CONCORDANCE OF MARGINAL KERATODERMA OF THE PALMS AND FAVRE-RACOUCHOT DISEASE IN MZ TWINS

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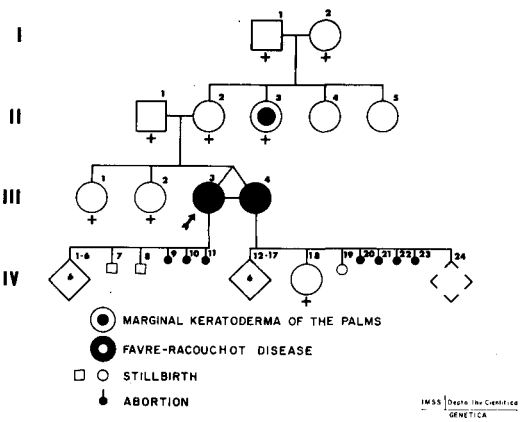
Marginal keratoderma of the palms (MKP, Cozzolino 1951) and Favre-Racouchot disease (FRD, Favre and Racouchot 1951), are mild dermatoses which onset in adulthood. Concordance of both diseases in a set of MZ twins surmising a genetic etiology, is here presented.

CASE REPORT

The proposita (III-3 in the Figure) was 46 years old when first studied. She was a gravida 11, and had had 6 normal children (4 males, 2 females), 2 stillbirths and 3 abortions. The main skin lesions were firm and thick papules following the lateral edges of the fingers, thenar and hypothenar of both hands, preferentially in the union of the dorsal and palmar skin. The papules (1 to 4 mm in size) showed rombic or oval configurations. In the malar regions papules, small cysts and comedos were observed. Microscopic examination of a skin biopsy from a hypothenar region revealed hyperkeratosis of the horny layer and disaggregation of the elastic fibres in the dermis. The malar skin biopsy studies showed poral hyperkeratosis, cysts and disaggregation of the dermal elastic fibers. Laboratory studies including blood cell count, urinalysis, blood and urine chemistries, X-chromatin, and karyotype, were normal or negative.



Figure

Genetic Data (cf. Figure). The proposita's twin (III-4), showed the same clinical and histopathological features. The mother (II-2), deceased at 28 years of age, and the father (II-1), deceased at 40 years of age, were nonconsanguineous and not known to have had similar skin problems. A maternal aunt (II-3), deceased at 60 years of age, was indirectly ascertained to have had the same clinical dermatologic manifestations in the hands.

Monozygosity was established on the basis of physical resemblance, concordance for seven blood group systems (O, ccDEe, MMSS, kk, Jka, Fya, D_1b) and Hp phenotypes, fingerprint analysis ($P_{MZ} = 0.78$, Slater 1963), mixed lymphocyte cultures to evaluate morphological transformation and mitotic index (Bach and Hirschhorn 1964), and a successful reciprocal skin transplantation (performed in 1971).

DISCUSSION

Most MKP reports have been on single cases (Macotela-Ruiz and Pinaud-Bustamante 1969) However, Jung (1973) has found a familial occurrence suggesting autosomal dominant inheritance. Although genetic heterogeneity cannot be excluded, "de novo" mutations, partial penetrance, or the overlooking of minor clinical forms, could explain the sporadic observations. FRD is a mild disorder of unknown etiology apparently more frequent in males than in females (De Graciansky and Boulle 1953). The concordance in MZ twins suggests a genetic etiology. Whether Mendelian or multifactorial, cannot be established by the present observation. Therefore, it is adviceable to be aware of a genetic causal component when studying further cases.

The coexistence of MKP and FRD in a same individual has not so far been reported. However, both dermatoses share common characteristics: Localized, bilateral and symmetrical presentation in exposed areas; onset in adulthood; and a disaggregation of the elastic fibers as a

histopathologic feature. Therefore, it is permissible to suggest a genetic predisposition of the elastic components of the dermis to exogenous factors as the pathogenesis of both diseases.

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