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FAMILIAL BIPHASIC CUTANEOUS AMYLOIDOSIS

K G Singh, Anoop K Tripathi, S C Gupta, A K Bajaj and Rajeev Sharma

A Bengali family had familial biphasic cutaneous amyloidosis in 6 of the 11 members in 3 generations. The propositti developed intense itching, thickening and pigmentation of the skin associated with discrete, brownish-black papules bilaterally on the legs, ankles, popliteal fossae and extensor aspects of the thighs. Face, neck, scalp and oral mucosa were spared. There was hyperkeratosis of the palms and soles. None of them had lymphadenopathy, macroglossia, hepato-splenomegaly, neuropathy or gingival hypertrophy.

Key words: Familial, Biphasic cutaneous, Amyloidosis.

Primary cutaneous amyloidosis (PCA) is characterised by deposition of amyloid in previously normal skin without any systemic involvement. Clinically, it manifests as pruritic macules or papules. Sometimes, both types of lesions are present simultaneously in the same person, and this type is called the biphasic form.[1] Though the rare familial type of PCA is reported in the world literature,[2-6] the paucity of such reports from the Indian subcontinent has prompted us to document the present case.

Case Report

A Bengali female, 22 years of age, developed intense itching, thickening and pigmentation of the skin during the last 19 years. The itching tended to increase in winters. Over the past one year, there had been marked thickening of the skin over the dorsa of the feet and knuckles of the hands. Discrete brownish-black papules, 1-4 mm in size were present bilaterally on the legs (Fig. 1), ankles, popliteal fossae and extensor aspects of the thighs. On the lateral side of upper extremities, upper back and chest, there were several, poorly demarcated, tenuous, brown macules, irregularly interspersed with normal skin and hypopigmented areas. Face, neck, scalp and oral mucosa were spared. There was hyperkeratosis of the palms and soles. There was no lymphadenopathy, macroglossia, hepato-splenomegaly, neuropathy or gingival hypertrophy. There was no evidence of nephrotic syndrome or cardiac failure.

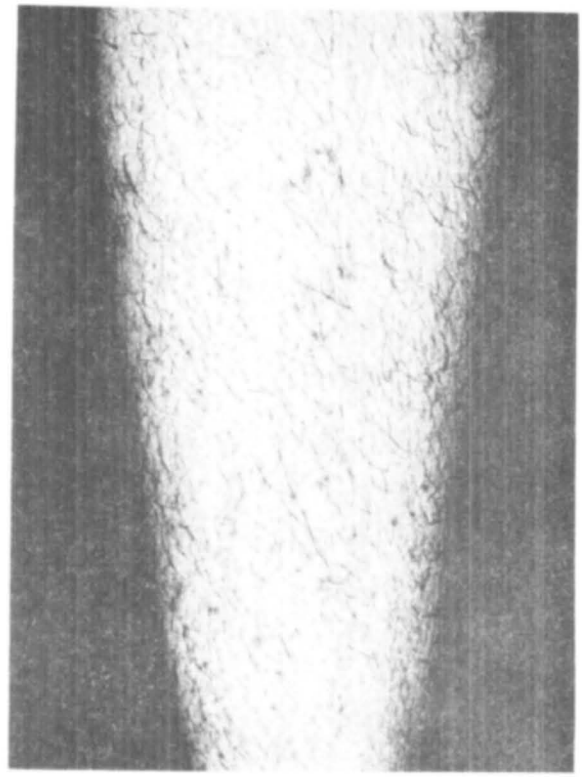

Fig. 1. Discrete, brownish black papules on the legs.

From the Departments of Dermatology and Venereology, and Pathology, MLN Medical College, Allahabad (UP), India.

Address correspondence to: Dr. S.C. Gupta.

The blood counts and urine analysis were normal. Bence Jones protein was absent. Serum levels for IgG, IgA and IgM were 1586, 255.50 and 216.07 mg percent as against their normal ranges of 1030 to 1251, 198 to 238 and 100 to 142 mg percent respectively.

Two biopsy pieces, one each from a papule on the pretibial area, and a macule on the lateral side of left upper thigh were taken. The former showed pronounced hyperkeratosis and moderate acanthosis with discrete masses of faintly eosinophilic homogeneous material in the dermal papillae (Fig. 2). The latter showed normal epidermis with deposits of amyloid material in the papillary dermis. In both the sections, the amyloid showed metachromasia with crystal violet. Various topical medicaments could neither appreciably alleviate the pruritus nor the morphology of the lesions was altered.

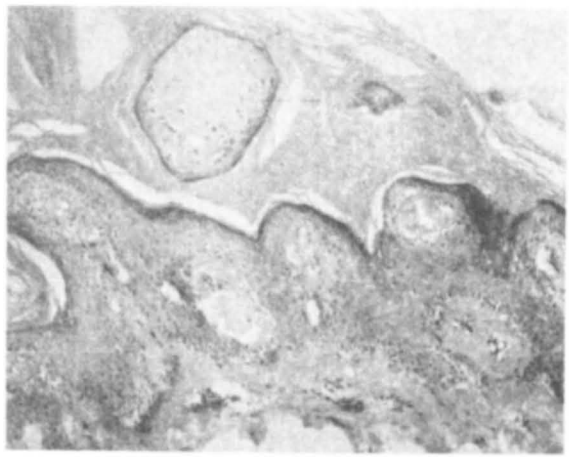

Fig. 2. Pronounced hyperkeratosis and moderate acanthosis with discrete masses of faintly eosinophilic homogeneous material (amyloid) in the dermal papillae (H&E × 100).

Family study of 3 generations could be possible. Six out of 11 members including propositus were found to be affected. Four were males and two females (Fig. 3).

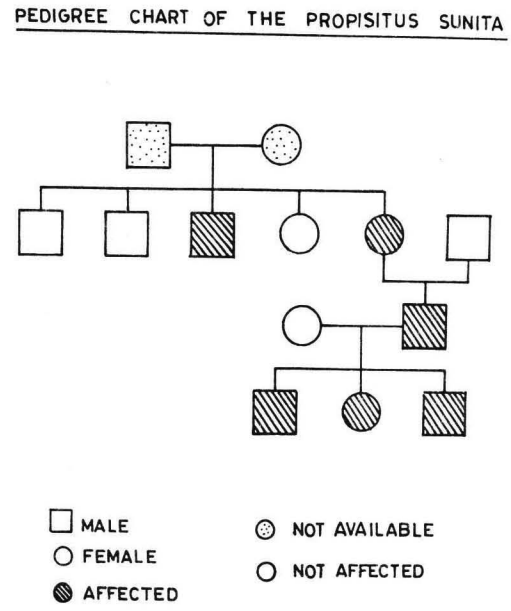

Fig. 3. Pedigree chart of the propositus.

Comments

Familial cutaneous amyloidosis normally manifests either in early childhood[5] or around puberty.[6] In the present case, the disease started at the age of 3 years. The lesions had classical distribution,[5,6] the face and oral mucosa were found to be spared and there was no evidence of systemic amyloidosis.

Brownstein et al[1] for the first time in 1972, proposed the term biphasic for the coexistence of macular and papular forms of the disease in the same person. Since then various reports of the biphasic cutaneous amyloidosis have appeared in the literature.[7,8] Five of the 7 cases of FPCA of Vasily et al[5] and 3 of the 19 cases of Rajagopalan and Tay[6] were of the biphasic types. In the present case also, there were macular as well as papular lesions.

Brownstein et al[1] had opined that papular and macular forms were two presentations of the same disease. The nature of itching, the site of involvement and the susceptibility of the area governed the type of the lesion. They

also found that macular lesions tended to localise on the back and thighs, while papules were more prominent on the legs. The distribution of the lesions in this case was not different from what has already been described.

Serum IgG, IgM and IgA were all found to be elevated. Tasanapradit et al[9] had found elevated serum IgG levels in a high percentage of patients with lichen amyloidosis. On the contrary, Vasily et al[5] had reported no change. The histological findings are in accordance with those of others.[1, 7, 10] Brownstein et al[1] suggested that intense itching was one of the important factors causing the epidermal hyperplasia found in lichen amyloidosus as they were able to convert the papular lesions to macules with almost normal epidermis by therapeutic alleviation of pruritus. But Marta and Mario[11] were of the opinion that macular primary amyloidosis and lichenoid amyloidosis were distinct clinical manifestations of the same disease and itching did not cause transformation of MPA into LPA. In the present case also, like those of Vasily et al[5] numerous therapeutic attempts using topical therapy failed to alter the lesion morphology or relieve the pruritus significantly.

Occurrence of the disease in 6 cases in 3 generations of this family further strengthens the genetic theory of primary cutaneous amyloidosis. Earlier, Isaak,[2] Sagher and Shanon,[3] Tay[4] and Vasily et al[5] had reported cases of familial primary cutaneous amyloidosis. Rajagopalan and Tay[6] studied 19 cases of FPCA in 4 generations of a Chinese family and proved that the disease was transmitted as an autosomal dominant trait with variable penetrance.

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