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Reed's syndrome: segmental piloleimyomas type 1 and uterus myomatosus

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Abstract

Background: Cutaneous mosaicism is a possible cause of segmental skin diseases. Cutaneous leiomyomatosis represent a spectrum of conditions ranging from single lesions to disseminated wide distribution. Reed's syndrome, is an autosomal dominant disorder characterized by multiple cutaneous and uterine leiomyomas.

Main observation: We observed a segmental cutaneous piloleimyomatosis type 1 in a 55-year old female who had an uterus extirpation because of uterine leiomyomas. The cutaneous lesions were moderately painful and localized on her left upper trunk. They presented as firm nodules and small plaques in a linear arrangement. Renal cancer was excluded.

Conclusion: In patients with multiple cutaneous (pilo)leiomyomas a search for underlying systemic diseases is necessary in order to not miss benign or malignant tumors of internal organs. (*J Dermatol Case Rep*. 2014; 8(3): 67-69)

Introduction

Cutaneous leiomyomas are tiny benign tumors of an uncharacteristic clinical appearance. They belong to a group of painful tumors of skin like eccrine spiradenoma, neuroma, dermatofibroma, angiolipoma, neurilemmoma, endometrioma, glomus tumor, and granular cell tumor.[1]

Leiomyomas can be differentiated into solitary, segmental and diffuse or disseminated subtypes. Histologically three different tumors can be identified: Piloleiomyomas are tumors of the smooth arrector pili muscle, leiomyomas of the nipple and genital leiomyomas arising from mammary muscles or tunica dartos of the scrotum, and angioleiomyomas from smooth muscles of blood vessels.[2]

Multiple cutaneous leiomyomas are caused by deleterious mutations in the fumarate hydratase gene.[3,4] Multiple cutaneous leiomyomas with uterine leiomyomas (MCUL; MIM 150800), also known as Reed's syndrome, is a rare disorder that may predispose to renal cancer.[5]

Case Report

A 55-year-old female presented to us with multiple brownish firm nodules and linear plaques arranged segmental in the left supra- and infraclavicular region (Fig. 1). They were asymptomatic except when the patient fastened a safety belt while car driving.

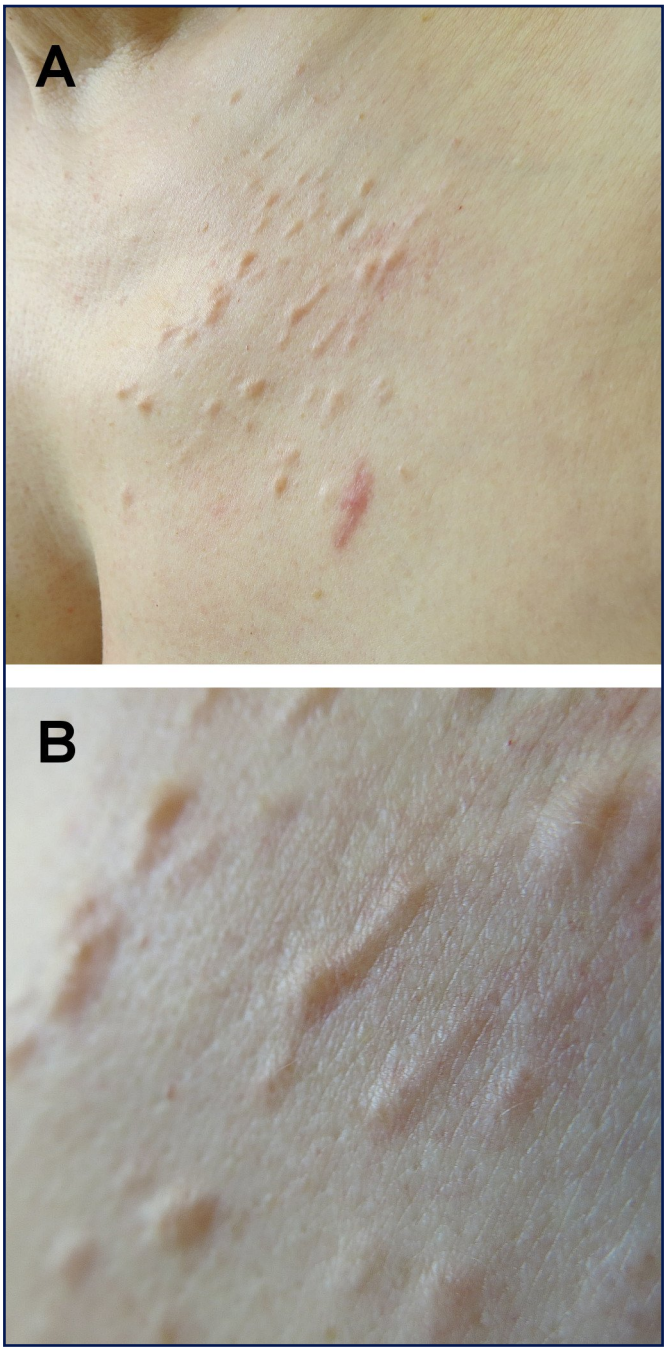
She had a history of uterine extirpation due to multiple leiomyomas (uterus myomatosus) five years ago. Otherwise, her personal and family medical history was unremarkable.

The patient's idea was that of an irritant dermatitis due to the car's safety belt.

A skin biopsy was taken from a lesion (Fig. 2). A circumscribed dermal tumor composed of interlacing and whorled bundles of smooth muscle cells was identified. By van Giesson stain this tumor appeared yellow-orange. The tumor cells were reactive for smooth muscle actin.

The diagnosis of piloleimyoma was confirmed.

Together with the history of an uterus myomatosus with



multiple leiomyomas the final diagnosis was Reed's syndrome or MCUL, type 1. Screening for renal cell cancer by diagnostic ultrasound and contrast-enhanced renal computer tomography were normal. No symptoms indicative for renal cell cancer were found.

Discussion

Segmental (pilo-)leiomyomas with uterine leiomyomatosis is known as MCUL or Reed's syndrome.

The genetic background are mutations of fumarate hydratase gene.[3,4] Two segmental types have been described:

- type 1 with a segmental distribution of cutaneous tumors due to heterozygosity for the mutation(s);
- type 2 where loss of heterozygosity leads to homo- or hemizygosity, with a pronounced segmental arrangement in conjunction with disseminated tumors.[6]

It is important to screen patients with multiple leiomyomas for uterine leiomyomas and renal cell cancer – both occurring in Reed's syndrome.[3-5] In the present patient no renal cancer could be detected. The case seems to be a sporadic mutation since no other family members are affected so far.

Multiple leiomyomas may occur in other syndromes as well. One is Alport syndrome with ocular, cochlear and renal manifestations due to mutations of COL4A5 and COL4A6 genes.[7] Another one is von Recklinghausen's disease, where multiple extracutaneous leimyomas may develop.[8]

Treatment of segmental piloleiomyomas is either surgical or symptomatic. The size of the lesion in our patient would have needed a skin graft with limited esthetic outcome. In case of more painful lesions, symptomatic drug therapy with nifedipine, gabapentin, doxazosin, pregabalin or duloxetine is possible.[9-11]

Figure 1

Segmental piloleiomyomas of the chest.
(A) Overview. (B) Detail with linear arranged mostly skin colored to slightly brownish papules.

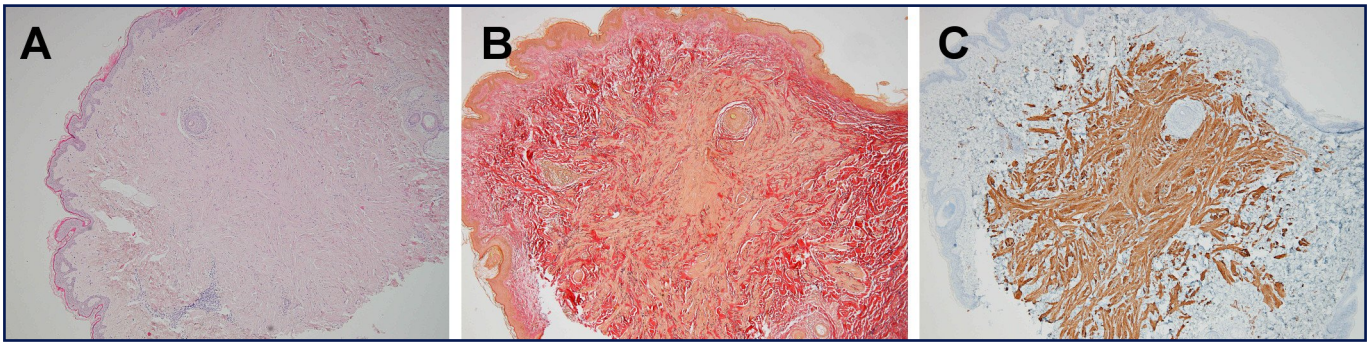


Figure 2

Histology of piloleiomyoma. (A) Overview demonstrating a dermal tumor with whorled and interlaced muscle bundles (hematoxylin-eosin x 4). (B) Van Gieson stain (x 4). (C) Smooth muscle actin (peroxidase staining, x 4).

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