PACHYONYCHIA CONGENITA

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Pachyonychia congenita is an infrequent genodermatosis, characterised by nail dystrophy, hyperkeratosis of the palms and soles, follicular keratosis and leukoplakia. It was seen in two male patients aged 12 and 35 years respectively. The younger patient had nail changes, palmo-plantar keratoderma, eye changes, hypotrichosis and mental retardation, while the elder one had minimal nail changes, keratoderma and leukoplakia.

Key words : Pachyonychia congenita

Jadassohn and Lewandowsky reported the first case of pachyonychia congenita (PC). Since then, approximately 100 cases have been reported[1] including a few cases from India.[2-5] Apart from the classical presentation, abnormalities like neonatal teeth, epidermal inclusion cysts, hoarseness of voice, hypotrichosis, micro-ophthalmos, cataract formation, hyperhidrosis and bony abnormalities have been described.

Case Reports

Case 1

A 12-year-old male child, born of a non-consanguineous marriage presented with large, painful horny growth of nails, and callosities on the palms and soles. The nail changes started in the second week of life, while the callosities on the palms and soles were present since the age of 2 years. The nails were extremely hard, greatly thickened, opaque, brown, lusterless, laterally curved and distally elevated. The free edge of the nail was raised by a thick horny mass of subungual keratosis. Paronychial inflammation was also present. Along with the nail changes, the patient had hypotrichosis, and plamo-plantar keratoderma which was more marked on the pressure areas. The eye changes consisted of micro-ophthalmos with bilateral horizontal nystagmus. The patient was mentally retraded with an IQ of 65.

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Case 2

A 35-year-old muslim male patient, born of a non-consanguineous marriage, had a painful palmo-plantar keratoderma that affected his gait. The finger nails were only slightly thickened while the toe nails showed the characteristic changes along with multiple, small, follicular, keratotic lesions on the extremities and extensive oral leukoplakia. The painful skin lesions from the sole were excised, and a full thickness graft was used to cover the area. The grafted area showed no hyperkeratosis, but 6 months later, developed a peripheral hyperkeratotic band which was treated with keratolytics. The pain on walking was reduced by the use of special footwear (MCR foot wear used by leprosy patients).

Comments

Pachyonychia congenita (PC) is an autosomal dominant genodermatosis,[6] though, Haber and Rose[7] consider it to be autosomal recessive. Several family studies have been reported.[4-5-7] Schonfeld[8] divided the PC syndrome into three types : Type I (Jadassohn-Lewandowsky syndrome) which consists of, (a) Symmetrical, hard, thickening of all finger and toe nails, (b) Keratosis palmaris et plantaris, (c) Palmar and plantar hyperhidrosis, (d) Follicular keratosis, especially on the knees and elbows, (e) Blister formation, especially under and around the callosities, (f) Leucokeratosis of oral mucosa, and occasionally of the laryngeal mucosa which can produce hoarseness, and (g) Hair abnormality. Type 2 (Jackson-Sertoli syndrome) in which in addition to type I changes,

neonatal teeth and multiple epidermoid cysts are seen, but leucokeratosis of oral mucosa is not seen. Type 3 (Schafer-Brunauer syndrome) which is like type 1, with associated leucokeratosis of the cornea.

Most of the cases so far reported had constant involvement of the nails. Pinkus[9] had described these nail changes as resembling a miniature horse's hoof. One of our patients (case 2) had a minimal involvement of the nails. A similar observation was made by Schonfeld.[8] The exact site of nail pathology in PC is not yet well-defined. According to Pinkus and Mehregan,[9] nail plate and proximal nail matrix are hyperplastic and papillomatous, producing a large quantity of abnormal horny material with hyaline masses.

Our other case (case 1) had associated mental retardation, which has not been reported so far. He had small, dry, lusterless brittle scalp hair and loss of eye-brows. Rook[6] has recorded hypotrichosis with a complete loss of hair in the occipital region.

Extensive benign oral leukoplakia was seen in one patient. Carl et al[10] described leukoplakia in four hereditary mucosal syndromes involving multiple sites (Darier's disease, hereditary benign intra-epithelial dyskeratosis, white sponge nevus and PC).

Many treatment regimes have been tried in PC with a varying success, like surgical removal of the nail[11] or removal of nail with skin grafting.[12] In case 2, skin grafting was done for the painful plantar lesion. Though new lesions did not appear at the site of graft, a band type hyperkeratosis appeared around it. Jagavkar et al[4] removed palmo-plantar keratosis with dermabrasion and applied topical retinoic acid with beneficial effect.

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