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Case Report

# An Uncommon Case of Multiple Cutaneous Leiomyomas Without Systemic Association

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## ABSTRACT

**Background**: Cutaneous leiomyomas are rare benign smooth muscle tumors that may occur sporadically or as part of Hereditary Leiomyomatosis and Renal Cell Carcinoma (HLRCC) syndrome.
**Case Presentation**: A 46-year-old woman presented with multiple, slowly enlarging, firm, non-tender nodules over the shoulder and bilateral arms for three years. Excision biopsy, initially performed for a presumed lipoma, revealed well-circumscribed dermal nodules composed of smooth muscle cells arranged in interlacing fascicles with cigar-shaped nuclei and eosinophilic cytoplasm. Immunohistochemistry showed positivity for SMA and Desmin, and negativity for S100, confirming multiple cutaneous leiomyomas.
**Management and Outcome:** Surgical excision led to symptomatic relief. In view of multiple lesions, evaluation for HLRCC was advised, including genetic counselling and FH gene testing. Renal imaging showed no associated neoplasms. The patient remains asymptomatic and under regular follow-up.
**Conclusion**: Recognition of multiple cutaneous leiomyomas is important to exclude syndromic associations such as HLRCC.

***Keywords***: *Cutaneous Leiomyoma, Hereditary Leiomyomatosis and Renal Cell Carcinoma, Smooth Muscle Tumor, Fumarate Hydratase (FH) Gene, Immunohistochemistry.*

## BACKGROUND

Cutaneous leiomyoma or piloleiomyoma, are benign smooth muscle tumor arising from the arrector pili muscles of the skin, vascular smooth muscle, or dartos muscle . It is a relatively uncommon neoplasm, accounting for a small fraction of all benign soft tissue tumors. (1)

These lesions typically present as firm, skin-colored to reddish-brown nodules in the dermis or subcutaneous tissue, most commonly involving the extremities, trunk, or genital region.

They may be solitary or multiple, with multiple lesions often associated with hereditary leiomyomatosis and renal cell carcinoma (HLRCC) syndrome, linked to fumarate hydratase (FH) gene mutations (2)

Although benign, these lesions are important to recognize due to their potential association with genetic syndromes, their painful nature, and the need for differentiation from other spindle cell tumors

**CASE PRESENTATION**

A 46-year-old woman presented to the surgery outpatient with multiple cuatneous lumps on the shoulder, right and left arm . These were present for the last 2-3 years and were slowly growing in size . On examination they measured between 2-3 cm were firm , mobile and non tender . The patient did not give any history of uterine fibroids .

Complete blood counts, urea, creatinine, electrolytes, liver function tests were all normal and an excision biopsy was performed for all three lumps with a clinical diagnosis of Lipoma given the multiple nature of the lesions and there location

**Histopathological Findings**

Biopsies from all three lesions displayed comparable features. The sections showed skin with intact epidermis, dermis, and subcutaneous tissue. The dermis contained a well-defined nodular proliferation of smooth muscle cells arranged in interlacing fascicles. The tumor cells possessed elongated, blunt-ended ("cigar-shaped") nuclei, fine chromatin, and eosinophilic cytoplasm with indistinct cell borders. No cytologic atypia, necrosis, or significant mitotic activity was identified. (Fig 1 )

Immunohistochemistry was positive for smooth muscle active (SMA) and Desmin and negative for S100 ( Fig 2 )Given the patient's clinical presentation and her histopathological findings and immunohistochemistry markers , a diagnosis of multiple cutaneous leiomyomas was made .

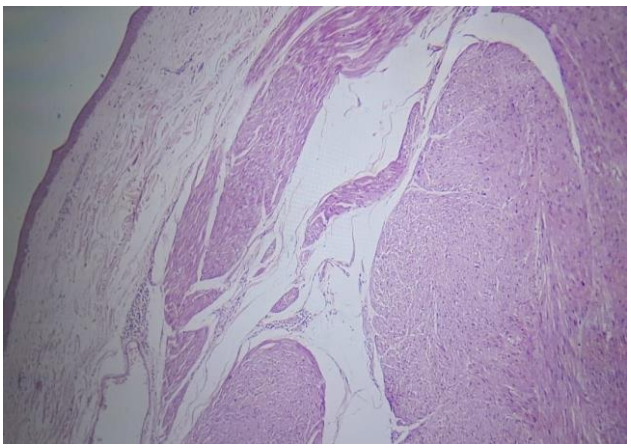

**Fig 1 H &E shows demal nodule of spindle cells (10 X)**

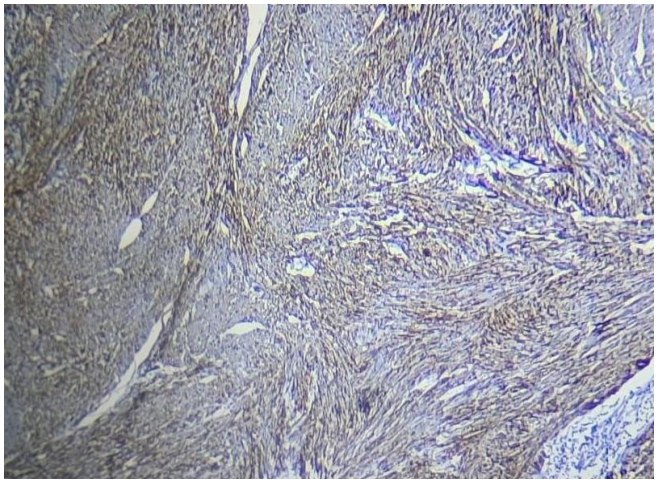

**FIG 2 Diffuse positivity for SMA and Desmin on IHC (40x)**

**Differential Diagnosis**

Clinically, cutaneous leiomyoma may mimic several other painful dermal or subcutaneous nodules. The important ones being dermatofibroma , Neurofibroma or lipoma .

In the present case diagnosis was based on hitopathology findings and immunohistochemistry markers .

**Treatment**

The treatment of cutaneous leiomyomas is dictated by the number of lesions and the degree of discomfort or cosmetic nuisance. When only a few lesions are present, surgical excision is the gold standard for complete removal .

**Outcome and follow up**

The patient reported significant symptomatic relief following surgical excision of the lesions. Considering the presence of multiple cutaneous leiomyomas, evaluation for Hereditary Leiomyomatosis and Renal Cell Carcinoma (HLRCC) syndrome was advised. Genetic counselling and FH gene testing were recommended. Renal imaging performed during follow-up revealed no evidence of renal tumors, and there was no clinical or imaging correlation with uterine leiomyomas. The patient continues to remain under periodic surveillance one year after treatment

## DISCUSSION

Reed's syndrome, also known as multiple cutaneous and uterine leiomyomatosis (MCUL), is a rare genetic disorder characterized by the presence of multiple smooth muscle tumors primarily involving the skin and uterus. The condition results from mutations in the fumarate hydratase (FH) gene, which encodes an enzyme critical to the citric acid (Krebs) cycle. (3)

In the present case, the patient developed multiple cutaneous leiomyomas over the shoulder and hand, which had gradually increased in size and number over several years. Histopathological examination and immunohistochemistry confirmed the diagnosis of smooth muscle tumors. However, no associated uterine or renal lesions were detected on imaging studies. Genetic counselling and FH gene testing were advised, and the patient remains under regular clinical and imaging surveillance

The treatment of cutaneous leiomyomas is dictated by the number of lesions and the degree of discomfort or cosmetic nuisance. When only a few lesions are present, surgical excision is the gold standard for complete removal (4)

**Learning Points**

- Cutaneous leiomyomas, though rare, are important to recognize due to their potential clinical significance.
- The presence of multiple lesions should prompt evaluation for Hereditary Leiomyomatosis and Renal Cell Carcinoma (HLRCC) syndrome.
- Genetic counselling and FH gene testing play a crucial role in identifying syndromic associations.
- Regular surveillance is essential to detect possible systemic malignancies, particularly renal tumors.
- Histopathological confirmation remains the gold standard for accurate diagnosis and for distinguishing leiomyomas from other cutaneous neoplasms.

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