Pachyonychia Congenita Project

15 March 2005

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We hope that making available the relevant information on Pachyonychia Congenita will be a means of furthering research to find effective therapies and a cure for PC.

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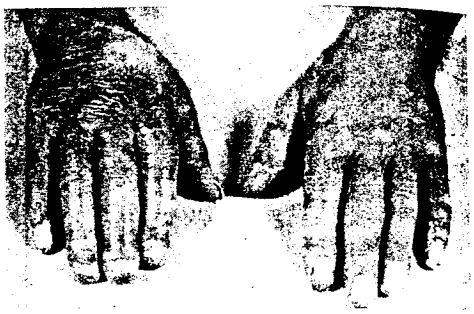

Fig 6.—Clinical aspect of lichen planus in an unsual clinical form on the dorsa of the hands.

Fig 7.—Clinical aspect of lichen planus in an unsual clinical form on the palms.

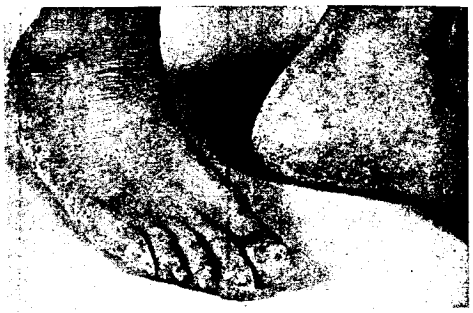

Fig 8.—Clinical aspect of lichen planus in an unsual clinical form on the feet.

aces. The nails of the right hand and some f the toes are dystrophic in the form of yperkeratosis, white-yellow in color and paque.

Histopathologic examination of biopsies rom the palate, the left arm, and right hand Fig 9) revealed alternations of acanthosis nd atrophy, hyperkeratosis, hypergranulo- is, liquefaction of the basal layer, edema nd subepidermal bulla formation, and a ense infiltrate of small round cells in the pper cutis and midcutis.

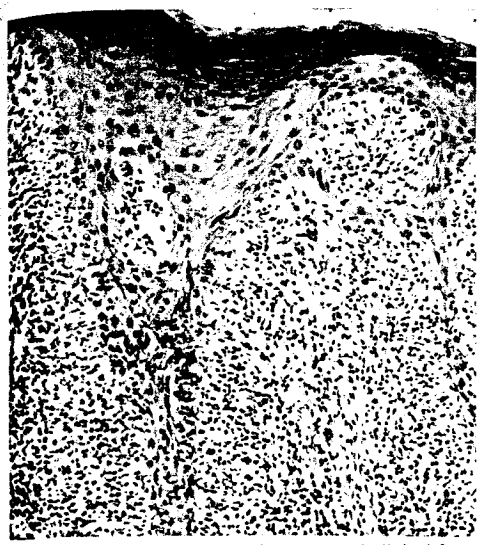

Fig 9.—Histologic aspect of an unusual clinical form of lichen planus.

Fig 10.—Clinical aspect of lichen planus in an unusual clinical form on the left arm.

DR. CHARLES WOLF: The clinical appearance presents so many atypical features that it is almost impossible to make lichen planus out of it.

DR. COHEN: The static course of the condition for six years is as inexplicable as are the clinical lesions. Like many other conditions, clinical variants are what add interest and challenge to medical complacency.

DR. GELLIN: The diagnosis of lichen planus is inescapable from the histopathologic appearance. Six years ago, Dr. Arthur B. Hyman read a biopsy from this patient as bullous lichen planus. The clinical appearance of the hands and feet and the lesion on the left arm (Fig 10) is more suggestive of discoid lupus erythematosis, psoriasis or superficial fungous infection. Only in the mouth is lichen planus more acceptable as a clinical diagnosis. Nevertheless, the histopathology is convincing of lichen planus everywhere.

NEW YORK DERMATOLOGICAL SOCIETY
Jan 24, 1967

Polykeratosis of Touraine. Presented by DR. CARL T. NELSON.

A 14-year-old white girl had a small hemangioma on the forehead at birth, which involuted and disappeared by the fourth month of life. At 6 months, reddened hyperkeratotic patches appeared on the thighs and these were followed by similar involvement of the buttocks, lower legs, feet, arms, and hands. The trunk remained clear. With the passage of time, the involved areas have become less reddened and somewhat less hyperkeratotic. At the age of 11, the hyperkeratotic patches gradually disappeared from the thighs, buttocks, and arms.

Fig 1.—Distribution of symmetrical hyperkeratotic plaques on backs of the legs of patient with polykeratosis of Touraine.

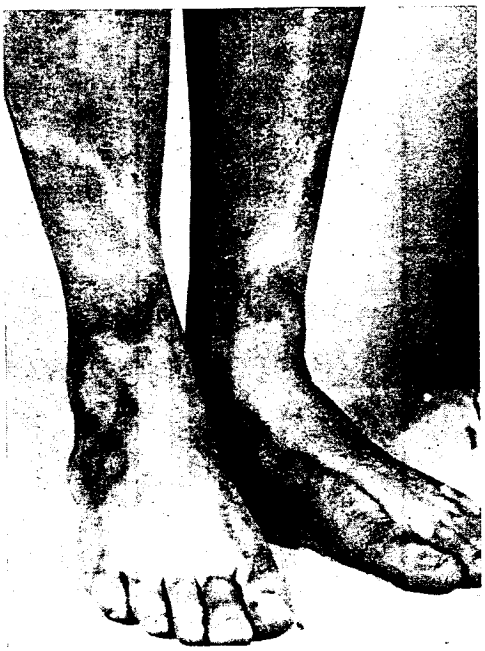

Fig 2.—Distribution of symmetrical hyperkeratotic plaques on legs and feet of patient with polykeratosis of Touraine.

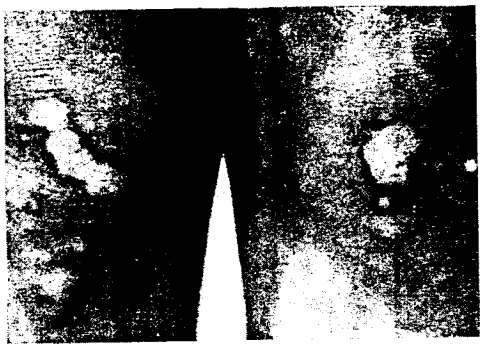

Fig 3.—Distribution of symmetrical hyperkeratotic plaques on knees of patient with polykeratosis of Touraine.

The patient has remained active and healthy with no manifestations of underlying disease.

Description.—The patient now shows symmetrically distributed hyperkeratotic brownish plaques on the dorsa of the feet, lateral surfaces of the lower legs, knees, elbows, and knuckles (Fig 1, 2, 3, and 4). Other parts of the body are clear.

Laboratory Data.—Results of general laboratory studies have been within normal limits. Biopsy in early 1966 showed marked hyperkeratosis with acanthosis in some areas. There was some nonspecific lymphocytic infiltration in the papillary dermis.

Fig 4.—Distribution of symmetrical hyperkeratotic plaques on hands of patient with polykeratosis of Touraine.

Course and Treatment.—Except as noted in the history, the lesions have changed only slightly in recent years. Usually the plaques improve in summer and become more noticeable in winter. Vitamin therapy by mouth and local therapy with keratolytics and sodium chloride cream have not been effective.

Discussion

DR. ORLANDO CANIZARES: This patient is improving slowly. Lesions have disappeared from the thighs and buttocks and in other areas they are less raised and thickened than before. I have the impression that this is a spontaneous recovery and not the result of therapy. We may conclude that the prognosis of this condition is benign.

DR. NELSON: Biopsy of one lesion showed only hyperkeratosis with acanthosis in some areas. There was also some nonspecific lymphocyte infiltration in the upper dermis.

DR. ANTHONY DOMONKOS: The hand lesions are not unlike those of epidermodysplasia verruciformis. Recently, Ruiter demonstrated inclusion bodies by electron microscopy in epidermodysplasia verruciformis. Perhaps that might be done in this case also.

DR. NELSON: Touraine, at the International Congress in Stockholm in 1957, presented a rather lengthy classification of congenital hyperkeratoses. He subsequently published this, slightly modified, in the article cited by Andrews and Domonkos (*Ann Derm Syph* 85:257, 1958). Briefly, Touraine classifies the congenital hyperkeratoses into three main groups. In the first, to which this patient belongs, the abnormality is limited to the process of keratinization. Here, there is simply an overproduction of apparently normal keratin. It is to this group that serodermia and ichthyosis belong, and here, one would also place hyperkeratosis in symmetrical plaques, as well as systematized hyperkeratosis, ie, polykeratosis of Touraine.

The second group includes hyperkeratosis with changes in the dermis, such as vascular hyperplasia or chronic erythema. Congenital ichthyosiform erythroderma and Darier's disease would belong in this category.

The third large group is made up of hyperkeratosis associated with developmental defects in the epidermis. Here one would include familial benign chronic pemphigus (Hailey-Hailey) and even, according to Touraine, such conditions as acanthosis nigricans.

Kyrle's Disease (Hyperkeratosis Follicularis et Parafollicularis in Cutem Penetrans)

Presented by DR. RUDOLF L. BAER.

A negro woman, age 46, five months ago noticed several dark raised lesions on the tibial surface of the right leg. A month later similar lesions were noted on the left leg and subsequently she also developed lesions on the forearms. The lesions were asymptomatic, but more recently some of them have become tender. Some of the lesions have disappeared.

The patient is in good general health, except for diabetes, for which she is receiving tolbutamide (Orinase) and neutral protein Hagedorn (insulin) (NPH). She has no history of nutritional problems or previous dermatoses. Nobody else in her family, to her knowledge, has similar skin changes.

Examination shows lesions on the tibial lateral, and medial aspects of both legs, on the flexor aspects of the wrists, and the adjoining parts of the forearms. The lesions are firm dark brown-black papules, 2 to 3 mm in diameter. They are nontender and some have a scale at the surface. Often these papules form a confluent group 6 to 8 mm in diameter. Many of the papules show a central plug. Where this plug is no longer pres

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Microscopic examination shows several areas of horn plug formation, compressing the surface epidermis and the hair follicle and sweat duct openings. In a few sections, the deeper layers of the epidermis near the horn plugs show rare isolated keratinized cells. At the level of these epidermal alterations, the basal layer is disrupted, the horn plug invading into the superficial dermis. These alterations are not seen in the horn plugs corresponding to the follicle and sweat duct openings. In most of the horn plugs, there are areas of parakeratosis and accumulation of nuclear debris, suggesting a periodic epidermal reaction to the keratinized cells formed in the lower layers. The rest of the epidermis shows moderate acanthosis and elongation of the rete ridges.

The upper dermis shows some increase in capillaries, a few of them dilated, and a moderate amount of perivascular inflammatory infiltrate predominantly in the areas of epidermal alterations. The inflammatory infiltrate consists of lymphocytes, histiocytes, very rare polymorphonuclear leukocytes, a few macrophages containing hemosiderin and a few melanophages. In the areas where the epidermis is disrupted, there is fragmentation of the collagen fibers. The elastic tissue appears normal and there are no amyloid deposits.

Discussion

Dr. Lewis Shapiro: Histologically, there was no actual penetration of the keratin plug into the dermis, but I do not think this rules out Kyrle's disease, since a few sections taken from one lesion may simply not show an area of penetration. Many of the published cases have not been accepted by other authors and the absolute histopathologic criterion of penetration and clinical correspondence to what Kyrle described may be interpreted more or less strictly by different individuals. They may not see penetration in one section of one biopsy; how closely must a particular case agree with what Kyrle published? I have looked at Kyrle's article, and I think this is a very good resemblance. Therefore, I accept the presenter's diagnosis.

Dr. Perry Sachs: This is a case of Kyrle's