DERMABRASION AND RETINOIC ACID IN THE TREATMENT OF PACHYONYCHIA CONGENITA

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Summary

A typical case of pachyonychia congenita in a female is presented. Of the sixty cases of pachyonychia congenita reported in the literature, only one case is a female Cases have been seen in families upto five generations. In our case five family members belonging to two generations manifested the disease. The hyperkeratotic plantar lesions were painful and incapacitating to the patient. With combined dermabrasion and topical retinoic acid the patient was made rapidly ambulatory. This new modality of treatment seemed gratifying in that the symptomatic relief to the patient was excellent.

Pachyonychia congenita is an unusual congenital and sometimes familial malady characterised by dystrophic changes in nails, hyperkeratosis of palms and soles, follicular keratoses, especially about knees and elbows, hyperhidrosis of palms and soles, and oral leukoplakic lesions.

The disorder was first reported by Jadassohn and Lewandowsky in 1910.[1] Sixty cases have been reported in the literature. Jackson and Lawler[2] reported six cases in one family. Kumer and Loos[3] described 23 cases of pachyonychia congenita in a five generation family comprising 70 members. Bhakta Viziam[4] reported this condition in 5 of 10 children of consanguinous parents. The malady is of dominant heredity transmitted probably as a single autosomal dominant gene. We are reporting a typical case of pachyonychia congenita in a female. In her family, five members in two generations showed manifestations of the disease.

Paper read at V Annual Conference I.A.D.V.L. January, 1977, Ahmedabad.

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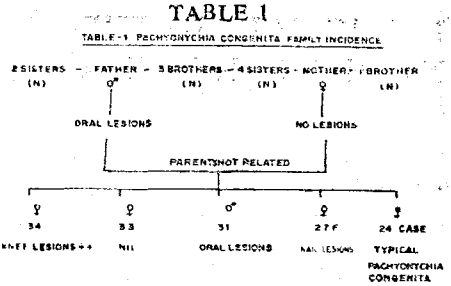
Received for publication on 13—6—1977.

Case Report

A 22 year old female patient attended the O.P.D. of St. George's Hospital, Bombay on 1-4-1974, with the complaints of painful lesions over the soles-deformed nails, stomatitis and diffuse alopecia. She was the 5th and youngest child of non-consanguinous parents and a normal F.T.N.D. child. According to the mother she noticed at birth that the nails of the child were tubular, long and deformed. Verrucous lesions on the lips were noticed at the age of 2 years and thickening of the skin over palms and soles at $4\frac{1}{2}$ years. Mile stones were normal. Patient was of averege intelligence having studied upto S.S.C. Personal history was not contributory. There was history of cervical lymph-adenopathy in the past probably of tuberculous etiology for which patient was treated with 30 injections of streptomycin and I.N.H. tablets.

Family History

TABLE 1



The parents were not related. Father had 2 sisters and 3 brothers. Father showed mild stomatitis. No one else had any manifestations of the malady. Mother had 4 sisters and 1 brother. No lesions were seen in the mother or her family. Patient had 3 sisters and one brother. The eldest sister showed papular lesions on her knees, the second sister had no lesions, the third, her brother showed mild stomatitis and the fourth, another sister showed early nail changes. Patient was the fifth sibling and showed typical manifestations of pachyonychia congenita.

On examination, patient was fairly built and nourished. She had diffuse alopecia of the scalp (Fig 1) which had started on 1974 and gradually progressed so that by 1977 the occipital area had become totally bald. The lips were hyperkeratotic, scaly and at places ulcerative and the tongue magenta coloured with white streaks and opaque plaques (Fig 2). Similar changes were seen on the buccal mucosa. There was no hoarseness of voice, eye or teeth changes. The nails of the fingers and toes were affected and appeared greatly thickened, opaque, lustreless and folded longitudinally (Fig 3). Besides, they were narrow, laterally curved and distally elevated. The nail plates were thick and wedge shaped. The distal edges of the nail plates were several mm thick and the free edge of the nails raised by a thick horny mass of subungual keratosis. No nails were lost.

Patient also showed acneform and follicular lesions on elbows and knees. They were thick, scaly, gray and consisted of erythematous papules with central horny plugs. Occasional verrucous lesions were seen on elbows. No sebaceous cysts were seen. Palms and soles showed gross hyperkeratosis and verrucous lesions (Fig. 3). The verrucous lesions especially on the soles were painful, making walking difficult if not impossible. Thus patient was crippled and almost bedridden. Hyperhidrosis and bullae were absent in this patient. Systemic examination did not reveal any gross abnormality.

Investigations

Routine urine, stools, Hb, WBC, T.D. blood urea and blood VDRL showed no abnormalities. ESR was raised being 115 mm./1 hour and fasting blood sugar was 118 mg%.

Histopathology

Histopathology of the skin showed hyperkeratosis with areas of parakeratosis and occasional follicular plugging. Hypergranulosis and acanthosis were evident with areas of dyskeratosis. The dermis showed mild inflammatory reaction. Mucous membrane biopsy was not done because patient was not cooperative.

Management

In the past, this patient had been treated extensively with high doses of Vitamin A, Vitamin E, B Complex factors and multivitamins, but with negligible response. The nails were cut short after immersing them in KOH. Futile attempts had been made to reduce the hyperkeratotic lesions by application of salicylic acid (5% – 20%). We therefore decided to remove the hyperkeratotic lesions by dermabrasion[5] and follow it up with topical application of retinoic acid.

Method

Patient was given a short acting general anesthesia and the hyperkeratotic lesions were excised. Skin over

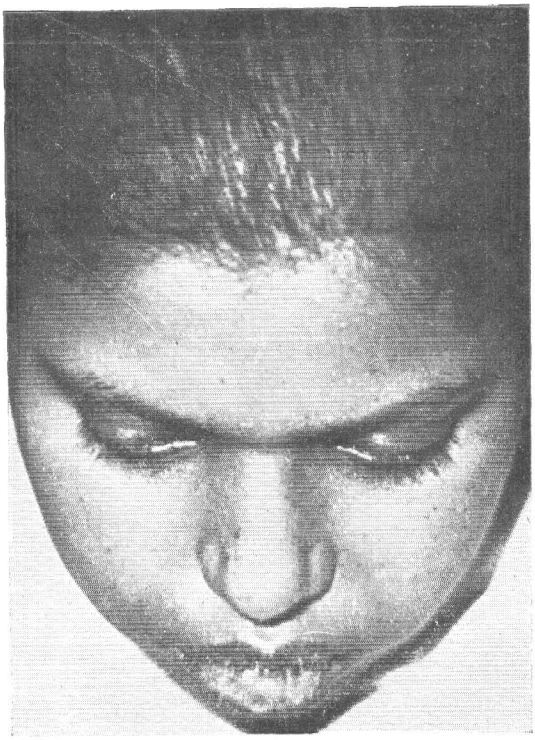


Fig. 1 Showing difference alopecia

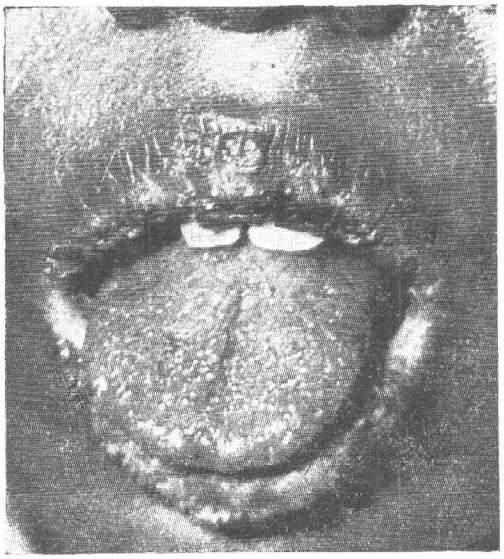


Fig. 2 Showing lips and tonque before treatment

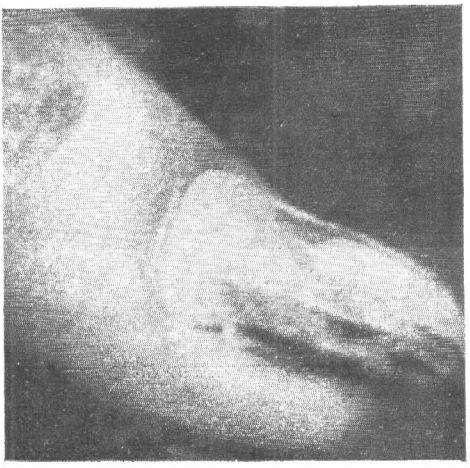


Fig. 3 Showing the nail

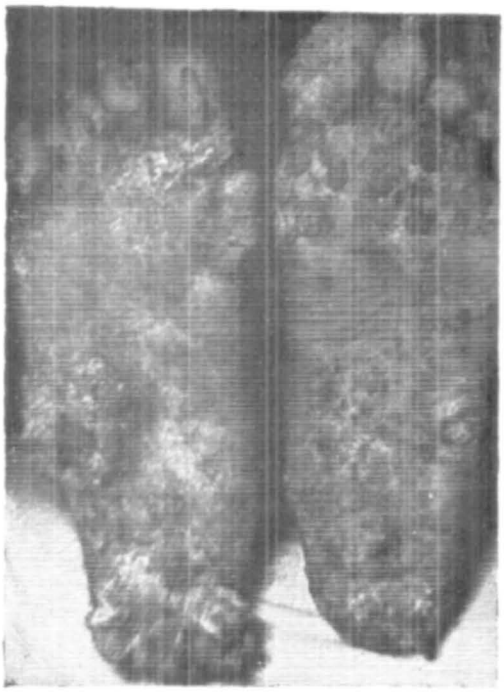


Fig. 4 Showing painful verrucous lesions on soles before treatment

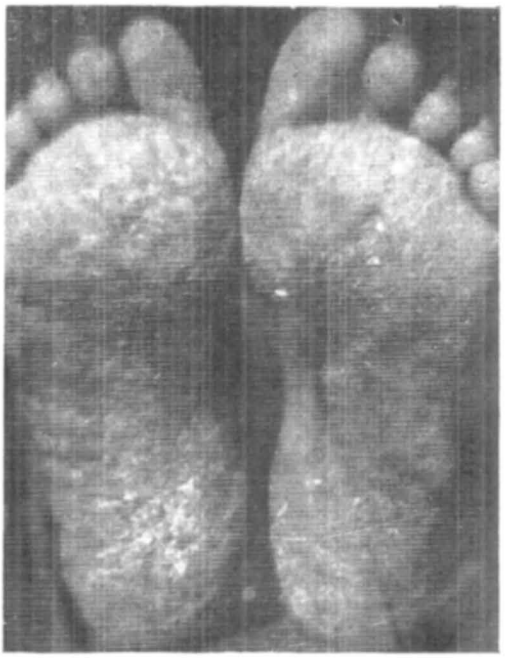


Fig. 5 Showing healed lesions on soles after dermabrasion and retinoic acid application

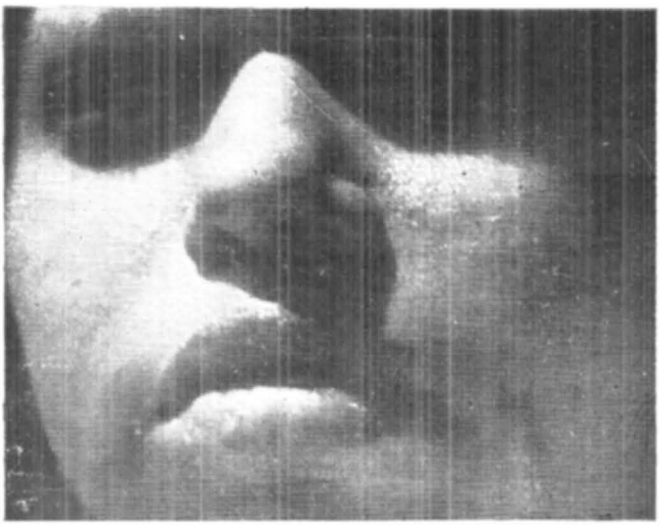


Fig. 6 Showing healed lesions on the lips following application of retinoic acid 0.05%

the palms and soles was dermabraded and made smooth and plain. Antibiotic dressings were applied and continued upto a week. When the lesions healed, topical retinoic acid 0.1% was applied on palms and soles twice daily, and 0.05% on lips. Patient made a rapid recovery and was able to walk about freely (Fig No. 4). Pain subsided completely. Patient was followed up for six weeks after operation with application of retinoic acid. No new hyperkeratotic lesions developed during this period. (Fig No. 5.)

Discussion

Of the sixty cases of pachyonychia congenita reported so far, one was a female. This rarity in females prompted us to report this case. We were able to detect involvement in the father who showed stomatitis and oral lesions. Patient's brother and two sisters showed minimal changes of pachyonychia congenita. The patient herself showed all the features of pachyonychia congenita except eye changes, bullae and hyperhidrosis. There was no loss of any nail.

Pachyonychia congenita has to be differentiated from dyskeratosis congenita. Table 2 shows the characteristic differentiation.

Some workers however believe that both maladies are variations of the same disorder. The disease is not lethal but it makes life miserable. The hyperkeratotic lesions on soles make walking difficult. Stomatitis and leukoplakic lesions make eating difficult, and most of the time patient has to be on a bland diet. Loss of hair, deformed nails and papular lesions on body disfigures the patient who is thus likely to develop psychic complexes. Corneal dyskeratosis may result in opacities and partial blindness.

Management of these cases is generally symptomatic and treatment has to be persistent. Oral vitamins and

TABLE 2

Pachyonychia Congenita and Dyskeratosis Congenita – Characteristics

	Dyskeratosis congenita	Pachyonychia congenita
Inheritance	Autosomal or sex linked recessive	Autosomal dominant
Onset	During 1st 2 years of life	Nail changes at birth others 1st 2 years of life
Pigmentation	Fine maculopapular linear reticular lace-like gray to brown	No pigmentation
Nails	Dystrophic or absent	Greatly thickened
Verrucose lesions	None	Elbows knees popliteal areas, legs, ankles.
Leukoplakia	Pronounced and extensive in mouth and anus : Pre-malignant	Mild and not extensive not premalignant
Eyes	Atrophy of tear ducts ; ectropia with bullous conjuctivitis at times	Corneal dyskeratosis and cataracts leading to blindness at times
Complications	Squamous cell carcinoma of mouth, anus, rectum and leukemia when associated with Fanconi's disease	None

Common factors : More in males, hyperhidrosis, bullae, hyperkeratoses, teeth changes and thinning of hair or alopecia

B. complex factors play doubtful role. Major efforts should be made to reduce the painful, thickened and isolated keratosis of soles. Specially made rubber based foot moulds and shoes have been employed for such lesions[6]. Hypnotherapy has been reported to produce marked improvement of the plantar hyperkeratosis.

Treatment of nails poses another problem. The nail plate being hard is difficult to cut. Therefore they are immersed in KOH to make them soft before cutting. Salicylic acid 2–20 % application is futile. Surgical removal of nails together with scraping of matrix to prevent regrowth has been frequently employed. The disability produced by pachyonychia is so great that amputation of distal phalanges have been advocated.

Vitamin A in high doses is frequently given to reduce follicular lesions and hyperkeratosis. The crusted bullous lesions are treated with compresses, soaks, gentian violet, icthammol ointment and wet dressings with cysteine hydrochloride.

It was our impression that this new modality in treatment i.e., dermabrasion and retinoic acid combination in the treatment of pachyonychia congenita served well in making the patient ambulatory. The result of treatment was gratifying and excellent.

Acknowledgment

We thank the Superintendent, St. George's Hospital, Bombay for allowing us to use the Hospital records. We are indebted to Ethnore Ltd., Bombay for the liberal supply of retinoic cream.

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