Retinoblastoma and Retinal Astrocytoma: Unusual Double Tumor in One Eye

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Abstract- Retinoblastoma is the most common intraocular neoplasm in children. Glial tumor of the retina and optic nerve head are considered to be congenital and are therefore classified as hamartomas. Concurrent occurrence of these tumors in one eye is uncommon and by reviewing the studies, a few cases have been reported. We report a 9 years old boy with eye enucleation and concurrent occurrence of retinoblastoma and astrocytoma in one eye as two separate and different masses. Although retinoblastoma and astrocytoma are two distinct tumors and their concurrent occurrence in one eye is rare, concurrent occurrence of these tumors may suggest differentiation of these two tumors from a neuroectodermal primary cell.

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Introduction

Retinoblastoma is the most common intraocular neoplasm in children. It is generally believed to be congenital and derived from primitive neuroectodermal cells exhibiting retinal differentiation.

Retinoblastoma is seldom recognized until considerable growth has taken place and is usually diagnosed between the ages of 16 months and 2 years.

Approximately 60% of the cases are sporadic, and the other 40% are familial, whereas the predisposition to tumor development is transmitted in an autosomal dominant pattern. Retinoblastoma will develop in 80% to 90% of persons who carry any one of a variety of mutant alleles associated with a predisposition to the tumor. The gene responsible for familial retinoblastoma is located in chromosome 13q14 and designated the retinoblastoma (Rb) gene.

Gene mutations in both alleles are necessary to produce inactivation of the Rb protein, which is a negative regulator of cell growth. Patients with hereditary retinoblastoma have a germ cell mutation in one allele and develop retinoblastomas as a result of a somatic mutation in the second allele, whereas in patients with sporadic retinoblastoma both mutations are somatic.

Retinoblastoma characteristically presents as a leukocoria (white papillary reflex) or less often as a strabismus when the tumor is in the macula. In rare cases, extraocular extension with the formation of an orbital mass is the presenting manifestation. Bilaterally is present in 30% of all cases and in over 90% of familial cases (1).

Glial tumor of the retina and optic nerve head are considered to be congenital and are therefore classified as hamartomas. Rarely new tumors have been observed to develop in clinically normal retina.

Optic nerve astrocytoma is a rare tumor which occur under 15 years old patients. Clinical features of this tumor are proptosis, loss of vision, nystagmus headache and vomiting (2).

The majority of astrocytoma of retina is associated with either tuberous sclerosis or less often, neurofibromatosis.

Sporadic cases account for less than 30% of cases. Long standing lesion, often become extensively calcified and thus may radiologically mimic retinoblastoma (3).

Case Reports

The patient is a 9 years old boy suffering from bilateral strabismus from birth. At the age of 1 year and 3 months, right eye was enucleated following the diagnosis of retinoblastoma. At the age of 7 he experienced decreased eyesight and blurred vision in his left eye for which he underwent 5 chemotherapy and 2 radiotherapy sessions. In examination of the left eye, visual acuity was reduced to the extent of not recognizing the light; blurred cornea; lens with

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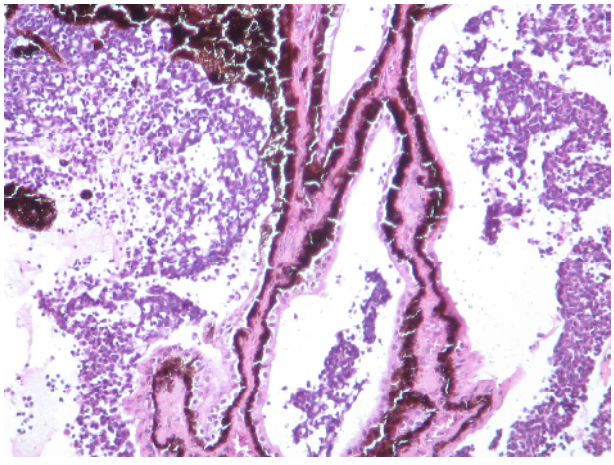

Figure 1. Intraocular tumor, retinoblastoma. H&E staining (X 100).

cataract; anterior chamber contained tumor cell; weak red reflex and involvement of the vitreous with tumor. The patient finally underwent enucleation of the left eye with the diagnosis of retinoblastoma.

There was a family history of enucleation in his father and sister with the diagnosis of retinoblastoma.

The sample was sent to pathology department and contained an eyeball with maximum diameter of 3 cm and white cornea. In resection, two tumors were evident. The first mass was a soft anteriorly located mass that was grey in colour and the second mass that was cream in colour with the diameter of 1 cm located in the posterior portion.

In microscopic examination, anterior mass consisted of tumor cells originated from retina with average differentiation including cells with round and hyperchromatic nuclei and small cytoplasm associated with necrosis, which invaded ciliary bodies and choroids.

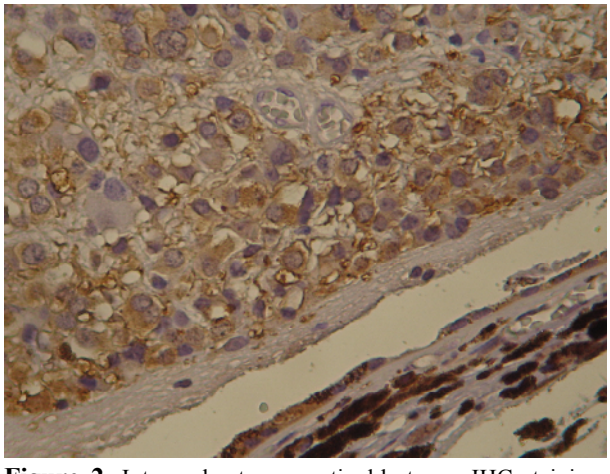

Figure 2. Intraocular tumor, retinoblastoma. IHC staining shows positive reaction for synaptophysin (X 400)

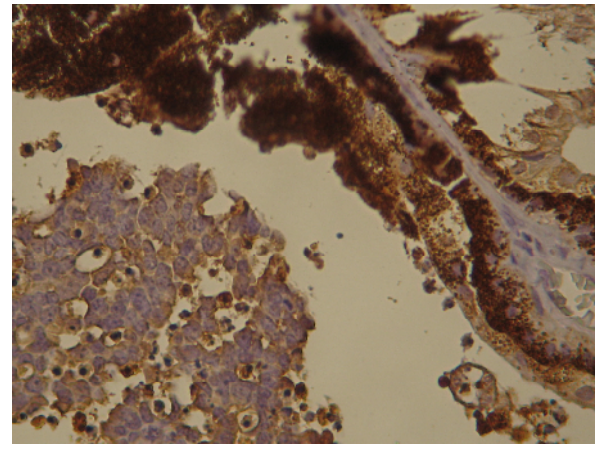

Figure 3. Intraocular tumor, retinoblastoma. IHC staining shows positive reaction for NSE (X 100).

In immunohistochemical (IHC) examination, these cells were positive for neuron specific enolase (NSE) and synaptophysin, and negative for glial fibrillary acidic protein (GAFP) (Figure 1-3).

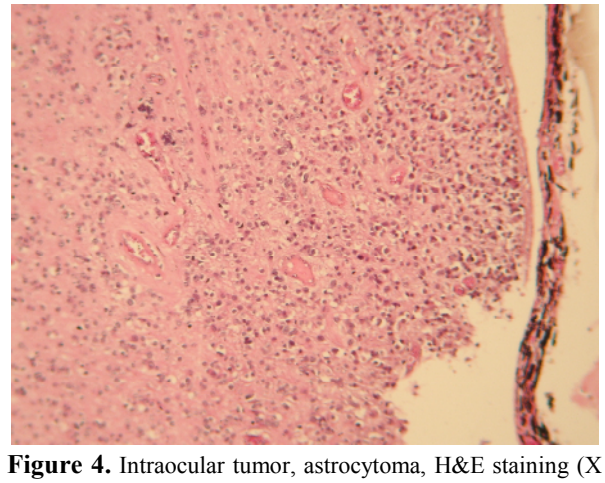

Figure 4. Intraocular tumor, astrocytoma, H&E staining (X 100)

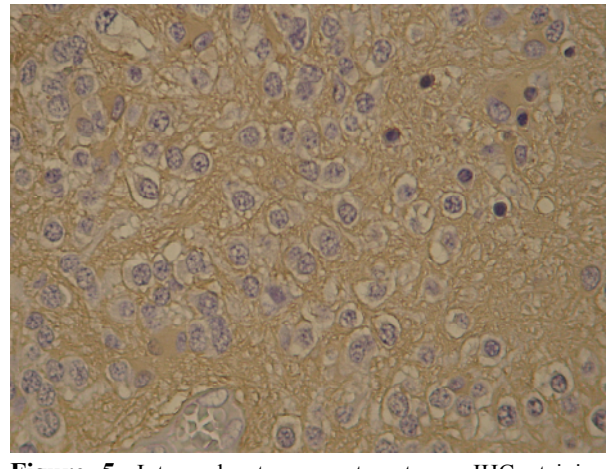

Figure 5. Intraocular tumor, astrocytoma. IHC staining shows positive reaction for GFAP (X 400).

The second mass, located in the posterior portion, consisted of cells of glial origin arising from retina together with abundant vessels indicating for GFAP marker and negative for NSE and synaptophysin (Figures 4 and 5).

In resections obtained from optic nerve, invasion of tumor cells was evident.

Discussion

As mentioned earlier, retinoblastoma is the most prevalent intraocular neoplasm in children diagnosed between age of 16 months and 2 years. It usually manifests as strabismus. The disease occurs bilaterally in 30% of cases and is inherited in 90%.

Retinal Astrocytoma is a rarely occurring tumor diagnosed in children especially girls, at the age of 5. This tumor appears with proptosis symptoms.

Concurrent occurrence of these two tumors in one eye is uncommon and by reviewing the studies, only one case has been reported by Imhof *et al.* in 2002 (9). In Farabi Hospital which is one the main ophthalmology referral units in Iran, one case of concurrent occurrence of retinoblastoma and retinal astrocytoma is reported. Both morphological and histological studies revealed that these two tumors are completely different.

As mentioned before, retinoblastoma tumor was negative for GFAP and positive for synapthophysin and NSE. However, the astrocytoma glial tumor was positive for GFAP and negative for NSE and synapthophysin.

Our study does not necessarily prove that these tumors are completely different. There is a possibility that these tumors may be originated from a common primary cell. Various studies indicate that retinoblastoma is a tumor with neural origin that, of course, can differentiate into glial route (4-8). In conclusion, although retinoblastoma and astrocytoma are two distinct tumors and their concurrent occurrence in one eye is rare, concurrent occurrence of these tumors may suggest differentiation of these two tumors from a neuroectodermal primary cell.

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