VOLUME-7, ISSUE-10, OCTOBER-2018 • PRINT ISSN No 2277 - 8160

Original Research Paper

Radiology

BILATERAL RETINOBLASTOMA IN AN 11 MONTHS OLD BOY: CASE REPORT

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KEYWORDS :

INTRODUCTION

Retinoblastoma is rare malignant congenital intraocular tumour arising from primitive photoreceptor cells of retina[1]. It is the most common intraocular tumour in children[1,2,] with an incidence of 1/15,000–20,000 live births[3]. It may occur at any age but most often it occurs in younger children, usually before the age of two years[3].

Retinoblastoma is the first disease for which a genetic etiology of cancer has been described and the first tumor suppressor gene identified. Loss or mutations of both alleles of the retinoblastoma gene *RB1*, localized to chromosome 13q1 are required to develop the disease[3].

Sixty per cent of retinoblastomas are unilateral and most of these forms are not hereditary (median age at diagnosis two years). Retinoblastoma is bilateral in 40% of cases (median age at diagnosis one year). All bilateral and multifocal unilateral forms are hereditary[3].

The two most frequent symptoms revealing retinoblastoma are leukocoria and strabismus. Hyphema, buphthalmia, orbital cellulites and exophthalmia may also be observed3. Other rare presenting signs of retinoblastoma include differences in pupil size (anisocoria), differences in iris color (heterochromia), and abnormal eye movements (nystagmus).

Disease complication includes metastasis to meninges, bone marrow, lung, liver and lymph nodes[1,4].

Here presented is an unusual case of bilateral retinoblastoma, presenting with rare feature of jerk nystagmus. This case report highlights some of the uncommon presenting symptom of a rare disease, which will hopefully add value to the existing knowledge of the disease epidemiology.

CASE REPORT

A.S is an eleven (11) months old infant who presented with history of "whitish eye" bilaterally and left ocular deviation for six (6) months prior to presentation. Patient is said to bump into objects occasionally when crawling. No associated history of fever or trauma before the onset of symptoms. Patient is a product of full term booked pregnancy born to 22 year old primipara. Labour and immediate post natal period were uneventful. Developmental milestones are said to be appropriate for patient's age. No history of similar ailment in the family. Parents are not related (i.e. marriage not consanguineous).

General examination revealed a playful infant, afebrile and not dehydrated. Visual acuity exam reports that patient moves eyes towards light. There was jerk nystagmus, worse on the left eye. Eyelids were normal bilaterally. Cornea was clear on the right while cloudy on the left. The pupils were both dilated (worse on the left) and unreactive. No red reflex detected on fundoscopy bilaterally.

On account of the findings of bilateral leukocoria and other supporting history and examination findings, a diagnosis of bilateral retinoblastoma was made by the ophthalmologist and hence referred to radiology department for ocular B-scan and Magnetic Resonance Imaging (MRI) examination.

High resolution ultrasound scan of the orbits shows normal sized globes, measuring 21.7 and 22.2mm in their anterior-posterior dimension, on the right and left sides respectively. There is however, mixed echogenic mass lesion seen arising from the retina, almost occupying the entire vitreous segment of the left eye, protruding beyond the equator into the anterior segment. There is associated extensive area of calcific echo-density seen within the mass. Similar but smaller mass is seen on the right eye, arising from posterior-lateral wall of the retina (figure 1).

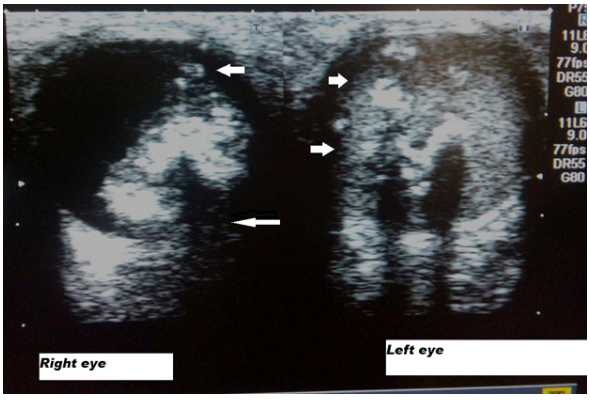

Figure 1: High resolution sonogram of the right and left eye showing mixed echogenic masses filling the entire globe on the left and filling almost half of the right globe (arrows). There are multiple foci of calcific echodensities with posterior acoustic shadowing within the masses.

Complimentary transfontanelle ultrasound scan was then performed through patent anterior fontanelle, which showed no intracranial mass. Brain MRI was not done due to unavailability and financial constrain. The care givers opted for brain CT scan instead.

The CT scan confirmed the calcification within the globe masses bilaterally (figures 2, 3 & 4). No clinical or CT sign of proptosis. No extension into the adjacent optic nerves or extra-ocular muscles was seen. No mass was seen in the region of pineal gland or anywhere in the brain.

Based on the clinical and imaging findings, final diagnosis of bilateral retinoblastoma (worse on the left) was arrived at. Parents were counselled and the child underwent left eye enucleation. The histology confirmed retinoblastoma, limited to the globe. Patient is currently being prepared for chemotherapy to save right globe and if possible, the right eye vision.

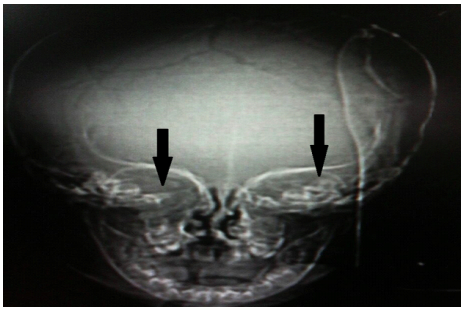

Figure 2: Frontal scanogram showing foci of calcifications projected over the orbits bilaterally (arrows).

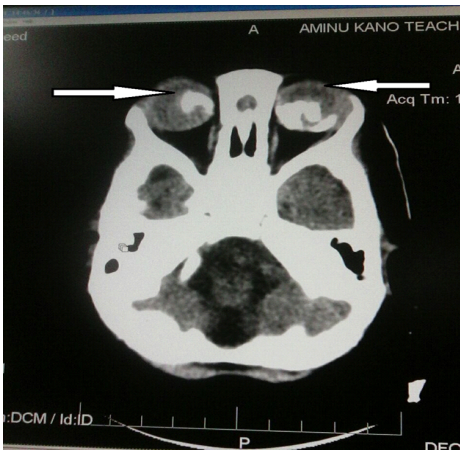

Figure 3: Contrast enhanced axial computed tomographic scan of the brain at the level of orbits, showing mixed density poorly enhancing masses (arrows). The extra-ocular muscles appear within normal limits.

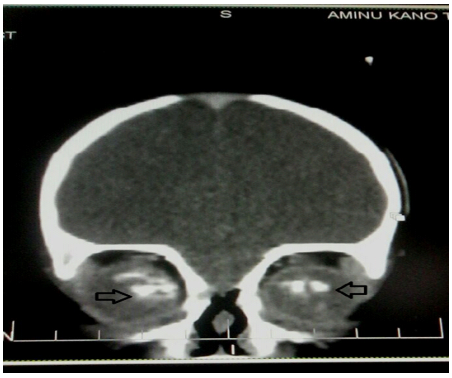

Figure 4: Coronal reformatted CT image at the level of orbits, showing a larger mass with more extensive calcification on the left eye.

DISCUSSION

The incidence of retinoblastoma is not distributed equally around the world. It seems to be higher (6–10 cases per million children) in Africa, India, and among children of Native American descent in the North American continent. The increased incidence in those groups occurs primarily in unilateral cases. Whether these geographic variations are caused by ethnic or socioeconomic factors is not well known. An increased incidence of retinoblastoma has been associated with poverty and low levels of maternal education[5].

Retinoblastoma is by definition a tumor of young children, and the age at presentation correlates with the risk of bi-laterality. Patients with bilateral retinoblastoma tend to present at a younger age (usually before 1 year of age) than patients with unilateral disease (often in the second or third year of life)[5]. This trend was exemplified by the index case who presented with bilateral disease before his first birthday.

The index case presented with leukocoria (loss of the normal red retinal reflex), which as highlighted earlier, is the commonest presenting feature. It is however nonspecific and can be found in retinoblastoma, Coats' disease, toxocariasis, optic nerve drusen, retinopathy of prematurity, persistent hyperplastic primary vitreous (PHPV), and phthisis bulbi[4]. History of term-delivery, findings of normal-sized globes (i.e., no micropthalmia) and dense calcification rules out the possibility of prematurity-associated retinopathy (retrolental fibroplasia). Phthisis bulbi and PHPV are usually associated with microphthalmia and are therefore less likely in this index patient. Furthermore, PHPV and toxocaraiasis (which is usually infectious in nature and often a unilateral eye disease), rarely calcify[1,4].

Strabismus (squint) is being described as the second commonest presenting symptom of retinoblastoma[1,3,4,6], which is explained by tumor in the region of the macula disrupting the sensory input needed to keep the globes aligned[4]. The index patient in addition, presented with nystagmus, a feature rarely seen in retinoblastoma.

Hereditary retinoblastoma also occurs in conjunction with midline intracranial masses, usually pineoblastoma, of either the suprasellar or the pineal region (trilateral disease)[6] or in both suprasellar and pineal regions (quadrilateral disease)[1]. The incidence of trilateral retinoblastoma is reported to be between 1.5% and 5% of patients with retinoblastoma[6]. Rodjan F et al[7] showed that trilateral retinoblastomas detected synchronously with the bilateral disease on routine (baseline) brain screening were significantly smaller, more frequently asymptomatic, and could have a better prognosis compared to those found after the diagnosis (metachronous trilateral retinoblastomas). Complimentary brain imaging done in this patient (transfontanelle USS and brain CT scan) helped to rule out the presence of trilateral or quadrilateral disease. It also provided the opportunity for early detection of possible trilateral disease.

The diagnosis is usually established by the ophthalmologist on the basis of fundoscopy and ultrasound scan. CT provides important information about the presence of calcifications, the size of the globes, and the contrast enhancement pattern that can help narrow the differential considerations[4]. Magnetic resonance imaging (MRI) may contribute to pre-treatment assessment, diagnostic confirmation, detection of local tumour extent, detection of associated developmental malformation of the brain and detection of associated intracranial primitive neuroectodermal tumour (trilateral retinoblastoma)[2].

Management of patients with retinoblastoma must take into account the various aspects of the disease: the visual risk, the possibly hereditary nature of the disease, the life-threatening risk. Enucleation is still often necessary in unilateral disease; the decision for adjuvant treatment is taken according to the histological risk factors. Conservative treatment for at least one eye is possible in most of the bilateral cases[3]. Factors to be considered in the treatment decisions include intraocular and extra-ocular stage, laterality, and potential for vision. Ocular salvage treatments include systemic or intra-arterial chemotherapy, aggressive focal treatments (photocoagulation, thermotherapy, cryotherapy, and brachytherapy), and external beam radiation therapy[5]. The successful management of retinoblastoma depends on the ability to detect the disease while it is still intraocular; disease stage correlates with delay in diagnosis. In developing countries, late referrals are strongly associated with orbital and metastatic disease[5]. The index case underwent left eye enucleation and chemotherapy as part of conservative management to salvage the right vision.

Calcification is a favorable prognostic sign, while contrast enhancement is unfavorable[1]

SUMMARY

An unusual case of bilateral leucokoria due to retinoblastoma in an

11 months old infant was presented. The role of radiology in diagnosis and evaluation of the extent of the disease as well as ruling out associated intracranial primitive neuroectodermal tumor (trilateral/quadrilateral disease) was highlighted.

Early diagnosis is essential in salvaging the vision of the affected eye.

Children with bilateral disease are at high risk of developing second malignancies and therefore need to be followed closely. Radiation therapy is avoided whenever possible in this group of children5.

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