Dermatopathia pigmentosa reticularis: A rare reticulate pigmentary disorder

Vinay Shanker, Mudita Gupta

ABSTRACT

Dermatopathia pigmentosa reticularis is a rare ectodermal dysplasia with a triad of generalized reticulate hyperpigmentation, noncicatricial alopecia, and onychodystrophy. We report a case of a 21 year old woman who had generalized reticulate pigmentation, diffuse noncicatricial alopecia and onychodystrophy of finger and toe nails. Along with this triad she had palmoplantar keratoderma and poorly developed dermatoglyphics. There was no evidence of involvement of other ectodermally derived organ.

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

INTRODUCTION

Dermatopathia pigmentosa reticularis (DPR) is a rare ectodermal disorder with the diagnostic triad of generalized reticulate hyperpigmentation, noncicatricial alopecia, and onychodystrophy. Since first described by Hauss and Oberste-Lehn[1] in 1958, approximately 12 cases of DPR have been reported.[2] There has been a single case report from India by Brar et al.[3] Goh et al.[4] noted a patient of Malay ancestry with DPR secondary to a recurrent KRT14 p.R125C mutation.[4]

CASE REPORT

We report a case of a 21-year old woman who presented with onset of darkening of skin since 6 months of age, which increased progressively to whole of the body within a span of 2-3 years. There was also progressive thickening of skin of palms and soles. Diffuse thinning of hair on scalp was present [Figure 2]. She had never had a hair-cut and hair length was never more than up to lower end of neck. Pubic and axillary hair were also poorly developed. No history of photophobia or any other eye complaints was there. Hearing and sweating were normal. There was no history of similar problem in the family. She was born to a second gravida by normal vaginal delivery. She had 2 siblings who were normal. The morphology of the hair shaft was normal on clinical and microscopic examination. Generalized reticulate hyperpigmentation was present [Figure 1]. Scalp hair were short and there was diffuse thinning [Figure 2]. Nails were dystrophic [Figure 3]. Hyperkeratosis of palms and soles was there [Figure 4]. Poorly developed dermatoglyphics were there [Figure 5]. Oral mucosa and teeth were normal. No ocular or auditory involvement was seen. Her intelligence quotient and stature was estimated to be in the normal range. There was no delay in secondary sexual character development. Routine investigations in the form of complete hemogram, liver function test, renal function test, and chest radiography were normal. Histopathology revealed basal layer degeneration and absence of skin adenexa and melanophages in dermis [Figure 6] and interface dermatitis.

A diagnosis of (DPR) with a differential of Naegeli-Franceschetti-Jadassohn (NFJS) was done. Absence of enamel defects and the presence of diffuse non-scarring alopecia favour DPR. She was prescribed keratolytics topically for hyperkeratosis.

DISCUSSION

DPR is an autosomal dominant ectodermal dysplasia characterized by a triad of widespread reticulate pigmentation, non-scarring alopecia and nail changes.[5] Other associated findings include adermatoglyphia, hypohidrosis or hyperhidrosis, palmoplantar hyperkeratosis, and acral dorsal...
The reticular pigmentation of DPR occurs at birth or during early childhood and persists throughout life.[6]

Other genodermatoses associated with generalized reticulate pigmentation are dyskeratosis congenita (DKC), NFJS, X-linked nonscarring blisters.[2] Other genodermatoses associated with generalized reticulate pigmentation are dyskeratosis congenita (DKC), NFJS, X-linked nonscarring blisters.[2]
reticulate pigmented disorder, Dowling-Degos disease, reticulate acropigmentation of Kitamura and Haber's syndrome. Pigmentation is at different skin sites, depending on the specific underlying disease.

Reticulate hyperpigmentation, mucosal leukoplakia, bone marrow dysfunction, cytogenetic instability, and a predisposition to malignancy are characteristic of DKC. These patients can have dental findings, reticulate hyperpigmentation, adermatoglyphia, palmoplantar hyperkeratosis, and nail anomalies similar to NFJS and DPR patients.

In Kitamura’s disease, palmar pits and breakage in palmar ridges and acral hyperpigmentation, especially on the backs of hands and feet can be observed. Haber’s syndrome is characterized by verruciformous papular lesions of the trunk and a distinct facial erythema and telangiectasia, most commonly presenting in childhood. In X-linked reticulate pigmentary disorder in females, pigmentation occurs along the Blashcko’s line. Hereditary bullous acrokeratotic poikiloderma of Weary-Kindler has some striking similarities to NFJS but poikiloderma is present. Flexural reticulate hyperpigmentation occurs in Dowling-Degos disease. Additional findings, such as dark hyperkeratotic follicles, pitted perioral scars, and comedo-like lesions may occur. Galli-Galli disease also shows macular and papular reticulate pigmentation of flexures.

NFJS and DPR are ectodermal dysplasias. They are inherited in an autosomal dominant fashion. Both manifest with poorly developed dermatoglyphics, reticulate hyperpigmentation of the skin, hypohidrosis, and heat intolerance. Palmoplantar keratoderma, nail dystrophy, and enamel defects are common in NFJS, whereas diffuse alopecia is only seen in DPR. Teeth are always severely affected, leading to early total loss in NFJS. In some NFJS pedigrees, the reticulate pigmentation fades after puberty and may disappear completely in old age. In DPR the hyperpigmentation persists throughout life, showing no tendency of spontaneous fading. The reticulate network of hyperpigmented macules occurs particularly on the trunk, neck, and proximal areas of the limbs.

Dereure[7] noted that NFJS and DPR are 2 allelic ectodermal dysplasias related to mutations of dominant gene coding for keratin 14. Severe keratin 5 and 14 mutations induce down-regulation of junction proteins in keratinocytes, which likely underlies all of these diseases.

Abnormalities of dermatoglyphics are divided into 4 main categories; ridge aplasia, ridge hypoplasia, ridge dissociation, and ridge off the end.[7] Embryologic development of dermal ridges occurs in conjunction with the eccrine glands and sweat pores are found on the ridges. Ridge hypoplasia refers to poorly formed dermal ridges while ridge dissociation is characterized by a discontinuous pattern in which dermal ridges are broken into short segments. Ridge hypoplasia and dissociation are the abnormalities that can be seen in DPR as well as NFJS.

No specific laboratory changes are seen in DPR. The typical histopathologic picture of DPR shows liquefaction degeneration of the basal layer and dermal pigmentary incontinence. That is, an interface dermatitis can be present. No specific treatment exists for DP, except for symptomatic management of some of the associated conditions, such as palmoplantar hyperkeratosis. For hyperkeratosis, topical retinoic acids and keratolytics may be beneficial. Nonscarring blisters are generally transient and self-healing. Cold compress may suffice.

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