

Case Report

Familial leiomyoma: a case report

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ABSTRACT

Cutaneous leiomyomas are benign tumors that can be exquisitely painful. Comprise three distinct types such as piloleiomyoma, angioleiomyoma, and genital leiomyoma. Piloleiomyomas, derived from arrector pili muscle, are solitary or multiple firm papulonodules located mostly on the extremities and trunk; genital leiomyomas, derived from dartos, vulvar, or mammary smooth muscle, are solitary or papulonodules located on the scrotum, vulva, or nipple; and angioleiomyomas, include solid, cavernous, or venous subtypes, are derived from the tunica media of small arteries and veins and typically present on the extremities. Excisional biopsy is required for diagnosing all cutaneous leiomyomas. Histology shows interlacing fibers of spindle cells with moderate amounts of eosinophilic cytoplasm and a blunt-ended, elongated nucleus with perinuclear halos. Surgical excision is curative for cutaneous leiomyomas, with other management options including medical or destructive therapy. A 36-year-old male patient presented with a 13 years history of painful, multiple lesions over the back. Based on the histopathological examination, imaging, and past medical records, a diagnosis of familial leiomyoma was made. His brother had similar disease. The patient was started on gabapentin 300mg at night. Patient advised for carbon dioxide laser excision. The present case report has been reported for its interesting clinical presentations and rarity.

Keywords: Familial, Leiomyoma, Leiomyoma cutis

INTRODUCTION

Cutaneous tumors with smooth muscle differentiation can be categorized according to origin: (a) arrector pili muscles, (b) smooth muscle of genital skin (dartos muscle), vulva, nipple and areolar region, and (c) walls of blood vessels.

Each of these can give rise to benign superficial smooth muscle tumors (leiomyomas) as well as their malignant counterparts (leiomyosarcomas).¹

We report a case of familial leiomyoma in a 36-year-old male patient.

CASE REPORT

A 36-year-old male patient presented with a 13 years history of painful, multiple lesion over the back. The lesions were first few in number and gradually increased in number over the years. Patient complains of pain on pressure and exposure to cold environment. Pain was sharp, shooting type.

No spontaneous resolution was seen over years. No significant medical history. There is similar history seen in a family member, his brother. He had similar lesions over the back (Figure 4). However he was not available for examination.

On examination: Physical examination revealed multiple, discrete pink to dark brown colored firm, shiny, tender nodules ranging from 2 mm to 20 mm present over the left scapular region not crossing the midline with no significant surface change. (Figure 1 and 2)



Figure 1: Multiple, discrete pink to dark brown colored firm shiny nodules present over the left scapular region.



Figure 2: Multiple, discrete pink to dark brown colored firm shiny nodules present over the left scapular region.

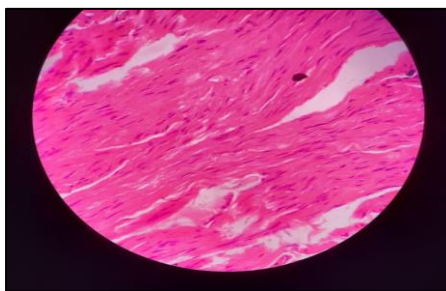


Figure 3: Section showing Interlaced fibers of smooth muscle cells with abundant eosinophilic cytoplasm and elongated nuclei with blunt ends.

Systemic examination was unremarkable complete blood count (CBC), erythrocyte sedimentation rate (ESR), fasting blood sugar (FBS), post prandial blood sugar (PPBS), liver function test (LFT), renal function test (RFT), thyroid stimulating hormone (TSH), lipid profile, urine routine, USG abdomen and pelvis was normal.

A skin biopsy was obtained from a lesion over the back. Histopathology examination revealed interlaced fibers of smooth muscle cells with abundant eosinophilic cytoplasm and elongated nuclei with blunt ends. (Figure 3)



Figure 4: Multiple, discrete brown colored firm shiny nodules present over left scapular region (seen in his brother).

On the basis of clinicohistopathology correlation, the diagnosis of multiple familial leiomyoma was made.

Patient was started on tab. Gabapentin 300 mg at bedtime. Patient was advised about carbon dioxide (CO₂) laser, but the patient was not willing. Patient also counseled about it and chances of recurrence.

DISCUSSION

The familial form of multiple leiomyomas, characterized by autosomal dominant inheritance and variable penetrance, was described by Virchow in 1854 and Koepfler et al. in 1858.^{2,3}

Clinically, cutaneous leiomyomas appear as multiple reddish violet, 'mother-of-pearl' colored papules or nodules. The lesions are firm, rounded and vary in size. They range in number from a few scattered lesions to more than 5,000.⁴

Multiple cutaneous leiomyomas are painful. Pain can be spontaneous or occur on exposure to cold, pressure, injury or emotional stress.^{5,6,7}

Our case is a presentation in a 36 year old male with family history of similar lesions in his brother. Our patient had visited the OPD in view of cosmetic concern, and pain.

The prognosis of the condition to recur was explained to the patient.

Patient was started on tab. Gabapentin 300mg at bedtime. Patient was advised about carbon dioxide (CO₂) laser, but the patient was not willing.

Associations have been seen between leiomyomas and uterine myoma, fibroma, epidermoid cyst, osteoma and intestinal polyposis.⁸

Recent evidence shows that most patients presenting with multiple cutaneous leiomyomas have a germline loss-of-function mutation in the fumarate hydratase gene (FH), encoding an enzyme that functions as part of the Krebs cycle by converting fumarate to malate.^{9,10}

Renal tumors present in approximately 10% to 16% of individuals with hereditary leiomyomatosis and renal cell cancer and tend to occur more commonly in adult females than in adult males.^{11,12}

CO2 laser ablation, cryotherapy, and electrosurgery are treatment modalities that used for cutaneous leiomyomatosis, where surgical excision is not feasible.¹³

Symptomatic treatment include nitroglycerin, nifedipine, phenoxybenzamine, gabapentin, intralesional botulinum toxin, and topical analgesics.¹⁴⁻¹⁸

CONCLUSION

Cutaneous leiomyomas are rare lesions and an important clinical differential diagnosis of painful papulonodules.

Current knowledge of this cutaneous disorder is important in clinical practice, as comprehensive clinical assessments and patient education can provide positive outcomes to affected individuals.

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