PROGRESSIVE PIGMENTATION OF WHOLE BODY MIMICKING HEREDITARY RETICULATE PIGMENTATION IN A YOUNG GIRL: A RARE PRESENTATION OF AMYLOID CUTIS DYSCHROMICA

Medical Science

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KEYWORDS

INTRODUCTION

Primary cutaneous amyloidosis is a chronic, progressive disorder of skin, because of the amyloid deposition in the skin with no systemic involvement. Amyloid cutis dyschromica (ACD) is considered a rare variant of primary cutaneous amyloidosis with around 50 cases reported so far. Most cases are reported from Asia, majority with family history. Autosomal recessive inheritance in GPNMB encoding glycoprotein non metastatic gene B has been reported in many cases with few case reports of semidominant inheritance. It is usually asymptomatic condition as opposed to other types of primary cutaneous amyloidosis like macular or lichenoid variant which are associated with moderate to severe pruritus and photosensitivity. Chief complaint in majority of cases is cosmetic concern only. In view of the very few cases reported from India, we hereby report the case of a 28 years old female having similar dyspigmentation in one sibling.

CASE DISCUSSION

A 28 years old unmarried female presented in skin OPD with history of dyspigmentation on body since 12-15 years starting from dorsum of feet then extending to other parts in one year. On examination, multiple lesions were present on trunk and extremities in the form of hypopigmented macules of size ranging from 2mm to <1 cm hyperpigmented, dark brown colored macular lesions sparing face, palms and soles. Lesions were equally distributed on extensor and flexor surfaces. All other systems were normal. No significant medical history was present. She gave history of similar lesions in younger sibling since 8-10 years, his lesions started from trunk.

Dermoscopy showing striped, globular white monotonous spots (red arrow)

Dermoscopy showed striped, globular white monotonous spots with intermingled margins and variable hyperpigmented granules, dots and globules. Histopathological examination revealed extracellular eosinophilic hyaline deposits consistent with amyloid in superficial dermis. Sparse perivascular lymphocytic infiltrate admixed with melanophages was seen in papillary dermis. Orthokeratotic hyperkeratosis showed an occasional apoptotic keratinocyte overlying the foci of deposit. Congo red stain gave brick red colour to dermal deposits supporting amyloid.

DISCUSSION

Morishima, from Japan described this disorder in 1970 as a variety of localised cutaneous amyloidosis characterised by dyschromia. He described it as a diffused speckled hypo and hyperpigmentation with absence of telangiectasia, atrophy, papules and mild or no itching with prepubertal onset⁽¹⁾. In our case, the onset was almost at the time of onset of puberty. It is a rare disorder with around 50 cases reported till date, most cases reported from Asia with 3 cases reported in Caucasian population⁽²⁾ but very few case reports from India. Clinically it differs from other types of amyloidosis in terms of little or no itching, and absence of photosensitivity. 77% cases show familial history with no sex predilection. Lesions usually start from trunk, very few start from extremities as in our case⁽³⁾. The mean age of onset in familial cases is 6 years⁽⁴⁾, which is very young as compared to our case where patient gave the history of onset of dyspigmentation at around 13-15 years of age. Familial PLCA show dominant or recessive mutations in OSMR (Oncostatin M receptor Beta) or in IL31RA (less common). Whereas ACD show recessive mutation in GPNMB, some show dominant penetrance. This GPNMB encodes a transmembrane glycoprotein type 1 which is vital for melanosome formation and in vitro studies showed its knockdown induced apoptosis in keratinocytes. Although, thorough understanding of its role in ACD is not elucidated yet. UVB and UVC rays induced damage also thought to play some role in ACD^(1,5). Role of EBV has also been postulated in some cases of primary cutaneous amyloidosis⁽⁶⁾.

Histological examination shows homogenous eosinophilic deposits predominantly in papillary dermis with superficial deposition in reticular dermis along with presence of few melanophages. This presence of melanophages may suggest damage to the epidermis in ACD. It is also proposed that this papillary deposit of amyloid stretches the basement membrane, thus decreasing local density of melanocytes in that stretched area⁽¹⁾. ACD lesions are usually positive for AE1/AE3, CK5/6 and CK34βE12 immunohistochemical markers⁽²⁾. Amyloid gives homogenous pink color with Congo Red stain and apple green birefringence on polarised light.

Differential diagnosis are dyschromatosis universalis hereditaria (DUH), xeroderma pigmentosum (XP), dyschromatosis symmetrica hereditaria (DSH), epidermolysis bullosa simplex (EBS) with mottled pigmentation, idiopathic guttate hypomelanosis (IGH) and macular amyloidosis. Macular amyloidosis and IGH can be differentiated dermoscopically (if onset after puberty) with radiating streaks, leaf like projections in former and typical well defined patterns of hypopigmented lesions namely ameboid, patelloid in latter condition⁽⁷⁾. XP has severe photosensitivity and other system involvement like ocular in early age. replace conditions can be differentiated by histopathology.

No modality is 100% effective and there are no well recognised guidelines for treatment of ACD. It is thought to be treatment refractory condition. Various cases are reported to be treated with topical steroids, capsaicin, dimethylsulfoxide (DMSO), keratolytics, oral vitamin E and C and oral acitretin with variable response.

CONCLUSION

After the description of the disease in 1970, there is rise in reporting of the disease, specially from Asian region. Our case also signifies the importance of familial presentation of the disease with no sex predilection but with significant late onset of age as reported in earlier familial cases with a lapse of almost 7-9 years, signifying the varying age of presentation. More and more cases will give better understanding of epidemiologic profile and exact pathological factors implicated.

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