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Familial Dyskeratotic Comedones – Case report in four families - A Rare entity.

KEYWORDS: Comedones, Familial comedones, dyskeratosis, Autosomal Dominant.

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ABSTRACT *Familial dyskeratotic comedones(FDC) is a rare asymptomatic Autosomal Dominant condition with distinctive clinical and histopathological features. The condition usually starts at puberty as comedones or hyperkeratotic papules that are widespread mostly involving the trunk, arms and face. The lesions are usually asymptomatic showing worsening with time causing cosmetic disfigurement to the patient. Histology shows invagination of the epidermis with a lamellar keratinous plug and focal evidence of dyskeratosis. This condition shows poor response to treatment. We report four cases of this condition showing strong family history of the same.*

Introduction:

Familial dyskeratotic Comedones is a rare asymptomatic, autosomal dominant disorder. Rodin et al.[1] first reported this condition in 1967. Very few cases were reported since then. Only 15 patients from seven families have been reported in the literature.[2,3] It is clinically characterized by numerous, discrete, disseminate, hyperkeratotic papules, and comedones.[1,3,4,5,6,7,8,9,10,11]. Histopathology shows crater-like invaginations filled with keratinous material and evidence of dyskeratosis.[3,4,5,6,7,8,9]

Case Reports

Case 1

Male 58 years presented with 20 year history of multiple asymptomatic comedones, hyperkeratotic papules over trunk and face. Few pustules over the trunk. No similar history in his parents. His son who is 30 years started developing similar lesions for the last 10 years.

Case 2

A 52 year old male presented with a 25 year history of asymptomatic comedones , hyperkeratotic papules all over the body including the buttocks and Pock like scars over the face. There are Keloids over the chest. Few papules over the trunk show inflammatory changes. Patient gives history of seeing similar lesions in his father. Wife 51 years has similar lesions since 6 years. The patient has a history of 2° consanguinous marriage.

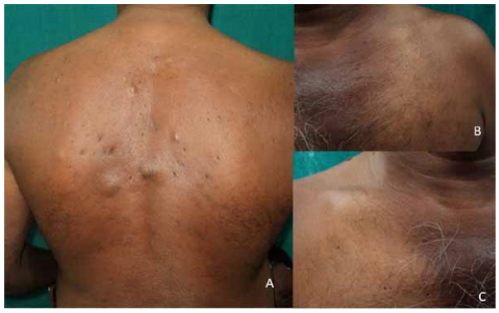
Case 3

A 22 year Unmarried, Female with a 10 year history of asymptomatic comedones and hyperkeratotic papules over trunk, arms and face. Face shows Pock like scars. There is history of similar lesions in her father.

Case 4

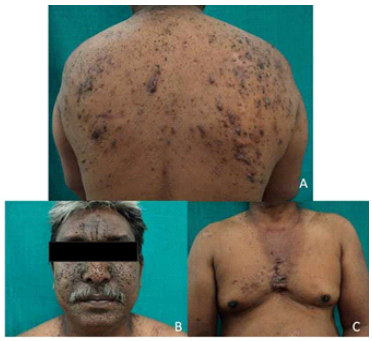
A 27 year Female with 5 year history of asymptomatic comedones over buttocks gradually increasing in intensity. History of similar lesions in her mother and female cousins who developed the lesions during their adolescence.

Tables/Charts:



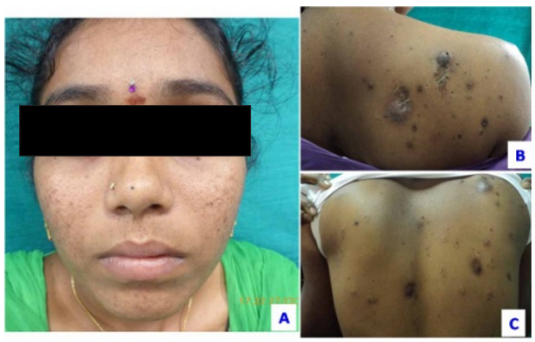
Case-1

Case 1- A. Multiple comedones, pustules on back B&C- comedones on chest.



Case-2

A. Comedones and Pustules on back
B. Comedones and Pock like scars on face
C. Keloids and Comedones on chest



Case-3

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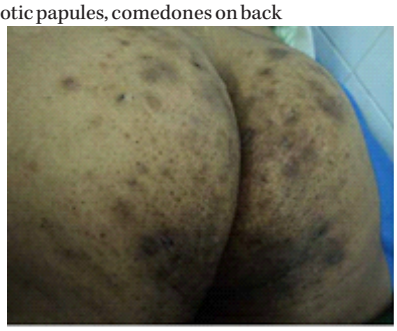
Case3

A. Multiple comedones and pock like scars on face B.& C.- Hyperkeratotic papules, comedones on back



Case-4

Case4- Multiple comedones on both buttocks

Discussion:

Carneiro et al. [4] proposed the term FDC when he first described a family of four members affected with this rare entity, which was based on the following distinctive features:

1. Lesions clinically resembling comedones
2. Occurrence in some family members
3. Presence of dyskeratotic changes on histological examination

The lesions usually appear around puberty and show a predilection for the trunk, arms, legs, face and shaft of the penis, sparing the glans, palms and soles. [10]Histologically, it is characterized by dyskeratosis and invaginations into the dermis, occasionally acantholysis may be seen. Dyskeratosis may not be seen in all patients. [13]

All our four cases have very strong family history of similar lesions. Like in many other studies, the occurance of the lesions was mostly in mid or late adolescence in all our cases.

Differential diagnosis includes Kyrle's disease, Reactive Perforating collagenosis, Keratosis pilaris, Perforating folliculitis, Nevus comedonicus, and Acne vulgaris.[12]

Treatment has always been unrewarding. Various treatment modalities including topical retinoids and oral isotretinoin have proved to be ineffective. [3,7,8,11] The pathophysiological process in FDC could be different from that of normal comedones in acne, thus explaining its lack of response to retinoid treatment. However, frequent sun exposure and carbon dioxide laser [7] have shown promising results.

Summary:

While FDC is a rare entity, its presence may be missed because of its close clinical similarity with Acne and Kyrle's disease. However a careful history and examination of the family members may be rewarding. The treatment for this condition remains empirical and largely unsatisfactory.

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