Darier Disease: Sustained Improvement Following Reduction Mammaplasty

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Darier disease is an autosomal-dominant inherited genodermatosis. A woman is described who had Darier disease and hereditary spherocytosis whose inframammary Darier disease had dramatic and sustained improvement following reduction mammaplasty. The postoperative resolution of the dermatosis beneath the patient's breasts introduces the possibility that, when appropriately indicated, breast reduction surgery may have a potential role in the management of severe inframammary Darier disease.

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Darier disease, also referred to as Darier-White disease or keratosis follicularis, is an autosomal-dominant inherited genodermatosis that can clinically involve the seborrheic and flexural areas (as warty brown papules and plaques), the hands and mouth, and the nails.[1-4] Hereditary spherocytosis is a hemolytic disorder that can have an autosomal-dominant mode of inheritance.[5-7] A woman is described who had Darier disease and hereditary spherocytosis whose skin condition dramatically improved following bilateral reduction mammaplasty. The possible role of breast reduction surgery as a treatment modality in Darier disease is introduced.

Case Report

A 46-year-old Hispanic woman had a history of hereditary spherocytosis and a recurrent dermatosis that initially appeared in her early 20s. The patient's mother, sister, son, and daughter also had hereditary spherocytosis. In addition, her mother, another sister, and 2 sons (including the son with spherocytosis) had a similar dermatosis. The woman also had chronic neck and shoulder pain related to her large heavy breasts.

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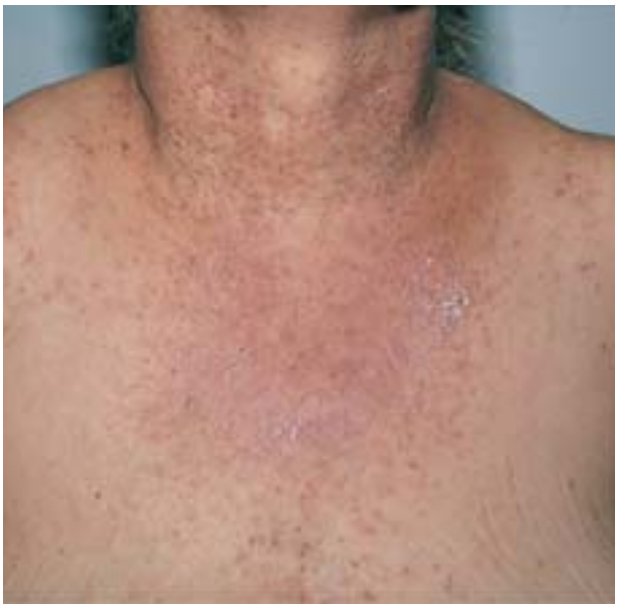

Figure 1. Keratotic papules of Darier disease on the anterior neck, chest, and proximal breasts.

Clinical examination showed a woman whose large breasts extended to just above her umbilicus. Pruritic, erythematous, keratotic papules were present on her posterior neck, anterior neck, and upper chest (Figure 1). Similar lesions also were present on her abdomen and between, beneath, and on her breasts (Figure 2). A longitudinal red streak was on the nail of her left index finger (Figure 3).

Microscopic evaluation of a lesional skin biopsy showed acantholytic dyskeratotic keratinocytes in the granular and horny layers of the epidermis (corp ronds) and in the stratum corneum and lacunae (corp grains). In the suprabasilar layers of the epidermis, there was cleft (lacunae) formation—dermal papillae, lined by basal cells, that stuck up into the lacunae to form villi. The pathologic findings, correlated with the morphologic features, confirmed the clinically suspected diagnosis of Darier disease.

Topical application of 0.1% triamcinolone acetonide cream twice daily provided symptomatic relief and eventual resolution of the patient's recurrent

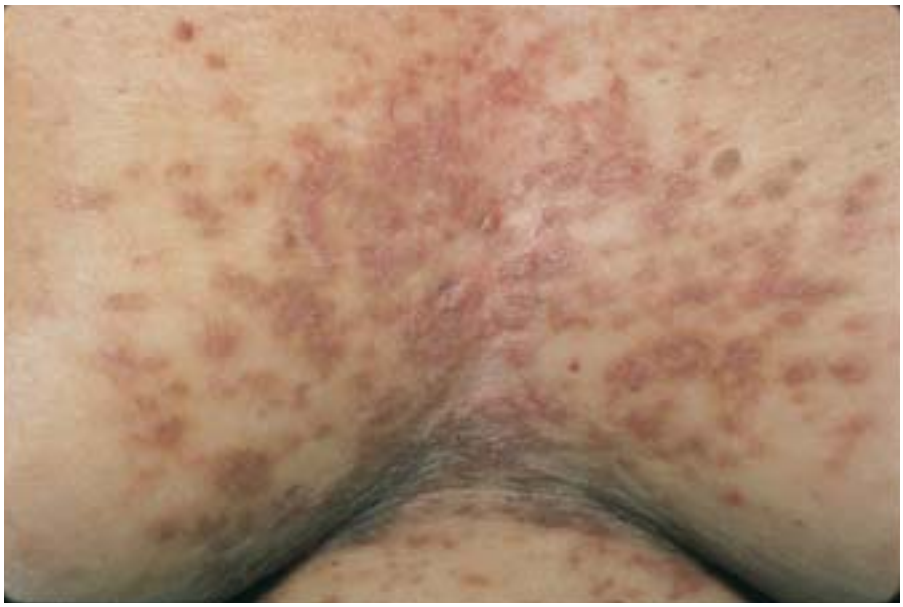

Figure 2. Extensively distributed Darier disease lesions between, beneath, and on the breasts, extending to involve the abdomen.

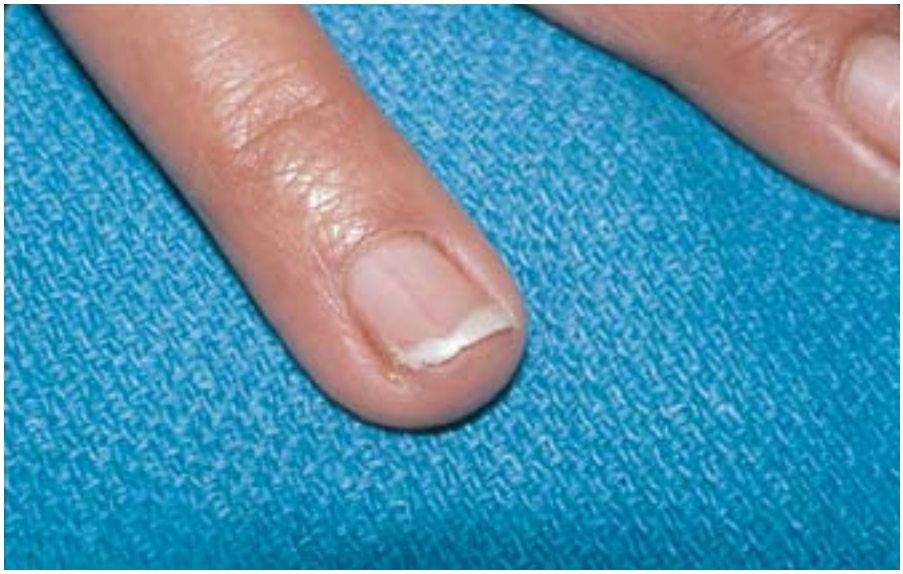

Figure 3. Darier disease-related longitudinal red streak involving the nail of the left index finger.

disease flares. To treat her neck and shoulder pain, she had a bilateral reduction mammaplasty; 685 g were removed from the right breast and 760 g were removed from the left breast.[8-10] There was dramatic improvement and sustained near-complete remission of the Darier disease beneath her breasts after surgery. However, her postoperative course was complicated by necrosis and total loss of the right nipple-areolar complex (Figure 4).[11]

Comment

Darier disease and hereditary spherocytosis are autosomal-dominant inherited conditions. In contrast to Darier disease, in which the responsible genetic defect maps to chromosome band 12q23-24.1,[1,2,12-16] some patients with hereditary spherocytosis have disease-associated abnormalities of chromosome 8.[5-7] Therefore, though it is intriguing to consider that the concurrent occurrence of these disorders in 3 generations of our patient's family may indicate that they have a common mechanism of pathogenesis, it is more likely that their coexistence is merely coincidental.

The most effective medical treatment for Darier disease is oral retinoids, such as isotretinoin, etretinate, and acitretin. However, long-term retinoid therapy is required, and the dermatosis usually recurs after the drug is discontinued.[1,2] Topical cortico-

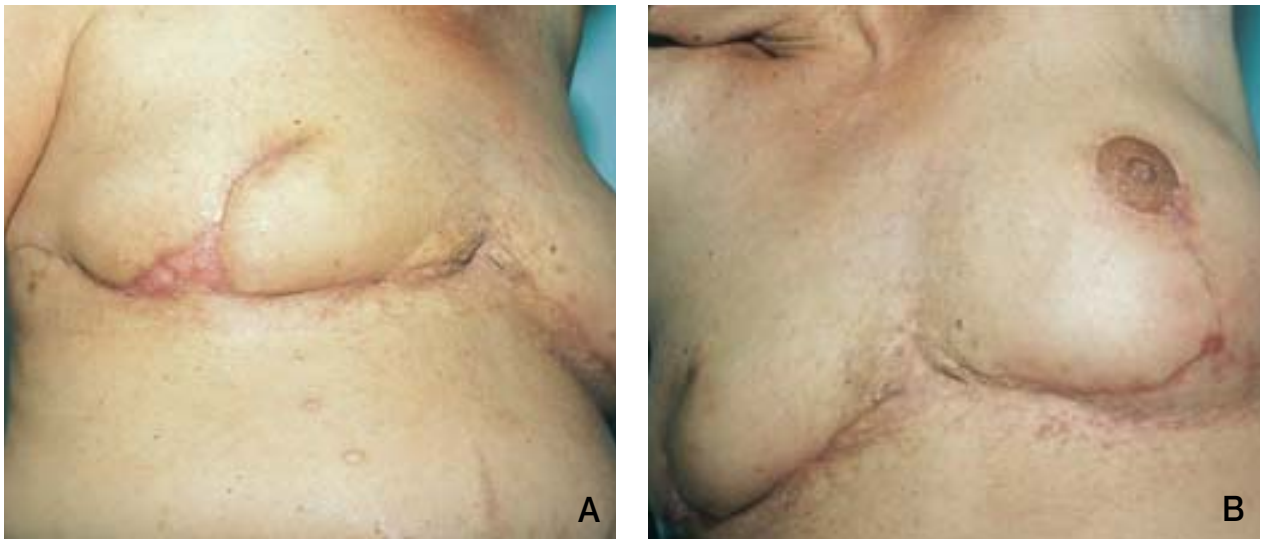

Figure 4. Right (a) and left (b) breasts, inframammary regions, and abdomen demonstrate the marked reduction of Darier disease lesions following bilateral reduction of breasts. The right nipple-areolar complex required excision secondary to postoperative necrosis.

steroids and oral antibiotics are conservative modalities for managing Darier disease.[3]

Radiotherapy (grenz rays) and surgical intervention (excision and grafting, electrosurgery, dermabrasion, and carbon dioxide laser vaporization) have been described only in a small number of patients.[3,17] The resolution of our patient's inframammary dermatosis following reduction mammaplasty was an unexpected, yet greatly appreciated, outcome. Based on the dramatic improvement of the patient's Darier disease, additional investigation into the potential role of appropriately indicated breast reduction surgery in the management of severe inframammary Darier disease in women with gigantomastia may be warranted.

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