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PHYSICAL DISABILITY DUE TO DARIER'S DISEASE: A RARE PRESENTATION

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ABSTRACT
Darier's disease is a rare inherited disease with characteristic skin, nail and mucosal involvement and also with learning disabilities. Here we report a case of physical disability (inability to walk) due to Darier's disease as it has not been previously reported in literature.

INTRODUCTION

Darier's disease, also known as Keratosis follicularis, Dyskeratosis follicularis, Darier–White disease or Benign dyskeratosis [1, 6] is a genetic disease with autosomal dominant trait affecting the skin, nails and mucous membrane. This was 1st described by Prince Marrow in 1886 and later in 1889 by Darier and White simultaneously [1, 6]. This is the first case of crippling physical disability (inability to walk) due to Darier's disease and is hence being reported.

Case report

A 45-year-old man presented to us with complaints of reddish-brown keratotic papules of 30 years duration with inability to walk (crippling physical disability) since 17 years. Initially the lesions which were painful appeared over palms and soles, gradually involving the axilla, groin, scalp and face over a span of 5 years. No other members in the family were affected. Physical examination revealed hyperpigmented, greasy, keratotic, crusted papules over the face, retroauricular area and scalp without any hair loss. Nasolabial folds, malar area and nose showed multiple pits (Figure 1). Malodorous warty plaques were formed in the flexures by coalescence of papules.

Grayish warty plaques and numerous pits were present over palms and soles, mainly affecting the pressure areas (Figure 2). Nail involvement was seen in the form of clubbing, subungal hyperkeratosis, angular nicking of free edge of the nail plate and longitudinal ridges with alternate red and white bands. Oral cavity showed fissured tongue, hypertrophic gums; cobblestone appearance of the hard palate, pharynx and larynx. Purplish macules were found over buccal mucosa. Genital examination revealed hyperkeratotic papules over shaft of penis and scrotum.

Musculoskeletal system examination revealed disuse atrophy of muscles of lower limbs with decreased power. Examination of other systems was within normal limits. Histopathologically, the biopsy revealed acantholytic dyskeratosis with corps ronds and grains.

Fig 1. Nasolabial folds, malar area and nose showing multiple pits and crusted papules

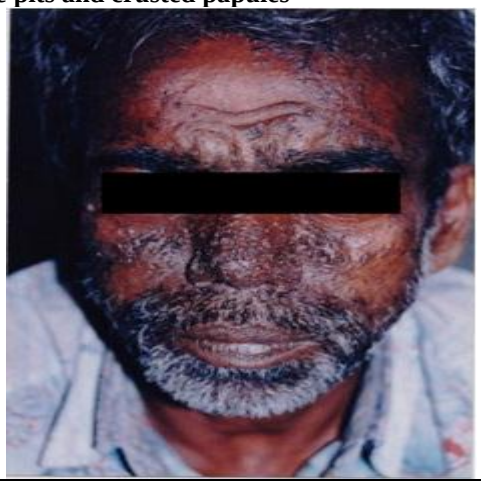
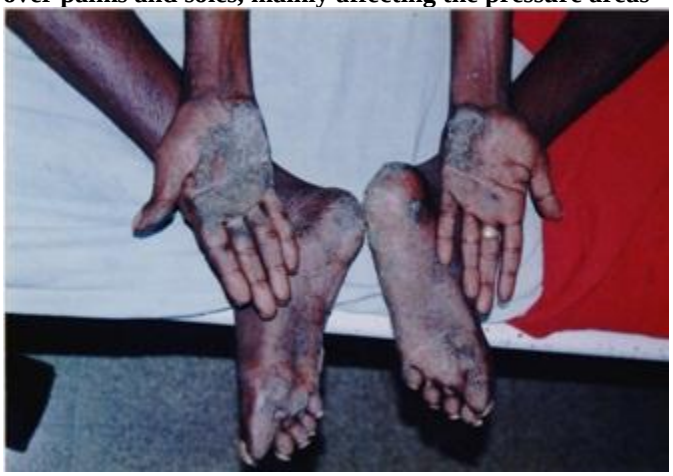


Fig 2. Grayish warty plaques and numerous pits present over palms and soles, mainly affecting the pressure areas



DISCUSSION AND CONCLUSION

Darier's disease is an autosomal dominant disease, with high penetrance and variable expressivity, with a prevalence of 1:100,000 and equal sex distribution [1, 2, 6]. But most of the cases are sporadic as a result of spontaneous mutation [2] and hence 47% of patients do not give a positive family history [1]. It is caused by mutation of genes ATP2A2 and SERCA2 [3], which affect the calcium balance in the epidermis.

Our case had many characteristic features of Darier's such as, keratotic papules in seborrheic distribution and less common features such as nail and mucosal involvement. But the distinguishing features were painful lesions over palms and soles [3], which made the patient to crawl and thus resulting in disuse atrophy of muscles. Another notable feature was the involvement of pharynx and larynx.

Although many rare forms of Darier's disease including zosteriform, localized [2], acral hemorrhagic [4] and its association with learning disabilities or neuropsychiatric disturbances [5] have been reported in the past, this is the 1st case of an association with physical disability. Hence we report this case and propose the inclusion of prenatal diagnosis and early treatment with systemic retinoids so as to avoid such crippling physical disabilities.

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