Dermatopathia pigmentosa reticularis: A report of a case with delayed onset alopecia and onychodystrophy

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INTRODUCTION

Dermatopathia pigmentosa reticularis (DPR) is an extremely rare autosomal dominant ectodermal dysplasia that occurs because of mutations in *KRT14*. It mainly affects the skin, nails, and hair, with a characteristic diagnostic triad of widespread reticulate hyperpigmentation that begins at birth or during early childhood, noncicatricial alopecia (usually mild), and onychodystrophy. Patients with this syndrome may also have adermatoglyphia, palmoplantar hyperkeratosis, hyperhidrosis or hypohidrosis, and acral dorsal nonscarring blisters.[1-5]

The first cases of DPR were reported and described in 1958 by Hauss and Oberste-Lehn.[1,6] To date, <20 cases of this syndrome have been reported worldwide.[2,6,7] We report a case of DPR, and to the best of our knowledge this is the first case of this extremely rare syndrome reported in Iraq. We are urged to report this case because of several factors: a rarity of reports in the literature, an uncanny similarity to other syndromes—in particular, Naegeli-Franceschetti-Jadassohn syndrome (NFJS) and dyskeratosis congenita (DKC)—and the unusual age of presentation of certain diagnostic clinical features in this case.

CASE REPORT

A 24-year-old Iraqi man presented to our outpatient clinic at Al-Sadr Teaching Hospital with diffuse brown hyperpigmentation associated with diffuse thinning of the scalp hair and discoloration and brittleness of many fingernails and some toenails. The hyperpigmentation was present at birth on the trunk and then spread to involve the proximal extremities, palms and soles, and head and neck, in addition to sublingual mucosa, all within a span of 1 to 2 years. Thinning of his hair began at 19 years of age, and the nail abnormalities occurred more recently, at 23 years of age. These lesions were associated with a lack of dermatoglyphics, hyperhidrosis, and heat intolerance. The patient had been unemployed for the last 6 years and denied any history of trauma to his fingers or exposure to chemicals.

According to the patient, he had 2 brothers and 6 sisters; 1 brother and 1 sister had the same symptoms, though no other first- or second-degree relatives were affected.

On examination, the patient had generalized reticulate hyperpigmentation involving the trunk, face, neck, proximal extremities, palms, soles, popliteal fossae, penile shaft, and sublingual mucosa, and there was increased pigmentation of the areolae (Fig 1). A physical examination of the hair revealed diffuse nonscarring alopecia involving the frontal, parietal, and occipital areas of the patient's scalp (Fig 2). A hair-pull test was negative. The patient also had onychodystrophy of all fingernails except for the nails of the fourth and fifth fingers of his left hand; some toenails were also involved, in particular the nails of the first and fifth toes (Fig 3). The dermatoglyphics were lacking (Fig 4). The patient's teeth were normal and there was no oral leukoplakia or pallor. Routine laboratory test results were normal.

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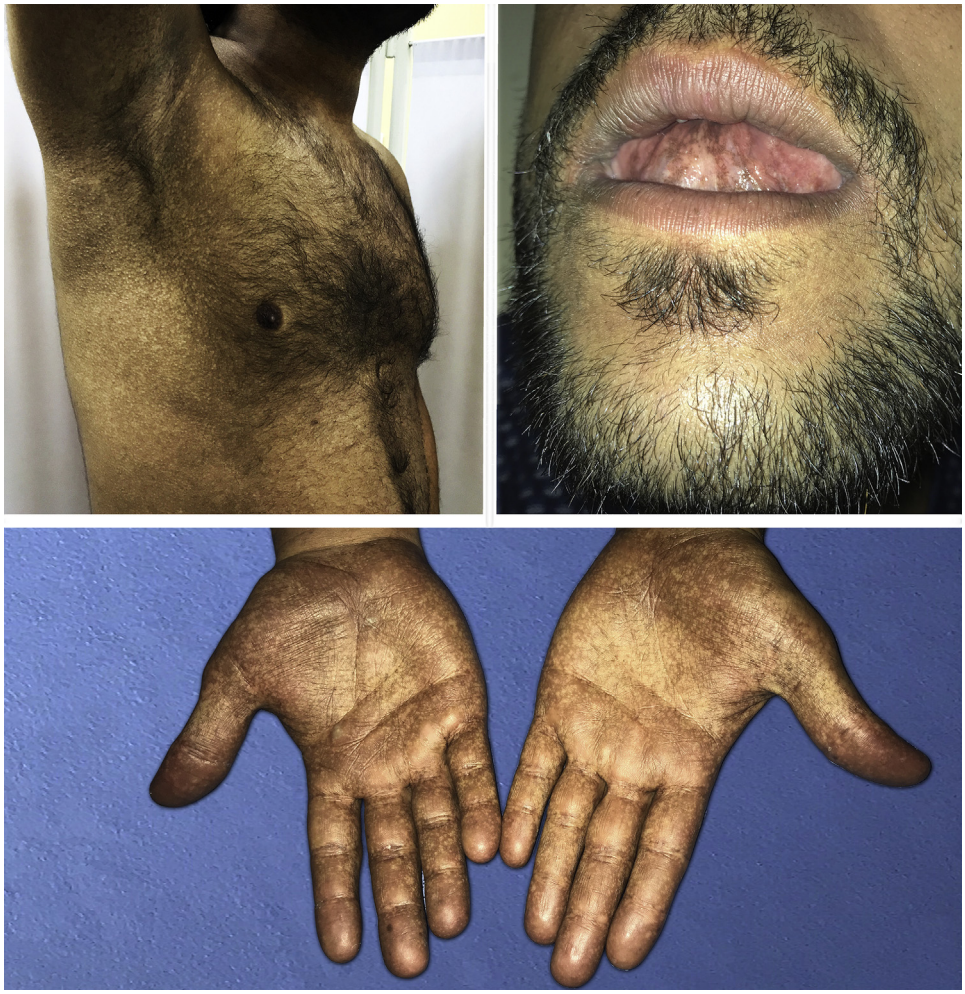

Fig 1. Reticulate hyperpigmentation involving the trunk, tongue, and palms.

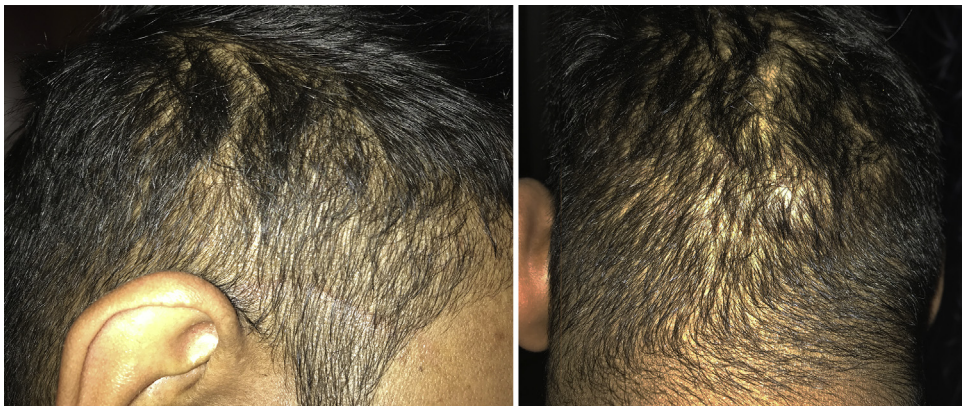

Fig 2. Diffuse nonscarring alopecia (lateral and posterior views of the patient's head).

His serum cortisol level was elevated at 730.6 nmol/L (normal range in the morning, 171-536 nmol/L). In addition, thyroid function tests, total testosterone, and dehydroepiandrosterone sulfate levels were normal. His blood film morphology and erythrocyte sedimentation rate were normal, and tumor markers were negative. An abdominal ultrasound revealed hepatomegaly. A biopsy specimen from the reticulate hyperpigmentation on the trunk revealed epidermal atrophy, increased basal layer

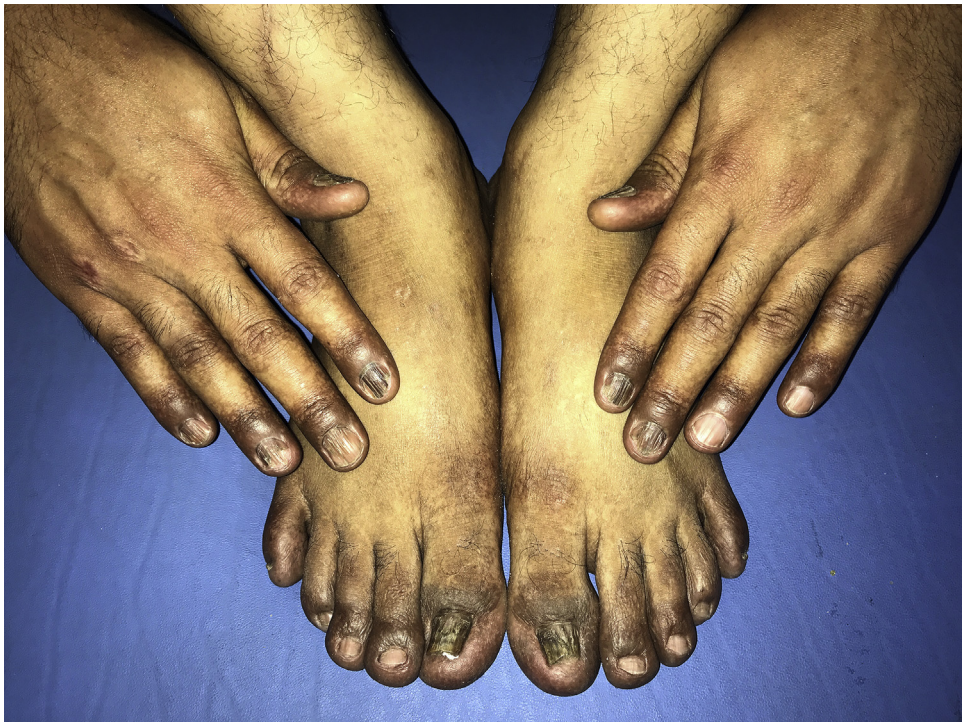

Fig 3. Onychodystrophy of the fingernails and toenails.

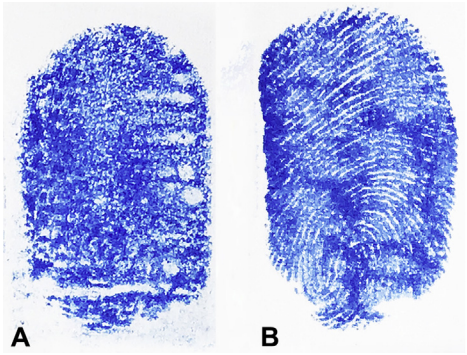

Fig 4. Fingerprint of the patient's left thumb, demonstrating adermatoglyphia (**A**) compared with a normal fingerprint (**B**).

pigmentation, basal layer degeneration, and the presence of melanophages in dermis.

DISCUSSION

We report a 24-year-old Iraqi man with diffuse reticulate hyperpigmentation that presented at birth and continues throughout adulthood that is associated with diffuse nonscarring alopecia together with onychodystrophy. The latter 2 features have had an onset after 19 years of age. These 3 clinical features collectively represent the diagnostic triad of DPR, which was first described by Hauss and Oberste-Lehn in 1958.[1]

The reticulate hyperpigmentation and nail abnormalities can also be seen in NFJS and DKC. The clinical features of NFJS include dental abnormalities and, in many cases, the hyperpigmentation fades or disappears after puberty or in adulthood; in addition, nonscarring alopecia is not a feature of NFJS.[4,6] In DKC, the hyperpigmentation is associated with leukoplakia of the oral mucosa, bone marrow dysfunction, and the patient may have abnormal dental findings.[4-6]

In addition to the diagnostic triad of DPR, our patient also has adermatoglyphics, sweating abnormalities, and heat intolerance, which are also features of this syndrome. The patient refused genetic testing. This is not an issue, because the diagnosis of DPR can be made clinically based on its characteristic diagnostic criteria—namely, generalized reticulate hyperpigmentation, noncicatricial alopecia, and onychodystrophy.[7-10] Genetic testing aids in confirmation of the diagnosis when not all diagnostic criteria are present. In our case, not only these diagnostic criteria are present, but also the other features of the condition mentioned above. These features, supported by the typical histopathology of the reticulate hyperpigmentation, compelled us to make the diagnosis of DPR.

Based on previous studies, our case differs in the late onset of both alopecia and onychodystrophy. This suggests that the reticulate hyperpigmentation may be the only presented feature of DPR before

puberty and, therefore, it is wise to consider the diagnosis of DPR in any child with diffuse reticulate hyperpigmentation that begins in early childhood, even in the absence of the other 2 sides of the diagnostic triangle (alopecia and onychodystrophy). Furthermore, an elevated serum cortisol level and hepatomegaly may be associated or coincidental findings. Therefore, correlation with future reported cases will be required.

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