Case Report

Amyloidosis cutis dyschromia: a rare form of primary cutaneous amyloidosis

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Dermatology Online Journal 20 (4): 5

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Abstract

Amyloidosis cutis dyschromia is a rare form of primary cutaneous amyloidosis. Amyloid deposition in the skin occurs without systemic manifestations and produces hypopigmented and hyperpigmented macules. A 19-year-old woman is presented with progression of this condition over 16 years.

Introduction

Amyloidosis cutis dyschromia (ACD) also known as familial generalized dyschromic amyloidosis cutis is a rare disorder of pigmentation; the abnormal gene or locus is unknown. It is considered to be a type of primary cutaneous amyloidosis, which is characterized by the deposition of amyloid in the skin with absence of systemic deposits. To date, there are about 26 cases reported in the medical literature. Asians are affected more than other ethnic groups. The disease presents with mottled pigmentation formed by hyperpigmented and hypopigmented macules and patches in a generalized distribution [1-14]. We report a case of ACD with diffuse symmetrical involvement of the skin sparing the face, palms, and soles along with xerosis of the involved areas. Histopathology showed amorphous eosinophilic deposits in the papillary dermis that stained positive for Congo red.

Case synopsis

A 19-year-old woman presented with progressive diffuse symmetric hyper and hypopigmented patches and macules that started to develop at the age of three years. The changes were asymptomatic and the patient denied any history of photosensitivity. Her past medical history was negative for any significant medical problems or disease. No history of prolonged or repeated infections could be elicited. The patient denied using any long-term medications, topical creams, or preparations before the onset of the skin changes. She had normal growth and development as a child. Her parents are first-degree cousins. One of her paternal aunts had a similar pattern of pigmentation.

Physical examination showed multiple hyperpigmented patches with intermingled multiple non-atrophic hypopigmented macules involving all the body, but sparing the face, palms, and soles. Some of the areas involved were dry and scaly (Figure 1-5). The dermatoglyphs were intact. No dental or nail abnormalities were noticed. Scalp and hair examination was within normal limits. Serum protein electrophoresis was negative and 24 hours urine collection for arsenic was within normal range. Skin biopsy from involved skin revealed eosinophilic globules in the papillary dermis (Figure 6-7). Congo red stain was positive (Figure 7-8). Topical emollients and steroids improved the skin dryness but not the dyschromia.
Figures 1-4. Hyperpigmented patches and macules with intermingled hypopigmented macules along with xerosis

Figure 5. HE X10: amorphous eosinophilic deposits (Amyloid) in the papillary dermis
Figures 6. HE X100: amorphous eosinophilic deposits (Amyloid) in the papillary dermis

Figure 7. Congo red stain: Amyloid deposits in the papillary dermas showing positive staining.

Discussion

Primary cutaneous amyloidosis is a group of cutaneous disorders that are characterized by deposition of amyloid in the skin without deposition in any other organs [3]. ACD is a rare form of primary cutaneous amyloidosis that presents with diffuse hypo and hyperpigmented patches and macules with occasional itching. Onset is almost always before puberty [1-14]. Atrophy and blisters are rarely seen [4].

The disease is generally not associated with systemic symptoms. However, there is one case report in which ACD was associated with generalized morphea [5], and another case report involved two siblings suffering spasticity, motor weakness, and atypical Parkinsonism [6].

Histopathological examination shows amyloid deposits in the papillary dermis. Amyloid stains positive for crystal violet and Congo red. In rare instances, amyloid is not detected with the former stains and immunochemistry for anti-cytokeratin antibodies should be obtained if suspicion remains [1-4].

The pathogenesis is poorly understood but the disease is thought to be caused by an inherited increase in sensitivity to UVB leading to defects in DNA repair. Amyloid is formed as a result of keratinocyte degeneration [15].

Ozcan et al used oral acitretin (0.75 mg/kg/day for 3 months) with significant improvement in hyperpigmentation [7]. In another retrospective study published by Qiao et al, 10 patients were treated with oral vitamin E and vitamin C with minimal improvement. Three patients received 20 mg of oral acitretin daily for three months. A good response was observed in two of the three patients. Long term follow up of patients treated with acitretin was not published. No other treatments to date are successful to treat ACD [8].

Other causes of dyschromia should be excluded. The differential diagnosis is summarized in Table 1.

Table 1. Clinical differential diagnosis of ACD

<table>
<thead>
<tr>
<th>Condition</th>
<th>Clinical Presentation</th>
<th>Associated Features</th>
<th>Histopathology</th>
<th>Treatment</th>
<th>Other</th>
<th>References</th>
</tr>
</thead>
<tbody>
<tr>
<td>Congenital</td>
<td></td>
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<tr>
<td>Dyschromatosis universis hereditatis</td>
<td>Generalized</td>
<td>hyperpigmented and hypopigmented macules distributed in a reticulate pattern</td>
<td>Case reports of mental retardation, learning difficulties, seizures, eye abnormalities, dwarfism, juvenile diabetes and thrombocytopenia</td>
<td>Focal increase or decrease in melanin in the basal layer.</td>
<td>No treatment available: Genetic Counseling</td>
<td>AD, AR,</td>
</tr>
<tr>
<td>Dyschromatosis symmetrica hereditaria (reticulate hyperpigmentation of Skin)</td>
<td>Hyperpigmented and hypopigmented macules distributed on the face and the dorsal aspects of the extremities</td>
<td>Case reports of dental abnormalities, neurological abnormalities and hypopigmented hairs</td>
<td>Increased/decreased melanin in hyperpigmented macules and hypopigmented hair respectively.</td>
<td>No treatment available: Genetic Counseling</td>
<td>AD, sporadic</td>
<td>[19,20]</td>
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<tr>
<td>Poliosis/leukoderma-like cutaneous amyloidosis</td>
<td>Poliosis/leukoderma like skin lesions, whitened papules, occasional blister and palmo/plantar keratosis</td>
<td>Short stature, photosensitivity</td>
<td>Amyloid deposition in the pigmented lesions and the leukoderma papules.</td>
<td>No treatment available: Genetic Counseling</td>
<td>AD, X-linked</td>
<td>[21-24]</td>
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<tr>
<td>Dyschromatosis congenita</td>
<td>Hyperpigmentation involving the upper chest and upper arms associated with hyperpigmented macules, occasional tinea capitis and epidermal atrophy, i.e. poliosis/leukoderma.</td>
<td>Leukoplakia, nail dystrophy, palmar plantar hyperhidrosis</td>
<td>Epidermal thinning, telangiectasias, areas of increased pigmentation within the basal layer</td>
<td>No treatment available: Genetic Counseling</td>
<td>X-linked recessive 90%</td>
<td>AD, AR &lt;10%</td>
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<tr>
<td>Xeroderma pigmentosum</td>
<td>Hyperpigmented macules in sun-exposed skin (Solar keratosis)</td>
<td>Increased risk of skin cancers, solar abnormalities, neurological abnormalities, developmental delays</td>
<td>Increased or decreased melanin in the basal layer, epidermal atrophy or hyperkeratosis, solar elastosis</td>
<td>Photo protection, oral retinoids</td>
<td>AR</td>
<td>[26-28]</td>
</tr>
<tr>
<td>Acquired</td>
<td>Hyperpigmented patches and macules &quot;sunlight pigmentation&quot;, areas of depigmentation</td>
<td>Hyperkeratosis of palms and soles, cutaneous and internal malignancies</td>
<td>Basal cell pigmentation in hyperpigmented lesions.</td>
<td>Chemotherapy</td>
<td>Acquired</td>
<td>[29,30]</td>
</tr>
</tbody>
</table>

AD, autosomal dominant; AR, autosomal recessive
Conclusion

We report a woman with ACD. Some of the conditions in the clinical differential diagnosis include cutaneous diseases presenting with congenital or acquired dyschromia (Table 1). The diagnosis of ACD may be suspected based upon the presence of progressive diffuse hypo and hyperpigmented patches and macules with or without itching before the onset of puberty. The diagnosis is confirmed by histopathological examination, which shows amyloid deposits in the papillary dermis that stain positively for amyloid stains such as crystal violet and Congo red. To date, a small number of patients have been successfully treated with acitretin. However, this was not an option in our patient or in women of childbearing potential.

References
