Case 2b

A 27 year-old woman from Nakornsritammarat

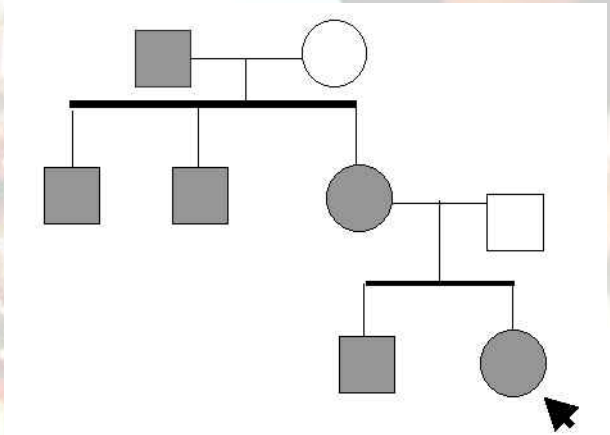
Chief complaint: progressive skin lesions on extremities and trunk.

Present illness She developed gradually progressive discrete minute skin colored papules distributed on both extremities and trunk. The lesions occasionally were pruritus. Application of topical steroids as well as salicylic ointment only resulted in temporary benefit.

Past history Generally she has enjoyed good health.

Family history Her elder brother, mother and grandfather are also similar affected.

The pedigree was shown as following.



Physical examination

General appearance: A healthy looking young woman with.

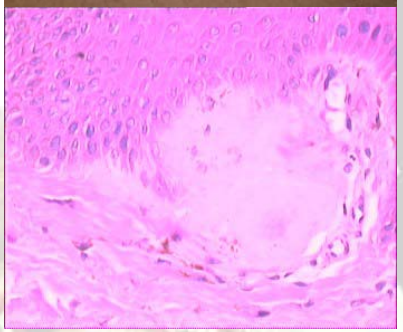
HEENT: no pale no jaundice, no macroglossia

Heart &lungs: normal

Abdomen: soft, no hepatosplenomegaly.

Skin examination

Multiple closely set discrete minute skin colored and hyperpigmented ,hyperkeratotic papules symmetrically distributed on both lower legs, trunk and upper extremities



Histopathology: slide No. S01-9246

- Papillated epidermal hyperplasia and compact hyperkeratosis
- Broadened dermal papillae
- Pale homogenous eosinophillic globules in the dermal papillae
- Melanophage and stallate fibroblasts assocciate within the globules

Diagnosis: Familial primary cutaneous amyloidosis

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Discussion

Primary cutaneous amyloidosis is a relatively common skin disease in Southeast Asia, South America, and Republic of China. Although most case are sporadic with unknown ethiology. Some patients have a family history, suggested that genetic factors may play a role in its pathogenesis. It transmitted in family as an autosomal dominant.

In some case the lesions of amyloidosis are associated with other genodermatoses and with other familial diseases as Hereditary multiple endocrine neoplasia(MEN 2a).dyskeratosis congenita, Paritington's disease and pachyonychia congenita .The syndrome of cutaneous amyloidosis and MEN 2A appears to be a clearly defined autosomal dominant hereditary syndrome. Whether this syndrome can be linked to chromosome 10 is not yet known. In clinical presenting lichen amyloidosis is the most common variant. Histochemically, H&E stain will indicate the diagnosis of amyloidosis , which can be confirmed with crystal violet, stain. There are various treatments both surgical and medical for this condition.

High potency topical or intralesional steroid combine with oral antihistamines can be used for relief symptoms. Oral etretinate or long-term cyclophosphamide appear to be helpful in relieving pruritus but relapsing occurred rapidly after termination. Dermabrasion and other surgical procedure resulted
in a long-term beneficial effect.

Reference

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3. Hartshorne ST : Familial primary cutaneous amyloidosis in a South African family. Clin Exp Dermatol 1999 Nov;24(6):438-42
4. Lee DD, Huang JY, Wong CK, Gagel RF, Tsai SF. Genetic heterogeneity of familial primary cutaneous amyloidosis: lack of evidence for linkage with the chromosome 10 pericentromeric region in Chinese families. J Invest Dermatol 1996 Jul;107(1):30-3