Dermatopathia pigmentosa reticularis (DPR): A rare case of late onset disease

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Sri Lanka Journal of Dermatology, 2009, **13**, 34-35

Abstract

Dermatopathia pigmentosa reticularis (DPR) is a rare inherited disorder presenting with the clinical triad of reticulate pigmentation, nail dystrophy and diffuse alopecia.

The usual presentation of DPR is at birth or very early childhood (<2 years). A few cases of late onset disease have been reported. We report a rare case of post pubertal onset disease which had started at the age of seventeen years.

Introduction

Dermatopathia pigmentosa reticularis (DPR) is a rare type of ectodermal dysplasia presenting with the clinical trial of reticulate pigmentation, nail dystrophy and diffuse alopecia. Associated variable clinical features include palmoplantar keratoderma, adermatoglyphia, hypohidrosis or hyperhidrosis and non cicatricial bullae. Autosomal dominant inheritance has been reported in the literature[1].

Most of the reported cases are from the western countries. There are only a few cases reported from the eastern world[2].

DPR usually starts at birth or in very early childhood. There are a few reported cases of late onset disease[2]. A 19-year old boy who developed the disease at the age of 17 is reported here.

Case report

A 19-year old boy presented to the Dermatology Clinic at the National Hospital of Sri Lanka with a history of asymptomatic abnormal pigmentation on the trunk, upper arms, palms and soles and in the oral cavity for 2 years duration. He also complained of discoloration, thickening and ridging of most of his finger and toe nails for the same duration. He had noticed diffuse thinning of his scalp hair for five to six months duration. He did not give a history of blistering of hands and feet or abnormal sweating. There is no consanguinity or family history of a similar disorder.

Examination revealed reticulated pigmentation prominent over the trunk and upper limbs. Abnormal pigmentation was also seen on the palms and soles and in the oral cavity. There was no flexural accentuation. Almost all of his finger and toe nails were dystrophic and thickened. There was diffuse thinning of scalp hair. No eye brow, axillary or pubic hair loss was noted. There was no evidence of oral leukoplakia, palmoplantar keratoderma, adermatoglyphia or palmar pits. His teeth were also normal.

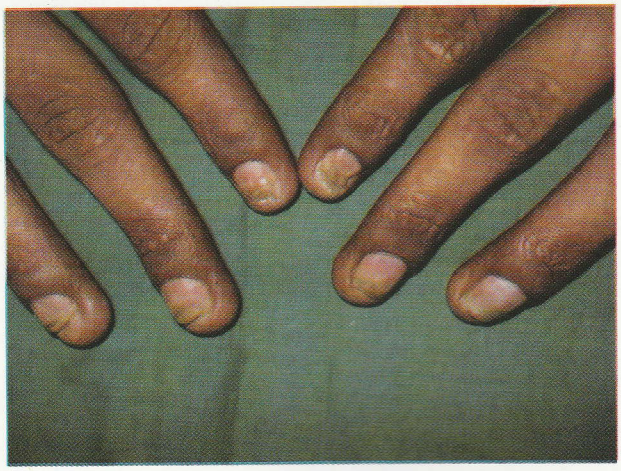

Figure 1. *Nail dystrophy and thickening.*

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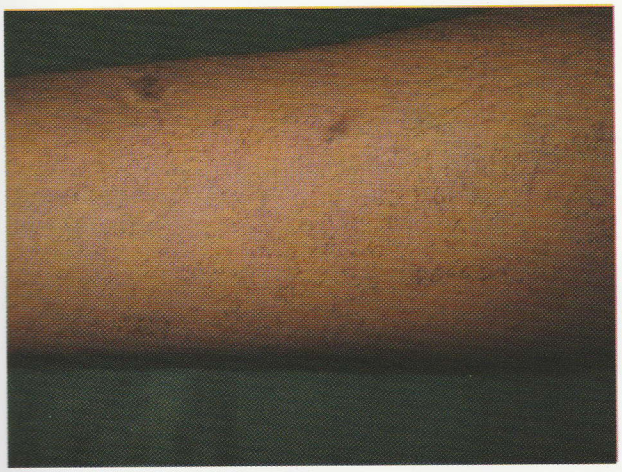

Figure 2. *Reticulate pigmentation of forearm.*

His routine blood investigations were normal. There was no evidence of bone marrow suppression. Thyroid hormone and serum cortisol levels were normal. Two skin biopsies showed pigment incontinence with no features of basal cell degeneration.

Discussion

DPR is a rare congenital disorder of pigmentation. Three primary features of DPR are reticulate pigmentation mainly in the trunk, nail dystrophy and non cicatricial alopecia. There are many other variable clinical features described in the disease such as adermatoglyphia[3,4] hypohidrosis, hyperhidrosis[4] palmoplantar hyperkeratosis, non scarring blisters in the hands[4] and darkly pigmented areolae. Our patient had remarkable pigmentation in the palms and soles and in the oral mucosa in addition to pigmentation in the trunk and arms. He did not have adermatoglyphia or palmar plantar hyperkerotosis. He denied any blisters in the hands and feet.

The age of onset of DPR is usually at birth or early childhood (less than 2 years). There had been a few cases reported in late childhood. Our patient had disease onset at 17 years of age which is unusual. This is the eldest reported case in the literature to our knowledge.

Differential diagnosis of DPR are Naegeli-Franceschetti-Jadassohn syndrome (NFJ) and dyskeratosis congenita (DC). NFJ and DPR are ectodermal dysplasias caused by mutations in keratine 14[5]. In NFJ dental abnormality is a primary feature whereas it is not a feature in DPR. In contrast to DPR the reticulate pigmentation tend to improve in adult life in NFJ[6,7].

DC is associated with nail dystrophy, palmar plantar hyperkeratosis and hypopigmented macules scattered among the pigmentation[8]. Leukoplakia and haematological disorders occur commonly. On the basis of the distribution of the lesions and the nail changes acropigmentation of kitamura is unlikely.

Since the onset of disease is at 17 years we also considered lichen planus and graft vs host disease (GVHD) in our patient. There was no other clinical or histological evidence of lichen planus. He never has had a blood or blood product transfusion. There were no sclerotic or lichenoid lesions or any evidence of GVHD in the histology.

The usual histology of DPR is localized pigment incontinence. Some cases of DPR has shown liquefactive degeneration of the basal layer. Our patient's histology showed pigment incontinence with no basal cell degeneration. Genetic analysis could not be carried out due to lack of facilities to detect the mutations in keratin gene.

We report this case due to its atypical late presentation.

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