

Retinoblastoma: A spectrum of manifestations in three cases on B-scan

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Abstract

Retinoblastoma is a primary intraocular neoplasm of childhood, presenting with leukocoria. We present a spectrum of three cases. First two cases are of bilateral retinoblastoma and last case is a retinoblastoma of right eye. All cases are confirmed to be retinoblastoma on operation and consequent histopathological study.

Key words: B scan, posterior segment, retinoblastoma

Introduction

Retinoblastoma is the commonest primary malignant tumor of childhood and comprises about 30% percent of all intraocular tumors. Nevertheless, it is rare, occurring in about 1 in 20,000 births. It is one of the commonest causes of leukocoria in children and infants. The USG B-scan helps in easy, noninvasive diagnosis of this intraocular tumor and is cost-effective. We present here a spectrum of three cases of retinoblastoma.

Case Reports

Case 1

A two-years old boy presented with bilateral white pupils. The -ray chest radiograph and ultrasound abdomen of the same patient were normal. B-scan of both eyes revealed hyperechoic intraocular lesions, even at low gain settings with irregular contours and a few calcium deposits within, seen predominantly in the posterior segments of both eyeballs [Figures 1, 2]. There was no extrascleral extension and optic nerve involvement. A diagnosis of bilateral retinoblastoma was made. A computerized tomography (CT) of the brain revealed no intracranial extension of the mass. The patient was advised excision of both eyes, which he underwent subsequently. Histopathological sections of the operated specimens confirmed the diagnosis of retinoblastoma.

Case 2

An 18-months old child presented with bilateral white

pupils. On B-scan [Figures 3, 4] there were bilateral hyperechoic intraocular tumors with irregular contours as well as multiple calcium deposits seen as highly reflective foci in the tumors. The tumor on the right was larger. There was no extrascleral extension and optic nerve involvement of the mass lesions. A suspicion of bilateral retinoblastoma was considered, which was confirmed on operative findings and subsequent histopathological study. The cut specimen of the left eye [Figure 5] revealed a whitish intraocular tumor, which was confirmed to be retinoblastoma on histopathological study. An option of chemotherapy was kept in mind, after enucleation of the eyes, but since follow-up USG did not show metastases, chemotherapy was not given.

Case 3

A three-years old girl presented with right-sided leukocoria. A B-scan of the right eye [Figure 6] revealed an intraocular lesion similar in appearance to those seen in the previous cases, with calcium deposits within the tumor. The child underwent excision of the right eye, confirming the diagnosis of retinoblastoma on histopathological study. The histopathology study of the tumor revealed small clear blue cells forming focal rosettes in the tumor arising from the retina extending into the vitreous [Figure 7].

Discussion

Retinoblastoma is the commonest primary malignant intraocular tumor of childhood, comprising 30% of all ocular tumors. It is nevertheless rare, with an incidence

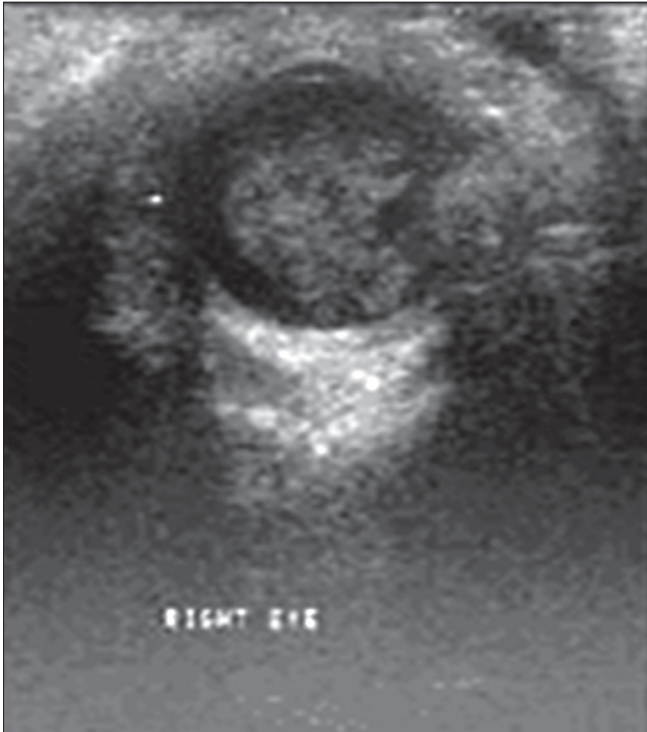


Figure 1: B-scan of the right eye of a two-years old boy reveals a hyperechoic solid intraocular posterior segment tumor

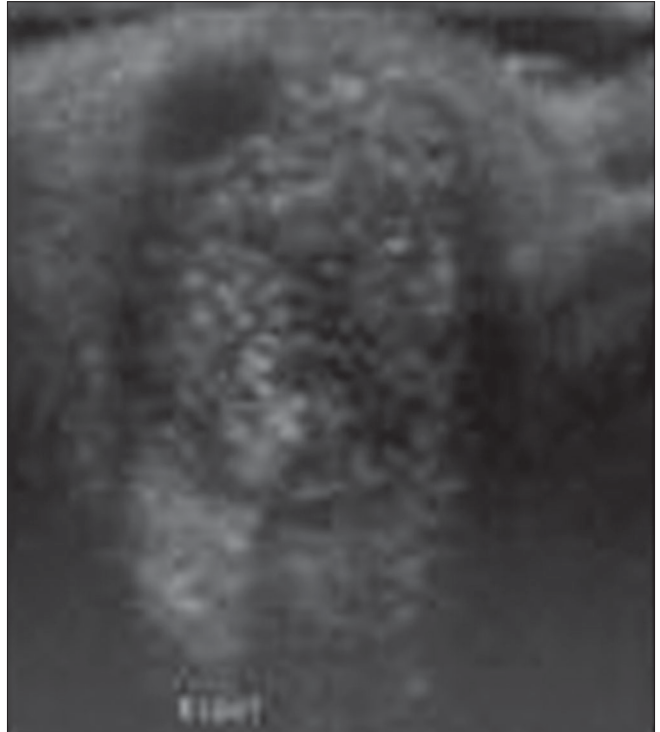


Figure 3: B-scan of the right eye of an 18-months old girl reveals a hyperechoic intraocular posterior segment tumor with an irregular surface and calcium deposits within



Figure 2: B-scan of the left eye of the same two-years old boy reveals a similar tumor in the posterior segment

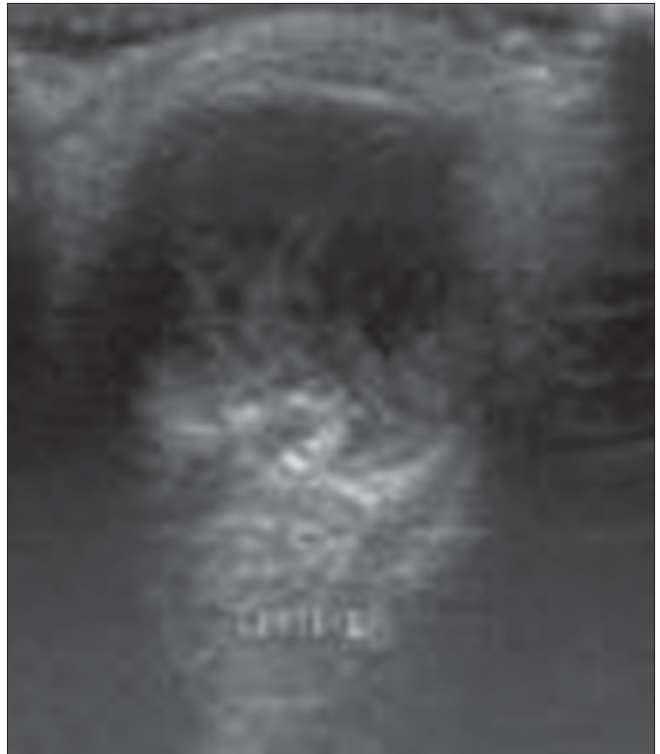


Figure 4: B-scan of the left eye of the same child reveals a similar lesion in the left eye

of 1 in 20,000 births. It arises from cells derived from the embryonic retinal epithelium of the primary optic vesicle. Retinoblastoma has the best prognosis of all childhood

tumors with an 80% survival rate. Both sexes are equally affected and the disease is commonly unilateral. It is bilateral in about a third of the cases.



Figure 5: Cut specimen of the left eye in an 18-months old child reveals a whitish intraocular tumor

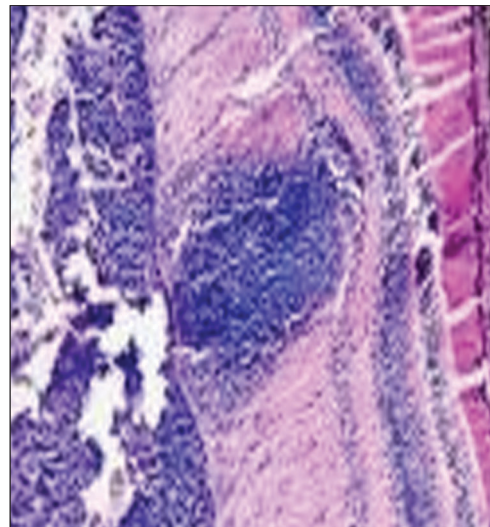


Figure 7: Histopathological study of a right eye tumor reveals small clear blue cells forming rosettes in the tumor, arising from the retina and extending into the vitreous

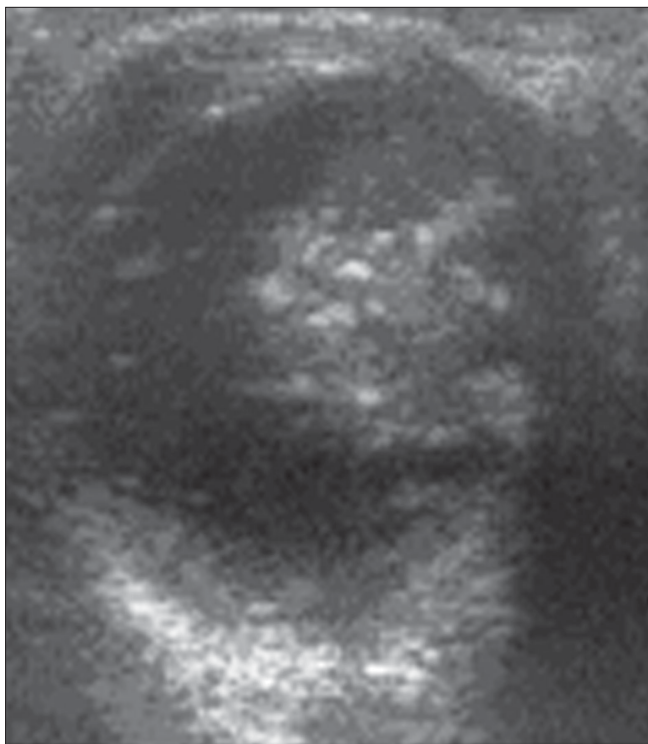


Figure 6: B-scan of the right eye of a three-years old girl reveals a hyperechoic posterior segment lesion with calcium deposits

The tumor presents before the age of three years, with the average age at diagnosis being 18 months. Six per cent of the patients have a positive family history of retinoblastoma. The mode of inheritance is autosomal dominant. The remaining 94% cases are sporadic, of which, 25% cases are due to germinal mutations which are inherited.

Retinoblastoma usually presents at a late stage with leukocoria. It presents with a white pupil (amaurotic cat's eye). Figure 8 shows an example of a child with leukocoria



Figure 8: An example of leukocoria (white pupil) in the left eye of a child known to have retinoblastoma

of the left eye. Other modes of presentation include squint, cataract, buphthalmos and proptosis. It presents in two forms viz. endophytic and exophytic. The endophytic variety of tumor projects from the retina into the vitreous cavity as a white or pinkish mass and is characterized by the presence of secondary calcification. The exophytic tumor grows into the subretinal space leading to a total retinal detachment. In such cases, USG reveals the hidden tumor.

On B-scan, the tumor is usually localized to one area of the globe, though it may be more extensive. The tumor is hyperechoic, even at low gain settings. Calcium deposits may be present, seen as highly reflective foci, sometimes associated with acoustic shadowing. The tumor outline is irregular and the calcium deposits are pathognomonic of

retinoblastoma. The lesion may be cystic due to blood clots. When small, tumors are smooth and dome-shaped and show low to medium reflectivity. As the tumor grows, it becomes irregular and more calcium accumulates, leading to a highly reflective mass.

B-scan helps to detect extrascleral spread as well as to detect optic nerve invasion. B-scan displays lesion with or without retinal detachment or vitreous seedlings. However, CT or MRI is more useful for evaluating extraocular spread and optic nerve involvement as well as for the demonstration of central nervous system spread or pineal gland involvement. Calcification occurs in 75% of the cases. The lesions may be bilateral. Sometimes, the pineal gland shows a tumor – the so-called trilateral retinoblastoma. If in addition, the suprasellar region shows a tumor, it is called a quadrilateral retinoblastoma. Optic nerve involvement beyond the point of surgical transection is associated with a 65% mortality rate, but if uninvolved, the mortality rate is just 8%. A CT scan is the most sensitive modality for detecting calcium.

B-scan is a useful, effective way for following the tumor as treatment is delivered. If left untreated, retinoblastoma runs through the same stages as choroidal melanoma viz. 1) quiescent stage – six months to a year, 2) glaucomatous stage, 3) the stage of extraocular extension, and 4) metastases.

Metastases occur in pre-auricular lymph nodes and neighboring nodes and later, to cranial and other bones. Liver metastases are rare. Baseline tumor measurements and tumor location are obtained and these parameters are monitored closely during and after treatment.

The differential diagnosis of leukocoria includes 1) Coat's disease 2) Retrolental fibroplasia 3) Toxocariasis, and 4) Persistent hyperplastic primary vitreous. All these conditions can be differentiated on a B-scan. Retrolental fibroplasia presents with a retrolental mass, due to neovascularization from the retinal periphery with resultant fibrotic changes. Coat's disease is a severe form of retinal telangiectasia with

large intra- and subretinal exudates, retinal detachment and a subretinal mass with low reflectivity in the anterior vitreous. Persistent hyperplastic primary vitreous (PHPV) is due to failure of regression of the primary vitreous and shows a retrolental mass in the anterior type, and a dense membrane containing the hyaloid artery, extending from the retrolental area to the optic disc, in the posterior type.

Small tumors are treated with light coagulation or suturing a cobalt disc to the sclera over the site of the neoplasm. Small tumors may also be treated with local cryotherapy (anterior lesions) and photocoagulation (posterior lesions). For larger tumors, tumors which are fungating and cases in which there is vision loss, treatment includes excision of the eye, the optic nerve being cut long (at least 1 cm). Prognosis if untreated is always fatal. In bilateral tumors, an option of combined therapy in the form of chemotherapy and external beam irradiation may be considered. When the tumor shrinks in size, then local forms of treatment may be instituted as stated above. If a child presents with enucleation of one eye for retinoblastoma and a retinoblastoma in the other remaining eye, the enucleation of the remaining eye is preferred. For larger unilateral tumors, combined therapy in the form of chemotherapy, external beam irradiation and brachytherapy may be considered. Prognosis is fair, if extraocular extension can be avoided. Owing to its familial tendency, the eyes of subsequent siblings or descendants should be carefully watched during childhood.

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