PACHYONYCHIA CONGENITA

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Summary

A rare case of Pachyonychia congenita in a Muslim male child of 5 years is presented. Pachyonychia congenita is a rare hereditary disorder affecting predominantly skin and mucous membranes. Most patients present only with thickened nails. In this paper we describe the various features observed in the case.

Case Report

A Muslim male child of 5 years was brought in April 1976 for bouts of excessive sweating, miliaria, impetigenous lesions and horny papular thickening of the skin over the face, neck, trunk and extremities since the child's third week of life. The hyperhidrosis and miliarial lesions subside once the severity of summer is over, only to recur again.

Various degress of thickening of more than half the finger and toe nails is present since birth. (Fig. 1 page 219). Other nail changes are yellowish discoloration and marked subungual hyperkeratosis with elevation of the free margins of the nail plates. Repeated paronychial inflammation with loss of nail and regrowth of another abnormal nail with thickening more marked at the distal end has been a constant feature since birth.

There is marked hyperhidrosis with miliarial lesions on the face, neck and trunk. Greyish-white papular thickening of the skin with maceration and fissuring on the forehead, angles of the mouth and the nape of the neck is very striking. (Fig. 2 page 219) Hyperkeratotic verrucous plaques on the back, body folds and anterotalarel aspect of the arms started during the late infancy.

Moderate callosities developed on the pressure points of the feet like heels and heads of metatarsals from the second year when he started walking.

Eyes, teeth, oral mucous membranes and hair are normal.

Systemic examination did not reveal any abnormality.

Discussion

Pachyonychia congenita is a rare genodermatosis first described by Jadassohn and Lewandowsky, in 1906[1]. It is inherited as a simple Mendelian dominant trait characterised by incomplete penetrance. Though the condition is sometimes associated with features such as keratosis Pilaris, Palmoplantar keratoderma, hyperhidrosis, leukokeratosis, ichthyosis, bullae, steatocystoma multiplex, corneal dyskeratosis cataracts, hoarseness and caries teeth, there are monosymptomatic cases wherein nail abnormalities are the

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most constant single finding. Hence the unsatisfactory term pachyonychia congenita is still accepted and retained[2].

Pachyonychia congenita has been adequately reported and described in Western literature. Kumer and Loose reported a family of five generations with 34 cases and classified the condition into three types[3].

Type I :— Symmetrical keratose of the hands and feet with follicular keratoses of the body.

Type II :— Like type I but with leukokeratosis oris (Reihl type) This is commonest.

Type III :— Like type II but with corneal changes.

In our case there are follicular keratoses of the body, hyperhidrosis with miliaria, symmetrical keratoses of the feet and nail changes. Hence it falls into Type-I of Pachyonychia congenita. No other member in the family is having similar lesions. This could have been due to the incomplete penetrance of autosomal dominant gene. Involvement of oral mucosa, tongue, larynx and anal mucous membrane occur from second decade onwards. It is possible that mucosae are not involved yet as the patient is very young. Leukoplakia of the oral and anal mucous membranes occurring in this condition is potentially malignant. Thus in this disease we find a premalignant condition of hereditary nature.

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