Scholars Journal of Applied Medical Sciences (SJAMS) **ISSN 2320-6691 (Online)**
Sch. J. App. Med. Sci., 2015; 3(9D):3375-3377 **ISSN 2347-954X (Print)**

(An International Publisher for Academic and Scientific Resources)
www.saspublishers.com DOI: 10.36347/sjams.2015.v03i09.047

Case Report

Extraocular Retinoblastoma: Case Report of a Late Diagnosis in a Child

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Abstract: Retinoblastoma (RB) is a highly malignant tumor of the retina and is believed to be the commonest childhood primary malignant intraocular neoplasm. The survival rate is very low in developing countries contrary to developed countries where it may reach 95 %. The case reported here was diagnose at an extraocular stage.
Keywords: Retinoblastoma, malignant tumor, intraocular neoplasm

INTRODUCTION

Retinoblastoma (RB) is the most common primary cancer of the eye in children [1]It is the most common primary intraocular tumor in children and results from mutations in the tumor suppressor retinoblastoma gene (RB1) located on chromosome 13 [2]. It is sporadic in 60% and hereditary in 40% [3]. Retinoblastoma as a highly malignant intraocular tumor requires an early diagnosis and immediate treatment [4]. The average age for diagnosis varies from 12 months in bilateral cases to 23 months in unilateral tumors [5]. The association of bilateral retinoblastoma with intracranial tumors, termed "trilateral" retinoblastoma, is a well recognized but uncommon syndrome. The intracranial tumor arises most often in the pineal region but can also be a suprasellar or parasellar tumor [6]. The clinical presentation of RB includes leucocoria, strabismus, conjunctival chemosis, proptosis, and even blindness, which are attributed to late presentation and diagnosis [7].

Management options include Laser therapy, chemotherapy, radiotherapy, enucleation, and exenteration. A clinical staging system is essential to enable proper definition, treatment plan, outcomes assessment of disease, and international communication [8].

In case of early diagnosis, conservative treatments like chemotherapy or laser may be enough to cure the disease. In developing countries, patients present commonly late; so oftentimes enucleation or exenteration are the ''harmless solutions''. We report a case which presented late. It was staged "E" according to the international classification of retinoblastoma [9].

CASE REPORT

A 4 years old female child was referred to our hospital for exophtalmos of the left eye. According to her mother it started with a leucocoria one year ago, then it increased progressively and the eye begun to push outward. The child was taken to a general practitioner who eventually referred her. No history of RB was found in the siblings. Ophthalmic examination showed a normal right eye whereas in the left eye there was a huge proptosis with conjunctival chemosis. Computed tomography (CT) scan of orbit revealed calcified mass of the vitreous and the retina with extension to the optic nerve.

Fig-1: Photograph of the child showing the extraocular RB in the left eye

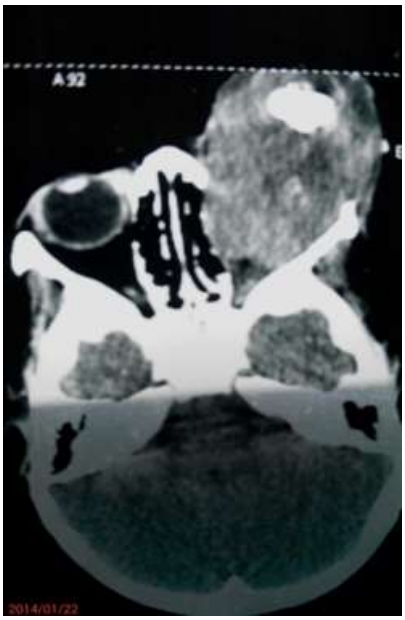

Fig.2: CT scan showing the calcifications in the left eye

DISCUSSION

Retinoblastoma is the most common intraocular tumor in childhood; characteristically aggressive, it initially involves the globe, with posterior extension to the cranium and central nervous system via optic nerve, orbit and metastatic spreading to the whole body excluding the lungs. Prognosis of extraocular retinoblastoma (RB) has remained relatively poor for both orbital and distant metastatic diseases [10].

The retinoblastoma gene encoded on chromosome 13q14 was first described tumor suppressor gene. It can be sporadic or inherited. Leucocoria is the most common presentation. Strabismus, poor visual tracking and glaucoma are other presenting features [11]. Orbital inflammation, hyphema and irregular pupil, fungating ocular mass are signs of advanced disease. In developing countries, retinoblastoma presents very late in its extraoccular stage either with an orbital mass (proptosis) or with distant metastasis in bone marrow, lymph node or central nervous system extraocular extension, either directly through the sclera or via extension along the optic nerve, is a poor prognostic factor [12, 13]

In more developed countries overall survival rates exceed 95%, a success attributable to both early detection and prompt access to enucleation services [14]. In less developed countries, retinoblastoma is still a life-threatening disease [15]. Survival rates are about 70% in low and middle income countries [16]

Therapeutic plan usually requires a multidisciplinary approach. In cases of unilateral disease with large tumor where no useful vision can be preserved enucleation must be performed early. In advanced disease and bilateral cases systemic chemotherapy includes vincristine, carboplatin and etoposide. Most tumors that are confined to one eye are cured [17]

Timely enucleation reduces risk of metastatic spread, morbidity, side effects of chemotherapy and focal laser treatment, and repeated examinations under anesthesia [18]

CONCLUSION

Despite the improving treatment methods of retinoblastoma, it is still a dreadful disease in developing countries where the survival rate remains low. Ignorance and poverty are among others the cause of the poor outcome.

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