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Case Report

Reticulate acropigmentation of Kitamura with Dowling Degos disease overlap

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Abstract Reticulate pigmentary dermatoses (RPDs) are group of uncommon genetic disorders which are inherited as an autosomal dominant pattern. Reticulate acropigmentation of Kitamura (RAK) and Dowling-degos disease (DDD) are considered to be a part of RPDs. A 23 year old female presented with bilaterally symmetrical multiple ephelid-like dark macules over dorsum of hands, forearms, dorsum of feet, shin, axillae and groin. The patient showed characteristic clinical and histopathological overlap of both the rare diseases (RAK and DDD), thus supporting the hypothesis that they represent two different features of a single entity with variable phenotypic expression.

Key words
Reticulate pigmentation, Dowling Degos diseases, reticulate acropigmentation of Kitamura.

Introduction

Reticulate pigmentary disorders (RPDs) are a group of uncommon genetic pigmentary conditions inherited in an autosomal dominant pattern.[1] They clinically present as multiple hyperpigmented macules arranged in a reticular pattern. RPDs include Reticulate acropigmentation of Kitamura (RAK), Dowling-Degos disease (DDD), Reticulate acropigmentation of Dohi (RAD), Galli Galli disease and Haber's disease.[2]

RAK and DDD are both uncommon genodermatoses that invariably affect the extremities and flexures, respectively and follow autosomal dominant pattern of inheritance with variable penetrance. There have been only a few reports in the past of the clinical phenotype of the overlap of the two conditions.

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Case report

A 22 year old female presented with multiple asymptomatic dark coloured macules on dorsum of hands and feet since she was 4 years of age. These lesions progressively increased in size and number to involve forearms, shins and neck. Later she developed multiple dark coloured macules over axillae and groin at around 18 years of age. Her parents were first degree relatives. She was delivered by normal vaginal delivery at home and had an uneventful childhood with normal developmental milestones.

Dermatological examination revealed multiple hyperpigmented macules in a reticulate pattern over dorsum of hands, feet, forearms, legs, axillae, groin and neck **(Figure 1-3)**. Multiple pits over both palms with breakage in palmar ridges were also present **(Figure 4)**. The rest of the general, systemic and cutaneous examination revealed no abnormality.

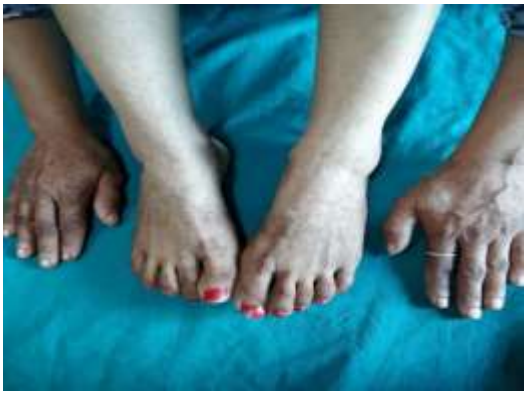

Figure 1 Reticulate pigmentation over the dorsum of hands and feet.

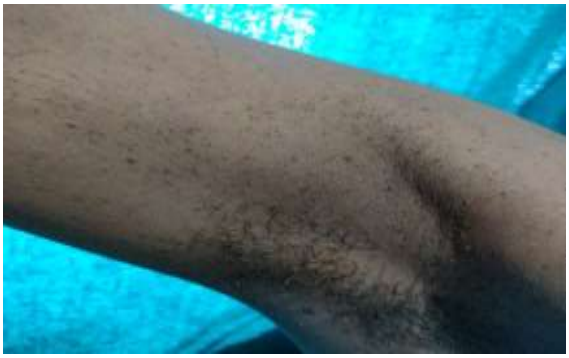

Figure 2 Reticulate pigmentation over the axilla.

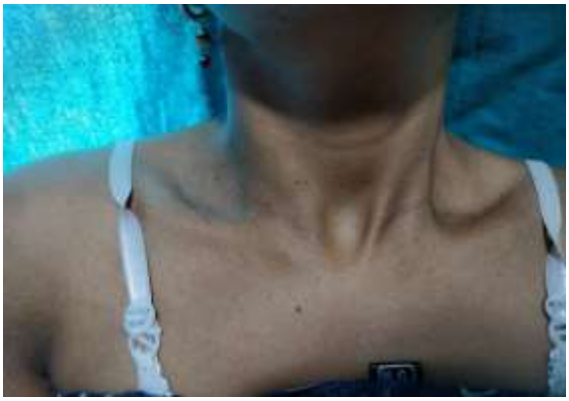

Figure 3 Reticulate pigmentation over the neck.

Histopathology showed variable elongation of the rete ridges with increased pigmentation and clusters of melanocytes at the tip of the rete ridges **(Figure 5)**. Based on the clinical features and histopathology, a diagnosis of overlap between Reticulate Acropigmentation of Kitamura and Dowling Degos disease was made.

Discussion

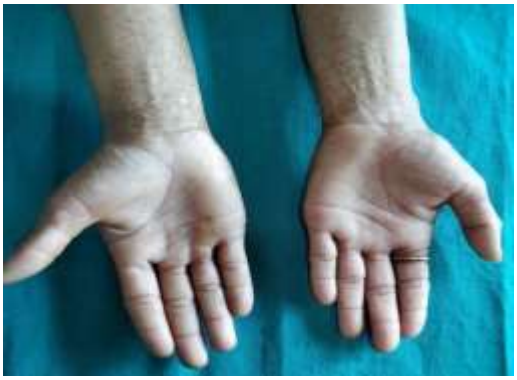

Figure 4 Breaks in dermatoglyphic pattern of the palms and pigmentation over the flexor aspect of the wrist.

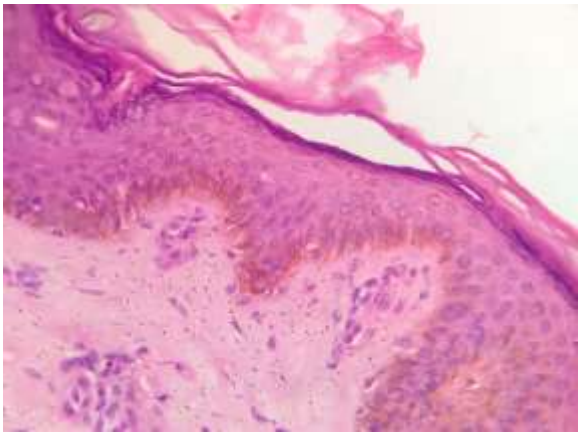

Figure 5 H&E staining (400X magnification) shows variable elongation of the rete ridges with increased pigmentation and clusters of melanocytes at the tip of the rete ridges.

Reticulate acropigmentation of Kitamura was first described by Kitamura and Akamatsu in Japanese patients in the year 1943. It is an entity that usually occurs in Japanese patients but the condition has been recognized in other countries as well. The age of onset is usually seen in first and second decades of life. Clinically the lesions first appear as ephelid-like hyperpigmented macules arranged in a reticular pattern over the dorsal aspect of hands and feet. Gradually the lesions may spread proximally. Pitting in the dermatoglyphics may also be seen in palms producing breaks. Histopathology includes filiform elongation of the rete ridges with clusters of melanocytes over them and hyperpigmentation of basal keratinocytes.[3]

DDD was described initially by Dowling and Freudenthal in 1938. In 1978, Wilson Jones and Grice described DDD as "demonstrating dusky dappled disfigurements and dark dot depressions and disclosing digitate downgrowths delving dermally". It is a rare genetic skin disease inherited in an autosomal dominant pattern. The condition usually presents in adult life. It is also known as "pigmented reticulate anomaly of the flexures". DDD is characterized by reticulate pigmentation of the flexures, perioral pitted scars and comedo like papules.

Table 1 Differentiating features between Dowling Degos disease and Reticulate Acropigmentation of Kitamura

Differentiating feature		*Dowling Degos disease*	*Reticulate Acropigmentation of Kitamura*
Genetic basis	Genes	KRT5 POFUT1 POGLUT1	ADAM10
	Chromosome	Chromosome 12 Chromosome 20 Chromosome 17	Chromosome 15
	Inheritance	Autosomal dominant/ sporadic	Autosomal dominant/ sporadic
Clinical features	Age of onset	Childhood (first to early second decade of life)	Adolescence to adult life
	Site of involvement	Flexures	Distal extremities
	Itching	May be present	Absent
	Involvement of anogenital area	Present	Absent
	Comedo-like papules	Present	Absent
	Pitted perioral scars	Present	Absent
	Break in dermatoglyphics	Absent	Present
	Palmoplantar pits	Absent	Present
	Atrophy	Absent	Early atrophy
	Photosensitivity	Absent	May be present
	Palmoplantar keratoderms	Absent	May be present
	Associated diseases	Hidredinitis suppurativa, squamous cell carcinoma, seborrheic keratosis, keratoacanthoma	None
	Alopecia	Absent	Partial alopecia
Histo-pathology	Epidermis	Hyprkeratosis or atrophy with thinning of suprapapillary epidermis	Epidermal atrophy
	Elongated rete ridges with basal hyperpigmentation	Antler-like' rete ridges with pigmentation at tips	Present
	Comedo-like cysts	Present	Absent
	No. of melanocytes	Normal	Clumps of melanocytes
	Involvement of follicles	Prominent	Not pronounced

Histopathological findings include acanthotic epidermis, irregular elongated "antler like" rete ridges with increased melanin pigmentation at the tips. There is no increase in melanocyte number.[4] There are multiple treatment options for these conditions but none of them are effective. Treatment with topical retinoids has been largely unsuccessful. Azelaic acid is a treatment option which has some potential. Erbium- doped yttrium aluminium garnet (Er: YAG) is another treatment option.[5]

Our patient presented with acral pigmentation and palmar pits resembling RAK but also with flexural involvement resembling DDD. There have been few previous reports on the coexistence of RAK and DDD in the literature.[6] In 1983, Crovato *et al.* first reported a family with features of RAK and DDD.[7] This has led many authors to conclude that the two diseases are different phenotypic expressions of the same disease because of their similar clinical presentation and histopathology.[8] However, recent genetic studies have shown there is defect in KRT5, POFUT1 and POGLUT1 genes in Dowling Degos disease and ADAM10 gene in case of reticulate acropigmentation of Kitamura.[9] The polygenic nature of DDD supports the possibility of RAK genes being involved in the same clinical spectrum.[10] We present this case because of the co-existence of the two rare conditions. Differentiating features between RAK and DDD are given in the **Table 1**.

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