Familial Amyloidosis Cutis Dyschromica: a Case Report

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Received: 25 Nov. 2012; Accepted: 29 Apr. 2013

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.

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.

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,
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
cutis dyschromica: DNA repair reduction in the cellular
3. Choonhakarn C, Wittayachanyapong S. Familial
amyloidosis cutis dyschromica: six cases from three
4. Vijaikumar M, Thappa DM. Amyloidosis cutis
dyschromica in two siblings. Clin Exp Dermatol
5. Bourke JF, Berth-Jones J, Burns DA. Diffuse primary