Familial Comedonal Darier's Disease: A Case Report

Dear Editor,

Darier's disease is an autosomal dominant blistering condition caused by mutated ATP2A2 gene on chromosome 12q23-24, which encodes the sarcoplasmic/endoplasmic reticulum calcium pump ATPase (SERCA2). It usually presents with chronic eruption of greasy hyperkeratotic papules and plaques, in a seborrheic distribution. The rarer subtypes include vesiculobullous, hypertrophic, cornifying, hypopigmented, linear and comedonal variants. We observed an unusual case of Darier's disease, which was familial and showed prominent comedonal lesions.

A 36-year-old male presented with multiple prominent open and closed comedones over the face, mid-chest and upper back since the past 15 years which leaves behind pitted scars. Cutaneous examination also revealed multiple discrete to coalescent hyperkeratotic skin colored to pigmented greasy papules in a similar distribution [Figure 1]. The palms, soles, nails, scalp and mucosa showed no abnormality. Family history revealed similar but less extensive lesions in the mother and the younger sister of the index case. There were no neurological or infectious complications in the index case or the family members.

Histopathology of skin lesion from the index case showed hyperkeratotic, acanthotic epidermis, and dilated follicular infundibulum with keratotic plugging [Figure 2]. Suprabasal acantholysis with dyskeratosis represented by corps ronds and grains [Figure 3a], leading to formation of clefts and lacunae with elongated dermal villi formation [Figure 3b] was present. Skin biopsy from the family members also demonstrated similar findings. According to the clinical and histopathological features, we finally diagnosed the case as familial comedonal Darier's disease. Isotretinoin was started at 0.5 mg/kg/day. However, the patient was unfortunately lost to follow-up.

Darier- White disease or keratosis follicularis is a disease of abnormal keratinization with variable penetrance described by Darier and White in 1889.[1] It is usually characterized by seborrheic distribution of chronic and recurrent greasy hyperkeratotic papules and plaques. Vegetating papules, hemorrhagic blisters or erosions may also be present. Histopathologic findings include dyskeratosis evidenced by corps ronds and grains, suprabasal acantholysis leading to clefting and single layer of basal keratinocytes overlying dermal papillae which project into the acantholytic cavity.[1]

Previously reported cases of comedonal subtype manifested as comedones, papules or nodules, on the face, upper

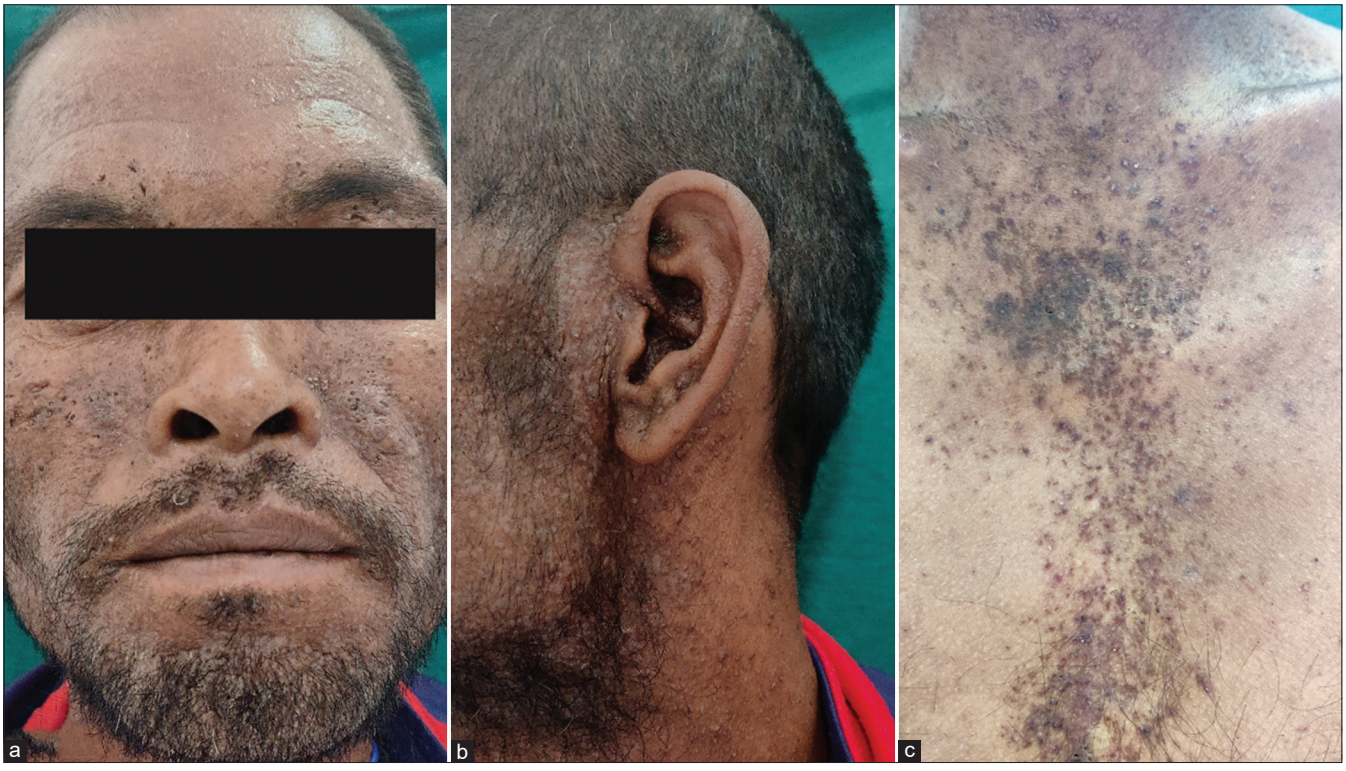

Figure 1: (a) Comedones and pitted scars over seborrheic areas of face (b) Peri-auricular area showing both open and closed comedones (c) Hyperkeratotic greasy papules over mid-chest

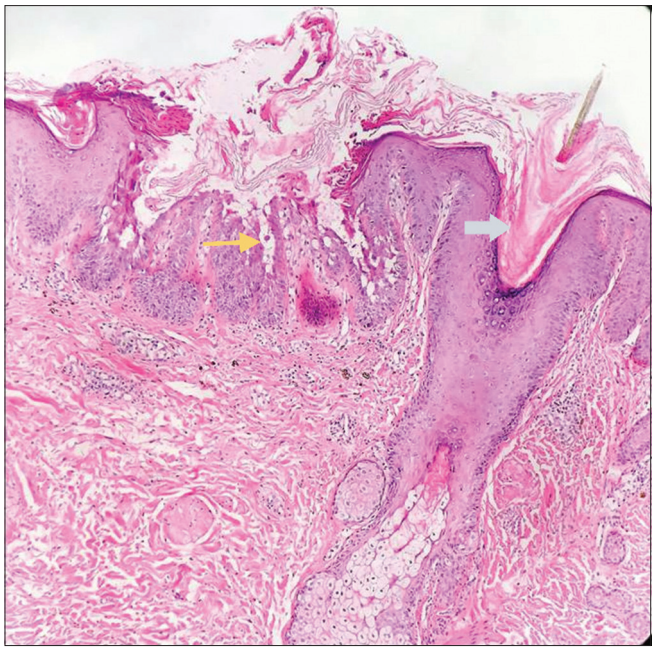

Figure 2: Hyperkeratotic, acanthotic epidermis with follicular plugging (grey arrow) and suprabasal acantholysis (yellow arrow) with formation of clefts and lacunae [H and E,100X]

trunk and scalp. Its association with other features of classic Darier's disease has been seen as in our case. The distinctive predilection of follicular involvement and formation of elongated dermal villi with papillary projections differentiates comedonal Darier's disease from classical Darier's disease. Disorders like familial dyskeratotic comedones, acne vulgaris, Favre-Racouchot syndrome, nevus comedonicus and trichoepithelioma should be differentiated from comedonal Darier's disease. Familial dyskeratotic comedones, in particular, has very similar features, but large open comedones on the forearms and thighs, with sparing of scalp, face and mouth usually differentiates it. Also, lacunae, villi and corps ronds are less prominent.[2]

The course of comedonal Darier's disease is variable. Topical treatments (retinoids, calcipotriol, steroids, tacrolimus, bufexamac, emollients) and systemic therapies (retinoids, antibiotics, biotin, Korean ginseng and anti-histamines) have all showed guarded success and are unsatisfactory.[3]

We found 22 cases of previously reported comedonal Darier's disease, including a case of linear comedonal variant.[3-5] However, only two of them had a family history.[5] Therefore, we report this interesting case of familial comedonal Darier's disease.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

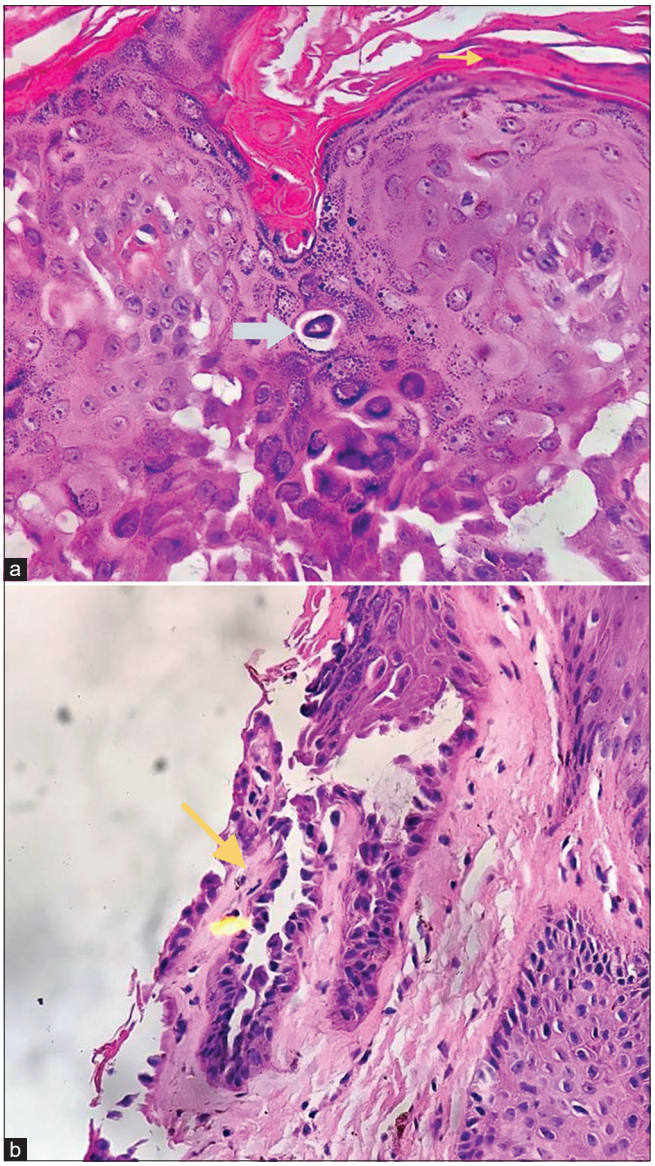

Figure 3: (a) Characteristic dyskeratosis: corps ronds (grey arrow) and grains (yellow arrow) [H and E,400X] (b) Elongated villus formation (yellow arrow) [H and E, 400X]

Financial support and sponsorship

Nil.

Conflicts of interest

There are no conflicts of interest.

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Website: https://journals.lww.com/idoj

Quick Response Code

DOI: 10.4103/idoj.idoj_667_22

How to cite this article: Bikash C, Verma S, Handique G, Dey B. Familial comedonal Darier's disease: A case report. Indian Dermatol Online J 2023;14:714-6.

Received: 12-Dec-2022. **Revised:** 11-Jan-2023.
Accepted: 26-Feb-2023. **Published:** 25-May-2023.