DARIER'S DISEASE

(Clinical study of fifteen cases)

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Summary

Fifteen cases of Darier's disease were studied. Males and females were in the ratio of 3 : 2. The disease appeared between the ages of 5 and 16 in majority (80%) of the patients. Family history was positive in only 60% cases. Itching was the main symptom in 2/3rd (66.6%) of cases. Seven cases experienced aggravation of the disease in summer. Recurrent skin infection and eczematisation were present in five cases. Majority of patients had typical lesions. Severity of the disease was directly related to its duration. Acrokeratosis verruciformis-like lesions over the hands and feet were present in all cases and 86.6% of cases had palmoplantar keratoderma. Mucosal lesions were seen in 73.3% of cases. These unusual findings are not often stressed in clinical practice and need to be highlighted. Leucoderma guttate, yet another unusual finding was seen in 26% of cases.

Darier's disease is a rare skin disease of familial origin[1]. Prince Marrow was the first man who described this in 1886[2], under the name of keratosis follicularis and Darier[3] was the first to recognise the phenomenon of dyskeratosis in this disease. The present paper is a study on the spectrum of clinical manifestations and other associated features of this disease.

Material and Method

Material consists of fifteen biopsy-proved cases of Darier's disease which attended skin OPD of JIPMER Hospital, Pondicherry during 1971-76. Their age, sex, complaints, family history, age of onset, aggravating factors, type and distribution of lesions, mode of spread, involvement of mucosa and nails, presence of keratoderma, leucoderma guttate, alopecia and other associated features were recorded.

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Observations :

Age, Sex and Onset - Out of fifteen cases, nine were males and six were females. Their ages ranged from 10 to 51 years. In twelve cases disease started between 5 and 16 years of age. In one patient it started at the age of 2 years. In two others disease started after forty years.

Family History :

Among 9 cases with positive family history, 6 had the disease in one of the parents and in 3 others it was present in

one of the children. In 6 patients, the family history was negative.

Symptoms :

In ten patients, the main presenting ymptom besides skin lesions was itching. In seven cases the itching and skin lesions used to get aggravated during summer months. Five patients used to get recurrent skin infection and eczematisation of the involved areas.

Morphology of the lesions and their distribution :

Typical lesions (papules with dirty brown crusts) were seen in eleven cases while skin coloured flat topped papules were seen in four cases. In all cases lesions were generalised but varying in number. Scattered and discrete lesions were seen in four cases and confluent lesions with eczematisation on face, neck and axillae were seen in five cases. In the remaining six cases lesions were numerous but discrete. The seborrhoeic sites (fig. 1) were more involved than the other areas.

In all cases warty lesions were present on the dorsa of hands and feet (fig. 2). They were devoid of dirty brown crust. In thirteen cases the palms and soles showed papules which were so closely set that they appeared like pebbled keratoderma.

Rare features :

1. Nail changes were seen in six patients in the form of thinned out nail plates, longitudinal red and white streaks, terminal notching, semilunar tips and subungual hyperkeratosis. These changes were present mostly on the finger nails.

2. Mucosal lesions - In eleven cases closely set papules forming violaceus and white plaques were present on the hard palate (fig. 3).

3. Leucoderma guttate - In three patients, small depigmented discrete pinhead sized to a few millimeters sized macules were seen on the lower abdomen, upper parts of thighs and genitalia. There were no papules in these areas. In another patient slightly bigger depigmented lesions were present on the abdomen, chest and back. These were intermingled with the keratotic papules of Darier's disease.

Other associated findings observed were partial alopecia scalp (one case) utero-vaginal prolapse (one case), recurrent pyoderma and eczematisation (five cases) and pseudoainhum (fig. 2) (one case).

Investigations :

Histopathology-Biopsies were done from (a) the typical skin lesions (b) palatal lesions (4 cases), (c) palms (6 cases) and (d) the dors of hands (6cases). All showed typical histopathology of Darier's disease.

Comments :

Sex incidence and age of onset of disease in our patients were comparable with those of earlier reported series[1,4,5]. The greater incidence in males has been attributed to their more frequent exposure to sun[1] ; but we were unable to explain the male preponderance on that basis. Although an autosomal dominant inheritance is common in this disease[1,6] evidence for such an inheritance was seen only in 40% of our patients. In the others, disease may have occurred as a result of mutation. Lesions on oral mucosa[1,4-12] oesophagus[13] rectal mucosa[14] and larynx[15,16] have been considered rare occurrences. In our series involvement of oral mucosa was present in 11 patients (73.3%). Similar high incidence of mucosal lesions were reported by Getzler and Flint[6]. Verma et al[10] detected mucosal involvement in 12.5% of their

Fig. 1 Showing typical lesions of Darier's disease predominantly over seborrhoeic sites

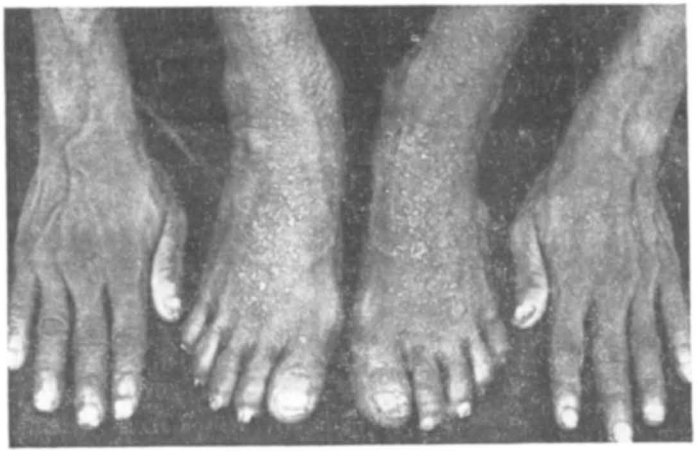

Fig 2 Showing

(a) Wart like lesions of Darier's disease over the hands and feet.

(b) Pseudo-ainhum left fifth toe

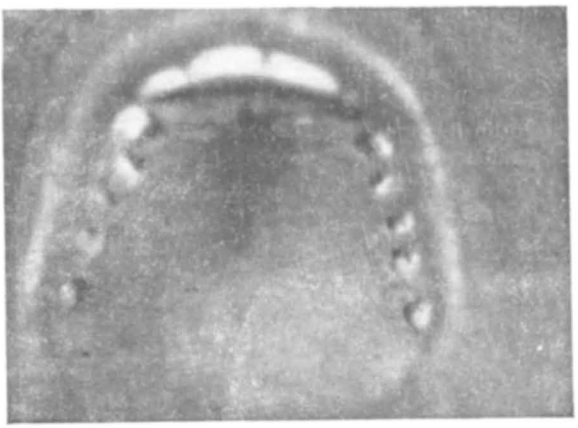

Fig. 3 Showing punctate whitish papules of Darier's disease over the hard palate

North Indian patients. Leucoderma guttate was described by Cornelison et al[17] and four (28%) of our patients manifested these lesions. We believe that leucoderma guttate is a feature of this disease. Acrokeratosis verruciformis like lesions described by some authors[18-21] were present in all our patients. 13 patients showed diffuse palmoplantar keratoderma.

Nail changes as described by Ronchese[22] and Zais et al[23] were seen in six of our patients but the typical red and white longitudinal lines were present only in two cases. Features like small stature[6], low intelligence[6], genital hypoplasia[4], psychotic episodes[6], diffuse fibrosis with nodulation effect mainly in the lower lobe of lungs[24] and cystic changes in bones[25] were not present in our cases.

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