Porokeratosis and Chromosomal Abnormalities

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Sir,

porokeratosis is usually divided in four types: porokeratosis of Mibelli, porokeratosis palmaris plantaris et disseminata, disseminated superficial actinic porokeratosis and linear porokeratosis. They all share the same histological hallmark but differ in the clinical presentation. This distinction however doesn't seem so sharp as unusual presentations [1 , 2] or combination of different variants in the same patient [3] have been recently described.

We would like to convey our experience on this disease describing the case of a patient suffering from both porokeratosis and vitiligo. This patient showed features of both porokeratosis palmaris plantaris et disseminata and disseminated actinic superficial porokeratosis, as many lesions appeared in unprotected vitiliginous areas in association with primary lesions of palms and soles. Moreover we studied clonal chromosome abnormalities in lymphocytes and cultured fibroblasts of unaffected skin from this case and from three sibs (classic type of porokeratosis of Mibelli) and we found that chromosome 3 was preferentially involved in all 4 subjects, with rearrangements mainly located in the region p 12–14 [4]. These breakpoints are consistently involved with epithelial neoplasms [5], which are indeed described as associated with all types of porokeratosis [6–16].

The clinical variety in this disease could therefore be the result of the influence of different agents (actinic rays, PUVA treatment, immuno-suppression and other unknown stimuli) acting on 'genes that, either directly or through the control function they exert, are essential in the proliferation and differentiation of human cells' [5].

In conclusion our clinical and genetic investigations support the unitary vision of this disease stating that 'the similarities of their histopathology and clinical appearance as well as their mode of inheritance make a strong case for considering the various types of porokeratosis different phenotypic expressions of a common genetic aberration' [17].

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Letters to the Editor

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