Original Research Paper

Volume - 11 | Issue - 11 | November - 2021 | PRINT ISSN No. 2249 - 555X | DOI : 10.36106/ijar

Dermatology

A RARE CASE REPORT OF ISOLATED FACIAL ANGIOFIBROMAS.

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ABSTRACT Tuberous sclerosis complex (TSC) is an autosomal-dominant neuro-cutaneous disorder affecting multiple organs, with hamartomas developing in the brain, skin, kidneys, heart, and eyes. Cutaneous manifestations include hypomelanotic macules, subungual fibromas, facial angiofibromas, fibrous plaques of the forehead, and shagreen patchesFacial angiofibromas (FA) occur in 80%–90% of Tuberous Sclerosis Complex patients. They commonly occur as bilateral symmetrical small brown to erythematous telangiectatic papules over cheek, chin, and nose. FAs with or without poliosis, as the only clinical manifestation of Tuberous Sclerosis Complex is rare. Here, we report a case of isolated presentation of facial angiofibromas without any other cutaneous and systemic features.

KEYWORDS : Facial Angiofibroma, Isolated, Tuberous Sclerosis.

INTRODUCTION:

Tuberous sclerosis complex (TSC) is an autosomal-dominant neuro-cutaneous disorder affecting multiple organs, with hamartomas developing in the brain, skin, kidneys, heart, and eyes. Cutaneous manifestations include hypomelanotic macules, subungual fibromas, facial angiofibromas, fibrous plaques of the forehead, and shagreen patches. More than 90% of patients with TSC have at least one cutaneous manifestation, though none are pathognomonic. [1] They commonly occur in bilateral distribution, but rarely they can be unilateral [2]. We report here, a case of isolated bilateral facial angiofibromas without systemic involvement.

CASE REPORT:

A 21-year-old female born of the non-consanguineous marriage presented to us with asymptomatic red to dark-colored raised lesions over face for the past 7 years. The lesions first appeared at 14 years of age, which progressively increased in number and size over the last 7 years. There was no history of seizures, headache, visual or auditory disturbances, mental retardation, or early puberty. None of the family members had similar complaints. Cutaneous examination revealed multiple, firm, well-defined, dome-shaped, reddish-brown papules present over bilateral cheeks, nose and on right upper eyelid. There were no periungual fibromas, shagreen patches, café-au-lait macules, forehead plaques, or hypopigmented patches. The rest of the cutaneous and systemic examination was unremarkable.

Based on history and clinical examination, differential diagnoses of bilateral facial angiofibroma, trichoepithelioma, fibrofolliculoma, syringoma, and sebaceous hyperplasia were considered. Dermoscopy of facial lesions revealed multiple yellowish-white dots distributed over a pinkish-grey background, suggestive of angiofibroma.

Histopathological examination of papule over the right cheek revealed the presence of concentric arrangement of collagen bundles around multiple hair follicles and dilated blood vessels in the upper dermis suggestive of angiofibroma. Extracutaneous involvement was excluded by carrying out appropriate investigations such as computed tomography of the brain, chest X-ray, abdominal sonography, echocardiography, and fundus examination. Genetic mutation analysis was not done due to a lack of resources. Based on history, examination, dermoscopy, and histopathology, a diagnosis of bilateral facial angiofibromas with no other features of TSC was reached.

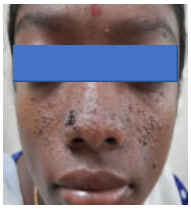

Figure 1 : Multiple, firm, well-defined dome-shaped, red- brown papules present over bilateral cheeks and the nose.

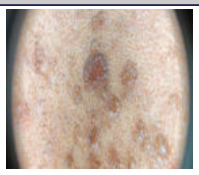

Figure 2: Dermoscopy of facial papule showing multiple yellowish-white dots distributed over a pinkish-gray background, suggestive of angiofibromas

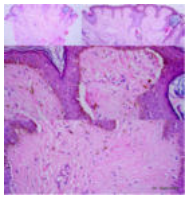

Figure 3: Histological examination of papule over right cheek showing irregular proliferation of fibrous tissue and blood vessels with the adnexae surrounded and compressed by concentric lamellae of collagen. Numerous spindled to stellate fibroblasts and melanophages are cited within the collagenous stroma.

DISCUSSION:

Adenoma sebaceum was first described as a distinct feature of TSC by Balzer and Menetrier [3] in 1885 and Pringle in 1890. However, adenoma sebaceum is a misnomer as these lesions are neither adenomatous nor sebaceous. The histopathologic evaluation has proven these lesions to be angiofibromas. [3] The classical triad of mental retardation, convulsions, and angiofibromas occur in 29% of TSC patients and 6% of them lack all manifestations. Facial angiofibromas occur in 80%–90% of TSC patients, typically presenting after 5 years of age. They commonly occur as bilateral symmetrical small tan to erythematous telangiectatic papules over cheek, chin, and nose. These lesions have also been reported in patients with multiple endocrine neoplasia type 1 and neurofibromatosis (NF). FAs with or without poliosis, as the only clinical manifestation of TSC is rare [4]. Some authors suggested that patients with isolated FAs need to follow up to look for the development of other extracutaneous manifestations of TSC. The dermoscopic features of FA were described by Bahera in 2017 as multiple yellowish-white dots distributed over a pinkish-grey background and crypts in few lesions, as seen in our case [5]. Histologically, the yellowish-white dots correspond to the follicular hyperkeratosis along with the presence of sebum, the pinkish-grey colour to the proliferating blood vessels along with pigmentary incontinence, dermal melanophages while crypts to the pseudofollicular opening. Isolated FAs without any other evidence, as in our case, are very rarely reported in the literature. Invasive procedures such as cryotherapy, radiofrequency ablation,

dermabrasion, excision, chemical peeling, and lasers, have been tried but all these procedures carry a risk of permanent scarring, the requirement of sedation, and incomplete removal. Recently, topical sirolimus has shown significant improvement of adenoma sebaceum. In our case, he was treated with cryotherapy, and there was a moderate improvement of lesions without any sequelae. We report this case because of its rarity to create awareness among dermatologists and to emphasize the need to follow up in such patients for the development of extracutaneous manifestations of TSC.

DECLARATION OF PATIENT CONSENT:

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published, and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

FINANCIAL SUPPORT AND SPONSORSHIP:

Nil.

CONFLICTS OF INTEREST:

There are no conflicts of interest.

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