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Non-Familial Retinoblastoma: Case Report

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Abstract

Retinoblastoma is a malignancy of the retina. It is most common intraocular tumor in children. Dominantly inherited disorder frequently found association with defect in RB1 gene. Approximately 40% of patients with retinoblastoma have inherited a germ-line mutation of the RB1 gene found 10% unilaterally while 60% are non-familial. Herewith we present 2 cases of non-familial, unilateral retinoblastoma.

Keywords: Non-Familial, Retinoblastoma, Sporadic

1. Introduction

Retinoblastoma is an embryonal malignancy of the retina and is most common intraocular tumor in children[1]. Retinoblastoma is an autosomal dominant inherited disorder with defect in RB1 gene located on chromosome 13q14. Approximately 40% of patients with retinoblastoma have inherited a germ-line mutation of the *RB1* gene (gene map locus 13q14.1-q14.2) (OMIM 180200) 85% are bilateral, 10% are unilateral[2] while 60% cases are non-familial (unilateral). Pathologically, it is a small, round blue cell tumor with rosette formation with necrosis and calcification. Bilateral involvement is more common in children under the age of 1year[1]. Racial or gender predilection is not known. We present herewith 2 cases of non-familial, unilateral retinoblastoma with varied clinical presentations.

2. Case 1

7 years old male child presented with gradually progressive swelling in right eye since last 1.5 year leading to proptosis with gradually decreasing vision. 4 months ago patient had redness and tenderness in the same eye and was diagnosed as panophthalmitis and Enucleation of right eye was done at his village but histopathology report was not available. On clinical examination, proptosis was present. Visual acuity of right eye had no light perception and visual acuity of left eye is central, steady, and maintained fixation. Patient was awake but irritable. Red reflex was absent. Ophthalmic examination s/o proptosis with glaucomatous change. Orbital inflammation was present. We did MRI Brain, which revealed a residual / recurrent retinoblastoma with intracranial extension (Figure 1). Patient was referred for radiotherapy; however, he went against medical advice.

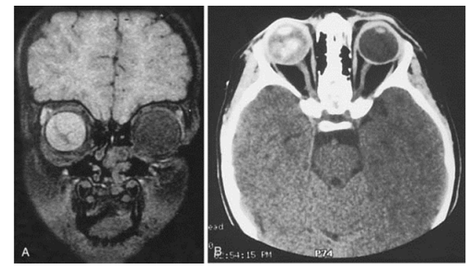

Figure 1. MRI Brain plain with intraocular growth.

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3. Case 2

4.5 years old male child came to us with fever, lethargy, generalized tonic-clonic convulsions and proptosis of left eye which gradually increased in size over the period of 6 months. On clinical examination, patient was unconscious and GCS was 3/15. Deep tendon reflexes were exaggerated. Ophthalmic examination revealed proptosis with prolapse of left iris, signs of raised ICT (Figure 2). Antibiotics and anti-cerebral edema measures were instituted, however, patient progressed and succumbed before stereotactic biopsy and radiotherapy could be given. Postmortem biopsy revealed Retinoblastoma of left eye.

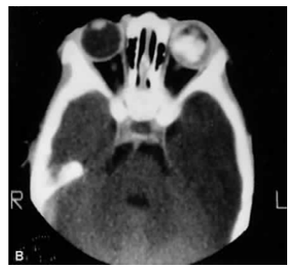

Figure 2. MRI Brain plain with proptosis and intraocular extension of the tumor.

4. Discussion

Retinoblastoma is a primary intraocular malignant tumor caused by mutations of the *RB1* gene. The *RB1* gene product, p-RB, is a cell cycle regulator with tumor suppressing activities[5]. Knudson et al[6] hypothesized that 2 events of mutations are required for the development of hereditary retino-blastoma: first, a germ-line mutation; second, a separate somatic mutation occurring in retinal cells. Patients with the disease can pass on the initial germline mutation as a dominant trait[3]. Twenty-five percent of patients with the inheritable form have a family history of the disease. Others can develop either a new germ-line *RB1* mutation or inherit the mutation from an asymptomatic carrier parent[7].

Both cases described in this report developed unilateral unifocal retinoblastoma despite no family history of retinoblastoma. Approximately 12% of sporadic cases of unilateral retinoblastoma involve a germline mutation[3]. This suggests that they are more likely due to inheritance from an asymptomatic carrier parent[8].

Leukocoria is the most common clinical presentation and strabismus, ocular inflammation, proptosis, hyphema, mydriasis and glaucoma are other manifestations.

Diagnosis depends mainly on thorough ophthalmologic examination supported by neuroimaging. Visual acuity, alignment, and general ocular health should be should be examined periodically. Various modes of therapy include enucleation, radiotherapy, cryotherapy / photocoagulation and chemotherapy. Screening with DNA analysis of parents and siblings is advised. The patient and parents should be questioned about and warned about signs of secondary nonocular tumors during these examinations.

The prognosis in retinoblastoma is good if they are detected and treated early. The cure rate is almost 90% if the optic nerve is not involved and enucleation is performed before the tumor passes through the lamina cribrosa. Survival rates decrease to 60% if the tumor extends beyond the lamina cribrosa even if the cut end of the nerve is free of tumor cells.

5. Conclusion

Proptosis is a more common presenting symptom in most underdeveloped countries. High index of suspicion is required in, children with proptosis and leukocoria as early diagnosis, institution of therapy will markedly improve the outcome. The possibility of mosaicism is important to consider while counseling parents of a child with retinoblastoma. Parents are usually reassured after diagnosis of a first child with retinoblastoma that the chance of having another child with the same tumor is extremely low.

6. References

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