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RESEARCH LETTER

WILEY

Dermoscopy of facial angiofibromas in Fitzpatrick II-III phototype subjects affected by tuberous sclerosis complex

Dear Editor,

Tuberous Sclerosis Complex (TSC) is an autosomal dominant multisystemic syndrome with a wide range of clinical features.[1–3] The diagnosis is generally easy and is usually based on the recognition during infancy of typical cutaneous and neurologic signs. However, skin lesions may show different age of onset and severity making the diagnosis more difficult.[1–3] In case of mild disease, TSC may remain unknown for years.

Facial angiofibromas (FAs) are benign hamartomatous skin growths occurring in nearly 75% of patients with TSC.[4,5] They appear as reddish, fibrous, dome-shaped papules mainly involving the central area of the face (especially around nasolabial folds). The usual onset is between ages 2 and 5 years.[2,4] The identification of FAs is important as they may represent the sign of a TSC not yet diagnosed. Although one or two isolated sporadic lesions may be observed within the general population, the presence of at least three lesions is considered as major criterion for the diagnosis of TSC.[5] When few in number or later in onset, FAs require biopsy to histologically confirm the diagnosis.

The aim of this study was to evaluate the dermoscopic features of FAs in order to establish if this technique[6] may enhance their recognition and, consequently, early TSC identification.

All patients with a diagnosis of TSC and the presence of FAs observed over a period of six months were evaluated by dermatologic examination and dermoscopy. In selected cases, skin biopsy for histopathology was proposed.

A total of 10 TSC patients ($M = 2$, $F = 8$; mean age: 19,8 years, range: 10–33 years) with Fitzpatrick phototype II (six cases) and III (four cases) affected by FAs were enrolled. In four cases, the diagnosis of TSC had been performed before birth by genetic test (to confirm

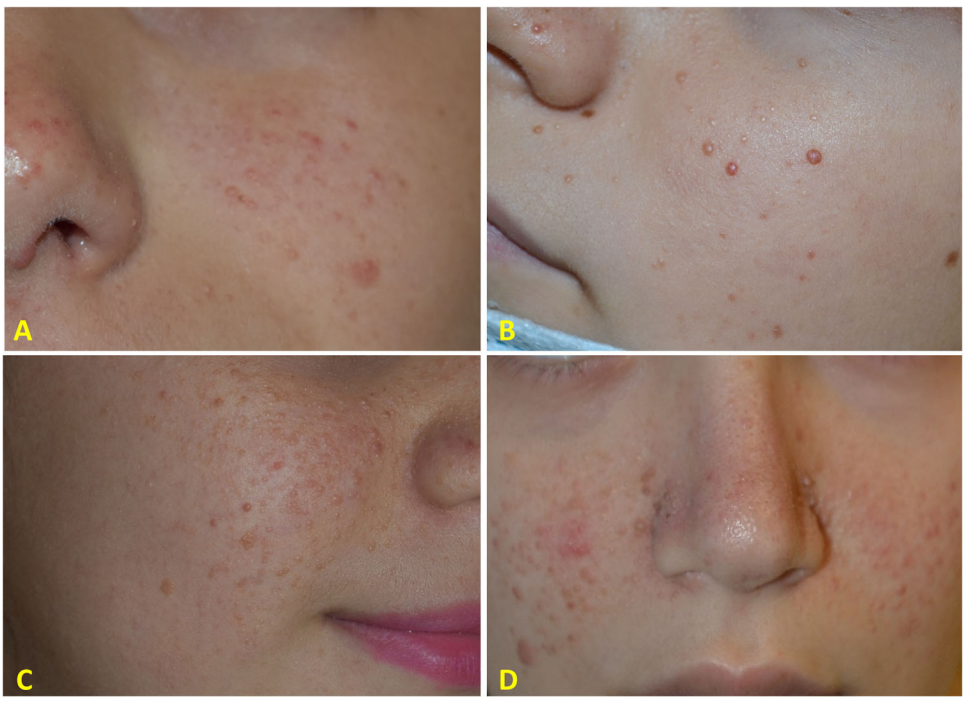

FIGURE 1 (A–D) Facial angiofibromas in four children affected by tuberous sclerosis complex: reddish, dome-shaped papules mainly involving the central area of the face.

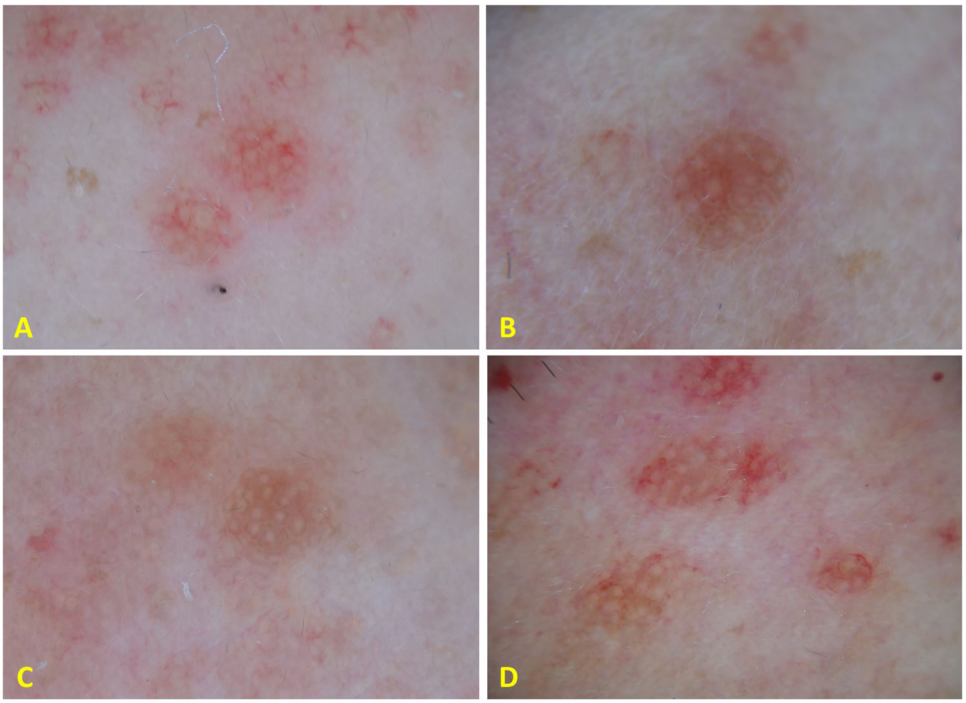

FIGURE 2 (A–D) Polarized dermoscopy of facial angiofibromas of patients as shown in Figure 1: multiple whitish globules over a reddish-brown background in a repetitive pattern; comma and/or hairpin vessels may also observed (A,D).

mutation of the TSC1 or TSC2 gene) following the observation of cardiac rhabdomyomas during the prenatal follow-up ultrasound. In six cases, the diagnosis had been made after birth: specifically in one case at birth, three within the first year of life, one during the second year of life, and one case at 16 years.

Clinically, in all patients FAs appeared as multiple reddish-pink papules located in the central-facial area (Figure 1). Patients and/or parents referred that FAs appeared at birth (one case), or during the first (one case), the third (four cases), the fourth (three cases), and the eleventh (one case) year of life. Polarized dermoscopy showed in all lesions a repetitive pattern, consisting of multiple whitish globules over a reddish-brown background; in the majority of cases, comma and/or hairpin vessels were also observed (Figure 2). The reddish hue tended to fade under pressure. In two cases, a skin biopsy revealed a dermal proliferation containing dilated vessels and fibrosis surrounding the hair follicles.

An early diagnosis of TSC is crucial to plan a proper management in order to avoid and/or delay the systemic manifestations of the disease, thus reducing the mortality and morbidity rates.[1–3] It is important also in mild and/or asymptomatic cases, as affected subjects should be made aware of the possible transmission of the disease to offspring. Early identification of FAs is important as they may be the sign of an undiagnosed TSC. However, they may be sometimes overlooked, especially in those patients with scattered lesions, and/or misdiagnosed as dermal melanocytic nevi, molluscum contagiosum, acne vulgaris, syringomas, etc. To date, the dermoscopy findings of FAs associated to TSC, consisting of a reddish-brown background, whitish globules, comma and/or hairpin vessels, gray-brown dots and crypts, have been described only in patients with skin of color.[7–9] In our cases, we observed in all lesions the same dermoscopic features except for gray-brown dots and crypts observed in the previous reports, probably related to the different phototype. Regarding the histopathological correlation, the whitish globules likely correspond to perifollicular fibrosis, and the reddish-brown background to increased number of dermal vessels and melanocytic hyperplasia.[7–10]

In conclusion, we described for the first time the dermoscopic aspects of FAs in patients with skin phototype II-III affected by TSC. Our case series confirms that dermoscopy. through the recognition of a characteristic pattern, is a simple, non-invasive and affordable tool that may enhance the clinical diagnosis of FAs in patients affected by TSC of all skin phototypes, ruling out other facial disease that may look clinically similar, but that have a different dermoscopic pattern.

DATA AVAILABILITY STATEMENT

Data available on request from the authors.

ETHICS STATEMENT

The procedures followed were in accordance with the ethical standards of the responsible committee on human experimentation (institutional or regional) and with the Helsinki Declaration of 1975, as revised in 1983. The patients in this manuscript have given written informed consent to publication of their case details.

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