Segmental tuberous sclerosis in a patient presenting as unilateral facial angiofibromas, periungual fibromas and Shagreen patch

Abstract. – Tuberous sclerosis (TS) known as Epiloia and Bourneville’s disease is an autosomal dominant inherited disorder that can affect many organ systems. Tuberous sclerosis has many forms of clinical presentation. Rarely, multiple facial angiofibromas of unilateral distribution have been reported. We describe a patient with unilateral facial angiofibromas, periungual fibromas and Shagreen patch.

Key Words: Tuberous sclerosis, Facial angiofibromas, Periungual fibromas, Shagreen patch.

Dear Editor,

Tuberous sclerosis (TS) known as Epiloia and Bourneville disease¹ is a hamartomatosis which mainly affect central nervous system, skin, kidney, liver and heart. Vogt, in 1908, delineated the classic triad of facial adenoma sebaceum, epilepsy and mental retardation².

Cutaneous lesions in TS include ash leaf hypopigmented macules, facial angiofibromas, periungual fibromas, Shagreen patches, forehead fibrous plaques, confetti hypopigmentation and poliosis³.

Multiple facial angiofibromas are thought to be a pathognomonic feature of TS. In contrast, it is rare to see multiple angiofibromas limited to one side of the face. Some Authors have speculated that this unilateral development may be derived from either a postzygotic mutation or from an abnormality in the highly conserved homeobox genes⁴,⁵.

Here, we describe a case of a 23-year-old woman who presented with unilateral facial angiofibromas, periungual fibromas and Shagreen patch without other features of TS.

A 23-year-old woman first presented for evaluation of right-sided facial papules that were first noted at 7 years of age. On examination, the patient has multiple, discrete, 2-4 mm, flat, skin-colored and erythematous firm papules involving the right side of the face (Figure 1). Interestingly, periungual fibromas were found in the right hand and foot (Figure 2). Also, Shagreen patch was seen in the right lumbosacral region (Figure 3). Soft fibromas was found in the right side of the neck (Figure 4). There were no cafe-au-lait macules or hypopigmented patches. The patient has no evidence of mental retardation and no history of seizures. There was no family history of dermatologic, neurologic, renal or cardiac problems.

Histopathology of the unilateral lesions on the face revealed a clearly differentiated zone of perivascular fibrosis in the epidermis and large stellate cells in the dermis. These findings are characteristic of angiofibromas.

Additional examinations were made to find other manifestations of tuberous sclerosis. Cranial CT, abdominal USG, EEG and radiography of the chest were normal. The fundus of the eye was also normal. Genetic study was not available.

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Facial angiofibromas are often arranged bilaterally in the nasogenian folds and cheeks. Some cases have been related with MEN 1 and neurofibromatosis but none of them were reported as unilateral. A few cases of unilateral facial angiofibromas have been reported.  

Their clinical significance is unknown. It has been related to the mutation of two genes, one of which is located in chromosome 16 and the other in chromosome 9. In normal conditions, these genes act as tumor suppressors but, once affected, they stop carrying out their function and the tumors characteristic of tuberous sclerosis appear.
Recently, it has been suggested that unilateral facial angiofibromas are a segmental, genetically defined form of tuberous sclerosis. Our patient has multiple angiofibromas affecting only right side of the face, periungual fibromas in the right hand and foot and Shagreen patch in the right lumbosacral region. No other characteristic features of TS were present.

Segmental manifestations of tuberous sclerosis have been reported to involve the brain, maxilla and mandible or one side of the face. Reported treatments for facial angiofibromas have included curettage, chemical peeling, cryosurgery, dermabrasion, shave excision and copper vapor, argon and carbon dioxide laser ablation. Our patient has responded well to cryosurgery.

In conclusion, our case of unilateral facial angiofibromas, periungual fibromas and Shagreen patches likely represents a rare clinical manifestation of tuberous sclerosis.

References


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