Dermatopathia Pigmentosa Reticularis

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Abstract

Dermatopathia pigmentosa reticularis is a rare ectodermal dysplasia that presents with a triad of reticulate hyperpigmentation, nonscarring alopecia, and nail dystrophy. We report herein a case of a 23-year-old male presenting with the characteristic triad associated with anhidrosis and palmoplantar keratoderma.

Key Words: Dermatopathia pigmentosa reticularis, ectodermal dysplasia, reticulate pigmentation

Introduction

Dermatopathia pigmentosa reticularis (DPR), a rare form of autosomal dominant ectodermal dysplasia, was first described by Hauss and Oberste-Lehn in the year 1958. Defect in the N-terminal part of keratin molecule results in loss of inhibition against proapoptotic signals. KRT14 mutation has resulted in a case of dermatopathia pigmentosa reticularis with wiry scalp hair and digital fibromatosis. While reticulate hyperpigmentation involving the whole body, nail dystrophy, and nonscarring alopecia forms the classical triad of dermatopathia pigmentosa reticularis, other clinical features such as hypohidrosis or hyperhidrosis, palmoplantar thickening, and loss of dermatoglyphics and ainhum-like constrictions have also been reported.

Case Report

A 23-year-old male presented to us with the complaints of absence of sweating associated with discomfort during the summer months and gradually progressive darkening of the skin for the last 17 years. Additional complaints were thickening of the palms and soles along with thinning of hair. The patient’s past medical history was otherwise unremarkable. There was no complaint of similar illness in the family. Examination revealed the presence of generalized reticulate pigmentation most pronounced over the trunk and proximal part of the upper limb [Figures 1 and 2] along with miliaria-like lesions. Palmoplantar keratoderma with a yellowish tinge was also seen [Figure 3]. Nail dystrophy [Figure 4] along with nonscarring alopecia involving the frontal area, madarosis, and loss of eyelashes were other findings [Figure 5]. The skin was generally dry. Systemic review was noncontributory. Routine blood investigations were within normal ranges. A 10% KOH mount from one of the fingernail scrapings was positive for fungal elements. Biopsy and subsequent histopathological examination revealed the presence of hyperkeratosis, parakeratosis, follicular plugging, and basal cell melanization [Figure 6]. Based on the clinical and histopathological features, a diagnosis of DPR was made.

Discussion

DPR is a rare disorder of ectodermal dysplasia inherited in an autosomal dominant pattern, presenting with the characteristic triad of generalized reticulate pigmentation, nonscarring alopecia, and nail dystrophy. The reticulate pigmentation has its onset in early childhood and remains throughout life. Other associated features are absent or poorly developed dermatoglyphics [Figure 7], sweat gland dysfunction, palmoplantar keratoderma, and nonscarring blisters over the acral areas.

Goh et al. reported a patient of Malay descent having wiry scalp hair and fibromatous thickening of digits. Goel et al. described an Indian girl with reticulate pigmentation and corneal changes in the form of Salzmann’s nodular degeneration. Shanker and
Gupta noticed diffuse alopecia with ill-developed pubic and axillary hair in an Indian female.\(^7\) Maso et al. reported a case of reticulate hyperpigmentation with a history of seizures and neurofibromas, without any other features of von Recklinghausen’s disease.\(^8\) Melanin-laden macrophages scattered in the papillary dermis with an overlying normal epidermis and basal layer degeneration have been the usual histopathological findings.

Reticulate hyperpigmentation starting in infancy or childhood can be attributed to a horde of disorders,
including dyskeratosis congenita, incontinentia pigmenti, epidermolysis bullosa simplex, and X-linked reticulate pigmentary disorders, besides DPR/Naegeli–Franceschetti–Jadassohn syndrome (NFJS). Other genodermatoses having reticulate pigmentation are Dowling–Degos disease and reticulate acropigmentation of Kitamura, with onset in adulthood. Focused history taking and careful clinical examination can differentiate DPR from these close differentials.

Incontinentia pigmenti, an X-linked dominant disorder, evolves sequentially in four stages, with reticulate pigmentation along the lines of Blaschko observed in infancy. Affected individuals develop dental, ocular, and CNS abnormalities as well. Epidermolysis bullosa simplex presents with widespread or focal blistering at birth or shortly afterward, which often heal with a postinflammatory mottled reticulate pigmentation. Palmoplantar hyperkeratosis may be an accompanying feature. Dyskeratosis congenita, commonly inherited in an X-linked recessive manner, is a multisystem disorder characterized by reticulate hyperpigmentation, onychodystrophy, poikiloderma, cytogenetic abnormality, and leukoplakia with a tendency toward malignant transformation. Dowling–Degos disease typically presents in adolescence with reticulate pigmentation of the flexures without any nail or hair changes. Acropigmentation of Kitamura presents with acral pigmentation, palmar pits, and broken palmar ridges.

NFJS and DPR may be distinguished on the basis of absent dental anomalies, partial alopecia, and persistent pigmentation in the latter.

NFJS and DPR are two allelic disorders attributable to mutations in the KRT14 gene, with mutation occurring in the nonhelical (E1/V1) head domain resulting in premature termination of protein synthesis. Apoptosis may play a key role in the pathogenesis of these conditions as evidenced by increased apoptotic activity in the basal cell layer expressing keratin 14. A similar presentation due to a variety of missense mutations in gene for keratin 14 has also been reported.

No specific treatment is available for DPR. Symptomatic management of hyperkeratosis and other secondary changes can be done with keratolytics, topical steroids, and emollients.

**Declaration of patient consent**
The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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**Conflicts of interest**
There are no conflicts of interest.

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