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(CASE REPORT)

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Atypical and voluminous skin manifestation of Bourneville tuberous sclerosis: A case report of giant scalp hamartoma

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Abstract

Bourneville tuberous sclerosis (BTS) is an autosomal dominant phakomatosis characterised by multisystemic involvement, particularly of the skin. Although facial angiofibromas are the most common lesions, severe or atypical forms affecting the scalp are rare. We report the case of a 26-year-old female patient presenting with a giant, highly progressive lesion of the scalp associated with BTS. The gradual increase in volume, the tumour-like appearance, and the major aesthetic impact prompted surgical intervention. Extensive excision followed by appropriate reconstruction was performed with good aesthetic and functional results. This case illustrates the value of plastic surgery in the management of severe and unusual cutaneous forms of tuberous sclerosis complex. It highlights the importance of reconstructive surgery in advanced forms of TSC.

Keywords: Bourneville tuberous sclerosis; Hamartoma; Scalp

1. Introduction

Bourneville tuberous sclerosis (BTS) is an autosomal dominant phakomatoses linked to mutations in the TSC1 and TSC2 genes, leading to the formation of multiple hamartomas in various organs (1). Skin manifestations are one of the first clinical signs of the disease and are reported in more than 90% of patients, including angiofibromas, fibrous plaques, cutaneous hamartomas, and nodular lesions that can become quite large (2,3).

In severe forms, these skin lesions can become giant, disfiguring, and a source of haemorrhagic complications or infections, causing major functional, aesthetic, and psychological repercussions. The management of these advanced forms often requires a multidisciplinary approach in which plastic surgery plays an essential role, particularly for excision and reconstruction of tissue loss (4).

2. Case presentation

A 26-year-old female patient with no previous significant pathological history, followed up for BTS, who has had a large, progressive lesion in the parieto-occipito-temporal region of the left scalp since infancy. The tumour-like lesion caused significant functional and aesthetic discomfort and had a notable psychological impact. Clinical examination revealed a well-defined, firm mass measuring 12 cm in diameter, with no signs of inflammation, covering a significant portion of the scalp. No associated neurological deficits were noted.

Preoperative evaluation included imaging (CT scan) to assess the extent of the lesion and its connection to subjacent structures. Due to the size and tumour-like appearance of the lesion, extensive surgical excision was performed,

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followed by reconstruction using a thin skin graft. Postoperative healing was satisfactory, with excellent aesthetic and functional results, and no recurrence was observed in the short term.

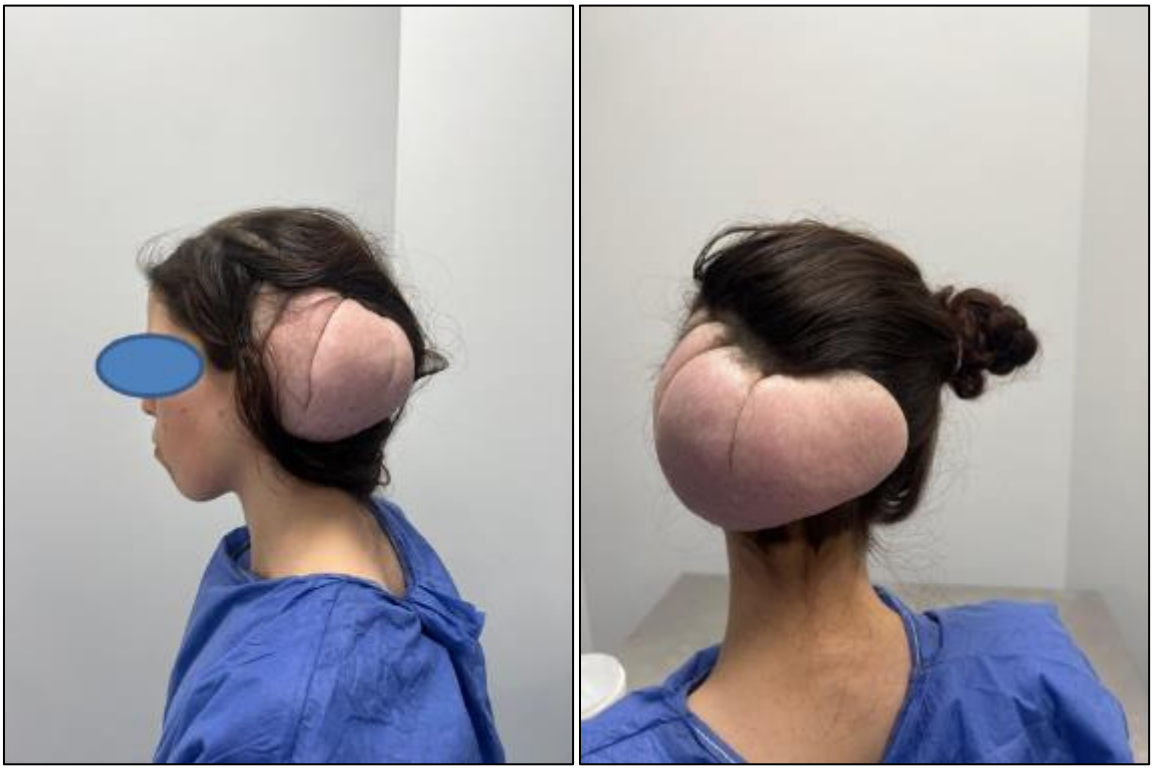

Figure 1 26-year-old female patient with a follicular sebaceous hamartoma**.**

Figure 2 26-year-old female patient with a follicular sebaceous hamartoma seen from the side

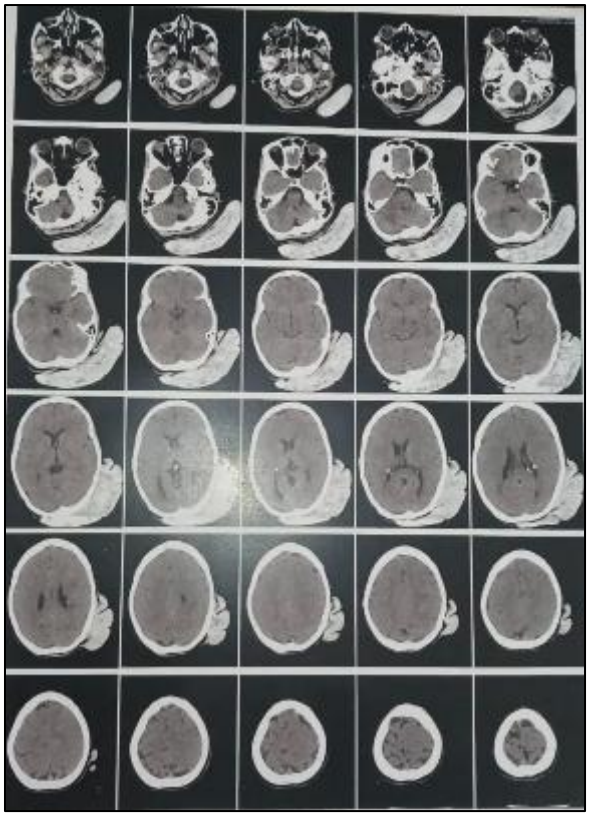

Figure 3 Brain CT scan showing parieto-occipito-temporal hamartoma.

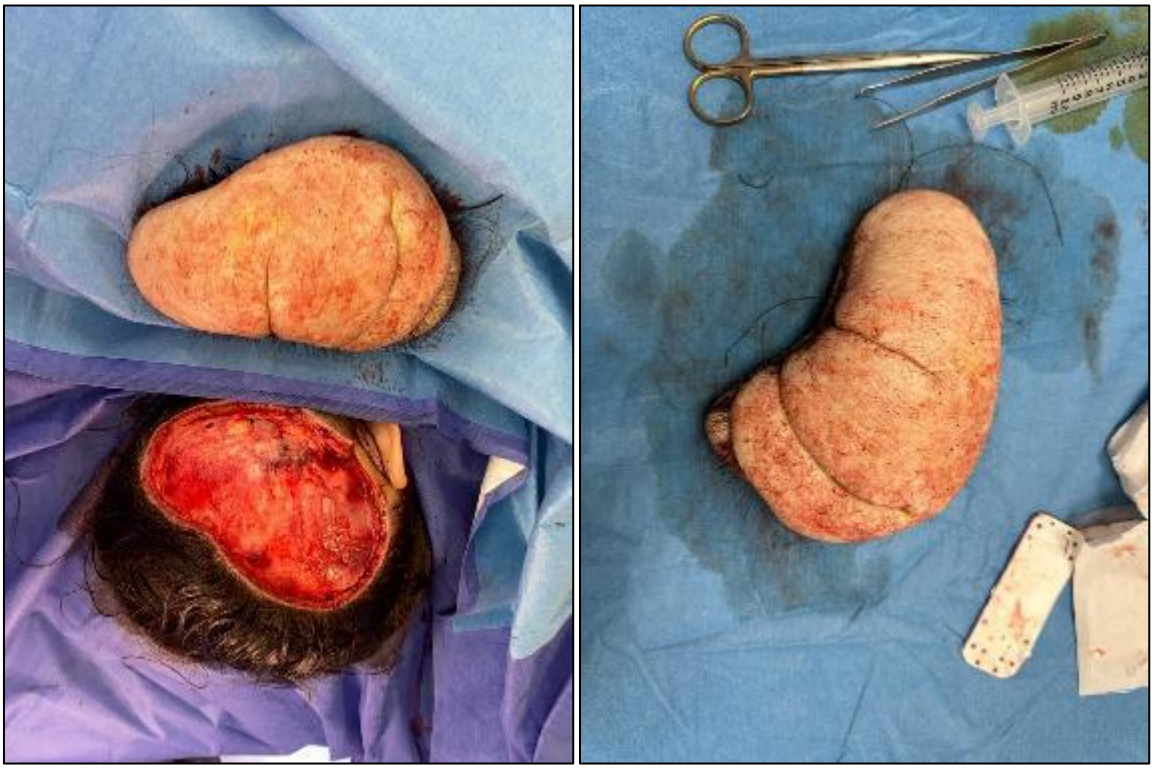

Figure 4 Per-operative images of tumour excision

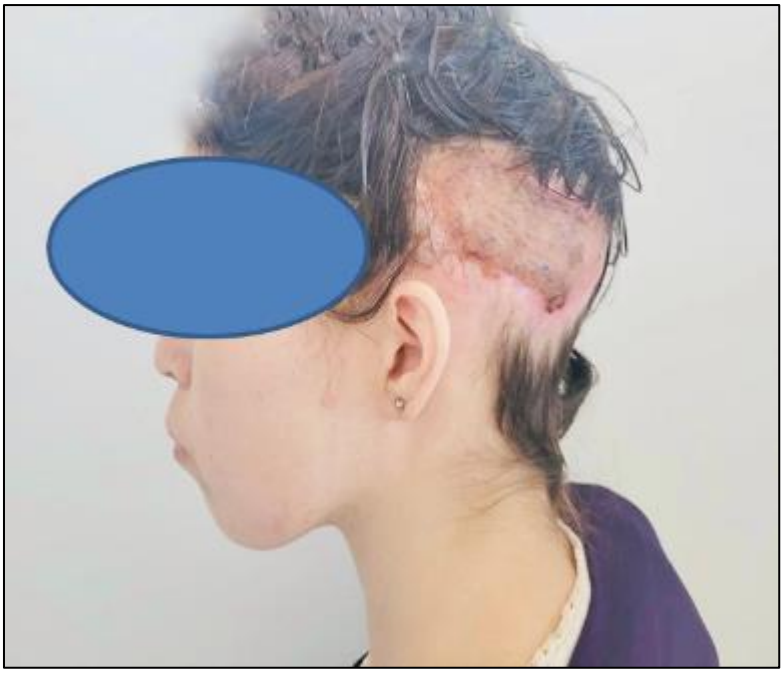

Figure 5 Post-operative image after skin grafting

3. Discussion

Bourneville tuberous sclerosis (BTS) is an autosomal dominant genetic disorder characterised by the formation of benign tumours in several organs, including the skin, brain, kidneys and heart. Among the skin manifestations, facial angiofibromas are the most common, while large lesions on the scalp remain rare [5].

3.1. Characteristics of skin lesions in BTS

Skin lesions associated with BTS can vary greatly in terms of location, size, and progression. Hypomelanotic plaques, nail fibromas, angiofibromas and facial fibromas constitute most skin lesions [5]. Giant scalp lesions, as in our case, are rarely described in the literature, making them atypical and complex to manage [6]. These lesions may progress slowly but can cause significant deformity, psychological discomfort, and, in some cases, a risk of local complications (ulceration, infection).

3.2. Therapeutic approaches

Surgical treatment remains the treatment of choice for large, disfiguring lesions. Complete excision minimises the risk of recurrence and improves appearance [7]. Post-excision reconstruction may require different techniques:

- Local flaps: adapted to moderate tissue loss or areas with sufficient mobile skin.
- Skin grafts: useful for covering large zones when skin mobility is limited.
- Tissue expansion: sometimes used to provide aesthetic skin coverage on the scalp.

In our case, a thin skin graft was sufficient to achieve effective reconstruction, with a satisfactory functional and aesthetic result.

3.3. Complementary medical approaches

mTOR inhibitors (such as everolimus or sirolimus) have been shown to be effective in reducing the size of certain skin and internal lesions in BTS [5,6]. However, their effectiveness on large lesions of the scalp remains limited, and surgery is still necessary to restore aesthetic appearance and avoid local complications.

Optimal management of patients with BTS requires collaboration between dermatologists, neurologists, plastic surgeons and, where appropriate, nephrologists or cardiologists. The aim is to assess multisystemic involvement as a global entity, plan surgical management, and ensure appropriate post-operative follow-up.

4. Literature review

Skin lesions of the scalp in tuberous sclerosis complex remain poorly described in the literature, with most publications focusing on facial angiofibromas, frontal fibrous plaques, and periungual fibromas [1,2]. Large scalp lesions with a pseudo-tumoural appearance are particularly rare and constitute a minority of the dermatological manifestations reported in BTS.

In previously reported cases, scalp lesions most often present as localised fibromas of moderate size. Giant forms, reaching several centimetres in size and altering the contour of the scalp, are exceptional. These cases are generally characterised by slow but continuous growth [3].

In terms of treatment, the literature agrees on the necessity of complete surgical excision for large lesions. Several reconstructive techniques have been described:

- Local rotation or transposition flaps are used when there is sufficient skin surrounding the lesion in terms of quantity and mobility.
- Skin grafts, reserved for extensive tissue loss or in areas where the scalp is not lax to provide primary coverage.
- Tissue expanders, indicated for major tissue loss, but requiring prolonged operating time and rigorous follow-up.
- Combined reconstructions, combining excision, tissue expansion, and local flaps, in the most complex cases.

Most authors consider local flaps to be the technique of choice in terms of aesthetics, as they preserve the texture, colour and pilosity of the scalp [2]. However, in situations where there is excessive loss of substance, skin grafting remains an effective alternative, although its main disadvantage is the absence of pilosity at the graft site.

In our case, extensive excision resulted in a loss of substance that could not be satisfactorily covered by a local flap alone. A thin skin graft was therefore performed to ensure complete skin coverage. This choice is in line with the options mentioned in the literature for extensive scalp lesions. Despite the aesthetic limitations inherent in this technique, mainly the absence of hair regrowth, the result obtained was satisfactory, with proper healing and no significant post-operative complications. Compared to the cases described above, our observation is distinguished by:

- The exceptional size of the lesion, rarely reported in cutaneous forms of BTS.
- The necessary use of a thin skin graft, whereas some similar cases have benefited from local flaps or tissue expansion.
- A satisfactory scar result, despite the absence of hair at the graft site, demonstrates the relevance of this strategy when scalp mobility is limited.
- This case therefore makes an additional contribution to the literature by illustrating that, in giant scalp lesions associated with BTS, skin graft reconstruction can be a reliable and effective solution when local flaps are not an option.

Table 1 Comparison of the results of our study with other research

Year / Reference	Age / Gender	Lesion type	Location	Surgical technique	Result	Remarks
2021 — Freitas et al. (Atypical cutaneos presentation *Giant angiofibroma on the scalp*)	26 / F	Giant angio-fibroma	Occi-pital scalp	Large excision + reconstruction/flap	Satisfactory aesthetic result, no recurrence reported in the short term.	Case very similar to our presentation (large size)
2014 — Sharma et al. (BMJ Case	23 / F	Scalp fibroma (hamar-tome)	Scalp	Surgical excision; primary closure	Satisfactory healing, no major	A rare lesion as a cutaneous manifestation

Reports : Scalp fibroma)					complications reported	
2012 — Kacerovska et al. (Giant angiofibromas in TSC)	Several cases	Giant angiofibromas	Face/scalp	Excision ± reconstruction (flaps/grafts depending on loss of substance)	Generally favourable results	Series describing the rarity and variability of 'giant' forms.

Our case	26 / F	Giant lesion of the scalp	Scalp pariéto-occipito-temporale	Large excision + reconstruction using a thin skin graft	Satisfactory healing; excellent aesthetic and functional outcome; no recurrence in the short term.	Very rare lesion

Positioning of our case in relation to the literature:

Our observation is clearly distinct from the cases described in the literature in several significant ways:

- An exceptionally large lesion, with a volume far exceeding that is usually reported in skin lesions associated with BTS. Descriptions of giant scalp masses associated with this condition remain extremely rare.
- A major aesthetic impact, comparable only to the most severe forms published, causing significant psychosocial discomfort and justifying surgical treatment.
- An appropriate reconstructive strategy, combining complete excision and thin skin grafting, which differs from certain publications where the authors favoured.

In our case, the extent of the loss of substance did not allow for direct closure or coverage by flap alone, making grafting essential.

Satisfactory post-operative results, characterized by:

- Complete and stable skin coverage.
- Good graft take.
- No immediate complications.
- And no recurrence in the short term.

Although the graft inevitably results in an area of hair loss, the overall result remains harmonious given the initial extent of the lesion, in line with the conclusions of the authors, who emphasise that, for massive forms, the priority remains the restoration of acceptable volume and cranial contour.

This case therefore enriches the literature by documenting a particularly rare and spectacular form of scalp lesion associated with BTS. It also highlights that, even in giant forms, effective reconstruction can be achieved without systematically resorting to invasive techniques such as tissue expansion, provided that thin skin grafting is judiciously indicated.

5. Conclusion

This case of giant cutaneous hamartoma of the scalp is an extremely rare presentation of Bourneville tuberous sclerosis. The therapeutic approach to lesions of this size requires radical surgical excision for aesthetic and functional reasons, supplemented by reliable and effective reconstruction.

Compliance with ethical standards

Disclosure of conflict of interest

No conflict of interest to be disclosed.

Statement of informed consent

Written Informed consent was obtained from the patient for publication of this case report and accompanying images.

References

[1] Curatolo P, Moavero R, de Vries PJ. Neurological and neuropsychiatric aspects of tuberous sclerosis complex. Lancet Neurol. 2015;14(7):733-45.

[2] Northrup H, Krueger DA; International Tuberous Sclerosis Complex Consensus Group. Tuberous sclerosis complex diagnostic criteria update. Pediatr Neurol. 2013;49:243-54.

[3] Kingswood JC, et al. Tuberous Sclerosis Complex: Review of the clinical features, diagnosis, and management. Orphanet J Rare Dis. 2021;16:1-15.

[4] Schwartz RA, Fernández G, Kotulska K, Jóźwiak S. Tuberous sclerosis complex: Advances in diagnosis, genetics, and management. J Am Acad Dermatol. 2007;57(2):189-202.

[5] Northrup H, Krueger DA. Tuberous Sclerosis Complex Diagnostic Criteria Update: Recommendations of the 2012 International Tuberous Sclerosis Complex Consensus Conference. Pediatr Neurol. 2013;49:243–254.

[6] Jozwiak S, Kotulska K, Domanska-Pakiela D, et al. Clinical and molecular aspects of tuberous sclerosis complex. Lancet Neurol. 2008;7:222–235.

[7] Bissler JJ, Kingswood JC. Renal manifestations of tuberous sclerosis complex. Kidney Int. 2004;66:158–167.