CASE REPORT

Amyloidosis Cutis Dyschromica: A Rare Case Report

Avantika Jain, Preeti Doshi, Purva Kulkarni, Parineeta Shelke, NS Mani, Reena Bharadwaj

Abstract

Amyloidosis cutis dyschromica (ACD) is an extremely uncommon variant of cutaneous amyloidosis (PCA) which is a pigment disorder in the form of macules with or without pruritis and amyloid deposition in papillary dermis. We report a rare case of 50-year male patient with multiple dyschromic skin patches over abdomen, limbs and back.Histopathology & special stains with polarizing microscopy confirmed the diagnosis of ACD.

Key Words

Amyloidosis Dyschromica Cutis, Congo Red Stain, Pigmentation Disorder

Introduction

Morishima *et al*., first described Amyloidosis cutis dyschromica (ACD) in 1970.[1] It is an uncommon variant of primary cutaneous amyloidosis, which shows varying sized macules which are pigmented in form of hyperpigmented, hypopigmented or depigmented lesions. Pruritis could be associated with the lesions. Either macular, lichenoid, biphasic or nodular form may also seen.[2]Men and women are equally affected. The papillary dermis shows amyloid deposition on skin biopsy. The exact pathogenesis remains unknown. ACD are been associated with mutation of the glycoprotein non-metastatic melanoma protein B (*GPNMB*).[3] We report an uncommon case of ACD in a 50-year-old, male.

Case Report

A 50 year, male presented with dyschromic skin patches all over the body since 18 years of age. On multiple oral and topical treatment, no resolution was seen.

General physical examination revealed no significant abnormality. Cutaneous examination revealed diffuse hyperpigmentation with mottled spotty, depigmented and hypopigmented patches over chest, abdomen, back, limbs, neck, and face (Fig A). Hair, nails, and oral mucosa were unremarkable. Systemic examination was unremarkable.

A 4mm skin biopsy was done from the lesion and sent for histopathological examination.On microscopy, the epidermis reveled increase in melanin pigment in melanocytes and keratinocytes. The papillary dermis showed extracellular clumps of eosinophilic deposit. The rest of the dermis and dermal appendages were normal (Fig B). These eosinophilic deposit gave a brick red colored on the amyloid depositionin the papillary dermis when stained by Congo red special stain (Fig C) and also showed apple green birefringence under polarized light microscopy (Fig D). All the above features lead to a diagnosis of ACD.

Discussion

Vijaikumar & Thappa [1] , may be the first in India to report cases of Amyloidosis cutis dyschromica in two siblings. Amyloidosis cutis dyschromica (ACD) is a rare skin condition characterized by progressive, asymptomatic changes in skin pigmentation, resulting in both hyperpigmentation and hypopigmentation. This condition involves the extracellular deposition of amyloid material in the skin, with no associated systemic involvement, making it a form of primary cutaneous amyloidosis.

There are three primary types of cutaneous amyloidosis: systemic cutaneous amyloidosis, secondary cutaneous amyloidosis, and PCA. Lichenoid, macular, and nodular are the three subtypes of PCA.[4] Other atypical variants are ACD, bullous, vitiliginous, familial poikiloderma like cutaneous amyloidosis and anosacral type.[5]

ACD is an uncommon variant, clinically by widely distributed, hyperpigmented, hypopigmented, or depigmented macules of different sizes.These are linked

Department of Pathology, Bharati Vidyapeeth (DU) Medical College, Pune, India.

Correspondence to: Dr. Preeti Doshi, Professor, Department of Pathology, Bharati Vidyapeeth (DU) Medical College, Pune, Maharasthra, India.

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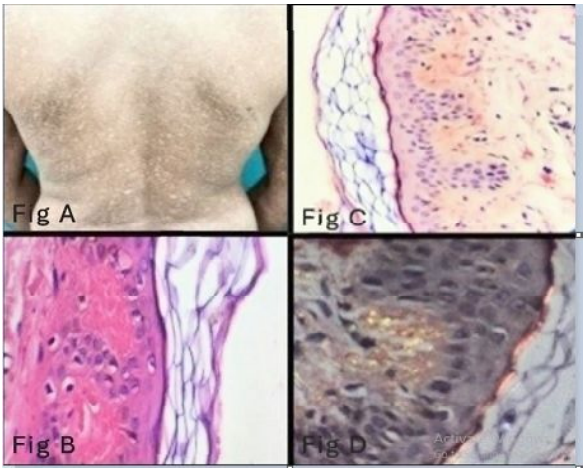

Fig A: Diffuse Hyperpigmentation with Mottled Spotty, Depigmented and Hypopigmented Patches.Fig B: (400x H&E) Papillary Dermis with Eosinophilic Deposits. Fig C: (100x Congo red) Brick Red Positivity. Fig D: (400x Polarizing light) Apple Green Birefringence.

to prepubertal onset, with little to no pruritis. The clinical and histopathological findings in our case were similar to those reported in the literature, with amyloid deposition in the papillary dermis.[6]

ACD is commonly recognized as a primary cutaneous amyloidosis, there have been a few instances where it has been linked to other diseases. [7]The gene mutated in this disorder is GPNMB, which encodes for transmembrane glycoprotein NMB, which has been reported in many cells including melanocytes and keratinocytes. UVB and UVC rays cause apoptosis of keratinocyteand poor DNA repair in genetically predisposed individuals. Histiocytes or fibroblasts phagocytose these apoptotic keratinocytes. Amyloid material is produced by the cytokeratin of lysed keratinocytes,which is deposited in the papillary dermis layer of theskin.[4]

The clinical diagnosis of amyloidosis cutis dyschromica also requires the exclusion of a small number of other cutaneous dyschromic disorders. Numner of differential diagnosis which includes: Dyschromatosis-universalishereditaria, xerodermapigmentosum, poikiloderma like amyloidosis, and progressive macular hypomelanosis.[7] Amyloid deposition is not detected in all the above mentioned diseases other than poikiloderma-like amyloidosis.[8] Poikiloderma-like amyloidosis can be differentiated from other conditions based on defining features like: these lesions predominantly appear on the limbs, presenting as raised, flat-topped papules and blisters. The condition typically emerges during adulthood. Affected individuals often experience heightened sensitivity to sunlight, with skin changes worsening upon exposure to UV light. Short stature or delayed growth is commonly observed in individuals with this condition. There may be a mild to moderate thickening of the skin on the palms and soles, though it is usually less severe compared to other forms of keratoderma. These characteristic features help differentiate poikiloderma-like amyloidosis from other conditions with similar clinical presentations, emphasizing its unique combination of skin changes and systemic associations.[3]

Histopathological evaluation with specific Congo red stain is the best clue for the diagnosis of ACD.[9] Immunohistochemistry can also be used for ACD, dermal deposits are positive for Cytokeratin CK34bE12 and CK5/6. In literature reported are many frameshift mutations, non-sense mutations, missense mutations, and splice site mutation found in ACD. [10]

Conlusion

ACD is an exceptionally rare form of primary cutaneous amyloidosis (PCA). Diganosis can be confirmed with histopathology, special stains with polorazing microscopy.

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