GENETICS AND GENODERMATOSES

A POLYMORPHIC RASH REVEALING THE DIAGNOSIS OF TUBEROUS SCLEROSIS

Y Soua [1] - K Belhareth [1] - M Daldoul [2] - M Ben Salem [3] - B Sriha [4] - M Korbi [1] - I Lahouel [1] - H Belhajali

University Hospital Of Monastir, Fattouma Bourguiba Hospital, Department Of Dermatology, Monastir, Tunisia [1] - University Hospital Of Monastir, Fattouma Bourguiba Hospital, Department Of Dermatology, Sousse, Tunisia [2] - University Hospital Of Monastir, Fattouma Bourguiba Hospital, Department Of Nephrology, Monastir, Tunisia [3] - University Hospital Of Sousse, Farhat Hached Hospital, Department Of Anatomopathology, Sousse, Tunisia [4]

Background: Tuberous sclerosis (TS) is an autosomal dominant disease characterized by an extreme variability of clinical presentations. Recently, several dermatosis have been reported in patients affected by TS.

Observation: Patient S.M aged 39, presented a generalized polymorphic rash evolving for 3 weeks. Physical examination revealed erythematous scaled lesions on the face, trunk and upper limbs. We noted purpuric lesions with slight scaling in the lower limbs. She had no mucosal involvement. In addition, she presented multiple facial angiofibromas and koenen tumors on the toes. Clinical diagnosis of TS was made based on the characteristic lesions (in the face and nails) and the discovery of similar lesions in her son. Further explorations showed only the presence of three renal angiomyolipoma. A skin biopsy with direct immunofluorescence confirmed the diagnosis of atypical pityriasis rosea (PR). The patient was treated with topical corticosteroids with a complete remission.

Discussion: We present an original case of TS revealed by a polymorphic dermatosis. Our patient presented 3 major criteria of TS. For the polymorphic rash, it did not match with the spectrum of dermatological manifestations of TS. The diagnosis of subacute cutaneous lupus erythematosus or atypical PR was suspected. Concerning PR, it is a benign dermatosis easily diagnosed. However, some atypical forms may cause potential diagnostic pitfalls, as in our case. To our knowledge, the association PR and TS has never been reported in the literature. Further researches are necessary to shed more light into this association.
Recently, associations of TS with some dermatosis have been reported in the literature: 5 cases of systemic lupus erythematosus and one case of transient bullous pemphigoid in a 28 women affected by TS.