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SHORT COMMUNICATION

Intraocular astrocytoma without phacomatosis

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PURPOSE. *Astrocytic tumors occur in the retina or in the optic disc usually as a part of tuberous sclerosis complex or other phacomatosis and their isolated occurrence is rare. The authors present two adult patients in whom the diagnosis of intraocular astrocytoma was established but no signs of phacomatosis were revealed.(Eur J Ophthalmol 2004; 14: 350-4)*

KEY WORDS. *Intraocular astrocytoma, Isolated occurrence, No phacomatosis*

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INTRODUCTION

Astrocytic hamartoma of the retina and optic disc is a benign tumor that usually occurs as a part of tuberous sclerosis complex (TSC) or neurofibromatosis unilaterally (1-4) or bilaterally (5). Retinal lesions are found in more than 50% of patients with TSC and most commonly they consist of small, grayish, translucent tumors (5-7). Solitary astrocytoma occurs rarely (6, 8-10).

Clinically, three types of astrocytic hamartomas are recognized. The most common presentation is as a flat semitranslucent lesion of the nerve fiber layer. The second type is an opaque white nodular tumor often with calcification. In the third type, the features described above are combined; it is calcified and opaque in its central part and semitranslucent in the periphery (8). Astrocytomas are usually stable lesions without remarkable growth or major clinical symptoms. However, a flat semitranslucent type of astrocytoma may sometimes grow and change into the nodular type of tumor with calcifications (9, 11). Nodular astrocytomas tend to develop necrosis, which may lead to vitreous hemorrhages and the subsequent seeding of the tumor (12). Exceptionally, they may also cause retinal detachment (3).

This case report describes two patients who had histopathologically documented retinal astrocytomas, but no other evidence of phacomatosis.

Case reports

Case 1

Clinical history-A 52-year-old woman had a history of poor vision in her right eye during early childhood, but no tumorous lesion was suspected. In 1990, her best-corrected visual acuities were 20/20 and 20/30 in the right and left eyes, respectively. In 1991, hemophthalmus occurred in the patient's right eye after a car

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accident and a nuclear sclerosive cataract was extracted in 1992. After recurrent intraocular hemorrhages and the development of secondary glaucoma in March 1996, the patient was sent to the Department of Ophthalmology at Charles University, Prague, Czech Republic, with a diagnosis of intraocular tumor. An ultrasound examination of the eye demonstrated a mildly elevated tumor with a dome-shaped configuration on the optic disc and adjacent retina (Fig.1). Because of intraocular hemorrhage no other imaging than ultrasound was obtained. The eye was enucleated due to suspected intraocular melanoma. Before the enucleation and subsequent to the histopathologic findings in the eye, the patient underwent a complex and thorough medical examination, including a computed tomography (CT) scan of abdominal, retroperitoneal, and thoracal organs, magnetic resonance imaging and CT scans of the brain, ultrasonography of the heart, and thorough dermatologic and neurologic examinations. No signs of phacomatosis were revealed by these examinations.

Pathology-The enucleated globe was opened in a horizontal plane. An intravitreal hemorrhage was present, and in the region of the posterior pole, a yellow relatively flat tumor was discovered with a diameter of 11.5 mm and a thickness of 3.5 mm. It extended from the optic disc towards the adjacent retina, which was completely attached, with no signs of traction (Figs. 2 and 3). A light microscopic examination revealed the tumor, which consisted of slightly polymorphous glial cells with round to oval nuclei and cytoplasmic processes forming an interlacing meshwork, showing positivity for the S-100 protein and glial fibrillary acidic protein immunohistochemically. Calcospherites were observed in the tumor stroma and in the adjacent thickened retina. The surrounding retina was thickened and focally disorganized, but relatively spared. The inner neuroretinal layers were particularly very prominent. Focal hemorrhages of various ages were present within the tumor.

Case 2

Clinical history-A 45-year-old man noted a slowly progressing decrease in the vision of his right eye when he was around 20 years of age; at that time he was told that he had severe amblyopia. The patient thus did not expect any useful function in his right eye from that time on, and he did not have an eye examination for 25 years. At the age of 45, his right eye was ex-

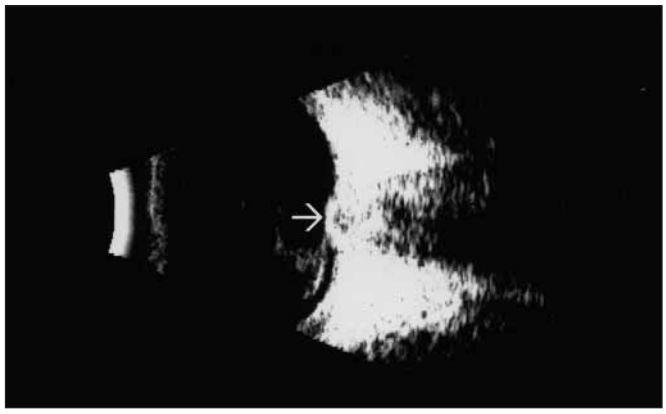

Fig. 1 - *An ultrasound picture of the right eye. A transverse B-scan echogram shows a mildly elevated tumor with a dome-shaped configuration on the optic disc.*

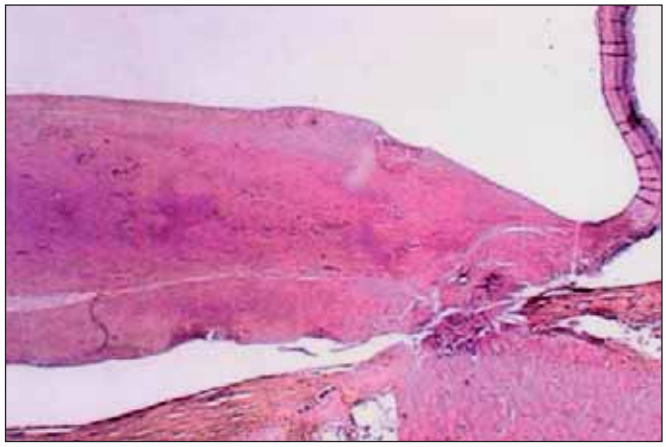

Fig. 2 - *Low power magnification of the histologic section through the tumor in the first case. A flat tumor growing from the optic disc and adjacent retina. Hematoxylin-eosin staining, original microscopic magnification 12.5 x.*

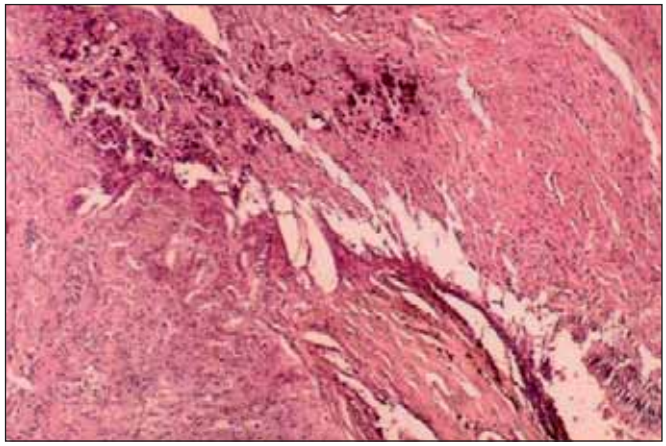

Fig. 3 - *Detail of the optic disc region from the first case with microcalcifications in the basal part of the tumor. Hematoxylin-eosin staining, original microscopic magnification 50 x.*

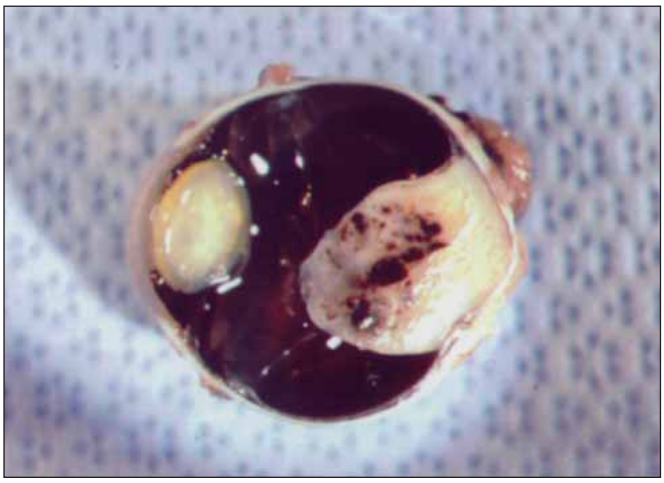

Fig. 4 - *Gross photography of the dissected enucleated eye in the second case. Bulky yellow tumor growing in the region of posterior pole.*

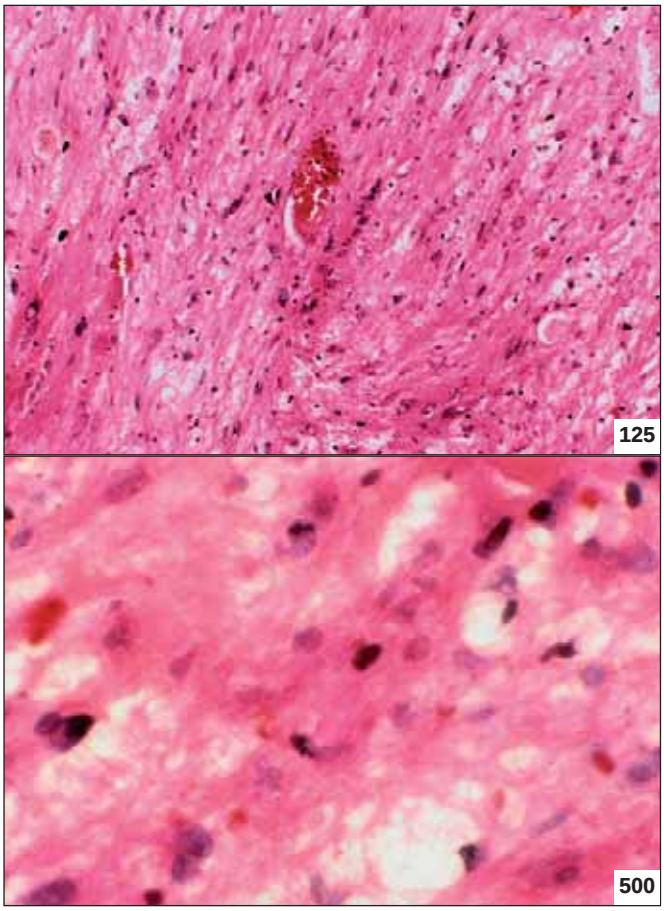

Fig. 5 - *High power magnification from the tumor in the second case. Round to oval nuclei of the tumorous cell, numerous cytoplasmic processes forming interlacing meshwork. Paraffin section, hematoxylin-eosin staining, original microscopic magnification 125 x 500.*

amined due to systemic hypertension. A large intraocular tumor was then noted, and the patient was sent to the Department of Ophthalmology at Charles University for consultation and treatment. In the left eye there were only mild changes related to systemic hypertension with no other findings, a natural vision of 20/20, and intraocular pressure was normal. Due to the presence of a large tumor in the posterior pole, and no useful vision, the eye was enucleated.

Pathology-The enucleated globe was opened in the horizontal plane. Consistent with the clinical examination, there was a nearly round, yellow tumor originating from the posterior pole, with a diameter of 12.0 mm and a prominence of 14.5 mm. It was growing from the optic disc and affected the adjacent retina (Fig. 4). The histopathology was identical to the previous case, with proliferation of slightly pleomorphic astrocytes, with round to oval nuclei and numerous processes, including the same immunohistochemical characteristics as in Case 1 (Figs. 5 and 6).

Subsequent to the histopathologic examination of the globe, the patient underwent a complete systemic work-up, and all tests were negative for any other signs of phacomatosis.

DISCUSSION

Intraocular astrocytic tumors are usually part of TSC or other phacomatosis (3, 5). Isolated retinal astro-

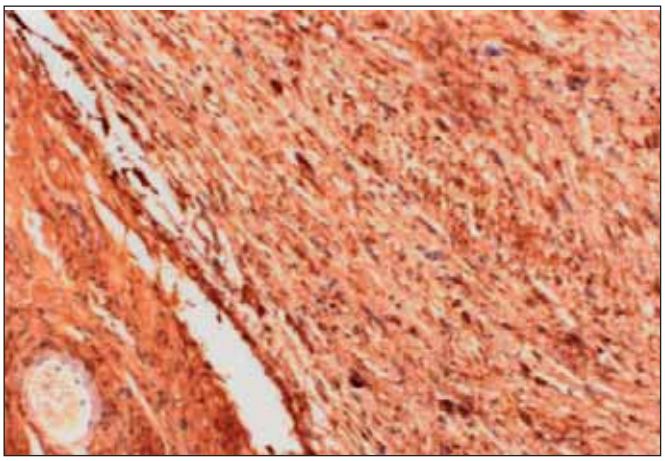

Fig. 6 - *Immunohistochemical positivity for the glial fibrillary acidic protein in the tumorous cells in the second case. Immunohistochemical peroxidase staining, original microscopic magnification 125 x.*

cytoma occurs rarely. TSC is an autosomally dominant disease known to be genetically heterogenous (3). Incompletely manifested TSC, which was thought to be an early form of disease, is nowadays more often diagnosed. The incidence of corresponding mutations is probably higher than was originally supposed. Due to large clinical variability in the expression of TSC, three types of clinical manifestations as diagnostic criteria were established: primary, secondary, and tertiary. For a definitive diagnosis, it is required that at least the presence of either one primary criterion or two secondary criteria or one secondary and two tertiary criteria are found. For a likely diagnosis, the presence of either one secondary with one tertiary criterion or at least three tertiary criteria is required. To suspect TSC, at least the presence of either one secondary or two tertiary criteria is needed. Solitary intraocular astrocytoma belongs to the group recognized as secondary criteria (13). The isolated existence of ocular astrocytoma without phacomatosis is considered rare (6, 11).

A solitary retinal astrocytoma as single, pedunculated lesions has also been reported (14). In infancy, the solitary, large masses may be confused with retinoblastoma (11, 14, 15). Other ocular lesions reported in TSC include eyelid and subconjunctival nodules, fundus depigmentation, iris depigmentation, and ocular colobomas and bilateral microphthalmos. None of these findings were present in our patients. In adults with retinal hamartomas, systemic manifestations are essential for a diagnosis of TSC; particularly the neuroimaging procedures to identify subependymal calcifications can be helpful. The absence of systemic TSC findings complicated the differential diagnostic process in our two cases. In the differential diagnosis of the first case, the massive retinal gliosis was also considered because of the patient's previous history. However, the absence of retinal detachment and absence of phthisis as well as a relatively spared retina indicated a diagnosis of astrocytic tumor. In the second case, other benign intraocular tumors such as leiomyoma were considered. The immunohistochemical characteristics, however, clearly indicated an astrocytic tumor.

As our two cases show, even these rare tumors should be considered in the differential diagnosis.

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