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Cutaneous Rosai–Dorfman disease in a 42-year-old woman: a rare case report

Giovanni Marco D'Agostino[1], Melania Giannoni[1], Giulio Rizzetto[1], Federico Diotallevi[1], Anna Campanati[1]✉, Marina Scarpelli[2], Laura Pepi[2], Annamaria Offidani[1]

[1]Department of Clinical and Molecular Sciences, Dermatology Clinic, Polytechnic Marche University, Ancona, Italy. [2]Department of Neurological Sciences, Anatomopathological Institute, Polytechnic Marche University, Ancona, Italy.

Abstract

Rosai–Dorfman disease (RDD) is a histiocytic disorder that has only a skin implication in a very small percentage of cases. RDD is usually painless and accompanied by disseminated lymphadenopathy. We present a rare case of a female patient that complained of grouped skin papules localized on the left leg, associated with a palpable deep nodular lesion. Initially, this was clinically mistaken for a soft tissue sarcoma, but after a total body CT and surgical excision it was identified as a non-Langerhans cell benign histiocytosis known as RDD. The patient had neither recurrence nor systemic involvement after 7 months of follow-up.

Keywords: histiocytosis, non-Langerhans cell benign histiocytosis, Rosai–Dorfman disease, skin cancer, surgery

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Introduction

Rosai–Dorfman disease (RDD), an eponymous designation for sinus histiocytosis with massive lymphadenopathy, is an uncommon non-Langerhans histiocytic disorder, usually presenting with painless cervical lymphadenopathy and other symptoms depending on the organs involved. This disease has been described as more frequent in men than in women, usually affecting dark-skinned people, with a mean age of 20.6 years (1).

Very rarely the disease is limited only to the skin (about 3%) without other involvement. This variant is known as cutaneous RDD (CRDD) and shows a different epidemiology, with a female predominance and a median age of 43.5 years (2). We report an uncommon case of a woman with pure CRDD completely resolved after surgical excision, and free from recurrence, complication, or other systemic involvement after 7 months' follow-up.

Case report

A 42-year-old Caucasian woman presented to our dermatology unit with a deep nodular neoformation of the anteromedial surface of the right thigh, which had been progressively growing for a few months. After an initial physical examination, no lymphadenopathy or further lesions were found, except for a small group of red papules located on the skin overlying the lesion (Fig. 1).

The patient underwent a total body CT with contrast, which did not show involvement of other organs, lymphadenopathy, or encephalic masses. The only formation showing an increase in irregular contrast with blurred edges was an expansive subcutaneous mass measuring about 5.3 cm, located in the medial region of the right thigh.

The patient underwent a needle-aspirated cytological examination to rule out any sarcomatous nature of the lesion and, after that, she underwent a biopsy, which showed polymorphic dermal and hypodermic infiltrate, with the presence of emperipolesis (lymphocytes within eukaryotic cells) and positivity for S100 and CD68, with negative CD1a and BRAF, suggestive of CRDD (Fig. 2).

Laboratory tests indicated that the patient's white blood cell count, erythrocyte sedimentation rate (ESR), and C-reactive protein (CRP) were in the normal range.

After surgical excision, the patient was referred to the hematology service for subsequent follow-up. After 7 months the patient had no complications, local or widespread recurrence, or systemic involvement.

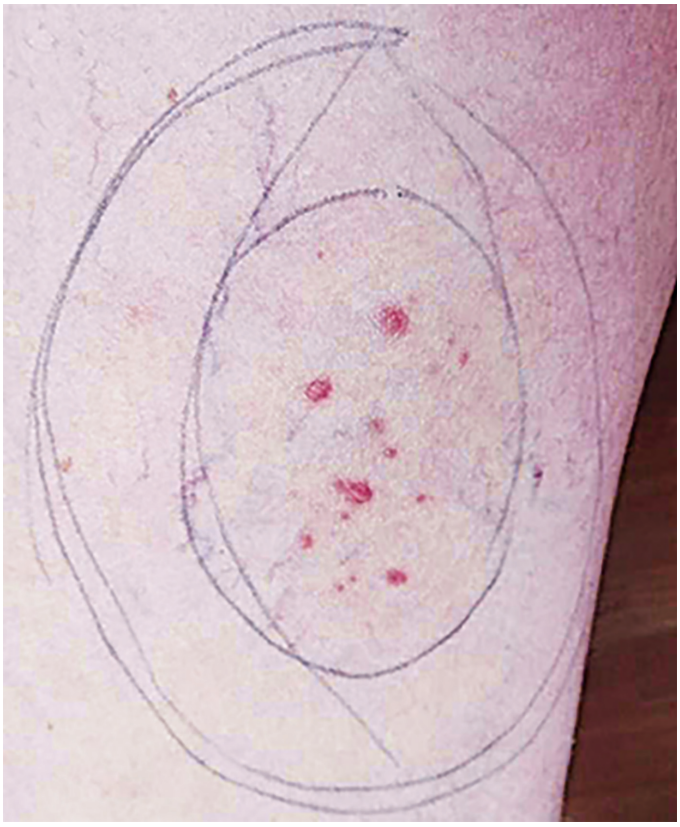

Figure 1 | Grouped skin papules localized on the left leg, associated with a palpable deep nodular lesion.

✉ Corresponding author: anna.campanati@gmail.com

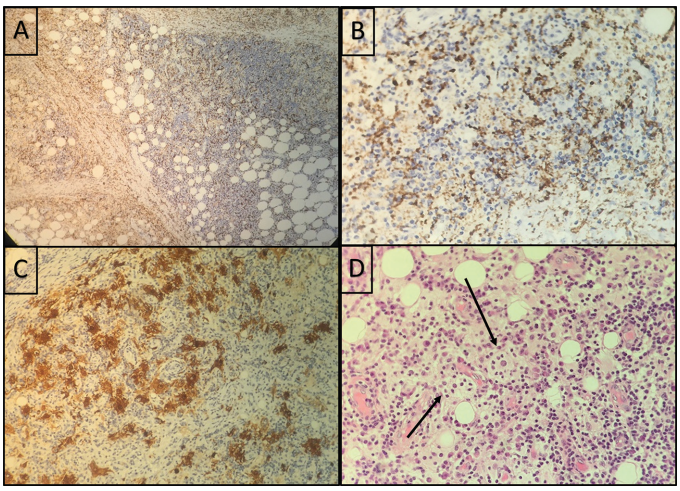

Figure 2 | Histological images showing a) CD68+ at 10×; b) CD68+ at 40×; c) S100+ at 20×; and d) emperipolesis (indicated by black arrows) at 40×.

Discussion

This case report describes a rare clinical variant of purely cutaneous RDD, which involves only 3% of patients with RDD (3).

From a clinical point of view, the diagnosis of CRDD can be very insidious and it may not be suspected because it presents as specific localized, grouped, or solitary papules, plaques, or nodules (4) without any other extracutaneous localizations, and it cannot be easily distinguished from inflammatory, cancerous, or other histiocytic proliferations (5). Laboratory tests should be performed to rule out the most common changes, such as increased ESR, anemia (hypochromic microcytic or normochromic normocytic), reduced serum albumin, and potential monoclonal gammopathy; a total body CT scan or brain CT only (depending on clinical symptoms) should also be performed to evaluate any differential diagnoses such as lymphomas, meningiomas, or Langerhans cell histiocytosis. The conclusive diagnosis derives from close collaboration between the dermatologist and pathologist.

In a review of 220 patients with CRDD, Aadil et al. (6) confirmed the demographic of previous studies: Asians are most affected by the purely cutaneous variant, whereas the systemic form affects Asians, Caucasians, and Africans equally.

Due to the extreme rarity of the disease, the etiology remains unclear, but in the literature it is possible to identify infectious hypotheses such as human herpesvirus 6 (7), a link to lymphoproliferative disease, and immunological disorders. All of these can be triggers of the disease, but there are no indications of direct correlation (6). ARAF, NRAS, KRAS, and MAP2K1 mutations have been reported in patients with CRDD (8–10), and one case was successfully treated with a MAPK-inhibitor drug (8). Treatment can be conservative, with intralesional steroid injection, but this has proven less effective than surgical excision (7). In agreement with our patient, we opted for surgical excision of the lesion.

In conclusion, dermatologists should be aware of the necessity for the patient to be followed for a few months from a clinical and hematological point of view to rule out skin localization being an early symptom of systemic disease. At the same time, they should reassure patients and family members because CRDD has been shown to be a pathology that resolves after the lesion has been removed.

In any case, it would be desirable to have a greater number of studies to more specifically define the possible etiological differences between the two forms of RDD.

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