Familial Amyloidosis Cutis Dyschromica: a Case Report

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Abstract- Amyloidosis cutis dyschromica (ACD) is a rare form of macular amyloidosis characterized by hypo and hyperpigmented macules. Here we described a 20 year old girl with diffuse hypo and hyperpigmentation since she was four years old. Five other members of her family are also involved. Biopsy of hyperpigmented lesions revealed increase of melanin in the basal layer, pigment incontinence and amorphous eosinophilic masses stained positive with Congo red in the papillary dermis. The histopathologic findings were consistent with amyloidosis cutis dyschromica. Other investigations were normal. Dermatologists should consider amyloidosis cutis dyschromica when visit a patient with diffuse hypo and hyperpigmentation.

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Keywords: Amyloidosis; Congo Red; Hypopigmentation; Hyperpigmentation

Introduction

Amyloidosis cutis dyschromica (ACD), that was first described by Morishima in 1970, is an uncommon variant of primary cutaneous amyloidosis, which is characterized by the following features: (i) dotted, reticular hyper pigmentation with hypo pigmented spots without papulation almost all over the body (ii) no or little itchy sensation (iii) onset before puberty and (iv) small foci of amyloid closely under the epidermis (1, 2). In this paper we reported a case of amyloidosis cutis dyschromica with family history.

Case Report

A 20 year old girl presented to our clinic because of mildly pruritic diffuse hypo and hyper pigmentation on her trunk and extremities. Her problem started since she was 4 years old. At first hyper pigmentation appeared on trunk and gradually both upper and lower extremities also involved. Simultaneously hypo pigmented macules developed and increased in number (Figure 1a and 1b). The patient was born to consanguineous parents. She didn't have a history of severe sun exposure, other systemic and cutaneous disease except for mild iron deficiency anemia. Similar skin lesions were also present in her aunt, mother, sister and two of her nephews.

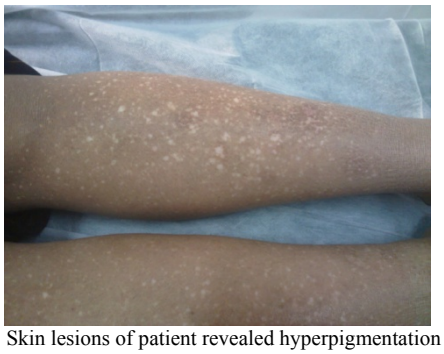

Figure 1a. Skin lesions of patient revealed hyperpigmentation with numerous hypopigmented macules on both legs

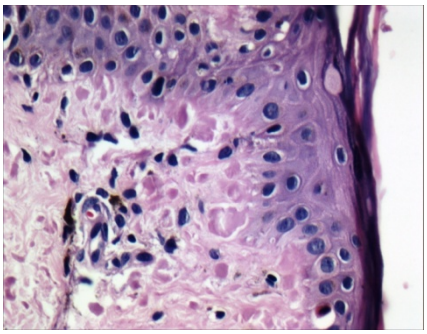

Figure 1b. Skin biopsy specimen from patient characterized by eosinophilic globular deposits of amyloid in the papillary dermis (H&E Staining, original magnification: x400)

In cutaneous examination diffuse hyper pigmentation surrounding hypo pigmented macules were seen on whole trunk and extremities. Dorsal aspect of both hands and feet were spared. Patient's hair, nails,

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teeth, mucosa, palms and soles were normal. Systemic examinations were unremarkable.

Skin biopsy taken from hyperpigmented lesions revealed mildly hyperkeratotic epidermis with scattered apoptotic bodies and basal vacuolar change along with small hyaline deposits of amyloid in the papillary dermis. There was a few melanophages beneath the epidermis and a sparse lymphohistiocytic infiltrate around small vessel (Figure 2a). The Congo red stain demonstrated pink-red colored amyloid materials in the papillary dermis (Figure 2b).

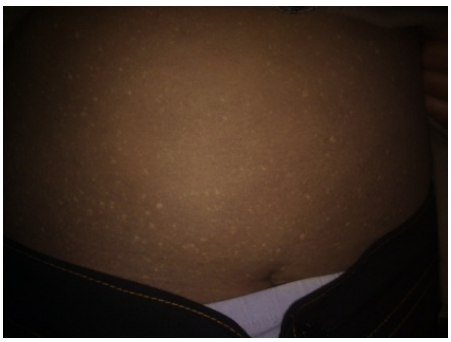

Figure 2a. Generalized hyperpigmentation intermingled with numerous hypopigmented spots on the trunk

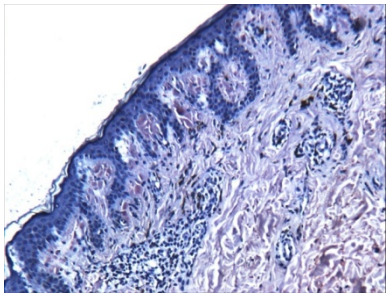

Figure 2b. Congo red staining revealed pink-red Congophilic deposits of amyloid in the papillary dermis
(original magnification: x100)

Laboratory examinations including routine blood test, biochemical profile, urine protein excretion, chest X-ray and abdominal ultrasound were normal. According to clinical and histopathological findings the diagnosis of amyloidosis cutis dyschromica was made.

Discussion

Amyloidosis cutis dyschromica is presumed to be a familial disorder with unknown pathogenesis. The etiology of primary cutaneous amyloidosis is believed to be multifactorial, including frictional and environmental (e.g. UV radiation) epidermal damage, immunology, and other factors. The source of the amyloid material is controversial. It is assumed that repeated damage by sun exposure with impaired ability of DNA repair results in keratinocytes destruction and triggers apoptosis (3-6), but in our cases there was no sun exposure history and they had overt lesions in unexposed locations. Here we reported familial occurrence of disease which presented in six members of this family. There are few reports of familial ACD in the literature (4,7-9) and only Choonhakarn and Wittayachanyapong had reported six familial cases of amyloidosis cutis dyschromica, one of them also had concomitant brownish lichenoid papules on the shins (3).

Main differential diagnoses are dyschromatosis universalis hereditaria, poikiloderma-like amyloidosis and xeroderma pigmentosum (8). Hypo and hyperpigmented macules are found in a generalized distribution in the dyschromatosis universalis hereditaria; it begins before puberty, at an average age of 6 years old. However, histologically it can be differentiated from ACD (10). Poikiloderma-like amyloidosis is associated with light sensitivity, short stature and blister formation or palmoplantar hyperkeratosis. Xeroderma pigmentosum may have a similar dyspigmentation but with marked photosensitivity. Also, there is evidence of premature actinic damage beginning in infancy and early childhood, and skin cancers often develop during the first decade of life (7).

In conclusion ACD should be considered in the differential diagnosis of patients with hypo and hyperpigmented macules. Histopathological examination is useful for confirming the diagnosis. Evaluation of family members is important for diagnosis of familial disorder.

References

1. Morishima T. A clinical variety of localized cutaneous amyloidosis characterized by dyschromia (amyloidosis cutis dyschromica). Jpn J Dermatol Series B 1970;80(1):43-52.
2. Moriwaki S, Nishigori C, Horiguchi Y, et al. Amyloidosis cutis dyschromica: DNA repair reduction in the cellular response to UV light.Arch Dermatol 1992;128(7):966-70.
3. Choonhakarn C, Wittayachanyapong S. Familial amyloidosis cutis dyschromica: six cases from three families. J Dermatol 2002;29(7):439-42.
4. Vijaikumar M, Thappa DM. Amyloidosis cutis dyschromica in two siblings. Clin Exp Dermatol 2001;26(8):674–6.
5. Bourke JF, Berth-Jones J, Burns DA. Diffuse primary cutaneous amyloidosis. Br J Dermatol 1992;127(6):641-4.

6. Wong CK. History and modern concepts. Clin Dermatol 1990;8(1):1-6.
7. Karadagi AS, Guler Şimsek G. Familial amyloidosis cutis dyschromica. Turk J Med Sci 2010;40(1):151-4.
8. Garg T, Chander R, Jabeen M, et al. Amyloidosis cutis dyschromica: a rare pigmentary disorder. J Cutan Pathol 2011;38(10):823-6.
9. Yang W, Lin Y, Yang J, et al. Amyloidosis cutis dyschromica in two female siblings: cases report. BMC Dermatol 2011;11(1):4.
10. Bulbul Baskan E, Tunalı S, Turan A, et al. Dyschromatosis universalis herediteria: a case report from Turkey. T Klin J Dermatol 2008;18(3):191-4.