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Short communication

Peripapillar retinal hamartoma associated with tuberous sclerosis. Case report☆

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ABSTRACT

Introduction: Tuberous sclerosis is a rare multisystemic disease with an autosomal dominant inheritance pattern. There are few documented cases in the literature of retinal hamartomas (astrocytomas) with aggressive progression in the context of this disease.
Case report: A report is presented on a case of a 31-year-old male with unknown history of ophthalmic or systemic conditions, who referred to a history of 6 months of blurred vision in his right eye. This was caused by a unilateral retinal hamartoma due to an undiagnosed tuberous sclerosis.
Discussion: Multidisciplinary management, with the cooperation of Internal Medicine and the Oncology Department, is needed in these cases, as well as genetic counseling for affected patients. Complications are directly related to increased tumor size. Treatment does not seem to have any influence on the natural history of the disease.

A propósito de un caso de hamartoma retiniano adyacente a cabeza de nervio óptico en un caso de esclerosis tuberosa

Palabras clave:
Hamartoma retiniano
Esclerosis tuberosa
Astrocitoma

RESUMEN

Introducción: La esclerosis tuberosa es una enfermedad multisistémica rara, con un patrón de herencia autosómico dominante. Existen pocos casos documentados en la literatura de hamartomas retinales (astrocitomas) con progresión agresiva en el contexto de esta enfermedad.

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Caso clínico: Se presenta el caso de un varón de 31 años sin antecedentes de afecciones oftálmicas ni sistémicas conocidas, que refirió una historia de 6 meses de visión borrosa en el ojo derecho causada por un hamartoma unilateral de retina, en un caso no diagnosticado de esclerosis tuberosa.

Discusión: Es necesario realizar un manejo multidisciplinario, con la colaboración del Departamento de Medicina Interna y de Oncología, así como ofrecer asesoramiento genético para los pacientes afectados. Las complicaciones están directamente relacionadas con el aumento del tamaño del tumor. El tratamiento no parece tener ninguna influencia en la historia natural de la enfermedad.

Clinic case report

Patient, age 31, who visited due to blurred vision in the right eye (RE). Examination produced corrected visual acuity (VA) in RE of 20/50 and 20/20 in the left eye. RE ocular fundus showed papillary edema with adjacent whitish lesions and presence of neo-vessels (Fig. 1). Subretinal fluid found from the papilla to the fovea. Macula presented hard exudates and the inferior temporal arcade showed retinal hemorrhages associated with neovessels. The other eye did not exhibit pathological findings.

Systemic study was initiated with analytics and serology as well as CAT image studies, cranial and orbit NMR. It was decided to maintain the patient in observation awaiting pointers to etiology or evolution.

Optical coherence tomography showed subretinal fluid from the papilla to the macula (Fig. 2). Fluorescein angiography showed 2 auto-florescent lesions adjacent to the papilla that capture and issue contrast in middle and late times. In addition, inferior temporal neovessels also exude contrast.

Serology showed negative results for toxoplasma, toxocara, virus herpes, CMV, syphilis, HIV and bartonella. Mantoux was negative. Chest X-ray showed significant increase of hilar adenopathies, for which reason a bronchial–alveole cleansing was performed due to suspected sarcoidosis. Subsequently, the study was completed with chest CAT and bone gammagraphy (due to suspected metastasis in the presence of lytic lesions in the ribs). NMR showed subcortical areas with altered signal, sub-ependyma nodules and sclerous lesions in the cranial socket. These findings, together with the observation of dermal neurofibromas in the nose and back, confirmed the diagnostic of tuberous sclerosis.

After the complex diagnostic, the patient was treated with oral glucocorticoids (prednisone, 1 mg/kg/d) and intravitreal anti-VEGF, in this case Avastin® (bevacizumab) for compassionate use, with initial improvements after 6 months of monthly injections (VA RE 20/32). Even so, the patient exhibited a relapse at 9 months (VA RE 20/200). A new cycle of anti-VEGF injections (3 per month) does not improve VA or exudation. Intravitreal dexamethasone implant (Ozurdex®, Allergan S.A.) is proposed to improve exudation, although the condition did not improve. Sixteen months after diagnostic, hemovitreous appeared (VA of hands movement and a 2 m). Ocular echography was taken (Fig. 3) to discard retina detachment, observing organized hemovitreous and hypo-reflective calcification with a surface of 2 mm^2 and posterior shadow adjacent to the nerve. NMR and CAT were repeated to check the size of the lesions. NMR does not show the tumors allowing for the observation of the indirect exudation signs that these cause. Orbit CAT shows calcification adjacent to the optic nerve (Fig. 3). Vitrectomy was proposed but rejected by the patient, who at present is in follow-up, has declined additional treatment and has been informed about the negative visual prognostic.

Discussion

Tuberous sclerosis is a multisystemic genetic disease, inherited through dominant autosomal pattern[1,2] and its incidence is of 1:6000 live births.[2] Over 50% of patients with tuberous sclerosis exhibit hamartomas in the retina,[1,3] half of which are bilateral.[3]

Retinal hamartoma is characterized by typically benign behavior without ocular compromise, locoregional or distance dissemination. Management is generally conservative, keeping track of tumor dimensions and periodic examinations.[1,2]

Exceptionally, said tumors could adopt an aggressive course.[3,4] Complications are directly related to increased tumor size.[1,3] Treatment of complications does not appear to influence the natural history of the disease.

There are few cases documented in the literature describing retinal hamartomas with aggressive behavior in the context of tuberous sclerosis, mostly among pediatric patients with voluminous lesions located in the posterior pole.[3]

The present patient developed neovessels and exudation in the posterior pole, initially treated conservatively with satisfactory initial response. However, exudation subsequently relapsed. An optional treatment could have been argon laser for managing this complication.

Multidisciplinary approach is recommended with Internal Medicine and Oncology. In the management of this disease, genetic counseling for patients in reproductive age[2,5] should be taken into account because 50% of descentants of a patient will inherit the disease.[5]

The literature reports cases of regression with radiotherapy[4] as well as with photodynamic therapy.[6]

Recent studies with sirolimus (mTOR inhibitor) and interferon have demonstrated to diminish tumor volume in relation to the use of said medicaments.[7]