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Case Report

TUBEROUS SCLEROSIS COMPLEX

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ABSTRACT
Tuberous sclerosis (TS) is an autosomal dominant disorder characterized mainly by mental retardation, epilepsy and adenoma sebasemia triad. All triads can be detected in only one third of the patients. Tuberous sclerosis complex (TSC) is a hamartomatosis that mainly carries the central nervous system, skin, retina, kidney and heart. Pathologies associated with renal involvement in patients with tuberous sclerosis; angiomyolipomas (70-80%), renal cysts (20%) and very rarely cancers. The most common neurological finding is epileptic seizures. In this article, we present a case report of a patient who applied to dermatological clinic with acne complaints and diagnosed as a tuberous sclerosis complex with skin findings, cortical tuber and angiomyolipoma in addition to radiological examinations.

Key words: tuberous sclerosis, çocuk, acne

ÖZET

Tuberoskleroz mental retardasyon, epilepsi ve adenom sebase tiradıyla karakterize otozomal dominant bir hastalıktır. Triadın tamamı olguların üçte birinde tespit edilebilir. Tuberoskleroz kompleksi, sinir sistemi, deri, retina, böbrek ve kalbi kapsayan bir hamartomatosistir. Tuberosklerozlu hastalarda renal tutulumla birlikteki patolojiler; anjiomyolipomlar (%70-80), renal kistler (%20) ve çok nadiren kanserlerdir. Bu makalede, akne yakınmasıyla dermatoloji kliniğine başvuran ve deri bulguları, kortikal tuber ve anjiomyolipom ile tuberoskleroz kompleks olarak tanı konan bir olgu sunuyoruz.

Anahtar kelimeler: tuberoskleroz, çocuk, akne

INTRODUCTION

Tuberous sclerosis (TS) is an autosomal dominant disorder characterized mainly by mental retardation, epilepsy and adenoma sebaceum triad. All triads can be detected in only one third of the patients. Tuberous sclerosis complex (TSC) is a hamartomatozy that mainly carries the central nervous system, skin, retina, kidney and heart. Pathologies associated with renal involvement in patients with tuberous sclerosis; angiomyolipomas (70-80%), renal cysts (20%) and very rarely cancers and the most common neurological finding is epileptic seizures.[1,2]

In this article, we present a case report who diagnosed as a tuberous sclerosis complex with skin findings like acneiform lesions and a radiologic examination with cortical tuber and angiomyolipoma.

CASE REPORT

A 24-year-old female patient applied to the dermatology polyclinic because of bubbles in the face area that was existed since the age of five. The mentioned lesions were found to be consistent with the nonspecific perivascular dermatitis resulting from biopsy in 2003. There was no family history of the patient. In routine blood tests, vitamin D was detected low and was replated. On dermatological examination, there were lesions compatible with angiofibromas but there were no shagren patch and periungal fibroma. Biopsy was obtained from the lesions. After the pathological assesment the spindle cells were noted and they were found compatible with early stage fibrous papules. The patient was evaluated with ophtalmology for the eye findings of tuberous sclerosis and the examination was normal. Her neurological examination was normal but because of her febrile convulsion story and for the asssesment of cognitive situation, electroencephalogram (EEG), contrast-enhanced cranial magnetic resonance imaging and neuropsychological test were obtained. On contrast-enhanced cranial MRI, bilateral frontal lobes especially more on the left side showed that cortical-subcortical T2A FLAIR hyperintense nodular signal changes. There was no contrast enhancement or diffusion limitation in the lesions (Figure 1) and lesions were evaluated primarily cortical-subcortical tuber. Her EEG examination was normal and the neuropsychological test was usual except maintain mildly simple and frontal attention difficulty. The patient was consultation with nephrology for tuberous sclerosis-related renal pathologies. In the renal USG, nodular appearances were seen bilaterally multiple which may be compatible with hyperechoic angiomyolipomas and the bigest one was measured 6 x 5 mm in the right middle pole. The patient was assessed with genetic and TBC2 gene was analysed.

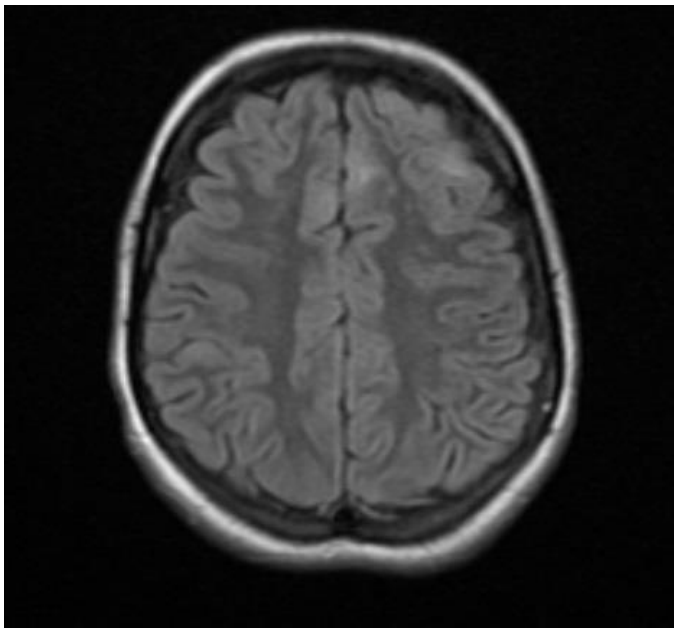

Figure 1. There is no contrast enhancement or diffusion limitation in the lesions.

DISCUSSION

Tuberous sclerosis is an autosomal dominant inherited neurocutaneous syndrome characterized by hamartomatous lesions mainly in numerous organs including skin, brain, eye, kidney and heart.[1] It is assumed that the genetic transition of the disease is due to gene loci on chromosomes 9 and 16. Mutations on chromosome 16 are more common and associated with more severe phenotypes. 75% of the patients have new mutations, and 1/3 have family history.[2]

The basic intracranial lesions of tuberous sclerosis are subependymal nodules and cortical tuberculosis. Tubers are benign lesions composed of cortical or subcortical dysmorphic neurons and cortical tubers may be calcified. Subependymal nodules occur in 95% of cases with TS. Lesions cause neurological symptoms by compressing where they are. Magnetic resonance imaging is more sensitive in detecting lesions.[3]

Adenoma sebaceum (AS), also known as facial angiofibroma, is the most characteristic skin lesion of TS. It is found in 75% of patients. The connective tissue nevi, commonly known as the shagreen patch, is usually seen in the lumbosacral region. It is found in 50% of patients and occurs within the first 10 years of life. Periungual fibromas are asymptomatic tumors of 5-10 mm in length in the flat, hard, horny, skin color of the hand and toe nail folds, also

called Coenen tumors and 50% of the patients were found[4] but there is only facial angiofibromas in our case.

CONCLUSION

TS is a disease with both skin involvement and systemic involvement. These patients should be evaluated in terms of brain, renal, heart, and eye involvement and should be followed closely. Although there are no neurological complaints, further investigations such as imaging methods may be necessary to demonstrate neurological involvement. Our case is important because it revealed that only in case of patients with dermatological complaints can be suspected tuberous sclerosis complex by the dermatologist, and may conduct further investigations and direct the patient to reveal other systemic involvements.

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