CD30, neuron-specific enolase, MIC2, and synaptophysin. These findings favored RMS (Figures 1, 2). The patient received the ICE chemotherapy regimen that consisted of ifosfamide (1.8 gm/m2 I.V. days 1 to 3 with mesna), carboplatin (450 mg/m2/day 1) and etoposide (100 mg/m2 I.V. days 1 to 3) every three weeks. Unfortunately after the second course of chemotherapy, his parents wanted to continue his treatment in Iraq. We have no additional information about the patient's management and outcome of his right eye.

Discussion

Many ocular and orbital tumors of childhood are congenital with early presentations. Developmental cysts comprise half of the orbital

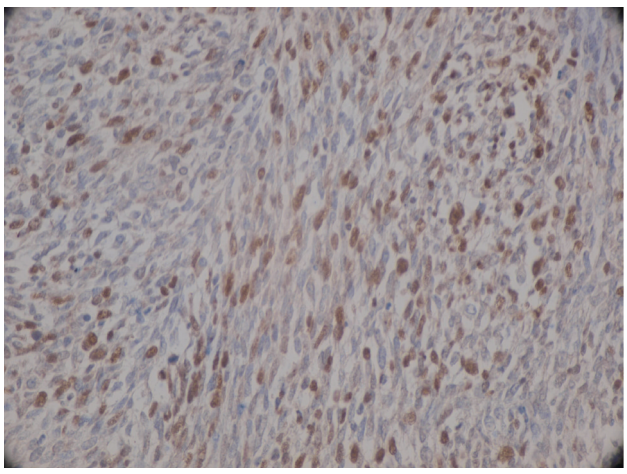

Figure1. Tumor cells with MyoD1 positivity (250×).

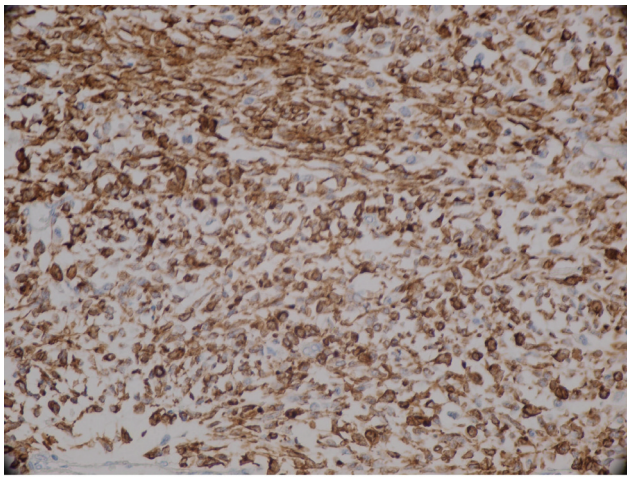

Figure 2. Diffuse desmin positivity in tumor cells (250×).

cases; capillary hemangioma is the second most common orbital tumor.[3] The most common intraocular malignant lesion is retinoblastoma.[2] Choroidal melanoma, which is common in adults, is extremely rare in children. The orbit and choroid are the most common locations for metastases in children and adults.

In a typical patient, the orbital tumor may present with crossed eyes, double vision, eyes that do not align, eye pain and redness, poor vision, and differing iris colors in each eye.[1,3]

Rhabdomyosarcoma is the most common primary orbital malignancy in the pediatric age group and can be found in the orbit, eyelid, conjunctiva, and uveal tract.[4] Rhabdomyosarcoma usually manifests clinically as rapidly progressive exophthalmos and displacement of the globe. On occasion it presents insidiously, mimicking other (benign) tumors both clinically and radiographically.

Diagnosis of RMS is based on biopsy, CT, and MR images.[5] In our patient, regarding the inferior displacement of the left eye ball, extraocular malignancies with ocular invasion such as rabdomyosarcoma could not be ruled out.

Davidson et al. reported a case of unilateral intraocular RMS who was the sibling of a patient with cerebellar medulloepithelioma.[6] In another study, Zimmerman et al. indicated that rhabdomyosarcomatous differentiation might occur in malignant intraocular medulloepitheliomas.[7]

An interesting aspect of our case was the involvement of both eyes. Clinical features and imaging findings indicated the presence of an intra-ocular tumor, most probably retinoblastoma. However, the IHC study favored RMS.

Pediatricians play a vital role in diagnosis of pediatric ocular tumors. Early recognition of signs and symptoms of pediatric ocular tumors is very important and should be undertaken for saving vision, prompt ophthalmologic evaluation, and treatment.

In this case enucleation was not the correct treatment strategy, whereas early chemotherapy might have preserved his vision.

In conclusion, according to the published literature, this was the first case of biopsy proven bilateral intraocular RMS.

Acknowledgments

The authors would like to thank Dr. Mohammad Reza Farahmandfar and Ms. A. Keivanshekouh at the Research Improvement Center of Shiraz University of Medical Sciences for improving the use of English in the manuscript.

Conflict of interest

No conflict of interest is declared.

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