

## Bilateral retinoblastoma in early infancy: A case report

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### Abstract

Retinoblastoma is the most common primary intraocular tumour of childhood<sup>1</sup>. Though congenital, it is not diagnosed at birth, and is usually seen between 1-2 years of age.<sup>2</sup> We here report a case of bilateral retinoblastoma with early presentation at the age of one month.

**Key words:** Bilateral Retinoblastoma, leukocoria, amaurotic cat's eye, Flexner-Wintersteiner rosettes

Retinoblastoma arises from the neurosensory retina in one or both eyes. It is a rare disease, affecting approximately 1 in 15,000 live births. Males have a slightly higher incidence than females (1.7 to 1). It results either from sporadic or heritable mutations in the retinoblastoma gene, RB1 on long arm of chromosome 13. Heritable forms greatly elevates risk of developing a second malignancy, commonly osteogenic sarcomas, lung and breast cancer.<sup>3</sup> In 25-30 percent cases there is bilateral involvement, although one eye is affected more extensively and earlier than the other.<sup>4</sup>

The association of bilateral retinoblastoma with intracranial tumours, termed "trilateral" retinoblastoma, is a well recognized but uncommon syndrome. The intracranial tumour arises most often in the pineal region but can also be a suprasellar or parasellar tumor.<sup>5</sup>

### Case report

A six months old female child was referred to our hospital for white reflex in both eyes for 5 months. The mother first noticed white reflex on the right eye when the child was one month old and since then it has been progressively increasing to have involved left eye within the span of 3 to 4 months. There was no history of trauma, fever, eye infections and lymphadenopathy. Review of systems was negative. Family history for retinoblastoma was negative. It was uneventful full term normal vaginal delivery. Developmental milestones are adequate for the age.

General examination is within normal limits. On Ophthalmic examination, slight proptosis is noted in both eyes with eyeball and cornea appearing relatively enlarged. Yellowish white opacity was seen on both the eyes behind the cornea [Fig 1].

Plain CT scan of the head showed no abnormality. Sella, parasellar and pineal region were normal. On CT scan of the orbits [Fig 2], calcified mass of the vitreous along the retina on both sides without any extraglobe extension was seen. Patient's parents refused to have further aggressive treatment.



**Fig1:** White reflex on both eyes



**Fig 2:** Calcified bilateral vitreous on CT scan of orbits

## Discussion

Retinoblastoma arises as malignant proliferation of the immature retinal neural cells called retinoblasts i.e., derived from photoreceptor precursor cells which have lost both antioncogenic genes either by deletion or point mutation.<sup>6</sup>

Clinically, leukocoria or yellowish white pupillary reflex (also called amaurotic cat's eye appearance) is the most common presenting sign. Parents may notice this with direct observation or on photographs. Other presenting signs are strabismus, abnormal pupils, nystagmus etc. If left untreated, it may progress to glaucomatous stage characterized by severe pain, redness and watering. Further, it may have extension into extraocular tissues, optic nerve or brain by direct invasion, or may metastasize by lymphatic spread or blood stream.

Grossly retinoblastoma appears chalky white, friable well circumscribed polypoid tumour with dense foci of calcification giving the typical 'cottage cheese' appearance. Tumours may grow from the retina into the sub-retinal space (exophytic) or may grow into the vitreous cavity (endophytic). Histopathologically, it consists of small round cells with large nuclei, resembling the cells of the nuclear layer of retina. Well differentiated tumours include Flexner Wintersteiner rosettes (highly specific of retinoblastoma), Homer-Wright rosettes, pseudorosettes, fleurettes formation and with areas of necrosis and calcification.<sup>1,4,7,8</sup>

The diagnosis is easily reached with the help of careful ophthalmologic examination, plain x-ray of the orbit and CT scan. The diagnosis of Retinoblastoma can be devastating. It means that the patient has a highly malignant childhood cancer and that other children in the family or future children are also at risk for the same cancer. Without treatment cancer will spread through the optic nerve to the brain and cause death within the first four years of life. Treatment may mean blindness or the loss of one or both eyes and patients who survive treatment of Retinoblastoma are at risk for second non-ocular cancers. Frequently in bilateral cases, the more severely affected eye was removed. More recently chemotherapy has been used to achieve reduction of local tumours so that a more conservative treatment would be possible for definitive tumour control. Currently the treatment modalities that we advocate

include enucleation, external beam irradiation, brachytherapy, photocoagulation, cryotherapy, chemotherapy, chemothermotherapy and chemoreduction.<sup>9,10</sup>

Prognosis is still in part dependent on early recognition. Although retinoblastoma is a malignant tumour, it can be successfully treated in many cases if early, accurate diagnosis and prompt referral are made.

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