Case 11

A 54-year-old Thai female from Bangkok

Chief complaint: Multiple, mild pruritic widespread, brownish hyperkeratotic papules on both arms, legs, and back for 40 years

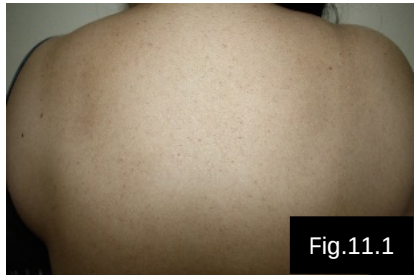


Fig.11.1

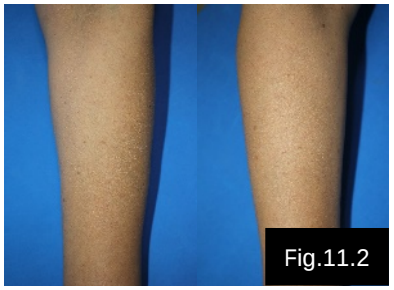


Fig.11.2

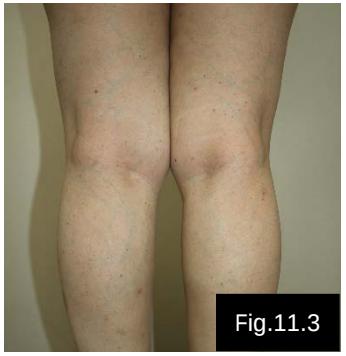


Fig.11.3

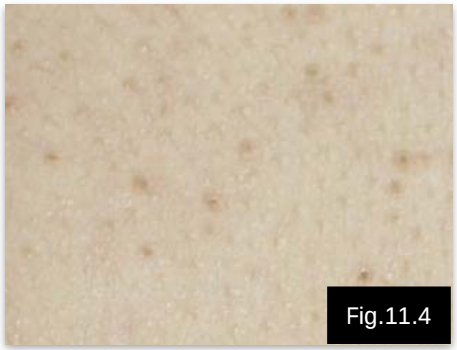


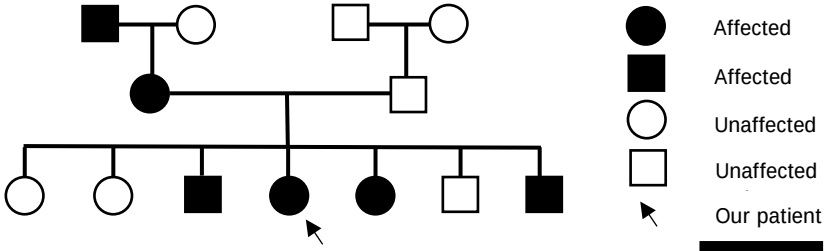
Fig.11.4

Present illness:

The patient visited the hospital with multiple mild pruritic widespread, brownish hyperkeratotic papules located symmetrically on both arms, legs, and back for 40 years. The lesions appeared when she was 12-year-old, initially over the extremities and subsequently spread to entire body. Mucosa, palms and soles were spared. She previously received some topical treatment form the other hospital with no clinical improvement.

Past history: Non-alcoholic Fatty Liver Disease (NAFLD), dyslipidemia (DLP), chronic gastritis, colonic polyp with pathological report of tubular adenoma and bilateral carpal tunnel syndrome

Family history: Information about family members who had similar lesions was noted as shown in pedigree chart. The lesions trended to develop around age of 10. She denied history of consanguinity among her parents (Fig.11.5).



Pedigree chart

Fig.11.5

Physical examination: Other systemic examination revealed normal

Dermatological examination:

- Multiple discrete, comedone-like hyperkeratotic brownish papules located symmetrically on both arms, legs, and back, sparing palms and soles (Fig.11.1-11.4)

Histopathology (S19-013969, left thigh):

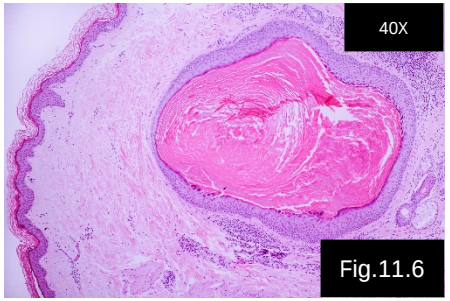


Fig.11.6

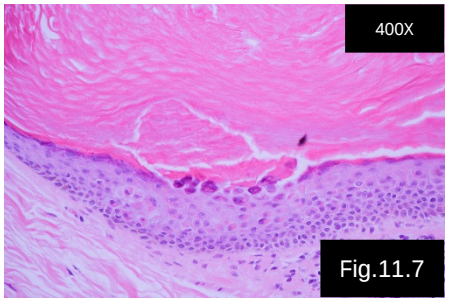


Fig.11.7

- Cystic dilatation with keratotic follicular plugging (Fig.11.6)
- Multiple zones of focal acantholytic dyskeratosis in the cystic epithelium (Fig.11.7)

Diagnosis: Familial dyskeratotic comedones

Treatment and outcome:

- Topical 10% urea cream apply twice daily
- Topical 0.025% retinoic acid apply twice daily
- Cetirizine 10 mg orally once daily
- She reported minimal improvement at 3-month follow-up visit

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Discussion:

Familial dyskeratotic comedones (FDC) is a rare autosomal dominant condition appearing around puberty, characterized by symmetrical, numerous, discrete, disseminated, hyperkeratotic papules and comedones on extremities, trunk, and face sparing palms, soles, and mucous membranes. Hair and nails appear normal.[1-5] The lesions are not spontaneously resolved and sometimes cause pruritus and inflammation.[3] Occurrence in two consecutive generation of this disease suggests autosomal dominant disorder.[1,3] The histopathological examination shows crater-like epidermal invaginations filled with lamella keratinous materials and evidence of dyskeratosis.[1-5] Acantholysis can sometimes be seen.[3] Electron microscopy reveals decreased number of desmosomal attachments within the stratum malpighii.[3] This condition has following distinctive clinical features which are: 1. Lesions clinically resembling comedones, 2. Occurrence in some family members, 3. Presence of dyskeratotic changes on histological examination.[1,4,5] Less than 50 cases of FDC have been reported in the literature.[5]

Similar clinical conditions that should be in differential diagnosis are acne vulgaris, nevus comedonicus, Darier's disease, Kyrle's disease, reactive perforation collagenosis (RPC), keratosis pilaris (KP), perforating folliculitis and a new entity of 'familial disseminated comedones without dyskeratosis. The lesion of nevus comedonicus develops shortly after birth or before age of 10 years, containing closely arranged, dilated follicular opening with keratinous plugs resembling classical comedones.[6] It usually occurs unilaterally in group, or bands, that present a 'sievelike' apperance, predominately over face and neck.[1] The Extensive bilaterally presentation can involve palms and soles. Large follicles containing lamellated keratin but absent of hair shafts can be seen in histopathology without dyskeratosis.[6] Darier's disease presents with follicular and extrafollicullar greasy, hyperkeratotic papules and plaques in seborrheic areas. The lesion also involves oral mucosa and nails. Diagnostic histopathology shows dyskeratosis with corps ronds and grain, suprabasal acantholysis and vill.[3] Acne vulgaris can be rule out by distribution of the lesion and histopathology difference. Keratosis pilaris has some similar histologic feature but lacks of dyskeratotic changes and has different clinical picture. Kyrle's disease, RPC, and perforating folliculitis can be easily differentiated by histopathology.[1]

In our case, the clinical presentation, positive family history and distinctive histopathology most suggest familial dyskeratotic comedones as diagnosis.

Management is difficult in this disease. Topical and oral retinoic acid derivative are ineffective and may aggravate pruritus. The pathophysiological process in FDC could be different from that of normal comedones in acne, thus explaining its lack of response to retinoid treatment. Unrewarding result can be explained by different pathophysiological process in FDC. However, sun exposure and carbon dioxide laser have shown good results.[3,7] Our patient received

topical retinoids, keratolytics and symptomatic treatment for relived pruritus but shown minimal improvement.

Data regarding the long-term follow-up of these patients are lacking.[8] Further studies might explore the molecular pathology, which may widen our gaze regarding the better treatment modalities.

References:

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