

Familial diffuse comedones: A Case Report

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ABSTRACT:

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Familial diffuse comedones is a rare autosomal dominant inherited dermatosis that usually appears in adolescents and characterized by multiple widespread comedone-like lesions. The distinct histology shows follicle filled with keratin plugs with no dyskeratotic and acantholytic change. The disease has good prognosis with no associated systemic involvement. No effective treatment is established. We report two unrelated families of Familial diffuse comedones from our institute.

Key word: Familial diffuse comedones, comedone-like, opened comedones

บทคัดย่อ:

สาลิณี โรจน์หิรัญสกุล, ธัญญา เตชะพิเชฐวานิช, ชนิตวณิช วิชาญชากร รายงานผู้ป่วยโรค FAMILIAL DIFFUSE COMEDONES วารสารโรคผิวหนัง 2013; 29: 236-241.

สาขาวิชาตจวิทยา ภาควิชาอายุรศาสตร์ คณะแพทยศาสตร์โรงพยาบาลรามาธิบดี มหาวิทยาลัยมหิดล

Familial diffuse comedones เป็นโรคที่ถ่ายทอดทางพันธุกรรมที่ไม่ค่อยมีรายงานและพบไม่บ่อย ลักษณะของรอยโรคทางคลินิกคล้ายสิวอุดตันที่สามารถพบได้ทั่วร่างกาย รอยโรคมักเกิดในช่วงเข้าสู่วัยรุ่น ลักษณะทางจุลพยาธิวิทยามีความจำเพาะสามารถแยกจากโรคที่คล้ายคลึงกันได้ โรคนี้มีพยากรณ์โรคดี มักไม่พบความผิดปกติในระบบอื่นๆ ของร่างกาย นอกจากรอยโรคทางผิวหนัง อย่างไรก็ตามเนื่องจากโรคนี้เป็นโรคหายากมีรายงานน้อยจึงยังไม่มีการรักษาจำเพาะในโรคนี้ รายงานนี้เป็นกรณีนำเสนอผู้ป่วยโรค Familial diffuse comedones จาก 2 ครอบครัวที่ไม่เกี่ยวข้องกันทางพันธุกรรม

คำสำคัญ: โรค Familial diffuse comedones, สิวอุดตัน, โรคคล้ายสิว

Case report

Case 1

A 17 year-old Thai female from Bangkok presented with recurrent multiple cysts at face, posterior neck, chest, shoulders and upper back. Initially, she was diagnosed as recurrent epidermal cysts and received multiple courses of antibiotics and surgical drainage from surgical department outpatient unit. Subsequently, she was transferred to our dermatological department because one of the surgeons obtained her family history of identical lesions. Additional history taking revealed that she had multiple asymptomatic blackish comedone-like papules since twelve; some papules become progressively enlarged, inflamed and painful, leaving discrete pock-like scars especially on her face and back. She was otherwise normal. She denied exposure to acnegenic substances, herbs, or medications. Many members of her family had similar lesions to the patient as shown in family pedigree. (Figure 1) The onset of lesions in all involved members was around the age of twelve as in our patient.

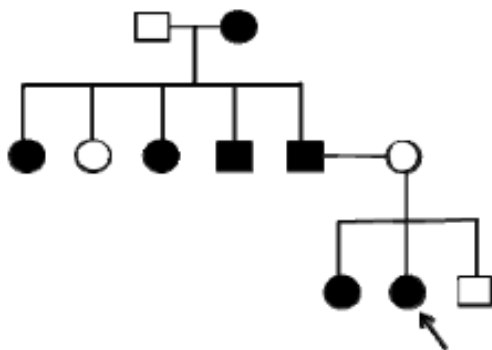


Figure 1 Family pedigree of case 1

Physical examination:

The skin examination revealed widespread, symmetrically scattered, comedone-like papules at face, trunk, arms and legs with discrete inflamed nodules and comedones over the back. Multiple pock-like scars were seen over the face and the back. (Figure 2)

Neurological, musculoskeletal, and eye examinations were normal.



Figure 2 Comedone-like papules on upper back and both forearms

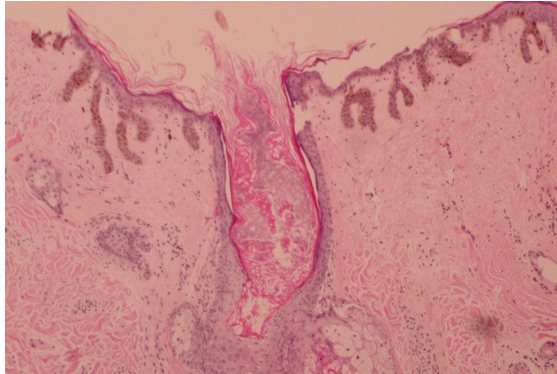


Figure 3 Follicular dilatation and delling of para-follicular epidermis lined by thin atrophic epithelium, with branching, pigmented, elongated and interconnecting strands. (H&E, x100)

Investigation:

Histopathology of the lesion from the comedone-like lesion showed follicular dilatation and delling of para-follicular epidermis lined by thin atrophic epithelium, with branching, pigmented, elongated and interconnecting strands. There was an infiltration of lymphocytes, melanophages, and fibrous in the underlying dermis. (Figure 3)

Case 2

A 16 year-old Thai female from Patumtanee came to our clinic with two-year history of multiple widely spread comedone-like papules. Her lesions initially appeared at her face and subsequently spread to upper chest, upper back both upper arms and also both legs. Apart from these skin lesions, multiple pock-like scars were seen over the face and the back. The lesions started as pinpoint dark papules, which gradually increased in number and extent of involvement as the

patient grew older. When fully formed, the lesions can be measured around 0.5 to 1 cm in size, which sometimes formed into painful swelling inflamed nodules over the face and back. The patient had not received any treatment and was in good health and otherwise normal. Further family history revealed that her father and older sister had similar lesions.

Physical examination:

There were multiple dilated follicular openings filled with blackish plugs diffusely involving the face, chest, upper back and upper arms. Some lesions developed inflamed erythematous nodules with central ostia. Discrete pock-like scars are seen. (Figure 4)

No abnormality of eye, musculoskeletal, and neurological system was detected by physical examination.



Figure 4 Multiple comedone-like lesions, inflamed nodules, and pock-like scar on right upper back

Investigation:

Skin biopsy was obtained from comedone-like lesion. There was a keratinous follicular plugging, and cystic dilatation of follicular epithelium lined by thin epithelium with branching and elongated rete ridges. The underlying dermis had few lymphocytic infiltration and fibrosis.

Discussion

Familial diffuse comedones is a very rare inherited dermatosis, characterized by widespread comedone-like lesions that can involve the trunk, arms, legs and face, sparing the palms and soles, in symmetrical distribution. The comedone-like lesions usually appear around puberty and progressively increase with age. The pattern of inheritance of this disease is suggested to be an autosomal dominant disease.¹ However, there are only few case reports of familial diffuse comedones.^{1, 2} To the best of our knowledge, only two families have been reported in the English language medical literature.

In 1967, Rodin et al described a 42-year-old woman who had widely distributed multiple comedones since adolescence; her mother and grandmother were also affected.² In 1978, Cantu' et al reported a large family in which 16 members had multiple relapsing comedones distributed on face, neck, anterior and posterior thorax, abdomen and extremities.¹ The onset was as early as 10 years of age, the activity of disease increased during adolescence. The pedigree analysis showed a definite autosomal dominant pattern of

inheritance since both male and female members of four consecutive generations were affected, including two instances of male-to-male transmission. The histopathology in previous reports disclosed follicles filled with keratin plugs. The keratin was lamellar without parakeratosis and the follicular ostium was dilated. No dyskeratotic change was observed.

We believe that there are three reasons why very few cases of familial diffuse comedones have been reported. Firstly, there are often confused with familial dyskeratotic comedones.^{3,4} In familial dyskeratotic comedones, the comedone-like lesions are less extensive and demonstrate acantholytic-dyskeratotic changes in the wall of follicular-like structure on histopathology. Second reason is that the disease can be sporadic. We found many reports of non-inherit case which had clinically similar to familial diffuse comedones under difference names such as "extensive nevus comedonicus"^{5,6}, "idiopathic disseminated comedones"⁷. Finally, since comedone is also a manifestation of acne vulgaris, a very common skin disease, it is often misdiagnosed as acne.

The differential diagnosis of widespread comedones is summarized in table 1. The two main differential diagnoses of familial diffuse comedones are familial dyskeratotic comedones and nevus comedonicus. In familial dyskeratotic comedones, clinical manifestation is similar to familial diffuse comedones, but differs in histological finding which shows follicle-like invagination with acantholytic and dyskeratotic changes

in the walls of a follicle-like structure.^{3,4} Nevus comedonicus is another disease with multiple

comedones, characterized by a group of comedones in linear or blaschko's line, and usually unilateral.⁸

Table 1 Differential diagnosis of widespread comedones

Disease	Inheritance	Distribution	Morphology /Arrangement	Histology	Systemic association
Familial diffuse comedones	Autosomal dominant	Generalized face, trunk, upper, and lower extremities	Diffuse open comedone-like lesions	Dilated follicle filled with keratin, no dyskeratosis	None
Nevus comedonicus	Sporadic	Usually unilateral in linear or blaschko's line of face trunk, and proximal extremities	Group of closely set comedone-like lesions	Same as familial comedones	Skeletal anomalies (scoliosis, hemivertebrae), seizure, cataracts
Familial dyskeratotic comedones	Autosomal dominant	Generalized, mainly trunk, upper, and lower extremities	Diffuse papules with central keratotic plugs	Invagination of epidermis filled with keratin, acanthosis and dyskeratosis in wall of follicle-like structure	None
Solar comedones (Favre- Racouchot disease)	Acquired	Sun-exposed area, mainly face	Comedones on background of solar elastosis	Large thin wall open and closed comedones with marked solar elastosis	None
Chloracne	Acquired	Face, postauricular, back, axillae	Open comedones, milia, cysts	Non-inflammatory follicular dilatation and plugging	CNS(headache, irritability), peripheral neuropathy, hepatotoxicity, persistent bronchitis

In summary, we reported two patients diagnosed as familial diffuse comedones, a very uncommon inherited dermatosis. Our case reports emphasize on the autosomal dominant mode of inheritance in these rare genodermatosis. Overall prognosis is favorable as the patients remain in good health, except disfiguring scars that may cause social problems especially in the adolescent female. The treatment of familial comedones has not yet been established due to the scant of reported cases. In our patients, oral isotretinoin 10 mg once daily were given, unfortunately, data regarding the long-term follow-up are lacking.

Reference

1. Cantu JM, Gomez-Bustamente MO, Gonzalez-Mendoza A, Sanchez-Corona J. Familial comedones. Evidence for autosomal dominant inheritance. *Arch Dermatol* 1978; 114: 1807-9.
2. Rodin HH, Blankenship JL, Bernstein G. Diffuse familial comedones. *Arch Dermatol* 1967; 95:145-6.
3. Van Geel NA, Kockaert M, Neumann HA. Familial dyskeratotic comedones. *Br J Dermatol* 1999;140:956-9.
4. Hall JR, Holder W, Knox JM, Verani R. Familial dyskeratotic comedones. A report of three cases and review of the literature. *J Am Acad Dermatol* 1987;17:808-14.
5. Beck MH, Dave VK. Extensive nevus comedonicus. *Arch Dermatol* 1980;116:1048-50.
6. Anderson NP. Comedonicus nevus of extensive distribution. *Arch Derm Syphilol* 1946;53:433.
7. Vano-Galvan S, Hernandez-Martin A, Colmenero I, Torrelo A. Disseminated congenital comedones. *Pediatr Dermatol* 2011;28:58-9.
8. Cestari TF, Rubim M, Valentini BC. Nevus comedonicus: case report and brief review of the literature. *Pediatr Dermatol* 1991;8:300-5.