CASE REPORT

Incidental finding of Darier's disease in a young Saudi female: A case report

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ABSTRACT

Darier's disease also referred to as dyskeratosis follicularis, maybe a rare disorder of keratinization. It is an autosomal dominant genodermatosis with a defect in the desmosomal attachment due to a mutation in the ATP2A2 gene, which encodes the Sarco/endoplasmic reticulum Ca2+ ATPase isoform 2 with high penetrance and variable expressivity. The condition is characterized by multiple hyperkeratotic papules predominantly in seborrheic areas on the face, neck, and trunk, with less frequent involvement of the oral mucosa. This paper reports a case of 34 years old woman who is presented at King Fahd Hospital, Madinah in the outpatient clinic with more than 18 years history of papules and greasy scaly lesions distributed on face, neck and trunk with palmoplantar pits. Furthermore, her daughter, 13 years old who was accidentally discovered during her visit with her mother, has scaly lesions that started in her neck, chest, and nose that appeared before six months.

KEYWORDS: Autosomal dominant, Darier disease, keratosis follicularis

INTRODUCTION

Darier's disease or keratosis follicularis is a rare autosomal dominant genodermatosis characterized by greasy, crusted, keratotic, yellow-brown warty papules and plaques over seborrheic areas. Although this is a genetically transmitted disease, according to large series, about 47% of patients had no apparent family history, presumably because of incomplete penetrance.[1] The disease is caused by mutations in the ATP 2A gene, which encodes the sarcoendoplasmic reticulum Ca2+ ATPase pump (SERCA2), leading to acantholysis and apoptosis, accounting for the characteristic pathologic finding of acantholytic dyskeratosis in Darier disease.[1] The disease usually starts around puberty and runs a chronic course with exacerbations induced by sun exposure, heat, friction, or infections.[2] This disease was first described by Prince Marrow in 1886 and simultaneously by Darier and White in 1889, independently. White was the first to recognize the genetic nature of keratosis follicularis by noticing that a mother and her daughter were affected.[3] In 1917, the first case with oral manifestation was reported by Reenstierna.[4] Clinically, the distinctive lesion is characterized by hyperkeratotic papules that coalesce into plaques and occur primarily in seborrheic and intertriginous areas.[5] Coalescence of the papules produces irregular warty plaques or papillomatous masses within the flexures, which become hypertrophic and malodorous with painful fissures.

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A 34-Year-old woman presented at King Fahd Hospital, Madinah, in the outpatient clinic with more than 18 years history of persistent pruritic skin lesions distributed over the face, neck, upper chest, and trunk. Sometimes associated with a bad odour. On examination, multiple skin-coloured yellow to brown greasy hyperkeratotic papules scattered over seborrheic areas (Face, nose, behind ears, cheeks, neck, upper chest, trunk) (Fig. 1,2). Punctate palmoplantar keratoderma was detected (Fig. 3). Nails show longitudinal streeks with distal V-shaped subungual keratosis (Fig.4). Furthermore, her daughter, 13 years old, who was accidentally discovered during her visit with her mother, has scaly time yellowish papules that are scattered over (Neck, Chest and Nose) that appeared before six months (Fig. 5,6).

Fig. 1 Multiple, skin-coloured to brownish greasy papules on the face and neck

Fig. 2 Multiple, skin-coloured to brownish greasy papules on the trunk

Fig. 3 Nails show longitudinal streeks with distal V-shaped subungual keratosis

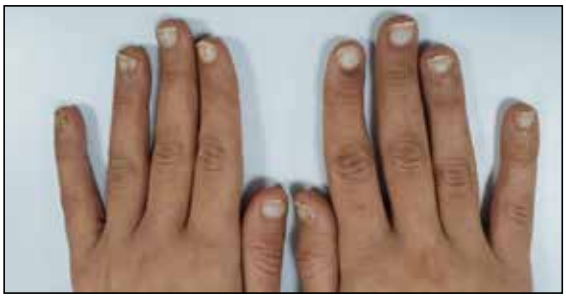

Fig. 4 Nails show longitudinal streeks with distal V-shaped subungual keratosis

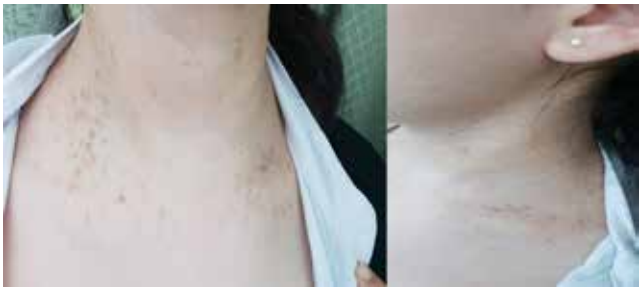

Fig. 5 Multiple, skin-coloured to brownish greasy papules on the neck

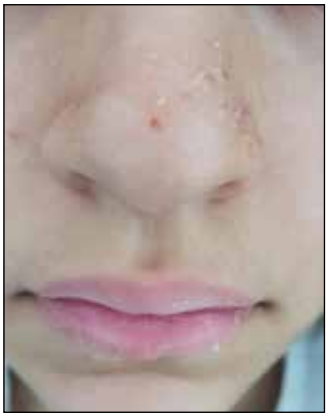

Fig. 6 yellowish papules that are scattered on the Nose

HISTOPATHOLOGICAL EXAMINATION

Skin punch biopsy 4mm from the abdomen of the mother (Fig.7), Right upper 2mm from the back of the daughter (Fig.8). Shows tiny focus of suprabasilar acantholysis with corps ronds columns of parakeratosis, reminiscent of cornoid lamellae above the affected foci.

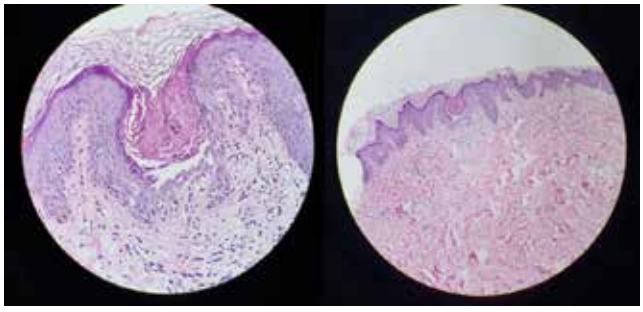

Fig. 7 Right upper 2mm from the back of the daughter

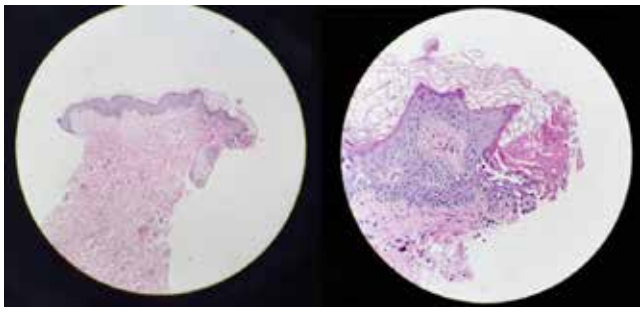

Fig. 8 Shows tiny focus of suprabasilar acantholysis with corps ronds columns of parakeratosis

DISCUSSION

Darier's disease was first described by Prince Marrow in 1886 and later independently described by Darier and White in 1889.[6] The condition is a genodermatosis with an autosomal dominant inheritance caused by a mutation in the ATP2A2 gene, at chromosome 12q23–12q24.[7] to a mutation in the ATP2A2 gene, which encodes the Sarco/endoplasmic reticulum Ca2+ ATPase isoform 2 with high penetrance and variable expressivity. This gene encodes the sarcoplasmic endoplasmic reticulum Ca2+ ATPase type 2 protein (SERCA2), which is a calcium pump widely expressed in the skin. The main function of SERCA2 is transporting Ca2+ from cytosol to the lumen of endoplasmic reticulum, where Ca2+ can be stored. A defect of SERCA2 leads to a deficiency of Ca2+ at the cell membrane, particularly in desmosomes, resulting in impaired cell-to-cell adhesion and induction of apoptosis. The chance of a child inheriting the abnormal gene if one parent is affected is 1 in 2 (50%) but not all people with the abnormal gene will develop clinical features of the disease.[8] The characteristic feature of Darier's disease includes the presence of multiple, skin-colored to yellow-brown, hyperkeratotic papules distributed in seborrheic areas, such as the head, neck, and trunk. Other clinical findings include focal acantholytic dyskeratosis with suprabasilar acantholysis with corps ronds columns of parakeratosis reminiscent of cornoid lamellae above the affected foci.[9,10]

STATEMENT OF ETHICS

The patient provided written informed consent to perform all necessary investigations, to take clinical photographs, and use them for research purposes and publication. The patient understood that her name and initials will not be published and due efforts will be made to conceal her identity. This case report was conducted ethically in King Fahad Hospital, Medina, Saudi Arabia.

DISCLOSURE STATEMENT

The authors have no conflicts of interest to declare.

FUNDING SOURCES

The authors have no financial funding sources to declare.

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