Case Report

SURPRISE INCIDENTAL FINDING OF RETINOBLASTOMA DURING RETINOPATHY OF PREMATURITY SCREENING: A CASE REPORT

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ARTICLE INFO

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Received:
January 2022
Accepted for publication:
May 2022

Keywords:

retinoblastoma;
retinopathy of prematurity screening;
preterm neonate

ABSTRACT

Retinoblastoma is one of the most common childhood malignancy and 3rd most common intraocular malignancy after uveal malignant melanoma and metastatic carcinoma. Retinoblastoma is most frequent in children less than 5 years old. Incidental retinoblastoma in neonates is rare. We presented a case of retinoblastoma in preterm patient during retinopathy of prematurity (ROP) screening. A premature baby was referred for ROP screening. The patient's gestational age at birth was 30 weeks 6 days and a birthweight of 1540g. He was under supplemental oxygen in intensive care unit. Dilated fundus examination of the left eye showed a central macular lesion approximately 2mm in diameter with a prominent feeding vessel infero-temporally. Patient underwent laser photocoagulation therapy to retinoblastoma tumor foci as well as barricade laser. Follow up examinations showed the tumor increasing in size with more prominent feeding vessel infero-temporally. Currently plan for systemic chemotherapy.

INTRODUCTION

Retinoblastoma is a most common childhood malignancy that arises from the retina. As an intraocular tumor, the tumor is rare but highly malignant, which may cause significant visual morbidity and death if left untreated. Most common presentations of retinoblastoma are leukocoria and strabismus which are usually noticed by the parents. In embryonic or fetal development, these kinds of cancer develop from immature tissues due to disruptions of cell proliferation and growth [1]. Mutations of the retinoblastoma gene RB1, which is usually inherited, are one type of defect that can result in such an early disturbance and thus begin cancer developing. In this case report, we discuss the issue of retinoblastoma in preterm patients as well as the challenges in managing the disease [1].

CASE REPORT

A male neonate born preterm, at 30 weeks 6 days gestational age with birth weight of 1540g was admitted to the neonatal ICU. Patient was under supplemental oxygen administration. The patient was referred for ROP screening. On examination, the anterior segments were normal. left fundus examination revealed a central elevated macular lesion of approximately 2 mm in diameter with an inferotemporal feeding vessel (Figure 2). The right fundus finding was unremarkable (Figure 1). Magnetic Resonance Imaging of Brain and Orbit, revealed a subretinal mass in the posterior pole involving the macula in the left eye. Scleral involvement was evident in T1 signals.

The provisional diagnosis was retinoblastoma. There is no family history of retinoblastoma. The family members were counselled regarding this condition and treatment options with laser photo ablation were discussed in depth with them. A laser photoablation was performed. After 2 weeks, examination showed regression of tumor size (Figure 3). Repeat laser therapy was given. However follow up examinations showed progression of the tumor with more prominent feeding vessel inferotemporally. Patient was planned for chemotherapy.

DISCUSSION

Retinoblastoma remains the most common childhood malignancy that arises in the retina and represents1%-3% of all childhood malignancies and 4%-29% of solid neonatal carcinoma, excluding hematological neoplasms and nonmalignant teratomas [1]. In spite of its rarity, it is an extremely

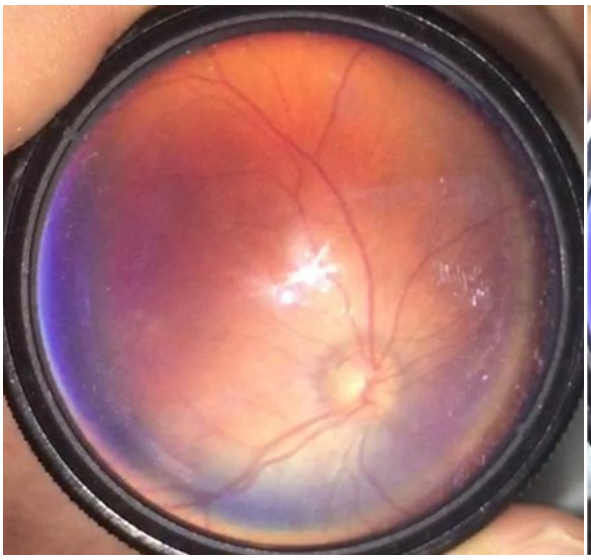

Figure 1: Right normal fundus

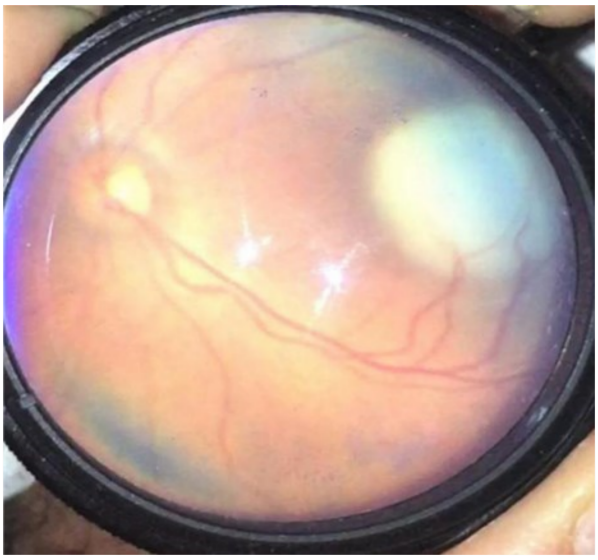

Figure 2: Left fundus: Central macular lesion with feeder vessels

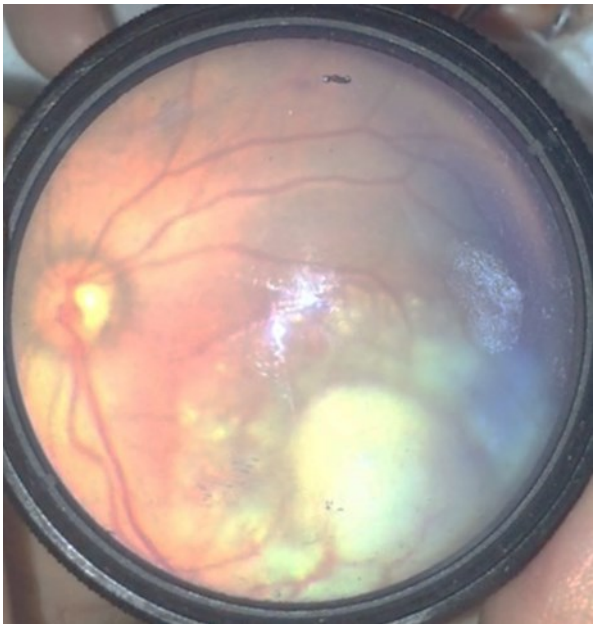

Figure 3: Left fundus: post laser ablation with less feeder vessels

malignant intraocular tumor that causes significant visual disability and even death if left undetected. Two-thirds are diagnosed before 2 years of age, overall 95% of cases occur before the age of 5 years. There are between 7000 and 8000 new cases of retinoblastoma per year worldwide with an incidence of 1 in 16,000-18,000 births per year. The incidence is 3.5 per million children under the age of 15 and 11.8 per million children under the age of 5 on an annual basis. There are an estimated 3000 to 4000 deaths annually due to retinoblastoma. The possibility in preterm and small for date neonates is more than 1 in 16,600 live birth [2].

The age of presentation of our case is 30 weeks 6 days gestational age, which is way below the average age. However, no genetic study was done in this case to further confirm the genetic correlation.

Retinoblastoma can be either heritable and associated with a germline mutation of the RB1 gene, or non-heritable. The clinical presentation varies according to the stage of the disease and the presence of one or several tumors in unilateral or bilateral eyes, with symmetrical or asymmetrical lesions. The tumor can present as an endophytic form, in which the tumor extends into the vitreous and exophytic form, associated with retinal detachment and subretinal infiltration or a mixed form, comprising vitreous involvement and retinal detachment [3].

Problems faced in treatment of retinoblastoma depend on presentation. In order to determine the

best treatment for each child, factors such as tumor laterality, the size and location of tumors, metastasis risk, subretinal and vitreous seeds, as well as the tumor relationship to nearby structures are considered. There are several criteria that determine whether small tumors are amenable to local ablative treatment. These criteria include tumour position greater than 3 mm from the fovea, greater than 1.5 mm from the optic disc, and smaller than 3 mm in height and diameter. Laser photocoagulation or cryotherapy can be used for ablation therapy, which results in 86% of tumors regressing [4].

The laser treatment may be repeated every 3 - 4 weeks, until signs of inactivity and complete tumor regression are evident. A clinically inactive tumor can be identified by indirect fundoscopy showing calcified, inactive tumor, as well as abnormalities such as subretinal fluid, subretinal seeds, or vitreous seeds. Intravenous chemotherapy is used for advanced tumors that can't be ablated locally (chemo reduction) [5]. Study by Chen M in Beijing, China suggested that primary Intra-arterial chemotherapy (IAC) treatment of retinoblastoma in infants less than 3 months old may be a feasible and promising treatment option [6].

CONCLUSION

Neonatal retinoblastoma is rare and presents a management challenge, in particular when there are added factors such as prematurity and small for gestational age. In light of the success of present treatment protocols, there is an increasing interest in examining local administration of chemotherapy, such as intra-arterial and subconjunctival chemotherapy, in order to improve tumor control in advanced tumors and minimize toxic systemic chemotherapy. Preliminary results have shown that super selective ophthalmic artery chemotherapy can successfully treat retinoblastoma tumors, including those requiring surgical removal, in a high level. A combination of therapies targeting hypoxia, angiogenesis, and cellular metabolism is effective in tumor control and should be evaluated and tested further in retinoblastoma.

CONFLICTS OF INTEREST

No conflicts of interest

ACKNOWLEDGEMENT

We would like to thank the Director General of Health Malaysia for his permission to publish this article.

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