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Clinical Records

Otological lesions in pachyonychia congenita syndrome

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Abstract
The authors report a case of a patient with pachyonychia congenita syndrome, a rare genodermatosis inherited as an autosomal dominant trait, who also had otological lesions beyond the other classic signs and symptoms of the syndrome. Many kinds of treatment have already been proposed, but all failed to show satisfactory results. A new, cheap and easy-to-use treatment was developed in this study, using keratoplastics interpolated with humectant lotion for 90 days. The results after three years of follow-up are still thoroughly satisfactory.

Key words: Ear, external; Keratosis.

Introduction

Pachyonychia congenita is a rare genodermatosis, inherited as an autosomal dominant trait with variable expressivity and high penetrance, that affects both sexes. The first cases of pachyonychia congenita were described by Muller (1904) and Wilson (1905). The literature reveals 168 cases since its first description in 1904 up to 1985. Numerous subdivisions of pachyonychia congenita have been suggested. The following classification, proposed by Feinstein in 1988 is based on a retrospective survey of 168 cases of pachyonychia congenita:

Type I – hypertrophy of nails, palmoplantar hyperkeratosis, follicular keratosis and oral leucokeratosis (56.2 per cent).

Type II – clinical findings of type I plus bullae and soles, hyperhydrosis of palms and soles, natal or neonatal teeth and steatocystoma multiplex (24.9 per cent).

Type III – clinical findings of type I and II plus angular cheilosis, corneal dyskeratosis and cataracts (11.7 per cent).

Type IV – clinical findings of type I, II and III plus laryngeal lesions, hoarseness, mental retardation, hair anomalities and alopecia (7.2 per cent).

Among concomitant diseases the most common is steatocystoma multiplex. Other diseases associated with the syndrome are vitiligo, depression, schizophrenia, systemic infection with herpes simplex, migraine headaches, diabetes mellitus, epilepsy, polydactyly and microcephaly. There has been no report in the literature about otological skin lesions until the present.

In this paper, the authors report a case of a patient with pachyonychia congenita that presented with otological lesions. A new modality of treatment is described, which showed good results quickly and for a long period of time.

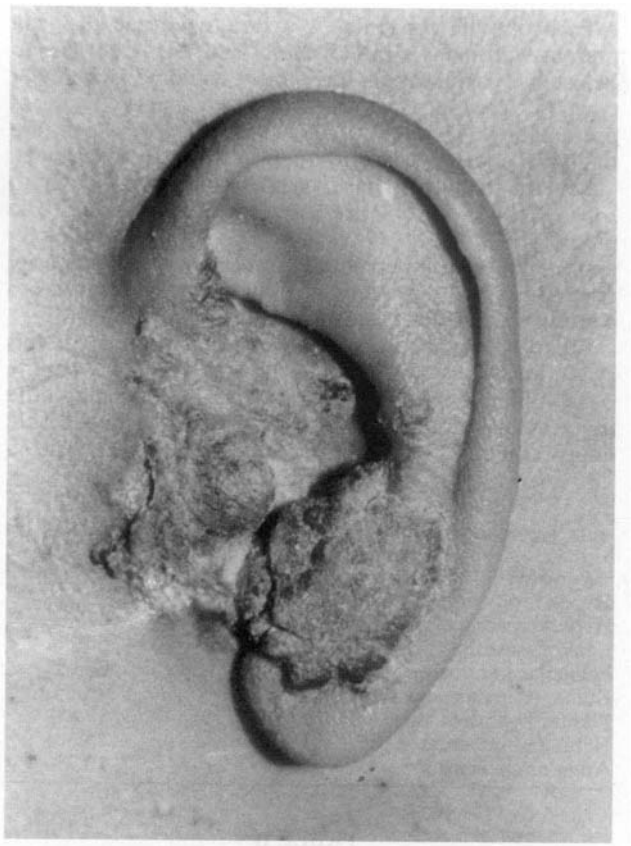

FIG. 1
Otological lesions in a patient with pachyonychia congenita syndrome before treatment.

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Case report

A 40-year-old man presented in our Department with a history of progressive hearing loss for the last four years. His general examination revealed thickened and distorted fingernails and toenails, palmoplantar hyperkeratosis, alopecia, hyperhydrosis, follicular keratosis, cataracts and diabetes. When this patient was born, he presented natal teeth. Throughout his life he had been submitted to several surgical procedures on his hands and feet progressing to extremities deformities.

The lesions that affected the face were located in the nasal dorsum, surrounding the mouth, both ears and external auditory canals. The lesions surrounding the mouth, compromised the speech and the mastication by limiting muscles movements related to these functions. The lesions on the external ear and in the external auditory canal closed the cartilaginous part of the external auditory canal (Figure 1) and led to an auditory dysfunction. These lesions also resulted in aesthetic and social problems.

The histological study of the lesion presented in the external auditory canal was follicular keratosis with hyperkeratosis that blocked the openings of the hair follicles. The same pattern was observed in compromised skin of this patient. The lesion in the external auditory canal was similar to the lesions on the dorsum of the nose and surrounding the mouth.

Discussion

Pachyonychia congenita is a rare syndrome that can present with many signs (Tables I and II). According to Feinstein's classification, this patient could be included in type IV group, which is the less common form of presentation (Feinstein, 1988). The focused lesions are the facial ones, especially those on the external ear canal.

Several treatments have been formulated in order to solve the skin lesions presented in this syndrome. The modalities with X-ray radium irradiation, ultra-violet application and ammoniated mercury had no beneficial results. The reports with the use of local steroid creams are controversial (Kumer and Loos, 1935; Forslind, 1973).

Some authors recognise the use of oral etretinate in this syndrome (Feinstein, 1988). Although its effect on the facial and the external auditory canal lesions is of little significance, the histological work-up showed remarkable improvement. This kind of treatment has its use restricted by the high cost, important collateral effects and the quick return of the lesions after discontinuing the medication.

TABLE I

MAIN PATHOLOGIC SIGNS OF PACHYONYCHIA CONGENITA AND ITS FREQUENCY OF APPEARANCE

Hypertrophy and distortion of nails	100%
Hyperkeratosis of palms and soles	62%
Leukokeratosis	60.2%
Follicular keratosis	36.9%
Bullae on palms and soles	36.1%
Plantar hyperhydrosis	19.8%
Natal and neonatal teeth	15.6%
Angular cheilosis	10.7%

TABLE II

LESS COMMON SIGNS OF PACHYONYCHIA CONGENITA AND ITS FREQUENCY OF APPEARANCE

Hair anomalies	9.6%
Corneal dyskeratosis	7.8%
Hoarseness	6.6%
Cataracts	6.0%
Mental retardation	4.2%
Alopecia	2.4%

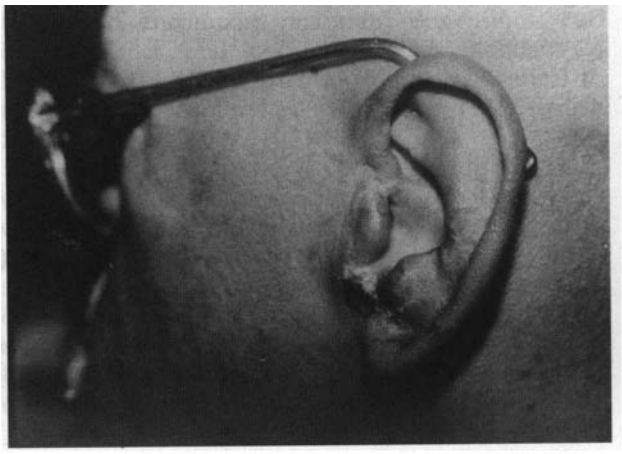

FIG. 2

The same patient after three months of treatment.

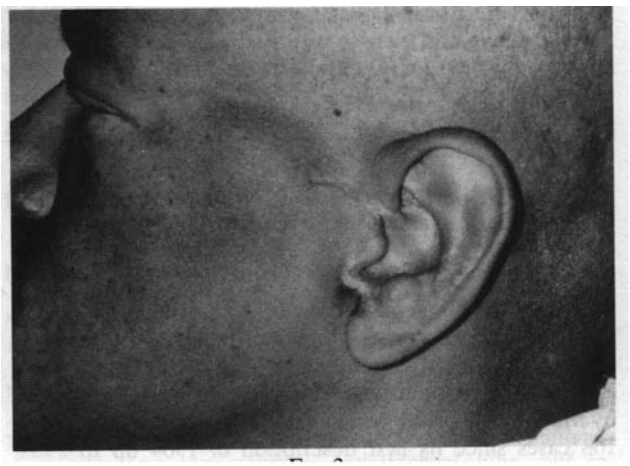

FIG. 3

The same patient after one year of follow-up, showing no reoccurrence of otological lesions.

The previous treatment of the lesions on the external ear canal for 20 years had been only their mechanical removal. Although recovering audition, as confirmed by audiometry, it was a painful process with re-establishment of 70 per cent of the lesions in a short period of time (seven days). In view of this rapid recurrence a new kind of treatment was imperative.

The instituted therapy, which lasted 90 days, was the local use of salicylic acid 0.5 per cent (keratoplastic) in propylene glycol (emollient solution) once every two days, interpolated with the use of humectant lotion. The salicylic acid acts in the desquamation process by solubilization of the intercellular cement present in the corneal layer. The action of the propylene glycol simultaneously softens the skin and provides, on the corneal layer, an oil film that avoids the loss of water by evaporation, then dissolving from the surface to the adjacent layers of the skin.

The results with the use of keratoplastics in emollient solution were the mechanical removal of the lesions and re-establishment of the keratinization process, which was previously altered. This is a painless process with quick results. Within a week favourable results were observed. There was new epithelial formation and the lesions have not reappeared during 24 months, which was the observation period (Figures 2 and 3). In this concentration there is no harm to the tympanic membrane.

Local keratoplastics have no collateral effects and

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achieve good and persistent results within a short period of time. Their low cost and easy use make them the best choice of treatment for hyperkeratosis.

Conclusion

The use of local keratoplastics in emollient solution interpolated with use of humectant lotion in the follicular keratosis present in the pachyonychia congenita syndrome produces good and persistent results. This treatment has no collateral effects. Because of its low cost and easy use, it is now the best choice of treatment for hyperkeratosis.

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