Astrocytic Hamartoma in a Case with Tuberous Sclerosis

Tüberosklerozlu bir Olguda Astrositik Hamartom

Sevinç Arzu POSTACI[1], Burak TURGUT[2], Seda LİMAN UZUN[1], Murat ERDAĞ[1]

ABSTRACT

Tuberosclerosis is a multisystemic phakomatosis originating from embryonic ectoderm characterized by presence of hamartomatous tumours and central nervous system, skin, visceral organs and eye findings. The aim of this case report is detailed description of systemic and retinal findings of the disease with the help of investigations. The patient was a 4-year-old male under vigabatrin treatment and follow-up by neurology department of pediatrics for epileptic seizures. The patient was consulted to our outpatient clinic for control. Ophtalmologic examination showed visual acuities as 8/10 in both eyes. Chorioretinal depigmented areas were observed on the periphery of the fundus in both eyes . Fundus examination of the right eye showed an astrocytic hamartoma on the inferior temporal arch which was bulgy, hypopigmented, with the size of two optic discs and with no calcifications. Dilated venous vessels were observed on the proximal of this lesion. Also two astrocytic hamartomas with the size of half-disc were observed on the nasal of optic disc. OCT showed surface bulging on hamartomatous lesion, hyper-reflectance and disorganization on the inner retinal layers and shading on the outer retinal layers. In the management of tuberous sclerosis, genetic counseling and multidisciplinary follow up are necessary as the disease is multisystemic.

Key Words: Astrocytoma, Astrocytic hamartoma, Retinal case, Tuberous sclerosis, Child.

ÖZ

Tuberoskleroz santral sinir sistemi, deri, visseral organlar ve göz bulguları olan hamartamatöz tümörler ile karakterize multisistemik embriyonik ektoderm kaynaklı bir fakomatozdur. Bu olgu sunusunun amacı hastalığın sistemik ve retinal bulgularını tetkikler ışığında ayrıntılı şekilde tanımlamaktır. Olgu, 4 yaşındaki erkek hasta olup epilepsi nöbetleri nedeniyle pediatrik nöroloji anabilim dalında vigabatrin kullanımı ile takip edilmekteydi. Hasta kontrol amacıyla polikliniğimize yönlendirilmişti. Oftalmolojik muayenede hastanın görme keskinliği her iki gözde 8/10 düzeyindeydi. Hastanın her iki göz fundus periferinde koryoretinal depigmentasyon alanları izlendi. Sağ göz fundus muayenesinde alt temporal ark üzerinde kalsifikasyon göstermeyen, kabarık, hipopigmente, iki optik disk boyutunda astrositik hamartom izlendi. Bu lezyonun proksimalinde dilate venöz damarlar görüldü. Aynı zamanda optik disk nazalinde de yarım disk boyutunda iki adet astrositik hamartom izlendi. OCT'de hamartomatöz lezyonda yüzey kabarıklığı, iç retinal tabakalarda hiperreflektans, disorganizasyon ve dış retinal tabakalarda gölgelenme izlendi. Tuberosklerozda multisistem tutulumu olduğundan diğer branşlarla birlikte takibi ve genetik danışmanlık gereklidir. .

Anahtar Kelimeler: Astrositom, Astrositik hamartom, Retinal olgu, Tuberoz skleroz, Çocuk.

INTRODUCTION

Retinal astrocytic hamartoma (RAH) or astrocytoma is a benign glial tumor of the retinal nerve fiber layer (RNFL) that origins from retinal astrocytes. It usually manifests as a creamy-white, well-circumscribed, elevated lesion with a multilobulated, opaque and "mulberry" appearance. However, the lesion may be multiple or solitary and flat or semitranslucent. RAH occurs most frequently in the patients with tuberous sclerosis (TS), and rarely neurofibromatosis.[1] TS is a syndrome characterized by benign tumors or hamartomas of the brain, kidney, lung, heart, and eyes. Retinal involvement has been described by Van der Hoeve in 1920 . Fundus lesions of TS include astrocytic hamartomas of the retina and optic nerve and depigmented lesions of the retina.[1-2] We report this case because of its rarity.

1- Asist. Dr., Fırat University, Faculty of Medicine, Department of Ophthalmology, Elazığ, Türkiye

2- Prof. Dr., Çanakkale Onsekiz Mart University, Faculty of Medicine, Department of Ophtalmology, Çanakkale, Türkiye

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Yazışma Adresi / Correspondence Adress:
Sevinç Arzu POSTACI
Fırat University, Faculty of Medicine, Department of Ophthalmology, Elazığ, Türkiye

Phone: +90 554 860 9984
E-mail: postaciarzu@gmail.com

CASE REPORT

The patient is a 4-year-old male under vigabatrin treatment and follow-up by neurology department of pediatrics. The patient was consulted to our outpatient clinic for control. The brain tomography showed tubers on the bilateral frontoparietal and temporal juxtacortical area and subependymal calcific nodules on the lateral ventricles (Figure 1). The examination of the skin showed angiofibromas the biggest of which had a dimension of 0,5 cm, thickened skin plaques on the cheek and forehead and diffuse hypo-hyperpigmented areas on the whole body (Figure 2). Cardiac rhabdomyoma was detected with echocardiography. There were simple cysts on both kidneys on the abdominal magnetic resonance imaging. Visual acuities were 8/10 in both eyes. Anterior segment examination was bilaterally normal. Chorioretinal depigmented areas were observed on the periphery of the fundus in both eyes (Figure 3). OCT showed surface bulging on hamartomatous lesion, hyper-reflectance and disorganization on the inner retinal layers and shadowing on the outer retinal layers (Figure 4). Diagnosis of TS was

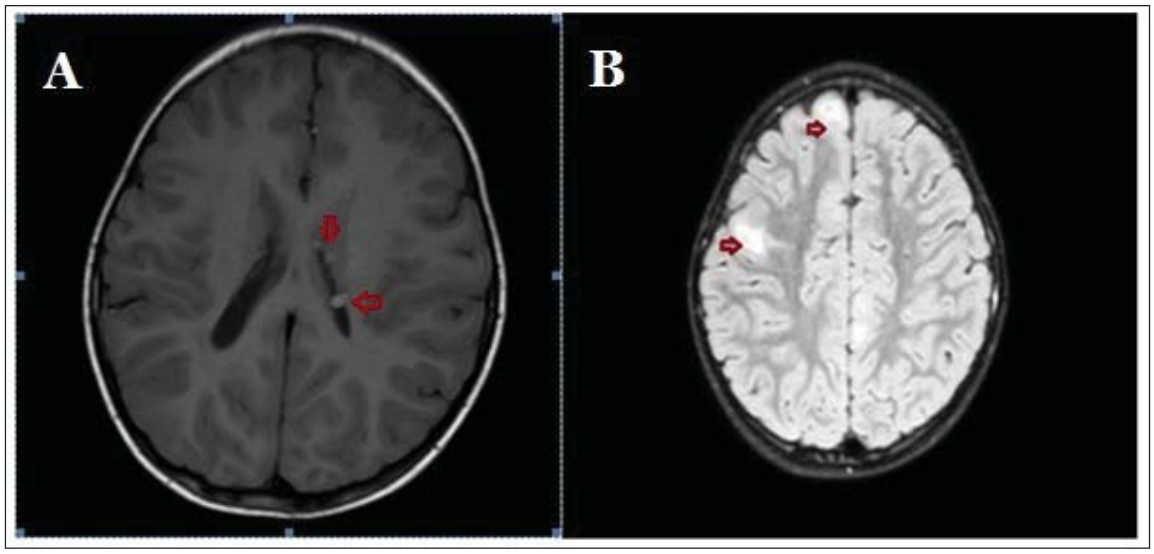

Figure 1. *Subependymal calcified nodules in the lateral ventricle* (**A**) *and subcortical intensities in the cerebral parenchyma* (**B**).

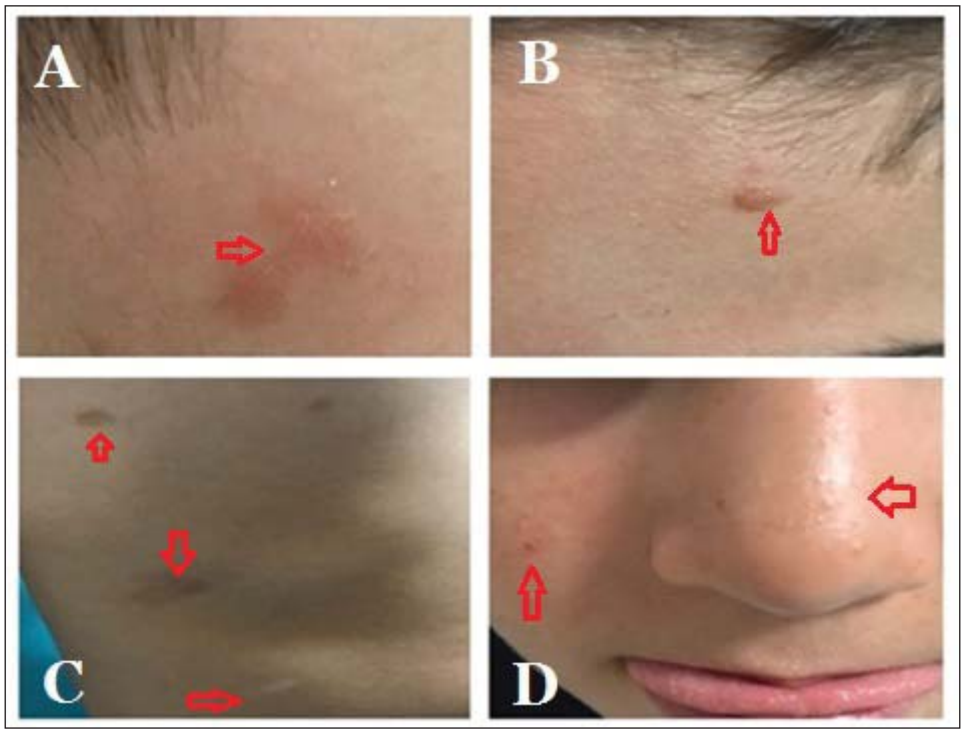

Figure 2. *Cheek and thickened skin plaques* (**A** *and* **B**)*, hypo-hyperpigmented areas throughout the body* (**C**)*, angiofibromas* (**D**).

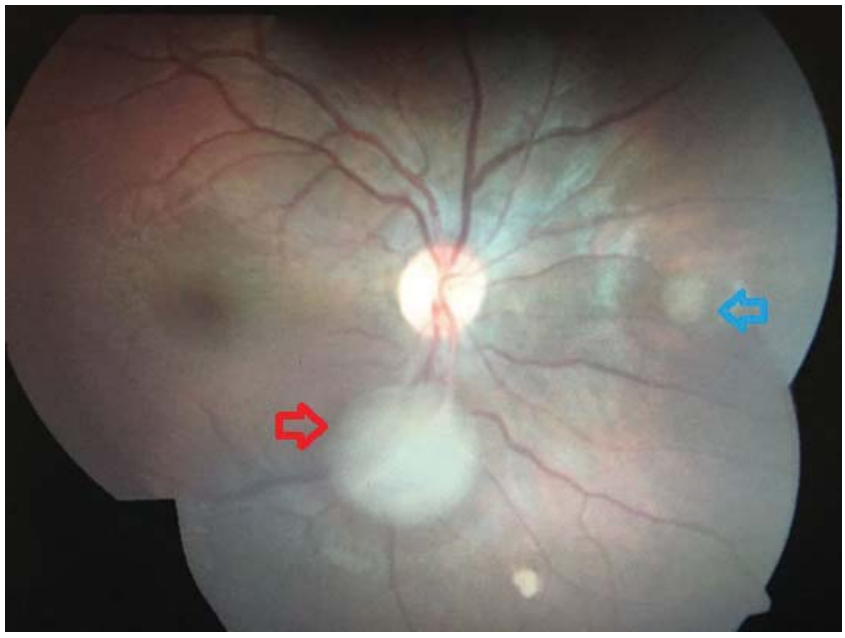

Figure 3. *Two astrocytic hamartomas one of which is bulgy, hypopigmented and with the size of two optic discs and on the proximal of which dilated venous vessels were observed on the right inferior temporal arch with no calcifications (red arrow) and the other one of which was on the nasal of optic disc with the size of half-disc (blue arrow).*

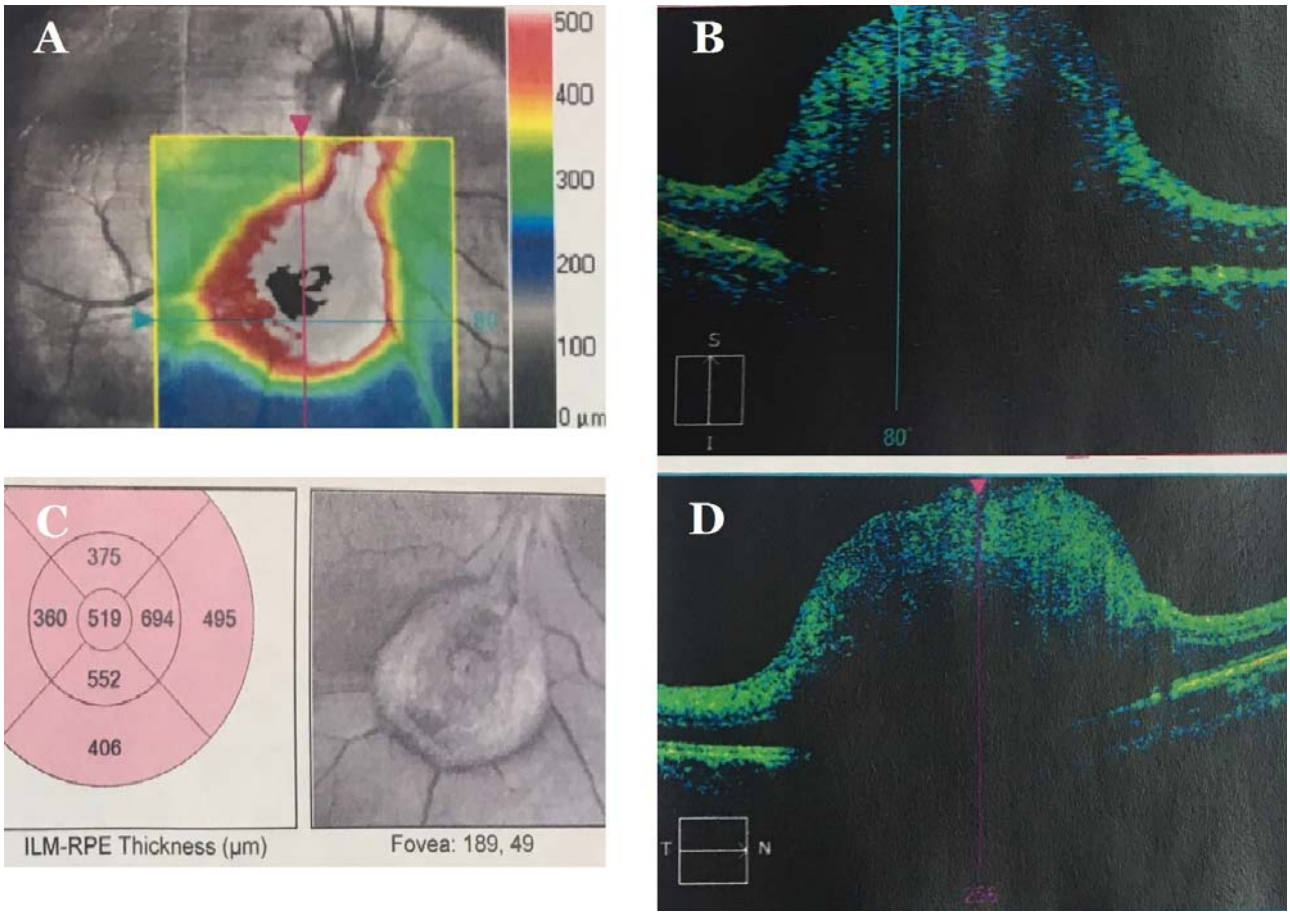

Figure 4. *Hamartoma with surface swelling, hyper-reflectance in the inner retinal layers and the shadowing in deep layers in optical coherence tomography.* **A**: *overlapped fundus image and topographic map of the lesion,* **B**: *a horizontal scan crossing the lesion,* **C**: *internal limiting membrane-retina pigment epithelium thickness in the lesion,* **D**: *a vertical scan crossing the lesion.*

performed based on clinical and radiological findings. The consent for the presentation was taken from patient's parents.

DISCUSSION

Tuberous sclerosis is a syndrome characterized by benign tumors or hamartomas of the brain, kidney, lung, heart, and eyes. Its prevalence is 1/10000. One-third of cases have an autosomal transition and the rest is sporadic.[1-2] Our case is sporadic . A consensus was achieved on clinical criteria for the diagnosis of TS in the 1990s. The suggested clinical classification is as certain, possible and suspicious TS. Clinical findings were defined as primary, secondary and tertiary according to the specificity of the disease. Primary findings are diagnostic and include adenoma sebaceum, ungual fibromas, cortical tubers, subependymal nodules or giant cell astrocytoma which are histologically shown, and multiple calcified subependymal nodules extending into the ventricle and multiple retinal astrocytomas which are demonstrated radiologically. Except for the signs of the central nervous system, skin, and eye, the findings of TS on visceral organs and skeletal system were also defined.[2] Ocular findings of TS can be classified as fundus lesions and non-fundus lesions. Non-fundus lesions include angiofibromas of eyelids, poliosis, iris-lens-choroid coloboma, focal hypopigmented lesions of iris and irido-ciliary hamartomas. Fundus lesions include astrocytic hamartomas of the retina and optic nerve and depigmented lesions of the retina.[1-2-3] Although RAHs are multifocal and bilateral, they may be solitary but have a lower diagnostical value. RAHs have two morphological types including wide, bulgy, white (calcified), mulberry shaped nodular lesions, and smooth, translucent (non-calcified), soft appearing lesions. If both types exist; it is called an intermediate type[2,4,5]

According to the classification criteria of TS, our patient can be classified as "Certain Tuberous Sclerosis" as he had 3 primary findings including adenoma sebaceum, multiple calcific subependymal nodules and multiple retinal astrocytomas, 4 secondary findings including cerebral tubers, shagreen patch, forehead plaque, hypopigmented patches in retina and 1 tertiary finding as hypomelanotic macules. Almost all RAH are endophytic. While endophytic tumors are hamartomas originating from the nerve fiber layer of the retina, exophytic tumors originate from subretinal space. These tumors are benign. They may grow slowly, may be calcified and increased calcification may cause progressive growth.[2] Changes of the retina pigment epithelium can be shown in OCT. Based on SD-OCT and clinical evidence, the independent examiners classified the RAH as 4 groups.[7]

Type I shows SD-OCT evidence of flat lesion and clinical evidence of no retinal traction .

Type II shows SD-OCT evidence of slightly elevated (height < 500 μm) lesion and clinical evidence of retinal traction .

Type III shows SD-OCT evidence of elevated retinal mass (height >500 mm) with inner retinal calcification and clinical evidence of mulberrylike calcification.

Type IV shows SD-OCT evidence of elevated retinal mass (height >500 mm) with optically empty cavity and clinical evidence of smooth noncalcified inner retinal mass. According to this classification, our case is considered as type II.

The real neoplasms may occur in kidneys and brain.[8-9] The malign transformation may occur in retinal hamartomas rarely.[1-2] Spontaneous regression of a giant RAH was reported in one case.[10] TS patients have a shorter estimated lifetime. The most common causes of death in these patients are renal failure, disorders of the central nervous system, the defect of cardiac conduction, failure of heart and lung and status epilepticus and pneumonia in epileptic patients.[2, 11]

In conclusion, in the management of tuberous sclerosis, genetic counseling and multidisciplinary follow up are necessary as the disease is multisystemic. The lifetime and the quality of life can be increased with the prevention of complications.

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