Case Report

Pigmented malignant teratoid medulloepithelioma of ciliary body – An extremely rare intraocular tumor in adult

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Abstract

Malignant teratoid medulloepithelioma of ciliary body is an extremely rare tumor usually occurring in children younger than 5 years of age and is usually amelanotic. Here, we report a very rare case of pigmented malignant teratoid medulloepithelioma in a 20-year old male patient who presented initially with pain, redness and proptosis of right eyeball. The right eyeball was enucleated. A histopathological diagnosis of orbital teratoma was made and no further treatment was given. Four months later, the patient developed another swelling in the same area. A review of the previous histopathology slides revealed a pigmented malignant teratoid medulloepithelioma. No nerve involvement was seen. MRI did not show any intracranial extension. The mass along with rest of the orbital tissue was removed. No adjuvant chemotherapy or radiotherapy was given due to the lack of intracranial extension. The patient was symptom free at 2-year follow-up and the repeat MRI also showed no evidence of recurrence.

Keywords: Malignant teratoid medulloepithelioma, Ciliary body, Pigmented

Introduction

Malignant teratoid medulloepithelioma in the eye is an extremely rare embryonal tumor of neuroepithelial origin.1 The majority of these tumors originate in the ciliary body epithelium1-3 and rarely from iris, retina and the optic nerve.1,2 Previously known by other names like “teratoneuroma” and “diktyoma”, the present term “medulloepithelioma” was coined by Grinker in 1931. Usually presenting as unilateral tumors, they do not have any predilection for particular, sex, race or laterality. These are regarded as congenital tumors with a median age of diagnosis being 3.8 years (range being 6 months to 41 years).2 Thus, extremely uncommon in adults, only around 11 cases of adult malignant teratoid medulloepithelioma have been reported.1 Histologically, the tumor is characterized by papillary, tubular or trabecular arrangements of neoplastic neuroepithelium which resemble the embryonic neural tube. Medulloepithelioma of the central nervous system is a highly malignant neoplasm while the ocular counterpart usually has a benign course.4 Medulloepitheliomas have been classified as non-teratoid and teratoid. Each variant can be either benign or malignant. Nonteratoid medulloepitheliomas account for 50–70% of all cases while the teratoid ones comprise of 30–50% of all cases.4 Most of these tumors are amelanotic arising from the non-pigmented part of the ciliary epithelium while only three cases till date have been reported as pigmented malignant medulloepithelioma of the ciliary body.5 Here, we report an extremely rare case of pigmented malignant teratoid medulloepithelioma in an adult patient.

Case report

A 20-year old male patient presented in an out-patient clinic with features of progressive proptosis of the right eye.
with loss of vision and pain for duration of two months. On examination, the right eye was protruded with no vision. The other eye was completely normal. A clinical diagnosis of retinoblastoma was made. The complete blood counts and the chest X-rays were unremarkable. The patient underwent enucleation of the right eye in a private hospital and the specimen was sent for histopathological examination in a private laboratory. A diagnosis of orbital teratoma was made. The optic nerve was free of tumor cells. No adjuvant treatment was prescribed to the patient. Four months later, the patient presented in the out-patient department of our hospital with complaints of a mass in the right orbital region. The previous histopathology slides were sent to the Department of Pathology for review. At the subsequent review the sections showed pseudo-stratified primitive looking epithelial cells arranged in papillary configuration, cords, nests, tubules and in diffuse sheets at places simulating the medullary epithelium arising from the ciliary body. (Fig. 1) The cells showed poor differentiation at places with individual cells showing significant pleomorphism, mitosis (approximately 8–9/hpf) and hyperchromasia. There was the presence of mature as well as immature cartilage. Some areas showed cystically diluted spaces containing an eosinophilic material. The most striking feature was the presence of pigment laden cells forming gland like structures (Fig. 2). The retina was not involved and the optic nerve was also not involved as was reported previously. A revised diagnosis of pigmented malignant medulloepithelioma of the ciliary body of the right eye was made. Subsequent MRI of the orbits showed evidence of a distorted mass (right orbital region) with mixed intense signals in T1 and T2 weighted images and mixed enhancement on contrast. The CT scan and MRI of brain revealed no co-existing CNS tumor or other anomaly and there was no evidence of any intracranial spread. The mass was removed and the histopathological features were found to be similar to the previous tumor being composed of primitive looking epithelial cells arranged in papillae, cords, nests, tubules with cystic areas, mature cartilage and pigment laden macrophages (Fig. 3). The resected margins were free of tumor. No chemotherapy or salvage radiotherapy was administered to the patient. The patient was free from relapse or any symptom at 2-year clinical and radiological follow-up.

**Discussion**

Medulloepitheliomas are extremely rare embryonal intraocular neoplasm occurring in young children with a median age of diagnosis being 3.8 years (range being 6 months to 41 years). They are still rarer in adult patients with only around 11 reported cases with the median age of diagnosis being 44 years (range 23–79 years). The majority of these tumors originate in the ciliary body epithelium and rarely from iris, retina and the optic nerve. In 1904, Verhoeff gave the first detailed historical description and he called the tumor “teratoneuroma”. The term “diktyoma” was coined by Fuchs in 1908. It was in 1931 that Grinker finally renamed the tumor as “medulloepithelioma”. Usually presenting as unilateral tumors, they do not have any predilection for particular, sex, race or laterality. Medulloepitheliomas usually present with poor vision and pain. They may also present with mass in iris, ciliary body, leucoria and proptosis. The present case initially presented with proptosis, blindness and pain in
the right eye. A clinical diagnosis of retinoblastoma was made which is a relatively common intraocular tumor. Medulloepitheliomas consist of multilayered sheets and cords of poorly differentiated neuroepithelial cells which may fold back upon themselves to form various structures which resemble the embryonic sensory retina or non pigmented ciliary body. Some folds surround fluid collections (composed of hyaluronic acid like vitreous humor) giving a net like appearance. Homer-Wright and Flexner-Wintersteiner rosettes may be seen. These tumors have been classified by the World Health Organization (WHO) as teratoid and non-teratoid, malignant and non-malignant. The teratoid variant may show heterotopic tissues in the form of hyaline cartilage, rhabdomyoblast, undifferentiated mesenchymal cells, neuroglial tissue etc. As described by Broughton and Zimmerman in 1978, the tumor is classified malignant if there are areas of poorly differentiated neuroblastic cells resembling neuroblastoma, increased nuclear pleomorphism and mitosis, atypical mitotic activity, sarcomatous areas which may resemble rhabdomyosarcoma, spindle cell sarcoma or even chondrosarcoma and invasion of uvea, corneas or sclera with or without extra-ocular extension. Most of the medulloepitheliomas are amelanotic with only three cases of pigmented malignant medulloepithelioma being described in the literature. In the present case, the undifferentiated cells showed significant pleomorphism, high mitotic rate (7–8/hpf), both mature and immature cartilage, focal retinoblastoma like areas, cystic spaces and a fair number of pigmented cells. Areas of hemorrhage and necrosis were noted. No calcification and no optic nerve involvement were noted. The tumor was limited to the orbit. Treatment of malignant teratoid medulloepithelioma is surgical enucleation which was done in our case. The case was initially misdiagnosed as orbital teratoma and it was in a subsequent pathological review that the present diagnosis was made. The tumor being locally aggressive recurred in four months following the enucleation. The tumor was surgically removed with surrounding tissue and due to the lack of any extra-ocular involvement no further treatment was given. The patient was clinically and radiologically free of the tumor at the 2-year follow-up. The prognosis of ciliary body medulloepitheliomas is generally better than their counterparts in retina, optic nerve or brain. The treatment of choice is enucleation, as already mentioned but the recurrence rate is high. The roles of chemotherapy and radiotherapy are unknown. However, aggressive tumors with extraocular extension and even distant metastasis to the lymph nodes, parotid glands, lungs and mediastinum have also been described. The recurrence in the absence of documented extra-ocular extension in this case may be related to the surgical maneuver of enucleation or it may occur due to extra-scleral extension into the orbit which was not evident in the sections provided.

Medulloepitheliomas are to be differentiated from the more common retinoblastoma by the cystic changes and possible calcification in CT scan or ultrasound, usually a more anterior development and a characteristic histopathology with frequent occurrence of heteroplastic elements in the former. Neuroblastoma also enters in differential diagnosis but has to be differentiated histopathologically by bipolar-like cell elements, hemorrhage, calcification and Homer-Wright’s rosettes. Malignant melanoma is usually differentiated with anaplastic pigmented cells with nuclear grooves, folds, prominent nucleoli and pseudo-inclusions. The awareness regarding this rare tumor is important for ophthalmologists as medulloepitheliomas are often mistaken as glaucoma or uveitis. Our case is unique being that of a malignant pigmented medulloepithelioma of ciliary body in an adult patient who presented with an unexplained local recurrence in the absence of documented histopathologic extra-ocular extension.

Thus, this case emphasizes that the recognition of this entity is important in any intraocular tumor irrespective of age group to plan a proper line of diagnostic modalities, treatment options and patient follow-up.

**Conflicting Interest**

The authors declared that there is no conflict of interest.

**References**