PACHYONYCHIA CONGENITA WITH ABNORMALITIES OF THE HAIR

(Case report)

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Summary

Two cases of Pachyonychia Congenita with hair abnormalities are reported. The family tree analysis shows an autosomal recessive mode of transmission. These cases are reported because of their rarity and certain unsual features.

Pachyonychia congenita is an unusual genodermatosis characterised by extensive hyperkeratotic abnormalities of the nails, skin, mucous membrane and hair. This syndrome was originally described by Jadassohn and Lewandowsky in 1910[1]. The first reported patient was a 15 year old girl who had nail changes from birth and callosities of the soles associated with painful bullae during summer months. In addition, there was hyperkeratosis of the palms, soles and nose and bright red papules with central follicular plugs on the elbow, knees, chin and tip of the nose. Her tongue was coated with irregular leukoplakic lesions. A younger brother exhibited only reddish papules and follicular plugging on the knees and elbows. Since then about 60 cases have been reported in the literature[1]. Epidermal cysts and abnormalities of the hairs and premature eruption of the teeth have been reported in some cases[2,3].

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Case Reports

A girl aged 16 years had been attending the hospital since 1969 for recurrent vesiculo bullous lesion on various parts of the body. On examination she was found to be an ill nourished girl with scattered secondarily infected bullous lesions – mainly affecting the limbs especially the palms and soles. The palms and soles in addition showed circumscribed symmetrical hyperkeratoses with normal looking intervening skin. (Fig. a & b Page No. 177). The extensor surfaces of the knees, elbows forearms and back of neck showed follicular papules with keratotic plugging. The skin was generally dry and scaly. Hyperhidrosis was not present. Nails of both hands and feet showed the following changes. The proximal portion of the nails looked normal. In some of the nails the distal portions were arched transversely and pushed upwards from their beds by hard adherent, keratinous material. The nails were firmly attached to the nail beds. The nail plate at the free edge was thickened and hard protruding beyond the tip of the digits, some of them

arching upwards and some backwards. The portion of the nail plate which was raised from its bed was opaque and lustreless.

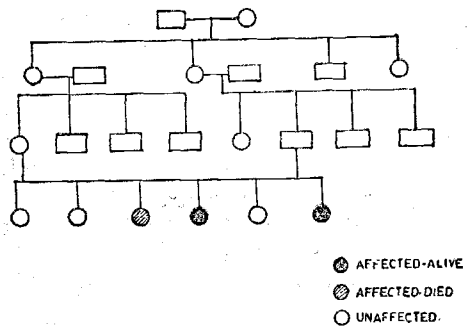
The hair on the scalp was sparse, thin, kinky and negroid (Fig. Page No. 177). There was no lesion on the mucous membrane of the oral cavity. There were no dystrophic changes in the teeth, and the eyes were normal. Intelligence was normal. There was no bony or joint involvement. Case 2 younger sister of case 1 aged 12 years presented a very similar picture.

Investigations

urine — N. A. D

Case 1. Hb. 10.5 Gms, T. L. C. 16000/cmm. D. L. C. P58 L30 E12.

Case 2. Hb. 11 Gms, T. L. C. 14500/cmm. D. L. C. P60 L24 E16.



Family tree – suggestive of a recessive mode of inheretance.

Family history: These patients were the children of consanguinous parents. One elder sister had similar features but died at the age of 4 months. Three other sisters were healthy and unaffected (family tree).

Discussion

The above clinical features justify the diagnosis of Pachyonychia congenita. In these cases there was no involvement of the mucous membrane. Involvement of the mucous membrane though a feature of the syndrome need not be present in all cases[4]. Although the first reported case was a female this syndrome predominently affects the males. In our cases all the affected children were females. The hairs in our cases were thin, kinky and negroid. Similar hair abnormalities have been reported [2,3]. This syndrome is accepted to be transmitted as a dominant trait, but in the present cases there was a history of consanguinity in the parents and the parents or their siblings were not affected. This may suggest an autosomal recessive inheritance.

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