Do you know this syndrome? *

Você conhece esta síndrome?

Tatiana Mara Costella [1]
José Roberto Paes de Almeida [3]
Ângelo Sementilli [5]
Ney Romiti [2]
Sandra Lopes Mattos e Dinato [4]
Thais Pacheco Lessa Ciofi [6]

CASE REPORT

A 40-year-old white female presented with brown papules of varying sizes and fibroelastic consistency on the upper limbs and upper trunk that she had had for over 20 years. The lesions had grown more painful over the previous 2 years due to exposure to cold weather. She denied any improvement with the use of nifedipine. Had previous history of hysterectomy for uterine fibroids. CT screening was conducted in search of kidney cancer but nothing abnormal was detected.

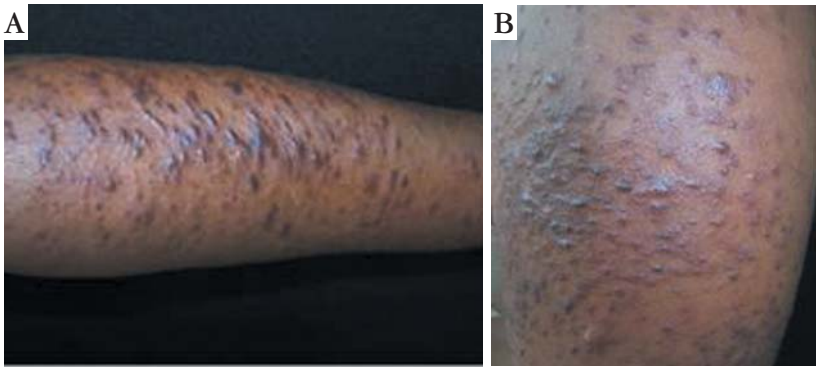

FIGURE 1: A. Proliferative brownish papules distributed linearly on the right forearm. **B.** Brownish papules on the left arm

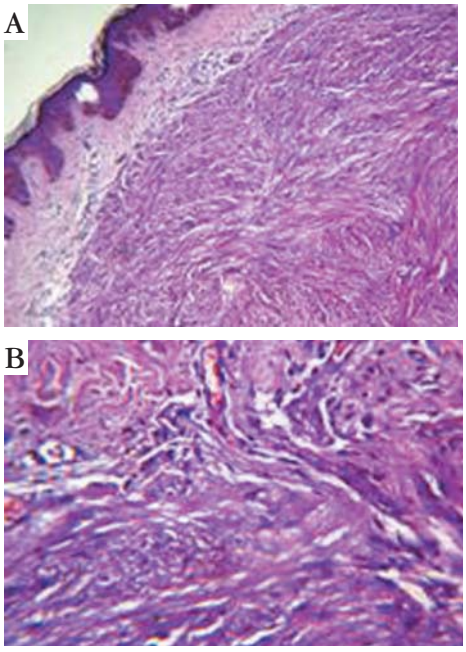

FIGURE 2: A. Skin section containing preserved epidermis and dermis with well-delineated, non-encapsulated nodule composed of spindle cells arranged in bundles. (HE 40x) **B.** Detail of the dermal nodule, which shows uniform nuclei and ill-defined eosinophilic cytoplasm. The bundles are parallel and located in the lower half of the photo (HE 100x)

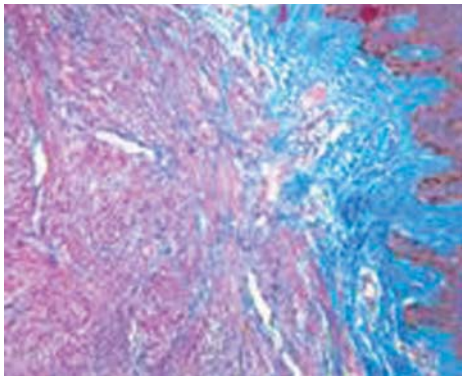

FIGURE 3: Specific staining shows superficial region of the dermis in blue, characterizing collagen, and the dermal nodule, with red-colored cells, which are smooth muscle fibers. (Masson's trichrome 40x)

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[1] Specialist in clinical medicine; Dermatology Intern at Fundação Lusíada (UNILUS), São Paulo (SP), Brazil.
[2] Senior Professor of Dermatology, Fundação Lusíada (UNILUS), São Paulo (SP), Brazil.
[3] Master´s Degree in Dermatology awarded by the Federal University of São Paulo (UNIFESP), São Paulo (SP), Brazil.
[4] PhD, Professor of Dermatology, Faculty of Medicine,University of São Paulo (FMUSP), São Paulo (SP), Brazil.
[5] PhD, Professor of Anatomical Pathology, Federal University of São Paulo (UNIFESP), São Paulo (SP), Brazil.
[6] Dermatology Intern at Fundação Lusíada (UNILUS), São Paulo (SP), Brazil.

DISCUSSION

Reed's syndrome (RS) is an autosomal dominant disorder with incomplete penetrance, characterized by cutaneous and uterine leiomyomas. [1] It was first described in 1954 in a woman of 45 years. [2]

The dermatological manifestation of RS is the presence of cutaneous leiomyomas, benign tumors consisting of fine muscle fibers emerging from the erector muscles of hair, from the genital dartos muscle or from the fine vein muscles. The tumors are characterized clinically as skin-colored or pinkish-brown papules or nodules, with a varied morphology and 0.2 to 2.0 cm in diameter. They may be single or multiple, asymptomatic or painful when pressed or subjected to cold. [3]

Cutaneous leiomyomas are divided into three categories: piloleiomiomas, from the hair erector muscles; angioleiomiomas, which originate from vascular smooth muscle; and dartoide leiomyomas, originating from the dartos muscle of the genitalia, areola and nipple. [4] Cutaneous leiomyomas generally affect the extremities, particularly the extensor surfaces, followed by the trunk, face and neck. They present as firm erythematous or erythematous-brownish, intradermal nodules adhering to the skin but not to deep tissues. [5]

The combination of familial multiple piloleiomiomas with uterine leiomyomas is known as *leiomyomatosis cutis et uteri* or Reed´s syndrome. [6] In these cases, erythrocytosis can result from erythropoietic activity of the tumor. [5]

The predisposition to RS was located on chromosome 1q42.3-43 in the gene encoding fumarate hydratase. This enzyme catalyzes the conversion of fumarate to malate in the Krebs Cycle, but it is believed that it also acts as a suppressor of tumor genes. [7] Several different mutations in this fumarate hydratase gene have been described.

The link between this syndrome and renal cell carcinoma has recently been highlighted. [8,9] However it should be noted that no definitive association was found between the type and location of the mutation and the risk of developing renal cell carcinoma. Isolated cases have been reported linked to endocrine neoplasia type I, [10] to rheumatoid arthritis, [10] and to breast and prostate cancer. [11]The fact that the number of leiomyomas is significantly higher in women than in men suggests a hormonal influence. There are no other known genetic or environmental factors. [12]

In histopathology, discrete bundles of muscle fibers intermingled with collagen can be seen in the dermis. These muscle fibers consist of cells with elongated nuclei and eosinophilic cytoplasm. [13] Immunohistochemical analysis of cutaneous leiomyomas, unlke the uterine leiomyomas, showed no estrogen or progesterone receptors. [7]

Treatment is symptomatic. Excision of painful or unsightly lesions can be performed providing they are small in number. For more extensive and symptomatic cases drugs can be used to block calcium influx to the smooth muscle, such as nifedipine (starter dose of 30mg in three equal doses).Gabapentin, used to relieve chronic neuropathic pain, has been a good therapeutic option which triggers fewer side effects. [14] Electrocoagulation and cryotherapy have been shown to be ineffective. Recurrences are frequent, particularly in patients with multiple lesions. However, it is argued that recurrence might result from partially excised or new lesions. [5]

The Brazilian literature carries little information about Reed´s syndrome. ❑

Abstract: Report on the case of a patient with a combination of multiple uterine and dermatoleiomiomas and uterine miomatosis of long evolution. It is a rarely diagnosed disease known as Reed's syndrome. The study highlights the requirement for, and importance of, systemic investigation for kidney cancer, which is related to the syndrome.

Resumo: Relata-se o caso de uma paciente com associação de múltiplos dermatoleiomiomas e miomatose uterina, de longa evolução. Trata-se de doença pouco diagnosticada, conhecida como Síndrome de Reed. Destaca-se a obrigatoriedade e a importância da investigação sistêmica, em busca de neoplasia renal, que se relaciona com a síndrome.
Palavras-chave: Carcinoma de células renais; Histerectomia vaginal; Leiomioma; Leiomiomatose

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Mailing address / Endereço para correspondência:
Tatiana Mara Costella
Rua Oswaldo Cruz 197, Boqueirão
11.045-003 Santos- SP, Brazil
e-mail: tati_lostris@yahoo.com

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