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Discoid lupus erythematosus associated with chronic granulomatous disease

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To the Editor,

Chronic granulomatous disease (CGD) is a primary immunodeficiency disorder in which a reduced activity of nicotinamide dinucleotide phosphate (NAPDH) oxidase leads to defective reactive oxygen intermediates (ROS) and impairs intracellular killing of microorganisms. Patients have recurrent bacterial and fungal infections as well as granuloma formations, and have a higher risk of autoimmune and inflammatory disorders like lupus erythematosus.[1]

A 20-year-old caucasian male with a diagnosis of autosomal recessive CGD was referred to our hospital because of a pruritic photosensitive rash on the face and hands, present for more than a year. A month before the appearance of the first lesions, the patient had started treatment with voriconazole for pulmonary aspergillosis. Due to the possibility of voriconazole-induced photosensitivity, after two months of therapy, this regimen was changed to posaconazole, but the rash persisted. The physical examination revealed several erythematous plaques with atrophic white center on the frontal region, malar region and nose (Figure. 1), and eczematous plaques on the dorsum of both hands. There were no other cutaneous or systemic symptoms. Antinuclear antibodies and double stranded DNA antibodies were negative. The histopathological examination of skin biopsies of the face and hands showed focal vacuolar degeneration of basal keratinocytes, occasional colloid bodies at the dermal-epidermal junction and dermal perivascular and a perifollicular lymphocytic infiltrate (Figure. 2). Direct immunofluorescence was not performed. Chronic discoid lupus erythematous (DLE) associated with CGD was the likely diagnosis. Due to the pharmacological interactions of hydroxychloroquine and posaconazole, oral acitretin 0.5mg/kg/day was started, as well as topical corticosteroids, topical tacrolimus and adequate photoprotection. A significant improvement of the lesions was noted after six months of treatment.

Cutaneous manifestations are common in CGD including skin infections (eg, impetigo, carbuncles, abscesses) and, less frequently, inflammatory conditions such as lupus-like skin lesions.[1] DLE can appear in a subset of patients with CGD.[2,3] The pathogenesis is unclear, but individuals with CGD may be at higher risk of developing lupus due to the defective clearance of apoptotic cells, including keratinocytes, leading to the release of autoantigens, autoantibody production and persistent inflammation.[4] However, the majority of cases are reported in female carriers of X-linked CGD, followed by autosomal recessive CGD patients and only 3 cases in patients with X-linked CGD, although the latter is the most common form of CGD.[2,5] It has been proposed that the existence of a population of neutrophils with NADPH oxidase deficiency in female carriers is sufficient for increasing the risk of lupus.[4] The possible greater risk for patients with autosomal recessive CGD to develop DLE could be due to the significant residual ROS production, when compared to the absolute deficiency of functional neutrophils in X-linked CGD patients. [2] Additionally, in our case and 4 other

cases reported in the literature, the development of DLE lesions was chronologically related with the initiation of voriconazole.[2,6,7] Voriconazole photosensitive metabolites can damage keratinocyte DNA and could act as a trigger for DLE. This case highlights that DLE should be considered in the differential diagnosis of new cutaneous lesions in CGD patients.

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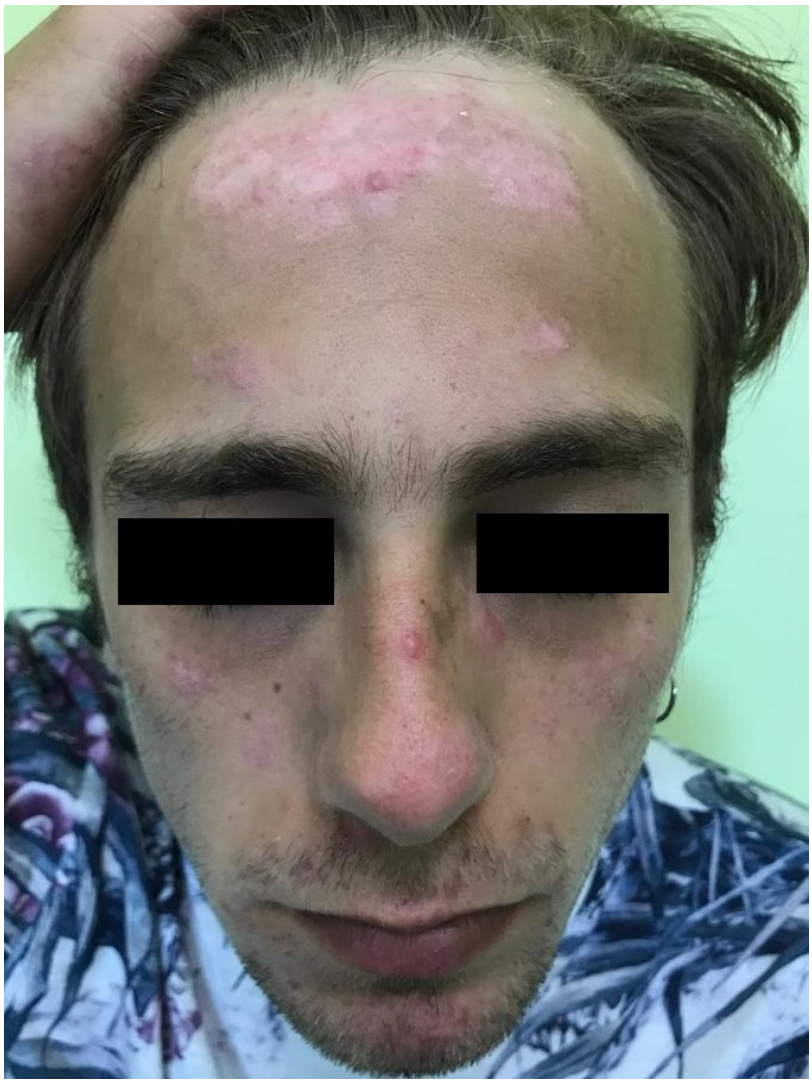

Figure 1. Erythematous plaques with atrophic hypopigmented center on the face.

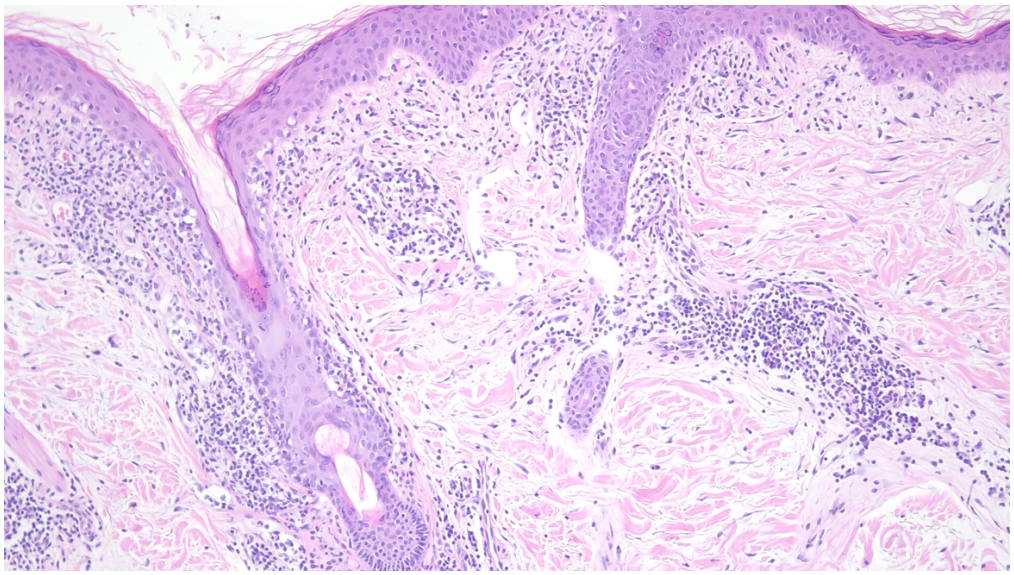

Figure 2. Histopathological examination showing perivascular and perifollicular lymphocytic infiltrate and focal vacuolar changes in the basal layer (HȣEx100).