CASE REPORT

Darier Disease associated with Cutis Verticis Gyrata

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ABSTRACT

Darier disease (DD) is an autosomal dominant genodermatosis characterized by cutaneous manifestations, such as keratotic papules present on the upper trunk and longitudinal erythronychia.

Cutis Verticis Gyrata, also known as pachyderma verticis gyrata, is a rare benign skin disorder characterized by the presence of convoluted folds, hypertrophy and skin hypermobility which generates deep grooves on the scalp of similar aspect to cerebral cortex gyri.

A 34-year-old male patient with no relevant pathological history or neuropsychiatric alterations presents with keratotic papules, developed during childhood, on the thorax (front and back), neck and intertriginous areas. During puberty, longitudinal folds on the head started to grow, extending anteroposteriorly and covering approximately 80% of the surface, without accompanying alterations on the scalp. This case presentation may consist of an association or a rare coincidence.

A skin biopsy for histological study is performed, exposing findings compatible with Darier Disease. The scalp does not reveal any histopathological alterations.

The importance of the present clinical case relies on the association of rare pathologies.

INTRODUCTION

Cutis Verticis Gyrata is a rare benign condition of unknown etiology, characterized by the presence of ridges and deep furrows on the scalp that mimic cerebral gyri. These folds may generate cutaneous maceration and superinfection. This disorder is classified, according to its presentation form, into primary and secondary.[1]

Darier disease, also known as follicular dyskeratosis, is an unusual form of genodermatosis characterized by the presence of hyperkeratotic plaques and papules which affect seborrheic areas. Such condition presents variable expressivity. Consequently, it carries discrete clinical features, leading to diagnostic errors.[2]

CLINICAL CASE

A 34-year-old male patient with no relevant pathological history or neuropsychiatric alterations presents with keratotic papules, developed during childhood, on the thorax (front and back), neck and intertriginous areas (Figure 1). During puberty, longitudinal folds on the head started to grow, extending anteroposteriorly throughout the scalp and covering approximately 80% of the surface (Figure 2), without accompanying alterations, but causing social discomfort.

Skin biopsy of lesions on the anterior portion of the thorax is performed. Histopathology reveals focal acantholytic dyskeratosis with the presence of large round acantholytic keratinocytes primarily found in the stratum spinosum and granulosum, along with enlarged acantholytic cells in the stratum corneum and suprabasal fissures (Figure 3). Additionally, biopsy of the scalp is performed; no histopathological alterations are found.

Clinical and histopathological features lead to the diagnosis of Darier Disease associated with Cutis Verticis Gyrata. The dyskeratotic pathology was kept under control. Therefore, some general care measures are implemented, such as the use of synthetic detergents and emollients. In regards to the benign nature of the scalp pathology, the patient decides not to adopt any therapeutic measure.

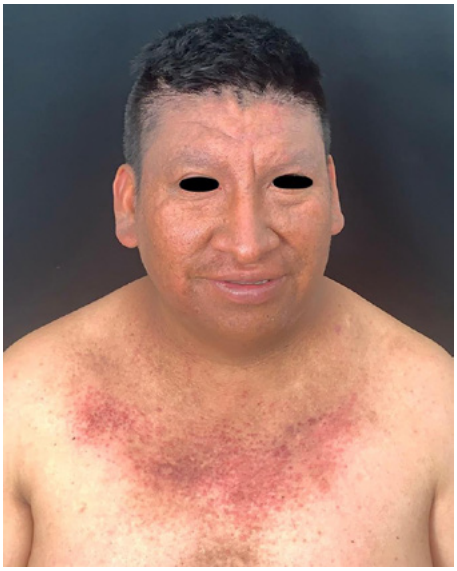

Figure 1. Hyperkeratotic papules at the anterior portion of the thorax.

Figure 2. Longitudinal folds (anteroposterior) on the scalp.

DISCUSSION

Darier Disease, also known as follicular dyskeratosis, was described in 1889 by French dermatologist Jean Darier from Saint-Louis Hospital in Paris and James C. White.[1,3]

This pathology affects both men and women in equal measure. Moreover, it manifests in the first or second decades of life.[4]

This condition consists of a dominant autosomal inherited genodermatosis caused by the loss of keratinocyte cell-adhesion as a result of desmosomal complex rupture. This is generated by a mutation in the ATP2A2 gene, at chromosome 12q23–12q24, which encodes the Ca (2+)-ATPase pump located in the endoplasmic reticulum (SERCA2), transporting cytosolic Ca2+ into the lumen. Mutations of this gene cause the desmosomes to exhibit calcium depletion, inducing apoptosis and intercellular adhesion alterations.[3,2,5]

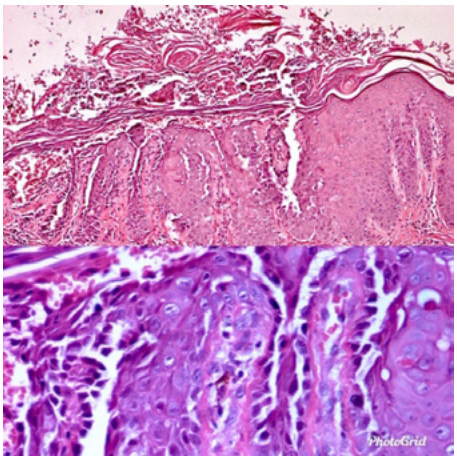

Figure 3. Focal acantholytic dyskeratosis with round acantholytic keratinocytes primarily found in the stratum spinosum.

This pathology is clinically characterized by the presence of multiple hyperkeratotic papules, which may appear as brown or brownish-red crusty entities distributed mainly on seborrheic areas, such as the head, face, temples, neck and trunk. Furthermore, facial lesions which are left untreated for a long period of time may develop into skin thickening with multiple papules and confluent hyperkeratotic plaques.[3,4,6]

The scalp exhibits thick crusts indicating seborrheic dermatitis. The dorsum of the hands and feet manifest clinically indistinguishable discrete papules from Hopf's acrokeratosis verruciformis. Ungueal changes include red or white longitudinal bands of variable thickness ending in a pathognomonic "v" or "y" notch at the free margin, as well as onychorrhexis and painful subungual hyperkeratosis.[4]

Lesions are often associated with pruritus and unpleasant smell; particularly, those which develop into frictional areas. Moreover, maceration and secondary infections may generate foul odor.[6]

There are topographic clinical variants: the generalized form with classic features and the localized or segmental form, which manifests relatively late in 10% of the population, constituting a genetic mosaicism.[4]

Apart from topographic variants, there are atypical morphologic variants (enumerated from most to least frequent): disseminated crusted-papular, blistering-pustular, hypertrophic, acral and hemorrhagic.[4]

Typical histopathological manifestations of DD include the presence of dyskeratosis (premature keratinization) with dyskeratotic cells (round and granular entities) found in the stratum Malpighi and stratum corneum, suprabasal cleavage, loss of epidermal adhesion (acantholysis), raised irregular projections of dermal papillae within cleavage lacunae (villosities) and hyperkeratosis.[7]

Differential diagnoses consist of seborrheic dermatitis, acanthosis nigricans and Hopf's acrokeratosis verruciformis. These pathologies were ruled out due to clinical and histopathological findings.[6,8]

An integral treatment is required, including general skin care and topical or systemic therapy. Nonetheless, DD commonly persists throughout life with partial remission periods. Intermittent exacerbations are caused by heat, sweat, humidity and bacterial infections. Oral retinoids (acitretin, isotretinoin and alitretinoin) are the foundation of DD treatment, as they reduce hyperkeratosis. Nevertheless, side effects limit their use.[9]

This patient's dyskeratosis was kept under control. Therefore, synthetic detergents and emollients were employed to ensure optimal supervision of the pathology, in conjunction with general care measures to prevent exacerbations.

Cutis Verticis Gyrata, also known as pachyderma verticis gyrata, was firstly described in 1843 by Robert, who referred to it as a benign skin condition characterized by the formation of furrows and folds on the scalp, simulating cerebral gyri over time. The condition's prevalence within men and women is estimated at around 1/100000 and 0.026/100000 respectively; indicating a 5:1 relation.[10,11]

It was classified by Polan and Butterworth in 1953. This division, based on the disease's evaluation, resulted in primary (essential or non-essential) and secondary. The primary non-essential and secondary forms are generally associated with abnormalities (neurological and ophthalmological). However, the primary essential form does not present with related comorbidities. This pathology's etiology is unknown. Additionally, men are 5 times more likely to manifest it.[10,12]

Clinically, it occurs during puberty and presents with 2 to 30 folds, which are 0.5 to 2 cm wide and approximately 1 cm deep. They are typically located on the occipital region, at the vertex. Nevertheless, they may cover the scalp in its entirety.[11,12]

The primary form of CVG presents symmetrical folds in an anteroposterior direction, while the secondary form manifests asymmetrical and variably disposed folds.[11]

Histopathology reveals that the primary form presents with normal skin or dermal thickening with adnexal hypertrophy. Secondary variants exhibit findings associated with the underlying disease process.[11]

The present case followed a CVG patient without osteoarticular disorders or any hormonal and psychiatric conditions. The classic manifestations of the pathology were present. Additionally, even though its manifestation coincided with Darier Disease, the condition was identified as primary essential, given that no clinical association reports have been observed in the literature.

The treatment of this pathology requires proper diagnosis, which would contribute to determine the subjacent etiology in each case of secondary CVG. The treatment for the primary form only consists of adequate scalp hygiene (especially the furrows) to prevent secretion accumulation.[13]

Surgical procedures are carried out solely if the patient presents psychological problems due to cosmetic complications caused by the condition; including simple excisions, tissue expansion or skin graft (conditioned by the size and locations of the lesions).[12,14]

CONCLUSIONS

Darier Disease is a rare and well-defined pathology whose common complications involve inflammatory processes, superinfection and foul smell. Some cases reveal more than the condition's usual findings and present with extensive manifestations, affecting the quality of life of patients to varying degrees.

At the present time, the references do not encompass any correlation between these two pathologies. Consequently, both the magnitude and intrinsic nature of their association are unknown and no physiopathological nexus has been identified. Nevertheless, the two entities are independently associated to neuropsychiatric alterations, which justifies the importance of the research.

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