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A rare case report of overlap of reticulate acropigmentation of kitamura with early onset dowling-degos disease

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Abstract:

Dowling-Degos disease (DDD) and Reticulate Acropigmentation of Kitamura (RAPK) are two distinct groups of rare genodermatoses, which fall under an umbrella of reticulate pigmentory dermatoses. Both are inherited as autosomal dominant trait with variable penetrance. Few studies have described DDD and RAPK in a single patient hypothesising that two conditions belong to a single complex disease. We describe a 32 years old male patient with hyperpigmented macules on neck, axillae, chest, lower back, forearms, hands and feet along with palmar pits and acneiform scars over face and comedo-like lesions over back since 10 years of age. The idea that both DDD and RAPK are distinct aspects of a single entity with variable phenotypic expression is further strengthened as our patient had features of both conditions. In addition, our patient had early onset DDD which is a rare feature.

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Introduction

Reticulate pigmentory dermatoses (RPDs) include congenital and acquired disorders.[1] Congenital RPDs are a group of rare pigmentory conditions inherited mainly as autosomal dominant pattern and are typically characterized by hyperpigmented macules coalescing in a reticulate pattern, few are associated with hypopigmented macules. Examples of RPDs are reticulate acropigmentation of Kitamura (RAPK), Dowling-Degos disease (DDD), reticulate acropigmentation of Dohi (RAPD), Galli-Galli disease (GGD), dyschromia Universalis herediataria (DUH), Haber's disease, Naegeli-Franceschetti-Jadassohn syndrome, dermatopathia pigmentosa reticularis (DPR) and dyskeratosis congenital (DC). DDD was first reported by Dowling and Freudenthal in 1938 and then by Degos and Ossipowski in 1954. RAPK was first reported by Kitamura and Akamatsu in 1943. Both dermatoses are characterized by progressive

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hyperpigmented macules affecting mainly flexures and extremities, respectively. In literature, there are few case reports of overlap of DDD-RAPK. We report a similar interesting case with the features of both disorders.

CASE REPORT

A 32 years old male presented with multiple asymptomatic dark coloured lesions over dorsum of hands and feet since 10 years of age. The lesions extended proximally in number to involve both forearms and shin. Patient also developed asymptomatic dark coloured lesions over neck, both axillae, upper chest, back and lumbosacral region starting from the age of 12 years. He had taken treatment from multiple doctors over the years without any satisfactory results. Patient was born of non-consanguineous marriage. There was a history of similar dark coloured lesions over intertriginous areas in father. Cutaneous examination revealed numerous symmetrical atrophic hyperpigmented macules over face, neck, bilateral axillae, upper chest, upper back, lumbosacral region, flexural aspects of bilateral arms and forearms, dorsa of bilateral hands and feet (Figure *1a-f*). These macules were arranged in reticular pattern. Multiple pitted scars were present over malar regions with no previous history of acne (Figure *1b*). Comedo-like lesions were present over upper back, lumbosacral region and upper chest (Figure *1c*). Multiple palmar pits were noticed with breaks in proximal palmar ridges (Figures *1g*). Hair, nail and mucosal examination showed no abnormality. Systemic examination was unremarkable.

Figure 1a-g: Multiple discrete atrophic hyperpigmented macules over face, neck, upper chest, upper back, dorsal aspects of bilateral hands extending proximally to forearms, dorsal aspects of bilateral feet extending proximally to shin, acneiform scars over malar regions (red arrow, fig. 1b), comedo-like lesions over neck (yellow circle, fig. 1c), multiple pits over palms (fig. 1g)

Figure- 1a, 1b, 1c

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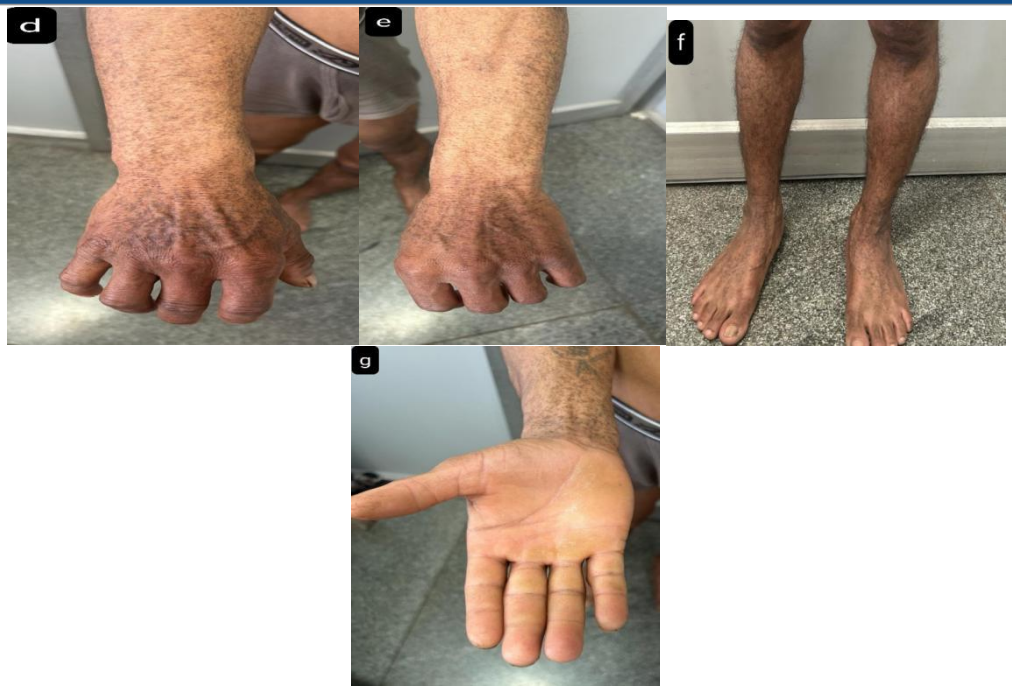

Figure- 1d, 1e, 1f, 1g

Dermoscopy of macules revealed numerous discrete hyperpigmented globules in irregular reticular pattern network (Figure *2a*). Dermoscopy of palm showed break in dermatoglyphics by black globules (Figure *2b*).

Figure 2a: Dermoscopy (using HEINE DELTA 20T) of pigmented macules showing numerous irregular pigment network
Figure 2b: Dermoscopy (using HEINE DELTA 20T) of Palm showing multiple pits along with break in dermatoglyphics by globules.

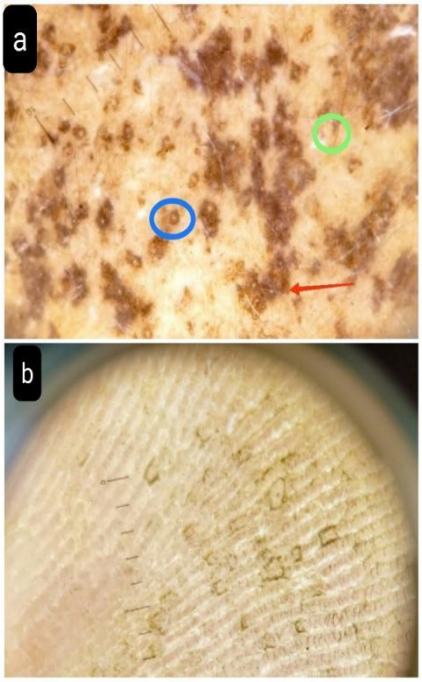

Figure- 2a, 2b

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Histopathological examination (Figure *3a* and *3b*) of macule over back revealed filiform elongation of rete ridges with increased pigment in the basilar keratinocytes creating an antler horn pattern. Dermis showed sparse lymphocytic infiltration suggestive of DDD.

Figure 3a: Histopathology of pigmented macule from back demonstrating filiform elongation of rete ridges with hyperpigmentation of basilar keratinocytes creating classical 'Antler horn pattern', keratin cyst (H and E stain, 10x)

***Figure 3b*: High power view of the same (H and E, 40x)**

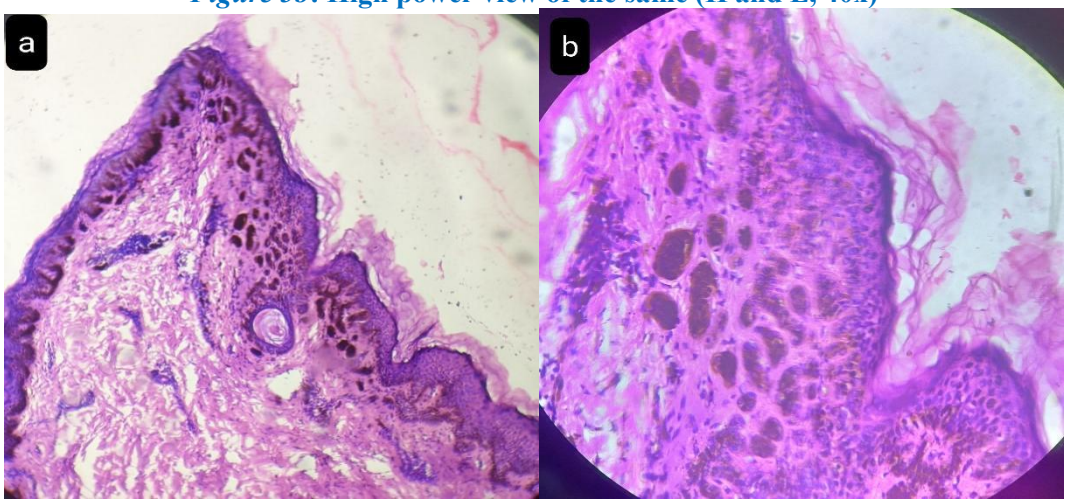

Figure- 3a, 3b

Genetic analysis could not be done due to financial constraints. The patient was advised for Nd-YAG laser treatment and topical retinoids to the patient. Patient lost to follow up after 3 months.

Discussion:

DDD is characterized by reticulated hyperpigmentation of flexures mainly neck, axillae, antecubital fossae, inframammary and intragluteal regions, inner aspects of arms and thighs and rarely groins starting after puberty. In our case, the lesions started during adolescence. We hypothesize it may be due to phenomenon called 'anticipation' in which signs and symptoms of a genetic condition tend to become more severe and appear at an earlier age as condition is passed from one generation to the next. The lesions are usually asymptomatic, pruritus may be sometimes present. Wilson Jones and Grice described DDD as "demonstrating dusky dappled disfigurement and dark dot depressions and disclosing digitate downgrowths delving dermally" in 1978.[2] Additional cutaneous features include pitted perioral acneiform scars, comedo-like lesions on back or neck. Conditions associated with DDD are hidradenitis suppurativa (HS), epidermoid cyst, pilonidal sinus, squamous cell carcinoma, seborrhoic keratosis, keratoacanthoma. Classic DDD (DDD1) is caused by loss of function mutation in keratin 5 gene (KRT5) on chromosome 12q13 leading to haploinsufficiency.[3] Mutation in DDD2 and DDD4 are present in POFUT1 gene on chromosome 20q11, and POGLUT1 gene on chromosome 3q13, respectively. Mutation in PSENEN gene has been reported in patients with DDD and HS. GGD is a rare acantholytic variant of DDD also caused by KRT5 mutation.

Usual onset of RAPK is in first or second decade of life and is characterized by hyperpigmented macules in a reticular pattern favouring dorsal aspect of hands and feet, rarely extending proximally over time. Progressive darkening of the lesions may also be present due to sun exposure. Other features include pits on palms, soles and dorsal phalangeal surface and disruption of dermatoglyphics. It is caused by loss of function mutation in ADAM10 gene (a disintegrin and metalloproteinase 10), which encodes for zinc metalloproteinase that activates Notch signaling.

The overlap between DDD and RAPK in a family was first reported in 1983 by Rebora and Crovoto.[4] In Indian literature, the overlap was reported by authors as summerized in table *1*.

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Table *1*:

	Age/Gender	RAPK onset	DDD onset	Interval between RAPK and DDD	Additional features
Thami et. al.[5] (Index case)	55/ F	15 yrs of age	NA	NA	Hypopigmented lesions over forearms and legs
Vasudevan B et. al.[6]	25/ M	22 yrs of age	NA	NA	Hypopigmented macules over trunk
Rathoriya et. al.[7]	28/ M	1st decade	3rd decade	15 yrs	Accessory tragus
Singh SK et. al.[8]	22/ F	10 yrs of age	? 10 yrs of age	0	-
Yadav A et. al.[9]	29/ M	7-8 months ago	?	14 yrs	-
Our case	32/ M	10 yrs of age	12 yrs of age	2 yrs	-

Table 1: Comparison of Indian studies of RAPK-DDD overlap cases with our study
(NA = Not available).

It is still controversial whether DDD, RAPK, RAPD and GGD are variants of a single disease entity. Distinguishing features are summerized in table *2*.

Table *2*:

	RAPK	DDD	RAPD	DUH	Haber's disease	DC	Naegeli – Franceschetti – Jadassohn syndrome	DPR
Pattern of inheritance	AD	AD	AD > AR	AD > AR	AD	X-LR > AD, AR	AD	AD
Gene mutation	ADAM10	DDD1 and GGD: KRT5, DDD2: POFUT1, DDD3: unknown, DDD4: POGLUT1	ADAR1, DSRAD	DUH1 & DUH3: ABCB6, SASH1, PER3; DUH2: KITLG	unknown	DKC1	KRT14	KRT14
Onset	1st/2nd decade	>puberty, adulthood	Infancy, early childhood	Early in life	Early adolescence	1st decade of life	By the age of 2 years	By the age of 2 years
Cutaneous features (reticulate pigmentation)	Reticulate hyperpigmentation	Reticulate hyperpigmentation	Reticulate hyperpigmentation and hypopigmentation (mottled pigmentation)	Reticulate hyperpigmentation and hypopigmentation (mottled pigmentation)	Reticalate hypedrpigmentation	Reticulate hyperpigmentation, sometimes hypopigmentation	Reticulate hyperpigmentation	Reticulate hyperpigmentation
Site	Dorsa of extremities	flexures	Dorsa of extremities	Generalised distribution, trunk, limbs, face	Trunk, axillae, proximal extremities	Neck, upper chest, upper arms	Neck, trunk, proximal extremities, axillae, groins	Trunk > proximal extremities
Other cutaneous features	Punctate pits over palms and	Pitted perioral acneiform	Freckle like macules over face,	Hair and nail abnormality,	Photosensitivity, persistent	Poikiloderma, nail dystrophy (pterygium),	Heat intolerance (ectodermal dysplasia), absent	Non scarring alopecia, onychodystro

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	soles, breaks in epidermal ridge pattern	scars, comedo like lesions, HS, epidermoid cyst, SK, KA, SCC		abnormal dermal connective tissue, nerve tissue	rosacea like facial lesions, keratotic papules, comedo like lesions, pitted sars, PPK	oral leukoplakia, malignancy (SCC)	dermatoglyphics, PPK, onychodystrophy, dental anamoly	phy, absent dermatoglyphics, hypo or hyperhidrosis, punctuate PPK
Systemic compications	-	-	neurogenic complication (rare)	Neurologic (seizures, mental retardation), hearing loss, ocular abnormalities, hypospadias	-	epiphora, bone marrow failure, pulmonary fibrosis, liver cirrhosis	-	-
Histopathologial features	Hyperkeratosis without parakeratosis OR epidermal atrophy, slightly elongated (club like) rete ridges with pigmentation of tip, perivascular lymphocytic infiltate	Characteristic thin branch like patterns of epidermal downgrowth with concentration of melanin at the tip forming antler horn pattern, no additional increase in number of melanocytes, perivascular lymphohistiocytic infiltrate. GGD: Prominent suprabasal nondyskeratotic acantholysis + above faetures	Hyperpigmented lesion: increased melanin within basal keratinocytes. Hypopigmented lesion: decreased melanocytes, degenerative vacuolation	Pigmented basal layer of epidermis, pigmentary incontinence in papillary dermis, melanophage and lymphocytes in upper dermis	Digitate elongations of hyperpigmented rete ridges	Early changes: hydropic degeneration of basilar keratinocytes, band like inflammatory infiltrate in upper dermis. Late changes: epidermis is marked by thinned and flattened melanophages present in upper dermis, telengectasias of superficial vessels	Clumps of melanin-laden melanophages in dermis in patchy distribution No overlying epidermal hyperpigmentation	Clumps of melanin-laden melanophages in dermis in patchy distribution No overlying epidermal hyperpigmentation

Table 2: Distinguishing features of congenital reticulate pigmentory disorders

(AD = Autosomal dominant, AR = Autosomal recessive, HS = Hidradenitis suppurativa, KA = Kearatoacanthoma, PPK = Palmoplantar keratoderma, SCC = Squamous cell carcinoma, SK = Seborrhoic keratosis, X-LR = X-linked recessive.)

Based on the location of hyperpigmented macules, presence of palmar pits, facial acneiform scars and comedo like lesions over back along with absence of hypopigmented lesions and typical histopathological finding with absence of acantholysis, our case is most likely to be DDD-RAPK overlap. There history of similar lesions in

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father, suggesting autosomal dominant nature of disease in our case.

Treatment options for DDD and RAPK are limited and include topical adapalene, azaleic acid, systemic retinoids and Er-YAG laser. Most of the treatment yield unsatisfactory results and provide only temporary improvement.

Conclusion

We report this interesting overlapping case of 2 rare genodermatoses, which adds on to the hypothesis that overlap between DDD and RAPK might be a part of single disease entity. Genetic study to identify possible mutation can confirm the diagnosis of the same.

Declaration of patient consent

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

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