Pictorial CME

Pachyonychia congenita (Three cases in a family)

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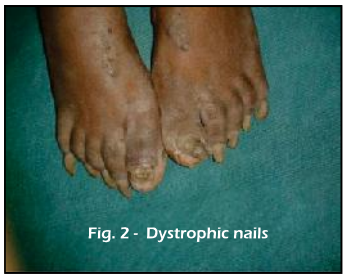
Fig. 2 - Dystrophic nails

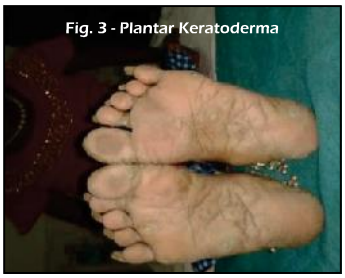
Fig. 3 - Plantar Keratoderma


Fig. 4 - Follicular Kertosis on elbows

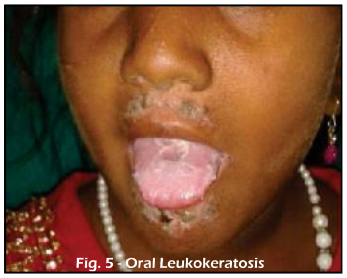
Fig. 5 - Oral Leukokeratosis


Fig. 1 - Thickened curved discovered nails

A 5yrs old female child born of a consanguineous marriage presented with finger and toe nails abnormalities since birth along with oral and multiple skin lesions. Her mother and brother had similar complaints. On examination all the fingers and toe nails were dystrophic. The nails were yellowish, thickened, curved with gross subungual hyperkeratosis causing upward angulation of the distal end (Fig 1 and 2). She had palmo- plantar keratoderma with thick plaques in areas of greatest friction (Fig 3). Multiple bilateral keratotic papules were present on elbows ,wrists, buttocks .(fig 4).Oral examination revealed white patches on dorsum of tongue (fig 5) .Examination of mother and brother revealed similar cutaneous findings The clinical findings of dystrophic nails, keratosis pilaris, palmo-plantar keratoderma, leukokeratosis, with a positive family history was consistent with the diagnosis of pachyonychia congenita type 1(jadassohn and lewandowsky).

Pachyonychia congenita (PC) described by jadassohn and lewandowsky in 1906 is a rare autosomal dominant disorder with predominant involvement of nails and palmo-plantar skin. The two main clinical variants of PC are PC 1(Jadassohn –lewandowsky) type and PC 2(Jackson –lawler type.).These variants of PC arise through mutations affected by genes encoding keratins 6a and 16 in PC 1 and 6b and 17 in PC 2.[1] Hypertrophic nail dystrophy is the most prominent and earliest clinical feature of both the types of PC present usually at birth. The nail changes in PC consist of three abnormal findings hyperkeratosis of nail bed, thickening of nail plate and distortion of curvature of nail plate[2] The nails are discoloured, thickened and firmly attached to nail bed. The nail bed is filled with horny yellow keratotic debris which may cause the nail to project upwards at the free edge.[3] Both the types of pachyonychia congenita are associated with variable degree of palmo-plantar keratoderma and follicular keratosis of elbows, knees and hips.Palmo-plantar keratoderma in PC is hard, non erythematous keratoderma that is accentuated in pressure points of the feet or in areas of chronic use of the hands. The clinical discrimination between the PC-1and PC-2 depends on more prominent oral leukokeratosis in PC-1 or findings of steatocytomas, pilosebaceous cysts, vellus hair cysts ,hair abnormalities (alopecia, pili torti) and natal teeth in PC-2.[2] Two more variants of PC have been described. Type -3 (schafter – branauer) is like type 1 with the addition of leukokeratosis of the cornea.[3] Late onset PC is known as pachyonychia congenita tarda.

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3) Genodermatosis and congenital anomalies.In: James JD, Berger TG, Elston DM, editors. Andrews Diseases of the Skin.Clinical Dermatology.11 ed.Saunders Elsevier, 2011, pg -561