PACHYONYCHIA CONGENITA AND MENTAL DEFICIENCY

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Pachyonychia congenita was seen in two different families. In one family the disease was present in only one child, while in the other family the disease was traceable in 5 generations involving 36 members. One individual had associated mental retardation.

Key words : Pachyonychia congenita, Mental deficiency.

Pachyonychia congenita is a distinct, inherited, predominantly muco-cutaneous disorder with occasional affection of other organs. This uncommon entity was first described by Jadassohn and Lewandowsky in 1906[1] and subsequent reports discussed below have established the familial nature of the condition. We describe two families, one with a single case probably resulting from mutation, while the other family had almost complete penetrance running for 5 generations. Thirty six members in the family were affected including a girl with mental retardation.

Case Reports

Case 1

An 8-year-old boy born after a full-term normal delivery of unrelated parents, had palmo-plantar keratoderma and nail dystrophy since birth. One month after birth there was some thickening of the nails over the right hand and by the age of 6 months this progressed to involve all the other nails. This was followed by 2 to 3 episodes of bullous lesions over the palms and soles which disappeared after the first year. Palmo-plantar keratoderma set in at the age of 2 years. Family history of a similar disorder was entirely negative. All the 20 nails were yellowish brown and thickened, being most marked distally. Scaly, keratinous plaques were seen over the heels and palmar prominences. Grouped horny papules were present over the knees, elbows and buttocks. Oral mucous membrane was normal. Milestones of development and eruption of teeth occurred at normal intervals. Hairs were normal. Systemic examination revealed no abnormality.

Case 2

A 58-year-old woman had thickening of all the nails and palmo-plantar keratoses. Nail changes had commenced a week after birth, involving all the nails by 8 months of age. Keratoderma set in at the age of 4 years. All the nails were discoloured and showed thickening which was maximal towards the free end. Circumscribed keratotic plaques were seen over the pressure-bearing surfaces of the palms and soles including the volar aspects of some of the fingers. Repeated maceration during house work gave way to frequent bouts of paronychia affecting predominantly the fingers of the right hand. There was no history of palmo-plantar hyperhidrosis, bullous eruptions, mucosal lesions or premature eruption of teeth. Hairs and eyes were unaffected. Family history (Fig. 1) revealed 36 affected persons and none

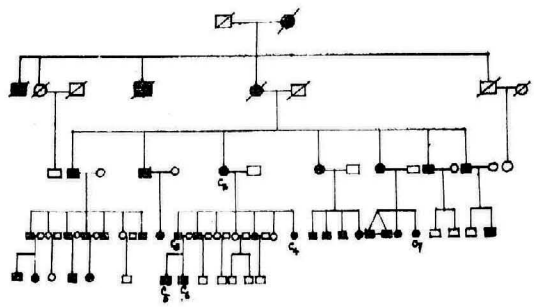

Fig. 1. The family tree.

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of the marriages were consanguineous. Since most of the people had settled elsewhere, only some members of the family could be examined. The details of others were compiled after thorough interrogation and scrutiny of available family photographs. The palms, soles and nails were affected in all the patients with the exception of one child (case 6). Noteworthy was the fact that out of all the affected children of the same parents in the 4th generation, 2 were monozygous twins and the youngest aged 13 years (case 7) had associated mental retardation; hospital records obtained from the parents revealed that this child examined a year ago was a FTND with delayed mile-stones of development and an IQ of 33 (Wechsler intelligence scale for children).

Comments

An extensive study[2] has revealed the lesions in pachyonychia congenita to be pachyonychia (96.8%), palmo-plantar keratoses (72%), follicular acneform keratoses (58.5%), and oral leukokeratoses (54.4%). Other manifestations include natal eruption of teeth, steatocystomas, epidermal cysts, hyperpigmentation, abnormal hair, corneal dystrophy, hoarseness of voice and a thickened tympanic membrane, all of which have led to a working classification of pachyonychia congenita into 3 types;[3-8] some have classified it into 2 distinct patterns[9,10] but uncertainty still lingers around the meaningful separation of pachyonychia congenita into the different varieties. Association of pachyonychia congenita with mental deficiency as seen in our patient has not been observed previously; it further supports the ectodermal nature of the defect as suggested earlier.[4]

Electron-microscopic studies have revealed abnormal keratinisation[11] which is seen under the light microscope as progressively increasing areas of perinuclear vacuolisation starting from the lower epidermis.[7,12] Genealogical observations[7,8,13-15] support the autosomal dominant nature of the defect, with age-dependent expressivity of some of the manifestations. Nevertheless, rare instances of autosomal recessive inheritance of this condition [16,17] and isolated case reports with an entirely negative family history have been recorded.[11,18-20]

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