Case Report

Image Features of Retinal Astrocytic Hamartoma in a Patient with Tuberous Sclerosis Complex

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Abstract

Purpose: To describe the image features of retinal astrocytic hamartoma in a 35-year-old male patient with tuberous sclerosis complex (TSC).

Methods: Fundus photography, fundus autofluorescence (FAF), fundus fluorescein angiography (FFA), and spectral domain optical coherence tomography (SD-OCT) were performed for this retinal astrocytic hamartoma.

Results: Fundus photography showed that the retinal astrocytic hamartoma presented as a well-circumscribed, mulberry-like lesion consisting of glistening yellowish spherules of calcification. FAF demonstrated dense hyper-autofluorescence spots corresponding to retinal astrocytic hamartoma. FFA revealed leakage from dilated retinal capillaries over the hamartoma. SD-OCT indicated moth-eaten optically empty spaces and hyperreflective dots within the lesion. The lesion surface was fluctuate.

Conclusion: FAF is a useful imaging modality for obtaining greater contrast between a retinal astrocytic hamartoma and the surrounding retina due to hyper-autofluorescence of calcification in the lesion. FFA is beneficial for monitoring the abnormal blood vessels in these lesions. SD-OCT is capable of visualization the structural details, such as the uneven surface and inner hyperreflective dots. (***Eye Science 2014; 29:223-226***)

Keywords: astrocytic hamartoma; retina; autofluorescence; optical coherence tomography; tuberous sclerosis

Introduction

Tuberous sclerosis complex (TSC) is an autosomal dominant disorder with an incidence of 1:10,000 to 1:25,000 births that is characterized by the development of benign hamartoma-like tumors, which are usually located in the skin, brain, and eyes[1,2]. More than half of TSC patients have retinal astrocytic hamartomas, which are mainly composed of well-differentiated glial astrocytes[3]. TSC is a rarely reported disease in the ophthalmological literature[1]. Here, we describe the image features of retinal astrocytic hamartoma in a patient with TSC.

Case presentation

A 40-year-old man presented to our ophthalmology clinic for a comprehensive eye examination. He reported that a physician found a mass in his right eye after a full physical examination. His best corrected visual acuity was 20/20 in both eyes. Facial angiofibromas around the patient's nose and chin were noticed, which presented as reddish spots or bumps in a butterfly distribution (Figure 1). A few well-circumscribed, rough, elevated, and. coffee-colored shagreen patches were also found. on the skin of the patient's back (Figure 2). Slit-lamp examination was normal for the anterior segment in both eyes. Intraocular pressure was 17 mmHg in each eye. Dilated funduscopic examination revealed a well-circumscribed, mulberry-like retinal astrocytic hamartoma consisting of glistening yellowish spherules of calcification in his right retina that bordered on the optic disc (Figure 3).

Fundus autofluorescence (FAF) and fundus fluorescein angiography (FFA) images were taken using a confocal scanning laser ophthalmoscope (SLO) (Heidelberg Spectralis HRA; Heidelberg Engineering, Heidelberg, Germany) with a 30-degree field of view. FAF demonstrated the multiple intensely hyper-autofluorescent spots corresponding to the glistening yellowish spherules of calcification in the retinal astrocytic hamartoma (Figure 4). Fundus flu-

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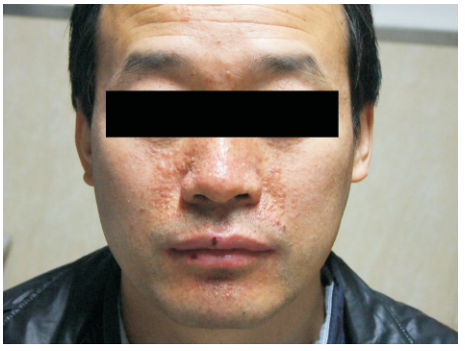

Figure 1 Facial angiofibromas in a butterfly distribution around the patient's nose and chin

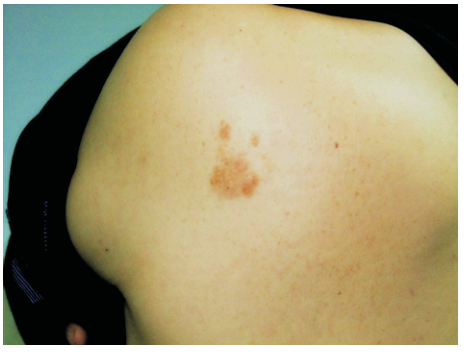

Figure 2 Well-circumscribed, rough, elevated, and coffee-colored shagreen patches on the skin of the patient's back.

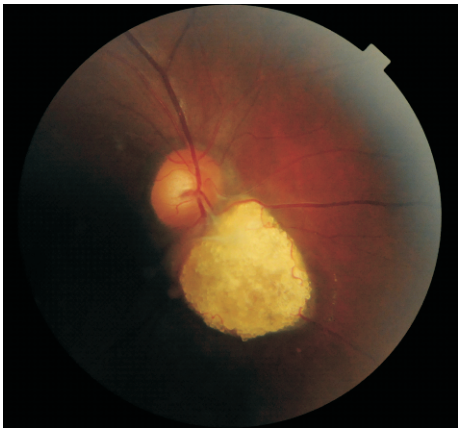

Figure 3 Fundus photography of the right eye showed a retinal astrocytic hamartoma bordering on the optic disk and presenting as a well-circumscribed, mulberry-like lesion consisting of glistening yellowish spherules of calcification.

orescein angiography (FFA) revealed the fluorescein leakage from dilated retinal capillaries over the hamartoma (Figure 5). Spectral domain optical co herence tomography (SD-OCT, 3D-OCT-TM system, Topcon Corporation, Tokyo, Japan) showed moth-eaten, optically empty spaces and hyper-reflective dots within the lesion, and the surface of lesion was uneven (Figure 6). FAF, FFA, and SD-OCT images of the left eye appeared normal.

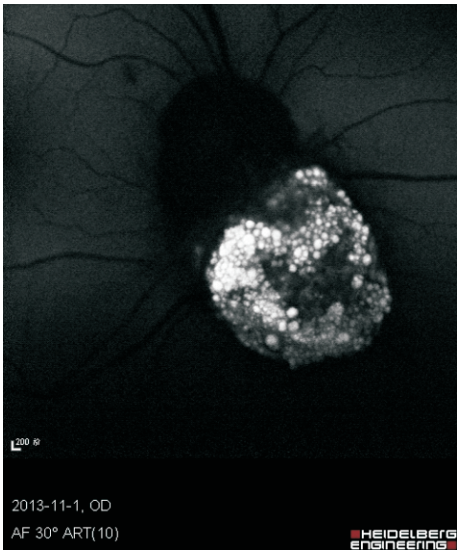

Figure 4 Fundus autofluorescence of the right eye demonstrated the well-circumscribed mass composed of multiple intensely hyper-autofluorescence spots corresponding to the glistening yellowish spherules of calcification in the retinal astrocytic hamartoma.

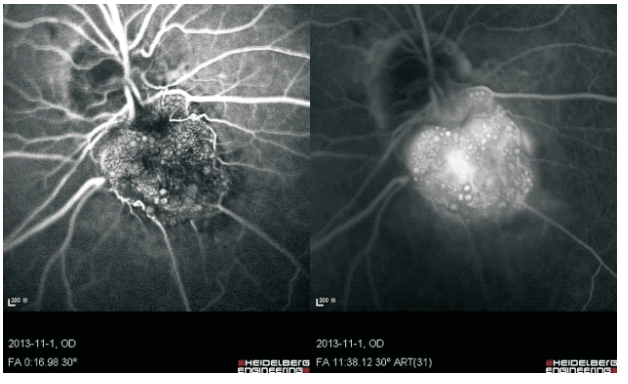

Figure 5 Fundus fluorescein angiography of the right eye revealed fluorescein leakage from dilated retinal capillaries over the retinal astrocytic hamartoma.

The diagnosis of TSC was made and we recommended that this patient undergo cranial magnetic resonance imaging (MRI) to screen for abnormalities in the brain. The cranial MR images were normal, and the patient denied any history of neurobehavioral abnormalities, such as epilepsy or learning difficulties. No treatment was given to this patient in the department of ophthalmology.

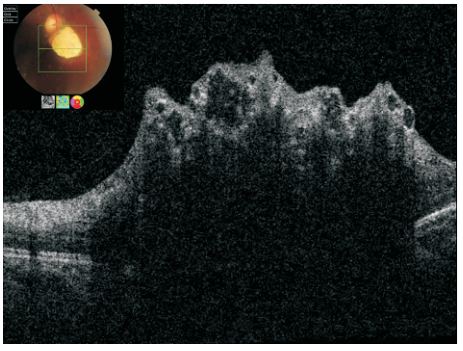

Figure 6 Spectral domain optical coherence tomography showed moth-eaten optically empty spaces, hyperreflective dots within retinal astrocytic hamartoma, and the uneven surface of the lesion.

Discussion

TSC is a multisystem disorder affecting children and adults that results from mutations in one of two genes, TSC1 (encoding hamartin) or TSC2 (encoding tuberin)[4]. TSC has a wide clinical spectrum of disease, and many patients have minimal signs and symptoms with no neurologic disability[5]. Currently, no cure is available for TSC. The diagnostic criteria for TSC were revised at the Tuberous Sclerosis Complex Consensus Conference in 1998[6]. According to these criteria, two major features or one major feature plus two minor features must be present. Major features for TSC include facial angiofibromas or forehead plaque, nontraumatic ungual or periungual fibromas, hypomelanotic macules (three or more), shagreen patch (connective tissue nevus), retinal astrocytic hamartomas, cortical tuber, subependymal nodule, subependymal giant cell astrocytoma, cardiac rhabdomyoma, single or multiple lymphan giomyomatosis, and renal angiomyolipoma. We made the diagnosis of TSC for this patient based on the following major features: facial angiofibromas, retinal astrocytic hamartoma, and shagreen patches. For ophthalmologists, differentiating retinal astrocytic hamartoma from other retinal lesions is helpful in diagnosing TSC.

Retinal astrocytomas can arise from any location in the retina, from the optic disc and macula to the extreme periphery. These benign retinal lesions are classified into three different morphological groups[7,8]. Type 1 lesions usually occur as a circular or oval-shaped solitary lesion with an average size of 0.5 disc diameter. These relatively flat, semitransparent, soft, light grey, glistening masses lie in the retinal nerve fiber layer without signs of calcification. Type 2 lesions are prominent, with multiple calcified nodular areas of variable size arising from the inner surface of the retina. These well circumscribed, multinodular lesions are characteristically found at the posterior pole. Type 3 lesions contain mixed features of both types 1 and 2. We classified the retinal astrocytic hamartoma reported in this case with TSC as type 2.

A type 2 astrocytoma contains multiple calcified nodules that are capable of emitting autofluorescence when excited by blue light[9]. In this case, the retinal astrocytic hamartoma presented as a well-circumscribed mass composed of multiple intensely hyperautofluorescent spots. This appearance confirms that FAF is a useful adjunctive test for obtaining greater contrast between the lesion and the surrounding retina for hyper-autofluorescence of calcification in retinal astrocytic hamartomas.

The FFA image revealed the vascularization of this hamartoma, which consisted of numerous abnormal vessels on the hamartoma surface. In the process of FFA, a gradual increase in fluorescein leakage was evident from these abnormal vessels, which lacked an adequate blood-retinal barrier function, or from fluorescein entrapped in the lesion, which made the hamartoma appear fuzzily circumscribed, and hyper-fluorescent. The FFA features that we observed demonstrate that the retinal astrocytic hamartoma is a highly vascular tumor[9]. This raises the possibility of treating retinal astrocytic hamartomas with anti-angiogenic medications in the future.

The use of SD-OCT in this case allowed visualization of the lesion, revealing a rough and uneven surface. The optically empty spaces observed within the retinal astrocytic hamartoma were likely due to the inner retinal hyper-reflectivity and complete shadowing of the deeper layers of the lesion. The hyperreflective dots in the SD-OCT image corresponded with the calcified nodules.

In summary, we report the FAF, FFA, and SD-OCT features of retinal astrocytic hamartoma in this patient with TSC. These imaging modalities provided

greater sensitivity for detection of this tumor. Nevertheless, the distinct mulberry-like form consisting of glistening yellowish spherules remains the diagnostic basis for retinal astrocytic hamartoma.

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