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PhotoDermDiagnosis

Hyperkeratotic, pigmented papules over the sides of neck in a girl.

Section Editor

Amor Khachemoune, **MD CSW,**
Suny Down State Medical Center, Brooklyn, NY, USA

Asher Ahmed Mashhood*, Shaukat Mehmood Qureishi, Tariq Khurshid***
* Department of Dermatology, **Department of Family Medicine, Combined Military Hospital, Multan.

Case report

Our patient a 15-year-old girl presented with one year history of eruption of multiple, small, firm and pigmented lesions over the sides of the neck, chest and upper back. These lesions were asymptomatic; however, she felt slight pricking sensation in the lesion when she was under direct sunlight or exposed to the heat of the kitchen. She also gave the history of abnormality of the shape of the nails of hand and feet with frequent cracks over the edge of the nails, since the same duration.

On examination, there were multiple confluent, pin-point, brownish, greasy looking papules over the sides of neck, upper chest and upper back. There were a few white, warty papillomatus masses on left side of her neck (**Figure 1**) and in right axilla. There were a few skin coloured discrete papules over the dorsum of both forearms. On the examination of hands, there were minute pits on the palmar surface of the fingers. Longitudinal red bands were seen on the finger nails of right little, ring, index fingers and thumb and left ring and little fingers. Nail notching was seen at the free margin of nails of right ring and index fingers and left ring finger (**Figure 2**). Examination of oral mucosa was unremarkable. The histopathological examination of the skin specimen revealed acanthosis, papillomatosis and hyperkeratosis. Several dyskeratotic cells were seen in the stratum granulosum and a few suprabasal clefts were visible. The dermis showed mild chronic inflammatory cell infiltrate.

Figure 1

Figure 2

Address for correspondence
Dr. Asher Ahmed Mashhood,
Consultant Dermatologist,
Combined Military Hospital, Multan.
Tel # 061-5316154
E mail: asherahmed67@yahoo.com

What is your diagnosis?

Diagnosis

Darier's disease

Hence based on the clinical and histopathological evidences the patient was diagnosed as a case of Darier's disease. Since the patient was a young girl she was prescribed emollients with mild keratolytics, which the patient used the treatment for 2 months with no improvement. After 2 months, the patient and her parents insisted on a more effective treatment. So they were explained in detail the option of the use of oral retinoids, their side effects and avoidance of conception one year after the end of the therapy. Having taken the consent, the patient was advised Cap Neotigason (Acitretin) 25mg x BD for 4 weeks and then 25mg x OD for another 2 weeks.

Discussion

Darier's disease is an autosomal dominant genodermatosis with variable penetration, characterized clinically by the presence of skin-coloured or brownish, greasy-looking hyperkeratotic papules over the seborrhoeic areas of the trunk, face, scalp and ears.[1] These papules may coalesce to form warty lesions and in the flexures form vegetating masses. The flexural areas most commonly involved are anogenital regions, groins and natal cleft. On scalp the lesions simulate seborrhoea but the spiny feel is characteristic. On limbs the lesions are in form of scattered papules simulating acrokeratosis verruciformis of Hopf.[2] Palms and soles show several minute pits, or in older subjects punctuate or filiform keratosis.[3] and the oral mucosa show white umblicated papules on the palate resembling nicotine stomatitis.[4] Confluent lesions in the oral mucosa may appear like leukoplakia. Nails are often brittle and show characteristic longitudinal white or red bands of varying width, often ending in a pathognomonic notch at the free margin.

The disease manifests mainly in second decade and start as a few lesions with nail changes. There are frequent exacerbations and remissions but the course is generally chronic. The disease is exacerbated by heat, sunlight[5] and by steroids.[6]

In most patients the disease is very typical, but a few present with incomplete forms such as isolated nail or palmar changes, vesicobullous lesions,[7] flexural or erosive disease,[8] multiple comedonal lesions[9] and naevoid form (acantholytic dyskeratotic epidermal naevus).[10] Unilateral focal form is one of the rare types of the Darier's disease. Two clinical and genetical phenotypes of the segmental Darier's disease have been recently reported: type 1- in which the skin outside of the localized manifestation is normal and type 2- showing segmental affection in combination with diffuse skin involvement.[11]

The disease in now believed to occur due to mutation in ATP2A2 gene at chromosome 12q24.1, which encodes the sarco- and endoplasmic reticulum calcium ATPase type 2 (SERCA2).[12] Cadherins are Ca++ dependent cell-to-cell adhesion molecules which play an important role in the cellular connection between normal cells. A defect in Ca++ transport ATPase will result in abnormality of the tonofilaments which

leads to the disease process.[13] Histopathology findings are very characteristic. They include hyperkeratosis, epidermal hyperplasia, acantholytic dyskeratosis resulting in *corps ronds*, suprabasal clefts or *lecunae* formation and irregular upward proliferation of dermal papillae with formation of *villi*.

Full blown disease can hardly be confused with any other dermatosis but mild cases may be confused with acne and seborrhoeic dermatitis. Flexural lesions may be mistaken with Hailey-Hailey disease, acanthosis nigricans or Dowling-Degos disease.

Patients with mild disease require mild emollients and advice to avoid sunburn. Topical tretinoin,[14] adapalene[15] and tazarotene[16] have been reported as effective. There are case reports of response with the use of cyclosporine[17] or topical 5-fluorouracil.[18] However, in patients with more severe disease, oral retinoids are the treatment of choice. Both isotretinoin[19] and acitretin[20] are effective.

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Erratum

In the article "Aplasia cutis congenita in two brothers – a rare occurrence" published in the July-September, 2005 issue of JPAD (*J Pak Assoc Dermatol* 2005; **15**: 275-7), the name of one of the authors was misprinted as **Naseem Raza**. His correct name is **Naeem Raza**.

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