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A RARE CASE OF FAMILIAL AMYLOIDOSIS CUTIS DYSCHROMICA IN THREE SIBLINGS

Dermatology

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ABSTRACT

Amyloidosis cutis dyschromica is a rare form of primary cutaneous amyloidosis with very few cases reported in literature. We report three siblings , 24 and 20 -year -old females and 22- year- old male presenting with asymptomatic reticulate pigmentation with hypopigmented and hyperpigmented macules, prepubertal onset and familial tendency. Based on histopathological findings, diagnosis of ACD was made. We present this case due to rarity of its occurrence and resemblance to other commonly occurring reticulate pigment dermatoses.

KEYWORDS

Amyloidosis cutis dyschromica , Familial, Reticulate pigmentation, Acitretin

INTRODUCTION:

Ever since its recognition by Morishima (1970), only <50 cases of Amyloidosis cutis dyschromica have been published worldwide.[2,3] It is extremely rare and distinct variant of primary cutaneous amyloidosis characterised by prepubertal onset, asymptomatic or minimally pruritic reticulate pigmentation and focal amyloid deposits in papillary dermis.[1,2]

Poikiloderma-like amyloidosis shows similar clinical cutaneous picture and amyloid deposits as ACD but with additional features of poikilodermic lesions, photosensitivity, blisters, lichenoid papules and palmoplantar keratoderma.[2,4,5] Various drugs tetracycline, diphenylcyclopropenone, thiazides, monobenzyl ether of hydroquinone and afloqualone can cause dyschromatosis.[6] Here in we report a case of peculiar reticulate pigmentation diagnosed histologically as ACD. The disorder is thought to be familial as her siblings had similar condition.[3,4,7]

Case Report:

A 24- year -old female born to non-consanguineous parents presented with progressive and asymptomatic mottled hyperpigmentation since 4-5 years. The lesion appeared initially over right leg later spread to involve other leg, extensor and lateral aspect of hands and forearms. The lesions have darkened over the years. There was no history of photosensitivity or extensive sun exposure. Patient was treated with topical corticosteroids with no satisfactory improvement.

General physical examination was normal. Her teeth, hair, nail and mucosa of the mouth, palms and soles and vulva were normal. Systemic examination was unremarkable. The mental and developmental milestones of the patient were normal.

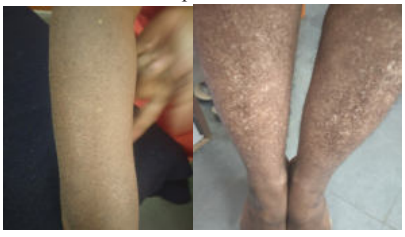

Fig 1,2 .Hyperpigmented macules with hypopigmented and depigmented macules of varying sizes involving both forearm and legs.

Her 22- year- old brother and 20- year- old sister have remarkably similar cutaneous manifestation of hyperpigmented macules associated with hypopigmented and depigmented macules of varying sizes involving only hands and legs. (Fig 1,2) Her siblings had less severe and localised disease compared to her. On examination, her younger sister had light coloured hair. Other family member had no systemic manifestation.

Dyschromatosis universalis hereditaria, xeroderma pigmentosum, poikiloderma-like amyloidosis and progressive macular hypomelanosis were considered in the differential diagnosis and a biopsy was taken from anterior aspect of leg. Histopathology showed increased melanin in basal layer of epidermis with melanin incontinence. The papillary dermis shows deposits of amyloid with globular appearance. These amyloid bodies were seen in direct contact with overlying basal cells of epidermis (Fig 3,4).

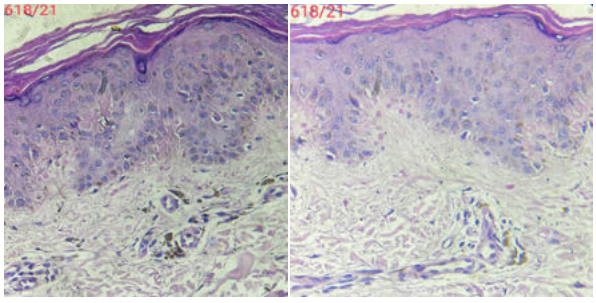

Fig 3,4. HPE showing increased melanin in basal layer of epidermis with melanin incontinence and globular amyloid deposits in papillary dermis.

Based on the clinical and pathological findings, diagnosis of amyloidosis cutis dyschromica was made. Patient was started with oral Acitretin 25 mg once daily, Topical corticosteroids and sunscreen. Patient was reviewed 1 month later, showed increased hair density over hands (Fig 5). Hence potency of steroids was reduced with continuation of oral acitretin.

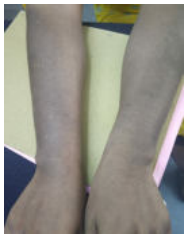

Fig 5 Increased hair density over hands post acitretin treatment

DISCUSSION:

Cutaneous amyloidosis has been classified into primary cutaneous amyloidosis, secondary cutaneous amyloidosis and systemic cutaneous amyloidosis. PCA is the deposition of amyloid in previously apparent normal skin with no systemic involvement.[3,4]

The uncommon type, PCA have three subtypes which are relatively common: Lichen or popular (most common subtype of PCA), macular, and nodular or tumefactive (relatively rare). Other rarer subtypes of

PCA have also been described in the literature which are dyschromic cutaneous amyloidosis or ACD, bullous, vitiliginous, familial poikiloderma -like cutaneous amyloidosis (FPLCA) and anosacral type.[1,6,7]

ACD is a very rare variant of PCD, first described by Morishima in 1970. It is characterized by the following four features i) mottled hyper- and hypo -pigmentation ii) little or no itching iii) pre-pubertal onset and iv) focal amyloid deposits below epidermis. The etiopathogenesis of ACD does not seem very clear but genetic factors leading to prolonged and defective UVB and UVC induced DNA repair and apoptosis of keratinocytes has been proposed. These apoptotic keratinocytes get phagocytosed by histiocytes or fibroblasts. The cytokeratins of lysed keratinocytes give rise to the amyloid material which gets deposited In the skin.[1,6,8,9]

Dyschromia being pathognomonic finding in ACD, existing literature does not clearly explain its mechanism. Increased basal layer pigmentation due to hyper melanized melanocytes, increased number of melanocytes, with or without melanin sequestration within papillary dermal melanophages leads to cutaneous hyperpigmentation. On the other hand, hypopigmentation or depigmentation results from destruction of melanocytes, decreased melanosomal transfer to epidermal keratinocytes or reduced melanogenesis in melanocytes .[6,10,11] A new hypothesis causing hypopigmentation was recently proposed by Verma K et al (2009 p46) , suggesting the possibility of stretching basement membrane and decreasing density of melanocytes per unit length of the basal layer of epidermis caused by papillary amyloid deposits.[2]

ACD is differentiated from dyschromatosis universalis hereditaria, xeroderma pigmentosum, poikiloderma-like amyloidosis and progressive macular hypomelanosis by the absence of amyloid deposits on histology. Poikiloderma-like amyloidosis shows additional features of poikilodermic lesions, photosensitivity, blisters, lichenoid papules and palmoplantar keratoderma.[2,4,5] Verma S & Joshi M (2015 p386) reported strong family history of vitiligo.[11]

Many therapeutic modalities have been proposed with variable success and anectodal reports of efficacy of topical steroids, keratolytics, capsaicin, dimethyl sulfoxide and CO2 laser.[3] Acitretin was given and seems to be effective by inducing apoptosis, thereby reducing the available keratinocytes, the most probable source of amyloid. Acitretin probably aids phagocytosis by macrophages, thereby ridding the dermis of amyloid.[7,12]

CONCLUSION

Amyloidosis cutis dyschromica is an extremely rare variant of primary cutaneous amyloidosis. Due to its rarity, there is paucity in literature to its guide in etiopathogenesis, diagnosis and treatment. We have tried to fill in these gaps in literature which no other case report has done to the best of our knowledge. However further research with adequate sample size is required to support our pathogenesis and treatment aspect.

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