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| RESEARCH ARTICLE

Surgical Management of Giant, Disfiguring Facial Angiofibromas in Tuberous Sclerosis

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| ABSTRACT

Background: Tuberous sclerosis complex (TSC) is a rare genetic disorder that affects many systems in the body. It is marked by benign hamartomas in many organs, especially the skin, brain, and kidneys. Facial angiofibromas are one of the most obvious skin problems that can lead to severe cosmetic damage and mental distress if not treated. Case Presentation: We are writing about a 41-year-old man who had a history of TSC, epilepsy, intellectual disability, and mental illness. He came in with multiple facial lesions that were getting bigger and causing severe disfigurement and partial airway obstruction. A physical exam showed many dark brown, round, soft tumours on the nose, cheeks, eyelids, ears, and forehead. The nostrils were completely blocked. The lesions were surgically removed from the patient, and then a split-thickness skin graft was done. Histopathology showed that there were more than one angiofibromas. Conclusion: This case shows how hard it is to treat large facial angiofibromas in TSC, especially in people who have neuropsychiatric problems and can't talk well. Surgical excision remains a viable option in advanced cases where conservative therapy is insufficient, offering both functional and cosmetic improvement.

| KEYWORDS

Angiofibroma, tuberous sclerosis, adenoma sebaceum, cutaneous

| ARTICLE INFORMATION

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1. Introduction

Tuberous sclerosis complex (TSC) is an inherited neurocutaneous disorder resulting from pathogenic mutations in the TSC1 or TSC2 genes, which cause abnormal activation of the mTOR pathway and the development of hamartomatous lesions in various organ systems (Curatolo et al., 2020). The estimated prevalence of TSC is about 1 in 6,000 live births, with varying levels of neurological and dermatological involvement (Northrup et al., 2021).

Cutaneous manifestations, such as hypomelanotic macules, facial angiofibromas, shagreen patches, and ungual fibromas, are essential diagnostic characteristics (Gupta & Agarwal, 2020). Facial angiofibromas, formerly known as adenoma sebaceum, generally manifest in childhood and progressively increase in size throughout adolescence and adulthood. Although frequently benign, extensive facial lesions may lead to cosmetic deformity, nasal obstruction, haemorrhage, and psychological distress (Sharma et al., 2019).

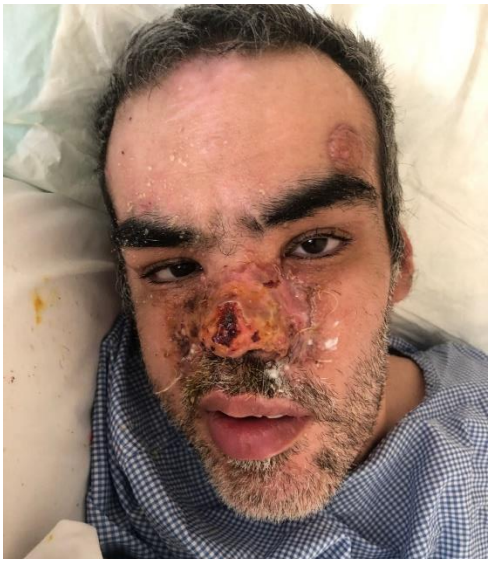
This report describes a case of a middle-aged male with untreated, extensive facial angiofibromas related to TSC, effectively treated through surgical excision and split-thickness skin grafting. The case underscores the multidisciplinary challenges in managing advanced dermatologic sequelae in neuropsychiatrically impaired individuals.

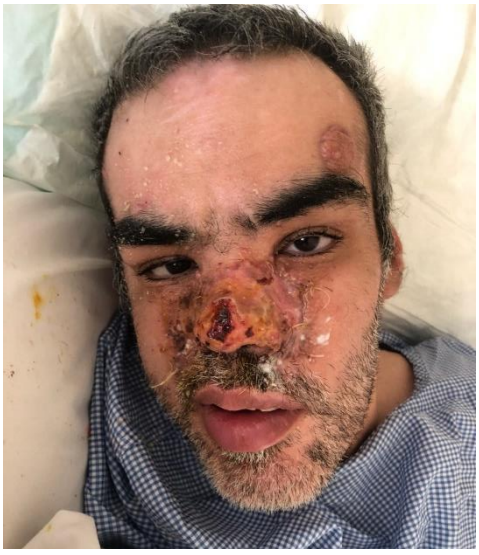
2. Case Presentation

A 41-year-old male, known to have Tuberous Sclerosis, Epilepsy, Intellectual Disability, and Psychiatric Illness, was referred from his psychiatric institution to the plastic surgery clinic with multiple painless, progressively enlarging facial lesions. The lesions had been growing gradually over several years without prior treatment.

On examination, the patient was bedridden with severe contractures of the lower limbs and was non-verbal, able to express only through sounds. Multiple spherical, soft, dark brown tumors were present on the nose, cheeks, eyelids, ears, and forehead. The lesions were thick, rough-surfaced, and disfiguring, completely obscuring the nostrils and causing the patient to breathe exclusively through his mouth. Oxygen saturation was normal on room air.

The patient underwent surgical excision of the lesions. The resultant defects were covered with split thickness skin grafts. Histopathological examination confirmed the diagnosis of multiple angiofibromas.





3. Discussion

1. Pathogenesis and Clinical Features of Tuberous Sclerosis

Tuberous sclerosis complex results from mutations in *TSC1* (hamartin) or *TSC2* (tuberin), which form a complex that regulates the mammalian target of rapamycin (mTOR) signaling pathway. Dysfunction leads to uncontrolled cellular proliferation and hamartoma formation (Henske & Jóźwiak, 2021). The clinical presentation of TSC is heterogeneous, with dermatologic findings often serving as early diagnostic clues.

2. Differential Diagnosis of Facial Angiofibromas

The clinical appearance of facial angiofibromas may mimic several dermatologic conditions, including trichoepithelioma, syringoma, sebaceous hyperplasia, and basal cell carcinoma. Trichoepithelioma typically manifests as small, firm papules during puberty, whereas syringomas present as flesh-colored papules on the eyelids in young adults (Chaurasia et al., 2020). Accurate differentiation is critical to avoid misdiagnosis and inappropriate management.

3. Treatment Modalities

Treatment of facial angiofibromas aims to improve cosmesis and prevent recurrence. Options range from non-invasive topical therapies to surgical interventions.

Topical therapies: such as rapamycin (sirolimus), timolol, and tacrolimus have demonstrated efficacy in reducing lesion size and erythema by inhibiting mTOR-mediated fibroblast proliferation (Wataya-Kaneda et al., 2018). These are particularly effective in early or small lesions (<4 mm) and have minimal adverse effects.

Procedural therapies: include shave excision, cryotherapy, electrodessication, dermabrasion, and laser ablation (PDL, CO_2, or Er: YAG lasers). However, these may lead to scarring, post-inflammatory pigmentation, and recurrence rates as high as 80% (Seibert et al., 2019).

Surgical excision: remains the mainstay for extensive, disfiguring cases such as this one. Excision with grafting or local flap reconstruction offers rapid functional and cosmetic restoration. In this patient, excision with split-thickness skin grafting provided satisfactory coverage and breathing improvement.

Table 1. Summary of treatment options for facial angiofibromas in Tuberous Sclerosis Complex.

Treatment Modality	Indication	Advantages	Limitations / Risks
Topical rapamycin/timolol	Mild to moderate lesions	Non-invasive, good cosmetic result	Requires long-term use, relapse possible
Laser ablation (PDL/CO_2)	Moderate lesions	Controlled removal, less bleeding	Hyperpigmentation, recurrence
Shave excision	Isolated lesions	Simple, outpatient	Risk of scarring
Surgical excision with graft	Extensive lesions	Definitive removal, functional restoration	Graft failure, visible scarring

4. Conclusion

Facial angiofibromas, though benign, can lead to severe aesthetic and functional impairment in patients with Tuberous Sclerosis Complex. Early diagnosis and treatment can prevent progression to disfiguring stages. In advanced cases, surgical excision with grafting provides an effective solution, offering significant improvement in the patient's quality of life. This case emphasizes the need for multidisciplinary collaboration in the management of TSC-related dermatoses.

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